



## **Genetic Testing and the Rush to Perfection**

**Part of the Bioethics and  
Disability Series**



**National Council on Disability**

October 23, 2019

National Council on Disability (NCD)  
1331 F Street NW, Suite 850  
Washington, DC 20004

***Genetic Testing and the Rush to Perfection: Part of the Bioethics and Disability Series***

National Council on Disability, October 23, 2019

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## National Council on Disability

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An independent federal agency making recommendations to the President and Congress to enhance the quality of life for all Americans with disabilities and their families.

### Letter of Transmittal

October 23, 2019

The President  
The White House  
Washington, DC 20500

Dear Mr. President:

On behalf of the National Council on Disability (NCD), I am pleased to submit *Genetic Testing and the Rush to Perfection*, part of a five-report series on the intersection of disability and bioethics. This report, and the others in the series, focuses on how the historical and continued devaluation of the lives of people with disabilities by the medical community, legislators, researchers, and even health economists, perpetuates unequal access to medical care, including life-saving care.

Scientific, medical, and technological advances over the past decade have made genetic testing more commonly known and widely accepted among healthcare professionals and the public. Entrepreneurs offer direct-to-consumer genetic testing for individuals seeking knowledge on everything from their ancestry to their chances of developing breast cancer. The potential for discrimination against people with genetic conditions by entities such as employers and health insurers was recognized with the passage of the Genetic Information and Nondiscrimination Act (GINA) of 2008, which provides protections from employer discrimination based on genetic factors. If and how prenatal genetic testing comprises discrimination against people with genetic conditions, however, has been more controversial and has had a much more limited federal and state response.

NCD is concerned that prenatal and adult genetic testing laboratories have exploded in terms of number in the United States, with very little regulation or oversight beyond the Centers for Medicare and Medicaid Services (CMS) ensuring that the laboratories function properly and the tests correctly measure the DNA components claimed. Additionally, healthcare providers involved in prenatal genetic screening and diagnosis have little time to provide in-depth counseling about the tests and the outcome of those tests. Genetic counselors, who would typically stand on the front lines of providing sufficient relevant information to facilitate informed reproductive choices for women, are subject to few requirements when it comes to disability cultural or social awareness and are increasingly being co-opted into the commercial genetic testing industry when they are directly hired by industry.

*Genetic Testing and the Rush to Perfection* examines the impact of genetic testing on people with disabilities and on disability communities, examines the range of scientific, commercial, medical/professional, and social factors that converge around prenatal genetic testing as it affects people with disabilities, and also provides an update on the interaction between genetic testing and employment

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discrimination. It concludes with recommendations aimed at greater federal and state oversight and quality control of genetic tests, and improving genetic counselor education on disability.

NCD stands ready to assist the Administration, Congress, and federal agencies to ensure that people with disabilities do not face discrimination in the area of genetic testing.

Respectfully,

A handwritten signature in black ink that reads "Neil Romano". The signature is written in a cursive style with a large, stylized "N" and "R".

Neil Romano  
Chairman

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(The same letter of transmittal was sent to the President Pro Tempore of the U.S. Senate and the Speaker of the U.S. House of Representatives.)

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## Acknowledgments

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This report is part of a series of reports on bioethics and people with disabilities which was developed through a cooperative agreement with the Disability Rights Education & Defense Fund (DREDF). The National Council on Disability (NCD) appreciates the work of those who contributed their expertise in its development, including Silvia Yee, Senior Staff Attorney, DREDF; Jennifer Mathis, Director of Policy and Legal Advocacy, Judge David L. Bazelon Center for Mental Health Law; and Erin Shea, Policy and Legal Advocacy Fellow, Judge David L. Bazelon Center for Mental Health Law. NCD also appreciates the work of Marilyn Golden, Senior Policy Analyst, DREDF, who shepherded the entire report series in cooperation with NCD. We also thank those who participated on the Advisory Panel, in interviews, and in the stakeholder convening, whose knowledge and willingness to share information helped make this series possible.



# Executive Summary

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## Purpose

This report examines the impact of genetic testing on people with disabilities and on disability communities. Scientific, medical, and technological advances over the past decade have made genetic testing more commonly known and widely accepted among healthcare professionals and the public. Entrepreneurs offer direct-to-consumer genetic testing for individuals seeking knowledge on everything from their ancestry to their chances of developing breast cancer. The potential for discrimination against people with genetic conditions by entities such as employers and health insurers was recognized federally with the passage in 2008 of the Genetic Information and Nondiscrimination Act (GINA), which provides protections from employer discrimination based on genetic factors. If and how prenatal genetic testing comprises discrimination against people with genetic conditions, however, has been more controversial and has had a much more limited federal and state response. This paper examines the range of scientific, commercial, medical/professional, and social factors that converge around prenatal genetic testing as it affects people with disabilities, and also provides an update on the interaction between genetic testing and employment discrimination.

## Background

Approximately 20 years ago, some disability rights advocates who had fought for full and equal access to all aspects of American life focused their attention on how genetic testing, particularly prenatal genetic testing, affected people with disabilities and disability communities. The disability critique of prenatal genetic testing prompted in-depth dialogue among, and writings by, bioethicists, healthcare professionals, people with disabilities, and parents of people with disabilities about the underlying purpose and result of providing information that was explicitly aimed at reducing the number of people who live with a genetic condition. In the years since, scientific and medical advances in prenatal genetic testing, and particularly the discovery of Noninvasive Prenatal Screening (NIPS), have made prenatal genetic testing more commonly available to women of different ages, socioeconomic backgrounds, and health histories than ever before, for an increasing number of genetic conditions. Moreover, rapid advances in prenatal genetic engineering, including inheritable changes in human genetic makeup, have dramatically raised the stakes not only for disability communities but for all of humanity, which must grapple with the implications of

human genetic experimentation for bioethics, the widening gap between those with economic power and those without, the value of diversity, and eugenics.

Genetic testing in general has historically led to barriers and discrimination against individuals who receive positive test results for a genetic condition, even if an individual is asymptomatic and may never develop the condition itself. Employers, particularly those who bankroll health insurance for their employees, can regard employees at risk of acquiring a disability as a financial loss that can best be avoided through dismissal. Such a workplace winnowing,

however, requires access to genetic information about employees.

The degree to which the Americans with Disabilities Act (ADA) and GINA continue to protect applicants and employees against discrimination, at a time when Congress and federal agencies have allowed broad interpretations of workplace wellness programs to undermine

the confidentiality of genetic testing information, is a critical question.

GINA was passed over 10 years ago. What has made it so difficult to address concerns raised by the disability community 20 years ago about the discriminatory implications of prenatal genetic testing for people with disabilities? One key difference is that prenatal genetic testing

provides information about a fetus in utero or an embryo that awaits implantation in utero, and women have a right to reproductive choice and privacy concerning the motives behind their choice. Respecting this right, while recognizing the concerns of disability communities who fear the ongoing dissemination of deeply rooted stereotypes about people who live with disability, eventually led to the passage of the federal Prenatally and Postnatally Diagnosed Conditions Awareness Act (PPDCAA) in the same year as GINA. The PPDCAA requires that healthcare providers and patients receive scientifically current, balanced medical and

social information about living with genetic conditions. A lack of funding sharply reduced the effectiveness of the PPDCAA. Without a funded effective mandate for balanced education about living with a disability, women and families who undergo prenatal genetic testing will continue to be subject to a “perfect storm” of rapid and generally unregulated

technological capacity, genetic testing information that is increasingly disconnected from clinical results or actionable knowledge, and professional standards of care that are subject to pressure by the growing involvement of for-profit commercial testing laboratories and an ongoing shortage of independent genetic counselors.

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*The degree to which the Americans with Disabilities Act (ADA) and Genetic Information and Nondiscrimination Act (GINA) continue to protect applicants and employees against discrimination, at a time when Congress and federal agencies have allowed broad interpretations of workplace wellness programs to undermine the confidentiality of genetic testing information, is a critical question.*

## Key Findings

It is possible to provide a balanced understanding of what it means medically and socially to have a child with a genetic condition, and what it means to live with a disability in the context of prenatal genetic testing. The necessity for doing so arises from the continued existence of deeply rooted social stereotypes and ongoing prejudice about disability, within and without the medical community. This paper's case study provides one example of *how* balanced, current medical *and* social education about disability can be achieved through provider and parent education. Unfortunately, there are no or exceedingly few incentives to promote such a balanced understanding in the current world of prenatal genetic testing. By extension, there are also few incentives to incorporate a disability rights viewpoint or understanding in the dawning world of prenatal genetic engineering, the first inheritable human incident of which made headlines all over the world at the end of 2018 when a scientist in China attempted germline editing on twin girl embryos to create resistance to HIV/AIDS, legally and socially recognized as a disability in the United States. From early manifestations of prenatal genetic testing in conjunction with preimplantation genetic diagnosis (PGD) to the latest NIPS tests, the impacts have been felt within disability communities, and are simultaneously difficult to trace given limited data collection on testing outcomes.

Healthcare providers involved in prenatal genetic screening and diagnosis have little time to provide in-depth counseling about the tests and the outcome of those tests. Genetic counselors, who would typically stand on the front lines of providing sufficient relevant information to facilitate informed reproductive choices for women, are subject to few requirements when it comes to disability cultural or social awareness and are increasingly being potentially co-opted into the commercial genetic testing industry when they are directly hired by the industry. Prenatal and adult genetic testing laboratories have exploded in terms of number and profitability in the United States, with very little regulation or oversight beyond the Centers for Medicare and Medicaid Services (CMS) ensuring that the laboratories function properly and the tests correctly measure the DNA components claimed. Over the last 10 years, industry has achieved technical leaps and bounds, developed sophisticated direct-to-market advertising materials and informational websites, and even coalesced into a lobbying arm that works with states to get Medicaid reimbursement for prenatal genetic testing. Genetic counseling, however, which is becoming more and more necessary as women and their partners receive commensurately greater amounts of unmediated test results, remains unrecognized by CMS as a profession that can independently

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*Unfortunately, there are . . .  
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genetic testing . . . or . . . in  
the dawning world of prenatal  
genetic engineering . . .*

bill, or that state Medicaid agencies will independently cover as “medically necessary.”

The federal PPDCAA is one attempt to ensure that providers and parents receive current information about living with genetic conditions and available supports, but the act is unfunded. Nineteen states have enacted similar legislation, though these vary in sometimes important details. Anti-abortion groups could target these information-oriented laws and propose amendments

that create barriers for termination of a pregnancy after a prenatal diagnosis of Down syndrome or another genetic condition. Most of these acts have no provision for data gathering after their implementation, so it is difficult to know if and how effective they have been since enactment, even if there were baseline data available for comparison.

Unlike prenatal genetic testing, genetic information concerning adults is a more regulated area. The ADA and GINA have been used to address workplace discrimination based on individuals’ genetic information. While GINA has often been dismissed as responding to a nonexistent problem of discrimination based on genetic information, studies have repeatedly shown that such discrimination is not uncommon. The ADA has limited application to this type of

discrimination, as an individual who may develop a genetic condition may not always meet the definition of a person with a disability protected by federal anti-discrimination laws. GINA, which addresses discrimination more directly, has been invoked in a relatively small number of circumstances to challenge adverse action taken by employers on the basis of employees’ or job applicants’ genetic information. GINA has been widely used, however, to address employees’

ability to keep their genetic information private or confidential from their employers. In an era where employers are increasingly seeking to make use of “big data,” those protections ensuring privacy of employees’ genetic information are important.

Indeed, workplace wellness programs have increasingly been used as a way to collect employees’ health and genetic information. The ADA and GINA protect

against the use of financial inducements to pressure employees to disclose such information, but in the past several years, Congress and federal agencies have made efforts to weaken those protections and allow large financial penalties for employees who choose not to provide that information. Those efforts have not been successful to date, leaving at least for now, the protections of the ADA and GINA in place.

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While state laws addressing genetic discrimination in the workplace are typically less protective than GINA, a state law proposal being considered by the California legislature has clear statutory language banning the use of financial inducements for employees to disclose health or genetic information in workplace wellness programs, and it may offer a useful model for states attempting to make their laws as clear as possible while maximizing protection for the confidentiality of genetic and health information in workplace wellness programs.

## Key Recommendations

### *Congress*

- Develop enforceable Sunshine and Conflict-of-Interest laws that will bring transparency to any financial relationships among genetic counselors, providers, and commercial laboratories.
- Fund the Prenatally and Postnatally Diagnosed Conditions Awareness Act, Pub. L. No. 110-374, 122 Stat. 4051 (2008).
- Incentivize the development of educational units on disability experience and exposure in genetic counselor education.

### *Department of Health and Human Services (HHS)*

#### **HHS, National Institutes of Health**

- Establish standing relationships with disability advocacy organizations and include individuals from them on genetic advisory panels.
- Encourage the attendance of advocates and representatives from disability communities at biomedical conferences by

offering scholarships that reduce or cover fees and expenses.

#### **HHS, Food and Drug Administration**

- End enforcement discretion and regulate LDTs, specifically, Noninvasive Prenatal Screening (NIPS), to establish and enforce standards for the accuracy of any claims made by prenatal genetic testing entities, and proactively work with the Federal Trade Commission to oversee marketing by genetic testing entities.

#### **HHS, Centers for Medicare and Medicaid Services**

- Recognize genetic counselors as health professionals who can receive reimbursement through Medicare and incentivize Medicaid payments for genetic counseling as an independent healthcare service rather than only reimbursing genetic testing.

#### **Federal Trade Commission**

- Actively oversee the marketing claims and practices of prenatal genetic testing companies as more tests with questionable clinical validity and utility enter the market as part of the “standard” testing panels that companies offer.

#### **Equal Employment Opportunity Commission**

- Leave wellness rules as they are now (May 2019) or, if EEOC does revise them, the agency should clarify that no financial incentives or penalties are permitted to induce employees to disclose health and genetic information.

## **State Legislatures**

- If genetic testing, and especially NIPS, is funded as a Medicaid service, the state should also ensure Medicaid funding for neutral genetic counseling *before* and after testing takes place.
- Where state Medicaid programs cover prenatal genetic testing, the state should ensure that it collects voluntarily provided information on patient demographics, including disability status, outcomes, and the quality of genetic counseling received before the testing, if any. This information will allow states and researchers to assess the use and results of prenatal genetic testing as a publicly insured service over time.
- Should consider enacting legislation, like that pending in California, that clarifies that no financial inducements are allowed for participating in or providing data to a workplace wellness program.

## **Professional Organizations and Training Accreditation Bodies of Healthcare Providers Engaged in Genetic Counseling such as the Genetics Society of America (GSA); American College of Medical Genetics (ACMG); American Board of Medical Genetics (ABMG); American Board of Genetic Counselors (ABGC); and the Association of Professors of Human and Medical Genetics (APHMG)**

- Clarify that disability education and cultural awareness extends beyond examining best practices for effectively communicating with patients with disabilities and includes a social and civil rights context for understanding disability.

- Ensure that the materials used for provider and patient education are passed through a consensus group of reimbursed stakeholders, including representatives from affected disability communities, to minimize the outsized influence of industry and investors in prenatal genetic testing.
- Professional standards of care for offering NIPS and other prenatal genetic tests should be established through consensus negotiations that include genetic counselors, obstetrics and gynecology care providers, and representatives from affected disability communities. Genetic testing entities should not be allowed to market or provide specific genetic tests that have not been vetted through a professional organization using a consensus process.
- Ensure that online and printed materials used for provider and patient education are fully communication accessible to people with a range of disabilities and diverse linguistic and cultural backgrounds.
- The Accreditation Council for Genetic Counselling (ACGC) must make disability education and cultural awareness mandatory and more consistent among genetic counselor programs, within a reasonable range of time and resources. The same holds true of professional ongoing education.

## **Genetic Testing Researchers**

- Propose ways to achieve better data over time to determine the link between prenatal testing outcomes and various factors in the field of genetic testing such as counseling, cultural conditions, social expectations, and

social determinants of health for particular disability communities.

- Research the relationship between women's choices after receiving pre-test counseling and after undergoing genetic testing, and how choices are affected by the kinds of genetic counseling information provided, who delivers it, and who is paying for the counseling.

## Methodology

The methodology for this paper includes a literature review using multiple academic sources including law reviews, disability studies articles, and science, medical, and social science journals. Nonacademic sources were also reviewed and include articles and analyses published in popular newspapers and magazines, as well as online sources ranging from information provided in official government websites such as the Centers for Disease Control, to blog posts published by nonprofit advocacy entities and marketing materials offered by commercial for-profit entities that offer direct-to-consumer genetic testing. Media and online sources for the public were also reviewed, as prospective parents and adults have increasing access to genetic testing outside of a purely medical context. For example,

commercial laboratories that offer adults services like health reports and ancestry searches from a saliva sample and are paid for out-of-pocket have no need for a healthcare provider's referral and are subject to little regulation. As a result, decisions about when testing takes place, who is tested, the consequences of testing, and what happens with the results are not only influenced by information found in health professionals' journals or peer-reviewed scientific articles, but in mainstream media as well.

Literature research was supplemented by phone interviews of five key stakeholders: a woman with visible disabilities who has undergone prenatal genetic testing, a Masters' Level Genetic Counselor with over a decade of professional experience, a representative of a Down syndrome advocacy organization, the founder of an organization that provides balanced medical and social information on Down syndrome to women receiving prenatal genetic test results, and a woman who has been involved in litigation involving allegations of genetic discrimination in the workplace. The first- and second-hand experiences they shared helped shape the direction of this paper, and particularly the recommendations that accompany it.



## Glossary<sup>1</sup>

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Amniocentesis	Prenatal diagnostic test performed in conjunction with ultrasound, in which the amniotic fluid (the fluid from the sac surrounding the baby) is mechanically withdrawn and analyzed.
Aneuploidy	Any deviation from the typical number of chromosomes, usually meaning a cell nucleus possessing too many or too few chromosomes.
cffDNA	Cell-free fetal DNA, which is used for prenatal testing, and is obtained from a blood draw of a pregnant woman.
CVS or CV	Chorionic villus sampling is a prenatal diagnostic test that examines material from the placenta.
DNA	Deoxyribonucleic acid—An extremely long molecular element that is the primary component of chromosomes, and carries information for the genetic characteristics of life forms.
Germline Engineering	Making changes in germinal (reproductive) cells such that those changes will be passed on to subsequent generations through reproduction.
LDTs	Laboratory-developed tests. A new genetic test usually comes to the market as an LDT where a single laboratory develops and performs the test and client, or provider-gathered specimen samples are sent to that laboratory to be tested.
Microdeletion	A “patch” or group of DNA that is typically present within human chromosomes or genomes, and that is missing from a particular individual’s chromosome or genome.
NGS	Next generation sequencing, which is an umbrella term for a number of new techniques that generate large masses of DNA and therefore allow for much faster sequencing of an individual’s DNA.
NIPS (or NIPT)	Noninvasive prenatal screening (or testing) that analyzes cell-free fetal DNA (cffDNA) obtained from a simple blood draw from a pregnant woman to determine the likelihood that a fetus carries particular genetic conditions.
Polygenic Risk Score	Analyzing multiple regions of DNA at once to derive a fetus or person’s likelihood of having or developing a certain trait or condition.

PPDCAA	Prenatally and Postnatally Diagnosed Conditions Awareness Act, Pub. L. No. 110-374, § 2(1)-(3), 122 Stat. 4051, 4051 (2008). Federal law aimed at ensuring that pregnant women undergoing genetic testing receive up-to-date information about raising children living with Down syndrome and other genetic conditions and evidence-based information about the accuracy of genetic testing.
UCEDD	University Centers of Excellence on Developmental Disabilities, which is part of the Association of University Centers on Disability (AUCD), is administered by the Administration on Intellectual and Developmental Disabilities (AIDD).

## List of Abbreviations

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ACA	Affordable Care Act
ABGC	American Board of Genetic Counselors
ABMG	American Board of Medical Genetics
ACMG	American College of Medical Genetics and Genomics
ACGC	Accreditation Council for Genetic Counselling
ACOG	American College of Obstetricians and Gynecologists
ADA	Americans with Disabilities Act of 1990
APHMG	Association of Professors of Human and Medical Genetics
CAPS	Coalition for Access to Prenatal Screening
cfDNA	Cell-free DNA
cffDNA	Cell-free fetal DNA
CMS	US Centers for Medicare and Medicaid Services
CRISPR	Clustered regularly interspaced short palindromic repeats
CVS or CV	Chorionic villus sampling
DNA	Deoxyribonucleic acid
EEOC	US Equal Employment Opportunity Commission
FDA	US Food and Drug Administration
FTC	US Federal Trade Commission
GINA	Genetic Information and Nondiscrimination Act
GSA	Genetics Society of America
HHS	US Department of Health and Human Services
HNPP	Hereditary neuropathy with liability pressure palsies
IVF	In-vitro fertilization
LDTs	Laboratory-developed tests
NDSC	National Down Syndrome Congress
NGS	Next generation sequencing
NIPS (or NIPT)	Noninvasive prenatal screening (or testing)
NPGD	Noninvasive prenatal genetic diagnosis
OI	Osteogenesis imperfecta
PGD	Preimplantation genetic diagnosis
PPDCAA	Prenatally and Postnatally Diagnosed Conditions Awareness Act
SCA	Sex chromosome aneuploidies
UCEDD	University Centers of Excellence on Developmental Disabilities



For disability rights advocates, the distinction between “perfectly normal” and “broken” is not an objective medical bright line in the sand, but a normative judgment that reflects human prejudice, disability stereotypes, and the social and physical barriers that place arbitrary limits on the length and quality of life of people with many different disabilities.



## Introduction

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With direct-to-consumer marketing of genetic tests for everything from ancestry to personalized medicine, common public views about the use of genetic testing characterize it as a success story that arises naturally and practically from the mapping of the Human Genome Project and researchers' increasing ability to link genetic markers with specific health conditions and diagnoses. According to these views, prenatal genetic testing provides information, more information leads to better prenatal/preventive care and more informed choices, and better care and more choice is always good.<sup>2</sup> Disability, on the other hand, is usually viewed as something that limits the choices of both parents and a prospective child.<sup>3</sup> It is understood as a negative health consequence that awaits amelioration through medical science. This simplistic view of prenatal genetic testing was challenged by bioethicists Parens and Asch almost 20 years ago in a 2-year project involving multiple stakeholders, including people with disabilities, academics, bioethicists, authors, and researchers.<sup>4</sup>

Since that seminal project, there have been many changes in genetic analysis, public and private healthcare coverage, the commercialization and marketing of prenatal genetic tests, disability rights laws, state and federal laws on genetic counseling, and medical

standards of care. The question raised by Parens and Asch, however, remains. What is the impact of genetic testing on the disability community, and on persons with disabilities as parents, as self-advocates, as employees, and as members of society who continue to experience discrimination and real barriers to fully entering the economic, social, and cultural mainstream of American life?

This paper evaluates the state of evidence on the implications of genetic testing for people with disabilities and presents a different view than the popular understanding of genetic testing as a way to avoid the "burden" of disability.<sup>5</sup> It considers recent scientific, commercial, social, and legal developments concerning genetic testing, as administered pre-birth or as used post-birth by employers with respect to job applicants or employees. It will examine the literature on how genetic testing policies and practices impact people with disabilities as both "objects" and users of genetic testing. It will also identify the various stakeholders involved in genetic testing, especially as technological developments such as Noninvasive Prenatal Screening (NIPS) have allowed private for-profit providers to market directly to potential parents without any known risk factors. All stakeholders operate within a complex scheme of professional guidelines, federal laws, and state regulations

that may ignore, consider, or co-opt disability rights concerns. The growing presence of private entities that hold genetic information about fetuses and their parents raises growing concerns about the confidentiality and use of such information. The increasing role of these private entities also warrants scrutiny of how effectively protections limiting the discriminatory use of genetic information are being implemented.

Chapter 2 also examines technologies that have emerged from and are related to genetic testing, but that have additional implications beyond the individual fetus or embryo that is being tested. These new technologies involve genetic engineering and prenatal and in vitro genetic manipulation, including the use of techniques leading to inheritable germline modifications. The expanding potential of genetic modification, and a consistent stream of news about yet another scientist who has broken away to push the envelope on human genetic engineering, has raised a firestorm of public, academic, and ethical comment.<sup>6</sup> Amidst the controversy are voices that recognize the need for an evaluative framework that holds scientific advancement accountable for advancing good, and not merely avoiding the bad. What social, cultural, legal, and political changes are needed to create “public policy regarding reproduction [that] can promote well-being, equality, and diversity”?<sup>7</sup> This is a higher and broader standard for evaluating a community response to new technologies that demands consideration for how traditionally

underserved groups have been left out of the conversation, because of race, disability, gender, sexual orientation, gender identity, or any number of other individual or intersecting characteristics. As one writer put it:

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*The growing presence of private entities that hold genetic information about fetuses and their parents raises growing concerns about the confidentiality and use of such information.*

While we struggle to protect women’s right to self-determination and control over procreation, we must also keep in mind that we are entering a new era of reproductive technology that does not necessarily parallel the needs of

women and families, but rather, follows private investors’ market predictions. Public

### **Foundational Principles**

NCD accepts the following as foundational principles:

- reproduction and child rearing are activities that concern all of society;
- disability is an inherent part of the diversity of life, and people with disabilities must be full and equal partners in society;
- decisions if, when, and how to have and raise children are the intensely private decisions of a woman and her family; and
- individuals and families should have full access to any resources, including medical, social, and practical information about living with disabilities, that they need to make informed decisions.

oversight is the best way to encourage the necessary thoughtful deliberation over these issues, because reproduction is an activity that concerns all of society.<sup>8</sup>

In practice, each of these principles can lead to laws or policies that will be in tension with the others. The current context of genetic testing in the United States features rapid and competitive scientific discovery, corporate profit motives, limited access to independent genetic counseling, and significant regulatory gaps. How can the disability community, as members of society, parents and potential parents, employees and employers effectively move from a position of defense to advocating for genetic justice? Genetic engineering does not necessarily raise the same potential as genetic testing for a direct conflict with reproductive rights advocates, but genetic analysis and manipulation raise a similar clash of divergent interests and opinions within the broader disability community.<sup>9</sup>

There are those within the disability community who argue strongly for urgency in society's use of new genetic technologies. One mother of a child with Duchenne Muscular Dystrophy wanted gene editing to be used like "an eraser on a pencil: correcting genes like correcting misspelled words on a page." She clarified, "We should use this technology to fix the most dangerous mutations that have no cure. . . . I oppose 'designer' gene editing—making changes to perfectly normal genes. . . . Fixing broken genes should take precedence over altering healthy ones."<sup>10</sup> For disability rights advocates, the distinction between "perfectly normal" and "broken" is not an objective medical bright line in the sand, but a normative judgment that reflects human prejudice, disability

stereotypes, and the social and physical barriers that place arbitrary limits on the length and quality of life of people with many different disabilities. Even if there is agreement on which genetic conditions invariably lead to very short life spans, high pain levels, and immediately limited or decreasing functional capacity, there is not agreement on the kinds of policies that can follow.<sup>11</sup>

While individuals and families make reproductive and treatment decisions, over time, professional practice in the application of genetic testing and behavior has led and will lead to the development of medical, legal, and social standards of professional behavior. These standards, in their turn, constrain and shape individual decisions as well as the decisions of other professionals. It is this circular cycle of influence and behavior that makes disability advocates and people with disabilities fearful, because the understanding of disability as a tragedy that requires a medical fix is deeply inscribed in ancient and recent history.<sup>12</sup>

NCD's final recommendations address knowledge, policy, and regulatory gaps that allow private commercial genetic testing entities to have significant influence over who gets prenatal genetic testing, the genetic conditions for which they get tested, and how genetic counseling is or is not delivered pre- and post-test. In the face of private entities' drive to prioritize profits, minimize rising healthcare costs, and emphasize prenatal genetic testing as a "preventive health" measure, disability communities and advocates can best offer their lives and experiences with disability. The recommendations directed at federal and state law and policymakers, genetic testing professions and provider accreditation bodies,

professional regulating bodies, and people with disabilities and disability advocates are intended primarily to “level the playing field” by exposing and limiting potential conflicts of interest among genetic counselors and providers by ensuring that providers and patients get accurate, balanced information about the medical and social functional needs that come when a child has a specific genetically inherited condition. Neither of these desired goals will occur without deliberate, thoughtful, ethical and

regulatory discussions that fully include people with disabilities. The existing “hands off” status quo is not an option in the highly dynamic, profit-driven field of genetic testing. The rapid pace of genetic technology, and the willingness of “rogue” scientists to experiment with human subjects force scientists, academics, and members of the public to confront a foundational question: Do we control genetic technology, or does it control us?<sup>13</sup>

# Chapter 1: Impact of Genetic Testing on Individuals with Disabilities and on Disability Communities

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**E**ric Parens and Adrienne Asch first wrote about genetic testing and its potential impact on the disability community in 1999:

As the ease of testing increases, so does the perception within the medical and broader communities that prenatal testing is a logical extension of good prenatal care: the idea is that prenatal testing helps prospective parents have healthy babies. On the one hand, this perception is quite reasonable. . . . On the other hand, as long as in-utero interventions remain relatively rare, and as long as the number of people seeking prenatal genetic information to prepare for the birth of a child with a disability remains small, prospective parents will use positive prenatal test results primarily as the basis of a decision to abort fetuses that carry mutations associated with disease and/or disability.<sup>14</sup>

The two decades since have shown their words were prescient. In the mid-1990s, Parens and Asch gathered healthcare professionals, disability advocates, advocates, bioethicists, and scientists for a “sustained dialogue”<sup>15</sup> of five 2-day meetings held over a 2-year period. The dialogue allowed this group to closely examine criticisms from a disability rights viewpoint

about how prenatal testing was offered and discussed, how testing results were presented to prospective parents, and how subsequent decisions concerning pregnancy affected people with disabilities as members of society. The group could not manage to agree on the genetic conditions for which testing would be reasonable, but was able to achieve greater consensus on how prenatal tests should ideally be offered and discussed.<sup>16</sup> For NCD, the main concern is the extent to which Parens and Asch’s fears have come true, and the degree to which their recommendations for providing genetic counseling that takes fuller account of the disability viewpoint have been successfully implemented in the specific context of Down syndrome.

## **Prenatal Genetic Analysis and Down Syndrome**

Human beings are typically born with 46 organized “bundles” of genes, called chromosomes. Down syndrome occurs when a fetus has an extra copy of the 21st chromosome, which then affects how the fetus, and the baby after birth, develops and grows.<sup>17</sup> People with Down syndrome may share some known physical characteristics, and typically have some degree of cognitive delay, as well as an increased risk of some medical conditions such as congenital

heart, respiratory, or hearing issues, childhood leukemia, thyroid conditions, and Alzheimer's disease. As treatments have been developed and more children with Down syndrome receive appropriate treatment, the expected life span for people with Down syndrome has increased significantly from 25 years in 1983 to 60 years today.<sup>18</sup> While Down syndrome occurs among all races and ethnicities, research has shown disparities such that the survival rate beyond infancy is lower for black children with Down syndrome than for non-Hispanic white children.<sup>19</sup>

Down syndrome is the most common chromosomal condition, affecting an estimated 1 in every 700 babies born in the United States, or approximately 6,000 babies per year.<sup>20</sup> The potential for having a fetus with Down syndrome increases with maternal age, and pregnant women over 35 years of age routinely undergo at least a NIPS as part of their maternal care. If NIPS shows that a fetus has an increased likelihood of having Down syndrome, then a more conclusive diagnostic test is recommended and can be performed as early as 15 weeks into the pregnancy; earlier use of the test usually carries increased risk of miscarriage or harm to the fetus. Nonetheless, 80 percent of babies with Down syndrome are born to younger women simply because most babies are born to women under 35<sup>21</sup> and testing for Down syndrome is far less routine in this age group. Tests such as amniocentesis have been used for five decades to indicate whether a fetus has Down syndrome,<sup>22</sup> but "[n]either screening nor diagnostic tests can predict the full impact of Down syndrome on a baby; no one can predict this."<sup>23</sup> This is true for many genetic conditions that can have a wide range of expression from mild to severe. Down syndrome advocate groups

also emphasize that however the condition manifests, "[q]uality educational programs, a stimulating home environment, good health care and positive support from family, friends and the community enable people with Down syndrome to lead fulfilling and productive lives."<sup>24</sup>

The prevalence of Down syndrome means that there are more parents of children with Down syndrome than with other health conditions detectable by genetic analysis. More affected families mean more Down syndrome advocacy and support groups to both provide accurate information about what living with Down syndrome looks like and to push for accurate tracking of information about babies born with Down syndrome, in the United States and internationally. The impact over time of genetic analysis, as well as social and economic context, on a specific disability community such as those living with Down syndrome thus becomes a useful stand-in for how genetic analysis affects the lives of people with many different genetically linked health conditions.

### **Accuracy, Prenatal Testing, and Live Births of Babies with Down Syndrome**

Numerous sources, including the Centers for Disease Control and Prevention and advocacy organizations, state that the absolute number of births of babies with Down syndrome in the United States is approximately 6,000 a year. However, the estimate is based on the findings from a study published in 2010 which used 2004–2006 surveillance data gathered in the National Birth Defects Prevention Network, to update a study that originally used 1999–2001 findings.<sup>25</sup> This is important because in 2007, the American College of Obstetricians and

Gynecologists (ACOG) issued Practice Bulletins No. 77 and 88, which recommended that less invasive new screening procedures for Down syndrome, involving nuchal translucency measurement and maternal serum analysis, should be offered to all pregnant women, and not only to women of “advanced maternal age” (35 years of age and older).<sup>26</sup> The question of whether the new Bulletins, which guides the standard of practice of American obstetricians and gynecologists, actually led to identifiable changes in how and when women receive prenatal testing for, and information about, Down syndrome cannot, therefore, be answered by looking at the updated study that used 2004–2006 findings.

Another, more recent study used a variety of datasets to derive the nonselective and live birth prevalence of babies with Down syndrome in the United States from 1900 to 2010.<sup>27</sup> This study places the annual rate of live births of babies with Down syndrome at 5,300 in the years 2006–2010, and an estimated rate of Down-syndrome-related elective pregnancy termination at 30 percent. While this study provided more updated information for the years immediately following the issuance of the ACOG 2007 Practice Bulletins, it did not capture the impact of the newer noninvasive prenatal screens (NIPS), which came on the US market in 2011 and use a simple first trimester blood draw from a pregnant woman to determine the likelihood that a fetus carries a copy of the Down syndrome

chromosome. However, this latest study could be useful in establishing baseline information with which to compare future research into live birth rates and selective abortions for Down syndrome. Such future research is needed to evaluate not only the 2011 commercial availability of NIPS as a key change, but also the influence of additional factors such as further professional practice bulletins issued by ACOG,<sup>28</sup> affiliations between commercial laboratories and genetic counselors, and federal and state laws that require disability-related information in genetic counseling (these last two factors will be

further discussed in the following chapters).

The above discussion illustrates the difficulty of making conclusive statements on how developments in the field of prenatal genetic analysis affect the choices and behavior of pregnant women and outcomes for communities with

specific genetic conditions or disabilities. Data collection, research, and professional guidance tend to lag behind technology, and in some cases, data on specific disabilities may not be available at all.

Nonetheless, there is data on Down syndrome that raises urgent concerns. A 2017 *Healthline* article pronounced that “nearly 100 percent of women in Iceland who receive a positive test for Down syndrome choose to terminate the pregnancy.”<sup>29</sup> From this first statistic, the author concludes that “[d]ue to abortions, only one or two babies with Down syndrome are born each

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year in Iceland.”<sup>30</sup> Pregnancy termination rates of 98 percent for Denmark, 77 percent for France, and 67 percent for the United States are also provided. It is not possible, however, to extrapolate the number of live births of babies with Down syndrome from a statistic about women who receive a diagnostic test for Down syndrome in their fetus. Not every woman who carries a fetus with Down syndrome, and ultimately gives birth to a

baby with Down syndrome, undergoes a prenatal diagnostic test for Down syndrome.

Some women may choose not to have prenatal genetic

screening, such as NIPS, at all. Some woman may agree to a screening, and then decide not to proceed with an actual prenatal genetic test such as amniocentesis or chorionic villus sampling, two common prenatal genetic tests that provide a more conclusive finding of Down syndrome but also carry a greater risk

of harm to the fetus. In the United States, where more women are also giving birth at an advanced maternal age,

and accounting for the rate of miscarriages that statistically occur with Down syndrome, all of these factors together indicate that the number of women who proceed to a live birth of a fetus with Down syndrome has fallen by a number closer to 30 percent rather than 90 percent.<sup>31</sup>

Even among those women who obtain a prenatal diagnosis of Down syndrome in the fetus, a 2012 review of 24 existing studies concluded that the abortion rate was 67 to 82 percent rather than a

previously reported 92 percent, and noted that rates varied between the United States and international studies,<sup>32</sup> as well as regionally within the United States.<sup>33</sup>

Accurate reporting and thinking about the consequences of prenatal genetic testing are important. Their absence gives rise to charges of exaggeration and can lead the public to simply dismiss the legitimate feelings and

eugenic fears of Down syndrome self-advocates as well as their families. Hyperbolic claims are also more likely to fuel different sides of a highly politicized abortion debate, while

failing to acknowledge the individuality of people with Down syndrome, the complexity of their lives, the variety of their circumstances, and how much remains unknown after a prenatal test indicates that a fetus is likely to have Down syndrome. Nonetheless, a

30 percent reduction remains a significant and serious quantitative impact on the Down syndrome community that is bolstered by

qualitative conversations as well. “For me, it’s just faces disappearing,” as one mother of a daughter with Down syndrome put it.<sup>34</sup>

## The Situation of Iceland

In Iceland, parents of children with Down syndrome are deeply discouraged by the reports of a 90 percent abortion rate because it bolsters their own belief about what is happening in providers’ offices. Tamara Pursley, the Programs

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and Partnerships Director at the National Down Syndrome Congress (NDSC) and the parent of a child with Down syndrome, describes NDSC's work translating into Icelandic informational materials and pamphlets on Down syndrome for prospective parents. Pursley describes a small number of people in Iceland who have children with Down syndrome, and says that they do work to promote awareness about the condition, but were not particularly receptive to the translated US materials. They thought the information was accurate and useful. Nonetheless, they thought the resources would be useless since "most doctors don't care" and will just encourage termination of the pregnancy if genetic testing detects Down syndrome. On the other hand, Ms. Pursley reports that adult Down syndrome

self-advocates in Iceland have become popular memes on social media as they try to encourage acceptance of Down syndrome and speak up for themselves.<sup>35</sup> The situation in Iceland also directly raises the additional complicating factor of how the larger culture, generational shifts, and fluctuating social attitudes about genetic analysis itself inevitably affect decisions about having a child with Down syndrome or any other type of intellectual disability.

A small island on the Northern edge of Europe, Iceland's "population remained isolated and quite homogeneous until recently."<sup>36</sup> In 1990, only 4 percent of Iceland's population was foreign-born, but by 2017 the proportion had risen to 11 percent of the country's population of almost 340,000. Furthermore, as recently as the 1980s, 70 percent

of immigration to the island was from other Nordic countries and the United States.<sup>37</sup> With such limited immigration “since the Norsemen first settled here in the 9th Century,” the genetic “purity” of Iceland’s small population makes it “much easier for scientists to isolate faulty genes than it is in larger multi-ethnic countries such as Britain or the United States. Iceland also has a database containing the genealogy of the entire nation dating back 1,100 years.”<sup>38</sup> Against this backdrop, deCODE, Iceland’s leading genetics company, has already gathered over one-third of Icelander’s genetic information in its database, and is aggressively seeking additional voluntary swab samples to double its count.<sup>39</sup> Salvor Nordal, Director of the Ethics Faculty at the University of Iceland, said that the country has never really had a “proper debate” about the uses to which genetic information should be put or whether individuals should be notified of their specific genetic analysis. The lack of a national conversation seems even more necessary given that, even without testing every single person:

[t]hey can fill in the missing gaps,” says Nordal. “DeCODE has collected so much information that we might become the first nation to be genome sequenced. Now it becomes much more than asking questions about an individual’s privacy—we are talking about group privacy . . . and whether we can be discriminated against as a member of that group.”<sup>40</sup>

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*The privacy and discrimination concerns . . . are exacerbated for some by the fact that deCODE is not a charitable, academic, or government enterprise, but a private entity that is owned by Amgen, an American biotech company.*

The privacy and discrimination concerns raised by Nordal are exacerbated for some by the fact that deCODE is not a charitable, academic, or government enterprise, but a private entity that is owned by Amgen, an American biotech company. Dr. Kari Stefansson, deCODE’s director, asserts pure motives: “Keep in mind my only goal—it is not manipulating the human genome but finding out which variant genes . . . are behind the common diseases of man.”<sup>41</sup> Iceland’s lawmakers were apparently convinced of that since deCODE had been granted default access to every Icelander’s health records until a woman sued

the company in 2003 on grounds of privacy, to prevent its acquisition of her deceased father’s records. DeCODE must now obtain individual consent before gaining access to medical records. Getting that permission may not, however, be a problem

for many Icelanders, as genetic testing and handing over the results has already become a part of the culture. One author conducted an informal survey in a university cafeteria and most people indicated that they had already willingly given samples of their DNA to deCODE, even though giving samples is no more mandated in Iceland than in other countries. One man stated that he had “a family member with a genetic condition who has suffered a lot,” asking, “Why should he suffer a lot if we know we have information that can help him? What is it to do with privacy?”<sup>42</sup>

The fact that one-third of Iceland’s population has voluntarily contributed DNA to a private

biotech entity may not be replicable in other countries.<sup>43</sup> Prenatal genetic testing is also *not* the same as genetic analysis performed on adult DNA and raises a different set of ethical, social, and legal implications. Nonetheless, Icelandic cultural attitudes about health and illness, the value of diversity, the value of the individual, and the social impact of discrimination are likely to also underlie how people in other countries understand and expect genetic analysis of all kinds to be used.

Professor Nordal's questions about "group privacy . . . and whether we can be discriminated against as a member of that [genetically identified] group" are the very questions that people with Down syndrome and other genetic conditions have been forced to grapple with in both prenatal and postnatal contexts ever since the initiation of the Human Genome Project. The essence of discrimination is being treated reductively and differently because of a personal characteristic, in this case, a genetic characteristic. There are some who insist that genetic analysis and manipulation is about getting rid of diseases or health conditions, not people, but inevitably those same conditions manifest in living persons. Disability rights advocates have long argued that when genetic testing has the primary goal or even the unintended consequence of discouraging the birth of people with disabilities, there is a negative impact on both those who are already living with disabilities

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*Disability rights advocates have long argued that when genetic testing has the primary goal or even the unintended consequence of discouraging the birth of people with disabilities, there is a negative impact on both those who are already living with disabilities and those who give birth to and raise children with disabilities.*

and those who give birth to and raise children with disabilities.<sup>44</sup> As one mother of a 2-year-old with Down syndrome says, "If all these people terminate babies with Down syndrome, there won't be programs, there won't be acceptance or tolerance. I want opportunities for my son. I don't know if that's right or wrong, but I do."<sup>45</sup> This viewpoint has been characterized as selfish or misguided for apparently "wishing disability" on other babies, or lacking actual evidence that links lower numbers to diminished services.<sup>46</sup> Nonetheless, it is worth noting that a core tenant of disability rights law is the fundamental

importance of people with disabilities entering mainstream life, to see and be seen as fully integrated rights-bearing citizens. An increasingly small and isolated disability community is not only subject to reduced services and supports, it also risks proliferation of false assumptions and stereotypes about

disability that can only be dispelled by genuine interaction between those with and without a wide range of disabilities.

### **The Issue of Abortion: How Decisions Are Made and Who Is Supposed to Help**

Another problem with using a bald statistic about a 90 percent reduction in births is that it tends to lead to a polarizing debate about abortion that, for many disability advocates, is not the point. Pursley emphasizes that the key point is

to communicate the kind of life that someone with Down syndrome can have. She says “[o]ur big position is . . . we’re not pro-life or pro-choice . . . we’re pro-Down syndrome.”<sup>47</sup> And finally, it will be extremely difficult to assess the impact of new genetic technologies, commercial marketing, provider practices, cultural influences, and regulatory action without an accurate baseline of data that reveals where gaps and limitations in knowledge exist. The anecdotes will continue and have their own value, but do not replace data.

The search for accurate information inevitably leads to complicated conclusions. As the new prenatal screening tests are becoming standard practice, it will be important to move beyond the reporting of numbers to a closer examination of the complex factors behind the numbers. One obstetrician posits multiple factors to consider before assuming that the number of births of babies born with Down syndrome may be going up or down. These factors include the racial/ethnic background of women having babies, the extent to which professional guidance on prenatal genetic testing has expanded to more women, and possible changes in the pre-existing inclinations of women who are now being tested.<sup>48</sup>

The most important factor for Down syndrome births in particular may be a “sea change in society’s approach to individuals with Down syndrome.”<sup>49</sup> J. L. Natoli, lead researcher for the paper that found lower rates of abortion for fetuses diagnosed with Down syndrome than first reported, notes that “from a social standpoint, women of childbearing age are from perhaps the first generation who grew up in an era where individuals with Down syndrome were in their schools or daycare centers—perhaps

not the mainstream integration that we see today, but still a level of exposure that was very different than in generations prior. They grew up watching kids with Down syndrome on Sesame Street.”<sup>50</sup> These observations notably posit a direct connection between the reproductive decisions made by women, and observable changes in society that have been wrought by disability rights laws such as the Individuals with Disabilities Education Act and the Americans with Disabilities Act of 1990, which operate to integrate people with disabilities into society and stop the unnecessary institutionalization of people with disabilities. Other original research highlights the multiple demographic and cultural factors that play a role in the prenatal screening and diagnostic testing decisions made by women, showing “findings that older maternal age, greater socioeconomic status, identification with particular races and religions, decreased strength of religious beliefs, and prior experience with genetic testing correspond to willingness to contemplate abortion.”<sup>51</sup> This last study chose to administer surveys to the general public, rather than only women of child-bearing age, recognizing that various members of society may not wish to, or be able to, bear a child, but “still influence prenatal decision-making via complex interpersonal, social, cultural, and political relationships.”<sup>52</sup>

The various themes that have been raised in this close examination of prenatal genetic testing in the context of Down syndrome resound throughout the remainder of this study. The kinds of discriminatory attitudes and stereotypical assumptions encountered by a woman carrying a fetus with an elevated chance of having Down syndrome are shared by women who carry fetuses with other markers of disability. They are

experienced *as well* by women with disabilities who are pregnant or considering having children. Dr. Kara Ayers is the Associate Director of the University of Cincinnati Center for Excellence in Developmental Disabilities and a person with osteogenesis imperfecta (OI). She is also a strong advocate for disability rights and described one of her own genetic counseling experiences:

The most negative interaction I had occurred with a resident in training who briefly saw me at age 22. The appointment was somehow related to the rare genetic type of OI that I have but I don't remember how it was intended to serve me in any way. I took the opportunity to ask questions about my choices around parenting. The counselor drew the tree to explain I'd have a 50/50 chance of passing on OI. I knew enough to know that I needed to add my partner also had OI. She looked confused but drew a different tree explaining our 75% odds of passing on OI. The resident asked how my other family members felt about me considering pregnancy. I'm not sure why but I told him that my mom was opposed to the idea. He responded, "If I were your parents, I'd do whatever it took to get you to consider adoption." I remember thinking that he must have known something about my medical likelihood of a successful pregnancy that he wasn't telling me. It took a few years, but I eventually realized that his statement was entirely based on his attitudes around passing on a genetic condition.<sup>53</sup>

Ayers and her partner currently have two biologically related children, neither of whom

has OI, and one son, adopted at age 7, who has achondroplasia. They undertook prenatal diagnostic testing for their first biological child because "we were very fearful of double dominance," but "also looked at genetic testing as a way to prepare. There would be different types of equipment we'd need with a baby with OI (and parents with OI). We'd need to consider OI in thinking about childcare, plans for travel, and many aspects of getting ready for the baby . . . we generally expected to have children with OI. For our first, however, we were just more nervous and wanted to know."<sup>54</sup> Ayers says that "I didn't experience any pressure, but I also was very proactive in expressing that our family takes pride in disability. We had no plans to terminate on the basis of disability."<sup>55</sup> When asked about useful advice given by the genetic counselor after the ambivalent test results from her chorionic villus sampling (CVS) test, Ayers says "I didn't receive any advice other than some general support that these decisions are difficult. I really looked to my friends who are also parents with OI and other genetic conditions. I don't feel like professionals who aren't disabled really have perspective on the complexities when you identify as disabled and could also be having a child with that disability—especially when it involves some painful aspects."<sup>56</sup>

Overall, Ayers had a positive experience with prenatal genetic testing, but that is likely attributable in part to the fact that she has a visible genetic disability herself and is a strong self-advocate. She acknowledges that she feels "very conflicted about genetic counseling because I know it has a mixed history with our disability community. At the same time, I saw exploring these options as my responsibility as a

parent, especially if it could improve the outcomes, our response, etc. to our child.”<sup>57</sup> Ayers insightfully notes how her own use and experience of prenatal genetic counseling, as a woman with a disability who did not plan to terminate her pregnancy, likely differed from women who don’t have disabilities or the genetic condition for which a fetus carries the marker:

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*“When I think about this experience from the perspective of someone learning their child has my condition, but they’ve never heard of it, I’m reminded of the major missing piece: any connection to actual people with the diagnosis.”*

When I think about this experience from the perspective of someone learning their child has my condition, but they’ve

never heard of it, I’m reminded of the major missing piece: any connection to actual people with the diagnosis. The social aspects of functioning—how do people work with OI? Live in their community? Go to school? Have families? What do they think of their quality of life? There should be more referrals to other people in the community—which to me, means a greater need for community-based support.<sup>58</sup>

## Chapter 2: The Influence of Technological Advances

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### Noninvasive Prenatal Screening— The Game Changer

Chapter 1 looked at multiple factors that influence the impact of genetic technology on people with disabilities, and the complexity of how these factors interact, using data and analysis on the lives of people with Down syndrome and their families. Unfortunately, most of this data does not measure the specific impact of noninvasive prenatal screening (NIPS) on the birthrates of people with Down syndrome or any other genetic condition. Data on birth rates of people with disabilities is not made available in real time. Instead this section will focus on qualitative research looking at the attitudes, experiences, and decisions of women who undergo NIPS and prenatal genetic tests, and the providers who give varying amounts of genetic counseling pre-test, post-test, or at both time periods.

It would be hard to overstate the degree to which NIPS has altered how genetic testing and analysis is done. For women, the ease of a virtually risk-free blood draw from the mother is infinitely easier to undergo, physically and psychologically, than an invasive procedure that extracts genetic materials from the womb and places the fetus at risk. Furthermore, NIPS can be performed early in a pregnancy, at 9 weeks,

giving women options ranging from planning fun “gender reveal” parties to terminating a pregnancy before anyone else even knows about it. This compares to 15 weeks, which is the earliest point at which an amniocentesis can be performed with less risk to a fetus, at which point a woman is already well into the second trimester of pregnancy. As an essentially unregulated product, women who are pregnant or hope to be pregnant learn about NIPS not only when their healthcare provider raises the topic of genetic analysis, but when they search for information online. Commercial laboratories have developed a plethora of direct-to-consumer marketing that is often focused on *all* women, not just women who have risk factors for having a fetus with a chromosomal condition. NIPS laboratories frequently offer one tab for healthcare professionals and another for expectant parents.<sup>59</sup> The professional tab leads to more technical information and urges providers to use a particular laboratory, while the parent tab urges women to discuss NIPS with their providers. Even though NIPS does not provide the certainty of genetic diagnostic tests such as amniocentesis or chorionic villus sampling, it has established a high rate of accuracy and few false positives when used to assess the chances of a fetus having Down syndrome.<sup>60</sup>

As one of the first companies to develop the ground-breaking NIPS technology, Sequenom was initially focused on a blood test that would achieve the results of amniocentesis without the risks. That is, the company primarily sought a simple and effective way to evaluate the presence of the chromosomal characteristics for Down syndrome, but it also recognized that the technology could lead to testing for other “chromosomal abnormalities.”<sup>61</sup> Sequenom was the highest performing California stock in 2007 and 2008, with a meteoric increase in stock price from \$4.70 on opening day 2007 to \$22.76 by September 2008.<sup>62</sup> Given the very significant potential for profit, as well as the kind of renown that would come with developing a new diagnostic biotech tool that appeals to prospective parents, Sequenom and its rival companies have quickly moved to expand the possible conditions that can be captured by NIPS.

The discovery in 2012 of the capacity to “map” a fetus’s entire genomic makeup rather than just conduct a targeted search for genetic markers associated with specific chromosomes or DNA has opened up additional realms of scientific and financial potential, as well as ethical and practical dilemmas. As one science reporter observed in 2014:

. . . whole genome sequencing could provide parents with an avalanche of

unexpected and perhaps confusing data. Instead of targeted tests for a few dozen genes, future sequencing techniques could provide parents with three billion base pairs of data. The key issues inherent in any genome sequence work would plague fetal sequencing as well—namely, there is no guarantee that genetic mutations will actually result in a specific disease. And grappling with information suggesting that certain conditions may emerge in adulthood, or studying mutations with unclear significance, could be fraught with risks

and challenges—impacting parents’ decision threshold for deciding to terminate a pregnancy or influencing how they rear their child.<sup>63</sup>

Fast forward to 2019, and we have gained additional scientific and technological advances but are seemingly no

closer to resolving the larger ethical dilemmas that were raised years ago or figuring out a way to clearly communicate those dilemmas to prospective parents and society at large. So many different questions are raised, from those that center on what the tests actually provide, to concerns about the social justice and legal ramifications depending on how tests are distributed and how much they cost, to what happens to the genetic information derived from the tests, and whether and how tests should be incorporated into the

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*Advances in prenatal genetic testing keep raising the same fundamental question: How does more and more fetal genetic information, and particularly information that is not complete, fully accurate, or consistently predictive, actually help women make informed choices . . . ?*



medical standard of care. As another reporter stated:

Makers of those tests, though, are already pushing the technology beyond its recommended uses to flag a rapidly expanding list of the unborn's potential genetic flaws. But these bigger and bigger menus of genetic testing also come with less and less information about how predictive the data they reveal actually is. . . . As companies amass these valuable stockpiles of prenatal DNA, should there be limits to how much data they can report back to prospective parents? And what about people without the means to join in the genetic data sprint to the womb?<sup>64</sup>

If the two passages above, separated by over 5 years, sound similar, it's because they are similar. Advances in prenatal genetic testing keep raising the same fundamental question: How does more and more fetal genetic information, and particularly information that is not complete, fully accurate, or consistently predictive, actually help women make informed choices about continuing or terminating a pregnancy? This, in turn, leads to the fundamental question that prenatal genetic testing raises for many in the disability community: How do we make room amidst the flood of genetic information to share knowledge on the lived experience of disability and raising a child with disabilities? Many women and their partners, including the middle- to upper-class

white women of child-bearing age who primarily populate the ranks of genetic counselors over the past couple of decades,<sup>65</sup> have little personal experience with disability. How do pregnant women who are on the receiving end of increasing options for prenatal testing feel about the information they receive from that testing, who advises them on what their various results and "risk factors" mean, and how do they evaluate whether to be tested or which tests to take in the first place? And one final question that all of society must collectively consider: Does our ongoing social, ethical, and legal failure to fully grapple with and resolve the

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*How do we make room amidst the flood of genetic information to share knowledge on the lived experience of disability and raising a child with disabilities?*

hard questions raised by technical advances in genetic testing and manipulation place individual women under the crushing burden of being our default decision makers, even as they are given fewer

opportunities for thorough conflict-free counsel on increasingly complex and unknown genetic options? An increasing body of qualitative research on women who are receiving or who have received genetic counseling suggests an affirmative response to this last question. The issue of how "changing nothing" places a growing burden on women is a theme in this study and will continue to be raised in different contexts in subsequent chapters.

One survey of pregnant women relied on over 550 survey responses received over a 3-month period in 2017. The women were all patients at a large maternity hospital. They were asked to consider undergoing whole genome sequencing and discussing their preferences and reasons for

receiving different categories of genomic results. In their surveys:

- Respondents were most likely to want information regarding serious treatable childhood-onset conditions (89.7 percent) and least likely to want to receive information about nonmedical traits from prenatal whole genome sequencing (40 percent).
- The reason given most often for wanting medical prenatal whole genome sequencing results was “to prepare financially, medically, or psychologically for a child with special needs.”
- 10.5 percent of respondents overall wanted clear recommendations from clinicians about the categories of information that are most appropriate to test for, 44.7 percent wanted clear recommendations plus all options presented, 26.2 percent wanted all options presented and joint decision making, and 13.2 percent wanted all options presented and independent decision making.<sup>66</sup>

Another study, noting that the very low rate of false positives achieved when cell-free DNA (cfDNA) is used for Down syndrome screening, is *not* replicated when cfDNA screening is used for a range of other chromosomal and microdeletion syndromes, interviewed 40 recently pregnant women who received cfDNA NIPS results indicating a high risk of a chromosomal condition, a false positive, or inconclusive results.<sup>67</sup>

One-third of the participants would not choose to have cfDNA screening for additional pregnancies, and another one-third would only choose screening under particular circumstances or if they could limit the scope of the panel of tests. The women reported experiencing problems with the clinical interaction when screening is first offered, when results are reported, and when information is sought about false positives. Ultimately, many of the women interviewed “reported feeling misled by the information they received prior to accepting cfDNA screening or receiving their results. . . . These reports suggest that inadequate pretest discussion contributes to women’s experience of decisional regret

after receiving high risk, false positive, or inconclusive results.”<sup>68</sup> Reproductive decision making is always difficult and any woman may experience regret even after making a decision that she feels was the

right one. But emotions experienced because of a pregnancy or terminating a pregnancy *is not the same* as regret that arises specifically out of prenatal genetic testing where a woman may feel she didn’t understand the test, didn’t have help interpreting the results, or wishes she had not acted on the results.

## Regulation of NIPS

NIPS is not regulated by the US Food and Drug Administration (FDA). That is due to a matter of historical treatment of laboratory-developed tests (LDTs), not an issue of lack of jurisdiction. Currently, NIPS is part of the larger category of LDTs. In general, LDTs are developed

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and patented by a single laboratory, which is then also the entity that builds a market for the product and applies the test to samples that are sent to it. The FDA has the authority to regulate LDTs, but for the most part has historically exercised “enforcement discretion,” and has not generally applied premarket or other regulatory requirements to LDTs.<sup>69</sup> The Centers for Medicare and Medicaid Services (CMS) oversee laboratories and LDTs under the Clinical Laboratory Improvement Amendments program.<sup>70</sup>

There is a crucial distinction between *analytical validity* and *clinical validity*. Analytical validity questions whether a test consistently and accurately measures what it is supposed to detect. Clinical validity questions whether the measured change results in an increased risk or likelihood of having an identifiable medical condition.<sup>71</sup> Clinical

validity could be considered the application of an abstract biological relationship to real life. CMS only looks at analytical validity, and only in the context of a single laboratory that is already in business.<sup>72</sup> When CMS conducts its routine biennial surveys of Sequenom, Illumina/Verinata, Natera, Ariosa, or any other prenatal screening laboratories that have joined these “first four” laboratories, CMS determines that one particular laboratory conducts its LDT in safe conditions that enable the laboratory to reliably state that it found and measured chromosomes present in what is usually cell-free placental DNA.<sup>73</sup> By contrast, the FDA conducts premarket clearance and approval procedures that examine both

analytical validity and clinical validity. That is, the FDA would determine whether a NIPS accurately measures what it is supposed to, going into greater scope and depth, and whether that measurement reliably and consistently conveys the likelihood of a fetus having a specific genetic condition.<sup>74</sup>

The FDA’s enforcement discretion regarding LDTs means that there is no federal oversight or consequences when a company like Invitae claims on its website that genetic testing “helps make healthy pregnancies possible.”<sup>75</sup> Genetic testing is not a maternity vitamin. There is no direct scientific or statistically established causative link between genetic

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***Genetic testing is not a maternity vitamin. There is no direct scientific or statistically established causative link between genetic testing and healthy pregnancies.***

testing and healthy pregnancies. When a company asserts that “carrier screening is recommended for all individuals who are pregnant or planning a pregnancy”<sup>76</sup> it not only neglects to say who is

doing the recommending,<sup>77</sup> it also implies to consumers that a carrier screen will not merely identify chromosomes and microdeletions in one’s DNA but can positively identify the health outcomes that will arise out of that genetic inheritance. The FDA’s current enforcement scheme allows such a claim to stand, although it comes close to, or crosses the line, between analytical validity and clinical validity.

The status quo of not actively regulating due to enforcement discretion was potentially going to change several years ago after the House Energy and Commerce Committee’s Subcommittee on Health held a hearing on September 9, 2014, into the FDA’s proposed plan

to change its policy on regulating LDTs. However, new regulation of LDTs, including prenatal genetic tests, “has yet to take place due to the complexity of the task at hand,” according to the National Human Genome Research Institute’s website.<sup>78</sup>

The FDA has identified problems with a number of LDTs, including claims that are not adequately supported by evidence, lack of appropriate controls yielding erroneous results, and in a few cases, falsification of data.<sup>79</sup> These problems demonstrated a need for greater FDA oversight to assure both analytical and clinical validity of LDTs relied on by physicians and patients.<sup>80</sup> In response,

the FDA drafted guidance that addressed how it intended to regulate LDTs and verify their analytical and clinical validity, but unfortunately, it was not issued. The FDA informed NCD that it has been very active in this area and continues

to engage with Congress to find a legislative solution, but its decision not to issue guidance or regulations on the rapidly expanding NIPSS market, and other LDTs, is troubling, particularly in light of unified industry actions that push for changes such as greater public and private insurance coverage of LDTs (see next section).

The only other federal agency that has what is currently a peripheral role in regulating NIPS is the Federal Trade Commission (FTC). According to the FTC, the agency has general jurisdiction “to prevent fraudulent, deceptive, and unfair business practices in the marketplace and to provide information to help consumers

spot, stop, and avoid them.”<sup>81</sup> When it comes to genetic testing, the FTC primarily warns against the use of “direct-to-consumer” tests that consumers order and pay for entirely on their own, rather than prenatal genetic tests that generally require referral through a healthcare provider. Nonetheless, their web page has consumer advice that applies equally to prenatal genetic tests, such as “[a]sk your doctor or a genetic counselor to help you understand your test results” and “[g]enetic test results can be complex and have serious implications. It’s a bad idea to make any decisions based on incomplete, inaccurate, or misunderstood

information.”<sup>82</sup> The FTC does not appear to have directly considered prenatal genetic testing companies. The direct-to-consumer marketing undertaken by these companies means that the FTC’s expertise and watchdog functions are needed more than ever,

as the prenatal genetic testing field becomes increasingly saturated and competitive.

### **The Booming Business of Babies and Marketing NIPS**

NIPS is a product developed and sold by commercial laboratories. Providers may approach prenatal genetic screening as a healthcare issue, people with disabilities may view it as a community and ethics issue, women’s rights groups may understand it as a privacy rights issue, and lawmakers and lawyers may see it as a policy and regulatory issue. From a business point of view, NIPS is about investment, market

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share, competition, sales strategy, and lobbying. In 2012, a year after the first test hit the market, four companies offered the service. Today there are more than 40, and global revenues for the industry are expected to exceed \$2.5 billion by 2025. Earlier this month, at an international meeting of reproductive scientists in Paris, the inventor of NIPT, Dennis Lo, told the audience that 6 million women from 90 countries have so far been screened. Some experts say it is the most rapidly adopted test ever.”<sup>83</sup> Professor Henry Greely, the Deane F. and Kate Edelman Johnson Professor of Law at Stanford Law School, predicted in early 2011 that if the uptake of NIPS among women nationally reflected the use of NIPS among women in California, which offers a robust statewide prenatal screening and testing program that covers various modes of testing, the number of fetuses tested through NIPS would increase from less than 10,000 to 3 million.<sup>84</sup> Such a startling increase “would produce a heretofore unimaginable wave of women struggling to understand their tests, the implications of a positive result, and how to proceed.”<sup>85</sup> Professor Eric Topol at the Scripps Research Institute estimated that 800,000 women in the United States had a NIPS in 2014, saying, “That’s a lot—about 20% of the 4 million total babies born each year.”<sup>86</sup> Three million, nonetheless, pales in comparison to the potential market of all women of childbearing age, or at least, the estimated number of women who

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would consider having a child. A report from the US Census Bureau found that as of June 2012, “75.4 million women in the United States were aged 15 to 50, and 59 percent of them were mothers.”<sup>87</sup>

As commercial entities seek to build a larger market for NIPS, marketing materials and websites operated by NIPS laboratories have become increasingly sophisticated in their presentation. As noted by author and poet, Georges Estreich, there is both a soft focus and a presumptive underpin when it comes to disability. He wrote:

To convince people to adopt the test, you have to accomplish a number of things. You have to downplay any risk associated with the test itself. You have to establish the test as a “scientific” thing to do—hence the recommendations from professional organizations (some of which actually caution against misuse and overuse of the test, though the companies tend to ignore those passages), plus all the numbers, graphs, and general science-y feel of the websites. And you have to highlight the risk of not using the product. You establish a happy world on the product side, and a sad, anxious world on the not-product side, and then it’s clear where the consumer should go. . . . Expanding a market is both a persuasive and an interpretive act. It involves a delicate

balance between stigma and acceptance. If the condition is too stigmatized, people won't willingly admit they have it; conversely, if it's accepted as a normal part of being human, then people won't buy treatments for it.<sup>88</sup>

In an earlier piece, Estreich, who has a daughter with Down syndrome, closely analyzed the websites of some of the major companies that provide NIPS, and found numerous implicit messages about the lives of people with disabilities:

It's not that I think these tests shouldn't exist, or that women shouldn't have access to them—or the abortion they might lead to. It's more that in the process of selling the tests, the companies distort or obscure the lives of those with the conditions tested for. As a result, the values they project are at odds with the values I, like many people across the political spectrum, have come to hold: that, for example, true peace of mind comes from accepting a child for who she is. That we should lower the wall between "abnormal" and "normal," not raise and reinforce it. That disability should be represented accurately and fully, that we shouldn't take the disability for the person, and that we should listen to the way persons with disabilities represent themselves.<sup>89</sup>

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*Six US genetic testing companies have joined together to form the Coalition for Access to Prenatal Screening (CAPS), a lobbying organization that "seeks to improve access to state-of-the-art prenatal screening . . ."*

Researchers of online marketing have also noted the unique fact that, "unlike the case for other prenatal genetic tests, industry has had an important role in the development and introduction of NIPT."<sup>90</sup> With both patients and healthcare providers increasingly using the Internet as a research and resource tool, it is significant that popular Internet search engines all include the commercial NIPS websites in the first three pages when a search is conducted for "non-invasive prenatal testing."<sup>91</sup> After members of this research team independently evaluated and coded each commercial laboratory's website, the team concluded that

there was a real need for "clear, consistent, and evidence-based materials to educate patients and healthcare providers about the current and emerging applications of NIPT."<sup>92</sup> In particular, most websites were inconsistent

in providing accurate information about sex chromosome aneuploidies (SCA) and chromosomal microdeletions. This is worrying in the context of SCA because most people have less knowledge of SCA, and healthcare providers are less aware of the physical and cognitive characteristics associated with SCA, which may prompt them to put greater reliance on the commercial website information. For microdeletions, the researchers noted that at the time of investigation:

. . . clinical practice guidelines do not recommend the general use of NIPT for the

detection of microdeletions. Yet, three of the laboratories presented information on their websites about their specific platform's ability to detect certain microdeletions. The differences among the laboratories' microdeletion offerings and educational resources may be a reflection of market incentives for commercial NIPT laboratories to provide large and unique testing platforms before competing laboratories do (references omitted).<sup>93</sup>

In addition to inconsistencies with regard to the substantive scientific information presented, the research team found inconsistent references to such core legal concepts as informed consent. Only three of the five commercial laboratories offered information about informed consent, and only one of the three offered information to *both* providers and patients. Of the remaining two, one laboratory offered the information to patients only, while the other offered information to providers only. Unfortunately, there was consistency in at least one aspect: "None of the website content provided by the commercial laboratories addressed personal values and ethical considerations as they relate to the decision-making process for NIPT."<sup>94</sup>

While marketing is an area where the growing number of commercial genetic testing companies are in competition, as each company attempts to promote the predictive strength

of its own testing platform, there is one area where at least some commercial laboratories are in close cooperation. Six US genetic testing companies have joined together to form the Coalition for Access to Prenatal Screening (CAPS), a lobbying organization that "seeks to improve access to state-of-the-art prenatal screening using cfDNA-based NIPS that is easily accessible to all pregnant women who choose to pursue aneuploidy screening, regardless of their risk factors, income, age or geographic location."<sup>95</sup> CAPS is actively working on outreach to lawmakers to achieve legislative changes and encourage reimbursement coverage of

NIPS. For example, on March 18, 2019, CAPS held a policy briefing at the Washington State Capitol, sponsored by the Women in Government Foundation, which, for \$12,000, will help recruit state lawmakers and staff to attend a meal and panel presentation offered by industry

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*CAPS' presentation was titled "Access and Disparity Challenges for Non-Invasive Pre-Natal Screening." The invocation of healthcare access disparities in the presentation title implicitly places NIPS among medically necessary treatments and services.*

or other groups on a specific policy topic at a state capital.<sup>96</sup> CAPS' presentation was titled "Access and Disparity Challenges for Non-Invasive Pre-Natal Screening." The invocation of healthcare access disparities in the presentation title implicitly places NIPS among medically necessary treatments and services. It would be difficult to even raise questions about NIPS clinical validity, federal and state oversight, informed choice, ethical considerations, or the absence of information about the real lives of people with disabilities when the framing is neatly focused on how some women are unfairly

excluded from using NIPS. In the Washington briefing, CAPS sought support for coverage of NIPS in Washington's state Medicaid program.

Stephanie Meredith, the founder of Lettercase and the mother of a child with Down syndrome,<sup>97</sup> vividly recounted a similar industry "framing" experience when she attended a prenatal molecular diagnostic conference in September 2018. Meredith was at the conference to help commercial laboratories understand how they could help balance medical information on their websites by including resources that show what it is like to live with various genetic conditions and disabilities. She attended a session where one panelist, an investor in NIPS technology, said "It was unfortunate that

invasive chromosome microarray testing was not expanded to the entire population right away, because at "three thousand a test, all four

million pregnancies would cost \$12 billion, but even with a 50% termination rate, would save \$19 billion in estimated medical costs . . . and so many other costs families bear." [Meredith's paraphrase.] After the presentation, one woman, an obstetrician-gynecologist, spoke up to the panel of four men, saying that they could not just assume these kinds of savings because you can't just assume that women would undergo invasive testing and terminate pregnancies. The response from the investor, according to Meredith, was agreement that he couldn't and wouldn't make women do anything, but the smart thing for them to do would be to choose invasive testing, and he estimated half would terminate. Meredith found this exchange memorable for both the woman's courage in speaking up and the panelist's

dismissal of her comment as irrational, as evidenced by the fact that it contradicted economic sense, including his own personal financial interest. Meredith was also deeply stung by the power this investor wielded, almost as a casual afterthought, over the lives of people with disabilities when he likely had little or no personal knowledge of the lived experience of disability.<sup>98</sup>

Power is another emerging theme that permeates this issue and one that is central to the revolutionary role of NIPS as a catalyst in the field of prenatal testing. Industry has driven the demand for noninvasive genetic testing, and shaped much of the narrative about the product. Commercial laboratories wield economic power

and lobbying power, and with those come outsized influence over legislators and the professional bodies that update medical standards. The disability

community has the power of narratives and lived experience, but all too often lacks sufficient support to broadcast its narratives and knowledge to policymakers, medical professionals, and most importantly, the women who are ultimately responsible for making informed choices. A review of NIPS led by Dr. Christina Lockwood of the University of Washington in Seattle concluded that "NIPS is only a screening test. This means that, while a negative result can rule out the need for further testing, a positive result should not be acted on until confirmed with invasive testing."

Lockwood's group recommends that all patients undergoing NIPS receive pre-test and post-test counseling to ensure they understand this, as well as to help with overall interpretation of

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*Industry has driven the demand for noninvasive genetic testing, and shaped much of the narrative about the product.*



complicated NIPS results.<sup>99</sup> Genetic counseling is recognized as a critical step for women considering or undergoing prenatal genetic testing, but in practice, and for various reasons, genetic counseling and the presence of genetic counselors cannot be considered a kind of “magic bullet” that, through their mere presence, will enable women to make fully informed and ethical decisions for themselves.

## Genetic Counselors—A Profession Caught in the Middle

In the 2-year prenatal genetic testing project initiated by Parens and Asch, the only full consensus recommendations centered on genetic testing and the key role of genetic counselors.<sup>100</sup> The group recommended both disability training and education for genetic counselors, and disability-related education content for genetic counseling.<sup>101</sup> In the two decades that have passed, there has been some action on those recommendations but not a broad national move toward, or even conversation about, how to consistently implement disability training and education for genetic counselors, or ensure that users of prenatal genetic testing receive clear, reliable information about disability. Worse yet, industry changes, and especially the market incentives brought about by NIPS, now raise additional issues of potential conflicts of interest and lack of transparency for genetic counselors as they are increasingly directly employed by the

very commercial entities that succeed by selling genetic testing.<sup>102</sup>

According to the US Bureau of Labor Statistics' *Occupational Outlook Handbook*, there were 3,100 jobs for genetic counselors in 2016, and employment “is projected to grow 29 percent from 2016 to 2026, much faster than the average for all occupations.”<sup>103</sup> Genetic counselors can work generally or with narrower populations in particular topic areas, such as assisted reproductive technology, prenatal testing, pediatric testing, or specific adult areas such as oncology, cardiovascular, or neurology. The supply of certified genetic counselors is

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expected to increase 72 percent, to 6,562 between 2017 and 2026, but this demand projection was limited “to only those certified genetic counselors in direct patient care, as this group comprises approximately 65 percent of the overall certified genetic counselor

population,” even though industry makes up the next largest group of employers.<sup>104</sup> At a rate of one certified genetic counselor per 100,000 persons, supply will meet demand by 2026, but a more generous model of care of one counselor per 75,000 would push satisfaction of demand out to 2029 or 2030.<sup>105</sup> The focus on meeting the demands of direct patient care does not appear to consider how biotechnology can influence both patient demand for genetic counseling as well as the degree to which industry could assume a greater proportional role as an employer of genetic counselors. In other words, “neither of

[the] demand models account for largescale exogenous factors, such as the introduction of blockbuster tests and changes in commercial/public reimbursement,” even though “[t]here are approximately 60,482 genetic testing products on the market and an average of 8 to 10 new products enters the market every day.”<sup>106</sup> Of course, not all of the new products are prenatal genetic testing products, but prenatal genetic testing is integrally part of a rapidly expanding industrial field.

Katie Stoll, a certified genetic counselor with 15 years of experience in prenatal, pediatric, and adult genetics, currently works in Washington State and has written on the conflict of interest issues that arise for genetic counselors who work with industry. Stoll’s concerns rise from what she sees as a core conflict between the profession’s historical roots in the 1950s and 1960s, a time when the public was increasingly rejecting explicit eugenics laws and beliefs in the United States while also gaining awareness and support of women’s reproductive autonomy,<sup>107</sup> and the thriving job market for genetic counselors offered by an industry that attracts capital and shareholders through increasing the use of genetic testing. If a genetic counselor’s highest professional standard is to protect women’s autonomy and informed decision making, as well as the unique choices that families make for themselves, then how can this purpose be completely unaffected when the genetic

counselor is directly employed by a for-profit entity that markets its patented tests as part of a medical “gold standard” of care and reminds providers that they can be liable as a matter of medical negligence<sup>108</sup> for failing to discover or tell a patient about discoverable health conditions? As Stoll and others have written:

Uncertain information from many complex genetic tests can find some patients unprepared, so that results are devastating rather than empowering. Labs touting their “simple” and “trusted” tests

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*If a genetic counselor’s highest professional standard is to protect women’s autonomy and informed decision making, . . . then how can this purpose be completely unaffected when the genetic counselor is directly employed by a for-profit entity that markets its patented tests as part of a medical “gold standard” of care . . . ?*

encourage obstetrical care providers to recommend genetic testing to their patients by promising that genetic counseling is included. What may be lost in connecting with genetic counselors through these polished, pro-testing websites, however, is nuanced, informed

decision-making about whether to undergo a particular test in the first place.<sup>109</sup>

Stoll has seen the industry appear increasingly at professional conferences and commercial laboratories and place more and more job postings. She says “we are losing independently employed genetic counselors very quickly, and genetic counselors who work for the companies have a job of selling the tests.”<sup>110</sup> As Stoll notes, “there is a big difference between 45 to 60 minutes of

counseling to unearth how a woman or couple feels about testing and the results that can come from it, and 5 to 10 minutes of counseling after a test has been taken and unexpected results arise.” Stoll also notes how new genetic counseling graduates, mostly young women, may take these jobs without acquiring wider or more diverse genetic counseling experience first and simultaneously having little life experience of people living with the genetic conditions they studied in textbooks, or much understanding of disability rights or disability objections to the reflexive use of prenatal genetic testing to deselect fetuses with disabilities.

Natera, one of the first companies offering NIPS, has multiple videos on its website aimed at potential parents. Upon entering the patient services portal, the first menu tab offers “resources,” which comprise several videos under the heading “Learn from our team of genetic counselors.”<sup>111</sup> Most of the six videos are less than a minute, with “Should I have prenatal genetic testing” the longest at 1 minute, 13 seconds. In the video, a personable young woman paints a picture of how women fall into three groups when it comes to prenatal genetic testing: one group that chooses not to know anything and refuses all tests, and one group that undergoes invasive diagnostic tests with the risk of miscarriage because they must know everything for certain. The third group, which is “the vast majority of women” according to the speaker, choose NIPS because this test “can provide information about specific conditions in the pregnancy, but does not involve taking a risk of miscarriage for the pregnancy.”<sup>112</sup> This juxtaposition of most women who choose NIPS and those few who choose not to test invites the viewer to identify with the sensible majority.

The video is as notable for what is not said as for what is said. The young women all look confidently into the camera and speak in terms of choice, benefit, and information, but nothing is said about how a patient is supposed to choose what to test the fetus for, what the test results could show, and the further likelihood of invasive testing should the NIPS results indicate a chance that a fetus carries genetic conditions. There is no mention of how individual values, fears, and family expectations will all likely come into play because decisions about what to do after a NIPS screening are not dictated purely by science and medicine. All is simple.

The 37-second “How can I find out the gender of my baby?” video describes “a simple blood test” at 9 weeks gestation that “will give you a risk assessment for chromosome abnormalities and also will tell you [here the young woman’s voice slows, she breaks into a smile and her eyes twinkle] whether you are having a boy or a girl.”<sup>113</sup> Natera offers appointment slots if a parent has further questions, but this is exactly when a genetic counselor who is also a good Natera employee could, without any malice or conscious intent, steer a potential patient’s questions toward what a test means, how it’s done, and how it works, rather than questions about whether a particular women with no risk factors should be undertaking the test in the first place or how she can prepare herself for the results. After the gender video, for example, the first likely questions would be “What are chromosome abnormalities?” and “How common are their risks?” rather than “Can’t I just find out the gender of my baby without the other stuff?”

The potential conflicts of interest for genetic counselors have been observed for several

years. On the Natera site, it is, at least, obvious that the counselors in the videos are part of the Natera team. That clear connection may be missing for patients who discover that “some counselors offering them advice in hospitals and doctors’ offices work for the commercial genetic testing companies, not for the hospitals or doctors themselves.”<sup>114</sup> LabCorp is the parent company to a laboratory that, in 2012, had placed over 140 genetic counselors hired by the lab at over 200 service locations, mostly working in prenatal testing.<sup>115</sup> A LabCorp spokesman indicated that the counseling service was only an add-on to the testing and counselor salaries were independent of how many tests were ordered. On the other hand, “some doctors and executives in the testing business say LabCorp needs a certain volume of testing to justify placing a counselor in a doctor’s office. And the genetic testing industry certainly perceives that those counselors help bring testing to LabCorp.”<sup>116</sup> One rival lab began hiring genetic counselors to place in prenatal clinics because “You can’t compete if you don’t go in and do it.”<sup>117</sup> The competition factor could apply even if the counseling was completely neutral, but providers and clinics come to regard counseling as a valuable add-on that frees them from having to hire their own genetic counselors. However, this leads to a further negative impact on the genetic counseling profession as major public insurers such as Medicare and Medicaid,

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*[M]ajor public insurers such as Medicare and Medicaid . . . have begun to pay for genetic testing in some circumstances but “are less likely to pay for the counseling sessions, sometimes lasting an hour or longer, that can precede and follow such testing.*

as well as private insurers, have begun to pay for genetic testing in some circumstances but “are less likely to pay for the counseling sessions, sometimes lasting an hour or longer, that can precede and follow such testing. Most states do not license genetic counselors, and it can be hard for a nonlicensed practitioner to obtain reimbursement.”<sup>118</sup> The more laboratories present genetic counseling with genetic testing as part of a bundled service, the less likely it is that insurers will come to regard counseling in and of itself as a valuable and distinct reimbursable healthcare service.

Many of the genetic counselors hired by laboratories do not, in fact, deal directly with patients, working instead with doctors who are still the ones who order the tests. However, this fact exposes how laboratories can unduly influence genetic testing standards themselves, with more far-reaching consequences than merely influencing testing decisions made by one woman at a time. Professional societies such as ACOG issue guidance on the appropriate tests to conduct, but these are only guidelines and new tests are being developed all the time. Some counselors who are embedded in a hospital or clinic may subtly or overtly campaign for standard panels to be expanded simply because the test exists and has analytical validity. “Dr. Richard Fischer, chairman of maternal fetal medicine at Cooper University Hospital in Camden, NJ, while saying he was pleased overall with the LabCorp counselors, said that he [had] to resist

the company’s counselors to offer a test to all pregnant women that was not recommended by ACOG.”<sup>119</sup>

Conflict of interest issues raised for genetic counselors have not necessarily reached a level that impugns actual patient care provided by genetic counselors employed by commercial laboratories, but larger questions of systemic influence and priming NIPS as an intrinsic component of a professional standard of care is equally concerning to the disability community. If counselors themselves are uneasy with being “pawns in the marketing scheme”<sup>120</sup> of corporate entities, people with genetic conditions find themselves cast in a similar role, but without pay.

People with disabilities and parents of people with disabilities can feel that the commercial NIPS laboratories offer tests on the risk of living lives with disabilities, while failing to offer any context to counteract

existing social and cultural stereotypes and prejudice concerning disability. Genetic counselors potentially offer a counter to the operation of stereotypes and prejudice. This cannot be the case, however, if proliferating for-profit entities rely on a two-prong marketing strategy that implicitly sells NIPS as a means of avoiding disability and employs genetic counselors to affirm NIPS as a gold standard of medical care.

## Genetic Counselors and Disability

Given the potential linchpin role of the genetic counselor in preserving informed choice and autonomy, what do genetic counselors have to

learn about disability? According to the most recent accreditation standards for graduate genetic counseling programs in the United States, as adopted by the Accreditation Council for Genetic Counseling on February 13, 2013, the answer is not very much.<sup>121</sup> The 27-page document, which covers all aspects of accreditation from administration to curriculum to student evaluations and appeals procedures, mentions disability twice. The first time is to indicate that accredited programs cannot discriminate on the basis of listed personal characteristics, including “disabling status.” The second mention is to include “disability awareness” and seven other categories,

including multicultural sensitivity and competency, within a category of psychosocial content that itself is one of nine “general content areas required to support the development of practice-based

competencies in genetic counseling.” It is unclear whether the general content area is directed at equipping counselors to *communicate* with diverse patients rather than educate them about the *lives* of diverse patients. The standards do indicate that the competencies are a floor, and there are other subcategories of content such as health disparities, genetic discrimination, and related legislation under the “social, legal, and ethical” content area, or the delivery and evaluation of educational tools and materials under the “education” content area, where disability-related topics could be raised. But if a genetic counseling program did not have a pre-existing

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*If counselors themselves are uneasy with being “pawns in the marketing scheme” of corporate entities, people with genetic conditions find themselves cast in a similar role, but without pay.*

intention to provide its students with a grounding in disability awareness and rights, there is nothing in the current accreditation curriculum that would foster such an understanding.

Katie Stoll graduated from Brandeis's genetic counseling program in 2003 and recalls it as one of the rare programs that was initiated with a broader understanding of disability than just the characteristic for which genetic testing determines a risk. The program was founded not by a genetic counselor, but by a biologist who was also the parent of a child with a genetic condition/disability. Both the curriculum

and the practicum requirements had disability components and students had multiple contacts with children and adults with disabilities, conducted interviews, and wrote a reflective paper. Stoll recalls being "matched" with an adult who was

living with a genetic condition, and only realizing much later that it was coincidentally the same condition that a couple of relatives had in her own extended family; she had never thought of them as having a disability or herself as someone experienced with disability. In addition, Adrienne Asch, one of the very few bioethicists who was also a person with a disability and a disability advocate, taught at Wellesley College "just down the road, and was periodically invited to guest lecture in the program." Stoll recalls that lecture as a challenging and seminal experience in her genetics counseling education. Asch believed strongly that genetic counseling, while it should

not be directive, could and should involve genuine counseling, and help women and their partners to determine for themselves what they sought in having children, their expectations for family life, what they were prepared to sacrifice, and the rewards they expected. Perhaps in her talks at Brandeis she said something akin to the following:

It is crucial that prospective parents are offered both information about disability and the opportunity to explore the values, desires, fears, and dreams that enter into deciding what to do with prenatal

genetic information. Equally important is that, in accordance with the ethic of genetic counseling, professionals who make such offers honor both acceptances and refusals of these offers.<sup>122</sup>

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*But if a genetic counseling program did not have a pre-existing intention to provide its students with a grounding in disability awareness and rights, there is nothing in the current accreditation curriculum that would foster such an understanding.*

Unfortunately, not every genetic counseling program is as committed to fostering disability awareness. When asked about the extent of their disability-specific education, two genetic counseling graduates who chose not to be named, respectively 4 and 8 years out of different schools, each felt their training had fallen short in some measure. The first had come to genetic counseling from an existing healthcare background and described "exposure to individuals with a wide range of disabilities" via a mandatory 5-week clinical rotation at a home for adults with disabilities,

but still wished that more coaching on how to communicate with people with disabilities or families of people with disabilities had been provided.<sup>123</sup> The second graduate had spent 4 years being a nanny for a child with developmental and intellectual disabilities and attended a genetic counseling program that had provided a “broad overview” of disability, but felt others in the program who lacked such an in-depth experience would have benefited from “a day in the life” type experiences or stories.<sup>124</sup> The first graduate estimated that perhaps 25 percent of the disability information provided in the program was written or taught by people with disabilities.

Genetic counselors could therefore be expected to have no more particular understanding of the social model of disability or disability rights than any member of the public, even though they are expected to counsel that same public about the significance of living with disability. There is limited research on the beliefs and attitudes that genetic counselors hold toward people with disabilities, but a 2011 article that examined multiple studies on the topic found that genetic counselors make substantially different

prenatal testing decisions for themselves than the decisions of the general public.<sup>125</sup> For instance, 96 percent of counselors undertook invasive genetic diagnosis, not NIPS, when

they had a risk factor such as advanced maternal age, and 76 percent of counselors with no indication for prenatal diagnosis would nonetheless choose invasive prenatal testing.<sup>126</sup> Even more striking, a survey of different professions showed that 52 percent of genetic counselors

chose invasive prenatal diagnosis during their own pregnancies; this compares with 5 percent for nurses and 7 percent for high school biology teachers.<sup>127</sup> None of the studies, however, explored counselors’ reasons for their decisions, nor the choices that counselors made upon receiving their fetus’s diagnosis. For example,

genetic counselors may particularly value the ability to prepare for the birth of a child with a genetic condition.

The above studies focused on testing decisions that genetic counselors make for themselves as a proxy for their attitudes

toward disability and as a way to avoid the social desirability bias.<sup>128</sup> Studies based on practice behavior provide additional insight into disability beliefs among genetic counselors. For instance,

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*[A] survey of different professions showed that 52 percent of genetic counselors chose invasive prenatal diagnosis during their own pregnancies; this compares with 5 percent for nurses and 7 percent for high school biology teachers.*

An analysis of 93 transcripts of simulated prenatal genetic counseling visits from the Genetic Counseling Video Project [Roter et al., 2006] found that 95% of genetic counselors talked about the biomedical aspects of Down syndrome compared to 26% who described social aspects of life with Down syndrome [Farrelly et al., 2010]. When discussing options available to a client if a fetus were diagnosed with a disability, among the 93 genetic counselors whose transcripts were analyzed 86% mentioned pregnancy termination, 37% continuation of pregnancy and 13% adoption [Farrelly et al., 2010]. The majority (61%) of the prenatal counselors who participated in the Genetic Counseling Video Project had more than 5 years of experience in genetic counseling. . . .<sup>129</sup>

The authors of the meta-study conclude by suggesting “multiple ways that genetic counselors, on individual and organizational levels, can examine and strengthen the relationship between the genetic counseling and disability communities,” while also continuing “the profession’s support of reproductive freedom and access to prenatal genetic testing opportunities.”<sup>130</sup>

## Genetic Counseling in Uncharted Territory

While this chapter primarily addresses NIPS, the genetic analysis techniques used in NIPS can

and are continually being advanced toward increasingly complex predictive operations, as well as potential combination with other techniques such as preimplantation genetic diagnosis (PGD), in-vitro fertilization (IVF), and genetic engineering. Brave or not, a new world has arrived with staggering implications for people with and without disabilities. Late last year, Genomic Prediction, a US company announced that it now had a test that could screen embryos who potentially had a low IQ.<sup>131</sup> A test of intelligence is much more complex than the single cell chromosome tests used to screen for Down syndrome and related genetic

conditions. Intelligence involves multiple regions of DNA/cells and the resulting findings may have analytical validity but are much less indicative of specific real-life results. The company was reportedly already in talks with IVF clinics to make the test available to

consumers.<sup>132</sup> Company cofounder Stephen Hsu, for ethical reasons, has decided not to offer the test for selecting embryos with a high IQ for implantation, but apparently feels no compunction about offering “the option of screening out embryos deemed likely to have ‘mental disability’.”<sup>133</sup> The test “isn’t accurate enough to predict IQ for each embryo, but it can indicate which ones are genetic outliers, giving prospective parents the option of avoiding embryos with a high chance of an IQ 25 points below average,” says Hsu.<sup>134</sup> He also didn’t rule out making the test available for high

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*When discussing options available to a client if a fetus were diagnosed with a disability, among the 93 genetic counselors whose transcripts were analyzed 86% mentioned pregnancy termination, 37% continuation of pregnancy and 13% adoption.*



intelligence screening in the future, reasoning that if he doesn't do it, someone else will, whether in the United States or elsewhere.<sup>135</sup>

As with the intelligence test, we have the ability to screen for an increasing array of conditions, but the clinical validity of the genetic anomaly is often less clear, as is the specific manifestation of the health condition. When considering something such as the gene for breast cancer or many other conditions, how the gene will express itself over time is not necessarily known. How does one help guide a woman toward actionable choices when she is being told that a fetus has a certain percentage chance of developing a "mild to severe" form of a health condition that may or may not manifest during a person's lifetime? What if these options are replicated for multiple kinds of conditions or traits in a given fetus? The resulting information can be confusing and paralyzing to many women and couples. If the potential for human germline engineering is thrown in, there is the additional need to consider the implications of genetic tinkering that will change humanity's overall genetic heritage. Reporting on the intelligence test noted this point as well, observing that "we don't yet fully understand what other effects the many genes involved in traits like a higher intelligence or lower

risk of heart disease might have. For example, some studies have suggested that people with higher polygenic scores for academic ability are also more likely to be autistic."<sup>136</sup> Other writers observe that the genetic condition that results

in sickle cell anemia also provides protection against malaria"<sup>137</sup> There have also been discoveries of rare and desirable human traits linked to a known gene that "was previously assumed to be a 'junk' gene that was not functional."<sup>138</sup>

In a competitive for-profit testing market, commercial entities are neither inclined nor expected to voluntarily exhibit humility concerning what they *don't* know about their tests, including the impact of tinkering with a fetus or embryo's genetic makeup, for that individual fetus or for future generations. Just how much help can a genetic counselor be? In future scenarios, the existing technologies of IVF and PGD are combined with the nascent technologies of next-generation sequencing

(NGS) and "Easy PGD" so as to enable the generation of literally hundreds of embryos. Large scale sequencing of all these embryos would provide a woman and her partner detailed genetic information on

*each* embryos' chances of expressing a range of physical health conditions, intellectual disabilities, and personal characteristics, temperaments, and capacities. In such an

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*How does one help guide a woman toward actionable choices when she is being told that a fetus has a certain percentage chance of developing a "mild to severe" form of a health condition that may or may not manifest during a person's lifetime?*

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*In a competitive for-profit testing market, commercial entities are neither inclined nor expected to voluntarily exhibit humility concerning what they don't know about their tests . . .*

overwhelming scenario, genetic counselors and other providers are turning to algorithms to evaluate the “best” embryo to implant. But as pointed out by critics, an algorithm “may be worse than the problem it tries to solve by routinizing reproductive decisions based on hidden biases, reducing societal diversity, exacerbating ‘choice overload effects,’ challenging professional norms, and raising the specter of eugenics.”<sup>139</sup> These tools are created using certain assumptions about what most people view as socially valuable and good. Algorithms can hide all kinds of negative assumptions about disability and health that people with disabilities would dispute, backed up by lived experience.

One potential result of the rapid development and proposed use of polygenic genetic analysis is that it places more and more people on “the wrong side of the genetic tracks” as

it were. Whereas the default assumption of the medical profession and the general public is that genetic testing and engineering should be applied to disability, the broad potential application of such technologies to “normalize” or even “improve” *all* human beings has led many more to question what it is that we are trying to “fix” or “cure.” Back in 2007, when NIPS or noninvasive prenatal genetic diagnosis (NPGD) was thought to be just around the corner, some authors applauded the chance to “experience enormous savings as a result of

the decrease in severely ill babies.”<sup>140</sup> But at the same time, they recognized how easily the technology could lead to a slippery slope:

Might NPGD be utilized to prevent the birth of children with less severe disorders (e.g., an extra finger, bowed legs, cleft palate)? With merely the potential to develop a disorder (e.g., 40% chance of breast cancer, 60% chance of Alzheimer’s disease)? With merely undesirable traits (e.g., propensity for obesity) or for selecting the sex of their child? Or parents with disorders such as dwarfism or deafness who want to abort a healthy fetus in favor of raising a child that

is more like them? With clinical NPGD lurking just around the corner, the issue of the “slippery slope” will become especially pressing, necessitating a full and frank discussion—

with representatives from all sides of the debate—of the many weighty issues at stake.<sup>141</sup>

In the decade since the above words were written, there have been numerous attempts to hold full and frank discussions on prenatal genetic testing, but from the viewpoint of the disability community, those discussions have, for the most part, been incomplete, lacking the presence of people living with disabilities and genetic conditions.

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*One potential result of the rapid development and proposed use of polygenic genetic analysis is that it places more and more people on “the wrong side of the genetic tracks” as it were.*

## Chapter 3: Healthcare System Pressures and Legal Protections and Gaps

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### Brief Overview of the US Healthcare System

The US healthcare system includes a complex mix of providers, a diverse range of entities that deliver and/or administer healthcare services or products (increasingly comprised of large and growing for-profit corporations),<sup>142</sup> both public and private health insurance programs, and federal and state laws, regulations, and policies that may affect all of these players. Professional bodies police the behavior and ethics of any given health profession, but often, there is considerable deference given to individual education and training programs. Studies have found great variance among genetic counseling training programs accredited by the ACGC with respect to disability, with programs giving the subject anywhere from 10 to 600 hours.<sup>143</sup> Given this wide range of training, genetic counselors and primary prenatal providers likely devote minimal or no time to informing parents of the kinds of post-birth supports, services, and futures

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*Studies have found great variance among genetic counseling training programs accredited by the ACGC with respect to disability, with programs giving the subject anywhere from 10 to 600 hours.*

available to a child with disabilities. Since genetic counselors are relatively rare, primary caregivers such as practicing obstetricians typically try to fit counseling sessions within the time slots given in clinical practice. Other healthcare providers lack the specialized training that genetic counselors have received on both the science of genetics and intricacies of nondirective counseling. This means that professional genetic counseling can be largely absent where a NIPS kit has been ordered by a provider, sent from the lab, returned for analysis, and the results sent to the provider and patient. In this scenario, “Dr. Google” and the laboratory’s marketing materials may be the only resources readily available to prospective parents who do not know how to interpret or understand the implications of the genetic testing results. The risk of misunderstanding is heightened by the fact that such testing results are, in many cases, *not* a “diagnosis” but only an analysis of a fetus’s increased risk of having genetic anomalies associated with certain health conditions.

For-profit pressures already tend to place a premium on the amount of time that providers spend with individual consumers, and managed care entities and private insurers have a vested interest in trying to attract consumers without disabilities who are viewed as having fewer care needs. For-profit systems, as well as public healthcare programs subject to budget pressures, may discourage the kind of in-depth conversations about living with disability that women who receive genetic testing results may want or need. Moreover, it can be expensive to meet the ongoing healthcare needs of individuals with chronic conditions and disabilities, compared with what is spent on “healthy” individuals. These pressures can lead providers, and especially obstetricians and primary care providers, to be more directive in urging patients to take prenatal genetic tests and terminate pregnancies if genetic testing indicates the presence or probability of a disability.

In addition, providers who conduct genetic testing are likely to hold the same beliefs about the quality of life and limitations of people with disabilities that are held by other healthcare providers as revealed in various studies. For example, 18 percent of emergency room providers such as nurses, residents, attending physicians, and technicians said they would be glad to be alive after a spinal cord injury, compared to 92 percent of people who are spinal cord injury survivors who said they had a good quality of life.<sup>144</sup> Not surprisingly, only 22 percent of the providers surveyed indicated that they would want to be treated

with “everything possible to ensure survival” after a spinal cord injury. A more recent study of pediatric residents found that 71 percent questioned giving aggressive treatment to children with severe disabilities.<sup>145</sup> In the genetic context specifically, there are similar studies/observations as providers’ vocabulary choice and assumptions about next steps can profoundly affect their patients. Doctors “can influence the decision by using phrases such as ‘I’m sorry,’ or ‘I have some bad news to share.’ For instance, 34% of Dutch women who terminated their pregnancy after a Down’s [sic] syndrome diagnosis said that their doctors did not even mention the possibility of carrying the pregnancy to term when discussing their options.”<sup>146</sup>

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*[P]roviders’ vocabulary choice and assumptions about next steps can profoundly affect their patients.*

Genetic medicine and engineering have been capturing the public imagination at a time when law and policymakers are

increasingly concerned with rising healthcare costs. A case study by the American Consumer Institute explicitly and favorably compares the cost of one-time gene therapy cures to the costs of conventional treatments for such genetic diseases such as hemophilia and spinal muscular atrophy.

The costs of research and development are enormous, and many gene therapies are expected to be priced at more than \$1 million per patient to provide an adequate return on investment . . . policymakers and payers should look beyond the sticker-shock. Curing a disease, rather than treating it, can not only provide

an immense improvement in longevity and quality of life, it can also save insurers and taxpayers money in the long-run.<sup>147</sup>

To be clear, genetic therapies and treatments for people with disabilities are not the same as genetic prenatal testing, and raise an entirely different set of ethical, social, and legal implications. This study is not advancing any opinion or recommendation on whether, how, or why people with disabilities gain access to genetic therapies and treatments. This point is only analogous. Healthcare costs are both particularly opaque and difficult to control in a fragmented, multiplayer healthcare delivery system. Once genetic treatment is singled out as an effective way to reduce the expenses of “debilitating and deadly diseases” caused by “defective genes,”<sup>148</sup> it seems only a matter of time before the impetus to fix or cure disability evolves into a mainstream cost-based argument. If all women routinely receive prenatal genetic testing for all genetic conditions, who or what will protect the integrity of a woman’s choice to proceed with the pregnancy of a fetus that bears the markers for hemophilia and spinal muscular atrophy, for example? Would that choice bring with it an obligation to bear some or all of the medical and other costs of raising and maintaining the health of a child with genetic conditions when such costs could have been avoided through termination of a pregnancy? These are the kinds

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*[W]ho or what will protect the integrity of a woman’s choice to proceed with the pregnancy of a fetus that bears the markers for hemophilia and spinal muscular atrophy, for example? Would that choice bring with it an obligation to bear some or all of the medical and other costs . . . ?*

of questions that must be asked once we enter the universe of healthcare financing and cost, and they need to be asked imminently. As described in Chapter 2, some investors in NIPS technology already equate the use of NIPS testing with terminating the pregnancy of fetuses with disabilities, and the two processes together are characterized as a reasonable cost control measure.

## **Current Genetic, Nondiscrimination, and Consumer Protections in Federal and State Law**

### ***Pre-Birth***

There are currently two main sources of legislation that most directly have an impact on prenatal genetic testing and information. The first source stems from the 2008 passage of the federal Prenatally and Postnatally Diagnosed Conditions Awareness Act (PPDCAA).<sup>149</sup> The second source is rooted in state efforts to restrict the options available to women once they receive the results of prenatal genetic testing. Other relevant laws that have a possible impact on prenatal genetic testing are general disability rights nondiscrimination laws that have the potential to be raised against healthcare professionals who work in genetic testing. Finally, there are consumer protection topics that are regulated in other areas analogous to prenatal genetic testing, where commercial for-profit entities have an outsized interest over medical and technology

breakthroughs that directly influence patients. This section will address the above topics in order.

The PPDCAA is the most significant federal legislation to arise at the junction of disability rights and prenatal genetic testing. Sponsored by Senators Brownback and the late Ted Kennedy, the latter known as a long-time disability advocate, the bipartisan legislation worked its way quickly through Congress over 3 weeks in the fall of 2008, but lost a proposed 5 million dollars in funding during the amendment process.<sup>150</sup> Since its passage, the law “has been funded at only a fraction of the requested amount, and it has had minimal impact in providing families with the essential information of its aim.”<sup>151</sup> The law has three explicit goals:

- to provide women who have received a prenatal or postnatal diagnosis for Down syndrome or another condition with provider referrals for key support services, and “up-to-date information on the range of outcomes for individuals living with the diagnosed condition, including physical, developmental, educational, and psychosocial outcomes”;
- to strengthen existing networks of support through the Centers for Disease Control and Prevention, the Health Resources and Services Administration, and other patient and provider outreach programs;
- to ensure that patients receive “up-to-date, evidence-based information about the accuracy of the test.”<sup>152</sup>

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*[PPDCA] has been essentially ineffectual because of its lack of funding, but its existence has been critical nonetheless in prompting 17 state measures . . .*

Some of the activities that can be authorized by the Secretary of HHS under the second goal include the provision of telephone hotlines, improved outreach and peer-to-peer counseling programs, the creation of family registries willing to adopt children with genetic conditions, and establishment of education services for medical providers who “provide, interpret or inform” prenatal test results for pregnant women and their partners.<sup>153</sup>

The federal law has been essentially ineffectual because of its lack of funding, but its existence has been critical nonetheless in prompting 17 state measures<sup>154</sup> to ensure that prospective and current parents of children with genetic conditions receive accurate information and support for making informed choices about

prenatal and postnatal test results. Two other states, Virginia and Missouri, enacted laws similar to PPDCAA 2007, prior to the federal law.<sup>155</sup> While all 19 jurisdictions share language and goals

similar to the PPDCAA, there are some interesting differences. For instance, requirements in the federal law trigger upon a genetic diagnosis of Down syndrome or other prenatal or postnatal disorders, but most of the state laws tend to be narrowly focused on a Down syndrome diagnosis (or Trisomy condition, which is slightly broader), with the exceptions being Virginia, Missouri, Indiana, and Florida (Down syndrome and other *developmental* disability). The laws also differ in the degree to which they mandate information be given to actual parents. Most states indicate that information on genetic conditions and service

referrals must be given to providers involved in referring, providing, or counseling on genetic testing, but then do not mandate that the information be passed on to parents. Delaware's law seems to require only the posting of information on a departmental website for providers to access. Maryland, Illinois, Washington, and Tennessee indicate that information must be given to providers but gives providers the discretion to share information with parents; Maryland expressly changed from a mandatory to a precatory sharing requirement in a later version of its law. New Jersey simply requires providers to give parents access to the information on the providers' websites.

There are also some unique provisions among individual state laws that may reflect specific

local advocacy efforts

within those states, and/

or particular political

leanings. Kentucky

focuses on Down

syndrome but also pulls

in positive screenings for

spina bifida as a trigger

for the act.<sup>156</sup> Louisiana

prohibits "explicitly or implicitly presenting

pregnancy termination as a neutral or acceptable

option when a prenatal test indicates a probability

or diagnosis that the unborn child has Down

syndrome or any other health condition" as a

ban on engagement in discrimination based on

disability or genetic variation.<sup>157</sup> Texas protects

providers by stating that a failure to provide

required information nevertheless does not

result in grounds for civil and criminal liability,

and Tennessee also states that the act does not

create a duty of care. Texas and New Jersey

are the only laws that require translation of

information into Spanish. Only Florida, Missouri, and Maryland explicitly require the provision of information about the accuracy of genetic testing itself.

The second category of laws that have arisen in response to the broadening availability of prenatal genetic testing have been outright bans, either on genetic testing for certain conditions or outright restrictions on a woman's right to terminate her pregnancy after the fetus has received a screening for Down syndrome, a genetic condition, or is a particular gender. Between 2014 and 2018, numerous states attempted to enact legislation along these lines: South Dakota (2014), Indiana (2014), Ohio (2017), New Hampshire (2016), Oklahoma (2018), and Missouri (2016).<sup>158</sup> Most, but not all, of these

bills barred penalizing

the woman who was

seeking an abortion or

who attempted to get

an abortion, instead

imposing fines and/or

imprisonment on the

healthcare providers

who performed or

would have performed the abortion. Ohio

and Indiana's bills did not include an explicit

protection against penalty for the woman.

Planned Parenthood Advocates of Missouri

fought that state's 2016 bill as a restriction

on women's reproductive rights but also

acknowledged the relevance of disability rights

concerns, stating "[t]his is about politics and

taking away women's ability to make personal,

private, and often complicated decisions.

This bill does nothing to address the serious

underlying concerns about discrimination

against people with disabilities."<sup>159</sup>

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***Most states indicate that information on genetic conditions and service referrals must be given to providers . . . but then do not mandate that the information be passed on to parents.***

In 2015, a bill that would have prevented the Department of Health from providing written information on abortion after a Down syndrome diagnosis was introduced in the Texas House of Representatives. According to *Healthline*, the bill failed that year,<sup>160</sup> but as of 2018, Texas law included a provision that enacted a state version of the federal PPDCAA. That is, Texas requires current and accurate information about living with Down syndrome and service referrals to be provided to parents who have received genetic test results indicating a likelihood for the fetus having Down syndrome.<sup>161</sup> As well, the law says that information provided cannot “explicitly or implicitly present pregnancy termination as an option when a prenatal test indicates that the unborn child has Down syndrome.”<sup>162</sup> This example shows how the lines between the first and second categories of law can be blurred, and requires careful thought among the disability community when developing and advocating for laws that will help bring current, accurate information about living with a disability to those who undergo prenatal genetic testing. Information laws can easily be “co-opted” and amended by groups or lawmakers who wish to include barriers on women’s right to full reproductive information and their practical ability to act on their choices, including the choice to terminate a pregnancy for private reasons.

The third category of laws are disability nondiscrimination laws. Section 504 of the Rehabilitation Act of 1973 (Section 504) and the Americans with Disabilities Act of 1990 (ADA) prohibit disability discrimination in healthcare settings such as provider offices and hospitals.<sup>163</sup> Whether prenatal screenings such as ultrasound and NIPS, and higher risk prenatal diagnostic

tests such as amniocentesis or CVS, are provided by a Medicaid care provider, paid for through other public or private insurance, or paid for purely out-of-pocket, the providers performing them will generally be subject to federal disability rights law. Currently, there are no specific regulations or guidance dictating what those laws require. For example, must a NIPS company provide all the information that it sequences, and must it completely return all that information to the consumer?<sup>164</sup>

As well, there may be barriers to establishing when discrimination in genetic testing provides a sufficient basis for taking legal action. Even if a genetic counselor or other provider overtly advised aborting a fetus that screened or was diagnosed with disability, under most established reproductive rights cases and law, the fetus is not a person who could bring a Section 504 or ADA action or administrative complaint. Prospective parents of the fetus may be able to show that they were subject to discrimination based on their association with a future individual with a disability because they were advised to terminate the pregnancy, unless courts conclude that association with a future individual is too speculative. Successful actions have been brought against providers for failing to detect a child’s disability through prenatal screening and provide the prospective parents with genetic information and advice that would have led to abortion; these rulings should demonstrate that genetic testing and advice about pregnancy termination are part of a provider’s expected standard of care. Discrimination claims might also be brought against genetic counselors or providers who refuse to follow parental wishes to identify and implant a fetus that carries a particular genetic marker for a desired health



condition or disability, although the viability of such claims is unclear.

The Affordable Care Act (ACA) also contains a number of consumer protections that benefit people with disabilities in the genetic testing context. For example, ACA includes maternal health benefits among the 10 categories of Essential Health Benefits that insurance plans must cover. This means that prenatal genetic counseling is usually covered by insurance when it is prescribed or recommended by a physician. At this point, we are not aware of any trend among health insurers to *require* prenatal genetic testing, or condition future coverage upon parents making certain decisions such as ending a pregnancy upon receiving a high probability screen or diagnostic test that indicates disability in a fetus.

In addition, Section 1557 of ACA prohibits discrimination in benefit design, and ACA bans a refusal to issue insurance based on a pre-existing condition or to charge higher premiums when the beneficiary has a pre-existing condition. These factors may inhibit insurers from requiring beneficiaries to undergo prenatal genetic testing. Notably, ACA and Genetic Information and Nondiscrimination Act (GINA) consumer protections do not apply to life, long-term care, or disability insurance policies, so it is possible that these policies might carry a requirement for prenatal genetic testing. Furthermore, short-term limited duration health plans, and potentially Association Health Plans,

which are now more widely permitted under the current administration's rules, are often exempt from ACA's consumer protections.

The final category of laws to consider are consumer protections that have been enacted to increase transparency and restrain actual and perceived conflicts of interest by corporate stakeholders. These include "sunshine laws" and conflict of interest provisions that require pharmaceutical and other medical treatment or device commercial entities to disclose their relationship with prescribing healthcare providers.<sup>165</sup> Under the Sunshine (Open Payments) Act,<sup>166</sup> CMS operates an Open

Payments Database that is publicly accessible and searchable, with results that are downloadable.

The Act requires "manufacturers of covered drugs, devices, biologics, or medical supplies to collect detailed information about payments and other 'transfers of value'

worth over \$10 from manufacturers to physicians and teaching hospitals"<sup>167</sup> and record this on the Open Payments database. The database covers a wide range of providers who have an active license to practice, including doctors of medicine, osteopathy, dentists, podiatrists, optometrists, and licensed chiropractors. There is currently no requirement that places such an obligation on NIPS or other commercial entities and laboratories engaged in prenatal genetic testing. Nor are there any disclosure requirements that require genetic counselors to disclose that they are working for third parties other than the

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***ACA and Genetic Information and Nondiscrimination Act (GINA) consumer protections do not apply to life, long-term care, or disability insurance policies, so it is possible that these policies might carry a requirement for prenatal genetic testing.***

physician offices, prenatal clinics, or hospitals where they are embedded, but employed by a laboratory. While Sunshine Act and conflict of interest guidelines currently comprise a gap in regulation for the genetic testing industry, the laws themselves would be a second step. Unlike pharmaceutical companies, prenatal genetic testing companies are not currently regulated by CMS in any substantive way. CMS should first extend its jurisdiction over genetic testing companies, thereby clearing the way for federal and state legislation to foster needed consumer protections for genetic testing consumers.

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*. . . Sunshine Act and conflict of interest guidelines currently comprise a gap in regulation for the genetic testing industry . . .*

### **Post-Birth**

#### **How Effective Have Federal Anti-discrimination Laws Been in Protecting Individuals from Workplace Discrimination Based on the Results of Genetic Testing?**

Disability-based discrimination occurs not only in the process of prenatal genetic testing, but also with respect to genetic tests conducted after birth. That is, employers, insurers, and others have discriminated against individuals based on the presence of a genetic marker or a condition identified through genetic testing. As Mark Rothstein, Director of the Institute for Bioethics, Health Policy and Law observed, “[e]mployers have a tremendous economic incentive to discriminate based on perceived future health status. They’ve done it with other conditions and we have every reason to expect they would do it in the genetics area.”<sup>168</sup>

Both the ADA and GINA have been used to address such discrimination in the workplace, where it has raised some of the greatest concern. In addition, most states have laws prohibiting some types of discrimination based on genetic information, though there is a great deal of variation among those laws and most are not as protective as federal law.

As scientists began sequencing the human genome in the early 1990s, public concerns about the potential for genetic discrimination began to proliferate, and proposals to address this type of discrimination started to be introduced in Congress.<sup>169</sup> While opponents of genetic anti-discrimination legislation argued for years that instances of discrimination based on genetic information are extremely rare, studies over time demonstrated that concerns about such discrimination were well founded. In 1996, a study of individuals who were at risk, presymptomatic, or asymptomatic for a genetic disorder found that nearly 48 percent of the respondents reported that they had experienced some form of discrimination based on their risk status.<sup>170</sup> A 2000 Northwestern National Life Survey found that 15 percent of employers intended to obtain job applicant’s genetic information before extending a final offer of employment.<sup>171</sup> A 2004 study by the American Management Society concluded that:

Companies were genetically testing employees for risk of breast and colon cancer, Huntington’s disease, and susceptibility to workplace hazards. One in

six of the companies surveyed collected family medical histories of their employees. At least one-half of the companies testing for breast and colon cancer risk and Huntington's disease considered the results in their hiring, re-assigning, and firing decisions. One-fifth of the companies use the information gleaned from family medical histories, and more than one-half use the information from tests regarding susceptibility to workplace hazards in their employee decision-making.<sup>172</sup>

A number of high-profile cases have been brought under the ADA and GINA to challenge employers' efforts to take

adverse action against individuals based on genetic test results or to force employees to disclose their genetic information. Perhaps the most notable case, and one that played a role in creating momentum for the passage of GINA, was a case brought by the

US Equal Employment Opportunity Commission (EEOC) against Burlington Northern Santa Fe Railway (BNSF) challenging BNSF's actions of secretly conducting genetic tests on employees who had made internal claims of work-related carpal tunnel syndrome without the employees' knowledge.<sup>173</sup> The employees were told that they had to undergo medical testing "to ensure that all possible contributing causes of [carpal tunnel syndrome] were work-related." Employees were not told that BNSF's doctor would draw vials of blood to be shipped to a genetic testing facility

to determine whether the individuals might have a rare genetic condition called Hereditary Neuropathy with liability Pressure Palsies (HNPP). HNPP is a progressive neuromuscular disorder that makes individuals susceptible to nerve injury from pressure, stretch, or repetitive use and, according to the EEOC, is very unlikely to be found in people who have work-related carpal tunnel syndrome.<sup>174</sup> The EEOC brought the case after one employee, upon discovering what tests were being performed, refused to take the test. BNSF scheduled him to appear before an "investigation" panel for a hearing on insubordination and violation of safety rules, with the possibility of termination. This resulted in the union's involvement, and

the EEOC sought an immediate injunction to stop the testing.<sup>175</sup>

The EEOC argued that the testing violated the ADA's bar on medical examinations of employees that are not job-related and consistent with business necessity. The Commission argued that the test would not

be medically necessary unless it was to determine whether a condition interfered with an employee's ability to perform the essential functions of the job or whether the person posed a direct threat to health or safety. Instead, the test was designed to determine the cause of the carpal tunnel syndrome.<sup>176</sup> The case eventually settled, with BNSF agreeing to end the genetic testing of its employees and to pay \$2.2 million to compensate employees.<sup>177</sup> While the ADA bans employers from using medical tests that are not job-related and consistent with business

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With the passage of GINA in 2008, Congress intended to encourage people to participate in medical genetic testing and ensure that this would not lead to loss of a job or health insurance.<sup>178</sup> Members of Congress had begun introducing legislation to address genetic discrimination as early as the 1990s.<sup>179</sup> President Clinton issued an Executive Order in 1992 forbidding genetic testing as a condition of employment in executive branch agencies,<sup>180</sup> and by the time of

GINA's passage, most states had some type of legislation addressing genetic discrimination.<sup>181</sup> GINA's findings indicate that a national standard "is necessary to fully protect the public from discrimination and allay their concerns about

the potential for discrimination, thereby allowing individuals to take advantage of genetic-testing technologies, research, and new therapies."<sup>182</sup> GINA prohibits employers from discriminating based on the genetic information of employees or applicants, and also bars employers from requesting, requiring, or purchasing such genetic information, subject to a number of exceptions.<sup>183</sup>

It is difficult to gauge how much GINA has changed employers' actions to seek or use employees' genetic information, but a 2019 survey of the caselaw under GINA published in 2019 provides some insight into the types of claims that have resulted in court decisions. During GINA's first 10 years, there were few

court decisions. The study found court decisions in only 48 cases with plausible GINA claims.<sup>184</sup> While many lawsuits are settled or otherwise disposed of without reported court decisions, the number of decisions is sufficiently small to indicate that GINA lawsuits are rare. And GINA charges filed with the EEOC rose for several years and then began falling steadily.<sup>185</sup> The court decisions reviewed in the study included none alleging workplace discrimination based on genetic test results. Moreover, few involved successful claims establishing that employers took adverse action based on genetic

information.<sup>186</sup> The study authors point out, however, that GINA has been most successfully used as a workplace privacy statute, invoked to challenge employers' unlawful requests for employees' genetic information or employers' unlawful disclosure of

that information. Nearly 40 percent of the cases involved claims based on employers' requests for family medical history, requests for medical records or exams, or employers' disclosure of genetic information.<sup>187</sup> Notably, the Justice Department entered its first settlement involving GINA claims in 2018, resolving claims that the Minneapolis Police Department routinely requested and obtained genetic information from applicants for police officer positions.<sup>188</sup>

To be sure, there have been lawsuits challenging adverse employment actions based on genetic testing information despite the absence of reported decisions. For example, Pamela Fink, who filed the first charge with

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EEOC alleging employment discrimination under GINA, resolved her charge through a confidential settlement with her employer. Ms. Fink's charge alleged that after nearly 4 years at her company, with consistently glowing reviews, she was suddenly terminated after she underwent surgery for a double mastectomy and reconstructive surgery following genetic tests revealing that she had the BRCA2 gene and had an 80 percent chance of developing breast cancer. The day before Ms. Fink's reconstructive surgery, she received a scathing review, and within 2 months she was fired.<sup>189</sup>

One of the stakeholders interviewed for this report, April Kiser, also sued to challenge employment discrimination based on genetic testing information. After 18 years as a city employee working for the police department in Jacksonville, Arkansas, Ms. Kiser was tested for and found to have the BRCA1 and BRCA2 gene mutations. With a 90 percent chance of developing breast cancer, she sought to have the city's health plan cover a double mastectomy and reconstructive surgery recommended by her doctor. Ms. Kiser's claim was denied and her appeal required arguing before a panel of mayors, including her employer, the mayor of Jacksonville, who made belittling comments when she described her health history and hormone treatment. During the appeal, Ms. Kiser disclosed her genetic testing information and family history of breast cancer. Her appeal was denied and the city refused to cover her

surgeries. As a result, Ms. Kiser did not have the double mastectomy, which would have cost more than she made in a year. In addition, Ms. Kiser experienced workplace discrimination. She filed a charge of discrimination with the EEOC following her employer's belittling comments at her appeal hearing, and several days after the EEOC terminated its investigation, her supervisor eliminated her job and reassigned her from being a public information officer to scanning old files. Shortly afterward, her pay was reduced as well. Ms. Kiser filed a complaint in state court, challenging the city's actions under GINA as well as the Arkansas Civil Rights

Act's bar on gender discrimination. Her lawsuit is pending.

The more common usage of GINA to address workplace privacy concerns and prevent employers from seeking employees' genetic information is perhaps unsurprising

in a "big data" era when health and genetic information have become commodities and are increasingly being sought in a variety of contexts.

### **Recent Congressional and Agency Efforts to Reduce ADA and GINA Protections for Employees to Keep Their Disability and Genetic Information Private from Employers in Workplace Wellness Programs**

One of the ways that employers have sought to collect employees' genetic information, as well as disability-related information, is through the workplace wellness programs that employers

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have used in an effort to try to curb rising healthcare costs. The use of such programs began to expand significantly after ACA increased the level of incentives and penalties that could be used to pressure employees to participate in these programs, to disclose information, and to meet health targets, beginning in 2014.<sup>190</sup> By 2016, 90 percent of large firms that offered health benefits offered some type of wellness program, and 71 percent of large firms gathered information about employees' health status using health screening tools, such as health risk assessments and biometric screens.<sup>191</sup>

There is little public information about the extent to which genetic information is being collected through workplace wellness programs. However, as discussed below, recent years have seen legislative and regulatory efforts to promote the use of penalties or incentives to pressure employees to provide genetic information (as well as disability information) through workplace wellness programs.

Both the ADA and GINA place limits on how incentives or penalties can be used in workplace wellness programs—for example, these laws forbid an employer from seeking disability or genetic information through a wellness program unless the program is voluntary, meaning that individuals cannot be penalized for choosing not to disclose this information.<sup>192</sup> Thus, while ACA allowed significant penalties and incentives to push employees to disclose information in workplace wellness programs, the ADA and GINA apply simultaneously and prohibit such penalties or incentives where the information sought is genetic or health information. In fact, the EEOC's GINA regulations, which were issued after passage of ACA and the expansion of workplace

wellness programs, specifically state that an employer “may not offer a financial inducement for individuals to provide genetic information” in a wellness program and that any inducement for completing a health risk assessment must be offered regardless of whether an employee chooses to answer the questions about genetic information.<sup>193</sup>

Responding to pressure from business groups, however, the EEOC in 2016 issued regulations purporting to “align” worker rights under the ADA and GINA with ACA's provisions, allowing steep financial penalties for declining to disclose information in a wellness program.<sup>194</sup> The new GINA regulation permitted employers to impose penalties of up to 30 percent of the value of individual insurance premiums, amounting to thousands of dollars each year, on individuals who chose to exercise their rights under GINA to keep private genetic information that consisted of family medical information. The new ACA regulation permitted employers to impose similar penalties on employees who chose to exercise their rights under the ADA to keep disability information private. These new regulations ignored the plain language of the ADA and GINA. They also ignored the fact that the protections against disclosure to employers under the ADA and GINA were designed to address the potential for workplace discrimination—an entirely different purpose than ACA's wellness provisions, which addressed discrimination in *insurance* plans.

The new regulatory provisions allowing these steep financial inducements to disclose health and genetic information were invalidated by a federal district court in 2017. A legal challenge to the regulations brought by AARP resulted in the court finding that the regulations were arbitrary

and capricious and that EEOC had failed to offer an adequate explanation for its decision to construe “voluntary” wellness programs to allow these types of financial inducements.<sup>195</sup>

In addition to the EEOC’s unsuccessful move to weaken workplace protections against forced disclosure of health and genetic information, a number of legislative efforts aimed to do the same, in even more draconian ways. Most recently, in 2017, Representative Virginia Foxx, then Chair of the House Education and Workforce Committee, introduced a bill that would have largely eliminated ADA and GINA protections for employees in workplace wellness programs, allowing employers to use large financial penalties to force disclosure of this information.<sup>196</sup> This proposed legislation garnered significant negative press, particularly focused on its weakening of GINA protections. It was never voted on by the full House of Representatives.

While efforts to weaken ADA and GINA protections against forced disclosure of genetic and health information in workplace wellness programs have not succeeded, it is likely that such efforts may resurface. NCD believes it is critical that these ADA and GINA protections remain in place. Congress enacted those protections in order to minimize the potential for discrimination, recognizing that once employers have this type of information, it is difficult to guard against it being used in a discriminatory manner and it is often difficult for employees to prove that such discrimination has occurred.

## State Laws Barring Discrimination Based on Genetic Testing Information

Thirty-five states have state laws related to genetic discrimination in employment.<sup>197</sup> By and large, GINA is at least as protective as those state laws, and typically more protective.

Most state laws offer lesser protections overall than GINA. Delaware’s law, for example, bars employers from intentionally collecting genetic information of an employee or applicant or a family member, but offers a broad exception where the information collection is “job-related and consistent with business necessity.”<sup>198</sup> That exception stands in contrast to GINA’s narrow exceptions—for example, where the employer

inadvertently acquires the information, where the employer must collect family medical information to comply with the Family and Medical Leave Act, where the information

is used under certain conditions to monitor the biological effects of toxic substances in the workplace, and as part of a voluntary wellness program.<sup>199</sup> Kansas bars employers from obtaining or seeking to obtain or use genetic screening or testing information, but only for the purpose of discriminating, distinguishing, or imposing restrictions based on that information.<sup>200</sup> Maryland has no exceptions to its prohibition on employers requesting or requiring genetic tests, but the bar is only for tests used as a condition of hiring or determining benefits.<sup>201</sup> Florida provides protection from genetic discrimination in employment only to people with sickle-cell trait, prohibiting

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“screening or testing for the sickle-cell trait as a condition for employment.”<sup>202</sup>

Some states provide protections that are more specific than GINA’s in some respects, though typically those more specific protections seem to cover the same ground as GINA. For example, the District of Columbia specifically prohibits employers from administering a genetic test as a condition of employment,<sup>203</sup> but administering such a test would presumably run afoul of GINA’s bar on requesting or requiring genetic information. And unlike GINA’s protections, DC’s protections do not extend to the genetic or medical information of an individual’s family members. Michigan bars employers from acquiring genetic information “of an employee, applicant, or family member of an employee or applicant”<sup>204</sup> with only *one* exception: where the information is “voluntarily provid[ed]” by the employee and “is related to the employee’s health or safety in the workplace.”<sup>205</sup>

## Enforcement

Fourteen states and DC allow parties alleging discrimination to immediately bring civil suits,<sup>206</sup> and five states, like GINA, allow parties alleging discrimination to sue following exhaustion of administrative remedies.<sup>207</sup>

Fourteen states and DC establish a process to file a complaint alleging discrimination with a commission, state department of labor, or similar administrative body charged with investigating such matters,<sup>208</sup> while nine states grant enforcement authority to the attorney general, a commission, or similar, or establish penalties for violations, but do not explicitly establish a complaint process.<sup>209</sup>

## Wellness Programs

Only one state, Illinois, has specific provisions governing employers’ use of genetic information and genetic testing in workplace wellness programs.<sup>210</sup> The Illinois law provides a similar exception to GINA’s, allowing employers to collect information pursuant to a voluntary wellness program. Illinois’ statute prohibits an employer from using an employee’s genetic information or genetic testing “in furtherance of a workplace wellness program” unless the employer offers “health or genetic services”; the employee provides the employer with written permission; individually identifiable information concerning the results of these services are received only by the employee, a family member if the family member is receiving genetic services, and the licensed healthcare professional or licensed genetic counselor involved in providing such services; and that information is only available for purposes of such services and is not disclosed to the employer except in aggregate terms that do not disclose the identity of specific employees.”<sup>211</sup> Furthermore, an employee cannot be penalized by an employer for choosing not to “disclose his or her genetic information” nor can an employee be penalized for choosing not “to participate in a program requiring disclosure of the employee’s genetic information.”<sup>212</sup> This exception mirrors GINA’s voluntary wellness program exception.<sup>213</sup>

California is currently considering a law to provide strong protections for employees’ health and genetic information in workplace wellness programs.<sup>214</sup> California’s proposed legislation would go further than GINA, although the enforcement mechanism would be to file a



complaint with the state's labor commissioner, rather than to seek relief through the courts.<sup>215</sup> The proposed California legislation would bar employers from requiring participation in wellness programs "as a condition of employment," and from penalizing an employee or otherwise retaliating against an employee "in response to a matter related to a wellness program, such as an employee's election to not participate in a wellness program or the data collected through the wellness program about the employee."<sup>216</sup>

California's proposed legislation would bar employers from offering an incentive or reward to an employee based on adherence to a wellness program.<sup>217</sup> In addition, it would require that employers provide employees with "a written explanation, in clear and easily understandable language, about the basis of the wellness program, a description about the data collection process and which data will

be collected through the wellness program, policies and practices pertaining to the wellness program, and the employee's rights concerning the wellness program under federal and state laws and regulations."<sup>218</sup> The bill, however, would not explicitly prohibit an employer from *receiving* individually identifiable information, instead requiring that "[a]n employer shall not *share* any personal information or data collected through a wellness program" and that the employer maintain "compliance with state and federal privacy laws."<sup>219</sup>

While state laws addressing genetic discrimination in employment do not offer many advantages in comparison with GINA, California's workplace wellness bill offers a useful model for states wishing to adopt a law, or modify their existing law, concerning genetic information discrimination in employment; however, its enforcement provisions could be much stronger.



## Chapter 4: Finding a Way Forward Among Competing Concerns

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### Timing

The topic of genetic testing, even leaving out the particular concerns of the disability community, touches many distinct stakeholders and identifiable groups: reproductive rights and reproductive justice advocates, civil rights advocates, pro-life/anti-abortion advocates, genetic counselors, medical providers, parents and families, employers, commercial laboratories, industry investors, bioethicists, insurers, federal and state law, and policymakers. The list of concerned parties is long because genetic testing, particularly of embryos and fetuses, inevitably leads to the potential for genetic manipulation and control, thereby raising questions about the direction of the human race. However, one difference among the many stakeholders is the timeframe for their concerns. Individuals and families who are focused on the potential for genetic treatment of specific health conditions and short life spans have an urgent window on genetic analysis. Investors and employers have a short-term viewpoint, impelled by the need to develop a patentable product, develop a financially sustainable company, and/or

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maintain a reliable, healthy workforce. By comparison, individuals with disabilities and disability advocates who argue for caution and a social justice approach because of fears about the impact of genetic testing on current and future generations of people with disabilities may appear to have a lengthier timeframe. This difference in timing can be perceived by regulators and policymakers as a reason to delay changing a free-market status quo, or in the case

off employment, a reason to delay or backtrack on existing regulatory measures that would restrict employer actions in favor of preserving the nondiscrimination rights of people with disabilities. But the astounding pace of

genetic analysis and the fact that inheritable germline genetic engineering *has arrived*<sup>220</sup> means that law and policymakers face an urgent time frame for all stakeholders.

Moreover, the stake of the disability community in genetic testing has not benefited from a decades-long academic and policy discussion while genetic advances continue under laissez-faire control here in the US. Section 504 was enacted in 1973 and key

disability-related regulations have now passed their 40th anniversary. The ADA approaches its 30th birthday in 2020. Section 1557 of ACA freshly embeds nondiscrimination in healthcare. The disability community has seen significant advances in social integration, physical accessibility, educational levels, and the length of their lives. Yet none of these advances have had, or will necessarily have in the future, an impact on how the general public perceives the experience and quality of life of people who live with disabilities and genetic conditions, and the lives of the parents of children with disabilities. Deeply rooted stereotypes about the limited capacities and worth of people with disabilities are not so easily erased. As Adrienne Asch wrote in 2003:

I fear that the current climate in which prenatal testing takes place displays neither the rhetorical agility nor the social commitment to equality that I desire. If we are ever to make it at least as acceptable to bear and raise a disabled child as a non-disabled one, we must simultaneously commit ourselves to both social reforms that include all people, whatever their characteristics, and to accepting consumerism in the reproductive marketplace.<sup>221</sup>

Without a genuine and timely commitment to disability and social justice policy and regulatory measures in the field of genetic testing, the United States will continue to see two tracks that will increasingly diverge. One track will see

genetic testing and genetic engineering continue to hold the unnuanced view that the elimination of disability through genetic testing advances humanity's physical, social, and economic wellbeing. The other track will have people with disabilities advocating for the ongoing elimination of social, economic, and legal barriers to their full participation in society. Even if these tracks never meet, they can influence one another. The disability community has spent decades hoping that the second track of full inclusion in society would come to influence the first and propagate a fuller understanding of what it means to live a life with disability, but the opposite appears to be true as the field of genetics tends to prioritize disability elimination over treatment and equal participation.

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## Building Consensus

Many possible courses of action may reduce the adverse impact of genetic testing on individuals and communities with disabilities. These actions range from informal individual measures such as talking to and educating one's friends, neighbors, and colleagues about disability rights, to the development of nonbinding guidance for professional stakeholders, to the adoption of binding regulations, to the enactment of state laws that curtail abortion on the basis of disability.

This paper's introduction established a principle of respect for women's autonomy over their own reproductive choices. Particularly in the current polarized political climate, many disability rights advocates would be uneasy about promoting restrictions on a woman's right to choose to end a pregnancy for whatever reason.

The last action in the spectrum of action listed above is therefore eliminated. The first action, as well, can always be encouraged and should always be taking place, regardless of additional ongoing advocacy for guidance or regulation. In fact, bringing regulatory and policy proposals can provide the opening that will prompt conversation about what it means to live with disabilities. The middle option, of advocating for specific professional guidelines and state and federal policies and regulations to govern the players in genetic testing, will require outreach to various stakeholder groups who would not otherwise support such proscriptive steps. Some stakeholders, such as industry, may never come to support regulation of the field. While it might be possible to achieve compromise with industry on some issues, disability advocates must also keep in mind the importance of maintaining close communication and ties with other, more natural allies, such as civil rights, reproductive justice, and social justice advocates, who may also have distinct concerns with the advance of genetic testing and personalized medicine.

The disability community focus will most naturally be on guidance or regulations concerning the quality and breadth of genetic counseling and information provided to prospective parents upon the receipt of a disability-positive screen or diagnosis, maintaining independence for the genetic counseling profession, ensuring disability-related education for all professionals involved in genetic testing, and direct regulation of industry marketing, testing, and outreach practices. Stephanie Meredith raised the concern of how direct one-on-one sponsorship between one genetic company and a specific advocacy group could lead to actual or perceived undue influence

over that group's informational materials and advice on genetic testing. Imposing a more generic "tax" on all commercial laboratories that would go toward an advocacy fund for which groups could apply would help eliminate the risk and appearance of undue corporate influence.<sup>222</sup>

In the seminal Parens and Asch article from 1999, the experts who assembled for five multiday meetings over a 2-year period engaged in a robust discussion that elevated participants' knowledge and shared understandings, but the group still could not reach agreement on what kinds of disability-specific screens could or should be offered in prenatal testing. Every such attempt seems to splinter a highly diverse disability community, and raise difficult questions about how to balance individual and collective needs, rights, and values. As just one example, Deaf parents who choose to use genetic testing to screen *for* genes associated with Deafness, do so as part of a broader attempt to preserve Deaf culture and community. Is this comparable, or not, to the attempts of parents without disabilities to ensure a child with desired traits of appearance and talent? The latter attempts have been critiqued by disability advocates as avoiding the parental ideal of unconditionally loving and supporting the child that is born, irrespective of that child's characteristics. Does a collective goal rather than an individual one alter the nature of how genetic testing is being used?

## Changing Hearts and Minds

The disability community underwent critical lessons in building and maintaining unity during the battle to enact the ADA. Similar concerns will doubtless arise when seeking regulatory and legislative support in the area of genetic testing. Yet unity among the community is one of the

most, if not the most, effective means of not only achieving the support of state and federal lawmakers, but also reaching the “hearts and minds” of the public. Unity will also be a significant challenge for the community when it comes to genetic testing, which by its very nature identifies and evaluates the “risk” of particular genetic conditions. Efforts to agree on the kinds of health conditions that preimplantation genetic diagnosis (PGD) can be used to detect have been challenging for the over three decades since PGD was first used on

humans. The ease and wide availability of NIPS, especially as technology—and remuneration—for NIPS improves and genetic sequencing and polygenic risk scoring is added to chromosomal analyses, creates even more pressure to establish a “list” of

diagnoses for which genetic testing can be used, and by implication, a list of conditions for which genetic testing *cannot* be used.<sup>223</sup> One way or another, lines are being drawn all the time in the evolving world of genetic testing. This reality makes disability community unity essential, but such participation can also swiftly be co-opted when disability rights arguments are swallowed up by patient advocacy and promises of medical

cure. Ultimately, there is no need for a showdown between those who would advocate for broader genetic treatment of disability and those who would advocate for conditions to be placed on genetic testing. In both arguments, the lived experience and narratives of people with disabilities must help lead the way, not the latest technology.<sup>224</sup>

Each generation has its own counselors to educate, its own genetic, medical, and research professionals, disability scholars and advocates,

plus new entrepreneurs and corporate entities who enter an essentially unregulated space. At the heart of the disability community’s critique of genetic testing is the same critique that lies behind many, if not all, of the bioethics papers collected by NCD: the failure of the nondisabled

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world to accept disability as a natural part of life that does not and should not determine the lives of people with disabilities. The problem is that the widespread use of genetic testing, and the rapid development of further technologies such as human germline editing, may lead to such significant losses within disability communities that these truths will never reach the tipping point for broader acceptance.

## Case Study

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Like so many best practices, Lettercase's origins arise out of one woman's experience. Stephanie Meredith was 23 years old when she gave birth to a son with Down syndrome. Her son was diagnosed postnatally and she recalls getting good support at a progressive hospital where another mother of an older child with DS came in and showed her a picture of her child on a bicycle. It was seemingly a small thing but a significant turning point because it showed her and her husband that family life could still be *fun*.

After becoming involved with DS support groups, she read a study in 2006 that indicated women were not getting the support they needed after a prenatal diagnosis of DS. Shortly thereafter, ACOG guidelines were modified to indicate that all women should be offered prenatal screening. Stephanie and other advocates realized that if all women could receive screening, then they should also all be able to get good peer-reviewed medical and social information about DS. She quickly realized there was a dearth of such information. Fortunately, Meredith is a technical writer, and her husband a photographer. Pictures were a critical component

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of the materials they developed for women who received prenatal genetic testing. They wanted to show natural glimpses of life with a child with DS, people of different races and ethnicities from different walks of life, engaged in typical family activities. Once a draft was put together, the materials were run past a "consensus group" put together by a University Center for Excellence in Developmental Disabilities (UCEDD) that included representatives from professional organizations such as ACOG, as well as DS advocates,

practicing healthcare providers, and so forth. The group's feedback was incorporated and then the materials were made available.

Lettercase initially tried to continue a nonprofit but it wasn't

sustainable, and eventually the organization went under the umbrella of the Kentucky UCEDD. Over time, other genetic condition advocacy groups have formed partnerships with Lettercase, and the underlying consensus group has also evolved to include representatives with other genetic conditions, a representative from the American Academy of Pediatricians, a UCEDD representative, and so forth. Meredith is proud of the material template that they currently use.

They include family life outcomes, research, health outcomes, employment prospects, and go a bit into the adult years. For guidance, they use a published study that compared the information that parents said they wanted after receiving a prenatal diagnosis of a genetic condition and the information that providers think parents want. Lettercase tries to provide the information that *both* groups think is important, and include perspectives from parents and providers, as well as disability advocates.

Katie Stoll, a genetic counselor in Washington State, connected with Stephanie Meredith around 2011 because of Stoll's own interest in developing additional materials related to living with a disability. She recalls getting

opposition from other counselors when she discussed the inclusion of pictures of people with DS living their daily lives. Some counselors thought women would be unduly pressured if they were shown pictures of "cute babies with DS." Others just wanted to use pictures of chromosomes, as they had in the past. Pictures of chromosomes may help explain the genetic abnormality, but it doesn't help to explain what a child with that genetic condition could be like. When asked whether it is difficult to get good information about disability, Katie replied that "it is really challenging to build the needed bridges to people who are actually living with the particular conditions. As a consequence, it's hard to really know and advise on the condition." She personally had a time when she worked with Stephanie and recalls that "I used to call her up to ask real questions about raising a child

with Down syndrome, things involving care, reactions, learning. It's also hard to be Stephanie, the one giving advice and doing it from a position of personal vulnerability, knowing as well that another woman is facing a decision to potentially terminate a fetus and understanding the need to support the woman."

When Meredith was asked about her biggest challenges, she named two; keeping up with the rapidly expanding list of genetic conditions for which commercial laboratories are offering tests, and disseminating materials once they

are written. Of course, the underlying problem to both of these challenges is sufficient funding. The laboratories themselves occasionally offer "sponsorships" of a particular affected

genetic condition advocacy group, to help that group develop educational materials, but once the draft is completed, the sponsoring lab can ask to review it and there can be the attendant pressure to modify or soften some of the language in accordance with what the sponsor requests. Meredith felt that "some labs have been helpful to work with, but for the most part, they have no interest in putting their money into comprehensive education programs about living with genetic conditions. That isn't their priority." She also doesn't mind commercial entities being involved as one stakeholder among many in the drafting, but it must, ultimately, be a balanced group of all stakeholders that reviews materials. Even when laboratories try to be objective, they can still be misleading, "for instance saying a test has 99% sensitivity does **not** mean that the test has 99% predictive value."

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*Pictures of chromosomes may help explain the genetic abnormality, but it doesn't help to explain what a child with that genetic condition could be like.*



In terms of dissemination, Lettercase has a good relationship with all the professional associations, but it is nonetheless difficult to get word out to individual providers, even when material is free. The small number of genetic counselors makes it possible to reach most of them, but that isn't the case for other specialists. Translation and having the proper cultural lens for educational materials is also a part of the dissemination challenge. Translation is not naturally built into the budget. She just has to go out and get donations, or possible grant monies. Grants are also part of what sustains the work of developing materials for genetic conditions other than DS.<sup>225</sup> As a result, the prioritization of which conditions get worked on next may depend less on the public need for information on that condition or the number of people affected, and more on how work on a particular condition can best meet the conditions of the grant.

Lettercase has an excellent website (<https://www.lettercase.org/>) and provides current evidence-based information about the actual experiences of people and families living with specific genetic conditions, general developmental milestones and common health concerns, available social supports and services, and further resources. Information is provided

through text and photographs and directed at different stakeholders. It has clearly been a labor of love for Meredith and her family, who also built strong relationships with other professionals such as Katie Stoll, advocates, and ultimately the Kentucky UCEDD. But sustained, effective, and *replicable* advocacy cannot depend wholly on the determination of individuals, on the fortuitous forming of professional and personal relationships, and on funding and sponsorship that shows up when it is needed. When considering the funds, marketing power, and

organized lobbying effort of the genetic testing industry in comparison with the DS advocacy community, it's hard not to think about David and Goliath. There is a clear imbalance of funding and outreach capacity, and

this is in regard to what is probably one of the biggest genetic condition advocacy organizations. Smaller advocacy groups who rely on parent volunteers to get out information about rarer genetic conditions have little opportunity to be heard. Numerous authors have discussed the need for varying degrees of federal regulation over genetic engineering,<sup>226</sup> and genetic testing is no different in its potential to negatively impact the disability community and leave parents with *less* capacity for making informed reproductive choices.

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*Even when laboratories try to be objective, they can still be misleading, "for instance saying a test has 99% sensitivity does not mean that the test has 99% predictive value."*



## Conclusion

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Many would consider having and raising a child as one of the most hopeful acts in which humans can engage.

Childbearing is an act of hope not only because of the dreams parents hold for their children, but because from conception onwards, so much of the process is out of the hands of individuals, families and societies. We cannot fully predetermine how a particular child, or any child, will turn out. Every parent just tries their best, and we have no choice

but to hope. Genetic testing in itself, and even the genetic editing potential that has arisen with evolving clustered regularly interspaced short palindromic repeats (CRISPR)

technology, has not changed the fundamental truth that we cannot predetermine how our children will turn out. Nonetheless, genetic testing is deeply impacting what we understand as every parent's "best."

We have seen a repeating pattern with gene technologies: technological breakthrough, we get closer to genetic engineering, we worry about the implications, someone "goes rogue" and performs a new genetic operation on a

human being or beings. There is outrage and discussion and renewed attempts to draw some kind of ethical guideline in the sand. And then the pattern repeats all over again when, not if, another scientist makes another breakthrough or goes rogue. It's fair to say that as this pattern continues, we are all—internationally and in the United States—coming closer to editing out the genetic components that result in a wide range of disabilities. In the process, we

are also likely editing out DNA elements that may control or influence a myriad of characteristics that could potentially be critical to human survival. For all our technological sophistication, the

human race seems no closer to finding ways to equalize the position of people with disabilities, as well as other medically underserved groups, within the ethical, social, and legal debate around these technologies. There is also the contradiction inherent in wielding choice for women as the motivation for increasing genetic testing, but failing to support and implement policies that would achieve equal opportunities for employment, meet long-term care needs,

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*Genetic testing in itself, and even the genetic editing potential that has arisen . . . has not changed the fundamental truth that we cannot predetermine how our children will turn out.*

and ensure effective healthcare to people with disabilities, as well as increase support for families of children with disabilities.<sup>227</sup> These latter actions would give women greater choice to continue a pregnancy of a fetus with disabilities. Choice involves making private

decisions among equally viable alternatives, which is not the case if pregnant women are simply expected to assent to genetic testing, and are potentially judged for failing to terminate a pregnancy that implicates greater healthcare needs and costs.

# Recommendations

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## **Congress**

- Develop enforceable Sunshine and Conflict-of-Interest laws that will bring transparency to any financial relationships between genetic counselors, providers, and commercial laboratories.
- Fund the Prenatally and Postnatally Diagnosed Conditions Awareness Act, Pub. L. No. 110-374, 122 Stat. 4051 (2008).
- Incentivize the development of educational units on disability experience and exposure in genetic counselor education.

## **Department of Health and Human Services (HHS)**

### **HHS, National Institutes of Health**

- Establish standing relationships with disability advocacy organizations and include individuals from them on genetic advisory panels.
- Encourage the attendance of advocates and representatives from disability communities at biomedical conferences by offering scholarships that reduce or cover fees and expenses.

### **HHS, Centers for Medicare and Medicaid Services**

- Recognize genetic counselors as health professionals who can receive reimbursement through Medicare and incentivize Medicaid payments for genetic counseling as an independent healthcare service rather than only reimbursing genetic testing.

*(continued)*

## **Department of Health and Human Services (HHS), *continued***

### **HHS, Food and Drug Administration**

- End enforcement discretion and regulate LDTs, specifically, Noninvasive Prenatal Screening (NIPS) to establish and enforce standards for the accuracy of any claims made by prenatal genetic testing entities, and proactively work with the Federal Trade Commission to oversee marketing by genetic testing entities.

### **Federal Trade Commission**

- Actively oversee the marketing claims and practices of prenatal genetic testing companies as more tests with questionable clinical validity and utility enter the market as part of the “standard” testing panels that companies offer.

### **Equal Employment Opportunity Commission**

- EEOC should leave wellness rules as they are now (May 2019) or, if EEOC does revise them, the agency should clarify that no financial incentives or penalties are permitted to induce employees to disclose health and genetic information.

### **State Legislatures**

- If genetic testing, and especially NIPS, is funded as a Medicaid service, the state should also ensure Medicaid funding for neutral genetic counseling *before* and after testing takes place.
- Where state Medicaid programs cover prenatal genetic testing, the state should ensure that it collects voluntarily provided information on patient demographics, including disability status, outcomes, and the quality of genetic counseling received before the testing, if any.

*(continued)*

## **State Legislatures, *continued***

This information will allow states and researchers to assess the use and results of prenatal genetic testing as a publicly insured service over time.

- Consider enacting legislation, like that pending in California, that clarifies that no financial inducements are allowed for participating in or providing data to a workplace wellness program.

## **Professional Organizations, and Training Accreditation Bodies of Healthcare Providers Engaged in Genetic Counseling, such as the Genetics Society of America (GSA); American College of Medical Genetics (ACMG); American Board of Medical Genetics (ABMG); American Board of Genetic Counselors (ABGC); and the Association of Professors of Human and Medical Genetics (APHMG)**

- Clarify that disability education and cultural awareness extends beyond examining best practices for effectively communicating with patients with disabilities and includes a social and civil rights context for understanding disability.
- Ensure that the materials used for provider and patient education are passed through a consensus group of reimbursed stakeholders, including representatives from affected disability communities, to minimize the outsized influence of industry and investors in prenatal genetic testing.
- Professional standards of care for offering NIPS and other prenatal genetic tests should be established through consensus negotiations that include genetic counselors, obstetrics and gynecology care providers, and representatives from affected disability communities. Genetic testing entities should not be allowed to market or provide specific genetic tests that have not been vetted through a professional organization using a consensus process.
- Ensure that online and printed materials used for provider and patient education are fully communication accessible to people with a range of disabilities and diverse linguistic and cultural backgrounds.
- The Accreditation Council for Genetic Counselling (ACGC) must make disability education and cultural awareness mandatory and more consistent among genetic counselor programs, within a reasonable range of time and resources. The same holds true of professional ongoing education.

## **Genetic Testing Researchers**

- Propose ways to achieve better data over time to determine the link between prenatal testing outcomes and various factors in the field of genetic testing such as counseling, cultural conditions, social expectations, and social determinants of health for particular disability communities.
- Research the relationship between women's choices after receiving pre-test counseling and after undergoing genetic testing, and how choices are affected by the kinds of genetic counseling information provided, who delivers it, and who is paying for the counseling.



## Endnotes

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- 1 ACOG offers a “Frequently Asked Questions” document on prenatal genetic diagnostic tests that provides a glossary with succinct definitions for common scientific terms used in genetic testing at <https://www.acog.org/Patients/FAQs/Prenatal-Genetic-Diagnostic-Tests>.
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- 9 See, for example, Erik Parens, Paul Appelbaum, and Wendy Chung, “Embryo Editing for Higher IQ Is a Fantasy. Embryo Profiling for It Is Almost Here,” *STAT*, February 12, 2019, <https://www.statnews.com/2019/02/12/embryo-profiling-iq-almost-here>; Usha Lee McFarling, “Bad Press About Genetic Engineering Could Scare Off Those Who Would Benefit Most,” *Los Angeles Times*, March 6, 2019, <https://www.latimes.com/opinion/op-ed/la-oe-mcfarling-genetic-engineering-dna-sickle-cell-20190306-story.html>.
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  - 15 Parens and Asch, “The Disability Rights Critique,” S4.
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