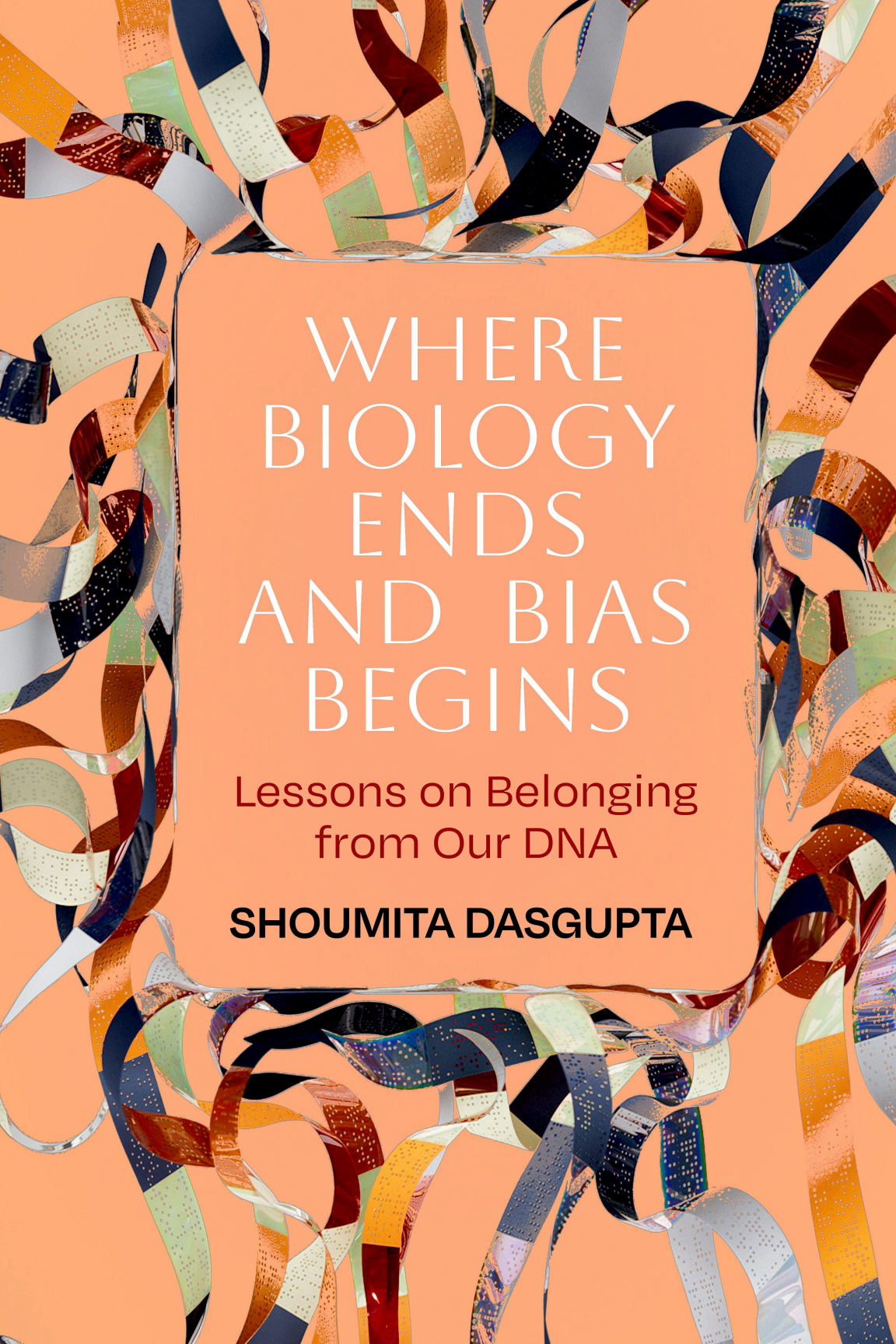




WHERE BIOLOGY ENDS AND BIAS BEGINS

Lessons on Belonging
from Our DNA

SHOUMITA DASGUPTA

*Where Biology Ends
and Bias Begins*

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LESSONS ON BELONGING
FROM OUR DNA

Shoumita Dasgupta



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Do the best you can until you know better.

Then when you know better, do better.

—Maya Angelou

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1 Why Me?

This book is not about me. But it is also inextricably linked to the lenses through which I see the world. As a scientist, I am trained as a truth seeker to weigh evidence and make sense of what it tells us about how the world works. A well-designed experiment can ask and answer complex questions and settle debates. Scientists can disagree, but ultimately, the data will lead us to the truth. When it comes to societal matters, however, things are rarely so easily settled. My own lived experiences as a first-generation American influence the ways in which I move through the world and consequently my views about the communities in which I hold space. For this reason, I thought it would be important to tell you a little bit about my personal history and how it ties into American history as well.

My parents were born and raised in South Asia in the 1940s. When my father was born in Chittagong, it was still part of India, but with the British partition of India in 1947, Chittagong became an anchor of East Pakistan. East Pakistan eventually became Bangladesh, and it was envisioned as a nation state for South Asian Muslims while India was meant to be the nation state for South Asian Hindus. My family is a Hindu Bengali family, so with the partition came emigration. My father's side of the family emigrated to what we now know as India, as part of a mass,

often violent, migration of Muslim and Hindu families across a new border, trying to establish new communities hundreds of miles from any place where they previously had roots.

My mother's side of the family was from Assam and then later the outskirts of Kolkata (known as Calcutta in those years of British rule), both areas where a Hindu majority resided, so they were fortunate to escape the traumas of forced relocation. Her father worked for the Indian Railways, and as a result, she had access to travel and experiences that many did not. She was able to attend boarding school and take advantage of educational opportunities that were not universally available to someone of her gender. When my mother's cousin married my paternal uncle, my mother came to know of my father. At first, he was just someone from stories, bookish, studying chemistry in Germany, having escaped much of the turmoil of India in those days, while my mother studied toward her masters in psychology closer to home. Plans were ultimately set in motion by both families to arrange a match between them, and before they knew it, they were planning a wedding and a future in America. My father came over first, scoping things out in Ohio and restarting his undergraduate studies in chemistry, ultimately earning a PhD in polymer chemistry from the University of Akron. My mother arrived soon thereafter, traveling with a group of Indian women all setting off to begin new lives in the West.

My parents' arrival in the United States in the 1970s was a direct result of the 1965 Immigration and Nationality Act. This act was passed at the height of the Civil Rights Movement and sought to eliminate previous immigration quotas based on nationality for their discriminatory practices. The act prohibited discrimination on the basis of nationality, race, sex, and other criteria, instead replacing this with a system of preference based on professional skills and family reunification. This was happening at a time when Indian educational systems were training large numbers of highly skilled graduates. As they could not all be employed locally, people started looking outward and contributed to a wave of immigrants from South Asia to the United States, doubling the South Asian population here every decade since the 1980s. Surely these immigration policies contributed to the image of South Asians as model minorities. My parents were at the leading edge of that wave of immigrants, and they were only able to be here due to the hard-fought battles for equity driven by Black

Americans in the 1960s. In many ways, I owe my own story to American civil rights leaders for paving the way for families like mine.

Because my parents were early members of the South Asian diaspora in the United States, they had to learn many things about resettling on their own. Although India was a British colony and English was one of the national languages, there was still a great deal about resettling in the United States that highlighted my family's foreignness. Navigating these unfamiliar systems is challenging even when language is not a barrier. Plenty continues to be lost in translation, and everything that may seem simple or routine to many people required relearning for my parents, who had uprooted everything to pursue the American Dream while somehow finding a way to send support back home, too. A key story from our family lore tells of my father in the post office striking up a conversation with a friendly gentleman from Ohio. That chance encounter led to my parents having an important connection to their whole family that has persisted over the decades. They became close family friends, supporters, and people who were there for every question and every moment, including after I was born. They started making my parents feel like we could belong here even though we looked nothing alike and our family histories were dramatically different.

When I was just a toddler, my parents moved to Central Pennsylvania, a predominantly White area in Pennsylvania, so that my father could accept a job as a chemist at AMP Incorporated. Soon thereafter, my brother was born. My parents found themselves again figuring out the lay of the land primarily on their own, and as other Bengali families found their way to Central Pennsylvania, they would open up the phone book looking for last names that reminded them of home. They would cold call us, and this would allow them to find out where we got our groceries, where there was a temple nearby, and most importantly, where they might find other immigrant families like ours. Those families became our chosen family. I still keep in contact with many aunts and uncles from those years of my life because they helped us feel less isolated in a place where, as hard as we tried, we didn't fully feel like we belonged. They also provided an important type of support for my parents, who were so far from home, raising children without the help of their families.

When this community of Bengali transplants would get together, there was a shorthand to our gatherings. The parents would speak Bengali in

one part of the house, and the kids would speak English somewhere else. We would eat foods we could only get in our kitchens, and my generation would compare our experiences as “American-born confused Desis.” Together, we found a community where we understood each other without having to explain the context. We shared the pressures to succeed academically because our parents gave up so much to give us these opportunities. Plus, we were all Bengali, and one of our major Hindu holidays was in celebration of the Saraswati, the goddess of knowledge, art, music, learning, and wisdom. Studying was, in fact, a cultural element of our existence.

Navigating school wasn’t always easy. Even with the waves of immigrants, South Asians are still only 2 percent of the US population, certainly less in my school years and in that part of the country. Kids made sure to point out exactly what they thought about my dark hair and eyes. I received anonymous calls before the days of caller ID, when kids would call and tell me to go back to where I came from, that hateful phrase so many immigrants have heard. Unsurprisingly, these views are learned from people who perhaps are threatened by the arrival of immigrants, fearing that opportunities they view as rightly theirs are being taken away from them. In some cases, people aimed their Islamophobia at a wide range of Brown communities after the events of 9/11, merging all identities into a monolith of America-hating terrorists. Or perhaps the hate is more diffuse, less defined in its purpose. Whatever the origin, we see it playing out in horrific and violent ways, such as when a shooter opened fire in a Sikh house of worship in Oak Creek, Wisconsin, in 2012 (Dirks 2022). Tragically, this is one of many examples of hate crimes targeting South Asian communities, especially Sikh communities, who are very visible in society due to their traditional attire, including turbans.

The dangers growing from racism and the inclusion of these harmful views in the national discourse by political leaders is thought to have emboldened people to act on these views. The impacts of racism on numerous BIPOC communities has been a common experience, particularly during the pandemic, as we’ve seen the deaths of George Floyd, Ahmaud Arbery, Breonna Taylor, and far too many other Black victims of gun violence; anti-Asian violence in response to the so-called “China virus”; anti-Latin American policies designed to remove them from our

country; and the examples go on and on. The parallel pandemic of recent years has been the propagation of hate. It hasn't been confined only to racial and ethnic groups, either. We see harmful actions targeting people defined by their gender, sexual orientation, disability, pregnancy status, and numerous other identities as a near-constant barrage of hate. As a person who has experienced harm due to misunderstood ideas of difference, I can't help but feel solidarity with all these groups. We have all experienced traumas in our lives and in our histories.

Because my father came to the United States in part to pursue higher education in the sciences, another consequence of being raised as a first-generation American in my family is a belief in the transformative power of education, as well as a true love of science. It is probably not a coincidence that I've ended up as a professor of genetics after many years of study. Ironically, many people who have embraced ideas that double down on difference and bias think that doing their own research is comparable to studying a topic professionally. It is my goal to apply my scientific expertise to create accessible resources that help people understand the unifying messages of genetic science that are embedded in our DNA. Science can help inform our understanding of the biases that marginalize groups and the misunderstandings that contribute to societal divisions. By engaging with the science in a meaningful way, we can work together to debunk the flawed thinking that is used to divide rather than unite us.

A critical component to a thorough examination of the issues that contribute to bias involves not only science but also an interdisciplinary approach where we look beyond the genetic code and delve into a study of identity, sociology, history, ethics, and other relevant fields. By taking this approach, we will be able to identify the questions that need to be asked, not only by scholars, but also by all members of society. As such, we will be exploring some very tough questions in this book. There will not always be simple answers that I can provide, but I hope that by inviting you to join me in this process, we will be more able to identify the potential for harmful biases and that together we can think about ways to promote equity in our various spheres of influence.

We will begin this journey by carefully considering what science tells us, beginning with a discussion of the scientific and neurological bases of bias, as well as the flawed premise of genetic essentialism. The framework

of genetic essentialism begins with the erroneous belief that members of a social group, like race, are homogenous not only in physical, cognitive, and behavioral dimensions, but also that those traits arise from fundamental genetic differences that distinguish them from other groups (Donovan 2022). From there, we will dive into what we actually know about the genetic variation that is seen in populations and contrast biologically derived traits with socially defined identities. Throughout, it is my goal to model use of inclusive language (Cho et al. 2022), so that in describing human difference, we show respect for the full range of identities. The terms used in this book are recommended not only by people who are professionals working in these fields, but also by people who hold these identities personally. In writing this book, I endeavor to use language and terms that invite people into the conversation. Because language evolves, however, it may be the case that some of these terms will be replaced by others with more precision. In that event, I ask you to forgive me for harms that may unintentionally emerge over time, and I also ask you to help our communities improve by using the favored terminology of the time and of the community.

Thank you for trusting me and joining me on this journey. I truly think that by learning together and engaging in open and honest dialogue, we can create a better and more just society. I am grateful to be part of that conversation.

PART II What Does Science Tell Us?

2 On the Origins of Bias

For anyone aware of the impact of bias on the lives of people from marginalized identities, the existence of bias in our surroundings feels pervasive. Once you see it, it is hard to unsee, and unfortunately, cases of overt bias and even identity-motivated violence are becoming all too common as expressions of bias are normalized by people in powerful societal roles.

To fully be able to counteract bias, we need to be able to characterize its origins. Understanding the roots of bias involves unpacking how our brains engage in sensemaking about our environments. Some stimuli in our environments require rapid, instinctual responses, and these reactions fall under the umbrella of system 1 thinking, an automatic sort of thinking. Other situations require carefully deliberate responses, using system 2 thinking, where the mind is actively engaging in working with incoming information (Kahneman 2012). System 1 thinking is engaged in familiar situations, such as driving a car on a routine commute. However, if we take that same thought process and move it to an unfamiliar context, such as driving on a different side of the road in another country, now we are engaging system 2 thinking, where you can almost feel the gears turning in your mind in real time as you exert noticeable effort to maintain this unfamiliar line of thought. Our minds require cooperation

between systems 1 and 2 in order to make fast judgments, particularly in potentially dangerous situations. For example, if an unexpected obstacle appeared on the road, system 1 would alert system 2 that help is needed to consider the next best action. The problem with these two systems, however, is that because system 1 is based on models that predict outcomes, the models are full of generalizations and biases derived from lived experience. Because our minds are designed to minimize effort in routine encounters, those biases, left unchecked, can influence our actions in negative, unintended ways.

When it comes to biases, they contain elements that are explicit and implicit. Explicit biases are those that we are aware of, that if asked, we would knowingly, even proudly report as part of our personal beliefs. Implicit biases are unconscious, often rooted in system 1 thinking, and are potentially even the opposite of our stated beliefs. With respect to implicit biases that do not align with our beliefs, it is possible to separate intent from impact. One may intend to act equitably and inclusively but may impact others negatively by one's automatic actions that perpetuate biases. For instance, imagine a teacher who has chosen to work in an under-resourced school with the expressed intent to help children who have not had access to high quality educational opportunities. As part of their training, they study classroom management techniques that provide examples on how to deal with "unruly children," and the examples shared by the professor are all students of color. Those hypothetical examples are internalized by this new teacher, who unintentionally provides more frequent and severe disciplinary consequences for their Black students. This unconscious racialized reaction to student behavior has lasting harm, labeling students as troublemakers and making them question their sense of belonging in a learning environment. If the intent is to help these children, however, then an administrative audit of behavioral management practices can turn attention to the demonstrated disparities and can lead to more intentional, equitable approaches to student discipline. If, however, the bias is an explicit one, where the teacher truly believes that Black students are less able to function in a classroom setting, this type of audit will be unlikely to catalyze change.

Another problem with the seemingly easy solution of interrupting implicit biases by turning conscious attention to potentially biased

decision-making is that we are all susceptible to biases, and these biases permeate so many decisions in our lives. Even if we were raised to believe in equity across races, genders, sexual orientation, ability, and other identities, we are not immune to cultural messaging that can be found everywhere, from communications from schools to movies and commercials. For example, is it necessary to portray older folks in advertisements as foolish or weak? Or in terms of entertainment, how often have you noticed that the criminal element in a movie is Black or Middle Eastern? The fact is that the media we consume and the messaging that permeates our society elevates White, cisgender, heterosexual, able men as the ideal against whom all else are compared. With these messages programming our system 1 thinking, we have to exert effort to keep those internalized messages from influencing our decision-making. And although it may seem counterintuitive, this applies to everyone, even people whose identities are featured in the biased messages of our culture, because all members of society internalize these harmful messages.

While it is true that we must take deliberate action against the seemingly constant onslaught of biased messaging, we are not powerless in this endeavor. To begin, we must intentionally consider our values and how our brains may be working against those values. A good place to begin is with measuring the existence of unconscious bias. One widely used tool to do so is the implicit association test (IAT). This test measures reaction time in a sorting task where images or words are selected and grouped in ways that are either congruent or incongruent with stereotypes. The underlying premise is that if your mind links two things, it will be able to sort them more quickly when they are colocated on the screen. For example, in an IAT designed to measure automatic preference by race, a person may be asked to sort images of people and good or bad words. The words and images are oriented such that they are congruent to harmful stereotypes in one test and incongruent in another test. Response times for the sorting task in congruent arrangements should reflect primarily system 1 engagement, an instinctual response informed by information absorbed by the participant. Response times for the sorting task in incongruent arrangements, however, can reflect the engagement of system 2 when the automatic, stereotypical response is not accurate. If response times are faster for the given congruent test than the incongruent test, that would

indicate an automatic preference for White people in the example race IAT. There are many versions of this test available to the public on the Project Implicit website (“Project Implicit” 2011) designed to help people assess their views about specific racial groups, skin tone, gender, sexuality, disability, religion, weight, age, and other attributes.

One of the first IATs I took was when I was participating in a training for Bias Reduction in Medicine (BRIM) (Carnes and Sheridan 2019). At this point, I had already been a professor for 15 years, not to mention my identity was and still is highly tied to being a woman in STEM. I experienced many of the biases encountered by women in academia firsthand, so it wasn’t a distant, theoretical concept to me. I had mentored hundreds of women who were training in the sciences. And I had actually volunteered to be part of the BRIM initiative at my university; this wasn’t something I was doing reluctantly. And yet, when I took the gender-science IAT (“Project Implicit” 2011), it said I had an automatic preference for men in science. I was more than a little mortified. I had quickly read the disclaimer at the start of the IAT—“If you are unprepared to encounter interpretations that you might find objectionable, please do not proceed further”—but I certainly didn’t expect it to apply to me. I’ve since turned this over in my mind many times. What could possibly explain this result, because it certainly doesn’t align with my core belief that women and gender-expansive individuals belong in science (and everywhere men are)? The best explanation that I can conclude from this experience is that we are all fallible and subject to biases internalized from the media. After all, a report from the Geena Davis Institute on Gender in Media and If/Then Lyda Hill Philanthropies (2024) determined that there is almost a 2:1 ratio of men to women portraying scientists in film, television, and streaming content and that those numbers have held steadily over a fifteen-year period. As much as I would hope to be born inherently unbiased, I am not uniquely immune to those external messages. Just as Ibram X. Kendi writes about anti-racism, “Racist ideas have defined our society since its beginning and can feel so natural and obvious as to be banal. . . . To be an antiracist is a radical choice in the face of this history, requiring radical reorientation of our consciousness” (Kendi 2019), the same is true of gender equity and other forms of bias as well. So, I had to set out to radically reorient my conscious thoughts about women in science to overcome

social programming and implicit biases. While that already came naturally to me in mentoring relationships, I had plenty to learn about the evidence basis demonstrating the value of diversity in professional spaces, as well as ways to improve equity in job searches, in supporting women's career advancement in academic medicine, and in faculty retention with an eye toward equity. I was grateful for the opportunity to learn from the BRIM program and continue to practice the equitable protections I learned in that program.

I recently retook the gender-science IAT while reflecting on these experiences, and I found that my result now is returned as neutral—no preference for any gender in science. The power in this approach of facing one's bias is perhaps less about the precision of the implicit association test and more about the process of being open to the possibility of bias in one's own actions. In fact, some studies have shown that implicit bias as measured by the IAT can decline after learning that contextualizes and explores biases and stereotypes (Rudman, Ashmore, and Gary 2001), perhaps indicating that formal study of this topic in the BRIM training allowed me to deliberately revise my unconscious framework. Working to understand implicit bias helps people understand the role of systemic cues absorbed by our minds and removes the issue of blame or a judgment on character from the discussion. With this knowledge and experience, it is easier to then adopt anti-oppressive safeguards to guide one's actions toward equitable outcomes. To be fair, prioritizing equity requires constant vigilance. Engagement in deliberate, system 2 thinking can be woven into processes like job searches, but it is much harder to bring this type of methodical process to everyday interactions. Moreover, people have gaps and oversights that they don't even realize are biased positions that can work against their best intentions.

It would be viewing the world through rose-colored glasses if I were to end this chapter without properly acknowledging the role of explicit biases in perpetuating societal inequities. Not all biases in the world result from the oversights of otherwise well-intentioned people. Some are personal views designed to reinforce the hierarchies that are the direct reason for structural inequities. To be sure, racism, sexism, homophobia, transphobia, disability bias, and many other forms of discrimination exist explicitly in society, and in these cases, the hierarchies of power are not a bug of the

system but a feature for those who create them. Shining a light on implicit biases focuses on an individual's role in inadvertently perpetuating bias, but we must also take on the formidable task of removing systemic barriers to equity (Pritlove et al. 2019). The focus can be less on academic categorizations of bias subtypes and more on how biases play out in society, and importantly, effective ways to interrupt those ripple effects. To fully realize equitable policy design and to benefit from contributions of all members of society, we must address not only individual perspectives, but also structural issues. For instance, if people hold the belief that caregivers to children and the elderly are not dedicated to their professions, then the workplace will not allow the flexibility individuals with caregiving responsibilities need to balance their duties. Similarly, if people from a marginalized race are viewed as less capable, then society will not make the effort to level the playing field and reduce the impact of structural barriers for them. For these reasons, it is important to debunk these wrongly held beliefs at the level of first principles. With respect to biological traits, some of the basis for disparate outcomes may be due to innate biological differences, whereas for social traits, we must look externally at the systems that disadvantage marginalized groups in seeking explanations for the observed inequities. A greater understanding of the root causes of these differences and their intersections will help us design more effective interventions in doing this work of moving toward justice.

In this first part of *Where Biology Ends and Bias Begins*, I aim to describe the current state of our knowledge of these topics. We will begin with a discussion of the concept of genetic essentialism and what it misses in trying to describe the genetic basis of the human condition. From there, we will do deeper dives on what science can help us understand when considering race versus ancestry, sex and gender, the biology of pregnancy, and disability. Note that we will be contrasting biological and social identities in each of these areas. To fully understand the nuances of human difference, we must be willing to use a variety of lenses, and as such, I aim to bring not only the biological and health sciences to bear, but also lessons from colleagues in sociology, anthropology, psychology, ethics, law, economics, and other related fields. This multidisciplinary approach will help us traverse from the edge of biological knowledge to where social biases and inequities take over. It will also help us have an honest conver-

sation about the limits of genetic knowledge so that we can ask hard questions and think more accurately about the ways in which we can work together to dismantle the systems that disadvantage members of our communities. These problems are bigger than us as individuals, and I hope in these pages you will begin to imagine the ways in which your roles in these complex systems can be leveraged to work toward justice. Let us continue toward this goal by building upon our bias framework to incorporate understanding of genetic essentialist ideologies that can contribute to biased actions.

3 The Flawed Premise of Genetic Essentialism

People are constantly using the phrase “it’s in their DNA” to describe a core part of someone’s personality, likes and dislikes, or identity (Birney 2022). It’s such a common phrase that it even appears in the dictionary with the definition “to be an important part of who someone is” (Cambridge University Press 2024). People use this idiom to describe everything from preferences, like which sports team one supports, to talents, like one’s fashion sense. Though the phrase is often used in harmless metaphorical contexts like these, it can also be used in ways that reflect harmful genetic essentialist thinking. Genetic essentialism is a reductive view that equates one’s genetic code with one’s individual traits, nature, or identity, as if one literally had a gene that made them a fan of the Barcelona Futbol Club. In casual conversation, it is unlikely that people using this phrase actually think there is a string of nucleotides in a person’s DNA that determines their athletic allegiances. However, with the rapid growth of personal genomics companies offering DNA genotyping to give folks a greater understanding of their ancestry, the scientific differences between race, ethnicity, and ancestry are getting muddled in popular discourse. Advertising campaigns for some of these products encourage people to go on identity-seeking journeys through DNA analysis, as if identity is hard-

wired in our DNA. In a twisted take on these ideas, White nationalists have started engaging in so-called “citizen science” to demonstrate racial purity using these test results (Zhang 2016, 2017; Boodman 2017; Panofsky and Donovan 2019). They then ritualize their results through a brotherhood of milk chugging to prove they have the trait of lactose tolerance (Harmon 2018), conveniently ignoring the fact that this trait is prevalent not only in European ancestries, but also East African populations. Notably, these tests can only measure similarity between variation in a test-taker’s genome and people who, at some point in history, lived in a given place on the planet (Foley and Wolfe 2019). I can actually demonstrate this point using my own genetic ancestry results from the All of Us research project. Based on comparison of my genomic results with those of people from around the world, genetic variation in my genome is similar to variants identified in people hailing from the South Asian nations of Bangladesh, India, and Pakistan. However, these are geopolitical borders that only surfaced during my parents’ generation, in 1947, with the partition of India, Pakistan, and East Pakistan (eventually Bangladesh), as India became independent of the British Commonwealth. These nations were hastily created to separate people by religion and caused a bloody migration of Hindus and Muslims across the newly defined borders. Seeing my genetic ancestry did not tell me anything new about my family roots even though, at first glance, they appear to show fragmented identity. When I go home to see my extended family, I go to India, but I know that our roots extend farther if we go back just a few years. My race and ethnicity are informed more by our shared language, our holidays and celebrations, our favorite foods, and our cultural observances than by my DNA.

One of the big concerns about the growing focus on using DNA as a tool to understand aspects of identity is that this appears to validate genetic essentialist beliefs and deterministic thinking that suggests our genetic makeup is immutable and fully responsible for aspects of a person’s character, behavior, and innate abilities. These deterministic views also assume that members of identity groups are uniform in these characteristics but different from other identity groups, setting up a hierarchy of harmful stereotypes between groups based on race, gender, ability, and many other characteristics. Not only does deterministic thinking reinforce stereotypes about uniformity within groups and about between-group

differences, but it also concludes that these outcomes are fixed and independent of influences outside of innate biological ones. In this case, it negates any potential benefit of structural interventions and considers these outcomes as the unavoidable moral result of inherent differences, an error known as the naturalistic fallacy (Dar-Nimrod and Heine 2011).

Camara Jones, a scholar of health equity, has an illustrative gardening allegory (Jones 2000) that helps debunk this error in thinking, this notion that outcomes are baked into our genomes from the start. In Dr. Jones's "Levels of Racism: A Theoretic Framework and a Gardener's Tale," she uses the analogy of a story of seeds for different-colored flowers with otherwise identical genetic potential. The seeds are planted in two different pots, one with top quality potting soil and fertilizer, and the other containing old soil, poor and rocky. As you might imagine, the seeds potted in the fertile, nutrient-rich potting soil sprouted beautifully, showing great vitality and beautiful red blooms. In contrast, the seeds potted in the old soil had far fewer plants successfully grow, with only the heartiest among them growing and yielding much less healthy pink flowers overall. By assuming that the different growth outcomes of the red and pink flowers are related to their innate genetic potential, the influence of external factors like environmental conditions is completely discounted. Now imagine that the gardener has a preference for one type of flower over another and deliberately chooses to put their favorite red flowers in the potting soil, nurturing these seeds with care, water, and sunlight. They simultaneously plant their less favored pink flowers in the other pot, paying little mind to the care of those seeds. Predictably, the red flowers thrive while the pink flowers struggle, and what is worse is that when these flowers go to seed, the red flowers will continue to dominate generation after generation. In this analogy, the gardener symbolizes institutional and structural biases. Frighteningly, this is a self-reinforcing cycle because the institution "learns" that their biases were right, that red is better than pink, and overlooks the reality that the differences result from inequities in the resources and care of the seeds. A genetic essentialist would continue devoting resources to the "superior" red seeds, whereas someone wise to the existence of bias would investigate the root causes of the observed inequities, ultimately realizing that a structural shift is needed in resource allocation.

In this allegory, it is straightforward to identify the factors beyond genetic potential that influence the growth outcomes of the red and pink flowers, but most human systems are far more complex. When it comes to

health outcomes, we must consider not only biological protections and susceptibilities, but also environmental exposure, housing insecurity, food insecurity, personal safety, wealth, language, health literacy, mobility, incarceration history, physical access to care, employment status, insurance status, sick leave policies, systemic biases writ large, and on and on. If in the study of health disparities people take the genetic essentialist shortcut, then the influence of all these factors that can dramatically impact health are not given appropriate attention, thus missing an opportunity to address systemic factors in the promotion of health, in other words, the vast majority of the entire field of public health!

To be fair, there are examples in which genes are the primary variables influencing biological outcomes. For instance, a textbook example of a scenario in which there is a very strong genotype-phenotype correlation is with Huntington's disease. This neurodegenerative condition results when the huntingtin gene's CAG trinucleotide repeat (a series of three repeating bases in the gene's DNA sequence) extends beyond a particular threshold. It is possible to carry out a genetic test where the precise number of CAG repeats is determined, and this can be used with strong confidence to predict whether a person will go on to develop Huntington's disease, losing control of intentional muscular movements over time. However, this example is more the exception than the rule. Conditions in which a single gene has an outsized impact on the development of a particular trait are very rare (about 2%) among diseases with a genetic influence. It is far more common that genes work in combination with each other and with environmental factors to impact biological outcomes, and that a genetic predisposition on its own is not definitively predictive of a particular outcome. For example, 71 percent of individuals with the APOE e4 allele associated with Alzheimer's disease never go on to develop the condition. Genetic essentialist thinking ignores the fundamental complexity of how genes work in the context of multifactorial systems.

In the absence of a nuanced understanding about the role of genes in biological outcomes, estimates of the influence of genetic factors are inflated to the extent that people erroneously discount the role of nongenetic factors. This perception can lead people to believe outcomes are inevitable despite interventions that can promote health, such as modification of diet or exercise habits. The flawed framework of genetic essentialism has also been linked to discriminatory practices and opposition to equity-promoting

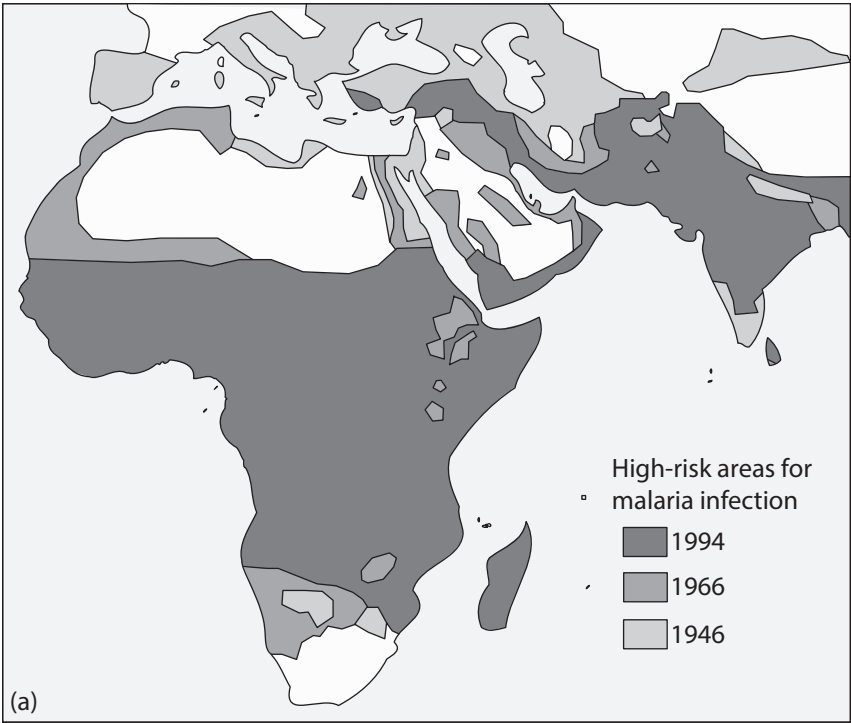


Figure 1. Comparison of regions where malaria is endemic (a) with observed sickle cell anemia allele (HbS) frequency (b).

policies because of the perceived inevitability of “natural” disparities. Encouragingly, by some measures, increasing people’s genetic knowledge has been shown to reduce genetic essentialist biases and racist views (Harmon 2019a; Dar-Nimrod et al. 2021; Donovan 2022; Donovan, Weindling et al. 2024).

Education is a powerful tool, and it is important to note that essentialist thinking can be either reified or debunked through the power of education. When genetics classes focus on differences in incidence of genetic conditions in racially defined groups, they unintentionally increase belief in racial genetic essentialism. For example, many people learn that sickle cell disease has a higher incidence in Black populations. The truth is a bit more complex than this broadly defined racial category. In the case of sickle cell disease, the oxygen-carrying hemoglobin in red blood cells

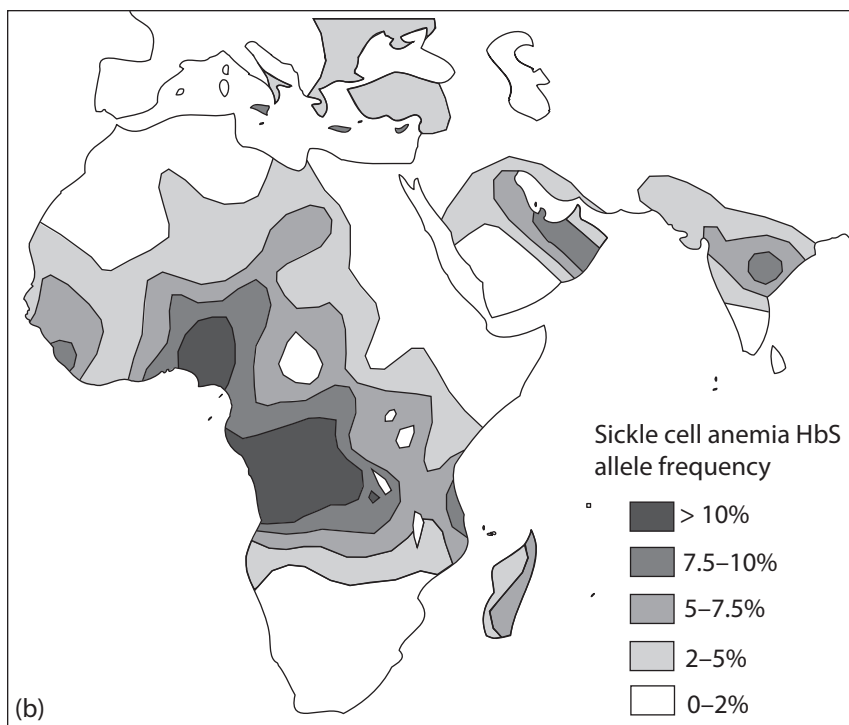


Figure 1. (continued)

deforms under conditions of low oxygen, leading to hemolysis, organ damage, and acute pain. One critical form of treatment for sickle cell involves pain relief during acute crises. Frustratingly, because some providers view this as a Black disease and also hold biases about Black folks' habits around the use of pain medication, people with sickle cell find themselves dressing up to try to present themselves professionally when simply trying to access care or avoid going to the hospital at all (Power-Hays and McGann 2020). The necessity of these protective strategies in the face of structural racism clearly reflects the origins of poor outcomes for Black patients with sickle cell disease. Moreover, it is important to recognize that sickle cell disease is not limited to Black populations. Rather, this devastating condition provides some protection to people who have one copy of the disease-causing allele (HbS) in areas where malaria is endemic (figure 1). Thus, due to

evolutionary selective pressure, pathogenic alleles are observed at elevated frequencies in a number of global populations hailing from regions in Africa, as well as Middle Eastern and South Asian peoples. In terms of diagnosis, the United States includes newborn screening for sickle cell disease in all 50 states, so for the vast majority of children born in the United States, regardless of race, they will be screened for this condition. However, their subsequent treatment and disease management is subject to many aspects of systemic racism.

When genetics is taught in this way, associating certain conditions with particular racial or ethnic groups, concepts of genetic essentialism are unintentionally reinforced, and students may jump to the conclusion that each race has its own genetic diseases. The natural extension of that concept may lead people to believe that these social groupings also have their own unique potential for intelligence, science ability, academic talent, athletic performance, and numerous other traits. Inevitably, concepts of racial superiority emerge from this type of thinking and contribute to dangerous societal divisions. This information needs to be contextualized to help people gain a more nuanced and accurate understanding of human populations, to help them understand that socially constructed groups are actually more biologically alike than different, and to help them appreciate that there are many cases in which, as the aphorism goes, one's zip code, reflecting social determinants of health, can have more to do with one's overall well-being than one's genetic code (Schroeder 2007).

In leveraging education to refute genetic essentialist beliefs about race, curricula that promote *humane* genetics literacy by examining subtleties of population genetics demonstrate an ability to reduce essentialist thinking about race by changing how students perceive inter- and intraracial variation (Donovan et al. 2019, 2021; Donovan, Weindling et al. 2024). This evidence basis underscores the importance of careful examination of these topics in both science and social science classrooms. In the absence of this type of educational intervention, people are going to engage in sensemaking of observations around them, which can lead to harmful, genetic essentialist conclusions in the absence of appropriate examination of the foundational science. For educators, this emphasizes the importance of providing guidance and context about these topics in a wide range of disciplines, not just the social sciences, because in its absence the virus

of bias will spread unchecked. The stakes are high when it comes to bias, and education is a powerful tool to correct the record by applying anti-essentialist and, therefore, anti-oppressive ideals. Now that we have a framework describing the neurological origins of bias and the flaws of genetic essentialism, we can incorporate a careful review of population, molecular, and clinical genetics into this context to understand what biology has to say about human difference, that is, where biology ends and bias begins. In the next few chapters, we will use this approach to explore the science behind ancestry, sex, reproduction, and disability as well as the social overlays of race, ethnicity, gender, bodily autonomy, and individual agency with the goal of disentangling the complex relationships between biology and bias.

4 Biology of Human Populations

RACE, ETHNICITY, AND ANCESTRY

The notion that race is a social construct is a frequently repeated refrain, but it is often stated almost as a sound bite without a detailed understanding of what background knowledge it is invoking. In this chapter, we are going to dive into the true meaning of the social construction of race, what variables it uses to group people, and how biological data gets inappropriately tied into this information. By exploring what science can actually tell us about human populations, we can begin to disentangle genetic ancestry from social groups like race and ethnicity.

We must reckon with the reality that races were originally conceived of as ways to group people and maintain systems of power and hierarchy (Smart and Saini 2022) in order to fully contextualize why it is important to consider the scientific understanding of human populations. Often these groupings coincided with continental boundaries, and by studying human differences across these arbitrary political borders, scientists began to systematize and legitimize these population definitions even though human migrational history describes a population of ancestors that went on to populate virtually all habitable regions of the globe (figure 2) (Henn, Cavalli-Sforza, and Feldman 2012). With each migrational event, a subset of the genetic variation from the ancestral population established a new

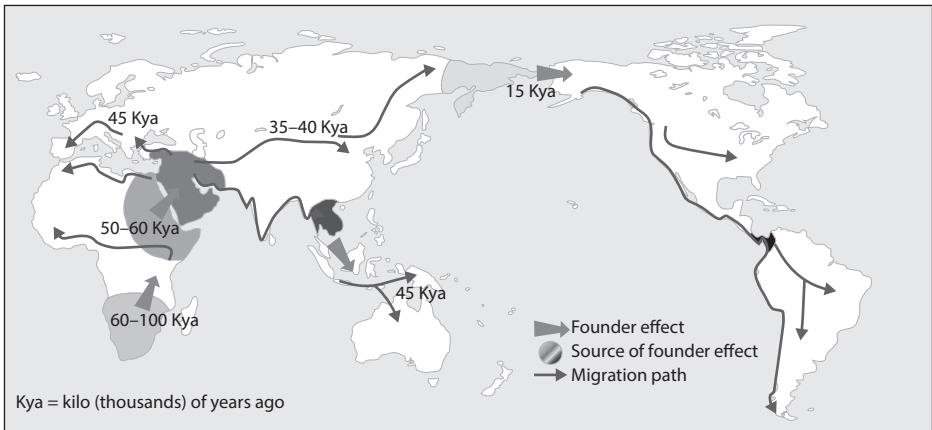


Figure 2. Patterns of human migration over the past 100,000 years. (Reproduced from Henn, Cavalli-Sforza, and Feldman 2012)

population, exhibiting a modest decrease in genetic diversity from the parent population (figure 3). As a result, human genetic variation is continuously, not discretely, distributed across the globe (Fujimura et al. 2014).

Despite the continuous nature of human genetic variation, there have been numerous scientists focused on sorting humans into discrete groups. Carl Linnaeus, famous for categorizing life on Earth, came up with categories for humans: Americas (red), Europe (white), Asia (yellow), and Africa (black) (Saini 2019). Similarly, Samuel Morton classified humans into five races that he termed Caucasians, Mongolians, Southeast Asians, Native Americans, and, lastly, Blacks or Ethiopians (Kolbert 2018). He believed people from these populations resulted from five different creation events and that the ranking stated above reflected a divine hierarchy. Using a collection of skulls from these populations, Morton carried out craniometry, measuring the skulls and concluding that Caucasians were “scientifically” the most intelligent of the races. These harmful racist ideas are embedded in the origins of our understanding of race, and, in fact, in the term *Caucasian* itself (Popejoy 2021), which is why I encourage you to refer to these populations as European ancestries or White populations.

Scientists historically have viewed genetics as a safe way of studying human difference, but unfortunately, those differences continue to be

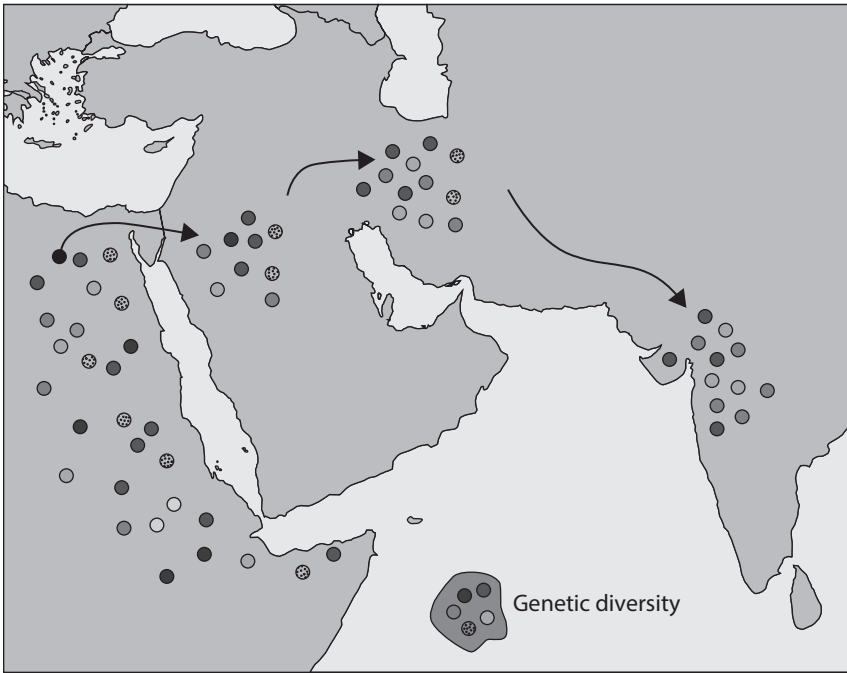


Figure 3. Genetic diversity as a function of migration and founder effect.
(Reproduced from Henn, Cavalli-Sforza, and Feldman 2012)

viewed through a hierarchical lens where populations of color are viewed through a deficit lens relative to others. White populations are framed as “normal,” intentionally ascribing a higher relative value to characteristics aligned with White populations, and they are the standard against which all else is measured, even scientifically in clinical trials. This framework was used to justify everything from slavery, apartheid, and colonialism to genocide and the Holocaust. In spite of these hard-learned lessons of history, we continue to fall into these habits of mind now. Using continentally defined ancestries in genetic studies (Cerdeña, Grubbs, and Non 2022; Lewis et al. 2022) has the potential to reify these harmful categories, essentially referring to scientific racism just by another name.

The work of Brian Donovan and colleagues establishes that lack of intentionality and precision in communication of genetic concepts can lend scientific credence to essentialist thinking (Donovan 2014). His

follow-up work has demonstrated the importance of humane genetic literacy, an approach to genetics instruction that helps learners understand the nuances of human difference (Donovan et al. 2021; Donovan 2022; Donovan, Weindling et al. 2024) and that emphasizes the fact that within-group variation is far larger than between-group variation. This concept is hard to grasp because in sensemaking about the world around us, our minds tell us that human difference is real by noting physical differences between people of different races. However, as Mark Thomas of University College London has said, “Variations in physical appearance, skin pigmentation, or other traits, make the gaps between groups feel far larger than they really are, genetically. Biologically, the differences really are just skin deep. It’s an error to assume that the internal differences are as profound as the external ones appear” (Saini 2019).



So how can we break free of these oversimplifications our minds are trying to make about the world around us? We can take a careful look at the emerging genetic information that describes human variation through this lens of humane genetic literacy with a focus on data from a collection of genetic studies (Rosenberg 2011) that has been shown to reduce racist views among learners (Harmon 2019a). Using this collection of data, more recent studies with updated genetic data (Bergström et al. 2020; Biddanda, Rice, and Novembre 2020), and Brian Donovan’s evidence-based method to reduce genetic essentialist beliefs, I will share a systematic approach to examining human genetic variation across populations.

First, we must begin with an understanding of what is meant by human genetic variation. Genes are housed in the DNA molecules we find in the nucleus of our body’s cells. The DNA is composed of a sugar-phosphate backbone and bases (adenine, thymine, cytosine, and guanine) that have the ability to pair across the DNA double helix. In humans, DNA is organized into a collection of 23 pairs of linear molecules called chromosomes. Each chromosome has between approximately 200 and 3,000 individual genes, and the genes average between 10,000 and 15,000 base pairs per gene. The entire collection of chromosomes is referred to as the genome, a blueprint that contains all the instructions needed to build our

bodies. Scientists have the chemical ability to read DNA molecules to determine the precise sequence of bases (A, T, C, and G, for short), and this technology was used in the Human Genome Project to essentially read the book of life. The sequence information initially released publicly in 2003 is composed of the anonymous genomic information from just a handful of donors in Buffalo, New York (*Human Genome Project Fact Sheet* 2022). This project was a 13-year, \$2.7 billion international effort. Now Guinness World Records has given the title of fastest complete DNA sequencing for the entire human genome to Professor Euan Ashley and colleagues, who completed a genome sequence in five hours and two minutes (“A New Frontier’: Fastest Ever DNA Sequencing Technique Achieved” 2022). With the reduction in time required to sequence a genome, there was a dramatic parallel reduction in cost. A single human genome initially cost \$300 million to sequence around the year 2000, compared to now, at the time I am writing this, when it is possible to sequence a genome for a few hundred dollars (“The Cost of Sequencing a Human Genome” 2021).

This rapid improvement in cost and sequencing technology put the possibility of sequencing many human genomes from around the world within reach. Through a range of projects, from the International HapMap project to All of Us, a more diverse group of people have volunteered to contribute their genomes to this global initiative. The results of these efforts are a treasure trove of human genetic information that allowed us to make comparisons across genomes. Positions at which the bases differ from person to person were identified using a computational approach similar to the compare-documents function in word processing programs. The first very striking example was just how similar human genomes are; comparisons have shown that humans are 99.9 percent identical at the genetic level. It never ceases to amaze me when I stop and think about that figure. Imagine the vast diversity in human biology and consider that 0.1 percent of our genomes is responsible for the differences that are encoded in our DNA!



Now that we understand what is fundamentally meant by human genetic variation, let's apply a data-driven approach to understanding human

genetic variation within and between populations. A 2011 study (Rosenberg 2011) used a particular type of genetic variant known as microsatellites (short repetitive DNA elements), and two 2020 studies used single nucleotide variants, single base pair differences, in these cases categorized by the 1000 Genomes Project (Biddanda, Rice, and Novembre 2020) and an extension of the Human Genome Diversity Project (Bergström et al. 2020). These data were also adapted and further analyzed for visualizing patterns of human genetic variation across populations (Kitchens and Coop 2023). Different versions of these sites of variation are known as alleles. Using information gathered about variant alleles across populations, I will use the core framework outlined in the initial paper by Rosenberg (2011), where he seeks to answer the following questions:

1. Are most alleles widely distributed, or are they largely confined to specific parts of the world?
2. Do there exist distinctive alleles for specific geographic regions that distinguish individuals in one group from those in other groups?
3. Of the genetic variants that exist in the human genome, how many are present within a given geographic region?
4. On average, how different are two individuals from the same local population, in comparison with two individuals chosen from any two populations anywhere in the world?

Let's now begin a deeper dive to answer these questions, which will help us develop a more nuanced understanding of genetic variation within and between human populations.

1. Are most alleles widely distributed, or are they largely confined to specific parts of the world?

Among the microsatellite variants examined at 783 loci (locations) in the genome for their presence in seven global regions (Africa, Europe, Middle East, Central/South Asia, East Asia, Oceania, America), 46.6 percent of all variants examined were present in all seven regions. The majority of the remaining variants were present across multiple regions as well, with over 65 percent of variants present in at least five global regions and over 92

percent present in two or more regions. If the variants were predominantly limited to individual ancestries, then we would expect the number of variants present across multiple regions to hover around 0 percent, and if the variants were equally spread across the globe, the percentage of variants across multiple regions should be approximately 100 percent. Thus, neither of these simplistic models of human genetic variation fully captures the reality of variant distribution across populations.

Examination of single nucleotide variants of the 1000 Genomes Project demonstrates that variants tend to fall into two major categories: common variants present across multiple populations and rare variants found in single populations. Nearly two-thirds (63%) of variants common in at least one region were found to be globally widespread across the five measured regions, rising to 82 percent of variants when we consider variants common in at least one additional region outside of Africa, consistent with out-of-Africa human migration histories. Rare variants found in single populations are more likely to be the result of recent mutation and thus would have arisen after the human migrations that populated other global regions. If between-group variation typically distinguished one ancestry from the next, there would be a different predominant category: common variants restricted to a single geographic region. However, this classification does not emerge from this analysis.

Both of these types of analyses of common variants support the conclusion that genetic variation is typically found widely distributed around the world, not isolated to single populations.

2. Do there exist distinctive alleles for specific geographic regions that distinguish individuals in one group from those in other groups?

As noted in the analyses above, there are a small number of alleles private to individually defined continental regions. In the microsatellite analysis, the percentage of alleles that are private alleles were 7.5 percent of the overall number of assessed alleles, and the mean frequency of those private alleles was 1.7 percent. That is to say, the private alleles are a small fraction of the overall observed human genetic variation, and in the populations where they are found, they are a rare, infrequent variant. If there

were greater between-group variation than within-group variation, we would expect both of these numbers to be much higher, approaching 100 percent, where private alleles would describe the majority of the observed variation and nearly all the members of a given population would harbor that variant. Conversely, if within-group variation explained all of human variation, then we would not expect to observe private alleles at all.

In the single nucleotide variant measures, there are a larger number of variants overall compared to the microsatellites. Of the over 44 million examined variants present in at least two individuals, the variants are classified as common (present in >5% of the population) or rare (present in <5% of the population). The vast majority of alleles that are present in a single continental ancestry are the rare variants. Furthermore, none of these are among the top 10 variant distribution patterns globally, corresponding to 34 percent of the 44 million characterized variants, but <1.7 percent of the overall alleles. Rare variants are more likely to be the result of recent mutation within a population or even a single family, which helps explain why private alleles tend to be rare. In Africa, there are a small number of alleles private to the continent that are present at somewhat higher frequencies, and these correspond to ancestral alleles that were lost outside of Africa once humans migrated out of the continent. If between-group variation explained human differences across populations, the single nucleotide variant data isolated to individual populations would reach much higher frequencies, approaching 100 percent, for both the fraction of variants unique to populations and their frequencies in those populations. If all variation was within-group variation, then there would be no such thing as a private allele.

Taken together, we see that there are not unique alleles present in all members of a particular population that are observed within that single group. The concept of widespread private alleles that define a particular group is not supported by human genetic data.

3. Of the genetic variants that exist in the human genome, how many are present within a given geographic region?

Evaluation of the presence of microsatellite alleles in each of the geographic regions finds a range of 57–87 percent of variant alleles present in

each region, with the highest proportion of variation found in Africa and the lowest in Oceania (or adjusting for sample size, the lowest variation would be found in the Americas). This pattern is consistent with the out-of-Africa migration history and subsequent genetic drift. The average variation found in any given region is 75 percent of the microsatellite alleles. Thus, again, the majority of variants are found within groups rather than between groups. If between-group variation was the predominant type of variation, we would be observing a smaller nonoverlapping subset unique to each region. In contrast, if within-group variation captured all variants, we would expect 100 percent of variant alleles to be present across all regions.

By examining the classes of single nucleotide variants identified in the 1000 Genomes Project, we can estimate that 32–64 percent of the human genomic variant alleles are present in any given continental region. Though this may at first glance look to be different from the microsatellite data reported above, it is important to recognize that the 1000 Genomes Project, because it was cataloging single nucleotide variants, was inherently better at identifying low frequency acquired mutations, which would ultimately define the previously described rare private alleles. As such, the apparent fraction of human genomic variants present in a given population may appear lower, but again, the common variants are generally found broadly. Repeating this analysis with a focus on single nucleotide variants common in at least one region, we observe 70–99 percent of variants present in each continental region. If between-group variation explained the majority of human genetic variation, we would expect a small percentage of variants to be represented in any given region, whereas if within-group variation reigned, then we would expect the percentage of variants present within a region to approach 100 percent.

Considering both microsatellites and single nucleotide variants, the presence of within-group variation is large across all examined groups and is far more frequent than between-group variants. Further, the distribution of within-group variation demonstrates a linear decline when beginning with an origin point in Africa and moving outward from there, indicating further support for an out-of-Africa human migration pattern.

4. On average, how different are two individuals from the same local population, in comparison with two individuals chosen from any two populations anywhere in the world?

In some ways, this question frames the essence of what people misunderstand—how alike are we actually, if we hail from different regions of the globe? Using microsatellite data to address this question, selecting two random people from different ancestries predicts a summation of both within- and between-group variation. If two people are instead randomly chosen from the same ancestry, that would represent only within-group variation. Using this methodology, the level of difference between two individuals from the same ancestry turns out to be almost as great as two individuals from different ancestries, with 92.7 percent attributable to within-group variation and only 7.3 percent between-group variation. If the majority of variation was actually between groups, we would expect those percentages to be reversed.

A similar pairwise analysis was done with single nucleotide variants. In this comparison, a range of 62–76 percent of globally common variants differed between each pair of individuals from different groups on different continents, relative to 54–76 percent of variants differing between pairs of individuals from the same populations. The examined variants were what we would call globally common, widespread variants, and of note, we see a great deal of overlap in the range of differences between individuals within and between groups. Importantly, in this representative sample, we do not observe an increase in the extent of variation when categorizing difference in pairs of individuals between populations as compared to pairs from within groups. Furthermore, because of the larger collection of variants among the African populations, scientists observed greater pairwise variation (54%) within this population than within or between others. That is to say, simply choosing to compare individuals who come from different ancestries does not automatically imply greater genetic difference than two individuals from the same ancestry. If between-group variation was responsible for the majority of human genetic variation, this would not be the case; in that case, comparing pairs of people from within the same ancestry would always generate more similarity than two people selected across populations. Likewise, if



Figure 4. A visual representation of the distribution of human genetic variation across populations. (Adapted from Donovan et al. 2019)

within-group variation was the only form of human genetic variation, then we would observe equivalent similarity whether two people were selected from within or across populations.

Examining these trends helps us see the wide distribution of most variants, particularly the common ones. The subset of genetic variants that are population-specific private alleles are rare with respect to both the fraction of variants they represent and their frequency in the population. When examining any particular region, we will be able to identify the majority of human genetic variation within a single group, with the largest collection of variants present in the African continental population, declining modestly with distance from Africa. Finally, when comparing two people’s genomes, we see that they are only ever so slightly more different when they hail from different populations than when they hail from the same original population. A summary of these findings by the framing questions is provided in table 1 and is visualized in the Euler diagram (figure 4).

I hope by my presenting this evidence you now feel like you have a stronger grasp of what the science is telling us and can confidently debunk common misconceptions. If you’d like to work further with the data to

Table 1 Summary of answers to framing questions

<i>Framing question</i>	<i>Observations and evidence</i>	<i>Interpretation</i>
1. Are most alleles widely distributed, or are they largely confined to specific parts of the world?	Over 92% of microsatellites are found in multiple geographical regions, and 82% of single nucleotide variants are common in at least one region outside of Africa.	<i>If variants were confined to specific parts of the world, the existence of variants in multiple regions would be vanishingly rare. The data support the conclusion that genetic variation is commonly distributed across multiple regions of the world.</i>
2. Do there exist distinctive alleles for specific geographic regions that distinguish individuals in one group from those of other groups?	Private alleles unique to a single geographic area are exceedingly rare in the human genome (7.5%) and even more rare at the allele frequency level within any single group (1.7%). The only variants that are found in a single continental ancestry are those that are categorized as rare variants by their small population frequency.	<i>Private alleles are only a small percentage of the overall alleles in the human genome. There are not any unique alleles present in all or even most members of any individual population, so these rare private alleles could not be used to distinguish between groups.</i>
3. Of the genetic variants that exist in the human genome, how many are present within a given geographic region?	Depending on which type of variant is being examined, 57%–87% of microsatellites or 32%–64% of single nucleotide variants can be found in any given continental region. When the focus is on common variants present at allele frequencies above 5%, the fraction of variants observed in any given geographic region goes up to 70%–99%, with the highest proportion found in Africa.	<i>Most genetic variation is within groups rather than between groups, and as populations migrated out of Africa, a significant subset of the original genetic variation migrated with the relocating populations. Overall, a large fraction of the globally common variants continue to be present in geographic regions.</i>

(continued)

Table 1 (continued)

<i>Framing question</i>	<i>Observations and evidence</i>	<i>Interpretation</i>
4. On average, how different are two individuals from the same local population, in comparison with two individuals chosen from any two populations anywhere in the world?	When comparing two people from different ancestries, in the case of microsatellite markers, only 7.3% of the differences are representative of between-group variants, and for single nucleotide variants, the between-group pairs have overlapping ranges of variation when compared to within-group pairs. In both cases, the majority of the differences found between individuals can be ascribed to globally common, within-group variants that could just as easily differ when two individuals from within the same population are randomly selected.	<i>When the genetic differences between two people from the same population versus two people from different populations are compared, the increased genetic variation observed across populations is marginally incremental.</i>

develop an even deeper understanding of these concepts, I encourage you to take a look at the interactive resources developed by Brian Donovan’s team at BSCS Science Learning (Donovan 2021) and by the American Anthropological Association (Artz 2012). Taken together, these data should help us understand that most human genetic variation exists within groups, that the identifiable differences between groups are not as significant as one might have thought, and that most human genetic variation can be cataloged even by examining individuals from within the same group.



While these conclusions are evident from a close examination of the data, it tends to seem counterintuitive to the sensemaking most of us engage in

based on personal observations. For example, how can it be that we are *this* genetically alike when we can see differences plainly in terms of skin color and hair texture, traits that do have some genetic basis? Let's consider skin color in particular. If you were to assess skin color within and across populations, you would see the same patterns of variation we see in terms of genetic variation. Entire populations are not a single skin color, and there are not discrete differences in skin tone from one population to the next. While, on average, people who come from equatorial populations have more melanated skin tones, there is no place on the globe where you can draw a line between People of Color and White People. Furthermore, the existence of a range of skin tones within a single population is the basis for yet another form of bias, colorism, where people with fairer complexions hold higher social status than those with more melanated skin.

At its core, race is a socially constructed way of grouping people, designed to maintain systems of hierarchy by using skin color, hair texture, and other physical differences to differentiate in-groups and out-groups. While skin tone and hair texture may have a genetic basis, the other traits that are lumped into the attributes defining racial groups do not. As we discovered in our deep dive on genetic data, we are far more alike than we are different. Unfortunately, this essential truth is frequently misunderstood, and genetic essentialist thinking leads people to believe that there is a fundamental biological difference between people based on race and many other attributes.

An example of racial genetic essentialism is embodied in people's views on pain perception. Many people falsely believe that Black people literally have thicker skin than White people and that this characteristic renders them less susceptible to pain. This falsehood was originally put forward as a justification for the brutalization of enslaved Black bodies and persists to this day, even among individuals training in the health care professions. In a study on views of pain perception (Hoffman et al. 2016), half of a group of White medical students and residents held false beliefs about biological differences between Black bodies and White bodies with respect to their ability to feel pain. This misled belief in the existence of specific biological differences between the races correlated with biased interpretations of pain perception and appropriate treatment recommendations when comparing Black and White patients. That is to say, these individuals with

medical training believed there was a biological basis for underestimating pain felt by Black patients and overestimating pain felt by White patients. The ramifications of just this one mistaken view can begin to explain health disparities, like the difficulties experienced by Black patients with sickle cell disease who are often labeled as drug seeking when they are just trying to alleviate excruciating pain associated with their chronic condition. It may also help us understand the widespread overuse of opiate painkillers among White patients. Furthermore, it connects to the same flawed notion that Dr. Marion Sims, sometimes referred to as the father of modern gynecology, used to justify his use of enslaved Black women as unanesthetized subjects for his experiments in gynecological research (Villarosa 2019). In one particularly horrifying example, Dr. Sims performed 30 unanesthetized procedures on an enslaved woman named Anarcha before declaring his repair of her vesico-vaginal fistula a success. It is frighteningly easy to draw a direct line from this type of mistaken thinking about pain perception to health care disparities and trauma, in this case, in failing to treat pain in Black patients. Imagine how many other instances of connections between biased thought and disparate outcomes exist beyond this case example.



By scientifically debunking the concept of genetic essentialism, the erroneous idea that our differences are encoded at the most essential level in our DNA, we can begin to move away from a deterministic view of genetic influences. I am so grateful that you, my readers, and our greater community of anti-racists are working to advance this critical work. However, the work is made unnecessarily more difficult when scientific leaders espouse harmful and wrong ideas in very public ways. In one of the most egregious examples of this type of behavior, Dr. James Watson, the scientist known for his Nobel Prize-winning work elucidating the molecular structure of DNA, has repeatedly said that he believes people of African descent are fundamentally inferior to others (Harmon 2019b). In putting forth his shameful views, he has said that he is “inherently gloomy about the prospect of Africa” because “all our social policies are based on the fact that their intelligence is the same as ours, whereas all the testing says, not

really.” This concept is not only blatantly racist, but it also willfully ignores the role of structural racism in contributing to disparities in IQ tests, learning assessments, educational attainment, and the numerous other proxies used to measure intelligence. Thankfully, scientists have begun having open and honest conversations about the harm they have been contributing to these discussions both intentionally, through divisive, inaccurate propaganda, and unintentionally, through less-than-careful communication on these nuanced topics. Scholars have also offered critiques to these messages, with helpful suggestions on how to get these conversations, categories, and labels right for all of us (Kahn et al. 2018; Lewis et al. 2022; Committee on the Use of Race, Ethnicity, and Ancestry as Population Descriptors in Genomics Research 2023). As 67 scholars wrote in their opinion piece, “There is a difference between finding genetic differences between individuals and *constructing* genetic differences across groups by making conscious choices about which types of group matter for your purposes” (Kahn et al. 2018). While this gives us a place to start, we still have a long way to go to unlearn the harmful ideas that have fed into widely held misconceptions.

To that end, let us continue on this journey together. Our next stop will be bringing a critical lens to sex and gender.

5 Biology of Human Populations

SEX AND GENDER

The extent to which gender is embedded in our cultural understanding of human identity is exemplified by the growing trend of gender reveal parties. These parties are occasions for expectant parents to celebrate their babies with loved ones by sharing what they have learned from standard prenatal testing. The celebrations include cakes that when cut open reveal blue or pink filling, balloons that are popped to expose pink or blue confetti, and even forest fire-causing pyrotechnics that explosively shower down pink or blue smoke (Morales and Waller 2020). Notably, the mere existence of these parties highlights multiple fundamental misunderstandings about what gender actually is and uses a restrictive binary frame of reference.

To properly discuss sex and gender, we must begin with a shared understanding of how sex and gender differ from one another, as well as how they are distinct from gender expression and sexual orientation. Sex is typically assigned at birth by a health care provider and largely reflects the variable that is most easily observable by a health care provider, a newborn's genitalia, rather than the more complete combination of an individual's sex-specific physical characteristics, including genitalia, chromosomes, and sex hormones. Gender is a person's innate sense of whether they are a girl/

woman, boy/man, a blend of the two, or neither. Gender expression has to do with how one's internal sense of self is outwardly expressed through clothing choices, haircuts, and chosen activities that are often categorized socially as masculine, feminine, or androgynous. And finally, sexual orientation has to do with who an individual loves, whether their sexual interests include people of the same gender, a different gender, multiple genders, or none of the above. Most importantly, while there may be common associations between some of these characteristics, none of them exist exclusively within binary categorizations, nor do they necessarily exist in strict correlation with one another. Counter to commonly held beliefs and to the binary sex and gender essentialism that is frequently taught in schools (Donovan, Syed et al. 2024), sex itself exists on a continuum, with intersex individuals, who have variations in primary and secondary sex characteristics that do not fit the binary definitions of male and female, present at an estimated prevalence of nearly 2 percent in the general population (Blackless et al. 2000). Intersex variation can involve sex chromosomes, genitalia, gonads and the sex hormones they produce, responsiveness to sex hormones, and internal reproductive anatomy. The very existence of intersex individuals proves that sex exists on a continuum beyond the binary. Furthermore, an increasingly large number of young adults identify as lesbian, gay, bisexual, or transgender (19.7% of members of Gen Z, born 1997–2004) (Jones 2023). Identity is a uniquely personal quality that reflects one's individual sense of self and cannot be dictated externally.

One might assume that with the rising number of young people feeling empowered to live authentically, the climate welcoming people of all identities would be improving, yet there still is nowhere on this planet where true equity of individuals by gender is demonstrated. The World Economic Forum quantifies different aspects of the observed gender gap using its Global Gender Gap Index framework that relies on measures of (1) economic participation and opportunity, (2) educational attainment, (3) health and survival, and (4) political empowerment (World Economic Forum 2022). According to these measures, while countries as a whole are moving toward equity, the starting points and rates of progress are highly variable, and on balance, it is fair to characterize most societies as patriarchal in structure. Observations like these have led many to erroneously conclude that there is something inherently different about the male

sex, that they somehow have a biological advantage over other sexes that explains their social advantages. Furthermore, the lens through which these gaps are viewed is often binary in nature and frequently conflates sex and gender. These oversimplifications further complicate the possibility of properly understanding the root causes of this gender gap.

In order to effectively address the gender gap, we need to know more about what science tells us about human development in terms of sex, as well as how gender differs from sex. In humans, the most common sexes are male and female. At conception, individuals typically have either XY (male) or XX (female) sex chromosomes. Early in development, the human reproductive system exists in an ambipotential state, that is, the precursors for either typical male or typical female reproductive systems exist (Ainsworth 2015). The genes on the sex chromosomes, especially the SRY gene on the Y chromosome, in conjunction with other genes in the genome, will help promote the development of sex-specific reproductive systems, including the gonads; males develop testes while females develop ovaries. The gonads have two critical roles in sex development: to grow the reproductive cells, called gametes (i.e., sperm and eggs), and to drive the next steps in sex development by producing sex hormones (i.e., androgens such as testosterone versus estrogens). Sex hormones control secondary sex characteristics, like the development of breasts, external genitalia, hair growth, muscle mass, and other traits. Sex hormones also are not exclusive to individual sexes. While males have more androgen and females have more estrogen, both types of hormones can generally be found at some level in all human bodies (Personal Genetics Education & Dialogue 2019). The combination of genetic or chromosomal sex, gonadal sex, and anatomical sex defines the key features of human sex, and these features are interrelated in that genetic sex influences development of the gonads, which in turn produce hormones that influence anatomical sex.

The process of sex development as outlined thus far describes the most common paths that these processes take. However, there is an entire spectrum of sex development that exists between these two poles and beyond the binary (Montañez 2017). In intersex development, also referred to as differences of sex development (DSD) and variations of sex characteristics (VSC), the combination of chromosomal, gonadal, and anatomical sex varies from the typical combinations outlined above. Broadly speaking,

these unique combinations can arise as a result of variation in sex chromosome constitution, sex hormone profile, or sex hormone response.

With regard to the sex chromosomes, some people inherit an atypical number of sex chromosomes, with either extra or missing full or partial sex chromosomes (Ainsworth 2015). Chromosomes segregate into gametes at the time of meiosis (a special kind of cell division that halves the genetic content in sex cells in preparation for fertilization between sperm and egg, which restores full genetic content). Sometimes, chromosomes can be missorted in this process, a phenomenon known as nondisjunction that is frequently associated with advanced reproductive age in oogenesis (the process of creating egg cells) but that can occur in sperm as well. Any variation from the expected number of chromosomes, that is, 23 full pairs of chromosomes, is referred to as an aneuploidy. Nondisjunction of sex chromosomes in meiosis can result in gametes with extra or missing sex chromosomes, which, upon fertilization, can lead to combinations such as X (Turner syndrome, where a second sex chromosome is absent), XXY (Klinefelter syndrome), XXX (triple X syndrome), and XYY (Jacobs syndrome). Because the sex chromosomes include genes important for sex development, many of these combinations of chromosomes are associated with variations from typical sex development, including development of intermediate gonads (gonadal dysgenesis/ovotestes), changes to external genitalia, and impacts on puberty and fertility. It is also possible for portions of chromosomes to be translocated (moved) onto their partner sex chromosome during meiosis; this can impact sex development if the SRY gene translocates from the Y chromosome to the X chromosome during spermatogenesis. An individual may inherit a Y chromosome missing the key gene SRY, which would prevent typical male development from fully occurring even though the individual appears to be XY. Likewise, an individual may inherit an X chromosome with the SRY gene transferred, which will influence sex development of this XX individual to express intersex characteristics.

The amount of and response to sex hormones can also impact sex development beyond the binary. For instance, individuals with androgen insensitivity syndrome (National Library of Medicine, MedlinePlus 2024) are XY and develop testes that are capable of producing androgens. However, the genes responsible for responding to those androgens have a variant

that prevents them from reacting to the presence of androgen. As a result, the secondary sex development does not involve the series of events usually seen in XY individuals, and the external genitalia will develop in a way more typically concordant with female sex while the testes will remain in the abdomen. In contrast, congenital adrenal hyperplasia (CAH) is a condition associated with androgen excess. Therefore, in an XX individual with CAH, ovarian development would proceed as expected, but the presence of excess androgen can lead to intermediate genitalia, such as a visibly enlarged clitoris. For individuals with CAH, there is some evidence for elevated risk for gonadal and adrenal cancers, which may necessitate risk-reducing gonadectomy and discussions about fertility preservation. However, careful examination of these risks, including malignancy risk percentages and appropriate timing of gonadectomy when medically necessary, is needed to avoid traumatizing intersex individuals through use of a medicalized model of care that does not center individual agency (Haghighat et al. 2023). Importantly, on the issue of timing, evidence may ultimately support early surveillance followed by postpubertal gonadectomy, which is a time frame when individuals can personally give consent or assent. There is also an additional collection of genes, including SOX9, WT1, and SF1, which can cause a shift toward typical female development in XY individuals when one copy of those genes harbors a particular variant allele. As should be concluded from these examples, biological sex does not exist within simple binary categories, but rather on a spectrum that includes a wide range of intersex possibilities.

There are four common points in time when sex is clinically ascertained. This can occur during prenatal development as part of the cell free DNA (cfDNA) prenatal screens. This type of testing can identify if someone has XX, XY, X, XXX, XXY, XYY, or another combination of sex chromosomes. Some patients refer to this as the “gender test,” which is an error conflating sex and gender and centering cis-normativity (Llorin and Zayhowski 2021), much like in gender reveal parties. Rather, this test just tells the parent the chromosomal sex of their baby. The second common time sex can be assessed is at birth, when health professionals examine the external genitalia. At this stage, particularly for folks who are intersex, transgender, and nonbinary, there is potential for significant error and harm. In the case where an intersex individual may have a visible genital

variation, such as an individual with CAH and an enlarged clitoris, it had been the case that the variation was frequently viewed through a medicalized lens, and it was common to recommend “normalization feminization or masculinization” surgeries (Liao and Baratz 2022), which were largely cosmetic in nature. The rationale for this intervention was focused on psychosocial implications for fitting into a society that is largely constructed in binary ways around gender. Importantly, it has been the case that these surgeries would be performed very early in development, before children could express their own preferences with respect to genitoplasty or their gender, so parents and providers would be making significant clinical decisions that would impact these children throughout their lives without their consent. The prevailing clinical recommendations are shifting to avoid nonconsensual surgical intervention until the individual can express their views on the decision that impacts their own body (Conway 2023; Jamal et al. 2024), particularly because these childhood procedures can have lifelong implications (Crocetti et al. 2020). However, recent indicators suggest little is changing with respect to the frequency of these surgical interventions and that parental advising continues to happen in the absence of psychosocial expertise (Liao and Baratz 2022). Importantly, though sex, a biologically defined variable, is not at all the same as gender, which is individually and socially defined, it has been observed that among people who are intersex, there is more gender and sexual diversity than among people without variations in sex characteristics (Jones et al. 2016). That is to say, the use of “normalizing” surgical interventions constructed through a cis-normative framework would have a significant chance of getting the individual’s preferences wrong without input from the patient themselves. A third point at which sex may be reassessed would be during adolescence, with the onset of puberty. For instance, for situations where prepubertal childhood development appears to fit within a binary gender construct, a condition like Turner syndrome may not become evident until someone assigned female at birth does not experience the onset of menstruation. This type of revelation has the capacity to vastly change a young person’s developing sense of self. Finally, aspects of sex variation may not come to light until individuals engage in family planning and experience infertility, which is often seen when gonads develop along intersex axes. The timing of each of these points in the life cycle during which sex is

ascertained has huge implications for the person in question, depending on their own views and the views of their providers and guardians.



An aspect of society that remains entrenched in a binary framework is that of competitive sports. It is routine to find athletes grouped according to binary sex designations, from Little League to the Olympics. In terms of elite sports, it was commonplace to invoke medical examination to verify athletes' indicated sex (Nyayieka, Macfarlane, and Shelley 2021). In the 1960s, this was carried out by physical examinations known commonly as nude parades, in which an athlete would be conferred a Certificate of Femininity (Padawer 2016). This humiliating practice was replaced with the "sex chromatin test," which was a way of assessing the identity of sex chromosomes present in an individual's genome (Cressey 2009). When María José Martínez-Patiño qualified to compete in the 1983 World Track and Field Championships in Helsinki, Finland, she represented Spain at the age of 22 and passed her first sex test, receiving a Certificate of Femininity (Martínez-Patiño 2005). In 1985, Martínez-Patiño went to the World University Games in Kobe, Japan. Unfortunately, she didn't bring her certificate along, and the sex verification used at the 1985 games was a genetic test that did not show presence of the expected two X chromosomes. Her testing had to be repeated, and it would not be completed in time for her to compete, so she faked an injury to avoid the public gaze while she sat out the competition. Ultimately, the karyotype analysis that examined all of Martínez-Patiño's chromosomes showed that she had XY sex chromosomes, more typically found in males. It turns out that she has androgen insensitivity syndrome, which, as we discussed before, renders her body unresponsive to testosterone. Martínez-Patiño developed breasts and a vagina, just like her athletic peers, and she identified as a woman. Ironically, she was probably at a disadvantage relative to her competitors due to her condition because females typically have some level of circulating testosterone to which their bodies, including their muscles, are able to respond. When news of her condition was leaked to the press as she won the 60-meter hurdles at the 1986 National Championships, she was expelled from the athletes' residence, she lost her

sports scholarship, her record times were erased in her country, and she was left by friends and her fiancé. This test cost her everything. Though she fought the disqualification with the help of experts, it was not until 1988 that her license to compete was reinstated by the chair of the International Federation for Athletics, Arne Ljungqvist, but by then she had lost momentum and failed to qualify for the 1992 Olympics in Barcelona, Spain, by one-tenth of a second. Although the reversal of this ruling didn't allow her to resume her career, Martínez-Patiño takes pride in the fact that she was able to help "other sportswomen with genetic variance participate without fear" (Martínez-Patiño 2005).

Sex testing to verify athletic eligibility has evolved from the use of sex chromatin testing thanks in part to Martínez-Patiño, but it still remains a flawed practice. The International Olympic Commission and the International Association of Athletics Federations jointly adopted a new set of regulations in 2011 centered around levels of circulating testosterone in the athletes' bodies (Marchant 2011). Females typically have 0.12–1.79 nanomoles of testosterone per liter, while males usually fall in the range of 7.7–29.4 nanomoles per liter (Ingle 2020). These ranges encompass roughly 99 percent of the range in the female and male sexes; however, as with any biological trait, there are natural variations and outliers with respect to these ranges. Hyperandrogenism (high levels of androgen, including testosterone) is often found in people with androgen insensitivity syndrome because the body, not sensing the presence of testosterone, believes it needs to increase its production. People like Martínez-Patiño would have passed this test due to an exception not holding people with androgen insensitivity syndrome to this standard, given the reality that the excess testosterone confers no biological benefit to these individuals.

When the appropriate receptors are present to convey signals from the hormone, testosterone influences physiological traits that may confer a biological advantage to athletes, including muscle development, height, and circulating hemoglobin levels (which will influence oxygen-carrying capacity in the blood). For this reason, there has been a great deal of controversy in athletics on how best to factor in variation in sex development that includes people who are able to respond to testosterone. Caster Semenya, a South African track star, has had this controversy shadowing her career for decades. She has been subjected to increased scrutiny

compared to most athletes in part because of her dominating performance in the 800 meters, including her two Olympic gold medals and three World Championship wins. After a 2009 finish where she blasted past her competitors, she was pulled for further testing amidst rumors about her sex and her eligibility. The suspicions were heavy with misogyny and racism, and the young athlete retreated from the public eye while the sex testing was carried out. Ultimately, tests confirmed that Semenya had hyperandrogenism (high testosterone) due to a condition known as 5-alpha reductase deficiency (National Library of Medicine, MedlinePlus 2017), in which the individual has XY sex chromosomes and testes. The testes will produce functional testosterone; however, the related hormone, dihydrotestosterone (DHT), is not produced at typical levels because the 5-alpha reductase deficiency limits conversion of testosterone to DHT. A shortage of the DHT hormone, which has a central role in male sex development, frequently results in formation of female external genitalia or intersex genitalia. In Semenya's case, she was born with female genitalia and assigned female at birth. At puberty, many people with 5-alpha reductase deficiency experience development of some secondary sex characteristics as a result of increasing male sex hormone levels. When tested, Semenya's testosterone levels fell in the low end of the typical male range, and together with the presence of the Y chromosome, resulted in further examination of her eligibility to compete. For the love of sport, she subjected herself to treatment with testosterone-lowering medications to retain her eligibility, and she competed, though not in full form as before. Since then, she has said she won't return to the medications she took from 2010 to 2015, stating that they made her feel constantly unwell. Subsequently, rules were created that seemed to single her out as an athlete, naming her specific events (middle-distance running) and caps of testosterone levels at five nanomoles per liter, just below her measured levels. To continue competing under the revised rules, Semenya was required to either change her events or take medical steps to reduce her circulating testosterone levels through surgical removal of her testes or through hormonal suppression. Given her previous negative experiences with the testosterone-lowering medications, she instead pursued the route of retraining for distance events and was beginning to regain her footing just as the World Athletics Council pulled the rug out from under her

again by both further lowering the testosterone caps to 2.5 nanomoles per liter for six months prior to competition and extending these caps to all the track events (Pells 2023). It is hard to see this moving eligibility target as anything but personal and motivated by exclusionary, biased thinking designed to dehumanize athletes just trying to compete as they were born. It also leaves us with the question of whether it is appropriate to police natural variation in testosterone levels differently from biological variants in unregulated traits, including oxygen-carrying capacity, limb length, joint flexibility, and any other qualities that could potentially contribute to athletic performance, particularly when the extent of the influence of natural testosterone levels is far from settled (Padawer 2016).

The more recent rules with further reduction in testosterone limits were created with transgender athletes in mind as well. Trans women who were assigned male at birth are being evaluated for athletic eligibility based not only on their current hormonal status but also on the timing of their medical transition relative to puberty. Swimmer Lia Thomas found herself navigating all these evolving rules as an athlete who began her collegiate swimming career in 2017 competing for the Penn men's swim team (Barnes 2022). As an NCAA athlete, she had four years of competitive eligibility during college. Her first three seasons were spent swimming with the men's team, even while she began hormone therapy between her second and third seasons. During her second season, she posted the men's team's top times in the 500-, 100-, and 1,650-meter freestyle events, and during her third season, after her medical transition, she competed against men in a women's suit. The mental health toll that Thomas's gender dysphoria was taking had a dramatic impact on her well-being, which led her to pursue her medical transition, even though she assumed it would mean the end of her swimming career. Thomas took a year off school after her junior year to maintain her NCAA eligibility when the COVID-19 pandemic resulted in the cancellation of collegiate competitive swimming. Upon return for her fourth and final collegiate season, she had lost muscle mass and was slower by 15 seconds in her signature event, the 500-meter freestyle, but it was worth it to Thomas to compete as her authentic self on the women's team. At the end of the season, Thomas became the first trans woman to claim a national title in swimming when she won the 500-meter freestyle at the NCAA swimming and diving

championships, and this resulted in increased complaints about the fairness of her competing against women (Powell 2022). It has long been a goal of Thomas's to swim in the US Olympic Trials, and though her performance levels suggest she would be competitive, the current Olympic eligibility requirements preclude Thomas's participation because she has gone through testosterone-driven puberty. While testosterone-driven puberty may have influenced her cardiovascular capacity, her wingspan, her pelvic width, and her muscle mass, some of this would be blunted by postpubertal testosterone suppression paired with hormone replacement therapy (Hamilton et al. 2024), and the research on the influence of naturally occurring endogenous testosterone is far less settled than the science behind doping with exogenous testosterone. There is enough variation among elite athletes with respect to hormone profiles as to undermine any definitive relationship between testosterone and athletic performance. As put by Louisa Thomas, if testosterone were a determinative characteristic that was predictive of performance, one would be able to rank athletes by measurements of their testosterone levels and use it to predict the outcomes of their competitions (Thomas 2022), giving no consideration to technique, strategy, training, dedication, grit, nutrition, coaching, and numerous other variables that feed into athletic performance. In other words, it is not as simple as presuming that a trans woman will automatically have an athletic advantage when competing in women's sports.

The regulations around competitive sports are only partly an attempt to create a level playing field for female athletes; they also reflect a larger culture war against transgender people. Hundreds of bills have been introduced in legislative bodies around the country that focus on limiting the access of transgender folks to bathrooms, health care, and yes, sports. In terms of sports, there is also the important question of whether youth sports should be governed differently than elite sports (Cretaz 2022). For trans youth, inclusion in sports is a critical opportunity to give them a sense of community, as well as a venue to develop self-esteem and leadership skills. This is especially important when we consider the huge risks trans youth experience with respect to violence, bullying, self-harm, suicide, depression, and housing insecurity, among others. Bills banning the participation of young people in sports based on their gender are centered on the fears of cis folks and not on the harms experienced by gender

diverse people. Importantly, we must also consider who is impacted by these exclusionary tactics and who is involved in policing their bodies. Frequently, enforcement of these regulations disproportionately targets people with intersectional identities of Black, Brown, trans, nonbinary, and otherwise marginalized identities that are measured against a cis-White normative “ideal” imagined by governing bodies that do not include folks from marginalized groups. The dehumanization of athletes subjected to these restrictions is rooted in bias rather than science and seeks to erase trans people from society. Brooke Forde, a Stanford swimmer who competed at the Tokyo Olympics, put it beautifully in her statement released ahead of her competition against Lia Thomas at the NCAA national championships: “I believe treating people with respect and dignity is more important than any trophy or record will ever be.”



All too often, it can feel like gender roles are hardwired into social norms. I remember picking my child up from preschool shortly after they had acquired a new play structure, an epic pirate ship that was the hot ticket on the playground. When I arrived to pick up my daughter, I was shocked to find the preschool teachers redirecting her from the ship toward another playground structure, encouraging her to play house rather than pirates. The ship, they explained, was for the boys. I remember feeling utterly incensed. I never imagined that I would find myself in the position of having to advocate for the right of my daughter to play act as a pirate, but that is exactly where I found myself in that moment. It made me wonder how many other times per day in countless other ways people were placing limits on her existence in both subtle and overt ways, even as young as she was.

Just as scientific racism was used to justify keeping people of color in subservient positions, the science exploring sex differences has been used to further parallel goals to maintain men's elevated status in society. People have extrapolated from studies examining differences among the sexes to put forth ideas about behavioral tendencies and intellectual capacity of the sexes. Scientists suggested that males were more aggressive and females were more nurturing. They have also suggested that males

have more intellectual capacity for subjects like math and science than females do. These theories have been repeated in harmful ways by people in power, including Larry Summers, who, while serving as Harvard president, suggested that perhaps the underrepresentation of women in STEM fields was somehow related to differences in innate abilities (Hopkins 2005) rather than acknowledging the myriad systemic barriers that explain the observed differences (Valian 1997; Cheney and Shattuck 2020).

The scientific sexism studies that have been used as rationalization for existing disparities among the sexes have significant flaws in their approaches and interpretation (Saini 2017). The collected data is high inference in that it is often many steps away from the suggested significance of the observed differences. For instance, examining newborns' time spent gazing at faces and then implying that any observed differences between female and male babies suggests a difference in adult empathy and caregiving tendencies requires a series of massive logical leaps and lack of attention to social conditioning in the intervening years. Furthermore, the data itself is highly overlapping to the extent that there really are not clear profiles associated with one sex and not another. Specifically, about 25 percent of the male babies preferred gazing at the face and 40 percent preferred the mobile, while 36 percent of the female babies preferred the face versus 17 percent preferring the mobile. Because of this extensive overlap, there is no predictive ability to use a data signature to determine the sex of the test subject or vice versa, to use the sex to predict a baby's preference. Notably, this mirrors the prior evidence we considered, demonstrating that sex itself is not binary but that it exists on a continuum. Also importantly, the absence of evidence for sex differences is a negative result and thus would be unlikely to survive the research article publication process, which, due to the nature of the system, will be skewed toward publishing findings of difference rather than the absence of difference. The combination of these factors discredits the existing literature on sex-based differences even while the patriarchal systems we navigate continue to use them as false justification for maintaining a binary hierarchy.

An important counterpoint to the comments of Larry Summers is the life history of Dr. Ben Barres (Barres 2006). Dr. Barres was assigned

female at birth. He had a distinguished scientific career involving training at MIT, Dartmouth, Harvard, Cornell, and University College London before becoming a faculty member of neurobiology at Stanford. Dr. Barres began his research career at Stanford presenting socially as a woman and transitioned to a man four years later. His lived experiences allowed him to comment on the bias he experienced when navigating academia as a woman, and he also describes the higher respect he received after transitioning by people who were unaware that he was a trans man. After his first seminar as a man, once scientist commented that Dr. Barres's seminar was so much better than anything previously presented by his "sister," who, of course, was Dr. Barres himself a few years earlier. It is hard to imagine a clearer demonstration of the powerful forces of bias and sexism while debunking the significance of sex-based difference.



A major source of the tensions described in this chapter is the inherent mismatch between the continuous nature of sex and gender and the social constructions of gender that tend toward the binary. True feminists will see the parallels in marginalization experienced by women and gender diverse folks, and we must ally along these lines of common experiences while advocating for the unique concerns of each other. In this chapter, we have explored how the concept of bodily autonomy, the right to live authentically in one's own body, is tied to sex and gender. This principle applies to both women and gender diverse people in many areas, particularly with respect to pregnancy, as we will see in the next chapter.

6 Pregnancy, Bodily Autonomy, and Reproductive Justice

From a scientific point of view, the events of pregnancy are simply awe-inspiring; that the information encoded in our genes can map out the journey from a single cell to a baby with tens of billions of cells, hundreds of types of specialized tissues, and dozens of organs is simply remarkable. From a parental point of view, the period of pregnancy is one filled with the strongest of emotions, from hope and optimism to fear and despair. Pregnancy can be a truly miraculous phase of life, but in a society with limited social support, such as in the United States, the fears of pregnant people encompass well-founded worries about things that feel like they are beyond their own control, including health, finances, childcare, employment, and bodily autonomy. In this chapter, we will be examining the typical course of preconception and prenatal screening, the complex choices faced by pregnant people, and the legal landscape of access to medical support.

In exploring these topics, I will also continue to model inclusive language, which has been important throughout this book, but is especially important in the context of pregnancy where cis- and heteronormative lenses are often assumed. Two terms that I will frequently be using in this chapter are pregnant people, a term that is inclusive of all pregnant indi-

viduals regardless of gender, and gestational carrier, which again is inclusive of all people carrying a pregnancy regardless of gender or intent to parent the child. As with all topics in this book, exclusionary language serves to build walls and creates unintentional omissions wherein we are unable to clearly see how policies and practices affect a wide range of interested parties. Disparities in quality of care and access to care will fall disproportionately on people from marginalized backgrounds, so we must take special care to not further exclude these communities by using divisive language.



Genetics and pregnancy are intimately intertwined; from screening for traits that impact health to decisions around pregnancy continuation, genetics is involved in reproductive health care from preconception to postpartum phases. At the preconception phase, there is an opportunity for risk assessment for genetic conditions, while during the prenatal and postnatal phases it is possible to diagnose genetic conditions at different stages of development. The ability to interpret genetic test results accurately and effectively at all these time points is influenced by the ancestry of the person being tested. To understand why this is the case, we need to know more about the origins of our knowledge of genetic disease.

When correlations between genetic variants and human disease first started to be elucidated, scientists were initially studying populations in which these conditions were common. Furthermore, the best characterized conditions were those that impacted European populations over others. For example, cystic fibrosis was one of the first conditions to be characterized at the genetic level. The cystic fibrosis transmembrane conductance regulator (CFTR) gene was first identified in 1989 (Riordan et al. 1989; Rommens et al. 1989). Further studies described how alleles (versions) of this gene that were associated with cystic fibrosis impeded normal chloride ion transport, which affected many tissues in the body, most notably the lungs. The imbalance of chloride ions in the lungs led to the accumulation of very thick mucus, which created an environment that bred respiratory infections and negatively impacted respiration. While this represented a huge leap in genetic knowledge, it is important to acknowledge that this

Table 2 Examples of elevated carrier frequencies by ancestral group

Disorder	Ancestral group	Carrier frequency
cystic fibrosis	European	1/25
spinal muscular atrophy	European	1/41
sickle cell anemia	Nigerian, South Asian, and the Democratic Republic of Congo	1/3–1/10
alpha-thalassemia	Nigerian, Ivory Coast, and Kenyan	1/3
alpha-thalassemia	East Asian/Southeast Asian	1/6 (some areas)
beta-thalassemia	Mediterranean	1/8–1/12
Tay-Sachs disease	French Canadian	1/30
Tay-Sachs disease and more	Ashkenazi Jewish	1/25

SOURCE: Hoffman 2022.

condition is found most often in European ancestries and that follow-up work characterizing the genetic mechanisms of disease have also focused primarily on patients of European descent. That is not to say we don’t have knowledge of conditions in other populations. Some examples of genetic conditions, the populations in which they are most prevalent, and the approximate carrier frequencies (the rates at which people may harbor a silent copy of the disease-causing variant) are indicated in table 2.

Genetic disease exists on a continuum, with some conditions resulting largely from a high-impact causative change in a single gene, whereas at the other end of the spectrum are conditions resulting from a collection of genes, each with a small individual impact, and environmental factors together. The first gene-disease correlations were discovered at the end of the spectrum where an individual gene was primarily responsible for a condition on its own. Those original gene-hunting approaches were often carried out in populations where a single, strongly predisposing gene was present at an elevated frequency, such as those described in table 2. This allowed identification of families with multiple people affected by the condition across multiple generations, a family structure that optimized the mathematical ability to detect gene-disease correlations. A complication

with this approach to the study of genetic disease is that genetic essentialist thinking began to creep into understanding of genetic disease and who is at risk. People without a nuanced understanding of how genetic variation is distributed over populations commonly made errors by assuming that people outside of the at-risk populations did not harbor any susceptibility to these conditions and that people in the at-risk population were all predisposed to the given condition in the same way. In our deep dive into human populations in chapter 4, we came to the understanding that the vast majority of variation is present across populations, so while the variants may differ in frequency, we would still expect to see most variants, and their associated genetic conditions, across multiple populations.

When clinicians came up with the recommendations on how to offer genetic testing to patients, they applied both scientific information and cost-benefit analyses. In the context of preconception carrier screening for prospective parents, testing was initially rather expensive, and the detection rates in patients in the general population was low. In the case of cystic fibrosis, for instance, while the carrier rate in White populations is about 1 in 25, in Black populations the carrier rate is roughly 1 in 132, and in Asian populations the carrier rate is even lower at 1 in 174 (Lazarin et al. 2013). For these reasons, the historical recommendation of the American College of Medical Genetics and Genomics (ACMG) was to offer genetic testing when the following criteria were met: when the genetic condition under consideration had a severe clinical presentation, when the screening methods had established clinical validity, when the genetic variants had a predictable impact on the health outcomes of the patients, when there were available prenatal diagnosis and reproductive options, and importantly, when there was a high prevalence of carriers in a screened population (Gregg et al. 2021). Simply put, this policy codified race-based screening. Furthermore, health insurance companies often look to professional organizations to determine what is standard of care and deserving of health insurance coverage. From a financial perspective, administrators viewed the return on investment for patients outside of the at-risk population to be not worth the commitment of funds and time. As such, people outside of the limited at-risk populations were routinely not offered preconception carrier screening, and if it was offered, it might not have been covered by their health insurance, if they were insured at all.

The impact of race-based screening for genetic conditions meant that patients in at-risk populations were receiving more thorough preconception counseling than the general population. For instance, Tay-Sachs disease is a fatal degenerative disease with an elevated carrier rate in the Ashkenazi Jewish population. When a child is born with two pathogenic variants in their *HEXA* gene, Tay-Sachs ultimately leads to blindness, deafness, seizures, paralysis, and eventual death of children as young as four years of age. When carrier screening became available for prospective parents, it was embraced so fully by the Ashkenazi Jewish community that the number of births of children with Tay-Sachs was reduced by over 95 percent since the screenings were initiated in the 1970s (Kolata 2003). The number of children born with Tay-Sachs in the United States has dropped from 50 per year to 5, with most of the current cases born to parents outside of the Ashkenazi Jewish community. Herein lies the complication. The success and efficiency of screening in the Ashkenazi Jewish population with the goal of reducing cases of this devastating condition is undeniable, but the fact remains that with resources focused on this community, other at-risk families in the general population are not identified until after their first affected child is born.

Growing awareness of the unintended consequences of race-based carrier screening and a careful self-examination of the field's contributions to racial health disparities, alongside the dropping costs of genetic testing, converged in a revision of the ACMG recommendations for carrier screening (Gregg et al. 2021). In these updated recommendations, the authors center the principle of equity in offering ancestry-agnostic carrier screening principles. They note that restricting carrier screening to certain ancestries is problematic not only for the reasons we have just explored, but also because identity itself is a complicated variable. For instance, there are issues about how the ancestry of prospective parents is determined at intake. Is it self-reported? Is it a best guess of a health care provider? Does it focus on race and ethnicity or true genetic ancestry? What categories are used? How does it account for people from mixed ancestral backgrounds? All these questions highlight the inherent inequities built into the process of using ancestry to determine eligibility for genetic screening. In revising their recommendations, the ACMG experts write, "It is important that carrier screening goes beyond commonly recognized

at-risk groups and includes diverse populations” (Gregg et al. 2021). Given the documented relationship between insurance status and access to genetic testing (Choi et al. 2022), it will be important to advocate for implementation of this updated professional recommendation at the level of insurance coverage of genetic testing.

While there has been significant progress on inclusivity in carrier screening along the axes of race, ethnicity, and ancestry, there remain significant inequities in preconception carrier screening when assisted reproduction is considered for patients who are building families headed by same-sex parents, for transgender patients who have gone through gender affirming therapies that impacted their ability to generate gametes, for single parents by choice, or for patients who are struggling with infertility. In any of these situations, the prospective parents may find themselves seeking out donor gametes to help them build their families. The donations can be obtained from a known donor or from an anonymous donor through a gamete bank. In some ways, this allows a unique opportunity for preconception carrier screening, since so much planning goes into the assisted reproduction strategy itself. The reality of traditional conception is that prospective parents are more often going through genetic screening after the conception takes place, but with assisted reproduction, it can be built into the fertility plan. Although assisted reproduction allows for conversations about screening early in the process, there are logistical hurdles that can adversely impact access to genetic services for these prospective parents.

In assisted reproduction, there is frequently a desire for the child to have a biological connection to the intended parents, but that might be complicated if the parents are the same sex or if a parent does not have viable gametes to contribute. In this case, one option is to ask for a gamete donation from a relative, for instance, a sister of a gay man who seeks to have a child with his husband. In this way, the resulting child could have a genetic connection to both fathers. When the issue of how to layer genetic testing into this scenario is considered, however, it begins to get more complex. In the case of gay men, a gestational surrogate is necessarily involved, and it is unlikely that most of these costs would be covered by health insurance (Weigel et al. 2020). In some cases, it is possible to work through infertility diagnoses to get some of the procedures covered, but

genetic testing of a relative could pose a complication in coverage. One would have to navigate whether the gamete donor could go through pre-conception genetic testing and gamete harvesting potentially through her own insurance to support the intended parents. Overall, the process of assisted reproduction is incredibly expensive, easily reaching six-figure costs depending on the number of pregnancy attempts required and whether a gestational surrogate is involved; gaps in insurance coverage would only increase these costs further.

In any of the aforementioned family structures, the prospective parents may opt to use an anonymous donated gamete. Sometimes this strategy is chosen out of biological necessity, and other times it may be chosen to simplify the roles of the extended family in the child's life. In the scenario where a gamete donor is being selected from a sperm or egg bank, the standard genetic screening utilized at the time of donation may not be up to current screening standards. It is complicated to determine what gaps in screening may exist, with the technology rapidly evolving and including more and more genetic conditions over time. Furthermore, if a parent themselves has a family history of a specific genetic condition, they may wish to determine if the donor has the same risk. In each of these scenarios, it would be reasonable to want to pursue further or updated prefertilization genetic screening in the donor; however, that would require the gamete bank to be able to reach the donor and for them to agree to provide a new sample, usually a blood sample, for genetic screening. Not only would this increase the associated costs, but it would require that the donor remain reachable by the bank.

In both cases, whether the donor is a known donor or an anonymous donor from a sperm or egg bank, the logistical, financial, and medical hurdles are significant and have the ability to amplify health disparities, particularly for gender and sexual minority groups. For LGBTQ+ individuals, they often do not meet the clinical definition of infertility, thereby disqualifying them for coverage of services. Similarly, for transgender individuals who have gone through gender affirming care, if fertility preservation steps were not taken at the time of their medical transition, their assisted reproduction options may subsequently be limited. In some cases, insurers will not consider gender affirming care a form of medically induced infertility (for instance, in comparison to a case of someone going

through chemotherapy), and thus will disqualify their coverage either for fertility preservation or for temporary suspension of gender affirming care to harvest gametes. While the American Society of Reproductive Medicine has stated that it is the ethical duty of fertility specialists to provide equitable care to LGBTQ+ patients, equally to heterosexual married couples, there remain many states where fertility preservation and assisted reproduction are not mandated to be covered for the LGBTQ+ community, and in fact, some states specifically and intentionally exclude LGBTQ+ patients from access to these treatments (Weigel et al. 2020).

Even if a patient is able to go through preconception carrier screening, there is still an important role for prenatal care that includes genetic screening to monitor the progress and status of the pregnancy. Routine prenatal care includes tests that evaluate both general health and specific factors that quantify risk for genetic conditions.

One of the original screens included in prenatal care was designed to quantify risk of there being a change in the genomic content resulting in extra chromosomes. This sometimes results when cells go through meiosis, a reductive form of cell division specific to producing gametes that halves the genetic content in anticipation of fertilization restoring full genetic content (first discussed in chapter 5). When this screening was initially introduced, it measured serum proteins found at different levels in pregnancies where there was an extra copy of chromosome 13, 18, or 21. Serum screening to test for changes in copy number of chromosomes or partial chromosomes (aneuploidies) was originally offered to gestational parents who were of advanced reproductive age (35 years old or older at the time of delivery). The rationale for this age-related screening was the observation that females 35 and older were at increased risk of experiencing mistakes during meiosis that resulted in changes in genomic copy number during egg formation (oogenesis). If that screen reported an elevated risk for aneuploidy, then a diagnostic test would have to be performed to reach a definitive result. This test is an invasive test involving sampling of amniotic fluid or the chorionic villus from the pregnancy itself, so it carried with it some risk of miscarriage. Since noninvasive screening was first routinely offered to pregnant people (Dungan et al. 2023), the screening technology has evolved in three important ways. First, cell-free DNA (cfDNA) circulating in the gestational carrier's blood

stream can be quantified to screen for differences in genetic copy number. This is possible because certain cells of the placenta release DNA fragments into the gestational parent's circulating blood, and this DNA can be purified for subsequent analysis. This allows people to detect amplifications or deletions of any DNA sequence, so there is no need to limit analysis to extra copies of specific chromosomes, though it is currently only recommended for a subset of these changes in copy number. Second, it is highly sensitive and can be offered as early as 10 weeks into the pregnancy, which is during the first trimester. Finally, prior generation serum screens quantified fetal proteins (e.g., alpha fetoprotein/AFP) and utilized a race-based adjustment for these measurements. Knowing that race is not a biological variable, accurate results can be obtained with adjustments based on weight of the gestational carrier and gestational age (Burns et al. 2023), or simply switching to the race-neutral cfDNA method, thereby reducing the potential for race-based disparities. Understanding why the serum screens employ a race-based adjustment factor while cfDNA does not isn't particularly intuitive. The serum screens quantify risk and therefore employ equations that seek to integrate values of a set of biomarkers, but in the absence of an equation that accurately predicts risk, some fudge factors, like race, are introduced to stand in for missing biological information. The cfDNA method, however, directly measures cell-free fetal DNA, which doesn't need any further adjustment to predict differences at the DNA level. As a socially constructed variable, race-based adjustment is not appropriate for risk assessment, and we will explore this in more detail in chapter 12. Importantly, results of cfDNA screens quantify DNA fragments released by the placenta and are therefore still reported as elevated risks requiring subsequent verification by diagnostic methods, such as amniocentesis and chorionic villus sampling, which directly test DNA found in fetal or placental cells. Because of these important advantages, cfDNA screening is now recommended for routine prenatal care of pregnant people, not just those of advanced reproductive age (Dungan et al. 2023).

The cfDNA screening approach is intended to be used for all pregnant people of general risk, but in some families, there are additional risks of particular genetic conditions, such as those caused by changes in single genes. If a DNA-based genetic test is available for a condition, it can be

employed in the prenatal stages by testing DNA derived from amniocentesis or chorionic villus sampling. Generally, it is recommended that directed, presymptomatic testing of minors be reserved for conditions with childhood age of onset rather than adult age of onset (Caga-anan et al. 2012; Committee on Bioethics et al. 2013; American Medical Association 2016; Ross et al. 2013; Botkin et al. 2015). The rationale for this recommended deferral is that the child has a right not to know, that is, to make their own decision about something that may occur in the future, and the landscape of treatment of a condition may be quite different by the time they reach adulthood. However, this line is quite blurry when considering recommendations for conditions whose age of onset spans the range of juvenile to adult, when risk-reducing interventions exist that can be initiated before adulthood, when the condition is life limiting and terminal, when the genetic status is determined incidentally to genetic testing initiated to answer different clinical questions, when consumer-facing testing is pursued directly by the patient themselves, and as the child matures and gains understanding of their own genetic risks (Hardart and Chung 2014; Anderson et al. 2015; Botkin et al. 2015; Hercher 2020). Similar rationale may be extended to prenatal testing, assuming that pregnancy management would not be impacted by a positive test result for an adult-onset condition; that is, it would not be recommended to test for adult-onset conditions prenatally, so that once the child is born they retain autonomy over the decision on whether to pursue testing and the right not to know. However, assumptions can be dangerous things to make, and even the parents themselves might not realize how a genetic test result would impact a decision about pregnancy continuation or termination until they have that result in hand.

The purpose of prenatal testing is not only to monitor the health and development of the fetus, but also to inform clinical and personal decision-making. Prospective parents may also choose to use this information to line up resources if their child will require complex care, or they may opt to make a decision on whether to terminate or continue the pregnancy. These decisions are emotional ones and are tied up with the realities of parenting a child with complex medical needs in the context of privatized medical systems, lack of subsidized childcare, limited family and sick leave, existing caregiving responsibilities, and so many other consequences

of an inadequate social safety net. Parenting children with complex needs frequently requires deep reserves of medical, emotional, financial, and temporal resources, which many are not privileged to have.

With respect to prenatal care focused on risk assessment for genetic conditions, it is important to keep the time line of care in mind. Even though cfDNA screening is available as early as 10 weeks, those tests must be analyzed, confirmed diagnostically by chorionic villus sampling or amniocentesis, and then, if desired, an abortion would be scheduled. The providers themselves are in short supply, with a national shortage of geneticists and genetic counselors, especially in rural areas, to help interpret these tests, thereby further contributing to delays in access to care. Moreover, the geneticists and genetic counselors that currently make up the workforce are overwhelmingly White people without visible disabilities, creating further barriers to accessing care among patients of color or patients with disabilities. Comparing the time line of confirmatory testing and genetic counseling steps with the limited windows during which abortion is available in states with timed restrictions that have taken effect since the Supreme Court ruling on *Dobbs v. Jackson Women's Health Organization* essentially makes abortion informed by prenatal testing inaccessible in many states enacting abortion bans and restrictions (Hercher 2022; Suter and Hercher 2022).

Although these restrictions affect anyone who has the ability to become pregnant, the impact of these laws will be felt disproportionately by those who are already disenfranchised, including BIPOC (Black, Indigenous, and People of Color) folks, trans people, and members of other vulnerable groups. In addition to the barriers all people with limited financial means face, people of color with limited means face further obstacles related to their identities. Under these emerging laws, the disparities existing for pregnant people with intersectional marginalized identities are likely to become amplified and to cause even greater harm. Imagine, for instance, the case of a pregnant person with limited financial means whose fetus tests positive for a life-limiting condition. She does not have the robust health insurance, social network, or financial capital to provide adequately for this child's needs. By the time this diagnosis is confirmed, she has passed the window of legal abortion in her home state, and she does not have the resources to travel out of state for this procedure. In cases like this one,

the inability to access abortion can make it even harder to break free of the cycle of poverty. Similarly, disparities in access to care and pregnancy outcomes are part of the reason why abortion is actually safer than pregnancy and childbirth, especially when considering people from Black, Indigenous, and Latin American communities (Goodwin 2020). Furthermore, even if the pregnancy ends with safe childbirth, the social safety net that exists to lift up families in need with children is dismal, with huge failings in domains including paid family leave, expansion of Medicaid, and minimum wage levels in the very states that have banned or are most likely to ban abortion (Badger et al. 2022). These are also the states in which we would expect to see a post-*Dobbs* increase in births of children with complex medical needs related to genetic diagnoses due to the inability to use prenatal care to inform pregnancy continuation decisions. Thus, the realities of intensive medical care for these children will disproportionately fall on people of color in states with limited social support from the government for families in need. In these states hostile to abortion, the measures of children in poverty, uninsured women and children, low birth weight babies, teen births, and infant and gestational carrier mortality already far outstrip those same measures in the states where abortion is likely to remain accessible. These laws prevent families from getting the help and support they need and will certainly continue to have a ripple effect on society as the effects of systemic racism and bias reverberate through their lives.

At its core, abortion access is a social justice issue (Kozhimannil, Hassan, and Hardeman 2022). Providers are increasingly experiencing moral injury when they can't adequately inform and protect their patients. They find themselves letting patients get sicker and sicker, wondering whether their patient's life is in sufficient jeopardy to justify an abortion under existing laws (Simmons-Duffin 2022). Professionals are now asking, "When these laws directly and immediately threaten the health of patients, should physicians collectively disobey them—that is, should they engage in professional civil disobedience?" (Wynia 2022). Pregnant people and their families are suffering (Grossman et al. 2023), and providers cannot deliver the care they were trained to deliver under these conditions. Doctors, nurses, and other health professionals are leaving states with these restrictions, further exacerbating issues of access for pregnant people in states hostile to abortion (Meek 2023; González-Ramírez

2023). For all these reasons, many professional societies, including the American Medical Association, the American College of Medical Genetics and Genomics, the National Society of Genetic Counselors, the American Society of Human Genetics, and the American College of Obstetrics and Gynecology (ACMG Board of Directors 2020; ASHG Board of Directors 2020; Hoskins 2022; Anderson 2022; Resneck 2022), have spoken out about the myriad harms that are already resulting from increasing restrictions on abortion care.

The intimate and complex relationship between biology, pregnancy, and bias also comes to bear when considering disability. Access to abortion is an important aspect of health care for people living with disabilities (Ducharme 2023). The medical risks associated with pregnancy for some folks with disabilities can be life threatening, and limiting their access to abortion can be especially dangerous. Furthermore, many of the genetic conditions for which prenatal testing is offered include some aspect of physical or intellectual disability. In the next chapter, we will explore the social and biological landscape of disability in depth to consider the impact of structural ableism.

7 Disability

Disability is often a matter of perspective, timing, and context. Disability can impact a range of activities that involve mobility, cognition, sensory perception, social/emotional engagement, and self-care. Likewise, numerous factors can cause disabilities, including accidents, war and violence, behavioral health forces, congenital conditions (ones that are present at birth), COVID-19 and other pathogens, substance use and other exposures, self-injurious behaviors, and pregnancy. In terms of disabilities with biological roots, while there are genetic syndromes associated with disabilities, there are also disabilities acquired later in life that are frequently associated with the natural course of aging. The dimensions of disability can exist anywhere on the continuum from visible to invisible, acquired to inherited, and physical to intellectual. Any one individual's experience with disability is unique and dramatically impacted by the way these variables combine and interact with the social and environmental contexts of their own lived experience.

The term *disability* itself encompasses such a wide array of differences that most people will experience some form of disability at one point in their life or another. In fact, the CDC estimates that 26 percent of adults in the United States live with a disability (Centers for Disease Control and

Prevention 2023), and the World Health Organization has found that 16 percent of the world's population (1.3 *billion* people) lives with a disability (World Health Organization 2023). Perhaps as we begin to understand long COVID in the aftermath of the greatest disabling force in modern times, we may find that these already high numbers are actually underestimates. Clearly, this is not a niche topic that only applies to “rare” genetic diseases, but by focusing on genetic and congenital conditions, we can explore how disabilities impact people throughout the life cycle. Let's begin with an exploration of the origins of developmental conditions that can lead to disabilities.

Human development is an awe-inspiring process. From a single cell with a single genome, over 200 unique cell types develop and grow into a human body estimated to have 37 trillion cells (Kramer 2015). The careful orchestration of cell fate decisions, cellular migration to form tissues and organs, and growth of the body requires activation of genes at the right time, in the right place, and in the right dosage. The fact that these innumerable individual decisions occur in concert to pattern a human from a single cell is a truly remarkable thing, miraculous even. But the fact of the matter is that occasionally the course of events isn't entirely predictable in the programs it pursues. Maybe a developing fetus was exposed to a harmful chemical or virus that interrupts fetal development of a particular anatomical structure, or maybe there was a variant in a gene that caused the resulting protein to be active when it should have been silent. In a process so intricate and strategic, any number of seemingly minor changes can have a significant impact on the ability of the fetus to develop in a typical fashion.

My choice of the word *typical* above is entirely deliberate. To recognize disability as a form of diversity, we must take care to use inclusive language that doesn't normalize one state while labeling any variance from this state as defective or abnormal. Importantly, the very concept of disability centers around who gets to define what is considered “normal.” For these reasons, it is important to bear in mind the impact of words and how these frames of reference impact our views of diversity in this realm (Cho et al. 2022). Disability advocates have increasingly called for a move from a medical model to a social model of disability (Pulrang 2021); the medical model emphasizes the biological limitations of one's body, while the

social model emphasizes the limitations on inclusivity in society. It would be an oversimplification to say that either model alone accounts for all aspects of disability, but consider these models in the context of accessibility. If an individual is a wheelchair user, and they want to enter a historic building that is only equipped with stairs, then the medical model would say that the individual's disability is preventing them from entering the building, while the social model would say that the building is not accessible and is a disabling environment.

Through the 1970s, disabling environments were prevalent throughout the United States. In those years, it was entirely commonplace to institutionalize individuals with disabilities, and society did not make any effort to accommodate people with disabilities in common spaces. Places like the Willowbrook State School on Staten Island housed folks with developmental disabilities throughout their lives. Even though it was named as a school, it was little more than a warehouse, and in this environment people were left unattended, filthy, overcrowded, undernourished, and abused (Weiser 2020). The horrific conditions of this school were brought to light in a graphic 1972 exposé, when reporter Geraldo Rivera brought television cameras into the Willowbrook facility ("The Story That Revealed Willowbrook's Horrors" 2016). These events were also occurring during a chapter of American history that included the early days of the Civil Rights Movement and the women's liberation movement. These examples of social activism paved the way for people like Judith Heumann and her contemporaries to begin advocating for change on behalf of the disability community (LeBrecht and Newnham 2020). They advocated for a world in which people with disabilities could just be people. Through their efforts and those of many partners, the Americans with Disabilities Act was passed in 1990, which finally codified the civil rights of people with disabilities, regulated requirements for employers to provide reasonable accommodations to employees with disabilities, and created accessibility requirements for public spaces. However, like all other aspects of civil rights, this is a journey that is still in progress, with documented abuses that have reverberations of Willowbrook continuing in group homes for folks with disabilities as recently as 2020 (Weiser 2020).

In order to fully understand the social context for people with disabilities, it helps to begin with the biological basis of disability. To realize how

places like Willowbrook came to be, it is important to conceptualize the decisions families who have children with disabilities are facing. Did they have an idea that their child might be born with a developmental difference? Was their child the first person like them in their family? Did they feel prepared to raise their child? What support existed to help them when they were in unfamiliar territory? How is disability viewed by their culture and community? All these questions grow from understanding how families experience disabilities, so that is where we will turn our attention next.



The biological origins of disability include exposure to teratogens (harmful chemicals) during critical periods of development, random changes in chromosome structure or number, and conditions that result from specific variants in developmental genes. We will consider multiple examples of disability and how different aspects of lived experience in each of these cases influence interactions with health care providers and broader society. Furthermore, although each disability has its own unique sets of lived experiences, values, and cultural norms, these communities are often lumped together into a single group, even though disability is far from a monolith. This oversimplification can cause harm, given the unique needs of each community. It is also important to recognize that one of the key issues raised in the context of disability is abortion. While some anti-choice rhetoric tries to pit disability advocacy against pro-choice beliefs, the disability community itself does not subscribe to that view because abortion is often a life-saving form of health care for them as well (Ciesemier 2022).

The case of thalidomide is one that highlights the connections between disability and abortion access. Thalidomide is a sedative that was prescribed as an anti-nausea medication in the late 1950s and early 1960s. Clinical research that preceded its approval in other countries was based largely on anecdotal data, and the drug was not adequately tested in pregnant people (McNeill 2017). Although the medication was not formally approved in the United States, pregnant people in other countries quickly discovered that when it was taken to treat morning sickness during certain vulnerable points of pregnancy, their children experienced significant

effects on their developing limbs, with severely shortened or absent limbs. In the United States, Sherri Finkbine, a children's television personality from Phoenix, took a sleeping pill her husband purchased over the counter in Europe during her fifth pregnancy in 1962. She then discovered that this pill contained thalidomide, and when she realized this was potentially harmful to her developing baby, she arranged to have an abortion in Arizona (Brennan 2018; Iorfida 2022). At that time, abortion was illegal in the United States unless the mother's life was in danger, but nothing about the fetus's life was described in these guidelines. Although her local hospital originally agreed to perform the abortion, after Finkbine spoke publicly about her plans in order to warn other prospective parents, the hospital reversed its decision. Ultimately, she had to travel to Sweden to have her abortion, underscoring the pre-*Roe* (and post-*Dobbs*) reality that legislation doesn't prevent people from having abortions when they have financial resources. Indeed, Finkbine's case was one that propelled a groundswell of support for *Roe v. Wade* in the days that followed and ultimately led to the legalization of abortion in the United States . . . for a time.

Pregnancy-related decisions necessitate access to accurate information about quality of life for people living with disabilities. When a person or couple finds out that their pregnancy is at risk for developing a disability, their questions are generally not about the biological mechanism, but are instead about things like whether their child will be able to go to school, play with friends, fall in love, and live on their own. To visualize the future for their child, providers must facilitate patient-centered conversations that provide them the information they seek and support the decisions they come to. The providers must be both knowledgeable and able to set aside their own opinions on what the appropriate course of action may be. While we might expect the overlap between genetic diagnoses and the development of physical or intellectual disabilities to lead to well-informed specialists in genetics and medicine, there remains significant bias among scientists and health care providers against people with disabilities. James Watson, one of the codiscoverers of the molecular structure of DNA, even said, "Seeing the bright side of being handicapped is like praising the virtues of extreme poverty" (Miller and Levine 2013). There is no room for misunderstanding the biased intent of Watson's words; his view of the

inferiority of persons with disabilities is clear, and when expressed by a leader in the field, is especially harmful.

Bias isn't always that explicit. Sometimes it emerges from a lack of representation, which leads to a gap in understanding the lived experiences of folks with disabilities. For instance, knowing that 27 percent of US adults have a disability, we see a drop-off in representation among people with doctorates in science, engineering, and health (10%) and among National Institutes of Health (NIH)-funded researchers (2%) (Swenor, Iezzoni, and Barnett 2023). Because the NIH is the most prominent domestic funder of biomedical research, this is worrisome and perhaps partially explains the ableist language in its mission statement (Gewin 2023). Specifically, the mission statement includes reference to the concept of "reducing disability" as a way of promoting health, but this language endorses the idea that disabilities are deficits, flaws requiring fixing. Indeed, it embraces a view of disability as an illness, which heavily utilizes the medical framing over a social framing of disability. An alternative mission statement suggested by the NIH Advisory Committee to the Director Working Group on Diversity is "to seek fundamental knowledge about the nature and behavior of living systems and the application of that knowledge to enhance health, lengthen life, and reduce illness" (Diaz Brinton and Bernard 2022; Iezzoni and Swenor 2023), a revised framing that doesn't automatically equate disability with illness. The group also recommends establishing an NIH Office of Disability Research, including disability advocacy in its DEIA (diversity, equity, inclusion, and accessibility) efforts, developing an NIH-wide effort to identify and address structural ableism, expanding efforts to include disability communities, formally designating folks with disabilities as a health disparity population, and collecting demographic data on disability in all NIH systems. In September 2023, the NIH took steps toward implementing these recommendations by formally designating the disability community as a population with health disparities (National Institutes of Health, National Institute on Minority Health and Health Disparities 2023). This was a hard-won battle for the community and its allies (Swenor and Kennedy 2023); the health disparity designation is significant because it encourages research focused on people living with disability in part by allocating resources for these studies, and thus can ultimately help improve access to health care as well as overall health.

Together, these measures can begin to right the imbalance and underrepresentation experienced by disabled communities.

Similar lack of disability representation is seen among health care providers, with only 17 percent of genetic counselors (National Society of Genetic Counselors 2022) or 3 percent of physicians (Nouri et al. 2021) self-identifying as having a disability. It is likely that this poor representation has directly contributed to a lack of empathy for and engagement with disabled patients, leaving many patients facing a search for a provider willing to see them (Lagu et al. 2022; Kolata 2022). The biases contributing to the experiences of folks with disabilities are a mixture of individual biases as well as systemic ones, including poor reimbursement from payers, loss of money on interpreters, and time pressure to complete appointments in very brief encounters, to name a few. Of note, the researchers who conducted these studies include scholars with mobility constraints and low vision, further emphasizing the importance of inclusivity in research to ensure these types of questions are studied.

Given that the evidence of biases among scientists and providers is robust, it begs the question of whether the motivations behind providing prenatal testing and opportunities for pregnancy termination for certain genetic conditions are rooted in a belief that individuals with disabilities are somehow defective. To study this, a team of researchers (including me) created a free educational module (Dasgupta, Stevens, and Sabatello 2020) that examines a participant's implicit biases toward individuals with disabilities and measures how likely they are to recommend prenatal genetic testing in hypothetical clinical scenarios where the patient's child was at risk of developing a disability. We targeted our studies to health care professionals and trainees, and in this population, we confirmed the existence of biases, determined that personal opinions can impact genetic counseling recommendations for testing, and found that with increased training the providers can improve in centering patient wishes (Vaimberg et al. 2021; Jungels et al. 2023). We also noted that previous professional experience with folks with disabilities reduced the influence of bias on clinical recommendations, providing further rationale for the importance of representation as well as service learning in these professions. These findings were encouraging to us because they suggested that there are effective interventions that are fairly straightforward in mitigating the

impact of disability bias. Unfortunately, the current state of medical education with respect to topics relevant to disability remains inadequate, but perhaps with growing evidence of the efficacy of these approaches, we can begin to integrate these topics more widely in medical education (Ratakonda et al. 2022).



Just as it is important for future health care providers to understand the lived experiences of folks with disabilities, it is important for all of us to be able to walk a mile in each other's shoes. In the following sections, I want to do just that, focusing on two case examples, dwarfism and deafness. I chose these two examples because they help us understand the social contextualization of disability in cases where accommodations are needed to engage with a world designed for people of typical height and for hearing people. Importantly, in both cases, there is no medical impact on lifespan, but there are still medicalized views of these disabilities where people seek to "fix" people of short stature or deaf people even though they can certainly live rich and meaningful lives without being "fixed." Furthermore, both communities have fostered their own vibrant culture that also informs how community members engage with health care professionals and how they make decisions about their lives.

You may have noticed that both of these cases fall into the realm of physical disabilities rather than intellectual or social/emotional disabilities. We will be returning to a full discussion of Down syndrome in chapter 9, when we discuss how individual decisions can collectively impact the existence of a whole community of individuals with intellectual disability. There are certainly other conditions associated with intellectual disability or neurodiversity with genetic underpinnings, but in many cases the genetic basis is poorly understood, as is true with autism. Importantly, even in the absence of highly predictive genetic signatures and clinical genetic testing for autism, it is notable that both the autistic community and Deaf community are less inclined to provide personal information for precision medicine genetic research or to receive the results of the research, findings that may suggest that they do not feel compelled to find a "cure" for their condition, that they do not subscribe to the medicalized

framework of autism or deafness, or that they are concerned about the potential for these findings to be used against their communities' interests (Sabatello et al. 2020). Thus, if scientists can appropriately partner with autistic individuals to identify genetic variants that are associated with autism in the future, we may expect some similarities between how autistic people and Deaf people choose to use this information in their lives and choices.

In addition, you may be thinking about the connection between chronic disease and disability. Indeed, there are significant overlapping interests between people with chronic illness and people with disabilities. Because the genetic basis of many of these chronic illnesses is not well understood, for the purpose of this discussion, I am focusing on cases in which the biological basis is well described so that we can more clearly disentangle the contributing forces at play.

For now, let us begin with these two cases, and we will circle back to a discussion of intellectual disability when we are able to properly frame it in historical context.



Let's begin our exploration of the social context of living with disability by considering dwarfism, first through the lens of the entertainment industry. While there are over 400 types of dwarfism, they share the common feature of bodies of shorter heights, often topping out at about one foot shorter than average adult height. Because of this difference in height, Little People often found themselves in roles as entertainers, sometimes typecast in very dehumanizing roles. For example, dwarfism was widely misunderstood and stereotyped, particularly in the context of circuses. P. T. Barnum was famous for featuring "freak shows" as part of his traveling show, where he would highlight people of short stature, people with endocrine differences resulting in atypical facial hair (the Bearded Lady and the Dog-Faced Boy), and individuals with other bodily differences, creating a popular spectacle that was intolerant of human difference (Hawkins 2014). The individuals who were part of these traveling shows found themselves trading acceptance and respect for a paycheck and fame. Though it may be tempting to say that this was a relic of the 19th century,

it would be dishonest to omit mention of the actors who played Munchkin villagers in *The Wizard of Oz* in 1939. These actors earned \$50 per week, only 40 percent of what producers paid Toto the dog (\$125), and they never saw their names in the credits (Abramovitch 2016). Furthermore, the Munchkin headhunters were specifically told to eliminate achondroplastic dwarfs and dwarfs of color from casting (Weinstock 2018). The racism at play in excluding dwarfs of color is obvious, but it may be less apparent why they were instructed to exclude dwarfs whose short stature results from achondroplasia. Achondroplasia is the most common cause of dwarfism, and it results in a disproportionate growth, where limbs would grow to be shorter than would be proportional with torso length. They were distinguishing between proportional and disproportional Little People, and with these policies, they were creating a hierarchy within a hierarchy. Since that time, many people of short stature with careers in entertainment continue to be caricatures in costume; think of the original Oompa Loompas, Ewoks, Austin Powers's Mini-Me, and the goblin bankers at Gringotts. While these roles didn't universally exploit people of short stature, they also didn't feature their talents, instead centering their height as the main characteristic or quality they brought to the table. Peter Dinklage, for his storied acting career, including his role as Tyrion in *Game of Thrones*, is the exception rather than the rule. At the same time, the dwarf community would like to retain the right to be cast in roles featuring people of short stature, so long as they do not portray them as parodies of average-height humans. Some dwarf actors have said they don't have the success that Peter Dinklage has, which allows him to be more choosy about which roles he takes. Seeing Hugh Grant, someone of typical height, cast as an Oompa Loompa in the *Willy Wonka* remake feels like a loss for the community in a different way (Kingsberry and Charles 2023). Just as it is not appropriate to cast a White actor in blackface to fill the role of a Black character, the dwarf community feels the same about being replaced by typical-height actors in portraying people of short stature.

Achondroplasia is a genetic condition resulting from a gain-of-function variant in the fibroblast growth factor receptor 3 gene. This genetic change causes the receptor to send a signal that inhibits proliferation of a particular type of cell (chondrocyte) in the bone, thereby reducing growth of the

	A	a
a	Aa	aa
a	Aa	aa

Figure 5. This Punnett square predicts the possible children resulting from a couple where one parent is of short stature (Aa) and one parent is of typical stature (aa), with “A” referring to the dominant achondroplasia allele and “a” referring to the typical height allele of FGFR3. The top row indicates the possible gametes from the parent of short stature, demonstrating that parent has a 50% chance of passing on the achondroplasia allele. The left column shows the possible gametes from the typical height parent. The cells at the junction of the left column and top row show the predicted combinations in their children. The Aa genotype combination is a child of short stature, and aa is a child of typical height; therefore, this couple has a 50% chance of having a child of short stature and a 50% chance of having a child of typical height.

long bones in the limbs. This can also lead to spinal cord compression and delayed motor development for physical reasons, not reasons related to intellectual development. Because the inhibitory signal is a gain-of-function change, one copy of the variant is enough to contribute to slowed growth, meaning it can be passed on in an autosomal dominant fashion. That is to say, a parent of short stature with a typical-height reproductive partner has a 50 percent chance of having a child with short stature and a 50 percent chance of having a typical-height child (see figure 5).

In the case where there are two parents of short stature (with the same form of dominantly inherited dwarfism, such as achondroplasia), the outcomes vary somewhat and carry some additional genetic risk (see figure 6).

Finally, even though short stature can be inherited from one generation to the next, the variant occurs at a high frequency due to new mutation. The significance of this observation is that over 80 percent of children with dwarfism have typical-height parents and siblings.

Each of these described family structures has a huge impact on how the family chooses to raise their children. In the case of new mutation, parents with no lived experience with short stature are making decisions about how to best accommodate their child’s height. The inverse is true in families where one or both parents are of short stature and find themselves

	A	a
A	AA	Aa
a	Aa	aa

Figure 6. This Punnett square predicts the possible children resulting from a couple where both parents have achondroplasia (Aa), with “A” referring to the dominant achondroplasia allele and “a” referring to the typical height allele of FGFR3. The top row and left column indicate the possible gametes from each parent of short stature, demonstrating that they each have a 50% chance of passing on the achondroplasia allele. The cells at the junction of the left column and top row show the predicted combinations in their children. The Aa genotype combination is a child of short stature like the parents, and aa is a child of typical height; therefore, this couple has a 50% chance of having a child of short stature and a 25% chance of having a child of typical height. However, the remaining 25% of the time the AA combination carries with it serious health concerns that are typically life limiting and result in premature death, either prenatally or neonatally.

raising a child of typical height. And in the case where a parent is of short stature, they may prefer to have a child of similar height to themselves. Consider the possibility of being the gestational parent who happens to be of short stature. It would be physically challenging to carry a pregnancy for a baby of typical height. Also, once the baby is born, if both parents are of short stature, the home may be customized for people of their height, with lower counters, sinks, toilets, shelves, pedal extensions on cars, and so on. Introducing a child of typical height into this physical environment might eventually require significant, costly alterations of their living spaces to accommodate that child.

When it comes to achondroplasia, there is debate within the community of people of short stature about whether the term *disability* applies to them. This is partly to do with the medical versus social construction of disability that we previously discussed. Through a social lens, the emphasis would be more on the accommodations to living and working spaces rather than on the medical view. It may also reflect lived experience in that a person who experiences more chronic pain, sleep apnea, or self-care complications related to the length of their limbs may find the condition more disabling than someone for whom these aspects of achondroplasia

do not weigh so heavily. It is worth mentioning, however, that dwarfism is recognized under the Americans with Disabilities Act, and in that context, the term *disability* is empowering because it unlocks access to accommodations allowing people of short stature to interact with the larger world.

While there is not a consensus among people of short stature about if this is a disability, there is a shared understanding of appropriate language. Referring to someone with dwarfism as a person of short stature, a dwarf, or a Little Person (a term claimed and coined by the community) are all acceptable. What is not appropriate is the term *midget*. The origin of this word is in reference to a small fly named the midge. Comparing Little People to small flies that are by all accounts a nuisance is insulting and degrading. Furthermore, the term *midget* is used in the context of dehumanizing activities that equate people of short stature with a sideshow, for example, “midget tossing” events, as depicted against the wishes of the community in the film *The Wolf of Wall Street*. The word used to have a specific meaning in the community, so you may come across historical documents contrasting midgets (people of proportional short stature) and dwarfs (people of disproportional short stature), but because of the root of the word *midget* and the attempt to create further inequity within the group, the term *midget* is now considered the “m word” and should not be used. Instead, dwarf or any of the other appropriate terms can be used to describe a person of short stature, whether or not they have proportional limbs.

Importantly, the culture created around people with dwarfism is increasingly one of “Dwarf Pride,” which influences how people of short stature interact with the medical community. In the context of the hypothetical family mentioned previously with two parents of short stature, we recognized that for a variety of reasons they may prefer to have a child of short stature like themselves (Stramondo 2017). There also are potential medical complications for the gestational parent with a pregnancy for a child of typical height or for a child who inherits two copies of the achondroplasia allele. Because the gene has been identified that causes achondroplasia, this family may wish to undergo prenatal or preimplantation genetic testing to select *for* a child of short stature. For many people of typical height, this application of genetic testing may not occur to them at first, but it is a potential application of genetic testing among people of

short stature. Depending on one's reaction to this, it may rightly cause one to question one's biases on which outcomes we consider "healthy" or "normal."

Until recently, treatment for people of short stature to try to increase height has been a very invasive process. Some people of short stature, more so in Europe than in the United States, have opted to go through limb-lengthening surgeries. These surgeries involve cutting bone, attaching a lengthening device to the limb, and gradually separating the bone at the cut junction to stimulate new bone growth to fill in the gap. This procedure occurs over a time span of months during childhood while bone growth is actively occurring, and it can result in a gain of about two inches of growth per lengthening cycle. The patient may experience pain in the first few weeks, but it is reported to diminish during the lengthening process. The nature of this procedure has likely been a big deterrent for its greater uptake in the United States, but with the advent of a new intervention, a drug that can be injected once daily to release the inhibition on chondrocyte proliferation, the question of whether or not to medically intervene to change one's growth potential to grow closer to the average-height adult (Saner 2020; Solomon 2020) is being reconsidered. There are so many layers to this question . . . first, as with the bone-lengthening surgeries, the injections must be administered during childhood while bones are actively growing. When a minor is being cared for, the decision is heavily influenced by the parents or guardians, which in this case carries several important perspectives to consider. For instance, for the 80 percent of people with dwarfism born to parents of typical height, do the parents have full understanding about whether this is something that would ultimately benefit the child? Could it just result in a person who doesn't quite fit in with the social community of people of short stature, nor with people of typical stature? What message is sent to the child if they feel their height is problematic and medicalized? Will the child have the maturity, sense of self, and agency to be able to contribute to this significant question about their body? Does reducing the incidence of dwarfism further make it harder for people who opt not to go through the intervention to receive accommodations? These questions and many more take me right back to the concerns about ableism in the NIH mission statement; these interventions have been developed in the name of

reducing disability, but it is unclear how widely the community will embrace this option.



Our next case for consideration is that of deafness. Newborn screening routinely includes testing for a baby's ability to hear. This is not the test that you might remember from the school nurse's office, where we would have to raise our right or left hand depending on which side we heard the auditory cue in the headphones. Instead, there are technologies that allow measurement of the brain's or the ear's response to sound, and these tests are frequently conducted in the context of a hospital shortly after birth. If a child does not pass this first test, they may need follow-up testing, including an audiology evaluation, testing for pathogens that can lead to hearing loss, or genetic testing. The rationale behind these early tests is, as with all other newborn screens, to maximize the possibility for early intervention for children who are found to be deaf or hard of hearing. The type of intervention that works best for each family is personal and reflects their approach to promoting language acquisition—including sign language—for the child.

As with other forms of disability that we have discussed, deafness can be viewed through a social or medicalized lens, and this influences which terms are embraced by the community (National Association of the Deaf 2023). Generally speaking, the term *deaf* refers to the audiological condition of profound hearing loss, and capitalized Deaf embraces not only the impact on one's hearing but also the culture of the Deaf community, including shared language and cultural norms. People who identify as Deaf are immersed in Deaf culture and community, in some cases born with hearing loss and learning American Sign Language (ASL) as their L1 or heritage language from parents and caregivers who passed on the shared culture, but people who lost hearing postnatally can also identify as Deaf culturally. The term *hard of hearing* generally refers to people with mild to moderate hearing loss. The lived experience of someone who is hard of hearing is complex because there are some elements that are similar to both the hearing and the Deaf communities; some people who are hard of hearing may view themselves as culturally Deaf, whereas

	A	a
A	AA	Aa
a	Aa	aa

Figure 7. This Punnett square predicts the possible children resulting from a couple where both hearing parents are asymptomatic carriers of a recessive deaf allele (Aa), with “A” referring to the dominant hearing allele and “a” referring to the recessive deaf allele. The top row and left column indicate the possible gametes from heterozygous carrier hearing parents, demonstrating that they each have a 50% chance of passing on the deaf allele. The cells at the junction of the left column and top row show the predicted combinations in their children. The Aa genotype combination is a hearing carrier of the deaf allele like the parents, and aa is a deaf child; therefore, this couple has a 25% chance of having a child with hearing loss, even though the parents themselves are able to hear.

others live entirely in the hearing world with various assistive devices. Folks who are hard of hearing or who acquired hearing loss with age have a different set of experiences, and because of their ability to hear to varying extents, may opt to use different accommodations, such as lip reading or hearing aids. These choices influence whether people who are hard of hearing view themselves as culturally Deaf, allies, or their own community. It is probably self-explanatory that historical terms like *deaf-mute* and *deaf and dumb* are offensive; importantly, the prevailing idea of the time that deaf people were incapable of complex thought was actually a reflection of the fact that they were being deprived of access to language and accessible communication. What may be less obvious is concern with the term *hearing-impaired*. Most in the community no longer accept the term *hearing-impaired* because it emphasizes what people *can’t* do, implying a deficit in their abilities, and relies on the medicalized view of their hearing loss. For these reasons, identity among people with hearing loss is a very personal thing, and the best way to honor their lived experiences with empowering terminology is to ask how they uniquely identify.

As alluded to above, deafness can be both inherited and acquired. The two primary forms of heritable deafness are syndromic and nonsyndromic deafness. Syndromic deafness results when a genetic change is responsible for a constellation of biological affects, including deafness, whereas non-

	a	a
a	aa	aa
a	aa	aa

Figure 8. This Punnett square predicts the possible children resulting from a couple where both parents are deaf (aa), with “a” referring to the recessive deaf allele. The top row and left column indicate the possible gametes from the parents, demonstrating that they each have a 100% chance of passing on the deaf allele, because in order to be deaf themselves, they must be homozygous recessive. As in the previous figures, the cells at the junction of the left column and top row show the predicted combinations in their children, with all combinations mirroring the parents’ genotype and leading to deafness in the child.

syndromic deafness is when a genetic change results in deafness on its own. Most genetic forms of deafness are recessive, meaning that both parents must contribute a deaf allele in the same gene to have a deaf child (see figure 7).

In the case where there are two deaf parents with the *same* form of recessively inherited hearing loss, we would predict that *all* their offspring would also be deaf (see figure 8). This is the family structure in which we would often expect to see the child raised in the Deaf culture, which would also be supported by early exposure to accessible language through sign language instruction.

In the case where there are two deaf parents with *different* forms of recessively inherited hearing loss, we would predict that *none* of their offspring would be deaf (see figure 9). This can occur because even though most forms of heritable deafness are recessive, if each parent has a different causative gene, then their children will have functional copies of each gene from the opposite parent. Because there are over 50 genes identified as being involved in hearing, this is a very plausible outcome when two deaf parents have a child. This hearing child is referred to as a child of deaf adults (CODA), a community term embraced in the Apple TV film *CODA*, starring Oscar-winning Deaf actors Marlee Matlin and Troy Kotsur as the parents.

Although 50 percent of deafness is genetic, over 90 percent of children born deaf have hearing parents and over 90 percent of deaf parents have

	aB	aB
Ab	AaBb	AaBb
Ab	AaBb	AaBb

Figure 9. This Punnett square predicts the possible children resulting from a couple where both parents are deaf (AAbb and aaBB) as a result of deafness variants in different genes, with “a” referring to the recessive deaf allele of gene A and “b” referring to the recessive deaf allele of gene B. Two copies of the same deaf allele lead to hearing loss, so each parent is deaf as a result of their homozygosity for either allele a or b. The top row and left column indicate the possible gametes from each parent, demonstrating that they each have a 100% chance of passing on their deaf allele together with a hearing allele of the other gene. As in the previous figures, the cells at the junction of the left column and top row show the predicted combinations in their children, with all combinations leading to hearing in the children, who have one hearing allele for both genes A and B, which is enough to allow hearing to develop.

hearing children (Mitchell and Karchmer 2004). The cultural implications of these various family structures are significant, particularly when the family has both hearing and deaf family members. When all family members are deaf, embracing Deaf culture is part of family life, but in the case where deaf children are born to hearing parents, many times families initially pursue a medical model of treatment, perhaps exploring the use of hearing aids or cochlear implants and enrolling the child in a hearing school, for example. It is very rare for hearing parents to simultaneously introduce sign language alongside their native spoken language. They may begin to connect to Deaf culture through Deaf social groups, early intervention programs, ASL classes, and eventually Deaf churches and Deaf schools like Gallaudet.

The intersection of deafness, genetics, and medicine raises a number of profound questions about how to plan for and raise children. When it comes to Deaf parents, for several cultural reasons, it may be preferable to ensure that their children are also Deaf. Preconception genetic testing can help distinguish between deaf parents that are likely to have deaf children (figure 8) and deaf parents that are likely to have hearing children (figure 9). With the resulting knowledge, the prospective parents, if they are likely to have hearing children, may opt to conceive through gamete donation or

to adopt a deaf child, though given the expense and logistical complexity of these approaches, it isn't always the first choice of prospective parents who can otherwise conceive without medical intervention. In one particular case, Sharon Duchesneau and Candace McCullough, two Deaf mothers who met while pursuing higher education at Gallaudet, were planning for their future child and inquired with the sperm bank about whether they could provide an anonymous deaf donor (Mundy 2002). The bank said no, that deafness is the type of condition that results in a donor being eliminated, so the women decided to ask a deaf friend to serve as a known donor. Going into it, they didn't know if Sharon and the donor had the same form of deafness, and based on family histories, the genetic counselor estimated the chance of their having a deaf baby at 50 percent. Because baby Gauvin was born at home, he was not screened for hearing in the hospital. They had to wait a couple of months for the full audiology evaluation to find out if their child was hearing or deaf. As Sharon put it, "A hearing baby would be a blessing. A deaf baby would be a special blessing." It turned out that Gauvin was mostly deaf, and his parents opted to go without hearing aids. Hearing aids and cochlear implants are sometimes viewed by members of the community as threats to the Deaf culture and ASL, though many in the hearing community have trouble understanding this perspective and the desire for children who are born deaf (Sanghavi 2006). At its root, though, this lack of empathy is an entrenched position of ableism, and to provide the best possible care, a patient-centered approach must be taken. The focus on patient-centered care applies differentially within the deaf community as well, as CODAs can struggle with their identity, and deaf children can perceive a reduced social status if they are using assistive devices to augment hearing or are not fully immersed in Deaf culture. The very definition of patient-centered care acknowledges that there is no one-size-fits-all approach.

With an understanding of the genetic basis of a given form of deafness, scientists are now beginning to create the tools needed to provide corrective gene therapy to restore hearing in folks who are deaf (Mullin 2023). The advent of this technology raises many questions about the choices people will make for their children and as adults when they are making their own health care decisions. The typical processes of neurological development suggest that timing of interventions designed to improve or

restore hearing are best carried out in the very young, often before age three, so parents will be making these complex decisions on behalf of their children. Importantly, early intervention need not be medicalized, as programs to improve access to sign language support for the families and the young person, social support groups for families, and other early interventions can also provide meaningful results. Undoubtedly, all these choices will be influenced by one's own identity as hearing, deaf, or Deaf, and when parent and child are members of different communities, how can one know they are making the right choice for their child?



Before we leave the topic of disability, we must circle back to the subject of reproductive freedom. These topics are intersectional because abortion is an essential form of health care, particularly for folks living with some disabilities that make pregnancy a life-threatening condition. Some anti-choice proponents have tried to co-opt disability advocacy to further their anti-choice rhetoric, but the disability community has staunchly distanced themselves from this position (Ciesemier 2022). The rhetoric is designed to use faux disability advocacy to win political points in favor of their restrictive views on abortion without actually supporting the medical or social needs of folks with disabilities or chronic conditions. Truly being prodisability means supporting families not just through the abortion decision point but also after birth by offering appropriate social supports to help families navigate complex social and medical needs. Unfortunately, it is the case that in states with the strictest restrictions or even outright bans on reproductive autonomy, the state does not step up to its responsibility to aid families now caring for the additional children who will be born. When we consider that some fraction of these pregnancies would have been terminated after prenatal testing, we can predict that these states will also have higher numbers of disabled persons. Some states are attempting to carve out exemptions to abortion bans for specific genetic conditions, but this process is riddled with bias in terms of who is determining what conditions are considered medical justification for an abortion, often without the input of the disability community. To this point, CDC estimates demonstrate an existing overrepresentation of people liv-

ing with disabilities in the South, where we also see legal threats to reproductive autonomy and some of the poorest measures of maternal mortality, infant mortality, and child poverty in the nation (Badger et al. 2022).



Over the last few chapters, we have considered numerous ways in which our understanding of genetics informs the ways in which our bodies develop, and we explored the boundary between biological traits and identity. However, there are numerous traits that are erroneously ascribed exclusively biological origins due to a combination of genetic essentialist thinking, multifactorial causation, and bias. In the next chapter, we will explore where biological explanations are oversimplifications and where we must delve deeper into systemic structures that reinforce societal hierarchies. Without integrating this knowledge into our views on human variation, we risk exacerbating disparities. An understanding of the true causes of inequity is essential to move in the direction of anti-oppression.

8 What Genetics Doesn't Fully Explain

SOCIOBEHAVIORAL GENETICS AND SOCIETY

What do our genes say about us? Depending on scientific versus romantic tendencies, the concept of what is encoded in our DNA falls somewhere between a simple blueprint for our bodies to the very essence of who we are. The reality is that it's never that simple, and in this chapter, we will explore what is known about the genetic basis of human traits. Let's begin with a little background on how nature and nurture combine to influence outcomes in our bodies, health, and behaviors.

When it comes to genetics, there are a range of potential combinations of factors influencing traits. In figure 10, you can see that some conditions are caused primarily by one gene (monogenic or single gene disorders) with minor influence from other weakly contributing genetic factors, but these conditions are relatively rare. As we move to the right in the figure, the number of genes contributing to a condition increases (oligogenic conditions, with involvement of a few genes, and polygenic conditions, with involvement of many genes), and each individual gene's effect is less impactful. Of note, these conditions are much more common in the population than single gene disorders.

Although this diagram was created to describe human disease, it is also relevant when we think about the influence of genetics on many traits that

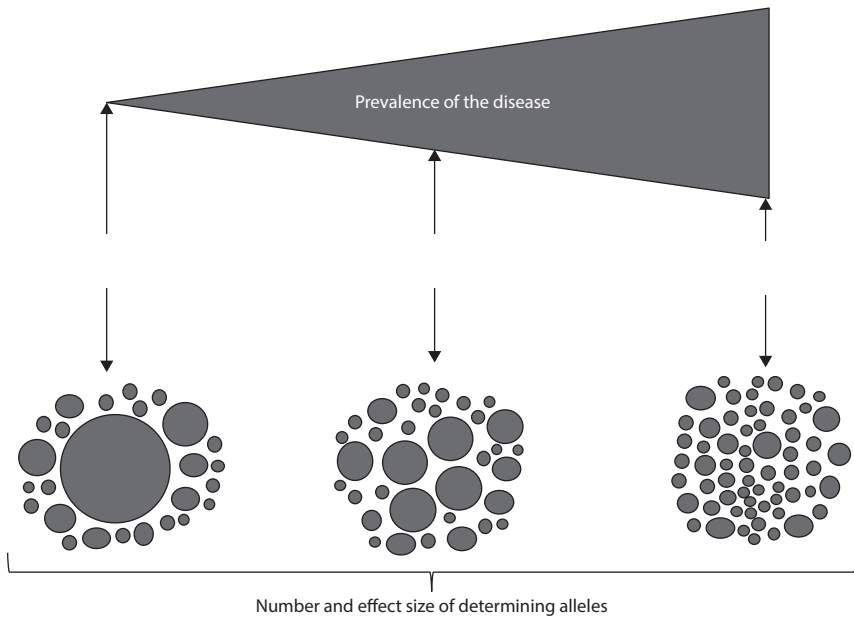


Figure 10. Genetic contributions to human traits. Each circle corresponds to a unique genetic contribution to a disorder. The size of the circle indicates the effect size. The width of the triangle at the top of the diagram indicates how common a condition is. (Image from Marian 2012)

are not related to health concerns, particularly when we think about traits that exist on a spectrum. For instance, skin color can be very fair or very dark and every shade in between. This trait is determined by a collection of genes, and it also reflects some environmental influence related to how much sun exposure a person has had recently.

When the Human Genome Project was completed, the sequence of the genetic code was determined, but the function of many of these sequences remained unknown. That means just sequencing the genome didn't tell us that a particular gene was related to a specific condition. Rather, statisticians and geneticists had to carry out studies to determine which sequences tended to be inherited along with particular conditions. This was easiest to do for conditions with a single strongly predisposing gene, as in figure 10, but as time has passed, people have started to define relationships between weaker contributing genes involved in oligogenic and

polygenic conditions, too. The vast majority of human traits fall into the categories of things influenced by a combination of weakly to moderately contributing genes and environmental effects, but this nuance is often not reflected in headlines declaring that the gene for trait X or behavior Y has been found.

To properly explain the complex interplays between genes and the environment, I want to take a bit of a detour to describe in broad strokes how scientists identify the genes correlated with disease and traits. Moving forward, I will refer to the various manifestations (phenotypes) of genetic variants that are characterized as both medical conditions and human variation by simply referring to them under the umbrella of traits. We will consider how genetic variants travel through generations and interact with environmental factors. Finally, we will look at a few examples where people have attempted to study the genetic basis of complex traits that have roots in social inequalities. I've chosen the examples to highlight how harms can emerge when easily measurable variables are substituted for actual biological outcomes.

In our exploration of the scientific approach to understanding the genetic basis of a complex trait, I will be focusing on Genome Wide Association Studies (GWAS) that have emerged as a way to identify overrepresentation of certain genetic loci in people living with a particular condition or who have a specific trait. Essentially, this approach uses statistics to identify a region of the genome that is present in a larger fraction of people with a trait than would be expected when compared with a control population that does not have the trait. Notably, these statistical associations help scientists zero in on a genetic region of interest, but that region can easily contain dozens of genes, so the direct mechanism, the causal variant that governs the presence of a trait, is not necessarily elucidated by this method. The combination of the Human Genome Project, powerful statistical methods, and collections of populations characterized for various traits have made running GWAS analyses rather straightforward, so they have become incredibly commonplace, with over 64,000 association studies described in the GWAS catalog as of 2024 (European Molecular Biology Laboratory et al. 2024). These associations have then been further analyzed to calculate a polygenic risk score (Petronio, Brockman, and Dron 2021; National Institutes of Health, National

Human Genome Research Institute 2022a), also known as a polygenic index, that describes an individual's overall genetic propensity to develop a particular trait by combining the predisposing and protective genetic signatures into a single risk number. These calculations are low-hanging fruit, particularly when all the information is present in a single platform, such as with consumer genomics companies that collect genetic, health, and trait information from their customers. When these companies share genetic risk with their customers, they are relaying polygenic risk scores that convey an individual's risk relative to the general population for developing a particular condition, and these probabilistic calculations have largely escaped the regulation that is normally associated with health-related products (Smart 2023).

A key limitation of the GWAS approach is what it does not capture. It fundamentally takes a genetic essentialist view, only quantifying genetic risk associations and not considering environmental factors, behavior, lifestyle, structural barriers, or systemic biases. Unfortunately, it is not as simple as just acknowledging that other studies need to be carried out to understand more about the social, environmental, and structural factors that can influence human traits because taking this approach at all carries with it the potential harm of overemphasizing genetic contributions and promoting genetic essentialist thinking. For this reason, scientists have started to think more carefully about their role, particularly in studying and communicating about social and behavioral outcomes through a genetic lens. Some scientists have taken the approach of including a “frequently asked questions” document with their publications (Martschenko et al. 2019), originally conceived of together with the Social Science Genetic Association Consortium as a way to ensure responsible reporting of their findings and first associated with a publication on educational attainment (Rietveld et al. 2013). The authors and other scientists decided on this approach in part because they believe that the work itself may not be dangerous but that its potential misuse certainly can be (“Dangerous Work” 2013). They explain in plain English the answers to some critical questions (Benjamin et al. 2013):

“Did you find ‘the gene’ for educational attainment? No. We did not find ‘the gene’ for educational cognitive function—or anything else. Educational

attainment, like most complex behaviors and outcomes, is influenced by myriad genes, each with effects that are likely to be tiny (as well as a huge host of environmental factors)."

"Does this study show that an individual's level of education is determined at birth? No, and this is probably one of the most common misconceptions about genetic research. Even if it were true—and it is certainly not—that genetic factors accounted for all of the variation across individuals in educational attainment, it would not follow that an individual's education is 'determined' at birth."

"What policy lessons do you draw from this study? None. Any practical response—genetic or environmental, individual or policy-level—to this or similar research would be extremely premature."

The view of the authors is that the potential misuse of findings like these is not a reason to refrain from producing the scientific insight itself. Rather, they indicate one potential benefit of this work is defining a modest upward boundary of what can be explained by genetics so that people can focus on other, perhaps more impactful, influencing factors. They believe that it is best for responsible scientists to lead the way rather than to abstain and hope for the best.

It is also worth pointing out that one of the reasons it was especially important to provide further explanation for this work on educational attainment is the high likelihood of these findings being equated with the discovery of genes related to intelligence. Importantly, these scientists calculated a genetic association with a measurable trait, that is, how many years of schooling one has completed. It is much harder to measure intelligence itself, given that even IQ tests and other tests of cognitive function have documented racial and other biases (Martschenko 2017). With respect to educational attainment, given the localized funding methods for K–12 public schools in the United States, there is a clear link between a family's socioeconomic status, the neighborhood in which they live, and the quality of schools that they can access. This does not even factor in things like ability to pay for private tutoring and college admission consultants. Furthermore, with financial stability comes freedom for the student to focus on learning rather than supporting the family. This is why the aphorism about one's zip code being more predictive than one's genetic code is especially apropos in this situation. At the end of the day, the iden-

tified genetic factors seem to impact educational attainment with very small effect sizes as measured in a few months of schooling, not years.

In terms of a progressive view on these findings, it has been argued that this information can be used to address social inequality and to provide individualized educational strategies for all learners (Lewis-Kraus 2021). What this view glosses over, though, is that educational attainment is affected far more by structural forces than by genetic ones (Meyer, Appelbaum et al. 2023). Generally speaking, sociobehavioral polygenic indexes are poor predictors of individual outcomes (Martschenko and Matthews 2020; Muslimova et al. 2023), and individual predictability of educational attainment from genetic signatures is noisy at best, and racist at worst. Efforts to level the playing field may be more effective if they focused on things like equitable funding for schools by decoupling funding from property values, assessing the impacts of racism on student punishment in schools, and generally studying the structural elements that contribute to educational disparities.

Although I stated earlier that GWAS methodologically does not measure things like the aforementioned structural barriers and systemic biases, they can certainly confound genetic results. As pointed out in an illustrative thought experiment (Coop and Przeworski 2022; Meyer, Appelbaum et al. 2023), if there was a society in which people with red hair were discriminated against and not permitted to attend school, the GWAS result would describe an association between the genetic loci influencing hair color and educational attainment, even though that is actually a social echo rather than a biological connection. For all these reasons, when setting out to study the genetics of particular traits, especially social and behavioral ones, we must consider how the results will be used and whether it makes sense to do this work in the first place.



This question of whether the work should be done is top of mind for me when I think about genetic studies of same-sex sexual behavior. As with the studies on educational attainment, a study on same-sex sexual behavior (Ganna et al. 2019) was accompanied by a robust press release detailing frequently asked questions and providing explainer videos (Broad

Communications 2019). One important addition to this study was a collaboration with the Out@Broad group to ensure representation of the community being studied in study design and in the authorship team. In terms of methods, the data being analyzed was derived from self-reported information concerning sexual behavior as to whether the study participant *ever* had a same-sex partner (Kaiser 2019). It did not look at self-reported sexual orientation or gender identity, and it did not distinguish someone who may have had a single sexual encounter with a person of the same sex from someone who consistently chooses partner(s) of the same sex. It also did not account for people living in environments where same-sex sexual behavior was considered criminal, though most of the participants were from the United Kingdom and the United States. As with the study on educational attainment, this study did not identify a single “gay gene,” nor did genetics even explain much of the behavioral trait. When I read the result that there appears to be some genetic correlation between the behavior characterized in this study and the previously described behavioral trait of risk taking, I started to worry about conclusions that people will draw about the LGBTQ+ community and the potential for these findings to contribute to further stereotyping and marginalization. I worry about how people might view these genetic associations through a deterministic lens to take harmful actions against young people, even though the findings show that you can’t predict one’s sexual behavior from DNA alone. This is certainly not an illness, and it does not require a medical intervention. Perhaps these results actually empower members of the LGBTQ+ community, enabling them to be able to point to scientific evidence and say that this is just part of the natural biological continuum of sexual behavior. It may also free members of the LGBTQ+ community from having to justify their private lives to homophobic people who view their lifestyle as unnatural. Does this then mean that scientists have a moral duty to carry out this work to describe a relationship between genes and the biological continuum of sexual behavior, or is there too much possibility for misinterpretation and harm? And how does this apply to scientists working to identify a genetic basis for gender dysphoria among trans folks? I suppose in my own internal debate on whether or not work like this is, on balance, beneficial, or whether by carrying out this study the scientists are pathologizing the behavior, I must remind myself, as one of

the authors states, “that bigotry needs no data. . . . It’s prejudice, not knowledge, that threatens us” (Phelps and Wedow 2019).

Given the reality that that cat is out of the bag with respect to many publicly available human genome datasets, it is important to recognize that these studies are unlikely to be halted, and guidance on responsible interpretation of these results is critical (Meyer, Appelbaum et al. 2023). To provide a nuanced framework around reporting sensitive genetic associations, the Hastings Center has identified some key limitations that generally bear emphasis for studies in social and behavioral genomics (Martschenko et al. 2019):

- “Genetic influences on behavioral traits are not immutable or deterministic.
- The mechanisms through which genetic variants are associated with social and behavioral outcomes is poorly understood and are not divorced from environmental or social processes.
- Genetic variants identified by GWAS have **miniscule** effects, typically ranging from .01–.03%.
- PGS for complex behavioral traits are not currently useful predictors of individual outcomes—i.e., polygenic scores are not ‘fortune-tellers.’
- The results of genomic studies listed [in their compendium] do not provide the genetic bases of ancestral, racial, or ethnic differences in human behavior.
- Genetic ancestry is not synonymous with the socially defined concept of race.
- Almost all genomic studies listed here were conducted in populations of European descent and cannot be meaningfully extrapolated to other populations.”

Why is it that PGS (polygenic scores) are not especially robust in predicting individual outcomes? It has to do with the fact that these scores only capture a small portion of the variation in the traits they describe. Furthermore, the score itself quantifies a probability of a certain outcome, but because this calculation is based on only a subset of the factors that contribute to the trait, it cannot accurately predict the specific outcome for an individual (Martschenko and Matthews 2020).

Given the reality that studies of these types have this lengthy list of inherent limitations, what is the best way for scientists to proceed, if at all? Scientists involved in social behavioral research summarized best practices to include the following (Meyer, Appelbaum et al. 2023):

1. Reflection on the risks and benefits of this research to consider whether to proceed with the planned research.
2. Engagement with individuals and communities impacted by potential findings.
3. Consideration of inclusion or exclusion criteria, particularly when they align with identity groups, and how these criteria may blur the lines between biologically and socially defined groups.
4. Justification of how they measure the studied phenotype.
5. Adequately powered and replicated studies.
6. Conducting within-family studies when possible to minimize confounding from environmental and assortative mating (a nonrandom mating pattern where individuals who are similar to one another are reproductive partners) variables, and reporting the corresponding effect size so as not to overstate the impact of genes.
7. Mindful study design to ensure potential results will not exacerbate existing disparities but will instead benefit all equitably.
8. Responsible communication of results with accuracy, care, and sensitivity toward relevant marginalized communities by providing proper context, deconstructing common myths, and discussing limitations of the work.

Unfortunately, though, even a study conducted and communicated responsibly can be appropriated to perpetuate harm. Though the reported facts may remain the same, the lenses and values through which a reader interprets them may differ. People with previously held values about superiority of one group over another have been known to access studies like these to twist results and confirm their own biases (Panofsky and Donovan 2019; Panofsky, Dasgupta, and Iturriaga 2021). For this reason, you'll recall that one of the recommended areas to emphasize in a FAQ section of a social and behavioral genetics paper points out that these studies "do not provide the genetic bases of ancestral, racial, or ethnic differences in human behavior" (Martschenko et al. 2019). This is because not only do

we understand that human populations are much more alike than different on the genetic level, but as we noted earlier, these polygenic scores capture only a small amount of the variation in these traits. When they are used in an attempt to explain behavioral differences along ancestral, racial, or ethnic axes, these citizen “scientists” are employing genetic essentialist frameworks to attempt to give scientific credibility to stereotypical beliefs.

In a particularly horrific example, the White supremacist who opened fire in a Tops supermarket in a predominantly Black neighborhood of Buffalo posted a manifesto prior to his attack detailing his willful misinterpretations and distortions of the educational attainment literature (Lee et al. 2018) in support of his erroneous view that Black folks are intellectually inferior to White folks (Carlson et al. 2022; Stemwedel 2022; Wedow, Martschenko, and Trejo 2022). In his mind, when he murdered 10 innocent Black shoppers at their neighborhood grocery store, his hateful actions were justified to ensure that White people wouldn't be superseded by Black people, a replacement theory that is commonly held among White supremacists (Molteni 2022; Smart 2022). One of the particularly insidious aspects of this attack is how the language of science was weaponized to legitimize his arguments, together with the fear that it would be easy for this to happen again.

Lest you write off the preceding cautionary tale as an example of a fringe extremist acting in a toxic corner of society, we must also acknowledge that these harmful views have been circulating for decades through works like *The Bell Curve*, where the authors contended that structural barriers to personal achievement have been removed and that efforts to eliminate inequities will fail because they result from biological differences (Allen et al. 1996). It is also essential to consider the subtle ways in which scientists inadvertently contribute to this discourse. When communicating genetic results in a way that can be taken out of context to support the erroneous view that racial groups are biologically different, scientists may be adding fuel to the fire. Indeed, the All of Us Research Project recently published intermediate findings from its diverse group of participants in its research study, and in this paper, the research team created a graphic through a statistical method that is known to distort and visually amplify difference between groups (Kozlov 2024). The harm of a

figure like this is it can be easily twisted to legitimize fringe beliefs that are not actually supported by careful scientific interpretation.

Downstream, principles about genetic determinism and hierarchy can also influence applications for emerging technologies in genomic medicine. In an era when genetic discoveries are frequent headlines, the questions of nature versus nurture and what constitutes heritability are often stretched to their limits, and as scientists, ethicists, and health care providers, it is our responsibility to attempt to anticipate the ways in which resulting technologies may unintentionally cause harm. To fully consider these questions and in order to attempt to avoid repeating mistakes of the past, we will reflect on ways in which scientists have previously caused harm. Please join me as we begin our conversations on scientific history and policy.

PART II History and Policy

9 Contemporary Eugenics and Its Historical Roots

The Greek roots of the word *eugenics* refers to “good birth,” that is, good inheritance from strong genes. Eugenic philosophy sought to improve the human gene pool (all the genetic variation found in a population group) by encouraging reproduction among people with “good” genes and discouraging or forcibly preventing reproduction among people with “bad” genes. Although the intentions of these policies were seemingly innocuous, they proved to reinforce biases held by the people crafting the policies.

Francis Galton is credited as the inventor, as it were, of eugenics (Galton 1904). He was an incredibly intelligent polymath and also the cousin of Charles Darwin. In the early 20th century, upon reading Darwin’s *On the Origin of Species* concerning evolutionary principles (Darwin 1859) and integrating the recently rediscovered work of Austrian monk Gregor Mendel on the fundamentals of inheritance from his work in pea plant breeding experiments (Mendel 1865), Galton came to believe that natural selection and evolution could operate more ideally with a little assistance in the form of selective breeding from people. A critical aspect of Galton’s framework for improvement of humankind was a strong belief that nature held far more sway than nurture; that is, he was a genetic essentialist in

his thinking. Galton was obsessed with human difference to the point where he even created a so-called “beauty map” of England after traveling around the country to tally his impressions of the attractiveness of women he observed. Remarkably, in his last year of life, he also embarked on writing a novel about a eugenic utopia where people were screened for their genetic quality, and if they were deemed unfit, they were sent to labor colonies where celibacy was enforced (Gillham 2001). His idea of utopia reads like a first draft of the screenplay for *Gattaca* (Niccol 1997), a dystopian film with uncanny similarities to Galton’s vision. Clearly, his judgment on what were considered ideal genetic specimens was hugely subjective and biased, and these biases became embedded in his proposed eugenic approaches.

It did not take long for Galton’s brainchild to inspire policy, making its way from London to America, where many were eager to embrace the approaches of eugenics. One of the major proponents of eugenic principles in the United States was Charles Davenport, who was an expert in human heredity, using pedigree diagrams to visually describe family relationships and inheritance of traits within families. After receiving a large donation, he founded the Eugenics Record Office (ERO) at Cold Spring Harbor (Cold Spring Harbor et al. 2000; Shotwell 2021), one of the leading institutes of biological research to this day. Field-workers for the ERO generated pedigrees from people in state hospitals, psychiatric hospitals, reformatories, and other venues. They were specifically seeking to identify the transmission of genetic degeneracy across generations within families, and in this way, they were generating a biased evidence basis to justify social engineering interventions (Micklos and Carlson 2000). Pedigrees documenting families with “feeble-mindedness,” promiscuity, pauperism, thievery, wanderlust, and other traits were used to flag a family as having degenerate stock (figure 11). Notably, eugenicists were particularly interested in following behavioral traits that, as we saw in chapter 8, are notoriously complex to study, and genes typically have only modest influence on outcomes for these traits.

Even though in retrospect these types of pedigrees are riddled with genetic essentialism, biases, and inaccuracies, the prevailing interpretation at the time was that “degenerate” families posed a threat to society’s gene pool. At the time, people were concerned that degenerate families were

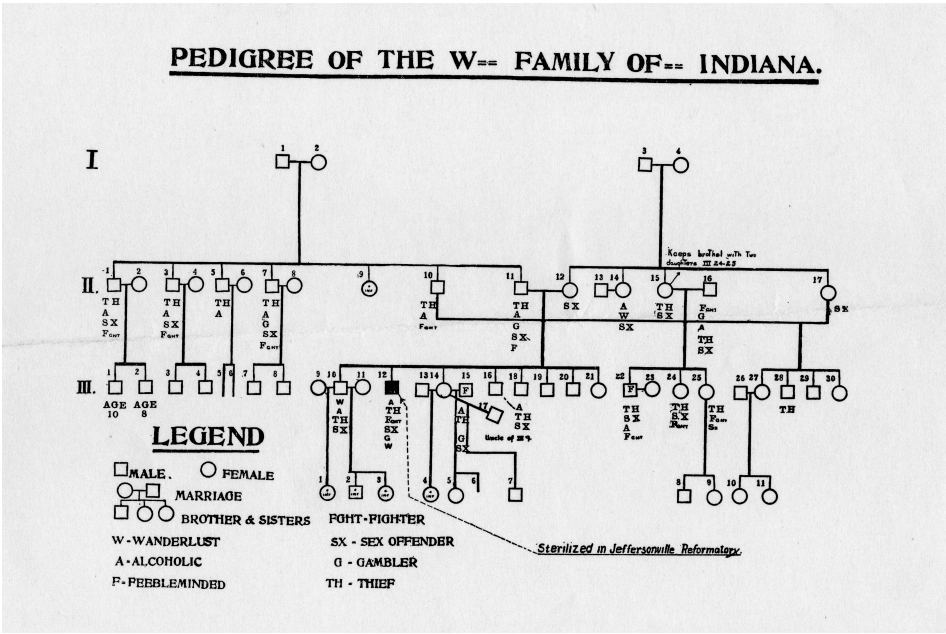


Figure 11. The W family of Indiana was a textbook case of a family described as having numerous undesirable “genetic” traits (Cold Spring Harbor et al. 2000). Circles indicate females, and squares indicate males. Horizontal lines are lines of mating, and vertical lines or branches are lines of transmission. One of the family members was ultimately sterilized for this reason.

reproducing faster than families with good genetic stock, and thus, action to protect the gene pool was necessary for the betterment of society. It was also a common view that degenerate families were not only producing many children but were producing children who would go on to be a burden to society. In the wake of the Great Depression, this economic argument played strongly on societal fears of enduring further financial burden (Stern 2016). Thus, to improve the genetic stock of human populations, principles of inheritance were misused by well-intentioned policymakers as the basis for social engineering using eugenics, and propaganda campaigns (figure 12) were launched to sell these ideas. Importantly, scientists were complicit in the shaping of these policies, with the histories of genetics and eugenics intertwined in complex ways; acknowledgment of the harms propagated by

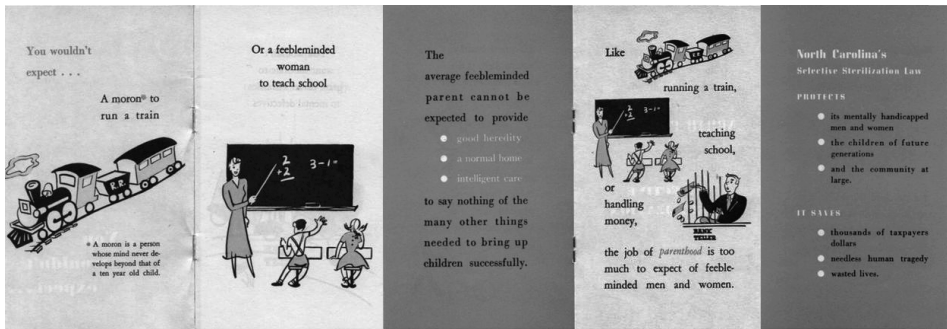


Figure 12. Excerpts from a pamphlet produced by an organization in North Carolina in 1950 as justification for selective sterilization programs operated by the state. (Human Betterment League of North Carolina 1950)

scientists and removal of their names from honorific prizes are early steps in unraveling these connections (Hanchard et al. 2023).

In the United States, these ideas began to take root in a range of settings, from the relatively innocuous to the inarguably harmful. As an example of a relatively harmless venue, “Fitter Family” contests were included at state fairs, the first being held at the 1920 Kansas State Free Fair (Cold Spring Harbor et al. 2000). The families participating in these contests were judged by American eugenicists, doctors, and specialists in the community based on their family health history and the psychological and health exams of the family members themselves. Unsurprisingly, those judged as having strong genetic stock were of western and northern European descent. In parallel, anti-miscegenation laws made interracial marriage illegal throughout much of the United States (Tanabe 2021) because race mixing was thought to pollute otherwise strong genetic stocks. These laws were not struck down in many states until the Civil Rights Movement, when the US Supreme Court took a unanimous stand, declaring these laws unconstitutional in the 1967 case *Loving v. Virginia* (Bates 2022). Ultimately, these eugenic policies and ideals contributed to the horrific Nazi platform of improving the strength of the German nation by purification of the Aryan race (United States Holocaust Memorial Museum, n.d.).

Many may not realize it now, but while Nazi Germany was the most extreme case of eugenic theory finding expression in state policy, these

policies were widespread well beyond the Third Reich. In fact, in the United States, Harry Laughlin, the superintendent of the Eugenics Record Office, managed to use his position as the expert eugenics agent on the House Committee on Immigration and Naturalization to enact severe immigration restrictions from anywhere outside of northern and western Europe (Micklos and Carlson 2000). Under Laughlin and Davenport's oversight, studies were initiated to calculate quantitative indexes for each race, meant to ascertain which races were most prone to certain degenerate traits or, conversely, which races were most inventive (calculated by patents per person, for example) (Cold Spring Harbor et al. 2000). These characteristics were then utilized to justify immigration quotas from different nations around the world into the United States. In other words, this "data" entrenched systemic racism further in US government practices. It is important to mention that these studies were essentially statistical analyses carried out at the population level, based on the scientifically unsubstantiated notions that particular behavioral traits had a genetic basis and that the prevalence of these traits would track differently in each racial category. Indeed, virtually nothing was known about the genetic basis of these traits at the time, yet overzealous policymakers sought to provide pseudoscientific support for their personal agendas.

In addition to setting policy restrictions, these eugenic principles were used to justify bodily harm in the form of forced sterilization procedures, most famously in the case of Carrie Buck (Cold Spring Harbor et al. 2000). Carrie was born out of wedlock to a "promiscuous" mother, and when she herself became pregnant after being raped by a member of her foster family in 1927, she was required to follow in her mother's footsteps to forced sterilization and life in the Virginia Colony for Epileptics and Feeble-minded. This treatment was lawful according to Virginia's Racial Integrity Act of 1924, and her forced sterilization was upheld at the Supreme Court level in 1927 when Justice Oliver Wendell Holmes declared that "three generations of imbeciles are enough." Thirty-two states went on to adopt eugenic laws, ostensibly to curb the rising tide of genetic "degeneracy" in the country (Stern 2016), and over 60,000 Americans (Bekiempis 2020) were forcibly sterilized under these policies (Stern 2016). As these harms continue to come to light, a small fraction of the states implicated in sterilization programs have begun

to distribute reparations for survivors and their families (Associated Press 2023; Morris 2021), but to date, many people victimized by the state still have not been able to access restitution due to technicalities in some cases on who signed off on the sterilization procedures (Mennel 2014; Chernoguz 2015).

Importantly, the idea that traits such as promiscuity, feeble-mindedness, wanderlust, and other behavioral attributes were genetic in nature took hold in the context of race as well. The described forcible sterilization programs disproportionately impacted women of color in the United States, whose intersectional identities put them at greater risk for this harm (Stern 2016). Furthermore, the laws codifying forcible sterilization in the United States were based on Laughlin's Model Eugenical Sterilization Law, which also served as a blueprint for one of Hitler's first hateful policies upon taking power in Germany ("American Eugenics and the Nazi Regime: The Eugenics Crusade," n.d.). The Nazi Racial Hygiene policies created a eugenics court consisting of a health official, a eugenics expert, and a judge (frequently a member of the Nazi party), who would determine which conditions were appropriate for sterilization. Under their oversight, about 375,000 people were forcibly sterilized from 1933 to 1939 (Sofair and Kaldjian 2000). In a sickening and horrifying turn, the Nazi regime found that this intervention was too expensive and too slow to protect the so-called purity of Hitler's ideal Aryan gene pool, a turn of logic that for his followers justified the subsequent genocide of Jewish peoples throughout the Holocaust.

These policies were widely accepted despite what we now see as their sinister and racist implications, and so it is worth emphasizing today that for Americans before the Second World War, racist eugenics was the norm, not just a policy being implemented by extremist groups like the Nazis. As science has progressed, we have begun to gain a more refined understanding of what genetics can tell us about human populations (see chapter 4), so it is tempting to think of these biased and harmful policies as elements of the distant past. Unfortunately, we continue to see egregious examples of coercive family planning and sterilizations, for example, in requiring use of long-acting reversible contraceptives like IUDs to allow Black, Indigenous, and disabled women to access charitable services and public assistance (Cappello 2021). Furthermore, forcible sterilizations were discovered in

privately operated ICE immigration detention facilities, in one case involving a physician known as the “uterus collector” who targeted Black and Brown detainees with limited English language skills (Bekiempis 2020; Dickerson, Wessler, and Jordan 2020). Though this heinous example is particularly harrowing and hopefully isolated, the widespread concepts of genetic essentialism and policing of fertility to reinforce hierarchy of groups through determinations of who is fit to have children remain a force to reckon with in the context of modern genomic practice.



With the development of prenatal and preimplantation genetic testing technologies, we now have the ability to peer into a crystal ball and make predictions about pregnancy outcomes before birth. When we discussed pregnancy and prenatal testing in chapter 6, we talked about the common prenatal cell-free DNA (cfDNA) screens for conditions resulting from changes in copy number of portions of the genome. These conditions alter the gene dosage of dozens or even hundreds of genes, and because of their broad genetic effects, are often associated with significant physical and/or intellectual disabilities that can be life limiting. Once confirmed with a diagnostic test, this information can potentially be used to decide to terminate a pregnancy. Similarly, for families with a risk for a heritable genetic condition, genetic tests can be performed on embryos created through in vitro fertilization (IVF) to select which ones to implant into the gestational carrier. In both cases, there is an inherent value judgment on which genes are worth passing on and on which lives are worth living.

Navigating these complex medical decisions requires the guidance of a knowledgeable professional who can help patients understand the accuracy of the testing approaches, the range of potential outcomes, and the impact on daily life, but in the end, the health care decision should remain patient centered. True autonomy in these decisions is hard to achieve because patients often look to experts for help evaluating the available options, allowing biases of providers to creep into the equation. As mentioned before, my work (Vaimberg et al. 2021; Jungels et al. 2023) and the work of others (Archambault et al. 2008; Hoffman et al. 2016) has demonstrated the existence of biases harbored by health care providers

and trainees in these fields. Preventing personal views from unintentionally influencing patients to make certain choices requires the disciplined framework of patient-centered care, but providers are not always successful in this endeavor. While the majority of genetics specialists (86%) and physicians (63%) do engage in patient-centered counseling by providing as unbiased information to patients as possible, there remain providers who emphasize either positive or negative aspects of a diagnosis involving disability to try to influence a particular choice or outright urge parents to make a specific choice (Parens and Asch 2000).

Some disability advocates view the very existence of prenatal genetic testing as morally problematic (Parens and Asch 1999). This is because it embraces the medical view of disability rather than the social view, wherein the individual's disability is something that requires fixing rather than a society that requires better accommodations for inclusion. In this argument, a prenatal genetic test result boils down all aspects of the fetus to their singular trait of disability. This view, however, takes a societal-level perspective on a family-level decision. That is to say, a particular family may be making a choice to terminate a pregnancy likely to result in a child with a disability because they worry about the child's ability to function in society, being able to provide for the child's complex needs, what will happen to the child after the parents pass away, and how this child may impact other children in the family (Korenromp et al. 2007). These family-specific concerns are not about a medicalized or socialized model of disability as much as they are about parenting in the context of a society with limited social support systems. Furthermore, as with any other community, people with disabilities have a range of views on the ways in which they interact with the larger world, and their perspectives influence how readily they would embrace medical intervention with respect to their disability (Hoffman-Andrews et al. 2019).

The inherent tension between individual decisions and societal impacts is particularly captured by the stories of people in the Down syndrome community. Down syndrome, also known as trisomy 21, results when an extra copy of the 21st chromosome is inherited. The presence of this extra chromosome results in a range of outcomes, most prominently intellectual disability, congenital heart defects that may require surgery, potential for early Alzheimer's disease, and possible leukemia. An individual with

Down syndrome may be highly independent, like a friend of mine who has a rich family life with lots of nieces and nephews, two silver medals from Special Olympics in downhill skiing, a passport overflowing with stamps, and a job at the Massachusetts State House. Some folks with Down syndrome are able to live independently and are increasingly in the public eye as actors, models, designers, bakers, and many other professionals. In contrast, while institutions like Willowbrook (see chapter 7) are no longer a common choice among families who have a child with Down syndrome, the child may require more assistance than others and may need to live with parental or guardian supervision or in a group home setting throughout their lives. In other words, even with a definitive prenatal diagnosis of Down syndrome, the journey may be hard to predict. As one parent of a child with Down syndrome put it, it is like planning for a trip to Italy with sites, restaurants, and language lessons all part of the itinerary, only to find that your plane has landed in Holland and that you must remain there (Kingsley 1987). To be sure, Holland is a lovely destination in its own right; it just isn't what that parent was expecting and will require a significant change in outlook to navigate this new path.

In Sarah Zhang's moving essay "The Last Children of Down Syndrome" (Zhang 2020), she explores the landscape of these decisions in Denmark, where universal prenatal screening for Down syndrome has been offered since 2004. As in many European countries, health insurance and social supports for individuals with disabilities and their families are provided through the government, and yet, 95 percent of pregnant Danes who receive a positive Down syndrome diagnosis choose to terminate their pregnancies. As a result, in 2019, only 18 people with Down syndrome were born in Denmark. The individual people making choices about their pregnancies and their families are not looking to eliminate Down syndrome from the population, but at the societal level, it is adding up to be very close to that. As asked by genetic counselor Laura Hercher in this piece, "If our world didn't have people with special needs and these vulnerabilities, would we be missing a part of our humanity?"

In the United States, one might expect the choices between pregnancy continuation and termination to not be equal. That is, I would expect an inextricable link between the relative lack of social support and the existence of an insincere autonomy of choice. Data at the national level from

1974 to 2016 show a ratio of 3,900 elective terminations to 5,100 live births of children with Down syndrome (de Graaf, Buckley, and Skotko 2015, 2022). These figures can't be used directly to calculate termination rate, however, because among the 5,100 live births, there may be cases where pregnant people declined prenatal testing for religious, moral, or other reasons; where they could not access prenatal testing; or where they received a false negative result. The current working estimate for the termination rate of pregnancies involving Down syndrome in the United States is about 74 percent (Natoli et al. 2012), though these estimates largely precede the advent of cfDNA screening, which greatly expanded prenatal testing for Down syndrome, particularly among parents of younger reproductive ages. Notably, in Massachusetts, a liberal state with a high density of health care providers, those numbers during the same time period are 126 elective terminations and 94 live births (de Graaf, Buckley, and Skotko 2016), exemplifying the observation that the highest termination rates in the United States can be found in the Northeast and West. Importantly, those same states are more likely to have protections in place for reproductive autonomy for pregnant people after the *Dobbs* Supreme Court decision (see chapter 6). Recall that this is significant because the restrictions being placed on abortion services in many states limit the time during which a pregnancy can be legally terminated, often to the period of a pregnancy that precedes prenatal genetic testing.

Consideration of the case of Down syndrome is instructive because it helps us understand how the principles of eugenics in some ways continue to haunt us today. Indeed, it is worth asking how modern approaches compare to past efforts to reduce the incidence of “feeble-minded” individuals in the gene pool. I am certainly not trying to say that people should bear pregnancies for the greater good in order to ensure we do not act according to eugenic principles; nor am I trying to say that I know what the greater good even is. Rather, these are complex issues, where ordinary people have the power to choose which lives are worth living. As we learn more about neurodiversity, we can gain an expanded understanding of naturally occurring neurological variation, and similar open minds should be brought to other traits that will be the focus of developing technologies in genomic medicine. As in chapter 8 where we considered the questions around whether a particular line of genetic research should even be conducted,

perhaps we should invoke a similar reflective practice about whether a particular technology should be developed for health care. If we were to adapt those principles, they might look something like this, with a number of similarities from the previous list:

1. Reflection on the risks and benefits of this technology to consider whether to proceed with its planned development.
2. Engagement with individuals and communities impacted by use of this technology.
3. Consideration of criteria for accessing the technology, particularly when they align with identity groups, and how these criteria may blur the lines between biologically and socially defined groups.
4. Justification of what value the technology adds to health care.
5. Economic analysis to consider the implications of having the technology delivered through insured and private pay mechanisms.
6. Mindful distribution and implementation that will not exacerbate existing disparities but will instead benefit all equitably.
7. Responsible communication and marketing strategies that convey the technological details with accuracy, care, and sensitivity toward relevant groups by providing proper context, deconstructing common myths, and discussing limitations of the technology.

The critical role of scientists and health care providers at this junction will be to continue to advocate for responsible use of developing technologies with incredible capacity for both great benefit and great harm. To responsibly and equitably use genomic technology, we must consider its potential to be used to forward eugenic goals in balance with the principle of reproductive autonomy. Furthermore, we must also carefully consider the issue of privacy because all these technologies involve accessing and curating personal genetic information, as we will see in the next chapter.

10 Personal Genetic Information, Privacy, and the Justice System

One of the things that is often overlooked with the rapid development of genetic and genomic technologies is the parallel acquisition of huge amounts of personal genomic information. This is a form of private health information because it is unique to each individual, and it can be used to make predictions about traits and health. For this reason, it is important to ensure that the information is safely stored and accessible only by people with appropriate permissions.

Genomic data can be generated in conjunction with consumer genomics products, clinical research, and clinical genetic testing. Depending on which technology is used to peek at one's genetic code, there is a different extent of information that is assessed. For example, if we use the analogy of the genome being the book of life, let's consider a particular page of the book with the following sentences: *My favorite molecule is DNA. That was probably obvious.* When genotyping technologies are used, only a subset (usually less than 1%) of that information is captured, whereas sequencing paints a much more complete picture of what genomic content is present (table 3). In the case of genotyping, the existence of a reference genome allows one to infer what content may be nearby. For instance, if genotyping identifies the letter *i* in the fourth position, then the scientist

Table 3 Contrasting the information acquired using genotyping versus sequencing technologies

Technology	Genomic content
Genotyping	<i>My favorite molecule is DNA. That was probably obvious.</i>
Sequencing	<i>My favorite molecule is DNA. That was probably obvious.</i>

NOTE: The two sentences are an analogy for the content of a section of the genome. The highlighted letters in the sentences are the ones that could be read by the given technological approach.

Table 4 Comparing and contrasting the approaches and applications of genotyping versus sequencing

Attribute	Genotyping	Sequencing
Extent of sequence determined	Examines specific positions in the genome	Can study the entire sequence of a genome or segment of the genome
Ability to identify previously undescribed variants	Typically identifies known sequence variants	Identifies known and unknown sequence variants
Typical technological applications	More prevalent in consumer genetics	More common in clinical genetics
Relative cost	Inexpensive	Becoming less expensive

can infer that the full word is likely *obvious*, though the further we get away from the genotyped nucleotide in the DNA, the less certain we can be about what information the genome contains. The approaches also differ not only in the percentage of the genome that is analyzed, but also in whether they are capable of identifying novel (previously undescribed) variants. The costs and applications are also linked in that the lower expense of genotyping approaches make them well suited for consumer genomics platforms that are offered directly to customers, whereas the more complete sequence determination, despite the increased cost, makes sequencing the tool of choice in most clinical contexts. These differences are summarized in table 4.

With this understanding of the basic differences between the technologies used to read DNA sequences, we can now begin to consider the contexts in which they are implemented. DNA sequence data is a commodity that has different implications and potential harms when accessible by consumer genetics companies, health care providers, employers, and the government. Let's begin by considering consumer genetics.



Consumer genetics is an umbrella term meant to encompass what many geneticists have come to think of as recreational genomics: applications like ancestry and perhaps some health-related traits, but also things with less scientific basis, like athletic abilities, individualized art derived from DNA sequences, and wine pairings for your personal palate. Before I go any further, I want to be clear that I have no personal connection to any of the companies offering these products; my goal is to try to give you a balanced sense of what these companies offer, what information they are collecting on you, and a sense of the interpretation and storage of this information.

Many people first engage with consumer genetics companies through an interest in genealogy. The companies that offer genealogical testing also often include analysis of the geographical origins of one's ancestors and, increasingly, some introductory analyses on health predispositions. For this level of analysis, genotyping, even though it covers less than 1 percent of the genome, generally provides enough information to identify living relatives with shared genetic elements, genetic similarity with people from certain parts of the globe, and a sense of some of the genetic determinants of health. As described in chapter 4, race is not a biological construct, though that isn't always obvious from the way some of these products are marketed, with a focus on using information encoded in your DNA to discover something about your identity. Genetic genealogy relies on similarity with modern-day individuals, either from public or company databases, to look for combinations of genotypes that are in common with the customer's sample. This can help determine if someone is a long-lost half-sibling or if it appears one's ancestors came from certain parts of the world. With roughly 1 in 25 Americans now having access to this type of

personal genomic data (Regalado 2019a, 2020), the ability to identify living family members and genealogical history is relatively robust.

Whether the consumer genomics information obtained is genotypes or sequences, some have speculated that the real product for the consumer genomics companies is not the clinical-risk interpretations offered to their clients, but rather the opportunity to sell their genomic data (Brown 2023). Depending on the platform, this may be structured as an opt-in or opt-out standard, and it isn't fair to characterize this as an entirely sinister breach of privacy. Often, the genomic data is paired with information provided by the client on their own traits and health issues. The combination can be sold to pharmaceutical companies to aid their own research into the genetic basis of traits and genetic conditions. This has the potential to accelerate drug discovery by removing the need to recruit study participants with a trait or condition of interest. The Michael J. Fox Foundation has endorsed this approach by subsidizing genotyping of people with Parkinson's on a consumer genomics platform to rapidly and inexpensively create a cohort of at least 125,000 people for whom genetic information and Parkinson's characteristics are known (Smolensky et al. 2020). Participants, like my childhood friend's father, longitudinally provide information about the course of their disease, and these data are available to researchers through institutional review board approval to protect the research subjects' interests. I know that for my dear friend and her wonderful dad, the opportunity to contribute to this research provided some actionability while navigating a very challenging situation.



Another potential use of genomic data obtained by consumer genomics companies involves allowing the consumer themselves to download the raw data. This data can then be uploaded into third-party platforms for further analysis. One of these platforms is GEDmatch, which accepts data derived from the major companies providing consumer genomic products and integrates them across platforms to help people find genetic relatives. Although the original intention of this platform was to aid genealogists, unanticipated uses of their databases in criminology have also come to light. In traditional law enforcement, there are forensic databases and

offender databases. Forensic databases contain information from DNA profiles collected as evidence at crime scenes. Offender databases include DNA profiles obtained from people who were arrested, who were compelled to provide a sample, or who committed a crime. When there is an overlap between the two databases, detectives can identify suspects in their cases. In a variation on this method, detectives uploaded forensic DNA information into GEDmatch in order to identify relatives of the man known as the Golden State Killer (Kaiser 2018). He had evaded capture for decades for his involvement in horrifying rapes and murders. Through this approach, law enforcement agencies were able to identify blood relatives who were roughly his third cousins, which then allowed detectives to connect the dots to identify him. From a public safety perspective, this is an important outcome. But from a privacy perspective, we must acknowledge that the people who granted permission for their genomic information to reside in GEDmatch are not necessarily the ones who are themselves at risk for privacy breaches with consequences. I don't mean to imply that we should elevate the importance of privacy for people who are committing violent crimes, but rather that guidelines should exist for what circumstances are acceptable scenarios where law enforcement could co-opt these databases. Minimally, we should consider whether it is appropriate to use this type of investigative genetic genealogy in nonviolent and victimless cases. As of 2019, the Department of Justice has an interim policy on investigative genealogy that limits its use to unsolved violent crimes and unidentified remains of suspected homicide victims (National Institutes of Health, National Human Genome Research Institute 2021). This is not merely an academic question, because a recent estimate indicates that 60 percent of White Americans could be identified by a combination of a DNA sample and a consumer genomic database with 1.3 million users (Erich, Shor, and Carmi 2018). And for reference, there are multiple databases in existence that meet and exceed that criterion already.

Concerns are also emerging about hackers directly gaining access to the genetic databases associated with consumer genomics companies. In 2023, over the course of few months, a data breach of the 23andMe databases exposed the personal genomic data of 6.9 million users when hackers accessed individual accounts and connected to user networks through the DNA Relatives and Family Tree tools (Carballo, Schmall, and Tumin

2024). The information accessed includes not only genetic data but also geographic location, birth year, family tree, and uploaded photos. This was especially concerning when it came to light that the hackers created curated lists of Jewish users and Chinese users that were then sold on the dark web before the customers were notified of the breach. The lists of Chinese user data were sold to an individual who goes by the name “Wuhan,” and the Jewish user data was offered up in the wake of the deadly explosion at Al-Ahli Arab hospital. The continuing anti-Asian bias that lingers after the COVID-19 pandemic and the geopolitical climate during the ongoing Israel-Hamas conflict make the existence of these curated lists even more alarming, particularly if this information was to find its way into the hands of extremists.



When it comes to clinical genetic testing, a genetic counselor begins by exploring the clinical utility of pursuing testing and ordering a genetic test. The patient sample is sent off to a laboratory, which carries out the testing, interprets the results, and shares the information with the clinician. This information then makes its way into a patient's electronic medical record, where it is protected health information that is safeguarded by HIPAA (Health Information Portability and Accountability Act). An element of complexity in this chain of events is related to the privatized health care and employer-based insurance systems in the United States. If one's health insurance is provided through one's employer, what protections are there to ensure that the employer does not access an employee's private genetic information?

The 2001 case of the Burlington Northern Santa Fe Railroad exemplifies the inherent conflict of interest between employer and employee with respect to personal genetic information. In this case, a number of union members who worked for the railroad were experiencing numbness and physical pain that was consistent with a carpal tunnel diagnosis (Girion 2001). They subsequently filed workers' compensation claims for carpal tunnel syndrome. What happens next is where it all goes sideways. Some of them were asked, coerced, or even told to provide a blood sample that was subsequently tested for genetic variants associated with hereditary

neuropathy with liability to pressure palsies (HNPP) (Chrestian 1998), a syndrome that shares many features with carpal tunnel. Some of the workers did not consent to this testing, and at least one was threatened with termination if he did not provide a DNA sample (Lewin 2001). The suspicion is that the railroad sought this genetic information to inform decisions about whether to provide workers' compensation payments, potentially denying compensation to people with a genetic predisposition for HNPP. It is unclear how prevalent these coercive intimidation tactics were, but there were enough concerns to warrant the Equal Employment Opportunity Commission suing the railroad. Ultimately, the railroad and unions settled out of court (Philipkoski 2001), so it is difficult to know the full extent of the harms, but at least the settlement required the railroad to destroy blood samples and test results for the affected employees.

In the Burlington Northern Santa Fe Railroad case, the lawsuit posited that the railroad's behavior violated the Americans with Disabilities Act (ADA), but at the time, there were no specific laws in place to protect people from genetic discrimination other than for employees of the federal government. Because prevailing interpretation of the ADA was relatively narrow, the passage of the Genetic Information Nondiscrimination Act (GINA) in 2008, after 13 years of legislative efforts, was an important step forward in prohibiting discrimination in health coverage and employment based on genetic information (Personal Genetics Education & Dialogue, n.d.). While some states have extended some of these protections to other forms of insurance, such as long-term care insurance, privacy remains a concern. The fact is that patients generally find doctors and scientists more trustworthy than health insurers and employers (Hudson 2007), which underscores the importance of offering legal protections in these areas. However, even with GINA in place, it isn't always clear that employers should never have access to the genetic health information of their employees. Consider, for instance, a commercial airline; should they know if their pilot has a genetic condition that predisposes them to seizures, to ensure safety of their passengers? The goal of these laws is not only to protect the privacy of people undergoing genetic testing, but also to ensure that people do not avoid genetic testing when it might be beneficial to them.



Although information derived from newborn screening is not protected by GINA, recent developments have demonstrated the harm that can arise from privacy violations associated with this type of population-wide testing for genetic conditions. Referring to chapter 7, newborn screening is carried out shortly after birth for conditions where the child may benefit from early intervention. We discussed deafness in this context, but there are a whole range of conditions that are encompassed within routine newborn screening. These conditions must be able to be detected dependably within the first few days of life, and there must be an effective intervention or treatment available. The American College of Medical Genetics and Genomics and the Department of Health and Human Services have a recommended uniform screening panel (RUSP) of 37 core conditions and 26 secondary conditions, as of 2023 (Health Resources & Services Administration 2023). The conditions on this list are significant ones. For instance, phenylketonuria is a metabolic condition where a person is unable to properly metabolize the amino acid phenylalanine. Amino acids are a building block of proteins and can be found in many different foods. When children are unable to break down phenylalanine, the amount of phenylalanine begins to build to harmful levels that can damage the body, especially the brain. Children exposed to high levels of phenylalanine can develop intellectual disability, seizures, and other significant outcomes if the intake of phenylalanine is not modified through a special diet. This diet needs to be implemented as soon as possible and over the course of a lifetime to protect the child from the impact of high levels of phenylalanine. In other words, this public health program is critically important for newborn health in children who have these conditions, and according to some estimates, roughly 13,000 babies are thought to be identified annually through newborn screening programs (Centers for Disease Control and Prevention 2020).

Though the RUSP list is nationally recommended, newborn screening is administered through state-run labs, so some states may screen for more than the RUSP conditions, and others may screen for fewer conditions. Generally, for children born in hospitals in the United States, a blood sample is collected from a heel prick during the hospital stay. The blood drop-lets are absorbed by and dried onto a thick paper card known as the Guthrie card, and these samples are used in newborn screening protocols.

Depending on the state, those cards with unused samples can remain in the state labs for years or even decades. Police in New Jersey took advantage of these stored samples when they subpoenaed a state lab for access to newborn DNA from an infant who was the child of a suspect in a 25-year-old cold case (Faife 2022). In a twist, the suspect became a client of the New Jersey Office of the Public Defender, who has filed a lawsuit to determine how common this practice is. The potential harm from this investigative workaround involving newborn screening is that families may begin declining newborn screening to the detriment of their child's welfare on the grounds of privacy. In a criminal justice system where Black folks are disproportionately incarcerated and harshly punished, the potential for this to exacerbate health care disparities is alarming.



The intersection between population screening and criminal justice is also relevant in consideration of scientists' role in justifying carceral punishment for people based on their genes. In the 1960s and 1970s, a number of scientists sought to understand the connection between genes and violent behavior, so they screened people in penal institutions and juvenile detention facilities for the presence of an extra Y chromosome (XYY syndrome) (Hanchard et al. 2023). These studies were carried out in populations where the majority of the research subjects were Black, where informed consent was not properly obtained, and where privacy and legal protections were not guaranteed. The findings of these studies were overstated and misstated, but follow-up studies with the intention of proving a biological basis for violence were conducted nonetheless, further creating a breakdown in trust between Black communities and scientific institutions.

Unfortunately, the strategy of using science in defense of incarceration and state-sanctioned violence continues with the example of sickle cell anemia. Sickle cell anemia occurs as a result of a variant in the beta-globin gene, which encodes a subunit of hemoglobin. This variant causes a structural change when red blood cells are in low-oxygen environments, such that the hemoglobin takes on a fibrous shape. This causes the cell to become crescent-shaped and rigid, making it difficult to navigate through small blood vessels like capillaries, which in turn causes intense pain

crises that we first learned about in chapter 4. The negative impacts of two copies of the sickle cell variant in one's genome are undeniable, but a single copy can offer some advantage in protecting the individual from the malaria parasite. Thus, people whose ancestors come from places where malaria is endemic tend to harbor higher frequencies of the sickle cell variant than other populations. This includes people from sub-Saharan Africa, the Mediterranean, the Middle East, and South Asia, and among Black folks, roughly 1 in 12 people will have a copy of the sickle cell variant. When it comes to Black men who are disproportionately the targets of state-sanctioned violence, when they die at the hands of police officers, their sickle cell status has been invoked as a way to give police some deniability for use of excessive force (LaForgia and Valentino-DeVries 2021). The argument put forward by the officers—and by physicians who were complicit in writing pro-police autopsy reports—is that if an individual had the sickle cell trait, that is, a single copy of the sickle cell genetic variant, then in low-oxygen conditions like a knee on the neck, the suspect may have an extreme reaction to the immobilization maneuver. Let me be clear: there is no scientific evidence to support this conclusion. By the time an autopsy is conducted, red blood cells may have taken on a crescent shape in the hours and days between these men's last breaths and the autopsy. Furthermore, the 47 Black men who died in police custody identified in a *New York Times* investigation (LaForgia and Valentino-DeVries 2021) all had a single copy of the sickle cell variant, not the two copies required for the men to develop full sickle cell anemia. These cases all highlight the state exercising its power and the potential for these power hierarchies to be reinforced along racial lines. This is what racism looks like. The only positive aspect of this story is the fact that when Derek Chauvin's lawyers attempted to use George Floyd's sickle cell trait status in his defense, the judge didn't buy it, and Chauvin has since been convicted for his actions that led to George Floyd's death. State-sanctioned violence and misuse of genetic information is a sickening combination used here to protect those in power from consequences for their actions. It is my hope that this is the beginning of a trend of accountability for these despicable actions and, more importantly, an end to racially motivated violence.



Sometimes, I think anger is a necessary emotion. Bearing witness to these acts, even just by reading this book, should make you angry. But anger does no good if it stops there. We saw anger transformed into a unified voice demanding justice during the Black Lives Matter protests of 2020. We must not let our voices be silent now, a few years down the line. It is our responsibility to continue to ask the hard questions. What is our role in this system? When have we intentionally or unintentionally looked the other way? How did we allow misconceptions to exist unchallenged? Because the stakes are literally life and death when people falsely conflate race and genotype, I urge you to hold on to these emotions and questions long enough to finish this journey with me. We have a few more stops, and in the next part, we will move to consider the use of genetics in science and medicine, beginning with the question of who gets left behind.

PART III Genetics in Science and Medicine

11 Who Gets Left Behind

As we begin this next part of the book considering the scientific and clinical implications of current genetic insight into human biology, it is critical to begin with a framework where we consider what is missing and who is left behind by the way science is structured. Though science is fundamentally the pursuit of truth, that doesn't make scientific understanding automatically infallible or even complete. The truth is that differences in the ways questions can be asked, data can be collected, and findings can be interpreted all flow through social lenses that are susceptible to the same structural issues that influence the ways in which we all move through the world. Scientists are themselves, by and large, well intentioned, often altruistic even. However, privilege can create oversights, and these oversights can make their way into how science is conducted, which in turn will create inequities in who benefits from the resulting findings.

The scientific and clinical workforces commonly are not nearly as diverse as the communities they serve. In the scientific community, it is recognized that we cannot realize the full potential of science in the absence of a diverse workforce (Bonham and Green 2021; Thomas et al. 2021). Diverse teams across numerous dimensions produce more innovative and creative science (Valantine and Collins 2015) and are able

to outperform homogenous groups of problem solvers (Hong and Page 2004). Similarly, patients have demonstrably better outcomes when they are treated by providers who have shared aspects of their identities (Alsan, Garrick, and Graziani 2018; Greenwood et al. 2020; Nguyen et al. 2020; Frakes and Gruber 2022; Wallis et al. 2022; Snyder et al. 2023), and additional evidence demonstrates reduced mortality when patients are cared for by female physicians (Tsugawa et al. 2017). Yet the availability of providers from marginalized backgrounds lags well behind the diversity of the patient population (Guevara, Wade, and Aysola 2021; Kamran et al. 2022). Given the growing realization that it matters *who* does the science and *who* cares for the patients, the 2023 Supreme Court of the United States ruling banning race-conscious admissions will needlessly risk the health and lives of people from marginalized backgrounds (Hamilton, Rose, and DeLisser 2023; Rubin 2023; Schmidt, Gostin, and Williams 2023).

With increasing awareness of the many negative consequences resulting from lack of representation, scientists and clinicians have begun asking hard questions of our fields to understand in what ways our privileges and oversights contribute to flawed science, poor care, and, ultimately, health disparities. In this chapter, we will take a deep dive into the scientific systems that generate incomplete evidence bases and gaps in knowledge that together have the potential to exacerbate health inequities. In particular, we will consider the groups who are excluded from these studies in the first place, frequently because members from those communities are not at the table with the scientists designing those studies. Toward this end, we will examine science's approach to studies involving race, sex, pregnancy, and disability and where these studies fall short.



When it comes to representation of people from a range of ancestries in genetic studies, it probably won't surprise you to learn that genetics has a diversity problem. When comparing data from the global population with people included in genome-wide association studies (GWAS), the kind of studies we first encountered in chapter 8 that are used to identify which genetic regions are associated with which traits or conditions, a clear mis-

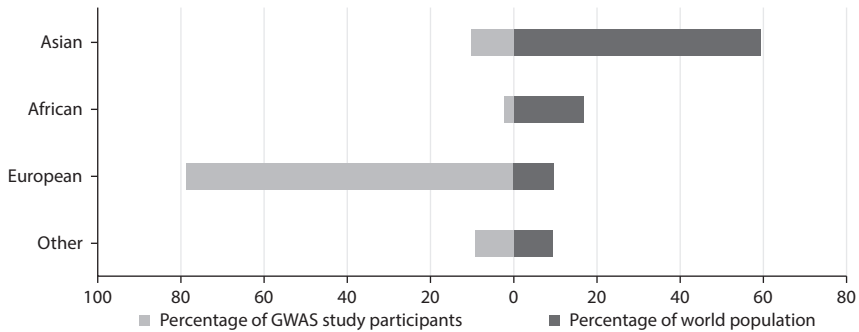


Figure 13. Two contrasting bar graphs, demonstrating representation across continental ancestries in the world population on the right and subjects in genetic studies on the left (Sirugo, Williams, and Tishkoff 2019). Both compare data from 2019.

match is observed (figure 13). The makeup of folks included in genetic studies is highly Eurocentric (Sirugo, Williams, and Tishkoff 2019), with nearly 80 percent of people participating in GWAS being European, even though the continent only represents roughly 10 percent of the global population.

I imagine some of you at this point are perhaps oscillating between disbelief and confusion. It's true, in chapter 4 we spent a great deal of time looking at human populations from a variety of perspectives and concluded that we are far more alike than different, that within population variation far exceeds between population variation. As such, you may be wondering about the apparent contradiction—why should we worry about who we include in genetic studies if we can capture the majority of human genetic variation by looking within populations? In other words, wouldn't in-depth study of a single population give us a solid understanding of most human genetic variation? The key words here are *in depth*. These studies are examining a small fraction of the genomes within any given population, so there are many low-frequency variants that will be missed using this approach. Importantly, the frequencies of variants can differ from population to population. This is true because as humans migrated throughout the globe, starting in Africa and moving outward from there, subsets of variants were represented in the gene pools of migrating populations (Henn, Cavalli-Sforza, and Feldman 2012). As we move farther

away from Africa, we see a decrease in genetic variation present in a population. Also, geographic barriers, bottlenecks related to disease and starvation that caused loss of some genetic variants, subsequent founder effects where a limited number of people started new populations, and unique selective pressures in new habitats that enriched or eliminated specific variants created new equilibrium points for the frequencies of the remaining variants. Imagine the genetic variants in Africa as a jar full of jelly beans. If you reach in and take a handful of jelly beans to populate another geographic area, you are unlikely to capture all the colors of jelly beans, and especially not at the same frequencies as in the original jar. All that is to say, a variant that might be rare in a European population may be quite common in a population from elsewhere on the planet. There will only be a small number of variants that are unique to specific populations; however, for African populations, because only a subset of the human genetic variation in the parent African populations was represented in the populations that migrated out of the continent to populate the rest of the globe, pursuing genomic studies in partnership with Africans and African scientists can yield as much as three to five times as many variants as studies from other ancestries (Wonkam and Adeyemo 2023). For this reason, the ambitious call to sequence three million African genomes (3MAG) has been described as a scientific imperative in service of health equity (Wonkam 2021). In sum, to capture a more complete picture of human genetic diversity, we need to also have diversity of genomic research participants who have input into the scientific goals that will best serve their communities (“Africa’s People Must Be Able to Write Their Own Genomics Agenda” 2020).



The significance of lack of data is that subsequent studies are based on these datasets, most notably GWAS, that attempt to link regions of the genome to functions that influence traits and health. You may remember from chapter 8 that GWAS relies on statistical analyses to determine if a particular genetic variant is overrepresented in populations with a trait or health condition of interest. The statistically identified genotyped variant

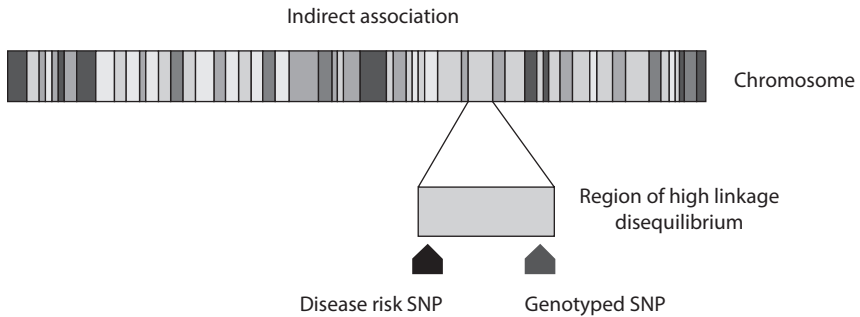


Figure 14. This diagram shows a linear chromosome in the upper rectangle. Each shaded segment of the chromosome corresponds to a region that has been inherited in a continuous stretch, a linkage disequilibrium block in genetic parlance. The lower rectangle is a zoomed-in image of a single linkage block. In order to show statistical correlation with a trait or health condition, a genotyped variant like a single nucleotide polymorphism (SNP) need only be within the same linkage block; it need not be the disease risk-causing variant itself (Bush and Moore 2012). Each linkage block can vary in length, but it can easily contain multiple or even dozens of genes.

is often not the variant that directly *causes* the genetic difference, as it may just be hitchhiking on a nearby stretch of DNA (figure 14) (Bush and Moore 2012). This seemingly minor difference has important implications when we connect it to the concept of human migrational history. As humans move into new areas, it is possible for the DNA stretches to get shuffled into new combinations such that the genetic variant identified as associated with a trait in one population may not be hitchhiking with the disease risk allele in a different population.

This may still sound a bit like inside baseball, so let me try to explain the significance of this plainly. Because the vast majority of GWAS studies have been carried out on people of European ancestries (Sirugo, Williams, and Tishkoff 2019), GWAS findings have significant limitations. Specifically, these studies identify a genomic region associated with a trait but not the causal variant itself, so we must recognize that the regional marker alleles identified in studies in one population may not be the same alleles that would be found at that same genetic position in another population. This means that when one goes to calculate a

polygenic risk score from these indirect measures of risk, it will be most informative in the population that was initially studied and less accurate in other populations (Abu-El-Haija et al. 2023). In other words, the polygenic risk scores that are calculated based on genetic studies of White folks will not be transferable to populations of color with precision (Ding et al. 2023; Lewis and Green 2021), and any resulting benefits will not be spread equitably (Martin et al. 2019; Meyer, Tan et al. 2023). The predominance of data from White populations therefore significantly restricts applicability of findings for populations of color.



Another potential consequence of the human migrational patterns we have discussed is the causative variants themselves may differ from population to population. For instance, maybe multiple variants arose independently after people moved into new geographical niches, and the cause of a particular disease within each population may then differ. For this reason, sampling a single population's genetic variation would also generate harmful knowledge gaps. In clinical genetic testing applications, this could lead to identification of an unknown variant in the previously uncharacterized population. This is called a variant of uncertain significance because, while the gene may be known to have involvement with a particular health condition in another population, if the specific genetic variant has not been previously described, it can be difficult to predict if it is likely to cause disease or not. Revisiting the analogy of the genome as the book of life, if someone was in the midst of reading a sentence and came across an unfamiliar term, they would probably want to turn to a dictionary to look up the definition. However, if this is a word that has not been entered in the dictionary, it will be difficult to know the precise meaning of the word. Similarly, if a scientist comes across a new and unfamiliar sequence in a gene, there may be no reference to which they can turn to elucidate its meaning, especially if that population has been excluded from prior studies. It might be possible to figure out a bit of the meaning by context. For instance, if it's a period at a spot earlier than one would normally find it, that would have a high likelihood of impacting the gene's function if it disrupts a large portion of the gene. However, if it is a smaller change that changes one word to a similar

one, like *affect* to *effect*, it can be tough to figure out if that change, though grammatically incorrect, is enough on its own to prevent the gene from functioning appropriately and to potentially contribute to a disease state.

As a result of the documented gaps in genetic databases, variants of uncertain significance are more prevalent in historically excluded populations (Landry and Rehm 2018). Not only would this scenario lead to difficult to interpret genetic test results in communities of color, but it would also lead to knowledge gaps related to whether the different variants cause a range in severity of disease or response to treatment. These scenarios specifically lead to poorer disease prediction and treatment options for historically excluded populations. For these benefits to be realized equitably, it is critical for inclusive study design and meaningful community engagement to be utilized at the outset.



Inclusive study design practices also come into play when evaluating visual clinical data, not just sequence data, related to genetic conditions. With this in mind, one frequent feature of genetic conditions is an impact on structural aspects of anatomy, often identified as differences in facial structure. A trained specialist can identify the nuanced differences and has the ability to distinguish variation in size, spacing, and arrangement of facial features. In many ways, this aspect of genetics, known as dysmorphology, has been more of an art than a science, but now people have started translating this analysis into artificial intelligence algorithms. The algorithms first train on images of faces from folks with known genetic syndromes. They can then quantify the same aspects of facial features from a simple photo of a patient and match them to the training dataset to come up with suggested diagnoses. The really impressive result of this algorithm is that it performs with similar accuracy to expert clinicians (Gurovich et al. 2019)! The significance of this finding is also related to workforce issues in that many people outside of major metropolitan areas with academic medical centers do not have easy access to clinical geneticists (Cosgrove 2020). If artificial intelligence can help nonspecialists make these diagnoses, then patient care will be improved even when a specialist is not readily available.

However, there is also a significant downside with these methods. Just as with genomic data, when the training and reference datasets are primarily from White folks, the subsequent ability to analyze results from other populations is limited. In this case, artificial intelligence will operationalize biased views on the ways facial structures may appear in specific genetic conditions. We know from observation that facial features vary across human populations, so if a genetic condition tends to result in narrow-set eyes, that may result in quantitative differences in the measurements themselves depending on the ancestry of the patient in question. As such, when an algorithm is trained on a homogenous dataset featuring White patient images, it performs less well for diagnosing patients of color (Porras et al. 2021). The net result of algorithmic bias in this scenario would be underdiagnosis in historically excluded communities. To address this gap, a working group at the National Institutes of Health has partnered with clinicians around the world to establish a repository of images of patients living with a range of genetic conditions in a variety of backgrounds (Adeyemo and National Institutes of Health 2021). This remains a work in progress, as many conditions still do not have images submitted for them, but efforts like these will help address disparities in care that result from bias baked into artificial intelligence. The bottom line about AI is that the algorithm will simply replicate any bias and limitations present in its training dataset.



As demonstrated by the preceding examples, gaps in knowledge can lead very directly to harms in being able to offer equitable health care to patients from communities that have been excluded from research. The importance of inclusive research has recently gained more attention, and national research initiatives are beginning to prioritize closing knowledge gaps by focusing on historically excluded communities. This is one of the driving factors behind the All of Us project, first mentioned in chapter 3, which seeks to collect genomic and other health information from historically excluded groups, especially from communities of color, sexual and gender minorities, folks with disabilities, and other minoritized groups (Mapes et al. 2020). The goal is to begin to fill the knowledge gaps that

result when most genetic and genomic studies are conducted on White populations with additional limited diversity in the dimensions of income, educational attainment, and geography.

While genetics researchers have begun to reckon with these historical policies of exclusion, significant barriers remain in the effort to diversify our genetic studies. In order to include people in these studies, they must trust that you have their best interests in mind, and this is justifiably not the case for many communities of color. The seminal example of this, the Study of Untreated Syphilis carried out at Tuskegee (1932–1972), allowed nearly 400 Black men to be studied for the untreated course of syphilis while withholding effective treatment from them (Gamble 1997). Clearly the men included in this study did not give their consent, nor was the research in their best interests.

In terms of genetic studies, the Havasupai, a Native American tribe who lives in the Grand Canyon, entered into a partnership with researchers from Arizona State University who planned to study the genetic basis of diabetes, a condition that is highly prevalent in the tribe (Harmon 2010). The researchers violated the trust of that partnership when they went on to use the same samples to study schizophrenia and tribal histories that were in conflict with their traditional stories. Although the scientists who were involved indicated that they did receive permission from the participants, there was clearly a disconnect in that the subjects did not understand the full scope of the possible genetic studies that would follow. With historical cases like this, it is not surprising that BIPOC communities would not easily embrace science without suspicion.

One of the ways in which trust was broken in these examples is that the folks participating in the research were not viewed as true partners in the science. Rather, the relationship was entirely transactional. The samples were given even though consent was not, at least not fully, and that is when the relationship ended. Benefits extended only to the scientists in terms of career advancement and accomplishment in academic halls, but the people who had trusted these scientists were left behind. Though researchers studying genetics have generally been viewed as trustworthy recipients of this personal information (Hudson 2007), I suspect that these trends would look different if results were disaggregated by race and ethnicity. Creating a structure of genuine partnership with equitable

sharing of benefits (Fox 2020) is a model promoted by Indigenous scholars in genetics. In this model, the relationship between scientists and research participants does not end with the donation of samples; rather, shared interests with respect to custody of samples, research pursuits, and subsequent application to clinical interventions would develop through collaborative processes whereby everyone with a vested interest in the research would receive some benefit.

A discussion of the financial benefits is an important one in this context because it is too often the case that minoritized communities are necessary for scientific advances, but they don't derive benefit from them. This is particularly the case for Henrietta Lacks, whose cervical tumor was biopsied without her consent to create the HeLa cell line. Ms. Lacks's treatment is an especially important example of a breach in community and family engagement. Ms. Lacks was a patient at the Johns Hopkins University "colored ward," where she was seeking treatment for her cervical tumor. The tumor was incredibly aggressive, and cells derived from her tumor at biopsy grew remarkably quickly in a lab setting. This was actually the first time human cells were successfully grown in the lab, so when she sadly lost her life to her tumor, a research technician was sent to the morgue to collect more samples (Skloot 2010). This was how the HeLa cell line came to be established. As you probably gathered by my omission, Henrietta did not give permission for this before she died, nor was her family approached after she passed. While her cells have contributed to over 70,000 research studies, were instrumental in developing the polio and COVID vaccines, and have even been to space, the ends do not justify the means. Rebecca Skloot, the author who wrote the life story of Henrietta Lacks and her cells, was able to advocate on behalf of the family to ensure that the National Institutes of Health would take steps to protect the interests of the family with respect to future research. And yet, in 2013, when cancer biologists wanted to understand what contributed to the aggressive nature of her tumor, they sequenced her cancer genome and published the sequence online without consulting the Lacks family. While the cancer genome will have alterations specific to the cancer, it will also contain information unique to Henrietta and her descendants, so their privacy and wishes were violated yet again. Of these violations, Jeri Lacks-Whye has said, "I look at it as though these are my grandmother's medical

records that are just out there for the world to see.” Rebecca Skloot returned to help the family advocate again, and now Henrietta’s cancer genome is only accessible to researchers upon approval by a review board that includes members of the Lacks family. However, this does not address the issue of reparations or the profits companies cleared on the biomedical products made from research involving the HeLa cell line. As is so frequently the case, Black people and people of color play a critical role in scientific advances, but they do not benefit equitably for their contributions.

The Lacks family story is one that highlights several problems related to inclusivity in biomedical research, and scientists are beginning to acknowledge their role in perpetuating these harms. To this end, the All of Us research program is seeking to build bridges to include communities that have been historically excluded from genomic research.

Further efforts to engage historically excluded study participants center around privacy of personal information to build trust with minoritized communities. The core values of reflecting the rich diversity of the United States, earning trust through transparency, giving participants access to their information, and elevating the importance of security and privacy all work together to promote a genuine partnership with participants. Safeguarding personal genomic information is key to protect anonymity and to prevent reidentification. These and other efforts to build trust have led to significant gains in representation of participants in All of Us and related initiatives in the dimensions of race/ethnicity, income, age, sexual and gender minorities, and education (Ramirez et al. 2022), which will reap benefits as the human pangenome across all these populations is refined (Attwaters 2023; Liverpool 2023).

Clinical genetic testing has had some similar challenges in uptake by minoritized communities (Armstrong et al. 2005). Given our exploration of the dimensions related to privacy that may contribute in part to these differences, development of improved protections for the privacy of vulnerable communities and reparations for the instances of broken trust may help us make inroads to wider uptake and benefits of clinical genetic testing.



Though Ms. Lacks's cells went on to fuel many fundamental scientific advances and led to many profitable products, her family received no compensation for any of this until 2023, a full 72 years after her tumor killed her ("Immortal Cells" 2023; Wadman 2023).

Importantly, these sorts of benefits cannot be realized without initial investment, and this is another example where racial bias influences outcomes. Take the examples of sickle cell disease and cystic fibrosis. Both can be found across many racial groups, but sickle cell is most commonly found in people from areas where malaria is endemic, whereas cystic fibrosis is most frequently found in people of European ancestry. As such, people taking a broad genetic essentialist view tend to think of these primarily as Black (sickle cell) and White (cystic fibrosis) diseases. Given this context, it is unsurprising to learn that though there are roughly three times as many sickle cell patients as cystic fibrosis patients in the United States, there is disproportionately high investment in cystic fibrosis research, both at the National Institutes of Health (NIH) and private foundation levels (figure 15). Together, the average research funding per affected person is \$943 for a sickle cell patient versus \$10,592 for a cystic fibrosis patient, which represents a 1,123 percent enrichment for the "White disease" (Farooq et al. 2020). This underinvestment then goes on to influence the time course of progress in research and development. The cystic fibrosis community is already the beneficiary of transformative therapeutic interventions, while the sickle cell community lags behind. Thankfully, though, there are very promising sickle cell treatments that have just recently been approved by the Food and Drug Administration (FDA), so hopefully this gap will begin to close in the near future.

Realizing equitable benefits of research goes beyond simple funding levels and extends into the nuts and bolts of study design, as well as publication practices that influence what research is publicly shared. As scientists grapple with research, development, and publication biases, working groups of experts, including foundational scientists, ethicists, clinicians, and editors, have come together to articulate best practices in these spheres (Brothers, Bennett, and Cho 2021; Committee on the Use of Race, Ethnicity, and Ancestry as Population Descriptors in Genomics Research 2023). The groups are in agreement that they are not recommending that we stop using racial categorization in research, but rather that we include

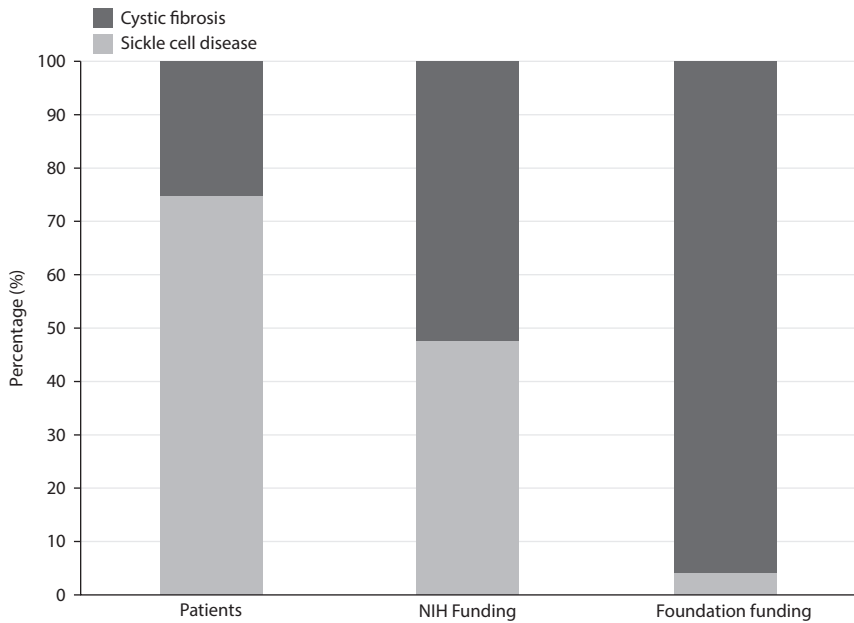


Figure 15. Comparison of prevalence of conditions with the fraction of federal and foundation research funding for the same conditions. When viewed across the populations most impacted by these conditions, these data exemplify underinvestment in communities of color. (Data from Farooq et al. 2020)

important guardrails to ensure that it is interpreted as a social variable only. It remains important to understand race-based disparities, but those must not be equated with biological ones. To this end, the experts have arrived at similar guidance for scientists with respect to the responsible use of population descriptors in scientific research (Text Box 1). They center the significance of highlighting the social and environmental contributions to defined populations; avoiding harmful terminology with connotations of hierarchy or superiority; using precision in language to prevent conflating race, ethnicity, and ancestry; partnering with invested communities; and transparently reporting all these methods.

The examples and principles we have discussed thus far in this chapter largely focus on populations defined by race, ethnicity, or ancestry, but many of the same issues plague studies that characterize people based on other criteria, such as sex, gender identity, sexual orientation, and

POPULATION DESCRIPTORS

The following recommendations from the editors of a genetics research journal (Brothers, Bennett, and Cho 2021) and a committee of the National Academies of Science, Engineering, and Medicine (Committee on the Use of Race, Ethnicity, and Ancestry as Population Descriptors in Genomics Research 2023) explore best practices in equitable approaches to describing populations.

TAKING AN ANTI-RACIST POSTURE IN SCIENTIFIC PUBLICATIONS IN HUMAN GENETICS AND GENOMICS

(Reproduced from Brothers, Bennett, and Cho 2021)

1. Race should be used in research studies involving health care delivery, etiologies of medical conditions, and health outcomes, but only as a sociopolitical category.
2. Genetic ancestry should not be used as a surrogate for sociopolitical race. Sociopolitical race should not be used as a surrogate for genetic ancestry.
3. Authors should avoid the use of terms that obscure the distinction between sociopolitical race and genetic ancestry, as well as terms that evoke historical conceptions of racial superiority.
4. Variables related to race as a sociopolitical category, as with all scientific variables, should be ascertained and described in a rigorous manner. Whenever possible, self-reported race and ethnicity should be used.
5. Authors should explicitly name racism when it is an underlying factor leading to health disparities and should further describe this racism in terms of its form, mechanisms, etc.
6. Authors examining the genetic contribution to health disparities should avoid framing health disparities in reductive terms.
7. Given the underrepresentation of Black, Latinx, Asian, and other non-White populations in genetics and genomics research, editors and reviewers should prioritize manuscripts with strong representation of these groups, even when findings replicate earlier findings in White populations.
8. Authors should carefully avoid structuring data tables and other representations of data in such a way as to treat White populations or European ancestry groups as the “normal” in group comparisons.

USING POPULATION DESCRIPTORS IN GENETICS AND GENOMICS RESEARCH

*(Reproduced from Committee on the Use of Race, Ethnicity, and
Ancestry as Population Descriptors in Genomics Research 2023)*

1. Researchers should not use race as a proxy for human genetic variation. In particular, researchers should not assign genetic ancestry group labels to individuals or sets of individuals based on their race, whether self-identified or not.
2. When grouping people in studies of human genetic variation, researchers should avoid typological thinking, including the assumption and implication of hierarchy, homogeneity, distinct categories, or stability over time of the groups.
3. Researchers, as well as those who draw on their findings, should be attentive to the connotations and impacts of the terminology they use to label groups.
4. Researchers conducting human genetics studies should directly evaluate the environmental factors or exposures that are of potential relevance to their studies, rather than rely on population descriptors as proxies. If it is not possible to make these direct measurements and it is necessary to use population descriptors as proxies, researchers should explicitly identify how the descriptors are employed and explain why they are used and are relevant. Genetics and genomics researchers should collaborate with experts in the social sciences, epidemiology, the environmental sciences, or other relevant disciplines to aid in these studies, whenever possible.
5. Researchers, especially those who collect new data or propose new courses of study for a dataset, should work in ongoing partnerships with study participants and community experts to integrate the perspectives of the relevant communities and to inform the selection and use of population descriptors.
6. Researchers should tailor their use of population descriptors to the type and purpose of the study, in alignment with the guiding principles, and explain how and why they used those descriptors. Where appropriate for the study objectives, researchers should consider using multiple descriptors for each study participant to improve clarity.
7. For each descriptor selected, labels should be applied consistently to all participants. For example, if ethnicity is the descriptor, all participants should be assigned an ethnicity label, rather than labeling some by race, others by geography, and yet others by ethnicity or nationality. If researchers choose to use multiple descriptors, each descriptor should be applied consistently across all individuals in that study.
8. Researchers should disclose the process by which they selected and assigned group labels and the rationale for any grouping of samples. Where new labels are developed for legacy samples, researchers should provide descriptions of new labels relative to old labels.

disability. Like race, ethnicity, and ancestry population descriptors, if these additional group labels are not used responsibly, they have the potential to create and exacerbate between-group disparities. In the final sections of this chapter, we will consider examples involving sex, pregnancy, and disability that demonstrate similar challenges facing scientists working in these areas and the extent of consensus recommendations that exist thus far.

For many years, when it came to sex, scientists viewed research in males as the gold standard. Females were considered too biologically complicated, given menstrual cycles and fluctuations in hormones. Thus, it was routine to design clinical studies entirely with males such that a drug could be approved by the FDA without a single study including females. In fact, in 1977, there was an FDA policy codifying the specific exclusion of people of childbearing potential in phase I and phase II clinical trials, which are designed to test the safety and efficacy of drugs (U.S. Food and Drug Administration 2018). This policy was not reversed until 1993 with the passage of the Revitalization Act, and even with this reversal, pregnant people are still not included routinely in these studies for things as fundamental as flu and COVID vaccines.



As the FDA previously excluded females from participating in clinical trials, it turns out that folks with disabilities are intentionally excluded from clinical trials as well. This doesn't come in the form of a national restriction, but rather at the level of individual clinical trials. Importantly, recognizing the importance of inclusion of a community on its own is not enough to foster participation (Sabatello 2018; Lewis et al. 2023). With respect to increasing representation of people with disabilities, trust issues exist between clinical and scientific professionals and disabled individuals, in part because of the contrasting medical and social models of disability that we discussed in chapter 7. Intentionally addressing barriers to participation, especially accessibility concerns such as transportation, space in health care and research facilities, and communication accommodations, must be incorporated up front in order to foster a true sense of inclusion (Sabatello, Chen et al. 2019; Sabatello, Blake et al. 2019).

Furthermore, study protocols must articulate inclusion and exclusion criteria to systematically decide whom they enroll in their research. A critical element of human research, as we've discussed, is informed consent. In this context, informed consent is sometimes used as a rationale for excluding individuals with disabilities from participating in studies. This can have truly detrimental effects, for instance, by preventing people with Down syndrome from participating in studies related to Alzheimer's, for which they are at elevated risk (DeCormier Plosky et al. 2022). Similarly, if the informed consent procedure is not accessible to people who are hard of hearing or blind, individuals with these disabilities will be excluded from the study. Of note, these *de facto* exclusions are often a simple matter of not providing accommodations for people with disabilities rather than a specific reason why a person may not be a good match for the research study itself. Furthermore, many studies have very broadly defined exclusion criteria that rely on scientist discretion around things like perceived ability to comply with study procedures, vulnerability, or complicating health conditions (DeCormier Plosky et al. 2022), opening the door for investigator bias to also prevent participation of folks with disabilities. Scientists could instead prioritize inclusion by allowing study participants to use supportive people or devices to facilitate informed consent and study protocol adherence. This might involve having a sighted companion or a text to voice device for a blind person or a trusted adult who can provide decision-making support for a person with intellectual disability during informed consent. Or it may mean training a partner to perform injections if someone with a physical limitation can't inject themselves during a study. There are many ways to envision supporting the inclusion of people with disabilities in research studies, and broad exclusion criteria simply reflect that this is not a priority of the scientists crafting the study protocols.

Just as omission of other populations from scientific research creates harmful knowledge gaps, the same holds true for the disability community. With the 2022 recommendations for the NIH to reckon with its ableist procedures and practices (Diaz Brinton and Bernard 2022), there is potential to address this concern. For instance, the recommendations include expanding efforts to engage with the disability community, which will allow folks with lived experience to inform NIH efforts. Furthermore,

the recent decision to designate folks with disabilities as a health disparity population, which will create additional funding opportunities and incentives to spur on research that can benefit the disability community, is an important step in the right direction. This recommendation goes hand in hand with the suggestion to create infrastructure to support people endeavoring to include folks with disabilities in their research (Swenor and Deal 2022). Together, these steps can begin to build a more complete understanding of the myriad ways in which people with disabilities navigate their health and access to health care.



An important community we have not yet discussed in the context of inclusion in research is the LGBTQ+ community. The disparities faced by members of this community are unfortunately poorly understood relative to the other examples we have considered so far. This is in part due to lack of inclusion in fundamental research studies, as with other populations, but it is even more basic than that. It comes down to the reality that people with identities that reside outside of the limited binary, cisgender, heterosexual categories have simply not been counted in many studies, let alone intentionally included (Streed, Lett, and Restar 2023). In a statement that should not be as provocative as it was, a National Academies of Science, Engineering, and Medicine Committee noted that “people deserve to count and be counted” (Committee on Measuring Sex, Gender Identity, and Sexual Orientation et al. 2022). Similar to recommendations from the previously mentioned population descriptors report, this report and others (Heidari et al. 2016) also focus on the importance of precision of language distinguishing biological criteria of sex and social contributions of gender. Additional significant principles from the National Academies report include respect for identity and autonomy, collection of necessary data only, beneficence, and importance of privacy and confidentiality.



We contain multitudes, and human identity exists in diverse and intersecting spectra. In this chapter, we have examined how numerous aspects

of humanity have been overlooked, leading to exclusion and marginalization of communities. The resulting knowledge gaps contribute to health care disparities for the most vulnerable members of society. One of our most powerful tools against these inequities is data, and in this chapter we have seen themes emerge across these populations about principles related to inclusion and community partnership to begin to address these gaps (Madden et al. 2024). As noted by Swenor and Deal with respect to disabled communities, “Medicine and public health are based on a framework of evidence. Data are necessary to support the allocation of resources and to advance policy. The lack of data on which to base policy and clinical decisions perpetuates inequities for people with disabilities” (Swenor and Deal 2022), but this could have just as easily been written about any historically excluded, minoritized community, and even more so, for people with intersectional identities and needs (Sabatello et al. 2023). In order to truly move forward, we must be able to apply the disability community’s maxim, “Nothing about us without us,” in order to build scientific knowledge on a foundation of trust.

In the absence of an inclusive scientific foundation, there is room for bias and oversimplification about what is considered a “normal reference” to influence the practice of medicine. These issues have come up repeatedly in the form of race-based medicine, which we will explore in the next chapter.

12 Race-Based Medicine

In chapter 11, we explored what happens when communities are excluded from research and the many ways in which results in members of those communities getting left behind. However, even well-intentioned researchers who make an effort to include minoritized communities sometimes inadvertently create harmful perspectives in the way they frame their research questions. This can absolutely be the case with work focused on describing race-based disparities, even when the research questions are carefully constructed. This often results when people who follow up on work that compares groups of people take an essentialist stance and consider only biological roots of differences. Consequently, the health disparities experienced by historically excluded communities are then pathologized and interpreted to result from some inherent biological “defect” (Cerdeña, Plaisime, and Tsai 2020).

Hopefully by this point in the book, you are able to look back on things we’ve learned about genetic essentialism (chapter 3) to poke holes in this type of thinking. And I hope you also know that I’m not advocating that we stop conducting research that examines disparities. Rather, we must do the harder work of contextualizing these studies and considering the multifactorial reasons—biological, environmental, and systemic—that

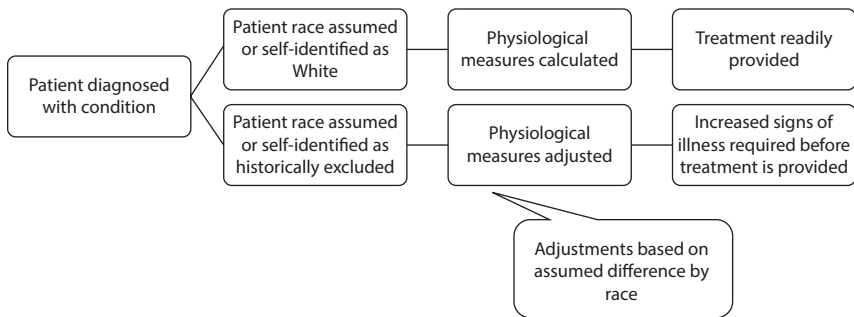


Figure 16. Race-based medicine: This flowchart demonstrates how White patients serve as the reference population upon which treatment of patients of color is modified.

contribute to observed disparities (Committee on the Use of Race, Ethnicity, and Ancestry as Population Descriptors in Genomics Research 2023).

To that end, disparities in health-related outcomes for people of different racial backgrounds speak to this complexity of contributing factors, but unfortunately, it is frequently flattened into a single biological dimension. Race-based medical practice can grow out of this oversimplification. Race-based medicine itself arises when groups are provided different clinical management based on their race, even in the absence of specific documented biological differences. An example of how this might play out is shown in figure 16.

In this model, patients of color are pathologized relative to the White reference, and adjusted markers of disease progress often underestimate disease burden carried by patients of color. This leads to issues around access to treatment and ultimately to poorer health outcomes for Black and Brown patients. To answer whether there is any true biological difference underlying these observed disparities, we will begin by examining the premise of whether there is any scientific basis for race-based medicine, both for single gene disorders and for multifactorial conditions.



Before we completely discount a race-based approach to medicine, let's consider if there is any legitimate rationale for differential treatment based

on patient race. After all, these treatments are generally based on an initial observation of some difference between the groups, so might there be a biological cause for that difference that substantiates a race-based approach? Let us consider this question in the context of two contrasting forms of genetic disease: single gene disorders and polygenic conditions (i.e., common complex disease).

To answer the question about whether racialized thinking about single gene disorders is accurate, we need to understand if certain conditions are only present in specific ancestries. Taking the example of cystic fibrosis, a single gene disorder resulting from pathogenic variants in the *CFTR* gene, CF is often thought of as a White disease. While it is true that there are more people who are carriers of a pathogenic variant that causes cystic fibrosis in European populations (1/28–1/29) than in African (1/65–1/132) and Asian (1/90–1/174) populations (Lazarin et al. 2013; Lopes-Pacheco 2016; Bono-Neri, Romano, and Isedeh 2019), it is important to note that Black and Brown populations are not free of cystic fibrosis altogether. Also, we know from chapter 4 that populations are not homogeneous, and as with many genetic conditions, there is not a single pathogenic *CFTR* variant that is associated with cystic fibrosis in any population. *CFTR* (or any genetic) variants differ in frequency and predominance across populations (Bono-Neri, Romano, and Isedeh 2019), which also influences how readily the variants are detected by genetic testing designed for and based on data from White populations. Thankfully, awareness of this issue has informed updated genetic testing protocols (Gregg et al. 2021) that now recommend DNA sequencing-based approaches that are technically poised to capture a wider array of variants, not just common variants found in single ancestries. Of course, we must acknowledge, as previously described in chapter 11, that the variants identified in communities of color will more often be variants of uncertain significance, creating a combination of situations that will hinder diagnosis of cystic fibrosis in patients of color. Furthermore, it is difficult for providers to shake the learned association of cystic fibrosis with White patients, such that newborns with false negatives or out-of-range test results suggestive of a cystic fibrosis diagnosis often receive delayed diagnostic confirmation and follow-up care if they are from minoritized backgrounds (McColley et al. 2023; Szabo 2024), and in one particularly

egregious case, Terry Wright, a Black man, was not diagnosed until age 54, eight years older than the average life expectancy of a child born with CF in 2017, the year he was finally diagnosed (Rubin 2021). Twenty years earlier, his physician said, “If you weren’t African American, I’d say you have the classic symptoms of cystic fibrosis.” According to Terry’s wife Michele, “That biased thinking caused him years of suffering and almost cost him his life several times over.”



Though we often think of genetic disease as those conditions determined primarily by a single gene, like cystic fibrosis, the vast majority of health-related traits are complex, influenced by multiple genes and environmental factors as well. Racialized thinking has been applied to both single gene disorders and common, complex diseases. To understand the ways in which genetic essentialism has reified racialized frameworks for multifactorial conditions, we can begin by considering the trait of blood pressure and hypertension. As with other complex traits, blood pressure results from a combination of protective genetic variants, predisposing genetic variants, environmental factors, behavioral elements, and systemic variables (Petronio, Brockman, and Dron 2021). The combined impact of these factors can determine where on the continuum one’s blood pressure lies, from low to high.

When it comes to blood pressure, there is a documented disparity between Black and White patients, with Black patients living with hypertension at disproportionately high levels compared to White patients. In an effort to explain this observation through a genetic essentialist lens, some scientists have proposed a salt hypertension hypothesis. People who subscribe to this hypothesis suggest that the Transatlantic Slave Trade was an evolutionary force that selected for survivors of the perilous journey who were able to retain salt, thereby also allowing them to retain water throughout their forced migrations, which exposed them to extreme heat, pathogens that caused diarrhea and vomiting, and dehydration. In this hypothesis, on the African side of the journey, the population included people with a range of salt-retention phenotypes, but by the end of the journey, the remaining survivors on average had much higher

salt-retention profiles. As the hypothesis goes, the individuals with the high salt-retention trait were also the ancestors of African American populations in the United States today, so the increased salt retention among Black Americans now, together with modern Western diets, leads to hypertension. Upon closer inspection, however, this hypothesis falls apart. Even if the horrific conditions of the Transatlantic Slave Trade resulted in increased survival of enslaved people with the ability to retain higher amounts of salt, this does not take into account that it is still a polygenic trait, and those individuals would harbor a mix of protective and predisposing genetic variants. In subsequent generations, the combinations of variants will randomly shuffle, and descendants with decreased salt retention will reemerge, even if at reduced frequency. An evolutionary selective pressure operating over a brief time scale, such as a single journey, is not enough to result in an irreversible change to the population. Rather, selective pressure that causes lasting evolutionary change to populations generally operates over multiple generations.

When it comes to hypertension, there are many environmental and behavioral factors that contribute to the development of high blood pressure. For instance, discriminatory lending practices known as redlining have created functional segregation of neighborhoods in many parts of the United States. The places where Black folks were able to afford housing were often the neighborhoods where they would have increased exposure to harmful chemicals, lack of access to fresh foods, and fewer safe areas in which to get out and move their bodies. These examples of structural racism and others contribute to poor health outcomes for Black populations, and one area in which this manifests is hypertension (Williamson 2021). Taken together, the significance of known genetic risk factors in explaining the disproportionate existence of hypertension in Black populations is quickly replaced by observations about the influence of social determinants of health.

Unfortunately, however, when health providers are determining how best to treat patients with hypertension, in spite of a lack of evidence for biological differences between Black and White hypertension patients, the therapeutic guidelines have long been stratified by race (Flack and Buhnerkempe 2022). Importantly, recent evidence demonstrates that these differential, race-based hypertension treatment methodologies are

actually not working well for anyone and are certainly not closing the health disparity gap between Black and White patients (Egan et al. 2022; Holt et al. 2022). New guidelines are needed that are based on treating the root causes of the hypertension itself rather than relying on race as a poor proxy for the actual biological processes at work. As noted by Dr. Francis Collins, one of the leaders of the Human Genome Project and former director of the National Institutes of Health, “A true understanding of disease risk requires a thorough examination of root causes. ‘Race’ and ‘ethnicity’ are poorly defined terms that serve as flawed surrogates for multiple environmental and genetic factors in disease causation, including ancestral geographic origins, socioeconomic status, education, and access to health care. Research must move beyond these weak and imperfect proxy relationships to define the more proximate factors that influence health” (Collins 2004). Steps like these will help fill in our gaps in understanding, such that we can move toward an individualized approach to care that will best serve patients, rather than painting them as members of a homogenous monolith. But even as we begin to take steps in that direction by defining polygenic estimates of biological risk, race-based frameworks of care remain deeply entrenched in providers’ approaches to patient care (Kerman et al. 2023).



Now that we have a sense of the limitations of categorization of human health on the basis of race, we can properly consider the ramifications and unintended consequences of race-based medicine using three illustrative examples: treatment of heart failure, assessment of kidney function, and estimation of neurological condition. These examples will help us assess the potential for harm when treating individual patients using methods informed by race in lieu of biological markers (Roberts 2012). In order to consider the ramifications of treating patients using race-based medicine, let us begin with the case study of BiDil (Kahn 2013). BiDil is a combination of two drugs used to treat heart failure and repackaged as a single pill for ease of administration. It originally failed a clinical trial and was not approved by the FDA as a general therapy for heart failure, but BiDil demonstrated significant benefit when the data were reanalyzed focusing on

the Black research subjects in the study. This observation was enough to launch a new clinical trial focused exclusively on Black research subjects, the 2004 African American Heart Failure Trial (A-HeFT) (Taylor et al. 2004). Folks who self-identified as Black, including people of mixed ancestry, were enrolled in the study, and the positive effects of the combination of medications in BiDil resulted in a dramatic 43 percent improvement in survival. This result was so positive that they actually halted the study to allow folks in the placebo group immediate access to the medication as well. Interestingly, BiDil's patent protection was set to expire in 2007, so the company NitroMed quickly pivoted to take out a new patent, a race-based patent on BiDil that would protect their market share through 2020 ("Illuminating BiDil" 2005; Saul 2005). The marketing of BiDil as the first race-based medicine occurred before more proximal causes of this observed benefit could be delineated. As such, Black patients whose heart failure may not stand to benefit from BiDil would be lumped in with the majority of Black patients; similarly, patients from other backgrounds who may benefit from BiDil would not be prescribed the medication based solely on their race. Given the exclusive focus on self-identified Black research subjects in the A-HeFT, it was not determined if there were other patients who might benefit from BiDil, nor was it determined what the predictors of responsiveness to BiDil actually were. This false reification of the biological nature of race can cause direct harm to patients by preventing their access to effective medications, much as we saw in the case of race-based approaches to hypertension treatments. In the end, corporate convenience did not benefit NitroMed, as many patients and physicians were skeptical about the so-called "race drug." The company never achieved profitability and was subsequently acquired in 2009, well before its race-based patent ran out (Krimsky 2012).



In addition to prescription decisions, as with BiDil, quantification of physiological function using validated calculations is also an area that has been "corrected" by race-specific modifiers. An example of this type of approach is best demonstrated using eGFR, or estimated glomerular filtration rate, a way of quantifying kidney function. Direct measures of kidney function

are laborious, invasive, and costly, so health care providers have instead routinely used methods that can estimate kidney function more conveniently (Trang 2022). The equations that are employed in this estimation frequently factor in serum creatinine, age, and sex—all biological variables—as well as race, which we know to be a social construct. The reason race is included is because it is being used as a proxy for muscle mass (Mohottige, Olabisi, and Boulware 2023). Creatinine is a by-product resulting from normal breakdown of muscle tissue, and other factors, from meat consumption to exercise habits, can also influence levels of serum creatinine. When kidneys are functioning well, this protein is not detected at elevated levels in the serum. In the case of Black patients, however, it is assumed that the presence of serum creatinine simply reflects higher initial muscle mass rather than kidney dysfunction. The use of race as a proxy for muscle mass is so crude as to seem ridiculous. Just as with all populations, Black folks exhibit the full range of body types, and to assume that their social identity somehow ties to increased muscle mass is absurd. As described, the Black coefficient mathematically adjusts the eGFR equations' output to suggest better kidney function for Black patients, thereby contributing to delays in diagnosis of chronic kidney disease, eligibility for certain lifesaving medications, referral to specialists, initiation of dialysis, or listing for kidney transplantation. Indeed, treatment can be delayed so substantially that health declines to the point the patient is no longer eligible to be on the transplant list. Not only does this contribute directly to increased mortality of Black patients from chronic kidney disease, but the eGFR underestimation of kidney disease in Black individuals also resulted in their increased incarceration during the peak of the COVID-19 pandemic (Goldstein 2022). While other folks were being released for medical reasons, Black folks defined by the limitations of the eGFR calculations were deemed healthy enough—by judges, not health professionals—to remain incarcerated.

Since that time, new information has come to light, identifying ways to estimate kidney function accurately and independently of race. The National Kidney Foundation and American Society of Nephrology reevaluated the data in 2021, and the task force recommended that a race-free version of the eGFR CKD-EPI equation be implemented immediately (Delgado et al. 2022), citing evidence that using an updated version of the

equation with more biological markers improves accuracy without the need for a race-based coefficient (Hsu et al. 2021; Inker et al. 2021). However, it will take time for clinics to embed these updated practices into their electronic medical records and patient care.

It is also important to acknowledge that this is just one example of clinical calculators, many of which continue to use a race “correction” as part of their approach (Vyas, Eisenstein, and Jones 2020). The nephrology community has made rapid, anti-racist progress in updating their methods for estimating kidney function, but many other fields of medicine continue to employ race-based calculations in their scope of practice, affecting everything from vaginal birth after Cesarean to cancer treatment and surveillance, as well as pulmonary function tests (Diao et al. 2024). There is a lot of work remaining to be done to fully decolonize medical practice.



For our final case study of race-based medicine in this chapter, we will consider race-norming and its use in assessing neurological function. Cognitive testing strategies include a defined cutoff score for dementia, but in the context of race-norming, different cutoffs are used for members of different races. Race-norming, similar to race-based correction as with eGFR, assumes a different baseline between members of different races. In the case of assessments for cognitive function, the neuropsychological tests themselves are considered biased instruments, originally written by White doctors conducting research on middle-class White populations (Hobson 2021), explaining why the distribution of Black patients’ cognitive scores skews lower than the scores of White patients. To properly demonstrate decline, trends in cognitive scores should be measured over time for a specific individual, but often the baseline scores are not available because assessments were not completed prior to the onset of symptoms. Therefore, to demonstrate decline, it is instead calculated relative to “demographic peers” as defined by age, educational attainment, and apparently, race. In other words, an individual’s decline is measured relative to a race-specific average score rather than a population-wide, race-neutral average.

To examine the impact of race-specific averages in race-norming, let’s study figure 17, which shows two overlapping but offset distributions of

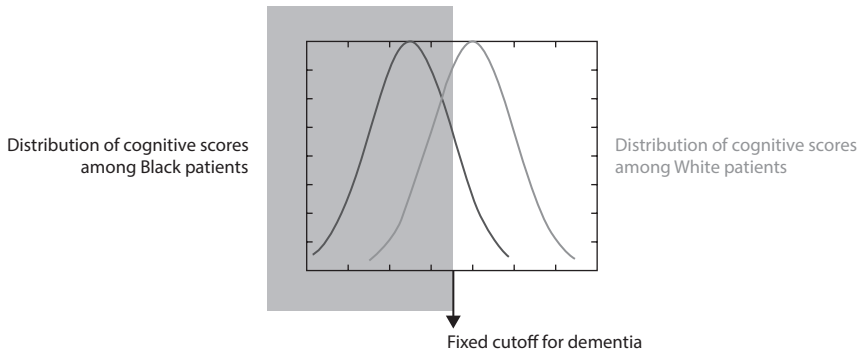


Figure 17. Fixed cutoff for dementia: When a race-neutral cutoff is used for dementia, a greater proportion of Black patients meets the clinical criteria of cognitive impairment.

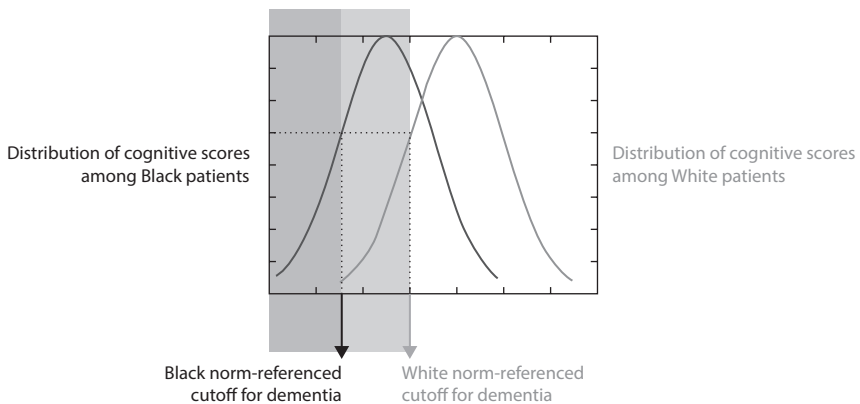


Figure 18. Norm-referenced cutoff for dementia: While the same proportion of Black and White patients are considered affected by dementia under this definition, the Black patients must score substantially lower on their cognitive assessment to be diagnosed.

cognitive assessment scores when comparing Black and White patients. In a scenario where race norms are not applied, a fixed cutoff for dementia holds true across populations. Using this fixed cutoff, we see that a greater proportion of Black patients meet the criteria of dementia than that of White patients. In contrast, figure 18 demonstrates the consequences of using race-specific norms, in which case the proportion of each population

that meets the definition of dementia remains the same, but in order to be considered affected by dementia, a Black individual must actually have much lower cognitive assessment scores than a White person.

The use of race norms was meant to correct, in part, for the biased origins of the neuropsychological tests, and it was also meant to protect Black patients from excessive medical intervention. However, when these scores and associated diagnoses of dementia were used to test the neurological health of athletes for the purpose of financial settlement, the implications were different. Specifically, this method of cognitive assessment was used for players of the National Football League who were seeking payment of claims related to settlement of a class action lawsuit. In the settlement, the NFL agreed to pay up to \$5 million to former players who developed neurological conditions, including Lou Gehrig's disease, Parkinson's, Alzheimer's, dementia, and death with chronic traumatic encephalopathy. As part of this settlement, the former players went through rigorous neuropsychological testing, which can easily cost thousands of dollars and is not always covered by health insurance. Therefore, the NFL hired psychologists to administer these tests for free to people who were part of the settlement. Damningly, some of the psychologists have indicated that they were specifically instructed to use race-norming methods, thereby making it more difficult for Black players to qualify for settlement payments (Hobson 2021). Given the NFL's history of disrespecting Black bodies, this policy just adds insult to injury in a sport where 50–70 percent of the players are Black. One attorney noted that White players' claims are approved at about two to three times the rate of the Black players' claims, demonstrating that Black players are actively being prevented from accessing payments meant to recognize the pain visited upon them and their loved ones in the name of sport. Thankfully, the settlement was sent back for renegotiation, and now the NFL has been instructed to eliminate race-norming from its settlement evaluation process (Associated Press 2021).



Each of the examples we have deconstructed in this chapter demonstrates how falsely equating race with biological traits systematically disadvan-

tages already marginalized individuals and further exacerbates existing health care disparities. To promote equitable access to health care, these practices require careful revision to ensure clinical decisions are based on biological criteria rather than social constructions. Using race in these scenarios is a harmful oversimplification. As Dorothy Roberts put it, “You see, race is not a biological category that naturally produces these health disparities because of genetic difference. Race is a social category that has staggering biological consequences, but because of the impact of social inequality on people’s health” (Roberts 2015).

Not only is it essential for us to take a careful look at existing medical frameworks like race-based medicine through the lens of inclusive practice, but we must apply similar critical thinking to developing technologies to anticipate exclusionary practices prior to implementation. In the next chapter, we will consider a few examples within the rapidly evolving field of genomic medicine to consider frontiers on which new disparities are beginning to take root and what we might do to interrupt these cycles of harm.

13 The Future of Genomic Medicine

I have a confession to make: I was never all that into history while growing up. I guess my younger self failed to see the connection between events of the past and contemporary issues. But honestly, that was incredibly shortsighted. The truth is, once you know how to recognize bias and echoes of past harms, you start to see it everywhere. Maybe you are already feeling that way at this point in the book, particularly as we have just considered the ways in which essentialist thinking has made its way into modern clinical practice. Over the course of this chapter, I want to highlight some emerging genomic technologies to demonstrate how these technologies are at the tipping point between the promise of health and benefit for all versus for just the elite few. Importantly, at a junction like this, we can't just hope for equitable access to these emerging technologies. Given the pervasive existence of bias, both implicit and explicit, without deliberate action to ensure equity in the practice of genomic medicine, the default will be to favor those with preexisting privilege.

All this is occurring in the context of a field in which it is incredible to consider the number of new diagnostics, technologies, and treatments that have been developed in this revolutionary period of genomic science. One recent study estimated that there are 10 new genetic tests entering

the market *daily* (Phillips et al. 2018). In this chapter, we will do a deep dive into a few examples of the types of technologies that stand to transform the practice of medicine, while also highlighting the challenges to ethical and equitable implementation of these developments. Specifically, we will consider new horizons in genetic testing, screening of newborns and embryos for genetic traits, and treatment strategies for genetic disease.



As we first saw in chapter 6, genetic testing initially relied on race-based criteria to access risk assessment, but newer approaches recommend population-neutral access to genetic testing (Gregg et al. 2021). Nevertheless, because clinical testing results must be compared to reference genomes, and because existing genetic data still heavily overrepresent European ancestries (Sirugo, Williams, and Tishkoff 2019), the probability of identifying variants of uncertain significance is higher for patients of color (Landry and Rehm 2018). These uncertain clinical testing results also plague emerging approaches to use genomic methods in newborn screening.

The most widely adopted forms of newborn screening rely on testing methods that quantify molecular markers correlating with the onset of disease. Predictive testing prior to the emergence of symptoms is more rarely used. The advantage of biomarkers for disease onset is that they don't have the same reference population limitations as genetic testing, whereas predictive genetic methods are only as good as the reference genomic data available for any given gene. However, on the plus side, not having to catch the patient just as their symptoms become evident takes timing out of the equation, which may ultimately improve the ability to capture a larger fraction of newborns susceptible to genetic disease. Furthermore, some argue that the ability to extrapolate benefits to the parents should also be considered, for instance, the ability to detect genetic cancer susceptibility syndromes in parents by examining the genome of their child (Molteni 2023a). To fully understand the relative pros and cons of newborn sequencing as a form of population-wide screening, a series of studies have been launched that seek to ascertain the impact and cost of sequencing healthy newborns, to develop ultrafast

turnaround for sequencing of babies in the newborn intensive care unit (NICU), to compare efficacy of traditional versus sequencing-based newborn screening methods, and to examine the benefits of these technologies for members of populations historically excluded from medicine. Preliminary results highlight the benefits of a genome sequencing-based approach for acutely ill newborns, demonstrating faster time to diagnosis, six-figure savings in health care costs, and improved outcomes (Heger 2019; Maron et al. 2023). In contrast, there is not a strong case for sequencing-based methods to displace traditional newborn screening methods just yet. There remain numerous cases of missed diagnoses when relying on exome sequencing alone because things like deletions and non-coding variants (outside of the region of the gene that directly encodes the protein) can easily be missed. Furthermore, when testing was done in a diverse population in California, fully one-third of the newborn sequencing variants were variants of uncertain significance. Overall, exome sequencing had a sensitivity of 88 percent, compared to the 99 percent sensitivity rate of traditional mass spectrometry methods (Adhikari et al. 2020). As such, sequencing may be helpful to verify mass spec results but not yet as a first-line newborn screen.

Though this technology is still in early stages, we can already see the relative benefit for well-characterized populations (i.e., White populations) relative to populations of color. At least there are already tried-and-true newborn screening methods that largely avoid these drawbacks. However, if we are to look beyond the newborn period and consider a potential future in genomic medicine where newborns are sequenced and this information is used to inform medical decisions throughout the life cycle, then addressing the many cases of uninformative sequencing results and their disproportionate impact on patients of color will be essential to ensure equitable access to care. This is already a priority for people taking a critical look at the original reference genome by assembling a pangenome that has greater representation of human genetic variation across populations (Attwaters 2023; Liverpool 2023). Availability of a tool like the pangenome is likely to significantly reduce the uncertain results often obtained when carrying out genetic testing in patients of color.



Another growing area of genetic screening is even earlier in development, that is, in conjunction with assisted reproduction at the preimplantation stage. In chapter 6, when we discussed prenatal screening, we focused specifically on testing during ongoing pregnancies, and in chapter 7, we considered the ramifications of testing for conditions associated with disability during the preimplantation and prenatal stages, effectively making it possible to choose which lives were “worth” bringing into the world. As we came to see, this type of decision-making opens the door for the influence of bias. I want us to now return to that context to reflect on the potential ramifications of combining preimplantation genetic screening with the evolving polygenic risk score market in an emerging technology termed *embryo screening*.

Embryo screening is a broader, more consumer-focused version of preimplantation genetic diagnosis (PGD). One of the key distinctions between the two is that PGD uses clinical genetic testing strategies to assess health-related genetic risk for embryos that may subsequently be implanted in a gestational carrier. These clinical testing strategies have robust predictive ability for clinical outcomes, meaning the tests have been approved by regulatory agencies and recommended by professional societies for use in the prenatal setting specifically because they offer reliable insight into pregnancy outcomes. It is important, however, to recall the caveat that they do overlay a medical framework over disability status, which can inject an element of bias into the risk assessment, as described in chapter 7. In contrast, the polygenic risk scores that are now starting to be offered as add-ons to assisted reproduction fertility services have less accepted clinical validity and are starting to dip their toes into social and behavioral traits.

When we discussed these types of social and behavioral genomic studies in chapter 8, we saw that the traits were often viewed through a genetic essentialist lens that overestimated genetic contributions while underestimating environmental and social influences (Meyer, Appelbaum et al. 2023). It is critical to recall this limitation in light of the increasing pairing of this screening strategy with assisted reproduction. For instance, in one case, a company has been offering screening for a range of traits, from diabetes and cardiovascular risk to intelligence and height, to prospective parents (Regalado 2019b). These estimates are calculated based on

polygenic risk scores created using data from White populations, so, as we saw in chapter 11, the screening will be most beneficial to White consumers, given the documented poor transferability of these calculations across populations (Lewis and Green 2021; Grebe et al. 2024). Furthermore, I hope the range of traits included on this list also caught your attention because it spans from health-related traits to things that some parents may simply find more desirable. For some of those traits that many would consider more questionable additions to the list, the company initially indicated that they would only report the embryos predicted to be in the lowest 2 percent of the distribution, for height or intelligence, for instance (Regalado 2017), but they have since moved away from testing of some of these more controversial traits (Ball 2021). This seems to be an attempt to provide some ethical guardrails, to prevent enhancement of embryos while prioritizing their health, but it's never that simple, especially when it is possible for the consumers to download their embryos' genetic data and do whatever analyses they please (Goldberg 2022).

Let's take the case of intelligence and examine it more closely. As we noted, the polygenic score is based on studies that look at things like IQ and educational attainment because those are more easily measured than actual intelligence. These measures are fraught with their own biases, as we discussed in chapter 8, and certainly do not accurately predict one's innate intelligence or capture the full range of influences on intelligence. At this point, the science behind this score is probably more like a crystal ball than a proper clinical genetic test. Furthermore, when these estimates are pitched to prospective parents, the "gains" in choosing the embryo with the highest polygenic score are frequently overstated, portrayed in relative rather than absolute terms. This is in part because the expected range of phenotypes is actually rather limited when considering the potential genetic combinations from a single pair of prospective parents rather than all the possible combinations within the entire gene pool (Turley et al. 2021). Moreover, if parents were selecting embryos for implantation on the basis of the potential intelligence of their future child, they may not be factoring in unintended consequences of making this selection. For instance, while focusing intently on the scores for intelligence, one might inadvertently select an embryo at increased risk for cancer because those scores aren't being carefully monitored. Or perhaps those genes that contribute to intelligence might also have

a role to play in psychiatric traits, whereby selecting for intelligence may also increase risk for bipolar disorder, as the math seems to suggest. It just isn't well enough understood to make these calls with any certainty, despite the marketing tagline "choice over chance." Just because a choice is being made for one trait, it doesn't mean that everything else will fall into place. Indeed, for the family that made the problematic decision to use the intelligence test option to have a child without autism after their first two children were diagnosed with autism, they are likely to find that not only is intelligence not the same thing, or even the opposite thing, as autism, but what is actually within our control is vastly overestimated (Regalado 2019b).

Though we have been focusing on the scientific ramifications of embryo selection, it is essential to also deeply consider the ethical and social consequences of this technology. We've already mentioned that these scores are most robust in their predictions for White folks. Furthermore, some of the social and behavioral traits included in these "embryo report cards" are highly subjective and align with social constructs of race, privilege, and other identities, thereby amplifying and normalizing racism and other forms of bias. Even given the inherent bias embedded in enhancing these traits, it seems there are many people who would be willing to use these types of genetic services to select children with higher educational attainment possibilities (Meyer, Tan et al. 2023). We also know that the scientific basis for these tests is relatively weak compared to more widely accepted clinical genetic tests. For this reason, health insurers have declined to cover this type of embryo screening, and the American College of Medical Genetics and Genomics has stated that it is premature to apply this approach clinically to embryo selection (Grebe et al. 2024). It follows that the folks who are able to pay for this add-on out of pocket (Carey 2022; Pells 2022), often on top of incredibly high assisted reproduction fees, are going to be members of society who already enjoy every advantage and who are using embryo selection to further cement their legacy and elite social status (Regalado 2023). When the founder of one of these companies gleefully compared his product to the 1997 dystopian film *Gattaca* (Niccol 1997), wherein eugenic norms define social hierarchies, he probably didn't quite realize how on the nose that comparison was.



When it comes to embryo screening, a couple is limited by the preexisting genetic variants present in their genomes, but genome editing technology vastly expands the universe of variants that can be introduced into genomes. Genome editing technology is based on a bacterial defense system known as CRISPR that normally cuts and destroys invading genomes. Scientific ingenuity has tweaked this naturally occurring machinery, allowing scientists to edit genomes in targeted and precise ways, akin to a pair of molecular scissors. Specifically, scientists have engineered this system to target specified sequences in the genome to make defined sequence alterations. This technology has revolutionized how scientists conduct research, and it has started to make its way into clinical spaces as well. Although the possibility of changing genomic sequences in a targeted way sounds almost like a godlike cure for all pathogenic genetic variation, clinical scientists are taking a measured approach to implementing this technology. One of the reasons for this caution is that CRISPR is not a perfect system for genome editing; sometimes it can have off-target effects at unintended genomic sites, and sometimes the edits themselves are imprecise. The prospect of introducing anything other than the intended edit is alarming in the context of clinical care because when toying with human health, “close enough” is not a viable option.

While scientists work at tweaking the molecular precision of CRISPR and related approaches, researchers have started to take careful steps forward to test clinical applications of genome editing technology (Kan and Doudna 2022). One of the first areas in which there has been some success has involved introducing gene edits into stem cells to provide a monogenic correction that restores gene function. Stem cells can mature into multiple cell types in the body, so being able to provide patient-matched stem cells that have been edited to correct pathogenic variants has the potential to restore gene function across a range of tissue types. For instance, in an experimental approach developed in 2021 to treat sickle cell disease (previously described in chapter 4) and beta-thalassemia, scientists devised a genome editing strategy that eliminated a repressor responsible for silencing fetal hemoglobin in adults (Frangoul et al. 2021). The edited cells were introduced into patients using a technique that is very similar to a bone marrow transplant. This procedure allowed fetal hemoglobin to compensate for nonfunctional or missing beta globin in

folks with sickle cell or beta-thalassemia well past the fetal period during which its expression is typically restricted. Beyond the convincing clinical markers of success describing the percent of cells expressing fetal hemoglobin and the like, this approach has been transformative in the lives of folks who volunteered for these early studies in numerous measures of quality of life. For instance, Victoria Gray, the first person to be treated with CRISPR for any genetic condition in the United States, has been able to live a posttreatment life free of the excruciating pain crises and frequent transfusions that used to dominate every aspect of her life with sickle cell (Stein 2019a, 2019b). In fact, the outcomes have been so positive that people have even started to tentatively whisper the word *cure* to describe just how effective this approach is in treating a condition that until recently was life limiting in so many ways.

Even though this revolutionary treatment made history in 2023 as the first genome editing therapeutic to be approved by the FDA (Kolata 2023a), it is not an instant or magical cure for the millions of people living with sickle cell and beta-thalassemia around the world (Kolata 2023b). It is important to consider the numerous logistical challenges involved in accessing this therapy, including the many hidden costs that would be borne by the patient (Kolata 2024). First, there are only a handful of specialized medical centers that are offering this treatment, so accessing this genomic medicine may involve significant amounts of time off and travel for the patient and their loved ones during the lengthy course of treatment. In Jimi Olaghere's case, the full treatment involved eight weeks of blood transfusions, treatment to release bone marrow stem cells into his bloodstream, editing of his harvested stem cells in the lab, chemotherapy to clear his marrows followed by a period of medically induced immunodeficiency, transfusion of his edited stem cells, and inpatient care while the new cells grew for a month and repopulated his marrow. All in, this amounted to 17 weeks of hospitalizations (Olaghere 2023). Importantly, the process of destroying existing stem cells by an aggressive chemotherapeutic regimen also damages other rapidly dividing cells, including gametes, thereby likely rendering the patient sterile (Molteni 2023b). Though fertility preservation measures were offered to some volunteers for the clinical trials, such measures are unlikely to be covered for many patients accessing the treatment through traditional means, especially those

covered by Medicare or Medicaid in the United States, leaving patients with the impossible choice between treating their sickle cell or retaining their ability to have children.



Genome editing is not the only way to restore function of a gene that is carrying pathogenic variation. Other emerging treatments for genetic conditions have been developing concurrently with genome editing approaches, most notably gene therapy. It wasn't clear in 1999 if gene therapy would ever be a viable way to treat genetic conditions. This is because of the tragic and unexpected death of 18-year-old Jesse Gelsinger, who was being treated via gene therapy for his ornithine transcarbamylase deficiency syndrome, a condition that could result in harmfully high levels of ammonia in his system and that would be lethal if untreated. Jesse's treatment was part of a clinical trial to see if providing a functional copy of the *OTC* gene could eliminate the need to maintain a low-protein diet and a grueling regimen of medication, which was usually working well enough to contain the disease for Jesse. He was signing up for the clinical trial not to cure his own disease, but to help infants born with a fatal form of the same disorder. As he said shortly before he died, "What's the worst that can happen to me? I die, and it's for the babies" (Stolberg 1999). In Jesse's case, he had an extreme inflammatory reaction to the weakened adenovirus, an engineered form of the cold virus, that was carrying the healthy DNA to his cells. Understandably, with the tragic loss of such a young life, the gene therapy field stopped instantly in its tracks (Stolberg 1999).

Since the tragic events of 1999, the field eventually regrouped with a renewed focus on safety. Many different viral delivery systems were tested, and approaches to giving gene therapy to specific tissues rather than systemically were developed. And with this measured, deliberate approach, successes started stacking up in treatment of a range of conditions, including sickle cell (again), retinal dystrophy (a form of hereditary blindness), hemophilia (a blood-clotting disorder), and severe combined immunodeficiency, to name just a few (Dunbar et al. 2018; Bulaklak and Gersbach 2020). In fact, the list has grown long enough that it is impossible for me

to attempt to provide you with a completely current list, as it will surely grow by the times these words are in print. But it is also important to acknowledge that these trials and treatments are not without risk. In fact, a more recent death after an adverse reaction to gene therapy administered at high enough dosages to penetrate the musculature and heart of a 27-year-old patient with Duchenne muscular dystrophy (Lek et al. 2023) has the potential to cause the already measured progress to slow considerably again. Importantly, some of the lessons learned in this field can also be applied to CRISPR-based treatments, for example, with respect to safe ways to deliver CRISPR molecular machinery, or another new category of genomic medicine therapeutics, antisense nucleic acids. Indeed, progress in all these related approaches has been in part related to the shared advances that can be combined in new, impactful ways.

Because the origin of the genome editing field is rooted in the early days of gene therapy, scientists working on genome editing have vocally supported efforts to proceed with caution, taking time to consider the ramifications of the work before diving headfirst into clinical applications. Various international bodies have suggested that the most responsible use of genomic editing technologies would follow this example: they focus on somatic cellular edits that cannot be passed on to the next generation, they address variants that have significant impact on health, they are used to restore function rather than enhance function, and they are only used when existing therapies are inadequate (ACMG Board of Directors 2017; Adashi and Cohen 2020; World Health Organization 2021). These guiding principles are actually reactionary in that a renegade scientist, He Jiankui, sent shock waves through the scientific community when he announced that he had carried out genome editing for three newborns to ensure they would be HIV-resistant. Unfortunately, in this case the edits to the *CCR5* gene were not previously tested and known to be safe, patient consent was not properly obtained, safer alternatives were in existence, and the edits were carried out in such a way that they could be passed on to subsequent generations (Regalado 2018). Furthermore, the edits to the *CCR5* gene, while they may decrease HIV susceptibility, may also contribute to increased influenza and West Nile virus susceptibility. There was no technical need to propose CRISPR methods that carried the risks associated with off-target genome edits and impact on other unintended traits

when there were already tried-and-true ways of minimizing the risk of HIV transmission. This really seemed to be much more about hubris and ego than the needs of the patients (Goodyear 2023), and because of his unethical approach to this work, the three babies He claims to have edited are apparently no longer being monitored by scientists. While that outcome addresses the reality that the experimental subjects never truly gave consent, instead believing that the work was actually an HIV vaccine development project, it also is worrisome that the children are carrying variants that are unexpected deviations from previously characterized genome edits and that no one who might have the expertise to ask the right follow-up questions is involved in their care. He's actions were widely condemned and led ultimately to his arrest, but now that his time served is complete, he seems to be returning to biology in the space of regenerative medicine (Regalado 2022). Clearly, careful governance of these technologies is an important aspect of ethical implementation, and from this example, it isn't clear that the field can be trusted to place appropriate guardrails around its own work (Regalado 2024; Kirksey 2023). The reality is that technology frequently outpaces its oversight, and sometimes ego and personality can find a way around.



While the medical promise of these treatments is genuinely inspirational, there are important concerns about equity related to the development of these flashy new therapies at the bleeding edge of technology. While the science itself is awe-inspiring, the resulting treatments carry incredibly expensive price tags and require access to specialty clinics for patients to even have contact with the appropriate experts in genomic medicine. One of these treatments, the recently approved genome-editing treatment for sickle cell, has been initially priced at the astronomical price of \$2.2 million (Kolata 2023a). Executives justify these high price tags by citing the cost of research and development and the relative savings of a one-and-done treatment when compared with the costs of management for a chronic condition like sickle cell, which some estimate to cost \$1.7–5.2 million over a lifetime (Johnson et al. 2023; Udeze et al. 2022), figures that do not even factor in lost wages due to repeated hospitalizations.

Even if you consider the savings realized by eliminating lifelong treatment of this chronic condition, these costs far exceed what the majority of people in the United States can afford out of pocket, and it is unclear how willing insurers will be to bear these costs and the ancillary expenses up front (Urnov 2022). Furthermore, with costs this high, low- and middle-income countries are left out of these striking advances right off the bat. Finally, it is critical to mention the inherent conflict of experimenting on Black bodies to develop medicines that Black patients may ultimately have difficulty affording due to generations of structural racism, redlining, and other social harms. Therefore, any conversation about the future of genomic medicine must also consider income inequality and how best to ensure that all people will be able to access emerging diagnostics and treatments independently of wealth, income, and insurance status (Vokinger, Avorn, and Kesselheim 2023).

Beyond cost, there is also the issue of what rises to the level of being an appropriate target for genome manipulation. Just as with polygenic screening of embryos, this technology has the potential to be used for the alleviation of disease as well as for human enhancement. While those two concepts seem disparate on their face, the line between them can sometimes be blurry. Who gets to decide where that line is drawn? Whose biases are intimately tied to the definition of “normal” that the field settles on?

We are at a tipping point with respect to the developing technologies of genomic medicine. There are a dizzying array of advances becoming available at an incredibly rapid pace. However, in the rush to monetize these developments and to beat competitors to market, important issues around equity and access frequently become an afterthought, if they aren’t forgotten altogether. For these reasons, it is essential that these topics become woven into the research, development, approval, distribution, and financial conversations around technological development in genomic medicine. Because after all, the true measure of a civilization is how it treats its most vulnerable members.

PART IV Integrating These Stories

14 Our Shared Responsibilities

Here we are, already at the end of our wide-ranging journey together. We've spent time considering the neurological processes behind bias; dismantling the fallacies contributing to hierarchical thinking around race, gender, sexual orientation, pregnancy, and disability; contrasting foundational biological knowledge with social identities; exploring historical harms of biased policies; and drawing parallels with contemporary issues related to medicine, social inequities, and access to developing technologies. But what do we actually *do* with this information, now that we have a more nuanced understanding of the intersectionality of these topics, how mistakes have been made in the past, and the origins of inequities? This journey would be incomplete if we didn't also talk about ways in which we can leverage both this newfound knowledge and our networks to enact positive change because we all have a role to play in promoting equity.

The work of ensuring equitable practice is much larger than a single person, so we cannot rely on or trust any individual invested party to take ownership of the multiple ways in which inequity and genetic essentialism can play out. An important place to begin is by asking ourselves the two following questions:

1. In what spheres and spaces do I have privilege or influence?
2. How can I leverage my privilege and influence for good?

I want to be clear that I am not trying to imply that this is only the work of the privileged, because if we rely only on allies, we would propagate oversights and omit the essential perspectives and voices of the marginalized. Nor do I want to suggest that it is solely the work of the minoritized, who already carry disproportionate burdens. Rather, we must foster true community partnerships that center the experiences of folks from minoritized backgrounds while working together toward equity. In this way, we can honor the lived experiences of people from marginalized backgrounds without further adding to their exclusion.

I also think that all of us, even those of us with multiple intersectional minoritized identities, can likely identify spheres and spaces in which we hold privilege or in which our voices are heard. For example, do you have the right to vote? Do you serve as a trusted adult to young people in Sunday school, on the baseball field, or in your family? Do you have a job where you can effect inclusive change? Are you a community leader? Any of these arenas are opportunities for advocacy and allyship.

Realistically, this work is not simple. There is a well-funded network of people pushing agendas of hate and division at work in America and other countries (Saini 2019; Serwer 2023; Breland 2024), and progress toward justice breeds backlash (Blackstock, Isom, and Legha 2024). In an era when legislators are increasingly sowing division on any number of fronts, from bathroom bills and youth sports to restrictions on reproductive care and immigration, we find ourselves entering a new era of separate as a threat to equal, an era wherein “the cruelty is the point” (Serwer 2018). Those who seek to create deep social divisions are relying on scientific racism and bias based on incomplete—or flat out wrong—notions to further their aims. By invoking science, albeit incorrectly, they give their arguments the sheen of legitimacy as they create genuine threats to our democracy and our humanity.

But you, dear reader, can see through that sheen of legitimacy. I hope I have shared with you enough examples such that you can identify bias and its flawed origins when it enters your spaces, and moreover, that you can

predict how people will be harmed in these situations. Beyond seeing bias, we must also consider strategies to interrupt it.

Let's begin by talking about the health of the American democracy. It only works when people exercise their right to vote and use their voices. If you're not old enough to vote or are not a citizen, you can still lend your time to assist candidates in their campaigns. In researching candidates to support, people must select scientifically literate representatives with values that embrace equity. One initiative meant to help with that is the 3.14 Action PAC, founded to elect "scientists who will use evidence and facts to fight climate change and fix our broken healthcare system" (3.14 Action, n.d.). And of course, once officials are elected, they are representatives who need to hear from their constituents, which gives us another opportunity to communicate the needs of the most vulnerable members of society. Importantly, the big-ticket races for president, vice president, senators, governors, and so on seem to get all the attention, but it is the local races that often have a huge impact on these issues. For instance, think about school committees and their role in book bans. With national organizations like Moms for Liberty organizing to ban books at schools around the country (Harris and Alter 2022), generally targeting books that teach about Black history, racism, LGBTQ+ inclusion, and other important social topics that can give minoritized students a sense of belonging, a strong counterpoint is needed to push back at both the national and local levels.

Many of us also serve as parents or other trusted adults to young people in a variety of settings. Knowing that socialization begins with the very young, we must be vigilant in identifying structures that divide kids or that reinforce hierarchies. We have to feel comfortable pushing back to suggest ways to reset these structures with more inclusive norms, like allowing girls to join scouting troops. Some of these practices are in place because that is just how it has always been done, but a gentle push to reconsider the unintentional harm baked into a traditional system may be just enough to effect change. Importantly, young people are constantly making observations about the world around them and are just doing their best to engage in sensemaking. If they see exclusionary practices around them, they need guidance to understand the forces at play and

how this negatively impacts people in their communities and elsewhere. Many often feel that these types of topics are too advanced or harmful for kids to consider, but the truth is they are already thinking about them, and without trusted advice, they may be susceptible to misinformation. If you come from a background where you don't have personal experience with bias, you may not know where to start, but that doesn't mean that it is best to remain silent. Rather, it is important to educate yourself on ways to promote inclusion through meaningful conversations with our young people (Murchison et al. 2016; National Museum of African American History and Culture 2017). Just because a conversation is hard to have doesn't mean we should shy away from having it.

Another important area in which advocacy work is invaluable is in the health care arena. As we talked about in chapter 12, there are numerous ways in which race factors into health care, and when it makes its way into health care decisions, it is often used as a risk factor, when in reality it is more of a risk marker for other factors that directly confer risk. Consequently, it is important for patients and caregivers to be aware of the ways in which race is used to adjust the recommendations of health care providers and to be attuned to situations in which these adjustments may limit access to care. Patients must advocate for care that prioritizes an individual's unique health concerns based on their proximal causes, and health care providers must be prepared to deliver equitable care with a firm understanding of how clinical shortcuts and racialized thinking can cause harm. This is an area in which I have some hope that the medical field is moving toward equity, but it will take time for the field to develop new evidence-based, race-agnostic approaches to quantifying risk in order to more fully decolonize the current decision-making algorithms. In the meantime, it is important to pick health care systems and providers that are aware of these issues. If possible, it may also be in the patient's best interest to choose a physician from a similar racial background or gender identity to them, as some studies demonstrate that racial and gender concordance leads to improved trust and communication between patient and provider (Alsan, Garrick, and Graziani 2018; Takeshita et al. 2020), and moreover, that meaningful outcomes like survival of Black babies during complex births are improved when overseen by a Black physician (Greenwood et al. 2020). Findings like these also highlight the impor-

tance of training a diverse health care workforce to ensure patients of color have access to providers who look like them and who understand the biased spheres they must navigate.

I hope from these examples it is evident that this is not about a culture of wokeness, but rather that this is about the well-being of society's most vulnerable members. Being woke should not be perceived as an insult, but rather as it was originally intended, as a reminder to "stay woke" to the forces of bias and racism (McWhorter 2021). However, the term has since become a political lightning rod, with pejorative implications that are used to distract from the goals of working toward inclusion. We can't let the taunts of wokeness serve the intended effects of the bullies, who use the term as an insult to stop inclusive efforts in their tracks.

It is clear to me that young people can see through these tactics and are often more aware of social inequities and essentialist thinking than other generations. Because of their insight and values, we need them to serve as champions for accurate discussion of these topics with folks who may not have been exposed to these ideas in classrooms, in places of employment, and with family members. It is a lot to ask of our younger community members, but they are the ones who give me, as an educator, the most hope for a better tomorrow. Together we must figure out how to do better as we work toward a more just, scientifically informed society. This is our lane; how will you use it?

Afterword

A CALL TO ACTION

Science has always been grounded in the seeking of truth. However, as we've seen throughout this book, some armchair scientists and even scientific luminaries have twisted this pure goal to further divisive beliefs and elevate their own statuses. With misinformation spreading unchecked through social media and other channels, the need for inclusive, accurate scientific information to dismantle agendas of division is more and more essential (Osborne and Pimentel 2022). Normalization of poisonous rhetoric and falsification or manipulation of science to further ideals of hate have become far too common. In this climate, bad actors are empowered to continue using these harmful tactics.

Some educators point to the pure origins of science in truth seeking when citing a need to stay in their lane and not comment on social justice issues. Instead, I believe we should be centering our roles as truth tellers and using the platforms we are privileged to hold, together with the knowledge we possess, to provide a scientific rebuttal to the prevailing dialogue about human difference that dominates current public discourse (Dasgupta 2020; Donovan 2022). It is my hope that after reading this text, if you are an educator, you are feeling empowered to initiate some of these discussions in your classrooms and offices. Even though higher edu-

cation is the lens through which I approach these topics because of the spheres in which I operate, I firmly believe that these conversations belong in broader contexts, such as at dinner tables, in community groups seeking to become more inclusive, in parent support groups serving children with disabilities, in groups celebrating LGBTQ+ and BIPOC identities, in religious communities, as well as in classrooms at all levels and disciplines, including the social sciences, the law, and beyond. Some of the activities I will describe in this section were created for a classroom setting but could easily be translated for use in the groups I just described and in other venues as well. Scientists and educators belong in these conversations (Duncan et al. 2024; Yamamoto 2024), but so do we all.

The study of genetics and genomics has always been about understanding the human condition. The DNA molecule is about more than inheritance. It captures the spirit of what makes us human, and through careful study, we can see that it tells a story of our shared humanity. There are many aspects of this common ground to which the study of genomics can lend its expertise, including across groups defined by race, gender, sexual orientation, disability status, and many other identities. Scientists and educators have a unique combination of skills, knowledge, and opportunities to discuss what can and cannot be explained by biology and how misunderstanding of this information can cause harm to marginalized groups, and it is my hope that what we have learned about our roles in fostering conversations about complex topics can be implemented well beyond our labs and classrooms.



To further the goal of correcting the scientific record in service of equity, this book was originally conceived of as a course, and in parallel, I developed this book to serve as a companion and to share these lessons with a wider audience outside of my university. In this book, I began by sharing my own positionality, lived experience, and potential biases in the first chapter, but in class, we used the time to explore the positionality of the entire group to help us identify individual and collective experiential gaps. Depending on the size and duration of the course or community event you are running and how comfortable the group members are with each other

on the first day you start these conversations, you may choose to run this perspective-taking exercise anonymously. Before we launched into the positionality questions, we began by setting some ground rules for our shared understanding around the following values:

- maintaining confidentiality
- making space for each other's thoughts
- being present in the moment and committed to the group
- allowing open, vulnerable communication
- listening, challenging, and supporting one another
- deliberately creating a safe space for questions
- assuming good intentions even when the impact may unintentionally harm
- calling in versus calling out to invite adoption of more inclusive language and stances
- critiquing ideas, not people
- suspending judgment
- identifying assumptions
- caring for themselves and each other

Once we set this framework, we thought a bit about our own identities, both given and chosen, before moving into our positionality exercise. This exercise is one where I posed a series of questions about individual identity. Students responded by annotating my slides using the Zoom tool (even though we were meeting in person, to foster authentic community). This allowed them to anonymously add a marker to a continuum I displayed for each identity conscious statement. I have also done this by inviting participants to physically line up in a four corners exercise, with each corner representing a different response (e.g., moderate and significant agreement or disagreement with each statement).

The statements start out fairly uncontroversial and get increasingly personal, as you will see in the following list. Everyone was told they could abstain from answering at any point, and I did allow them to vote on whether we would do this anonymously or with identifiable responses that would remain in our learning community. We quietly proceeded through

all the statements before debriefing at the end to allow us to suspend judgment and to take it all in before discussing the overall picture their responses to the statements painted.

Identity Conscious Statements

1. I came to graduate school right after undergrad.
2. I traveled a long distance for school.
3. I am younger than 25.
4. I have a significant life partner.
5. I plan to have children.
6. I am a parent.
7. I worry about balancing my career and my family.
8. I expect(ed) to take time off work when my first child is/was born.
9. I expect to make more money than my partner when I am 40+ years old.
10. I was not born in the United States.
11. English is the only language I speak fluently.
12. I am the first in my family to obtain a college degree.
13. I am a typical reflection of what a scientist looks like.
14. I belong in science.
15. I expect my research or professional work to be shaped by my identity.
16. I have felt threatened because of my race.
17. I have felt threatened because of my gender.
18. I have felt threatened because of my sexual orientation.
19. I have felt threatened because of my disability.
20. I have felt threatened because of my religious beliefs.
21. There are aspects of my identity that I do not share with people close to me.
22. There are aspects of my identity that I do not share with my peers and colleagues.
23. I have made derogatory jokes about a group because of its gender, sexual orientation, race, socioeconomic class, religious beliefs, disability status, or physical appearance.

24. I have been offended, but did not speak up, when someone made a derogatory joke about my gender, sexual orientation, race, socioeconomic class, religious beliefs, disability status, or physical appearance.
25. My race gives me an advantage over others.
26. My gender gives me an advantage over others.
27. My socioeconomic status gives me an advantage over others.
28. I feel I am culturally humble.
29. I have been uncomfortable at some point in this exercise.
30. I have not been entirely honest at some point in this exercise.

These questions are oriented toward adult learners, but they can easily be adapted for the age and context of your community. For instance, some questions can be posed about families rather than the learner if your group members are young. Or the question about moving a long distance for school can be reframed around how far they live now compared to where they were born. Moving through these 30 questions was a very powerful experience that helped us understand more of what our group would bring to the exploration of these complex topics and fostered a sense of respect for each other that we carried through the semester.

With each meeting corresponding to a chapter of the book, we frequently broke the two-hour sessions into background framework that I provided as an interactive presentation and a discussion about a reading. In these discussions, we considered questions of who the invested parties were, who was harmed, what the misconceptions were, how processes could have been changed to avoid harm, and what lessons we can learn to fix this moving forward. The topics and selected readings are listed in table 5. The readings are a mix of scientific articles and news or magazine articles. For younger learners or for nonscientists, some of the scientific articles can be replaced with less technical options from the news or videos. These discussions could also be structured as debates for a twist on these conversations.

The single most important thing I could do for these discussions was to create safe spaces, to help everyone understand that I cared about them as both people and learners. This can be done by articulating how some of

Table 5 Suggested discussion readings for topics from throughout the book

Topic	Reading(s) for class discussion
On the origins of bias and racism	Hoffman, Kelly M., Sophie Trawalter, Jordan R. Axt, and M. Norman Oliver. 2016. "Racial Bias in Pain Assessment and Treatment Recommendations, and False Beliefs about Biological Differences between Blacks and Whites." <i>Proceedings of the National Academy of Sciences</i> 113 (16): 4296–4301. https://doi.org/10.1073/pnas.1516047113 .
Sex and gender	Barres, Ben A. 2006. "Does Gender Matter?" <i>Nature</i> 442 (7099): 133–36. https://doi.org/10.1038/442133a .
Disability	Sanghavi, Darshak M. 2006. "Wanting Babies Like Themselves, Some Parents Choose Genetic Defects." <i>New York Times</i> , December 5, 2006. https://www.nytimes.com/2006/12/05/health/05essa.html .
What genetics doesn't fully explain	Coop, Graham, and Molly Przeworski. 2022. "Lottery, Luck, or Legacy. A Review of 'The Genetic Lottery: Why DNA Matters for Social Equality.'" <i>Evolution</i> 76 (4): 846–53. https://doi.org/10.1111/evo.14449 .
Contemporary eugenics and its historical roots	Zhang, Sarah. 2020. "The Last Children of Down Syndrome." <i>The Atlantic</i> , December 2020. https://www.theatlantic.com/magazine/archive/2020/12/the-last-children-of-down-syndrome/616928/ .
Genetics, privacy, and the justice system	Faife, Corin. 2022. "NJ Police Used Baby DNA to Investigate Crimes, Lawsuit Claims." <i>The Verge</i> , July 29, 2022. https://www.theverge.com/2022/7/29/23283837/nj-police-baby-dna-crimes-lawsuit-public-defender . LaForgia, Michael, and Jennifer Valentino-DeVries. 2021. "How a Genetic Trait in Black People Can Give the Police Cover." <i>New York Times</i> , May 15, 2021. https://www.nytimes.com/2021/05/15/us/african-americans-sickle-cell-police .
Who gets left behind	Brothers, Kyle B., Robin L. Bennett, and Mildred K. Cho. 2021. "Taking an Antiracist Posture in Scientific Publications in Human Genetics and Genomics." <i>Genetics in Medicine</i> 23 (6): 1004–7. https://doi.org/10.1038/s41436-021-01109-w .
Race-based medicine	Hobson, Will. 2021. "How 'Race-Norming' Was Built into the NFL Concussion Settlement." <i>Washington Post</i> , August 2, 2021. https://www.washingtonpost.com/sports/2021/08/02/race-norming-nfl-concussion-settlement/ .
The future of genomic medicine	Regalado Antonio. 2019. "The World's First Gattaca Baby Tests Are Finally Here." <i>MIT Technology Review</i> , November 8, 2019. https://www.technologyreview.com/2019/11/08/132018/polygenic-score-ivf-embryo-dna-tests-genomic-prediction-gattaca/ .

the concepts are complex and can easily lead to biased thought patterns when not intentionally interrupted. It can involve listening to how the content connects with their own lives and experiences. And it can be as simple as learning their names, understanding their backgrounds, and thinking about how different concepts will be relevant to each of them as individuals. Learning is best accomplished when we can bring our whole selves to our shared learning spaces and goals. The amount of love I felt for my students during these discussions cannot be overstated. This next generation gives me so much hope. They zeroed in on inequitable treatment, and they prioritized social justice at the center of our conversations. Whatever value I was able to add by highlighting these injustices for them was a true privilege.

Throughout the semester, I asked everyone to keep a running list of topics or questions that got lodged in their brains. Was there something that was personally meaningful that we discussed that they wanted to return to? Were there unanswered questions from class that they couldn't shake? Did related topics come to mind from our discussions that they wanted to explore in greater depth? These seeds grew into our final projects, one where I asked them to harness their inner changemakers by thinking of a creative way to explore the topics of interest and sharing a more accurate scientific message with a coworker, a loved one, or the public at large. The students led discussions on equity in sport with their athletic groups, facilitated conversations between health care professionals on race-based medicine, communicated with the Institutional Review Board on equitable practices in human subjects research, engaged with internet trolls by using memes to combat misinformation, and developed infographics and fact sheets to convey complex topics about equity. Though my students did these projects individually, groups of participants could take them on together. In my case, I wanted the world to see the amazing things these incredible community members had to offer, and they were eager to apply their learning and to advocate for change.



We do not have the luxury to remain silent on issues of social justice. Our predecessors and our contemporaries have committed harms by reifying

systems of hierarchies and marginalization to maintain power structures for their own benefit (Green et al. 2022). It is our social responsibility to restore balance in these conversations by setting the record straight, or we will be complicit in the oppression of historically excluded communities.

In order to explore the common foundation between marginalized identities from a scientific perspective with our learners, we must acknowledge the work we have to do to educate ourselves on topics that were largely overlooked in our own scientific training. Universities are increasingly seeking to bring attention to these issues through community dialogues and courses (Patel et al. 2024), and we can help catalyze those efforts. It is also important to bring humility to these conversations, particularly if you do not have lived experience with the marginalized identities that will be centered in these dialogues. While it is important to defer to people with lived experience, it is equally important not to burden them with the role of teaching others who have not had to navigate the same challenges.

I also want to acknowledge the growing reality that educators and community leaders do not always feel entirely free to discuss topics such as those included in these pages openly with their groups. For example, there are an increasing number of legislative challenges on what can be discussed in classroom settings (Greene 2022; Harris and Alter 2022), and this puts the educator between a rock and a hard place. Being anti-oppressive requires action, and simply avoiding difficult conversations will unfortunately allow misconceptions to spread. There are ways in which we can decolonize our curricula (Green et al. 2022), avoid harm through the use of unbiased language (Cho et al. 2022), and employ inclusive frameworks for our case examples (Bennett et al. 2022); ideally, we would extend further to actively engage in anti-oppressive actions. Having said that, if every educator who understands the principles that debunk essentialist thinking was removed from their classroom spaces for “activist” teaching, this would certainly be a negative outcome for the learners. Moreover, as the attacks on DEI have been particularly visible in academia, dissent grows louder in many realms across society, and community leaders of all sorts are facing pushback for delving into these topics. Every one of us will have to navigate this journey based on the complexities of our environments and our individual roles in society.

When I told you about one of my core beliefs in chapter 1, I described my faith in the transformative power of education. I do believe that knowledge is power and that it can help guide us to a more just society. By focusing on the foundational knowledge that defines biological traits, we can begin to tease apart those aspects of identity that are social in nature. That is not to say social identities are unimportant—far from it! Rather, our assumptions about the ways in which they contribute to inequities will differ.

While talent is equally spread throughout the population, opportunity is not. Though we are all human, we do not all enjoy access to the same benefits and rights. I do believe that if we take the time to learn why that is the case, we will have the tools to uplift each other. This may sound very idealistic, and it is. I imagine working toward a future with a truer sense of belonging and justice, but I also know the answers are not easy ones, or the problems would not have persisted for hundreds of years.

One final thought for us to consider is the fear of getting it wrong. These topics are nuanced and have the potential to offend the very people you are trying to uplift. I hope you will not let that fear prevent you from having these critical conversations with your communities. Don't be afraid to share your intentions, but also own the impact of your actions. If you unintentionally harm your learners with well-intentioned ideas that have limitations, acknowledge your mistake, correct your actions, and preserve those all-important relationships with your group members. Grow with them. And next time do better. This is how we all move forward together.

Acknowledgments

I wasn't thinking about writing this book when the project sort of fell into my lap. I was attending a conference sponsored by ELSIhub as a typical academic activity. Since the pandemic, many conferences had moved to online and hybrid formats; this one was online, and I was enjoying the side conversation that was happening in the chat while learning from the speaker. As part of that sidebar, I shared a few short pieces I had written for academic audiences. Little did I know that my future editor, Chloe Layman, was in the chat too, clicking on my links and hatching a plan. When she emailed a few days later to talk about a book project on anti-racist genetics education, my first thought was that this must be spam. Overcoming that initial misconception and scheduling the first exploratory conversation with Chloe put me on a journey that has been incredibly rewarding and fulfilling. Chloe has been such an incredible thought partner in our conversations as the plan grew from a book on anti-racism to a more expansive text on anti-oppression. I am so fortunate to have her as the first person who proposed this project and who has been in my corner ever since.

The UC Press, a nonprofit, progressive press, was the perfect home for this book given their dedication to social responsibility in the titles they choose to publish. I am thankful for their first generation grant to support my writing of this book and the program's mission to help amplify voices from a spectrum of perspectives. I am appreciative of all the members of the Press's team for supporting this labor of love.

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As a professor, I hope it was clear as I was writing this book how important my students are to me. I was fortunate to work with a group of incredible medical students on educational efforts related to dismantling racism in medicine, and bringing the work of our group to our community in June 2020 was an empowering experience, knowing that together we could do constructive advocacy work while navigating a time of nationwide pain and unrest. I started writing this book a couple of years later, just after I had decided to put together a course on these topics for my graduate students. That first cohort of students and those that followed fill me with hope. They earnestly wanted to give back, to use their science for good, and being part of that classroom community with them has been an incredibly special experience.

This book would not have been possible without the generous support, encouragement, and true enthusiasm I received from my friends and family, who have celebrated every step along the way to this book being in your hands. I had loved ones share their own book contracts, cook delicious meals for me, humor my perhaps too lengthy explanations of the contents of this book during our get togethers, and promise to read the book upon release. I am grateful to all my readers for your willingness to engage on these meaningful and challenging topics.

Most importantly, I want to thank my family. My mom deserves special recognition for her unwavering excitement about the project and for always reminding me how proud my dad would have been if he had lived to see this book come to fruition. My brother Deb is the best brother anyone could hope to have. And last but certainly not least, I had the support of my kids, Sonali and Nalini, and my spouse, Jason Fiorillo. The project started coming together in the fall of Sonali's senior year of high school and Nalini's first year at a new school. While Sonali navigated the stresses of applying to college, she always showed genuine enthusiasm for my work, even when it took me away from her own important efforts. Nalini showed great independence as well that year, making a big educational shift of her own, and she has been eager to offer feedback and her photographic

skills. I am so proud of both kids and their achievements, and I am thrilled that they are excited to read the final product. Finally and importantly, my deepest thanks to Jason, who stepped in to bridge those parenting gaps, who didn't complain (too much) when I spent many late nights in my home office trying to get a few more pages done after another full day of teaching, and who always believed in me and this project. His response to my "I didn't know I had a book in me" was perfect and without hesitation: "I always did."

Glossary

ableism	Beliefs or practices that rest on the assumption that being able-bodied is “normal” while other states of being need to be “fixed” or altered. This can result in devaluing or discriminating against people with physical, intellectual, or psychiatric disabilities. Institutionalized ableism may include or take the form of un/intentional organizational barriers, including lack of accessibility accommodations, that result in disparate treatment of people with disabilities (PwDs). (From Boston University Chobanian & Avedisian School of Medicine, Boston Medical Center, and Boston University Medical Group 2021)
abortion	Abortion is the termination of a pregnancy by removal or expulsion of a fetus before it can survive outside the uterus. It can occur spontaneously, known as a miscarriage, or be induced intentionally through medication or surgical procedures. Abortion is a routine form of health care that can be necessary to preserve the health of the gestational carrier, and studies have found that when performed legally and under the supervision of a trained professional, it can be safer than pregnancy and childbirth. Approximately 25 percent of women will have an abortion by age 45. (Adapted from Planned Parenthood, Action Fund, n.d.; Raymond and Grimes 2012)

accessibility	The “ability to access” the functionality of a system or entity and gain the related benefits. Also, the degree to which a product, service, or environment is accessible by as many people as possible. Accessible design ensures both direct (unassisted) access and indirect access through assistive technology (e.g., computer screen readers). Universal design ensures that an environment can be accessed, understood, and used to the greatest extent possible by all people. The social model of disability centers on accessibility challenges in one’s environment. The Americans with Disabilities Act set accessibility standards for public places, commercial facilities, and state and local government facilities for new construction, additions, remodeled settings, and virtual spaces. (Adapted from Boston University Chobanian & Avedisian School of Medicine, Boston Medical Center, and Boston University Medical Group 2021)
accommodations	A change in the environment or in the way things are customarily done that enables an individual with a disability to have equal opportunity, access, and participation. (From Boston University Chobanian & Avedisian School of Medicine, Boston Medical Center, and Boston University Medical Group 2021)
adenovirus	A member of a family of viruses that can cause infections in the respiratory tract, eye, and gastrointestinal tract. Forms of adenoviruses that do not cause disease have been manipulated for use in gene therapy, where they can carry an intact copy of a gene to restore function in an individual whose own gene is missing or has lost its ability to function appropriately. This is considered a viral vector, and there are a range of viruses beyond adenovirus being researched for use in the context of gene therapy. (Adapted from National Institutes of Health, National Cancer Institute 2011)
age of onset	Refers to the time period at which an individual experiences the first symptoms of a disease. Age of onset can vary, and conditions present themselves at various stages of life.
allele	An allele is one of two or more versions of DNA sequence (a single base or a segment of bases) at a given genomic location. An individual generally inherits two alleles, one from each parent, for any given genomic location where such variation exists. If the two alleles are the same, the individual is homozygous for that allele. If the alleles are different, the individual is heterozygous. (From National Institutes of Health, National Human Genome Research Institute 2022b)

allyship	An ally recognizes that though they are not a member of a marginalized group(s) they support, they make a commitment to better understand the struggle of another's circumstances. Sometimes an ally may have more privilege, can recognize that privilege in society, and be willing to work in solidarity with marginalized groups to help uplift their communities. Allies both understand that it is in everyone's best interest to end all forms of oppression and actively work to reduce their own complicity in structural oppression. (Adapted from Boston University Chobanian & Avedisian School of Medicine, Boston Medical Center, and Boston University Medical Group 2021; Gaines, 2019)
Alzheimer's disease	Alzheimer's is a type of dementia that causes problems with memory, thinking, and behavior. Symptoms usually develop slowly and get worse over time, becoming severe enough to interfere with daily tasks. (From Alzheimer's Association, 2024)
ambipotent	Ambipotent refers to a stage in sex development where both male and female developmental anatomical precursors simultaneously exist in an embryo.
amino acid	An amino acid is the fundamental molecule that serves as the building block for proteins. There are 20 different amino acids. A protein consists of one or more chains of amino acids (called polypeptides), whose sequence is encoded in a gene. Some amino acids can be synthesized in the body, but others (essential amino acids) cannot and must be obtained from a person's diet. (From National Institutes of Health, National Human Genome Research Institute 2022b)
amniocentesis	A procedure used to take out a small sample of the amniotic fluid for prenatal diagnostic testing. The amniotic fluid surrounds the fetus in pregnancy, and when sampled (by inserting a needle into the amniotic sac through the abdominal and uterine walls), fetal cells that are floating in the liquid are also obtained. The fetal cells can be grown in the laboratory and then tested for genetic variation. (Adapted from Cohn, Scherer, and Hamosh 2024)
amplification	Amplification refers to an increase in the number of copies of a gene or any other sequence in a genome. (From National Institutes of Health, National Cancer Institute 2012)
ancestry	Genetic ancestry refers to information about the people that an individual is biologically descended from, including their genetic relationships. Genetic information can be combined

	with historical information to infer where an individual's distant ancestors lived. Genetic ancestry can be thought of in terms of lines extending upward in a family tree, from an individual through their genetic ancestors. In practice, shared genetic ancestry is typically inferred by some measure(s) of genetic similarity. (Adapted from Committee on the Use of Race, Ethnicity, and Ancestry as Population Descriptors in Genomics Research 2023; National Institutes of Health, National Human Genome Research Institute 2022b)
aneuploidy	A condition in which there are extra or missing chromosomes or chromosomal segments in some or all the cells in the body. Human cells, except for egg and sperm cells, typically contain 23 pairs of chromosomes, for a total of 46 chromosomes in each cell. With aneuploidy, the number of chromosomes does not equal 46. For example, people with Down syndrome, also known as trisomy 21, have three copies of chromosome 21 in each cell. (From National Institutes of Health, National Cancer Institute 2012)
antisense nucleic acids	Antisense nucleic acids are DNAs or RNAs that are complementary to the mRNAs produced by genes as they direct the synthesis of a protein. Because of the antisense molecules' complementary sequence, they can bind to the mRNAs and block the ability of the mRNA to make protein or carry out its typical function. (From National Institutes of Health, National Human Genome Research Institute 2022b)
assisted reproductive technology (ART)	Procedures that use donor or nondonor eggs and sperm to create embryos in vitro. Examples of ART include in vitro fertilization (IVF), gamete intrafallopian transfer (GIFT), and zygote intrafallopian transfer (ZIFT). These procedures may be used prior to preimplantation genetic testing (PGT). (Adapted from Centers for Disease Control and Prevention 2019)
autonomy	Autonomy is an ethical principle defined by the right to self-determination and respects the individual's right to make informed decisions. (Adapted from Centers for Disease Control and Prevention 2022)
autosomal dominant	Autosomal dominant is a pattern of inheritance characteristic of some genetic traits. <i>Autosomal</i> means that the gene in question is located on one of the numbered, or nonsex, chromosomes. <i>Dominant</i> means that a single copy of the allele (from one parent) is enough to cause the trait to appear. A

	child of a person affected by an autosomal dominant condition has a 50 percent chance of being affected by that condition via inheritance of a dominant allele. By contrast, an autosomal recessive disorder requires two copies of the recessive variant (one from each parent) to cause the disorder. Huntington's disease is an example of an autosomal dominant genetic condition. (From National Institutes of Health, National Human Genome Research Institute 2022b)
base/base pair	A base pair consists of two complementary DNA nucleotide bases that pair together to form a "rung of the DNA ladder." DNA is made of two linked strands that wind around each other to resemble a twisted ladder—a shape known as a double helix. Each strand has a backbone made of alternating sugar (deoxyribose) and phosphate groups. Attached to each sugar is one of four bases: adenine (A), cytosine (C), guanine (G) or thymine (T). The two strands are held together by hydrogen bonds between pairs of bases: adenine pairs with thymine, and cytosine pairs with guanine. (From National Institutes of Health, National Human Genome Research Institute 2022b)
beneficence	Beneficence is an ethical principle related to minimizing or preventing harm and creating conditions that will help others achieve their maximum health potential. (From Centers for Disease Control and Prevention 2022)
biomarkers	A biological molecule found in blood, other body fluids, or tissues that is a sign of a healthy process, or of a condition or disease. A biomarker may be used to ascertain the progression of a particular condition or to see how well the body responds to a treatment for a disease. Also called molecular marker. (Adapted from National Institutes of Health, National Human Genome Research Institute 2022b)
BIPOC	An acronym that stands for Black, Indigenous, and people of color. This is a more recent term, developed to draw attention to the continued racialized harm visited on Black and Indigenous people within the term <i>POC</i>. BIPOC is used to promote visibility among Indigenous and Black people and to call attention to colonialism and anti-Blackness. (Adapted from Boston University Chobanian & Avedisian School of Medicine, Boston Medical Center, and Boston University Medical Group 2021)

bisexual	Describes someone who can be emotionally, romantically, or sexually attracted to more than one sex, gender, or gender identity, though not necessarily simultaneously, in the same way, or to the same degree. (From Boston University Chobanian & Avedisian School of Medicine, Boston Medical Center, and Boston University Medical Group 2021)
bodily autonomy	The right of a person to govern what happens to their body without external influence or coercion. This concept is particularly significant in the context of reproductive freedom and gender affirming care. (Adapted from Planned Parenthood, Action Fund, n.d.)
bottleneck	In studies of population genetics and evolution, the bottleneck event sharply limits population size. For example, an environmental disaster or hunting a species to the brink of extinction could serve as bottleneck events. Such events frequently produce changes in the genetic composition of a population because much of the genetic variation in the original population is lost as a consequence of the bottleneck event, a phenomenon referred to as genetic drift. In extreme cases, population-reducing events can eliminate genotypes from a gene pool, even genotypes not associated with reduced fitness. (Abel et al. 2015)
carrier	A carrier, as related to genetics, is an individual who “carries” and can pass on to their offspring a genomic variant (allele) associated with a disease or trait that is inherited in an autosomal recessive or sex-linked manner and who does not show symptoms of that disease or features of that trait. The heterozygous carrier has inherited the variant allele from one parent and an unaffected allele from the other parent. Any offspring of two carrier parents is at risk of inheriting a variant allele from both of their parents, which would result in that homozygous child having the disease or trait. (From National Institutes of Health, National Human Genome Research Institute 2022b)
carrier frequency	The proportion of individuals in a population who have a single copy of a specific recessive genetic variant. Carrier frequency also sometimes refers to the prevalence of variants in dominantly acting genes, such as BRCA1 and BRCA2. (From National Institutes of Health, National Cancer Institute 2012)

carrier screening	Involves testing to see if a person “carries” a genetic variation (allele) associated with a specific condition or trait. A carrier has inherited an unaffected and a variant allele for a disease or trait-associated gene, one from each parent. Typically carrier screening is performed in conjunction with preconception or prenatal care to look for recessively inherited alleles when the suspected carrier has no symptoms of the disease, but that person’s offspring could have the disease if the other parent is a carrier of a pathogenic variant in the same gene. (From National Institutes of Health, National Human Genome Research Institute 2022b)
Caucasian	This term was initially employed as a “polite” way to describe White populations of European ancestry, but the term itself has a racist history through its connection with scientific racist studies attempting to codify Caucasians as the superior biological race. In reality, race is a social construct, and the term was originally created to refer to people who lived in the Caucasus mountains and believed to be created in the image of God, while the other races were considered degenerate. Use of this term calls forward its racist history. <i>White populations</i> or <i>European ancestries</i> are preferred terms, but it is cautioned that the replacement of a single word does not correct the issue that White populations are frequently the standard against which all others are measured. (Cho et al. 2022)
cell fate	The fate of a cell describes its future identity, or the identity of its daughter cells, before it is phenotypically detectable through differentiation or division. Cell fate can be determined by endogenous developmental factors, interaction with adjacent cells, or external long-distance signals, such as morphogens and hormones. (From “Cell Fate—Latest Research and Reviews,” n.d.)
cell-free DNA (cfDNA)	Cell-free DNA is released during normal physiological functions as well as from a fetus during gestation and through the cell death programs of apoptosis and necrosis. It can be found in the plasma and other body fluids. Prenatal testing of cell-free DNA found in the gestational carrier’s bloodstream can be used to noninvasively test for DNA variants present in the fetus. Similarly, cell-free DNA testing can be used to characterize cancers and monitor cancer therapy. (Adapted from National Institutes of Health, National Human Genome Research Institute 2022b)

cell migration	Cell migration is a fundamental process that controls morphogenesis during development and inflammation. Its deregulation causes or is part of many diseases, including developmental differences, autoimmune syndromes, chronic inflammation, and cancer. (Adapted from National Institutes of Health, National Cancer Institute 2011)
chromosome	Chromosomes are threadlike structures made of protein and a single molecule of DNA that serve to carry the genomic information from cell to cell. In plants and animals (including humans), chromosomes reside in the nucleus of cells. Humans have 22 pairs of numbered chromosomes (autosomes) and one pair of sex chromosomes (XX or XY), for a total of 46. Each pair contains two chromosomes, one coming from each parent, which means that children inherit half of their chromosomes from each parent. Chromosomes can be seen through a microscope when the nucleus dissolves during cell division. (From National Institutes of Health, National Human Genome Research Institute 2022b)
chorionic villus sampling	A prenatal diagnostic test used to test for chromosomal and other genetic differences during pregnancy. During the test, a tissue sample is taken from the placenta and sent to a lab for analysis. The chorionic villi are tiny projections of placental tissue that look like fingers. They have the same genetic material as the baby and can be sampled slightly earlier in the pregnancy than the amniotic fluid. (Adapted from Johns Hopkins Medicine 2024)
cisnormativity	The assumption that everyone identifies as the gender that aligns with the sex they were assigned at birth, and those who do not, that is, transgender individuals, are considered “abnormal.” (Adapted from Planned Parenthood, Action Fund, n.d.)
cisgender	A term to describe a person whose gender identity aligns with the sex assigned to them at birth (i.e., a person who is not transgender). (Adapted from Boston University Chobanian & Avedisian School of Medicine, Boston Medical Center, and Boston University Medical Group 2021)
colorism	Also called shadism, skin tone bias, pigmentocracy, and color complex, which refers to the prejudiced attitude and/or discriminatory acts against people with darker color/shade/tone skin, typically among people of the same racial or ethnic group. This is a form of prejudice or discrimination in which

	people are treated differently based on the social meanings attached to skin color. White supremacy is upheld by colorism. (From Boston University Chobanian & Avedisian School of Medicine, Boston Medical Center, and Boston University Medical Group 2021)
common complex disease	A complex disease (or condition), when discussed in the context of genetics, reflects a disorder that results from the contributions of multiple genomic variants and genes in conjunction with significant influences of the physical and social environment. For this reason, complex diseases are also called multifactorial diseases. This stands in contrast to a “simple” genetic disease that is more directly caused by variants in a single gene. Common examples of complex genetic diseases include heart disease, diabetes, and cancer. (From National Institutes of Health, National Human Genome Research Institute 2022b)
common variant	A common variant is one that is found at a relatively high frequency (usually defined as >1%–5%) in the population. Genome wide association studies examine the impact of these variants on complex disease.
congenital	Congenital refers to a condition or trait that exists at birth. Congenital conditions or traits may be hereditary, may result from an action or exposure occurring during pregnancy or at birth, or may be due to a combination of these factors. (Adapted from National Institutes of Health, National Human Genome Research Institute 2022b; National Institutes of Health, National Cancer Institute 2012)
consumer genomics	Consumer genomics, also referred to as direct-to-consumer genetic testing and personal genomics, involves a consumer accessing genetic testing from a commercial company apart from a health care provider. The testing available through these means spans from genetic genealogy to health and traits. (Adapted from National Academies of Sciences, Engineering, and Medicine 2020)
copy number variant	Copy number variation (abbreviated CNV) refers to a circumstance in which the number of copies of a specific segment of DNA varies among different individuals’ genomes. The individual variants may be short or include thousands of bases. These structural differences may have come about through duplications, insertions, deletions, or other changes and can affect long stretches of DNA. Such regions may or may not contain a gene(s) and may also be associated with specific

CRISPR	traits or disorders. (From National Institutes of Health, National Human Genome Research Institute 2022b) CRISPR (short for “clustered regularly interspaced short palindromic repeats”) is a technology that research scientists use to selectively modify the DNA of living organisms. CRISPR was adapted for use in the laboratory and clinically from naturally occurring genome editing systems found in bacteria. Some liken this technology to molecular scissors, allowing for precise, deliberate manipulation of genomic sequences. Drs. Jennifer Doudna and Emmanuelle Charpentier shared the 2020 Nobel Prize in Chemistry for their discovery of CRISPR. (Adapted from National Institutes of Health, National Human Genome Research Institute 2022b)
deafness (syndromic and nonsyndromic)	Used to describe a person with total or profound hearing loss. Many only have mild or partial loss of hearing. The terms <i>deaf</i>, <i>hard of hearing</i>, or <i>hearing loss</i> are acceptable within the community, but <i>hearing-impaired</i> is generally not preferred as it emphasizes a deficit model through a medicalized view of disability. (Adapted from Boston University Chobanian & Avedisian School of Medicine, Boston Medical Center, and Boston University Medical Group 2021)
decolonize	May be defined as the active resistance against colonial powers and a shifting of power toward political, economic, educational, cultural, and psychological independence, and power that originates from a colonized nation's own Indigenous culture. This process occurs politically and also applies to personal and societal psychological, cultural, political, agricultural, and educational deconstruction of colonial oppression. (Adapted from Boston University Chobanian & Avedisian School of Medicine, Boston Medical Center, and Boston University Medical Group 2021)
deletion	A deletion changes the DNA sequence by removing at least one nucleotide in a gene. Small deletions remove one or a few nucleotides within a gene, while larger deletions can remove an entire gene or several neighboring genes. The deleted DNA may alter the function of any affected protein or proteins. (Adapted from National Institutes of Health, National Library of Medicine 2021)
development	The transformation of a single cell to a full organism involving changes in cellular identity and their three-dimensional structural arrangement along the way. This complex process is coordinated through the action of genes at the right times, in the right doses, and at the right locations.

developmental difference	Refers to any variation from typical development, including anatomical or physical development, intellectual development, and emotional development.
differences of sex development (DSD)	Differences of sex development (DSD) are a group of conditions where development of the reproductive system does not follow typical male or female trajectories. These differences involve genes, hormones, and reproductive organs, including genitals. The term was initially created to align with the DSD abbreviation for the term <i>disorders of sex development</i> , but because of the harmful connotations of the original term, the community has started to prefer the term <i>variations in sex characteristics</i> (VSC). See also <i>intersex</i> .
direct-to-consumer genetic testing	Direct-to-consumer (DTC) genetic testing provides people access to their genetic information without necessarily involving a health care provider or health insurance company in the process. After purchasing a test kit, customers send the company a DNA sample and receive their results directly from a secure website or app, or in a written report. DTC genetic testing, also referred to as consumer or personal genomics, spans applications from genetic genealogy to health and traits. (Adapted from National Academies of Sciences, Engineering, and Medicine 2020)
disability	General term for a functional limitation that affects one or more of the major life activities, including walking, lifting, learning, and breathing. Disabilities can be hidden (e.g., related to mental health, learning) or visible (e.g., mobility disability, sensory disability like low vision). (Adapted from Boston University Chobanian & Avedisian School of Medicine, Boston Medical Center, and Boston University Medical Group 2021)
DNA	Deoxyribonucleic acid (abbreviated DNA) is the molecule that carries genetic information for the development and functioning of an organism. DNA is made of two linked strands that wind around each other to resemble a twisted ladder—a shape known as a double helix. Each strand has a backbone made of alternating sugar (deoxyribose) and phosphate groups. Attached to each sugar is one of four bases: adenine (A), cytosine (C), guanine (G), or thymine (T). The two strands are connected by chemical bonds between the bases: adenine bonds with thymine, and cytosine bonds with guanine. The sequence of the bases along DNA's backbone

	encodes biological information, such as the instructions for making a protein or RNA molecule. (From National Institutes of Health, National Human Genome Research Institute 2022b)
DNA sequencing	DNA sequencing refers to the general laboratory technique for determining the exact sequence of nucleotides, or bases, in a DNA molecule. The sequence of the bases (often referred to by the first letters of their chemical names: A, T, C, and G) encodes the biological information that cells use to develop and operate. Establishing the sequence of DNA is key to understanding the function of genes and other parts of the genome. There are now several different methods available for DNA sequencing, each with its own characteristics, and the development of additional methods represents an active area of genomics research. (From National Institutes of Health, National Human Genome Research Institute 2022b)
double helix	Double helix, as related to genomics, is a term used to describe the physical structure of DNA. A DNA molecule is made up of two linked strands that wind around each other to resemble a twisted ladder in a helix-like shape. Each strand has a backbone made of alternating sugar (deoxyribose) and phosphate groups. Attached to each sugar is one of four bases: adenine (A), cytosine (C), guanine (G), or thymine (T). The two strands are connected by chemical bonds between the bases: adenine bonds with thymine, and cytosine bonds with guanine. (From National Institutes of Health, National Human Genome Research Institute 2022b)
dysmorphology	Dysmorphology is a discipline of clinical genetics that studies and attempts to interpret the patterns of human structures, congenital differences, and syndromes. (Adapted from Hughes and Maloy 2013)
embryo screening	Also known as preimplantation genetic diagnosis (PGD), embryo screening allows couples at risk of transmitting a genetic condition to increase the chances that their future children are unaffected by the disease without going through the process of prenatal diagnosis (i.e., testing of fetal tissue for the presence of disease genes) and being forced to make the difficult decision regarding pregnancy termination. PGD involves extracting a single cell from an eight-cell embryo (created via in vitro fertilization) and analyzing the DNA of that single cell for the presence of one or more disease-associated genetic variants. In the case of a single gene condition,

	only those embryos without the pathogenic variant are implanted in the mother's uterus. For polygenic traits, embryos with the highest scores are selected for implantation, though the outcomes are less predictable in these cases. (Adapted from Pray 2008)
endogenous	Produced inside an organism or cell. The opposite is external (exogenous) production. (From National Institutes of Health, National Cancer Institute 2011)
ethnicity	A sociopolitically constructed system for classifying human beings according to claims of shared heritage often based on perceived cultural similarities (e.g., language, religion, beliefs); the system varies globally. Race is not a synonym. (Adapted from Committee on the Use of Race, Ethnicity, and Ancestry as Population Descriptors in Genomics Research 2023; Boston University Chobanian & Avedisian School of Medicine, Boston Medical Center, and Boston University Medical Group 2021)
eugenics	Eugenics is a discredited belief that selective breeding for certain inherited human traits can improve the "fitness" of future generations. For eugenicists, "fitness" corresponded to a narrow view of humanity and society that developed directly from the ideologies and practices of scientific racism, colonialism, ableism, and imperialism. (From National Institutes of Health, National Human Genome Research Institute 2022b)
evolution	Evolution, as related to genomics, refers to the process by which living organisms change over time through changes in the genome. Such evolutionary changes result from mutations that produce genomic variation, giving rise to individuals whose biological functions or physical traits are altered. Those individuals who are best at adapting to their surroundings leave behind more offspring than less well-adapted individuals. Thus, over successive generations (in some cases spanning millions of years), one species may evolve to take on divergent functions or physical characteristics or may even evolve into a different species. (From National Institutes of Health, National Human Genome Research Institute 2022b)
exogenous	Found or coming from outside something, for example, from outside an organism or cell. The opposite is endogenous.

exome	An exome is the sequence of all the exons in a genome, reflecting the protein-coding portion of a genome. In humans, the exome is about 1.5 percent of the genome. (From National Institutes of Health, National Human Genome Research Institute 2022b)
explicit bias	Explicit bias is the conscious process in which we evaluate another person, deem them “acceptable” or “unacceptable,” and then treat that other person according to how we see them. Unlike implicit bias, which is most often unconscious or automatic, explicit bias is holding a clear and conscious judgment about groups of people based on their identity. (From Jed Foundation, n.d.)
female	A biological term referring to one’s sex, typically associated with XX chromosomes, ovaries, a uterus, and breasts. Females do not exclusively identify as women, nor do they only have male sexual partners.
fertility preservation	The process of saving or protecting eggs, sperm, or reproductive tissue so that a person can use them to have biological children in the future. (From National Institutes of Health, National Institute of Child Health and Human Development 2017)
fertilization	The union of two gametes. During fertilization, sperm and egg fuse to form a diploid zygote to initiate prenatal development. (From Siu et al. 2021)
fetus	An unborn offspring that develops and grows inside the uterus (womb) of humans and other mammals. In humans, the fetal period begins at nine weeks after fertilization of an egg by a sperm and ends at the time of birth. (From National Institutes of Health, National Cancer Institute 2011)
founder effect	A founder effect, as related to genetics, refers to the reduction in genomic variability that occurs when a small group of individuals becomes separated from a larger population. Over time, the resulting new subpopulation will have genotypes and physical traits resembling the initial small, separated group, and these may be very different from the original larger population. A founder effect can also explain why certain inherited diseases are found more frequently in some limited population groups. (From National Institutes of Health, National Human Genome Research Institute 2022b)

gain-of-function	A type of variant in which the altered gene product possesses a new molecular function or a new pattern of gene activation. Gain-of-function mutations are almost always dominant or semidominant. (Adapted from Jackson Labs, n.d.)
gamete	A gamete is a reproductive cell of an animal or plant. In animals, female gametes are called ova or egg cells, and male gametes are called sperm. Ova and sperm are haploid cells, with each cell carrying only one copy of each chromosome. During fertilization, a sperm and ovum unite to form a new diploid organism. (From National Institutes of Health, National Human Genome Research Institute 2022b)
gay	A sexual orientation describing people who are primarily emotionally and physically attracted to people of the same sex and/or gender as themselves. Men, women, and nonbinary people may use this term to describe themselves. (Adapted from Boston University Chobanian & Avedisian School of Medicine, Boston Medical Center, and Boston University Medical Group 2021; Human Rights Campaign 2023b)
gender	Gender involves how a person identifies along a broad, fluid spectrum, rather than strictly by the sex assignment they received at birth. Sex refers to the physical differences (e.g., chromosome composition, anatomy, and physiology) among people who are born male, female, or intersex. The term <i>gender</i> may also be used to refer to the social or cultural constructs of “roles” or “norms” typically associated with being masculine or feminine. Gender does not always directly relate to sex assigned at birth, and usually refers to those aspects of life that are shaped by social forces or to the meaning that society gives to perceived biological differences. Sex is not a synonym for gender. (Adapted from National Institutes of Health, National Human Genome Research Institute 2022b; Boston University Chobanian & Avedisian School of Medicine, Boston Medical Center, and Boston University Medical Group 2021)
gender affirming care	A supportive form of health care. It consists of an array of services that may include medical, surgical, mental health, social, and nonmedical services for transgender and nonbinary people to live authentically as their true selves. (Adapted from Human Rights Campaign 2023a)

gender dysphoria	Clinically significant distress caused when a person's assigned birth gender is not the same as the one with which they identify. (From Human Rights Campaign 2023b)
gender expression	External appearance of one's gender identity, usually expressed through behavior, clothing, body characteristics, or voice, and which may or may not conform to socially defined behaviors and characteristics typically associated with being either masculine or feminine. Gender expression may be masculine, feminine, and/or androgynous and does not necessarily align with assigned sex at birth or gender identity. (Adapted from Boston University Chobanian & Avedisian School of Medicine, Boston Medical Center, and Boston University Medical Group 2021; Human Rights Campaign 2023b)
gene	The gene is considered the basic unit of inheritance. Genes are passed from parents to offspring and contain the information needed to specify physical and biological traits. Most genes code for specific proteins, or segments of proteins, which have differing functions within the body. Humans have approximately 20,000 protein-coding genes. (From National Institutes of Health, National Human Genome Research Institute 2022b)
gene therapy	Gene therapy is a technique that uses a gene(s) to treat, prevent, or cure a disease or medical disorder. Often, gene therapy works by adding new copies of a gene that is broken or by replacing a defective or missing gene in a patient's cells with a healthy version of that gene. Both inherited genetic diseases (e.g., hemophilia and sickle cell disease) and acquired disorders (e.g., leukemia) have been treated with gene therapy. (From National Institutes of Health, National Human Genome Research Institute 2022b)
genetic counseling	Gives you information about how genetic conditions might affect you or your family. The genetic counselor or other health care professional will collect your personal and family health history. They can use this information to determine how likely it is that you or your family member has a genetic condition. Based on this information, the genetic counselor can help you decide whether a genetic test might be right for you or your relative. (From Centers for Disease Control 2024)

genetic determinism	The concept that an organism's phenotype is determined by genes alone and that the role of the environment is negligible to irrelevant. Sometimes also referred to as biological determinism or genetic essentialism. (Adapted from Sarraf and Woodley of Menie 2018)
genetic drift	Genetic drift is a mechanism of evolution characterized by random fluctuations in the frequency of a particular version of a gene (allele) in a population. Though it primarily affects small, isolated populations, the effects of genetic drift can be strong, sometimes causing traits to become overwhelmingly frequent or to disappear from a population. (From National Institutes of Health, National Human Genome Research Institute 2022b)
genetic essentialism	The tendency to infer a person's characteristics and behaviors as based on their genetic makeup is termed <i>genetic essentialism</i> . The environment is not viewed as influencing the development of these traits. Sometimes also referred to as biological or genetic determinism. (Adapted from Dar-Nimrod and Heine 2011)
Genetic Information Nondiscrimi- nation Act	The Genetic Information Nondiscrimination Act (abbreviated GINA) is federal legislation in the United States that protects individuals against discrimination based on their personal genetic information, as it applies to health insurance and employment. These protections are intended to encourage Americans to take advantage of genetic testing as part of their medical care. GINA was signed into law on May 22, 2008. (From National Institutes of Health, National Human Genome Research Institute 2022b)
genetic screening	The process of testing a population for a genetic disease in order to identify a subgroup of people that either have the disease or the potential to pass it on to their offspring. (From National Institutes of Health, National Human Genome Research Institute 2022b)
genetic testing	A laboratory method that looks for changes in genes, gene expression, or chromosomes in the cells or tissue of a patient. This can be carried out in presymptomatic, predictive, diagnostic, management, or reproductive settings, among others. (Adapted from National Institutes of Health, National Cancer Institute 2012)
genitalia	Refers to external sex organs, such as the penis and vulva.

- genitoplasty A form of gender affirming care sometimes referred to as bottom surgery that reconstructs existing reproductive tissue into new genitalia that aligns with one's gender identity. Historically, these procedures were performed on children without their consent to try to enforce binary constructions of sex on intersex children.
- genome The genome is the entire set of DNA instructions found in a cell. In humans, the genome consists of 23 pairs of chromosomes located in the cell's nucleus, as well as a small chromosome in the cell's mitochondria. A genome contains all the information needed for an individual to develop and function. (From National Institutes of Health, National Human Genome Research Institute 2022b)
- genome editing Genome editing is a method that lets scientists change the DNA of many organisms, such as plants, bacteria, and animals, including humans. Editing DNA has recently been used to treat hemoglobinopathies like sickle cell and beta thalassemia. (Adapted from National Institutes of Health, National Human Genome Research Institute 2022b)
- genome-wide association study (GWAS) A genome-wide association study (abbreviated GWAS) is a research approach used to identify genomic variants that are statistically associated with risk for a disease or a particular trait. The method involves surveying the genomes of many people, looking for genomic variants that occur more frequently in those with a specific disease or trait compared to those without the disease or trait. Once such genomic variants are identified, they are typically used to search for nearby variants that contribute directly to the disease or trait. (From National Institutes of Health, National Human Genome Research Institute 2022b)
- genomic medicine Genomic medicine is a medical discipline that involves using a person's genomic information as part of their clinical care. Other similar terms include *individualized medicine*, *personalized medicine*, and *precision medicine*. For some conditions, genomic information can be used to help diagnose disease, predict outcomes, and guide treatment. (From National Institutes of Health, National Human Genome Research Institute 2022b)

genotype	A genotype is a scoring of the type of variant present at a given location (i.e., a locus) in the genome. It can be represented by symbols. For example, BB, Bb, and bb could be used to represent a given variant in a gene. Genotypes can also be represented by the actual DNA sequence at a specific location, such as CC, CT, and TT. DNA sequencing and other methods can be used to determine the genotypes at millions of locations in a genome in a single experiment. Some genotypes contribute to an individual's observable traits, called the phenotype. (Adapted from National Institutes of Health, National Human Genome Research Institute 2022b)
genotyping	A laboratory process in which an individual's DNA is analyzed for specific nucleotides or bases to determine whether certain variants are present. Genotyping differs from sequencing, in which all the nucleotides comprising a specific length of DNA are assessed (e.g., within a gene, exome, or genome). (From National Institutes of Health, National Cancer Institute 2012)
gestational carrier	This term was originally used in reference to a female who carries and gives birth to a baby for another person (i.e., a synonym for gestational surrogate). Eggs from an egg donor are fertilized in the laboratory with sperm from a sperm donor to make an embryo. The embryo is implanted in the uterus of the gestational carrier, who carries the baby until birth. In the original definition, the gestational carrier is not genetically related to the baby; however, this term can also be used to refer to a pregnant person who is carrying their own pregnancy but does not identify as a woman, in which case they may not wish to be referred to as the mother. (Adapted from National Institutes of Health, National Cancer Institute 2011)
gestational surrogate	A female who carries and gives birth to a baby for another person. Eggs from an egg donor are fertilized in the laboratory with sperm from a sperm donor to make an embryo. The embryo is implanted in the uterus of the gestational surrogate, who carries the baby until birth. The gestational surrogate is not genetically related to the baby. Can also be called gestational carrier. (Adapted from National Institutes of Health, National Cancer Institute 2011)
gonad	The part of the reproductive system that produces and releases eggs (ovary) or sperm (testicle/testis). (From National Institutes of Health, National Cancer Institute 2011)

gonadal dysgenesis	Atypical development of a gonad (ovary or testicle), often associated with a genetic syndrome. Can also present with infertility. This is an example of a variation in a sex characteristic (VSC).
hemoglobin	A protein inside red blood cells that carries oxygen from the lungs to tissues and organs in the body and carries carbon dioxide back to the lungs. (From National Institutes of Health, National Cancer Institute 2011)
heteronormativity	The assumption that heterosexuality is the standard for defining “normal” sexual behavior and that male-female differences and binary gender roles are the natural and immutable essentials in human relations. According to some social theorists, this assumption is fundamentally embedded in, and legitimizes, social and legal institutions that devalue, marginalize, and discriminate against people who deviate from its normative principle (e.g., LGBTQ+ individuals). (From American Psychological Association 2023)
heterozygous	Heterozygous, as related to genetics, refers to having inherited different versions (alleles) of a gene or genomic marker from each biological parent. Thus, an individual who is heterozygous for a genomic marker has two different versions of that marker. By contrast, an individual who is homozygous for a marker has identical versions of that marker. (Adapted from National Institutes of Health, National Human Genome Research Institute 2022b)
homozygous	Homozygous, as related to genetics, refers to having inherited the same versions (alleles) of a gene or genomic marker from each biological parent. Thus, an individual who is homozygous for a genomic marker has two identical versions of that marker. By contrast, an individual who is heterozygous for a marker has two different versions of that marker. (Adapted from National Institutes of Health, National Human Genome Research Institute 2022b)
Huntington's disease	Huntington's disease is a rare, inherited disorder associated with the progressive loss of brain and muscle function. Symptoms usually develop during middle age and may include uncontrolled movements, loss of intellectual abilities, and various emotional and psychiatric symptoms. Huntington's disease is inherited as an autosomal dominant trait, meaning that a single mutated copy of the responsible gene (called HTT) is sufficient to cause the disease. (From National Institutes of Health, National Human Genome Research Institute 2022b)

identity	An individual's sense of self defined by (a) a set of physical, psychological, and interpersonal characteristics that is not wholly shared with any other person and (b) a range of affiliations, group belonging, and social roles. Such a sense is derived from one's body sensations; one's body image; and the feeling that one's memories, goals, values, expectations, and beliefs belong to the self. (Adapted from American Psychological Association 2018)
implicit association test (IAT)	The implicit association test (IAT) measures unconscious attitudes and beliefs that people may be unwilling or unable to report. The IAT measures the strength of associations between concepts (e.g., Black people, gay people) and evaluations (e.g., good, bad) or stereotypes (e.g., athletic, clumsy). The test is designed as a sorting task that is easier to complete when items that trigger a similar personal view share the same response key. (From "Project Implicit" 2011)
implicit bias	Negative or positive associations people unknowingly hold, also known as unconscious or hidden bias. They are expressed automatically, without conscious awareness, providing unearned advantage to those in dominant groups and unearned disadvantage to those in marginalized groups. Many studies have indicated that implicit biases affect individuals' attitudes and actions, thus creating real-world implications, even though individuals may not even be aware that those biases exist within themselves. Notably, implicit biases have been shown to undermine individuals' stated commitments to equality and fairness, thereby producing behavior that diverges from the explicit attitudes that many people profess. The term <i>implicit bias</i> many times is used to avoid naming internalized racism, internalized sexism, and so on. (From Boston University Chobanian & Avedisian School of Medicine, Boston Medical Center, and Boston University Medical Group 2021)
in vitro fertilization (IVF)	A procedure in which eggs are removed from an ovary and combined with sperm outside the body to form embryos. The embryos are grown in the laboratory for several days and then either placed in a gestational carrier's uterus or cryopreserved (frozen) for future use. (Adapted from National Institutes of Health, National Cancer Institute 2011)

- incidental finding** Incidental findings are observations, results, or other findings that may occur during analysis but are unrelated to the initial goals of the analysis. The term was originally coined with respect to imaging studies, such as X-rays, CT scans, MRI scans, or ultrasound exams, but it is also relevant in the context of an unexpected result when conducting exome or genome testing, where the result is unrelated to the immediate clinical question. The term *secondary finding* is often used in the context of genetic testing. In either case, the incidental/secondary finding may be clinically significant and may require follow-up.
- informed consent** The process in which a health care provider educates a patient about the risks, benefits, and alternatives of a given procedure or intervention. The patient must be competent to make a voluntary decision about whether to undergo the procedure or intervention. Informed consent is both an ethical and legal obligation of medical practitioners in the United States and originates from the patient's right to direct what happens to their body. Similarly, informed consent is an ethical pillar of clinical research, as well. (Adapted from Shah et al. 2024)
- intellectual disability** Refers to neurodevelopmental conditions that affect functioning in two areas: (1) cognitive functioning, such as learning, problem solving, and judgment; and (2) adaptive functioning, that is, activities of daily life such as communication skills and social participation. These differences begin early in the developmental period, typically before age 18 years for diagnosis. Previously referred to as mental retardation, a term that has been retired due to the harm it causes. (Adapted from Luna 2024)
- intersectionality** First coined by legal scholar Kimberlé Crenshaw, this term describes the overlapping or interdependent systems of discrimination related to age, disabilities, ethnicity, gender, geographic location, sex, socioeconomic status, sexuality, and other identities that in combination create intersecting forms of oppression and inequality. (Adapted from Stanford University, n.d.)
- intersex** Describes a group of congenital conditions in which the reproductive organs, genitals, and/or other sexual anatomy do not develop according to typical female or male developmental patterns. *Intersex* can also be used as an identity term for

	someone with one of these conditions. The medical community sometimes uses the term <i>differences of sex development</i> (DSD) or <i>variations of sex characteristics</i> (VSC) to describe intersex conditions; however, the term <i>intersex</i> is recommended by several intersex community members and groups. Some estimates indicate that approximately 1.7 percent of the population is born with intersex traits. (Adapted from Boston University Chobanian & Avedisian School of Medicine, Boston Medical Center, and Boston University Medical Group 2021)
lesbian	A sexual orientation that describes a woman who is primarily emotionally and physically attracted to other women. Women and nonbinary people may use this term to describe themselves. (Adapted from Boston University Chobanian & Avedisian School of Medicine, Boston Medical Center, and Boston University Medical Group 2021; Human Rights Campaign 2023b)
LGBTQ+	An acronym that stands for lesbian, gay, bisexual, transgender, queer, intersex, asexual, and gender diverse, and/or those who identify on the spectrum of sexuality and/or gender identity. Sometimes written as LGBT, LGBTQ, LGBTQ2, which is inclusive of two-spirit. The plus sign signifies the limitless sexual orientations and gender identities used by members of this community. (Adapted from Boston University Chobanian & Avedisian School of Medicine, Boston Medical Center, and Boston University Medical Group 2021; Human Rights Campaign 2023b)
locus	A locus, as related to genomics, is a physical site or location within a genome (such as a gene or another DNA segment of interest), somewhat like a street address. The plural of locus is loci. (From National Institutes of Health, National Human Genome Research Institute 2022b)
male	This is a biological term referring to one's sex, typically associated with XY chromosomes, testes, and a penis. Males do not exclusively identify as men, nor do they only have female sexual partners.
mass spectrometry (mass spec)	An analytical technology capable of measuring a wide range of biomarkers by separating and quantifying ions based on their mass-to-charge ratios. This instrument reliably detects a range of metabolites and has thus been effectively used in newborn screening programs for numerous genetic conditions in neonates, especially those related to inborn errors of

	metabolism. (Adapted from American College of Medical Genetics/American Society of Human Genetics Test and Technology Transfer Committee Working Group 2000)
medical model of disability	The medical model views disability as resulting from an individual person's physical or mental limitations and is not connected to the social or geographical environments. Defines disability as a deficiency or abnormality that requires treatment or fixing of the individual. (Adapted from "Medical and Social Models of Disability," n.d.)
meiosis	Meiosis is a type of cell division in sexually reproducing organisms that reduces the number of chromosomes in gametes (the sex cells, or egg and sperm). In humans, body (or somatic) cells are diploid, containing two sets of chromosomes (one from each parent). To maintain this state, the egg and sperm that unite during fertilization must be haploid, with a single set of chromosomes. During meiosis, each diploid cell undergoes reductive cell division to yield haploid daughter cells—the gametes. (Adapted from National Institutes of Health, National Human Genome Research Institute 2022b)
microsatellite	Microsatellite, as related to genomics, refers to a short segment of DNA, usually one to six or more base pairs in length, that is repeated multiple times in succession at a particular genomic location. These DNA sequences are typically non-coding. The number of repeated segments within a microsatellite sequence often varies among people, which makes them useful as polymorphic markers for studying inheritance patterns in families or for creating a DNA fingerprint from crime scene samples. (From National Institutes of Health, National Human Genome Research Institute 2022b)
miscegenation	Marriage, sexual activity, or reproduction among individuals of different racial or ethnic groups, especially when this is emphasized by marked physical differences (e.g., skin color). The term has traditionally carried a heavy sense of disapproval and is now largely archaic and disfavored. Related terms include <i>exogamy</i> , <i>intermarriage</i> , and <i>outbreeding</i> . (Adapted from American Psychological Association 2018)
misinformation	False or inaccurate information. <i>Disinformation</i> is a related term that, in addition to being false information, is deliberately intended to mislead. (Adapted from American Psychological Association, n.d.)

misogyny	Hatred of women, often manifested in sexual discrimination, denigration, or violence against and sexual objectification of women. (From Boston University Chobanian & Avedisian School of Medicine, Boston Medical Center, and Boston University Medical Group 2021)
monogenic	Monogenic disorders are caused by a pathogenic variant in a single gene. These conditions and traits are generally rare in the population. See also <i>single gene disorders</i> . (Adapted from National Institutes of Health, National Human Genome Research Institute 2018)
moral injury	Refers to the strong cognitive dissonance and emotional response that can occur in response to events that violate a person's moral or ethical code. This term was originally meant to describe experiences of military personnel during wartime, but also applies to health care workers during traumatic events, for example, when they are unable to provide the standard of care that they strive to deliver. (Adapted from Williamson et al. 2021)
mutation	A mutation is a change in the DNA sequence of an organism. Mutations can result from errors in DNA replication during cell division, exposure to mutagens, or a viral infection. Germline mutations (that occur in eggs and sperm) can be passed on to offspring, while somatic mutations (that occur in body cells) are not passed on. (From National Institutes of Health, National Human Genome Research Institute 2022b)
natural selection	A nonrandom difference in reproductive output among replicating entities, often due indirectly to differences in survival in a particular environment, leading to an increase in the proportion of beneficial, heritable characteristics within a population from one generation to the next. (From American Psychological Association 2018)
naturalistic fallacy	Attributed to G. E. Moore, the naturalistic fallacy is an ethics principle that ascribes moral acceptability to things that are natural, as genetic phenomena are. For example, a behavioral trait is more likely to be viewed as good if it is perceived to result from a genetic predisposition. According to this fallacy, if someone is thought to be born a certain way, they are viewed more favorably than if they choose to act or live that way. (Adapted from Dar-Nimrod and Heine 2011)

neurodiversity	A term that is used to describe individuals with diverse or variant cognitive functioning. Both neurodiverse and neurotypical originated as a more comprehensive way to describe autism, and now neurodiverse is used more broadly to include other developmental differences. Neurodiversity is the concept that brain differences are just that: differences. Thus, conditions like dyslexia, autism, and ADHD are not “abnormal.” Though they may be disabilities, they are not flaws. They are simply variations of the human brain. (Adapted from Boston University Chobanian & Avedisian School of Medicine, Boston Medical Center, and Boston University Medical Group 2021)
newborn screening	Newborn screening is a set of laboratory tests performed on newborn babies to detect a set of known genetic conditions. Typically, this testing is performed on a blood sample obtained from a heel prick when the baby is two or three days old. In the United States, offering newborn screening is mandatory for a defined set of genetic diseases, although the exact set differs from state to state. Newborn screening tests focus on conditions for which early diagnosis is important to treating or preventing disease. (Adapted from National Institutes of Health, National Human Genome Research Institute 2022b)
noncoding variant	Noncoding DNA corresponds to the portions of an organism’s genome that do not code for amino acids, the building blocks of proteins. Some noncoding DNA sequences are known to serve functional roles, such as in the regulation of gene expression, while other areas of noncoding DNA have no known function. (From National Institutes of Health, National Human Genome Research Institute 2022b)
nonbinary	Describes a person whose gender identity falls outside of the traditional gender binary structure of girl/woman and boy/man. Nonbinary can also be used as an umbrella term to encompass identities such as agender, bigender, genderqueer, gender expansive, or gender fluid. Sometimes abbreviated as NB or enby. (Adapted from Boston University Chobanian & Avedisian School of Medicine, Boston Medical Center, and Boston University Medical Group 2021; Human Rights Campaign 2023b)
nondisjunction	The failure of two members of a chromosome pair to separate during cell division (meiosis or mitosis), so that both members of the pair are passed to a single daughter cell, while the

	other daughter cell receives no copies of this chromosome. (Adapted from Cohn, Scherer, and Hamosh 2024)
nucleotides	A nucleotide is the basic building block of nucleic acids (RNA and DNA). A nucleotide consists of a sugar molecule (either ribose in RNA or deoxyribose in DNA) attached to a phosphate group and a nitrogen-containing base. The bases used in DNA are adenine (A), cytosine (C), guanine (G), and thymine (T). In RNA, the base uracil (U) takes the place of thymine. DNA and RNA molecules are polymers made up of long chains of nucleotides. (From National Institutes of Health, National Human Genome Research Institute 2022b)
oligogenic	Oligogenic is an intermediate term between monogenic and polygenic. Conditions and traits that are considered oligogenic are influenced by a small number of genes, in contrast to monogenic conditions that are determined by single genes and polygenic conditions that can be modified by hundreds or more genes.
oogenesis	The process by which germ cells divide and differentiate to produce female gametes (ova). In human females, primary oocytes are formed in the ovary during embryonic development by the proliferation and differentiation of precursor cells called oogonia (singular: oogonium). The primary oocytes enter into the first division of meiosis, but then remain suspended at this stage of cell division until puberty. Thereafter, roughly once a month until menopause, one primary oocyte resumes meiosis and completes the first meiotic division to produce two unequally sized daughter cells: the larger one is the secondary oocyte, and the smaller is a polar body. Following ovulation, the secondary oocyte undergoes the second meiotic division to produce an ovum and another polar body. The first polar body might also divide to produce two tiny cells, resulting in three polar bodies, which are normally nonfunctional and degenerate. (Adapted from American Psychological Association 2018)
ovotestes	In typical sex development, the fetus develops two tiny organs called proto-gonads. Usually these proto-gonads will become either testes or ovaries. Sometimes an ovotestis develops instead, which contains some ovary-type cells and some testis-type cells. Although a person may be born with two ovotestes, it is much more common for a person to be born with a typical ovary or testis on one side and an ovotestis on the

	other. People with ovotestes may have a typical genital appearance at birth, with either a vulva and labia or penis and scrotum, and some are more intermediate. Because there may be many different chromosomal and internal anatomical combinations, children may need other tests to evaluate their unique situations. The testicular part of ovotestes may have an increased cancer risk, but this does not automatically mean that they need to be removed immediately upon identification of the organ. (Adapted from interACT, 2021)
pangenome	The collective whole-genome sequences of multiple individuals representing the genetic diversity of the species. (Wang et al. 2022)
pathogenic variant	A genetic alteration that increases an individual's susceptibility or predisposition to a certain disease or condition. When such a variant (or mutation) is inherited, development of symptoms is more likely, but not certain. Also called deleterious mutation, disease-causing mutation, predisposing mutation, and susceptibility gene mutation. (Adapted from National Institutes of Health, National Cancer Institute 2012)
patriarchy	Structural and ideological system that perpetuates the privileging of particular kinds of masculinity and cisgender men. A system in which cisgender men have institutional control and dominance. (From Boston University Chobanian & Avedisian School of Medicine, Boston Medical Center, and Boston University Medical Group 2021)
personal genomics	Personal genomics, also referred to as direct-to-consumer genetic testing and consumer genomics, involves a consumer accessing genetic testing from a commercial company apart from a health care provider. The testing available through these means spans from genetic genealogy to health and traits. (Adapted from National Academies of Sciences, Engineering, and Medicine 2020)
phenotype	Phenotype refers to an individual's observable traits, such as height, eye color, and blood type. A person's phenotype is determined by both their genomic makeup (genotype) and environmental factors. (From National Institutes of Health, National Human Genome Research Institute 2022b)
physical disability	A physical disability is a physical condition that affects a person's mobility, physical capacity, stamina, or dexterity. Examples of physical disability include multiple sclerosis, muscular dystrophy, chronic arthritis, cerebral palsy, chronic

	fatigue syndrome, fibromyalgia, spina bifida, loss of limbs, and spinal cord injury. A person with physical disability may have difficulties moving and coordinating parts of their body for everyday life. Some may also consider sensory disabilities (e.g., low vision, deafness) as a form of physical disability. The difficulties and barriers a person may face will depend on the type of physical disability, available accommodations, and other environmental factors. (Adapted from National Disability Services 2024)
placebo	An inactive substance or other intervention that looks the same as, and is given in the same way as, an active drug or treatment being tested. The effects of the active drug or other intervention are compared to the effects of the placebo, and the person taking the placebo generally is not aware if they are taking an active drug or the placebo. (Adapted from National Institutes of Health, National Cancer Institute 2011)
placenta	The organ that nourishes the developing fetus in the uterus. (From National Institutes of Health, National Cancer Institute 2011)
polygenic	A polygenic trait is a characteristic, such as height or skin color, that is influenced by two or more genes. Because multiple genes are involved, polygenic traits do not follow the patterns of Mendelian inheritance. Many polygenic traits can be influenced by dozens, hundreds, or even thousands of genes and are also influenced by the environment; thus they are called multifactorial. (Adapted from National Institutes of Health, National Human Genome Research Institute 2022b)
polygenic risk score (polygenic index)	A polygenic risk score (abbreviated PRS) uses genomic information alone to assess a person's chances of having or developing a particular medical condition. A person's PRS is a statistical calculation based on the presence or absence of multiple genomic variants, without taking environmental or other factors into account. The term <i>polygenic index</i> has also been proposed to remove a disease-associated connotation while still quantifying the contributions of multiple genes. (Adapted from National Institutes of Health, National Human Genome Research Institute 2022b)
positionality	Positionality refers to where one is located in relation to one's various social identities (gender, race, class, ethnicity, ability, geographical location, etc.). The combination of these identities and their intersections shape how we understand and

	engage with the world, including our knowledge, perspectives, and practices. (“Positionality Statement,” n.d.)
precision medicine	Precision medicine (generally considered analogous to personalized medicine or individualized medicine) is an innovative approach that uses an individual’s genomic, environmental, and lifestyle information to guide decisions related to their medical management. The goal of precision medicine is to provide a more individualized approach for the prevention, diagnosis, and treatment of disease, targeting the right treatments to the right patients at the right time. (Adapted from National Institutes of Health, National Human Genome Research Institute 2022b; Center for Devices and Radiological Health 2018)
preconception genetic testing	Preconception genetic testing, also known as carrier screening, occurs before having children and generally involves testing the prospective parents to determine if they are asymptomatic carriers of a recessive condition to better predict whether their future offspring could be affected and to offer reproductive choices. (Adapted from Andermann and Blancaquart 2010)
pregnancy termination	Pregnancy termination is also referred to as abortion. Abortion is the termination of a pregnancy by removal or expulsion of a fetus before it can survive outside the uterus. It can occur spontaneously, known as a miscarriage, or be induced intentionally through medication or surgical procedures. Abortion is a routine form of health care that can be necessary to preserve the health of the gestational carrier, and studies have found that when performed legally and under the supervision of a trained professional, it can be safer than pregnancy and childbirth. Approximately 25 percent of women will have an abortion by age 45. (Adapted from Planned Parenthood, Action Fund, n.d.; Raymond and Grimes 2012)
pregnant people	Pregnant people is a gender-neutral term to refer to anyone with a uterus who is pregnant, regardless of their gender identity. This is a more inclusive term than <i>pregnant women</i> .
preimplantation genetic testing	Preimplantation genetic testing comprises a group of genetic assays used to evaluate embryos created by in vitro fertilization before transfer to the uterus. Preimplantation genetic testing–monogenic is targeted to single gene conditions, and preimplantation genetic testing–aneuploidy is a broader test

that screens for aneuploidy in all chromosomes, including the 22 pairs of autosomes and the sex chromosomes X and Y. To test embryos that are at risk for chromosome gains and losses related to parental structural chromosomal alterations (e.g., translocations, inversions, deletions, and insertions), preimplantation genetic testing–structural rearrangements is used. (Adapted from American College of Obstetricians and Gynecologists 2023)

race	Race is a social construct used to group people. Race was constructed as a hierarchical human-grouping system, generating racial classifications to identify, distinguish, and marginalize some groups across nations, regions, and the world. Race divides human populations into groups often based on physical appearance, social factors, and cultural backgrounds. This social construct was created and used to justify social, political, and economic oppression of people of color by White people. In short, race is the product of racism rather than the other way around. As a social construct or a social invention with political, economic, and historical context, race is both an idea and a way of organizing society that varies over time and from place to place. (Adapted from National Institutes of Health, National Human Genome Research Institute 2022b; Boston University Chobanian & Avedisian School of Medicine, Boston Medical Center, and Boston University Medical Group 2021; Committee on the Use of Race, Ethnicity, and Ancestry as Population Descriptors in Genomics Research 2023)
race correction	The insertion of race into diagnostic algorithms and practice guidelines that adjusts or “corrects” their outputs based on a patient’s race or ethnicity. This practice is often carried out as a proxy for more immediate biological factors that explain any observed health care disparities; race correction can unintentionally exacerbate these disparities. (Adapted from Vyas, Eisenstein, and Jones 2020)
race-based medicine	The system by which research mischaracterizing race as an essential, biological variable translates into clinical practice, leading to inequitable care. (Adapted from Cerdeña, Plaisime, and Tsai 2020)
race-norming	The use of different cutoff scores in diagnostic assessments for patients from different racial groups. (Adapted from American Psychological Association 2018)

racism	A system of power and oppression, codified into laws, policies, and institutions, based on the socially and politically constructed concept of race, that advantages the dominant group (White people) and disadvantages nondominant groups (people of color). (From Boston University Chobanian & Avedisian School of Medicine, Boston Medical Center, and Boston University Medical Group 2021)
rare variant	Rare variants are defined as genetic variants with an allele frequency <1 percent in the population.
recessive	Recessive, as related to genetics, refers to the relationship between an observed trait and the two inherited versions of a gene related to that trait. Individuals inherit two versions of each gene, known as alleles, from each parent. In the case of a recessive trait, the alleles of the trait-causing gene are the same, and both (recessive) alleles must be present to express the trait. A recessive allele does not produce a trait at all when only one copy is present. This contrasts to a dominant trait, which requires that only one of the two alleles be present to express the trait. (From National Institutes of Health, National Human Genome Research Institute 2022b)
red blood cells	A type of blood cell that is made in the bone marrow and found in the blood. Red blood cells contain a protein called hemoglobin, which carries oxygen from the lungs to all parts of the body. Also called erythrocyte and RBC. (Adapted from National Institutes of Health, National Cancer Institute 2011)
redlining	Redlining refers to the practice of systematically denying various services (e.g., mortgage credit access and home insurance) to residents of specific neighborhoods, often based on race and primarily within urban communities. This racially discriminatory housing practice is responsible for the segregation of communities, leaving people of color in neighborhoods where they have increased exposure to health-related risk factors, and is linked to increased risk of diabetes, hypertension, and early mortality due to heart disease. (Adapted from Egede et al. 2023)
reparation	Reparation (or “reparative justice” as it is sometimes called) is a key element of providing justice to victims of serious human rights abuses or atrocities. Reparations can be cash payments, land grants, or other forms of compensation. Reparations for descendants of enslaved individuals and people who were forcibly sterilized by the government have been proposed in the

	United States, and some states have started distributing funds for victims of eugenic sterilization. (Adapted from U.S. Department of State 2016)
repressor	A repressor, as related to genomics, is a protein that inhibits the expression of one or more genes. The repressor protein works by binding to the promoter region of the gene(s), which prevents the production of messenger RNA (mRNA). Repressor proteins are essential for the regulation of gene expression in cells. (From National Institutes of Health, National Human Genome Research Institute 2022b)
right not to know	The “right not to know” refers to the idea that adults should be permitted to control whether they receive genetic information—particularly information about the risk of future illness—and that their desire not to know certain kinds of information should be respected. The justifications offered for a right not to know are twofold, grounded in respect for decisional autonomy and/or an interest in protecting individuals from receiving unwanted and potentially harmful information. An adult’s right not to know genetic information has implications for children; testing a child preempts the option for that child not to learn the information as a future autonomous adult. (From Berkman and Hull 2014)
safe space	A safe space is a physical space or setting where a person, especially one from a marginalized group, can feel secure, respected, and valued for who they are. These spaces also foster open and honest dialogue and facilitate a sense of belonging. Safe spaces are, at their core, inclusive and supportive. The term originally referred specifically to spaces that were safe for the LGBTQ+ community, but the term has since grown into an umbrella term that broadly covers all identities.
scientific racism	Scientific racism is a historical pattern of ideologies that generate and perpetuate pseudoscientific racist beliefs that lead to racial bias and discrimination in science and research. Leading scientists across scientific institutions in the 19th and early 20th centuries were proponents of such ideologies. By the mid-20th century, pseudoscientific racist beliefs were widely disproven. However, evidence shows that scientific racism persists in science and research. (From National Institutes of Health, National Human Genome Research Institute 2022b)

selective pressure	In population genetics, the operation of forces that determine the relative fitness of a genotype in the population, thus affecting organism survival and the frequency of an allele in the population. (Adapted from Cohn, Scherer, and Hamosh 2024)
sex	Sex is a multidimensional construct based on a cluster of anatomical and physiological traits that include external genitalia, secondary sex characteristics, gonads, chromosomes, and hormones. Sex traits are often assumed to be unambiguous and correspond to a single sex; however, sex traits may not all correspond to the same sex or align with these binary categories, and thus with sex assigned at birth. For example, people who are intersex or have variations in sex characteristics (VSC) have traits that correspond to multiple sexes. Sex should not be conflated with gender. Furthermore, the term <i>biological sex</i> has been weaponized by those seeking to operate within a binary sex framework. (Adapted from Committee on Measuring Sex, Gender Identity, and Sexual Orientation et al. 2022)
sex assigned at birth	Sex in Western cultures is typically assigned at birth by medical professionals as either male or female, based solely on visual inspection of external genitalia. Also referred to as birth sex, natal sex, or sex. It is not valid to use “biology” or “science” to claim sex assigned at birth or gender are fixed, binary concepts. Science demonstrates the fact that sex and gender are more complicated and involve much more than one’s sexual organs. (Adapted from Boston University Chobanian & Avedisian School of Medicine, Boston Medical Center, and Boston University Medical Group 2021; Committee on Measuring Sex, Gender Identity, and Sexual Orientation et al. 2022)
sex chromosome	A sex chromosome is a type of chromosome involved in sex determination. Humans and most other mammals have two sex chromosomes, X and Y, that in combination determine the sex of an individual. Females typically have two X chromosomes in their cells, while males generally have one X and one Y. (Adapted from National Institutes of Health, National Human Genome Research Institute 2022b)
sex hormone	Any of the hormones that stimulate various reproductive functions. Primary sources of sex hormones are male and female gonads (i.e., testes and ovaries), which are stimulated to

- produce sex hormones by the pituitary gland. The principal male sex hormones (androgens) include testosterone; female sex hormones include the estrogens, progesterone, and prolactin. Importantly, these sex hormones are not exclusively found in a single sex. (Adapted from American Psychological Association 2018)
- sexual orientation An inherent or immutable enduring emotional, romantic, or sexual attraction to other people. Note: an individual's sexual orientation is independent of their gender identity. Some common sexual orientations include gay, lesbian, straight, bisexual, pansexual, and asexual. (Adapted from Human Rights Campaign 2023b)
- sickle cell disease An inherited disease in which the red blood cells have a crescent shape due to beta-globin polymerization under low-oxygen conditions. These sickle-shaped cells have difficulty navigating small blood vessels and do not last as long as normal red blood cells. Blockage of the capillaries can lead patients to experience pain crises. Sickle cell disease is caused by a pathogenic variant (change) in one of the genes for hemoglobin (the substance inside red blood cells that binds to oxygen and carries it from the lungs to the tissues). It is most common in people from areas where malaria is endemic. (Adapted from National Institutes of Health, National Cancer Institute 2011)
- single gene disorders Single gene disorders are caused by a pathogenic variant in one causative gene. These conditions and traits are generally rare in the population. See also *monogenic disorders*. (Adapted from National Institutes of Health, National Human Genome Research Institute 2018)
- single nucleotide variant A single nucleotide variant (SNV) is a genomic variant at a single base position in the DNA. Scientists study if and how SNVs in a genome influence health, disease, drug response, and other traits. (From National Institutes of Health, National Human Genome Research Institute 2022b)
- social construct A classification system created by humans. The classification is *not* based on inherent biological differences that apply to all members of the community. The classification can instead be based on behavioral norms and expectations (e.g., gender) or a hierarchical structure designed to maintain power structures (e.g., race). When a classification system is created by people, we can also see that it varies from one society to the next.

social model of disability	The social model views disability as a consequence of environmental, social, and attitudinal barriers that may prevent people from fully participating in society. Defines disability as a difference in interaction between an individual and society that may benefit from making social spaces more accessible. (Adapted from “Medical and Social Models of Disability,” n.d.)
social/emotional disability	A person with a social/emotional disability has persistent and consistent behavioral responses that adversely affect performance in an age-appropriate environment. Examples include mental health conditions, such as anxiety disorders, or psychiatric conditions, as well as eating disorders, obsessive compulsive disorder, and others.
somatic	Somatic cells are the cells in the body other than sperm and egg cells (which are called germ cells). In humans, somatic cells are diploid, meaning they contain two sets of chromosomes, one inherited from each parent. DNA mutations in somatic cells can affect an individual, but they cannot be passed on to their offspring. (From National Institutes of Health, National Human Genome Research Institute 2022b)
spermatogenesis	The production of spermatozoa in the seminiferous tubules of the testis. Male germ cells (spermatogonia; singular, spermatogonium) lining the seminiferous tubules mature into primary spermatocytes, which undergo meiosis to eventually result in mature spermatozoa. Through reductive meiotic divisions, diploid stem cells give rise to haploid spermatids that then mature into spermatozoa. The process is controlled by pituitary gonadotropins, which promote the release of testosterone from the interstitial cells of the testis, which in turn stimulates sperm production. Spermatogenesis is continuous in human males after the onset of puberty. (Adapted from American Psychological Association 2018)
stem cell	Stem cells are cells with the potential to develop into many different types of cells in the body. (From National Institutes of Health, National Library of Medicine 2016)
stereotypes	A set of cognitive generalizations (e.g., beliefs, expectations) about the qualities and characteristics of the members of a group or social category. Stereotypes, like schemas, simplify and expedite perceptions and judgments, but they are often exaggerated, negative rather than positive, and resistant to revision even when perceivers encounter individuals with qualities that are not congruent with the stereotype. (From American Psychological Association 2018)

systemic racism	Racial bias across institutions and society over time. It reflects the cumulative and compounded effects of an array of factors, such as public policies, institutional practices, cultural representations, and other norms, that work in various, often reinforcing, ways to perpetuate racial inequity. Also referred to as structural racism. (Adapted from Boston University Chobanian & Avedisian School of Medicine, Boston Medical Center, and Boston University Medical Group 2021)
teratogen	A teratogen is any agent that causes a developmental change following fetal exposure during pregnancy. Teratogens are usually discovered after an increased prevalence of a particular congenital condition. (Adapted from Genetic Alliance and District of Columbia Department of Health 2010)
thalidomide	A teratogen that was used to treat morning sickness in the early 1960s. Exposure of the fetus to thalidomide during early prenatal development resulted in cases of phocomelia, where interruptions to typical limb development resulted in hands and feet that are attached to shortened or absent arms and legs. (Adapted from Genetic Alliance and District of Columbia Department of Health 2010)
trait	A trait, as related to genetics, is a specific characteristic of an individual. Traits can be determined by genes, environmental factors, or a combination of both. Traits can be qualitative (such as eye color) or quantitative (such as height or blood pressure). A given trait is part of an individual's overall phenotype. (From National Institutes of Health, National Human Genome Research Institute 2022b)
transgender	An umbrella term for people whose gender identity and/or expression is different from cultural expectations based on the sex they were assigned at birth. Sometimes abbreviated as trans. Although the word <i>transgender</i> and our modern definition of it only came into use in the late 20th century, people who would fit under this definition have existed in every culture throughout recorded history. Being transgender does not imply any specific sexual orientation. Therefore, transgender people may identify as straight, gay, lesbian, bisexual, and so on. Some transgender individuals undergo a process called “gender transitioning” to match their gender identity more closely with their outward appearance. This can include changing clothes, names, or pronouns to fit their gender identity. It may also include gender affirming health care needs,

	such as hormones or surgeries. (Adapted from Boston University Chobanian & Avedisian School of Medicine, Boston Medical Center, and Boston University Medical Group 2021)
translocation	A genetic change in which a piece of one chromosome breaks off and attaches to another chromosome. Sometimes pieces from two different chromosomes will trade places with each other, forming a reciprocal translocation. (Adapted from National Institutes of Health, National Cancer Institute 2012)
typical (instead of normal)	Typical is offered as an alternative to normal. When <i>normal</i> is used to reference a group, it suggests that anyone who is different is abnormal. Typical, for example, neurotypical as compared to neurodivergent, is more inclusive of a range of identities.
unconscious bias	Negative or positive associations people unknowingly hold, also known as implicit or hidden bias. They are expressed automatically, without conscious awareness, providing unearned advantage to those in dominant groups and unearned disadvantage to those in marginalized groups. Many studies have indicated that implicit biases affect individuals' attitudes and actions, thus creating real-world implications, even though individuals may not even be aware that those biases exist within themselves. Notably, unconscious biases have been shown to undermine individuals' stated commitments to equality and fairness, thereby producing behavior that diverges from the explicit attitudes that many people profess. (From Boston University Chobanian & Avedisian School of Medicine, Boston Medical Center, and Boston University Medical Group 2021)
variant	An alteration in the most common DNA nucleotide sequence. The term <i>variant</i> can be used to describe an alteration that may be benign, pathogenic, or of unknown significance. The term <i>variant</i> is increasingly being used in place of the term <i>mutation</i> . (From National Institutes of Health, National Cancer Institute 2012)
variant of uncertain significance (VUS)	When analysis of a patient's genome identifies a variant, but it is unclear whether that variant is connected to a health condition, the finding is called a variant of uncertain significance. In many cases, these variants are so rare in the population that little information is available about them. Typically, more

	information is required to determine if the variant is disease-related. Such information may include more extensive population data, functional studies, and tracing the variant in other family members who have or do not have the same health condition. Populations that have been historically excluded from genetic research have a greater incidence of VUSs because there is less reference information on which variants are most common in those populations. Also called variant of unknown significance. (Adapted from National Institutes of Health, National Cancer Institute 2012; National Institutes of Health, National Human Genome Research Institute 2022b)
variations in sex characteristics (VSC)	Variations in sex characteristics (VSC) are a group of conditions where development of the reproductive system and secondary sex characteristics does not follow typical male or female trajectories. These differences involve genes, hormones, and reproductive organs, including genitals. The term <i>VSC</i> was initially created to replace differences of sex development because its DSD abbreviation also applied to disorders of sex development. Given the harmful connotations of the original term, the community has started to prefer the term <i>variations in sex characteristics</i> (VSC). See also <i>intersex</i>.
woke	Aware of social problems, such as racism and inequality, as in the phrase “stay woke.” To be woke was to have a leftist perspective on how American society operates, especially with reference to the condition of Black America and the role of systemic racism within it. (McWhorter 2021)

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