
PRENATAL DIAGNOSIS: AN ETHICAL AND A REGULATORY DILEMMA

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“[W]e are compelled to drive toward total knowledge, right down to the levels of the neuron and gene. When we have progressed enough to explain ourselves in these mechanistic terms . . . the result might be hard to accept.”¹ - *Edward O. Wilson*

I. INTRODUCTION

With recent advances in medical technology allowing for non-invasive prenatal diagnosis through a simple blood test, the benefits and ethical concerns that come with prenatal diagnosis (PND) are likely to soon be at the forefront of health law.² Today, PND consists of a sometimes-risky procedure to determine whether the fetus has any chromosomal abnormalities, such as Down syndrome. But in the near future, physicians will be able to test for a huge variety of genetic variants, many of which are not thought to increase the probability of developing a serious disability. This comment examines the current state of the law and regulation of prenatal genetic testing, including wrongful birth and wrongful life claims, and how certain statutes provide some limited coverage of issues involved in prenatal genetic testing. Finally, this comment will analyze what decisions our society must make regarding PND to ensure that it reaches its potential of improving human life while avoiding some very serious potential

¹ EDWARD O. WILSON, *SOCIOBIOLOGY: THE NEW SYNTHESIS* 575 (Harvard Univ. Press 1975).

² Henry T. Greely, *Get Ready for the Flood of Fetal Gene Screening*, 469 *NATURE* 289, 289 (2011).

abuses by calling for comprehensive federal regulation of prenatal genetic testing.

II. PRENATAL DIAGNOSIS – FROM PAST TO PRESENT

Since 1956, we have had the technology to prenatally diagnose certain genetic disorders.³ A large number of single-gene mutations can currently be tested for, in addition to a variety of chromosomal abnormalities, through prenatal diagnosis.⁴ While some parents use prenatal diagnosis to prepare to provide for a child with a genetic disease or disability, the majority appear to use prenatal diagnosis in order to determine whether to terminate the pregnancy due to the presence of a disability.⁵

Although the technology for prenatal screening and diagnosis has been around for quite some time and has advanced significantly since its inception, less than two percent of pregnant women in the United States undergo prenatal diagnosis each year.⁶ There are probably multiple reasons for the lack of widespread testing, but the main one seems to be the risk of miscarriage due to the invasive nature of the tests.⁷ Because of the inadequacies of the current methods, much research has been done to identify non-invasive methods of prenatal diagnosis, but until recently, no serious breakthroughs had been made. However, very recently several papers have been published describing a non-invasive method in which a small amount of the woman's blood is drawn and fetal DNA is isolated then sequenced with the potential to analyze any number of genetic traits.⁸ If and when this technique makes its way into clinics, which some in the field estimate will occur in less than five years, we are likely to

³ See Fritz Fuchs & Povl Riis, *Antenatal Sex Determination*, 177 NATURE 330 (1956).

⁴ See Wylie Burke, *Genetic Testing*, 347 NEW ENGLAND J. MED. 1867 (2002).

⁵ See Nancy Press & C.H. Browner, *Why Women Say Yes to Prenatal Diagnosis*, 45 SOC. SCI. & MED. 979, 979 (1997).

⁶ Greely, *supra* note 2, at 289.

⁷ *Id.*

⁸ See generally Rossa W. K. Chiu et al., *Non-Invasive Prenatal Assessment of Trisomy 21 by Multiplexed Maternal Plasma DNA Sequencing: Large Scale Validity Study*, 342 BRITISH MED. J. 7401 (2011).

see a dramatic increase in the number of women who undergo prenatal diagnosis.⁹ In this section, I will first briefly describe the current invasive techniques used for prenatal diagnosis and their drawbacks, and then I will discuss the advantages and potential of the newly devised non-invasive methods.

A. Current Prenatal Testing Technology

In order to determine whether a fetus is at risk for diseases involving chromosomal defects, maternal age, ultrasonographic examination of the fetus, and biochemical measurement of specific proteins or hormones in maternal circulation are used.¹⁰ However, the false-positive rates of the current screening programs are about five percent, and in order to obtain a more definitive diagnosis, fetal cells must be obtained through amniocentesis or chorionic villus sampling (CVS) and then analyzed.¹¹ The choice between CVS and amniocentesis depends on several factors, including the likelihood to proceed to selective feticide and the gestation at presentation.¹²

1. Amniocentesis

Amniocentesis is an invasive method of prenatal diagnosis, which requires entry into the mother's uterus.¹³ This procedure, typically performed during the second trimester, involves a needle puncture through the skin into the uterus and amniotic cavity so that amniotic fluid can be withdrawn.¹⁴ These cells are then stained in the

⁹ Greely, *supra* note 2, at 290.

¹⁰ Zarko Alfirevic et al., *Amniocentesis and Chorionic Villus Sampling for Prenatal Diagnosis*, COCHRANE DATABASE OF SYSTEMATIC REV., 2009, at 1, 2–3.

¹¹ *Id.*

¹² A. Antsaklis et al., *Second-Trimester Amniocentesis vs. Chorionic Villus Sampling for Prenatal Diagnosis in Multiple Gestations*, 20 *ULTRASOUND IN OBSTETRICS & GYNECOLOGY* 476, 478 (2002) (explaining that the main factor is gestation at presentation, with CVS almost never being used after the first trimester due to increased risks associated—CVS is also more likely to be used in the first trimester if the screening presents a relatively definitive positive diagnosis for a severe condition).

¹³ Zarko Alfirevic et al., *Amniocentesis and Chorionic Villus Sampling for Prenatal Diagnosis*, COCHRANE DATABASE SYSTEMATIC REV., 2009, at 1–3.

¹⁴ *Id.* at 2; Antsaklis et al., *supra* note 12, at 476, 477.

laboratory and analyzed for chromosomal defects, a process that takes several weeks to perform.¹⁵ After amniocentesis is performed, the rate of miscarriage is approximately 0.05%.¹⁶ There is also the possibility of damage to the fetus without causing miscarriage and the potential for maternal infection.¹⁷

2. Chorionic Villus Sampling

CVS is another invasive procedure in which placental tissue is obtained either vaginally or through the abdominal wall.¹⁸ The procedure can be performed during the first trimester, much earlier than is possible for amniocentesis.¹⁹ The chromosome analysis can also be performed immediately after fetal cells are harvested, unlike amniocentesis.²⁰ This procedure is potentially more risky than amniocentesis; the rate of miscarriage is approximately one percent.²¹ Because of the risk of miscarriage that both CVS and amniocentesis entail, few women, except for those with a high risk of having a child with a chromosomal abnormality or genetic disorder, elect to have these procedures performed.²²

¹⁵ NAT'L DOWN SYNDROME CONG., *Position Statement on Prenatal Screening and Diagnosis*, http://ndsccenter.org/worpsite/wp-content/uploads/2012/02/Prenatal_Screening_Diagnosis.pdf (last visited Apr. 2013). The National Down Syndrome Congress is a professional organization that advocates for and supports individuals with Down syndrome. They do not typically issue recommendations on the use of reproductive medicine.

¹⁶ *Id.*

¹⁷ *Id.*

¹⁸ *Id.*

¹⁹ *Id.*

²⁰ *Id.*

²¹ *Id.* It is more difficult to perform early-term CVS than second-trimester amniocentesis, but when performed by a highly experienced practitioner, the risk of miscarriage was close to equivalent for CVS and amniocentesis, contrary to most other studies. See Antsaklis et al., *supra* note 12, at 479.

²² Peter A. Benn & Audrey R. Chapman, *Practical and Ethical Considerations of Noninvasive Prenatal Diagnosis*, 301 J. AM. MED. ASS'N 2154, 2154 (2009); Women who screen positive through ultrasound and blood testing, women with advanced maternal age, and women with hereditary genetic disorders are typically fully counseled about prenatal diagnosis. See Antsaklis et al., *supra* note 12, at 477-78.

B. Cell-free Fetal DNA Isolation and Sequencing

Fortunately, the days of invasive testing may soon be coming to an end. The discovery of cell-free fetal DNA in maternal plasma, comprising the entire fetal genome, has opened up new possibilities for non-invasive prenatal diagnosis.²³ Several papers by two different research groups have been published recently describing techniques in which a single five-to-ten milliliter sample of maternal blood could be used to diagnose not only chromosomal abnormalities like Trisomy 21 (Down syndrome) but also mutations causing diseases such as β -thalassemia.²⁴ These new techniques have come about primarily due to the advent and advancement of massively parallel sequencing technology,²⁵ which can “identify and quantify millions of DNA fragments in biological samples in a span of days.”²⁶

In order to test for the chromosomal abnormality that causes Down Syndrome, the Chui lab used massively parallel sequencing to assay for the presence of elevated levels of chromosome 21 sequences in maternal blood because there are three rather than two copies of fetal chromosome 21.²⁷ Using this technique, Down syndrome could be ruled out in ninety-eight percent of pregnancies. This number is likely to increase as the technology improves and the cost of sequencing is reduced.

The Lo lab employed a slightly different and more complicated technique that allows for the detection of mutations and the inheritance of specific alleles in the fetal genome by comparing isolated fetal DNA fragments to the maternal and paternal genomes using massively parallel sequencing.²⁸ Because the entire fetal genome is present in maternal plasma in the form of DNA fragments, a genome-

²³ See Yuk-Ming Dennis Lo et al., *Maternal Plasma DNA Sequencing Reveals the Genome-Wide Genetic and Mutational Profile of the Fetus*, 2 SCI. & TRANSLATIONAL MED. 1 (2010).

²⁴ See, e.g., *Id.*; Rossa W. K. Chiu et al., *Non-Invasive Prenatal Assessment of Trisomy 21 by Multiplexed Maternal Plasma DNA Sequencing: Large Scale Validity Study*, 342 BRITISH MED. J. 7401 (2011), available at <http://www.ncbi.nlm.nih.gov/pmc/articles/PMC3019239/pdf/bmj.c7401.pdf>.

²⁵ See, e.g., Lo et al., *supra* note 23; Chiu et al., *supra* note 24.

²⁶ Chiu et al., *supra* note 24, at 2.

²⁷ *Id.*

²⁸ Lo et al., *supra* note 23, at 91.

wide scan for diagnosing fetal genetic disorders, or any genetic trait for that matter, could potentially be performed.²⁹

With these non-invasive tests becoming available in the near future, we should expect to see far more pregnant women electing to have prenatal diagnosis performed. Because many women choose to terminate their pregnancy after a positive diagnosis, widespread use of these technologies raises a host of ethical, legal, and social issues.³⁰ As a society, we need to be prepared to determine the best way to provide genetic counseling, informed consent, which traits and disorders may be tested for, and equity issues for what promises to start as a relatively expensive test.³¹

III. REGULATORY FRAMEWORK OF PRENATAL DIAGNOSIS

A. Regulation by Professional Organizations

Currently in the U.S. there are no uniform standards regulating the use of prenatal genetic testing. The federal government does not have direct jurisdiction over the practice of medicine, but thus far, no state has passed laws directly addressing prenatal genetic testing.³²

Several federal agencies have limited oversight of prenatal screening and diagnosis, including the Centers for Medicare and Medicaid Services (CMS), the Food and Drug Administration (FDA), and the CDC.³³ The CMS administers the Clinical Laboratory Improvement Amendments of 1988 to the FDCA (CLIA), which generally apply to clinical laboratories and set standards and testing for laboratory proficiency, personnel, and equipment.³⁴ However, CLIA

²⁹ *Id.*

³⁰ P. R. Reilly, *Commentary: The Federal 'Prenatally and Postnatally Diagnosed Conditions Awareness Act'*, 29 *PRENATAL DIAGNOSIS* 829, 831 (2009) (Currently, ninety percent of children prenatally diagnosed with Down syndrome are aborted).

³¹ Lo et al., *supra* note 23, at 9-10.

³² THE GENETICS AND PUBLIC POLICY CENTER, *Reproductive Genetic Testing* (2010), http://www.dnapolicy.org/policy.international.php?action=detail&laws_id=63 (last visited Jan. 2012).

³³ *Id.*

³⁴ Michael J. Malinowski, *Choosing the Genetic Makeup of our Children: Our Eugenics Past—*

does not provide any assurance of utility from a medical point of view, and CLIA has had significant reporting and enforcement deficiencies.³⁵ Furthermore, CMS has taken the position that laboratories that perform prenatal genetic testing are not considered “clinical laboratories” under CLIA.³⁶

The FDA regulates products used to diagnose a disease or condition, including tests sold as in vitro diagnostic devices.³⁷ However, the majority of tests used in prenatal diagnosis are developed by the genetic laboratories themselves and are, therefore, unregulated by the FDA.³⁸ The FDA does have the authority to regulate the safety and effectiveness of “biological products,” which include tissues that are manipulated or used in a manner different from how they function in the human body.³⁹ However, thus far, the FDA has opted not to regulate tissues used in prenatal genetic testing, and it is unclear whether they would have such authority.⁴⁰ Finally, the FDA does not have authority to regulate the practice of medicine; thus, physicians have significant discretion when using FDA-regulated products.⁴¹

In most cases, standards are set by guidelines established by professional organizations or by the healthcare practitioners and clinics that are directly responsible for providing these services.⁴²

1. American Congress of Obstetricians and Gynecologists

The American Congress of Obstetricians and Gynecologists (ACOG) is one such professional organization. In 2007, the ACOG released a set of “Clinical Management Guidelines” on screening for

Present, and Future?, 36 CONN. L. REV. 125, 184 (2003).

³⁵ *Id.*

³⁶ Susanah Baruch, *Preimplantation Genetic Diagnosis and Parental Preferences: Beyond Deadly Disease*, 8 HOUS. J. HEALTH L. & POL’Y 245, 263-64 (2008) (“CMS has taken the position that [prenatal genetic diagnosis] . . . is an assessment of a product and therefore falls under FDA’s oversight of reproductive tissue”)

³⁷ *Id.* at 262-63.

³⁸ THE GENETICS AND PUBLIC POLICY CENTER, *supra* note 32.

³⁹ Baruch, *supra* note 36, at 263.

⁴⁰ *Id.*

⁴¹ THE GENETICS AND PUBLIC POLICY CENTER, *supra* note 32.

⁴² *Id.*

fetal chromosomal abnormalities, making a number of recommendations concerning the appropriate standard of care.⁴³ Among other things, the ACOG recommends that “[s]creening and invasive diagnostic testing for aneuploidy [a chromosomal abnormality] should be available to all women who present for prenatal care before 20 weeks gestation regardless of maternal age. Women should be counseled regarding the differences between screening and invasive diagnostic testing.”⁴⁴ The ACOG also states that “[s]pecific training, standardization, use of appropriate ultrasound equipment, and ongoing quality assessment are important to achieve optimal . . . [d]own syndrome risk assessment, and this procedure should be limited to centers and individuals meeting these criteria.”⁴⁵ Their proposed performance measure is the “[p]ercentage of patients with documentation of discussion regarding Down syndrome screening.”⁴⁶ Again, these guidelines are by no means binding, and healthcare professionals must voluntarily opt to adopt these criteria.

2. *National Down Syndrome Congress*

Another professional organization, the National Down Syndrome Congress (NDSC), takes a much different position on prenatal screening and diagnosis.⁴⁷ In contrast to the ACOG’s position, the NDSC recommends that “[u]nless specifically requested by a pregnant woman or expectant parents, Obstetricians are not justified in rendering any opinion regarding the ‘potential value’ of the life that has been created.”⁴⁸ The NDSC believes that the ACOG guidelines constitute a form of discrimination against those with disabilities, specifically against those with Down syndrome, by targeting those with Down syndrome while making “only cursory mention of other

⁴³ ACOG Comm. on Practice Bulletins, Clinical Management Guidelines for Obstetrician-Gynecologists: *Screening for Fetal Chromosome Abnormalities*, ACOG PRAC. BULL. (ACOG), Jan. 2007.

⁴⁴ *Id.*

⁴⁵ *Id.*

⁴⁶ *Id.*

⁴⁷ THE NAT’L DOWN SYNDROME CONG., *supra* note 15.

⁴⁸ Such as “other autosomal or sex chromosome anomalies, large deletions or duplications, and chromosomal mosaicism.” *Id.* at 7.

detectable chromosomal conditions.”⁴⁹ The NDSC takes the position that the recommendation for early and thorough prenatal genetic testing for Down syndrome encourages feticide of those with the disability and that this encouragement “blatantly contributes to the devaluation of life in fetuses with chromosomal anomalies including trisomy 21.”⁵⁰

To combat this perceived discrimination, the NDSC has made a number of its own recommendations.⁵¹ The NDSC recommendations revolve around education and support for those considering prenatal genetic testing, especially for those who have tested positive for Down syndrome. For example, the NDSC desires to “[i]mprove the regulation of informed consent and disclosure of information regarding prenatal screening and diagnostic testing for all pregnant women” and to “[e]nhance training about Down syndrome for Genetic counselors, Obstetricians, Pediatricians, and students in training.”⁵² It is clear that these very different opinions on prenatal screening and diagnosis ethics lead to a lack of uniformity in the standard of care among healthcare professionals administering these tests.

B. Statutory Regulation of PND

1. The Prenatally and Postnatally Diagnosed Conditions Awareness Act

Although there is little federal regulation of prenatal genetic testing, one law has been passed by the U.S. government directly addressing the issue. In 2008, President Bush signed the Prenatally and Postnatally Diagnosed Conditions Awareness Act into law.⁵³ The Act focuses on post-diagnosis treatment and education, and it is likely meant to discourage feticide after a positive diagnosis.⁵⁴ The Act states its three purposes are to:

⁴⁹ *Id.*

⁵⁰ *Id.*

⁵¹ *Id.* at 1.

⁵² *Id.*

⁵³ Prenatally and Postnatally Diagnosed Conditions Awareness Act, Pub. L. No. 110-374, 122 Stat. 4051 (2008).

⁵⁴ *Id.*

(1) increase patient referrals to providers of key support services for women who have received a positive diagnosis for Down syndrome, or other prenatally or postnatally diagnosed conditions, as well as to provide up-to-date information on the range of outcomes for individuals living with the diagnosed condition, including physical, developmental, educational, and psychosocial outcomes; (2) 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; and (3) ensure that patients receive up-to-date, evidence-based information about the accuracy of the test.⁵⁵

In order to fulfill these purposes, the Secretary of Health and Human Services (HHS) may “oversee certain activities, including the awarding of grants, contracts or cooperative agreements to eligible entities” to undertake certain activities, including the dissemination of evidence-based information about Down syndrome and other prenatally or postnatally diagnosed conditions and to provide support and education to parents who receive a positive diagnosis.⁵⁶ It is easy to see the parallels between this Act and the position taken by the NDSC. Indeed, some commentators believe that the NDSC played a major role in bringing Public Law 11-374 into existence.⁵⁷ The lobbying for and passing of this bill all seem to stem from outcry against the ACOG’s recommendation in Practice Bulletin 77 that invasive testing should “be available to all women who present for prenatal care before 20 weeks of gestation regardless of maternal age.”⁵⁸

It seems that the primary intention of Public Law 11-374 is to provide information to parents with a positive diagnosis of Down syndrome (or other prenatally diagnosed condition) about the life experiences of those with said condition, the services available to aid families who have members with said condition, and the availability

⁵⁵ *Id.*

⁵⁶ *Id.*

⁵⁷ Philip R. Reilly, *Commentary: The Federal ‘Prenatally and Postnatally Diagnosed Conditions Awareness Act,’* 29 *PRENATAL DIAGNOSIS* 829, 830 (2009) (“On 13 October 2008, when the National Down Syndrome Congress . . . announced the new law, it did so with a lead claiming, ‘Down Syndrome Advocates Prevail.’”).

⁵⁸ *Id.* at 829.

of adoption services.⁵⁹ When providing background for this Act, the House Republican Conference website states that, “[c]urrently, ninety percent of children prenatally diagnosed with Down syndrome are aborted,” implying that the Act was intended to reduce these numbers through education and support.⁶⁰ Unfortunately, the Secretary would have to take money from other programs to fund this Act because it does not contain an appropriation clause.⁶¹ Because of the budget issues that Congress is currently facing, it seems unlikely that this Act will reach many of its intended targets.⁶² It is not yet clear what effect, if any, the passage of this Act will have on the reproductive decision making of parents expecting a child with a disability and, in particular, one with Down syndrome.

2. *Americans with Disabilities Act*

The Americans with Disabilities Act of 1990 (ADA) and the ADA Amendments Act of 2008 (ADAAA) are statutes that do not directly address prenatal screening and diagnosis, but they have important implications for expecting parents with a positive diagnosis of certain conditions that might be considered “disabilities.”⁶³ The intention of the ADA is “to provide a clear and comprehensive national mandate for the elimination of discrimination against individuals with disabilities.”⁶⁴ Furthermore, “Congress recognized that physical and mental disabilities in no way diminish a person’s right to fully participate in all aspects of society, but that people with physical or mental disabilities are frequently precluded from doing so because of prejudice, antiquated attitudes, or the failure to remove societal and institutional barriers.”⁶⁵ Under the ADAAA, Congress defined a “disability” as

⁵⁹ *Id.*

⁶⁰ *Id.* at 830–31 (citing House Republican Conference, 2008).

⁶¹ *Id.* The bill originally contained an appropriation clause and required all pregnant women who receive prenatal screening and diagnosis to be educated on Down syndrome and other conditions.

⁶² *Id.*

⁶³ See Americans with Disabilities Act of 1990 (ADA), 42 U.S.C. §§ 12101–12213 (2000); ADA Amendments Act of 2008, 42 U.S.C. § 12101 (2008).

⁶⁴ 42 U.S.C. § 12101(b)(1).

⁶⁵ 42 U.S.C. § 12101(2)(a).

“a physical or mental impairment that substantially limits one or more major life activities of such individual; a record of such an impairment; or being regarded as having such an impairment.”⁶⁶ In order to accomplish this, the ADA prohibits disability discrimination in employment through Subchapter I, guarantees access to public services through Subchapter II, and guarantees access to public accommodations through Subchapter III.⁶⁷

One might think that the passage of the ADA would decrease the number of parents who choose to terminate a pregnancy because of a positive test for a disability; however, the opposite trend has been observed.⁶⁸ By 1993, the Down syndrome birth rate “fell by between 13 and 18 per 100,000 relative to the pre-ADA period when controlling for demographic and medical care variables.”⁶⁹ Additionally, this decline “coincided with steady amniocentesis rates.”⁷⁰ There are several theories as to why we see this paradoxical decline after passage of an Act that should help to improve the lives of those with disabilities.⁷¹ It could be that increased access to employment and public accommodations by the disabled led to “interaction strain” between the disabled and non-disabled, causing the non-disabled to form a negative opinion (consciously or subconsciously) of the disabled.⁷² It is also possible that popular media coverage of the ADA’s passage focused on the struggles and hardships of the disabled, which then reinforced an idea that a disabled child would be a burden on prospective parents.⁷³ This is not to say that the passage of the ADA was not a commendable effort, just that it apparently had

⁶⁶ 42 U.S.C. § 12102(1). “[M]ajor life activities include, but are not limited to, caring for oneself, performing manual tasks, seeing, hearing, eating, sleeping, walking, standing, lifting, bending, speaking, breathing, learning, reading, concentrating, thinking, communicating, and working.” 42 U.S.C. § 12102(2)(A).

⁶⁷ See Americans With Disabilities Act of 1990, 42 U.S.C. § 12102–12213 (1990).

⁶⁸ See Dov Fox & Christopher L. Griffin, *Disability-Selective Abortion and the Americans with Disabilities Act*, 3 UTAH L. REV. 845, 871 (2009).

⁶⁹ *Id.*

⁷⁰ *Id.*

⁷¹ See *id.* at 864–66.

⁷² *Id.* at 868–70.

⁷³ *Id.*

unexpected impacts on the rate of pregnancy termination among those with a positive prenatal diagnosis of Down syndrome.⁷⁴

3. *The Genetic Information Nondiscrimination Act*

The Genetic Information Nondiscrimination Act (GINA) is another piece of legislation that does not directly address prenatal genetic testing, but could affect the decision making of parents deciding whether to screen and those with a positive diagnosis for a genetic condition.⁷⁵ Title I prohibits consideration of genetic information by health insurance companies, and Title II prohibits employers or potential employers from considering genetic information in their decisions.⁷⁶ Because genetic information discrimination is not yet happening on a large scale, GINA has been hailed as the “first predominately forward-looking antidiscrimination statute.”⁷⁷

The primary motivation for the passage of GINA seems to be to assuage fears of genetic information discrimination in order to encourage more people to undergo testing.⁷⁸ This could have several implications for prenatal screening and diagnosis. First, parents may be more likely to undergo prenatal diagnosis, especially when it is no longer invasive, if they do not have to worry about their future child being discriminated against because they were tested. Parents who simply want to prepare for life with a child who has a certain genetic condition can rest assured that their decision to test the fetus will not be a reason for health insurance companies to deny coverage or for future employers to deny or alter terms of employment.⁷⁹

Secondly, GINA may cause parents who would otherwise have terminated a pregnancy because of the presence of a certain genetic condition—or a propensity to develop a certain condition (such as BRCA 1 or 2)—to keep the child, knowing that the child will be safe

⁷⁴ See *id.* at 871.

⁷⁵ See Genetic Information Nondiscrimination Act of 2008, Pub. L. No. 110-233, 122 Stat. 881 (codified in scattered sections of 26, 29, and 42 U.S.C.) [*hereinafter* GINA].

⁷⁶ *Id.* at 883, 907.

⁷⁷ Jessica L. Roberts, *The Genetic Information Nondiscrimination Act as an Antidiscrimination Law*, 86 NOTRE DAME L. REV. 597, 600 (2011).

⁷⁸ *Id.* at 603-04.

⁷⁹ GINA, *supra* note 75, at I & II.

from discrimination despite this predisposition or condition.⁸⁰ However, it should be noted that if the child is born with a condition, insurance coverage decisions related to treatment may be subject to decisions by the insurance carrier.⁸¹

It is not yet clear what effect, if any, GINA is having on the rate of termination of pregnancies due to diagnosis of a genetic condition or predisposition,⁸² but parents must be aware of GINA's existence for it to have any impact at all. This is also true of the ADA, and parents considering prenatal genetic testing should be fully informed of the existence of these statutes and their implications for their lives and the potential life of their child.

IV. LIABILITY FOR MALPRACTICE IN PRENATAL SCREENING AND DIAGNOSIS

A. Wrongful Birth and Wrongful Life

When physicians and geneticists commit malpractice by failing to meet the standard of care when performing prenatal genetic testing and the parents either terminate a pregnancy that they otherwise would not have or keep a pregnancy that they otherwise would have terminated, many people believe that there should be some recourse. One such recourse is the highly controversial "wrongful birth" cause of action.⁸³

⁸⁰ Nat'l Cancer Inst. at the Nat'l Insts. of Health, *BRCA1 and BRCA2: Cancer Risk and Genetic Testing*, NAT'L CANCER INST. (May 29, 2009), <http://www.cancer.gov/cancertopics/factsheet/Risk/BRCA>; see GINA, Titles I & II.

⁸¹ Nat'l Coalition for Health Care Educ. in Genetics, *Case 4: GINA and Prenatal Diagnosis*, http://www.nchpeg.org/index.php?option=com_content&view=article&id=187 (last visited Jan. 2012).

⁸² Nancy Press, *Genetic Testing and Screening*, in *FROM BIRTH TO DEATH AND BENCH TO CLINIC: THE HASTINGS CENTER BIOETHICS BRIEFING BOOK FOR JOURNALISTS, POLICYMAKERS, AND CAMPAIGNS*, 73, 74 (Mary Crowley, ed., 2008), available at <http://www.thehastingscenter.org/Publications/BriefingBook/Detail.aspx?id=2176#prenatal>.

⁸³ LORI B. ANDREWS et al., *GENETICS: ETHICS, LAW, AND POLICY* 324 (West Group, 2nd. Ed. 2002). This is a hotly contested issue in some states. In 2005, nine states had prohibited wrongful life/birth actions. Wendy F. Hensel, *The Disabling Impact of Wrongful Birth and Wrongful Life Actions*, 40 *HARVARD CIV. RIGHTS-CIVIL LIBERTIES L. REV.* 150, 162 (2005). Since that time, Oklahoma and Arizona passed similar provisions. Julie Bisbee, *Oklahoma Abortion*

A wrongful birth cause of action exists when physicians fail to warn prospective parents that they are at risk of conceiving or giving birth to a child with a serious genetic disorder.⁸⁴ Parents can also bring a wrongful life cause of action on behalf of the child.⁸⁵ The controversial nature of this cause of action is due to the fact that the parents must argue that they were harmed by the birth of that child due to the presence of the condition in a wrongful birth claim; in a wrongful life claim the child must effectively argue that he or she would be better off having not been born.⁸⁶ Wrongful birth claims are met with more success than wrongful life claims, which almost never succeed because courts are very reluctant to find that a child would value nonexistence over life no matter what the circumstances.⁸⁷ These causes of action have been widely criticized because the birth of a child has never been considered an “injury” in the traditional sense and because they reflect an inaccurate and inappropriate attitude in society toward life with a disability.⁸⁸

Because of these issues with wrongful birth and wrongful life causes of action, there is much uncertainty and a lack of uniformity among courts concerning not only whether the claims should be allowed but also as to what kinds of damages should be awarded.⁸⁹

Bills Become Law, NEWS OK, April 27, 2012, http://newsok.com/oklahoma-abortion-bills-become-law/article/3457167?custom_click=lead_story_title#ixzz0mKGleHeb; *Bill blocks wrongful-life suits in defect cases*, ARIZONA DAILY STAR, Feb. 9, 2012, http://azstarnet.com/news/local/govt-and-politics/bill-blocks-wrongful-life-suits-in-defect-cases/article_7a8bed6d-675a-50d1-ba91-d90a3347940d.html. And most recently, on April 11, 2013, Kansas signed a prohibition into law, and in March 2013, the Montana House has passed a bill that is being considered in the Senate. *Brownback Signs 22 More Bills. Here are the Newest Laws*, SALINA POST, April 11, 2013, <http://salinapost.com/2013/04/11/brownback-signs-22-more-bills-here-are-the-newest-laws/>; *Montana Senate Panel Hears Wrongful Birth/Life Bill*, GREAT FALLS TRIBUNE, March 12, 2013, <http://www.greatfallstribune.com/view-art/20130312/NEWS05/303120006/>.

⁸⁴ See ANDREWS et al., *supra* note 83.

⁸⁵ *Id.*

⁸⁶ *Id.*

⁸⁷ Jeffrey R. Botkins, *Prenatal Diagnosis and the Selection of Children*, 30 FLA. ST. UNIV. L. REV. 265, 273–74 (2003).

⁸⁸ See Bernadette Kennedy, *The Trend Toward Judicial Recognition of Wrongful Life: a Dissenting View*, 31 UCLA L. REV. 473, 474 (1983); Ronen Perry, *It's a Wonderful Life*, 93 CORNELL L. REV. 329, 335 (2008).

⁸⁹ See *Azzolino v. Dingfelder*, 337 S.E.2d 528, 534 (N.C. Sup. Ct. 1985).

To date, the majority of states have recognized a wrongful birth cause of action.⁹⁰ In contrast, eight states have statutorily barred wrongful birth causes of action.⁹¹ But even the states that prohibit wrongful birth actions may allow other causes of action when prenatal screening or diagnosis is performed negligently.⁹²

B. Other Causes of Action in Prenatal Diagnosis

Some courts have recognized other causes of action in place of wrongful birth and wrongful life to avoid the ethical issues associated with them. For example, in *Grubbs ex rel. Grubbs v. Barbourville Family Health Center*, the Kentucky Supreme Court recognized a cause of action for breach of contract when a physician failed to recognize a birth defect during an ultrasound.⁹³ The court held that a physician who undertakes diagnostic or screening procedures has a responsibility to provide an accurate and a non-negligent diagnosis.⁹⁴ Otherwise, physicians could legally charge patients—and be paid—for services they did not perform.⁹⁵

Some other courts may also find a claim for damages from emotional distress, provided the plaintiff can first prove negligence.⁹⁶ However, this cause of action presents many of the same problems as wrongful birth and wrongful life actions because it requires the parents to plead that they were damaged, emotionally no less, by the

⁹⁰ Hensel, *supra* note 83, at 151–52.

⁹¹ See IDAHO CODE § 5-344 (1986); MINN. STAT. § 145.424 (1987); MO. ANN. STAT. § 188.130 (1987); 42 PA. CONS. STAT. ANN. § 8305 (1990); S.D. CODIFIED LAWS 21-55-2 (1987); IND. CODE ANN. § 34-12-1-1. MICH. COMP. LAWS ANN. § 600.2971

⁹² See, e.g., *Sejpal v. Corson, Mitchell, Tomhave & McKinley, M.D.'s, Inc.*, 665 A.2d 1198, 1200–01 (Pa. Super. Ct. 1995) (affirming denial of appellant's claim for wrongful life under 42 PA. CONS. STAT. ANN. §8305(b) but permitting a cause of action for lack of informed consent, in that but for "appellees' prenatal failure to discover" that appellant's child had Down syndrome, appellant would not have undergone sterilization after the birth and would have tried to have another child).

⁹³ *Grubbs ex rel. Grubbs v. Barbourville Family Health Ctr., P.S.C.*, 120 S.W.3d 682, 688–89 (Ky. Sup. Ct. 2003).

⁹⁴ *Id.* at 691.

⁹⁵ *Id.*

⁹⁶ See *Azzolino v. Dingfelder*, 337 S.E.2d 528, 530 (N.C. Sup. Ct. 1984).

birth of their child.

C. Liability After Non-invasive Prenatal Diagnosis

With the commercialization of non-invasive prenatal diagnosis, the number of wrongful birth and wrongful life causes of action filed should only increase. In the absence of future regulation, as we gain a better understanding of what each gene is responsible for, practitioners will begin offering tests for more and more genetic variants. The courts will need to address what exactly constitutes a disability or genetic defect that can give rise to a wrongful birth or wrongful life cause of action. Of course, as we have seen, many courts may choose not to recognize these causes of action at all. As one court stated:

To our ears, at the close of the twentieth century, this talk of the “unfit” and of “defectives” has a decidedly jarring ring; we are, after all, above such lethal nonsense. But are we? . . . [W]hen scientists map the human genome, they will unveil many more potentially harmful genes in each of us Will we then see the tort of wrongful birth extended to physicians who neglect or misinterpret genetic evidence and thereby fail to extend the option of a eugenic abortion to the unsuspecting parents of the genetically “unfit” or “defective” child?⁹⁷

While these issues may seem like science fiction now, they appear to be approaching more rapidly than one might have imagined. The human genome has been mapped,⁹⁸ and scientists are elucidating the function of more genes and genetic variants all the time.⁹⁹ In addition to determining which, if any, genetic variants should be tested for, we will have to decide: 1) whether parents who request prenatal diagnosis should have some recourse when that diagnosis is performed negligently, and 2) exactly what recourse can best prevent

⁹⁷ Grubbs, 120 S.W.3d 682, 690–91 (Ky. Sup. Ct. 2003) (quoting Taylor v. Kurapati, 600 N.W.2d 670, 690 (Mich. Ct. App. 1999)).

⁹⁸ See the Human Genome Project 2003, The 1000 Genomes Project.

⁹⁹ See Michael J. Malinowski, *Choosing the Genetic Makeup of Children: Our Eugenics Past—Present, and Future?*, 36 CONN. L. REV. 125, 172–74 (2003) (The Human Genome Project is a government-funded, \$3 billion project with the goals of assembling a complete map of the human genetic framework and identifying the base pairs of nucleotides—comprised of the building blocks of adenine, thymine, cytonine and guanine—for each gene. The basic genetic map was completed in 2003, identifying 30,000 genes in the human genome. *Id.* at 172; see also NAT'L HUMAN GENOME RESEARCH INST., NAT'L INSTS. OF HEALTH, <http://www.genome.gov/11006943>).

discriminatory attitudes toward the disabled.

V. FUTURE REGULATION PRENATAL GENETIC TESTING

As we have seen, there is no real comprehensive regulation of prenatal genetic testing. With the advent of non-invasive technology, we will likely be forced to confront this issue soon. We will have to decide whether to allow these screenings at all, and if so, whether we should allow screening for any condition or for certain conditions only.¹⁰⁰ We will also have to decide who should regulate these screenings—federal agencies, the states, or professional organizations. We will also have to decide how to fund these screenings so those who can least afford to provide for a disabled child have the option of prenatal genetic testing.¹⁰¹

It is apparent that stricter regulation will be necessary to promote uniformity and efficiency, to ensure that all prospective parents are fully informed in order to make the decision that is best for them, and to prevent the exacerbation of discriminatory and negative associations with the disabled community.

A. Standard of Care

When non-invasive prenatal diagnosis becomes widely available and reasonably affordable, the standard of care will have to adapt to harness the benefits of this new technology. Currently, most women who receive screening are given limited information prior to the screening test and are rarely asked to provide informed consent.¹⁰² Typically, only women found to be at high risk for Down syndrome due to relatively high maternal age are fully counseled about the significance of the tests and the risks associated.¹⁰³

Due to the risks of miscarriage inherent in amniocentesis and CVS and the lack of accuracy of the current screening procedures,

¹⁰⁰ Greely, *supra* note 2, at 289, 291.

¹⁰¹ *Id.* at 290 (mentioning the declining cost of DNA sequencing).

¹⁰² Peter A. Benn & Audrey R. Chapman, *Practical and Ethical Considerations of Noninvasive Prenatal Diagnosis*, 301 J. OF AM. MED. ASS'N. 2154, 2154 (2009).

¹⁰³ *Id.*

cell-free fetal DNA isolation and sequencing is likely to become the standard of care soon after commercialization.¹⁰⁴ Even if the accuracy of this technology does not reach diagnostic levels, it should become the standard of care because it is extremely safe, both for the mother and for the fetus. This standard of care will probably be developed in much the same way they are now, through professional guidelines issued by groups like the ACOG and the NCDS.¹⁰⁵

In order to provide the best care possible, obstetricians and genetic counselors must provide more information to parents considering prenatal genetic screening and those who receive a positive diagnosis so that they can make the best informed decision. Pregnant women should be informed of the existence of GINA so that they do not refuse testing for unjustified fears of genetic discrimination. Those receiving a positive diagnosis should be fully counseled about the implications of the diagnosis and should be given an unbiased perspective on the life experiences of the disabled and what to expect, as is the intention of The Prenatally and Postnatally Diagnosed Conditions Awareness Act. Finally, the significance of the ADA should be explained so they can be reassured that their potential disabled child would have access to the same employment opportunities and public services that those who are not disabled enjoy.

B. Where to Draw the Line

The question of which genetic variances should be tested for, and also which genetic variances can give rise to a wrongful birth cause of action, will need to be answered in developing the standard of care for non-invasive prenatal diagnosis.

Individuals have filed wrongful birth suits for a variety of conditions, ranging from life-threatening conditions like Tay-Sachs disease, to less serious ones like Down syndrome and congenital blindness.¹⁰⁶ At some point, a line will have to be drawn with regard to what conditions are actionable, but it is unclear exactly where that line should

¹⁰⁴ C.J. Chachkin, *What Potent Blood: Non-invasive Prenatal Genetic Diagnosis and the Transformation of Modern Prenatal Care*, 33 AM. J. OF L. & MED. 9, 37-38 (2007).

¹⁰⁵ *Id.* at 33-35.

¹⁰⁶ Hensel, *supra* note 83, at 181-82.

be drawn and who should make that decision.¹⁰⁷

If only certain conditions are to be actionable, then the line should probably be drawn by the testing centers rather than the courts. By allowing only certain genetic variances to be tested legally, it will become clear which missed conditions are actionable if the courts continue to accept wrongful birth causes of action. This would also help to avoid going too far down the path of positive eugenics.

With increasing awareness of female feticide among certain ethnic groups in Asia, which is now apparently becoming an issue in the United States and Canada, the notion that procreative liberty should be curbed in order to promote equality is becoming more common.¹⁰⁸ For example, one commentator recommends postponing “the disclosure of medically irrelevant information to pregnant women until after about 30 weeks of pregnancy—in other words, when an unquestioned abortion is all but impossible.”¹⁰⁹ Assuming we were to use this standard for prenatal genetic testing, the question then becomes: What information is medically relevant?

This is a very difficult question to answer, and is made even more difficult by the fact that the range of functioning among individuals with the same disability can vary dramatically.¹¹⁰ It may be difficult, if not impossible, to predict how badly an individual will suffer from a certain condition, and the manifestation of the condition could be benign enough to not be considered “medically relevant.”

Another argument against creation of such a list is that it would reinforce a message about the undesirability of the listed disabilities.¹¹¹ At least one commentator argues that permitting testing for a greater number of conditions is preferable so that reproductive decisions are left to parents rather than the government, the medical community, and insurance providers.¹¹²

¹⁰⁷ *Id.*

¹⁰⁸ Rajendra Kale, “It’s a Girl” – Could be a Death Sentence, 184 CAN. MED. ASS’N J. 387, 387 (2010).

¹⁰⁹ *Id.*

¹¹⁰ Hensel, *supra* note 83, at 183.

¹¹¹ Adrienne Asch, Disability Equality and Prenatal Testing: Contradictory or Compatible?, 30 FLA. ST. U. L. REV. 315, 339 (2003).

¹¹² *Id.*

Some also argue that a list of genetic variances to be tested for might eventually lead to a society that is less willing to provide aid to the disabled.¹¹³ Although most people currently sympathize with those who have genetic defects and offer compassionate care, will people be as concerned about the disabled in the future when those disabilities are often preventable?¹¹⁴

However, this concern for our society's future outlook on disability must be balanced with the highly valued principles of patient autonomy and procreative liberty.¹¹⁵ And if no list of permissible changes is created at all, what is to stop people from choosing the sex of their child? And when the technology to rewrite the genetic code arrives, allowing the potential to create "designer babies," how will we decide which traits are "medically relevant" enough to justify their altering?¹¹⁶

A line must be drawn somewhere in order to make a compromise between autonomy and procreative liberty with the concerns addressed above. This list could consist of only those genetic variances determined to lead to "serious medical conditions," and it could be based on factors such as the average cost of care and the rate of early mortality. Some families may not be able to afford caring for a child with a serious disability. Care for a child with a disability can cost up to three times as much as care for a child without one. Higher costs correlate strongly with the severity of the disability.¹¹⁷ Certain disabilities might also require more constant care, potentially preventing a parent from working when they otherwise would.¹¹⁸

¹¹³ M.J. Malinowski, *Choosing the Genetic Makeup of Children: Our Eugenics Past – Present, and Future?*, 36 CONN. L. REV. 125, 200–01 (2003).

¹¹⁴ *Id.* at 209.

¹¹⁵ *Id.* at 203–04.

¹¹⁶ Although this technology sounds like science fiction, it may be here sooner than expected. Gene therapy and virotherapy have already been used to replace defective genes with functional ones to treat cancer, SCID, and Parkinson's disease. See generally Donald B. Kohn & Fabio Candotti, *Gene Therapy Fulfilling Its Promise*, 360 NEW ENG. J. MED. 518 (2009).

¹¹⁷ T. Donna Anderson et al., *The Personal Costs of Caring for a Child with a Disability: A Review of the Literature*, 122 PUB. HEALTH REPORTS 3, 8 (2007).

¹¹⁸ Eileen M. Brennan & Ana Maria Brannan, *Participation in the Paid Labor Force by Caregivers of Children with Emotional and Behavioral Disorders*, 13 J. EMOTIONAL BEHAVIORAL DISORDERS 237 (2005).

Finally, certain conditions, such as early onset Tay-Sachs disease, are nearly always fatal at a young age.¹¹⁹ It can be emotionally damaging for parents to experience the death of a child, not to mention the pain that the child must go through.¹²⁰ These reasons justify the use of at least some prenatal genetic testing, but it must be carefully regulated so as to prevent future abuses.

C. How Should Prenatal Genetic Testing Be Regulated?

As we have seen, prenatal genetic diagnosis is loosely regulated by a patchwork of federal and state law, but it is left mostly to professional organizations and to the healthcare professionals themselves to determine how best to administer prenatal genetic testing.

Several commentators have suggested that comprehensive federal regulation is necessary to promote uniformity, to provide patients with the best possible care, and to prevent future abuses.¹²¹ An expansion of agency authority may be appropriate. If the FDA determines that it does not have the authority to regulate cell-free fetal DNA isolation and sequencing as an “in-vitro diagnostic test,” its authority may have to be expanded by Congress.¹²² Likewise, laboratories that perform prenatal genetic screening, which are now “essentially immune to federal laboratory-quality assurances,” should be regulated by the CMS through CLIA.¹²³ A list of severe genetic conditions to be tested for should be enforced, with penalties imposed against parents who receive a negligent diagnosis in order to deter bad practice of medicine and to prevent the need for wrongful birth and wrongful life causes of action. With laws like these in place, a compromise can be made between patient autonomy and the preven-

¹¹⁹ *Tay-Sachs Disease – Topic Overview*, WEBMD.COM, <http://children.webmd.com/tc/tay-sachs-disease-topic-overview> (last visited Apr. 11, 2013).

¹²⁰ Naomi Brower & Kimber Peart, *Coping with the Loss of a Child*, Families and Communities, Utah St. University Cooperative Extension, Sept. 2010, http://extension.usu.edu/files/publications/publication/FC_Family_2010-01pr.pdf.

¹²¹ Baruch, *supra* note 36, at 264–65 (discussing regulation of preimplantation genetic diagnosis, which is regulated in much the same way as prenatal diagnosis); Hensel, *supra* note 83, at 192–93.

¹²² Baruch, *supra* note 36, at 263.

¹²³ Hensel, *supra* note 83, at 192.

tion of abuse of this new technology, all while ensuring respect for the disabled community.

VI. CONCLUSION

Future technology that once seemed impossible is quickly being realized, and with it comes the potential to prevent much pain and suffering. But this technology also brings the responsibility to make sure that it is not abused in a way that may lead to harm for any social group. Prenatal diagnosis will have to be more strictly regulated at some point, and it would benefit us all if the government was proactive, rather than reactive, when enacting this legislation.