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# The Routinization of Prenatal Testing

Sonia Mateu Suter†

*The technology was available, so why not use it? – Words of an expectant father*

By now everyone is familiar with the recent accomplishments of the Human Genome Project. Accomplished in ten, rather than the initially expected fifteen, years, the human genome has been fully sequenced. Genetics is in its golden age. A product of the technology era, genetics has, in a short time, offered vast amounts of information. This increased knowledge promises potential benefits for our understanding of the disease process and, ultimately, treatment and prevention. The rapid flow of information, however, presents complications and challenges. It can complicate the decision making process for those involved in genetic testing, not only because our knowledge of genetics is more complex, but because there is so much more potential information to obtain. The sheer quantity of information for both researcher, clinician and especially patient, can be overwhelming. In addition, our ability to glean predictive or susceptibility information has vastly exceeded our ability to develop cures for diseases; patients can increasingly identify risks for conditions that they can do little to avoid.

The expansion of genetic information has created a new class of genetics patients: the adult at risk for late-onset conditions. Once a field devoted primarily to expectant or future parents, genetic counseling increasingly includes adult genetics. With this growing area have come concerns about the costs of getting genetic information. A woman might learn that she has the BRCA1 gene, but data are still uncertain about precisely what this means in terms of her risk of breast or ovarian cancer and what she can do to reduce her risk. People with a family history of

Huntington Disease (HD) may choose to find out whether they have the gene that virtually guarantees they will develop the disease, if they live long enough. But it is currently impossible to predict the age of onset and no cure or prevention is available.

As a result, anxiety and distress can be associated with finding out one does or does *not* have a late-onset gene.<sup>1</sup> Geneticists and ethicists have therefore urged caution and care in the decision making process. More importantly, many have suggested that knowledge may not always benefit everyone. For some, it can be “toxic.”

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<sup>1</sup> Surprising and unexpected negative emotional reactions have occurred in people who expected to have a late-onset gene and found out they did not. *See infra* text accompanying note 28.

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The circumspection regarding knowledge and genetic testing in the context of adult genetics applies equally well in the reproductive context. Yet, the same level of restraint and caution does not exist. Instead, prenatal genetic testing and screening has become *de rigeur*; nearly as commonplace and widely accepted as some of the more routine aspects of prenatal care. The routinization of prenatal testing<sup>2</sup> has impoverished the informed consent process in many ways; little emphasis is placed on the many emotional and psychological ramifications of undergoing such testing, leaving patients unprepared for certain choices and emotional reactions. This Article argues that the same concerns about informed decision making with respect to genetic testing for late-onset conditions apply to prenatal testing.

Part I outlines the history and evolution of genetic testing. It explores some of the issues that arise with genetic testing for late-onset conditions (late-onset testing) and reviews recommendations that have been made to address these concerns. Part II describes the routinization of prenatal testing and screening and the role that healthcare professionals, patients, society and the legal profession have played in this process. Part III then describes the implications of this routinization and the ways in which it has impoverished the informed consent process. While caution has been exercised to address the psychosocial risks associated with late-onset testing, the field of reproductive genetics today increasingly minimizes the psychosocial risks of prenatal testing. Therefore, patients are often uninformed about the implications of undergoing such testing, which can result in anxiety or decisions inconsistent with their values or preferences. Part III concludes by exploring some of the broader social implications of this routinization, suggesting that in diminishing the informed consent process, it contributes to an inability to discuss the moral dimensions of reproductive technology today and in the future.

## I. A BRIEF HISTORY OF GENETIC TESTING AND COUNSELING

Genetic testing is a new phenomenon. It only became available in the latter part of the 20th century as we learned about the mechanisms of genetics and were able to isolate products of defective genes, chromosomes and ultimately the genes themselves.<sup>3</sup> In the earlier part of the last century, human genetics was a crude, illinformed science. Theories of inheritance were based more on stereotype and ethnic prejudice than on hard science.<sup>4</sup> The pioneer genetic counselors tended to be nonmedical geneticists or non-practicing physicians,<sup>5</sup> focused on societal rather than individual concerns. These geneticists, active in the then-respected eugenics movement, sought to clean the gene pool by urging reproduction among the ostensibly fittest and discouraging it among the putatively genetically inferior.<sup>6</sup> More

a form of social engineering than a science, the eugenics movement successfully lobbied for legislation that prohibited the genetically inferior from reproducing.

2 Much of my discussion of the routinization of prenatal testing could apply to ultrasounds or sonograms. Given that this symposium focuses on genetics issues, this Article concentrates primarily on prenatal *genetic* testing and screening.

3 ASSESSING GENETIC RISKS: IMPLICATIONS FOR HEALTH AND SOCIAL POLICY 30-31

(Lori B.

Andrews et al. eds., 1994) [hereinafter ASSESSING GENETIC RISKS].

4 DOROTHY NELKIN & M. SUSAN LINDEE, *THE DNA MYSTIQUE: THE GENE AS CULTURAL ICON* 19-37 (1995).

5 Ian H. Porter, *Evolution of Genetic Counseling in America*, in *GENETIC COUNSELING* 17, 23

(Herbert A. Lubs & Felix de la Cruz eds., 1977).

6 For example, to promote the eugenic ideal, the American Eugenics Society sponsored nationwide “mental and physical perfection contests” to find the fittest baby or family. NELKIN & LINDEE, *supra* note 4, at 27.

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More than thirty states enacted such laws, which were not only tolerated, but were applauded by most levels of society.<sup>7</sup> Indeed, the U.S. Supreme Court upheld Virginia’s sterilization efforts in the now notorious opinion of *Buck v. Bell*<sup>8</sup> in which Justice Holmes declared, “three generations of imbeciles are enough.”<sup>9</sup>

As the weak scientific foundations for eugenic theories were eventually exposed, many geneticists turned their attention away from human genetics to study less socially controversial subjects, such as plant and animal genetics.<sup>10</sup> Only in the 1950s did scientists return to the study of human genetics, this time with a much more solid scientific foundation and a focus on the molecular basis of disease. In its infancy, molecular genetics used biochemical analyses of proteins to diagnose such

conditions as sickle-cell anemia and phenylketonuria.<sup>11</sup> Technological advances eventually allowed scientists to identify defects in chromosomes and ultimately many of the genes themselves.<sup>12</sup> These techniques became useful for carrier<sup>13</sup> and prenatal testing.

Today prenatal diagnosis is available for a number of inherited disorders and chromosomal anomalies. The most common indication for prenatal testing is advanced maternal age, which has historically been thirty-five years of age or older.<sup>14</sup>

Amniocentesis, first developed in the 1950s to monitor pregnancies at risk for Rh incompatibility, became an accepted reproductive diagnostic test in the 1970s.<sup>15</sup> The procedure, usually performed around sixteen weeks gestation, involves extraction of amniotic fluid from the mother's uterus. DNA or chromosomal analysis can be performed on the fetal cells that circulate in the amniotic fluid. The procedure poses a risk of fetal loss that can range from 0.5-1% depending on the genetics center.<sup>16</sup> A

newer procedure, chorionic villus sampling (CVS), became a diagnostic tool in the

<sup>7</sup> Paul Lombardo, *Medicine, Eugenics, and the Supreme Court: From Coercive Sterilization to Reproductive Freedom*, 13 CONTEMP. HEALTHL. & POL'Y 1, 5 (1996). In addition, in 1924, Congress

enacted the Federal Immigration Restriction Act "to combat 'the rising tide of defective germ-plasm' carried by suspect groups migrating from Southern and Eastern Europe." *Id.*

<sup>8</sup> 274 U.S. 200 (1927).

<sup>9</sup> *Id.* at 207. Paul Lombardo has pointed out that *Buck* has not been formally overturned.

Moreover, it has provided a subtle strand in reproductive rights cases, including *Roe v. Wade*.

Lombardo, *supra* note 7, at 19.

<sup>10</sup> DANIEL J. KEVLES, IN THE NAME OF EUGENICS 205-06 (1995).

<sup>11</sup> ASSESSING GENETIC RISKS, *supra* note 3, at 63.

<sup>12</sup> *Id.* at 63-64.

13 Carrier testing identifies individuals who carry single copies of deleterious recessive genes and are at an increased risk of having an affected child. If two carriers have a child there is a 25% chance the child will inherit two copies of the gene or be affected by it. Couples who are carriers for the same disease gene may choose to accept the risk of having an affected child, may adopt or may use prenatal diagnosis to determine if the fetus carries two copies of the disease gene. Thus, carrier screening is related to reproductive decision making.

Although we all carry several recessive disease genes, some are more common in certain ethnic and racial groups. For example, 1/25 of Caucasians (particularly those of Northern European descent) carries the gene for cystic fibrosis, 1/12 of African Americans carries the gene for sickle-cell anemia and 1/30 of Jews of Askenazi descent carries the Tay Sachs gene. Asian, Mediterranean and Middle Eastern populations are at varying increased risks of carrying the gene for thalassemia. *Id.* at 70-71.

14 *Id.* at 75. With every passing year, a woman's chance of having a child with a chromosomal abnormality increases. In the mid-1980s, the American College of Gynecologists and Obstetricians issued a policy statement describing the offer of prenatal diagnosis to women of advanced maternal age as part of the standard of care. *Id.*

15 *Id.* at 76.

16 *Id.* (describing timing between fourteen and sixteen weeks gestation); Jane E. Brody, *Experts Explore Safer Tests for Down Syndrome*, N.Y. TIMES, Jan. 23, 2001, at F6 (describing timing between sixteen and eighteen weeks gestation).

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1980s.<sup>17</sup> It has the advantage that it can be performed earlier than an amniocentesis, generally between nine and twelve weeks of gestation. However, it may pose a slightly higher risk of fetal loss: about 1%.<sup>18</sup>

The advent of prenatal testing coincided with the women's movement and some of its hallmarks (e.g., the availability of the birth control pill and the constitutional right to abortion). Together these technological advances and legal developments

offered unprecedented reproductive choices for women. With these choices came the freedom to control reproduction, giving women the ability to time childbearing around careers or personal desires.

In light of changing social attitudes toward women and reproduction, prenatal testing boomed in the late 1970s and early 1980s.<sup>19</sup> At the same time, the field of genetic counseling emerged. Universities developed formal programs for masterslevel training in genetic counseling, a process aimed at educating patients about their reproductive options and helping them reach informed decisions.<sup>20</sup> Unlike their eugenic predecessors, who had a social agenda regarding reproduction, modern genetic counselors saw their mission as *offering choice* to women and couples planning for and experiencing pregnancies.<sup>21</sup> As the field of genetic counseling developed, the profession adopted a nondirective approach—i.e., counselors strove to inform patients of their options without recommending particular decisions.<sup>22</sup> Rather than advocate a particular choice—indeed, nondirectiveness prohibits direct advice—the goal of genetic counseling is to provide information to help patients make decisions regarding genetic testing and reproduction consistent with their values, beliefs, circumstances and life plans.<sup>23</sup> In theory, at least, the goal is to maximize choice and promote autonomy.

Until the 1990s, genetic counseling and testing primarily addressed reproductive decisions.<sup>24</sup> Individuals might seek carrier testing to determine their risk of having a child with a particular recessive condition. Pregnant women at increased risk of fetal genetic anomalies might seek prenatal testing. In most cases, the motivation for

<sup>17</sup> ASSESSING GENETIC RISKS, *supra* note 3, at 76-77.

<sup>18</sup> *Id.*

<sup>19</sup> See Rachel Nowak, *Genetic Testing Set for Takeoff*, 265 SCIENCE 464, 464 (1994) (noting that prenatal testing became “a medical boom industry” in the 1980s).

20 CHARLES S. BOSK, ALL GOD'S MISTAKES: GENETIC COUNSELING IN A PEDIATRIC HOSPITAL

156-57 (1992); Beth A. Fine, *The Evolution of Nondirectiveness in Genetic Counseling and Implications of the Human Genome Project*, in PRESCRIBING OUR FUTURE: ETHICAL CHALLENGES IN

GENETIC COUNSELING 101, 105-06 (Dianne M. Bartels et al. eds., 1993).

21 See generally Seymour Kessler, *The Psychological Paradigm Shift in Genetic Counseling*,

27 SOC. BIOLOGY 167, 168, 182 (1980) (discussing the shift from the eugenics paradigm, which focused on managing human heredity, to the current paradigm of psychologic medicine, which focuses on helping patients resolve problems and make decisions).

22 Various forces have contributed to genetic counselors' commitment to nondirectiveness, including an attempt to distance themselves from eugenics; the bioethics movement, with its commitment to autonomy; and the women's movement, with its support of reproductive liberty. See Sonia M. Suter, *A Fresh Look at Nondirectiveness in Genetic Counseling* (Jan. 1, 2000) (unpublished manuscript, on file with author).

23 See, e.g., Barbara Bowles Biesecker, *Future Directions in Genetic Counseling: Practical and Ethical Considerations*, 8 KENNEDY INST. ETHICS J. 145 (1998). How successful nondirectiveness

is at achieving that goal is a question in some dispute. Several commentators argue either that nondirectiveness is not achievable, see *infra* note 72, or that counselors are not really neutral about the choices patients make, see notes 67-68 and accompanying text. An even larger issue is whether neutrality is the proper way to promote these goals, something I seriously doubt, though a topic beyond the scope of this Article.

24 Until then, the vast majority of genes that express in adulthood (late-onset genes) had not yet been identified. See *infra* text accompanying note 25.

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genetic testing or interest in genetic information was related to reproductive decisions.

With the rapid identification of numerous genes through the Human Genome Project, scientists began to isolate genes associated with late-onset conditions, such as HD and inherited forms of cancer.<sup>25</sup> HD was one of the first late-onset genes identified.<sup>26</sup> As genetic testing became available for at-risk individuals, research was conducted to assess the interest in and effects of learning whether one carried the gene. Studies provided some surprising information. First, far fewer people actually chose to be tested than expected.<sup>27</sup> Second, not surprisingly, those who learned they were at a significantly increased risk experienced anxiety and distress. However, contrary to expectations, studies showed that roughly 10% of those with a significantly *decreased* risk of carrying the gene had psychological difficulties coping with their new status.<sup>28</sup> Researchers found that genetic information about a late-onset condition for which no cures are available posed a risk of psychological distress. Third, geneticists and patients soon worried about the potential insurance, employment or social discrimination that might result if someone were identified as carrying the HD gene.<sup>29</sup>

These concerns led professional groups involved in HD testing and diagnosis to develop guidelines for predictive testing.<sup>30</sup> The guidelines reinforced many of the norms of genetic counseling, in particular, the commitment to informed consent and the notion that only the individual can decide whether to be tested.<sup>31</sup> The guidelines however, went beyond the approaches of prenatal genetic counseling. For example, the guidelines state that the “participant should be encouraged to select a companion to accompany him or her throughout all stages of the testing process.”<sup>32</sup> The guidelines also recommend preliminary “[n]eurologic examinations and

<sup>25</sup> See, e.g., *infra* notes 26 and 37.

26 Marlene Huggins et al., *Ethical and Legal Dilemmas Arising During Predictive Testing for Adult-Onset Disease: The Experience of Huntington's Disease*, 47 AM. J. HUM. GENET. 4, 4 (1990).

HD is an autosomal dominant neuropsychiatric condition that typically manifests in mid-life. Its symptoms include “chorea, cognitive impairment, and affective disturbance.” *Id.* Before the actual gene was identified, researchers found markers linked to the HD gene in 1984. Virginia Morell, *Huntington's Gene Finally Found*, 260 SCIENCE 28, 28 (1993). Analysis of the presence and absence of markers in affected and non-affected family members could be used to determine whether people with family histories of HD were very likely to carry the gene. Linkage studies, by nature, may not be precise and require analysis of DNA from multiple family members. Virginia Morell, *Gene Discovery Points to Better HD Test*, 260 SCIENCE 29, 29 (1993). The identification of the actual HD gene in 1993 allowed for more direct and accurate testing—it no longer required involvement of other family members. *Id.*

27 “Before linkage studies were available, 70% of people at risk for Huntington's stated they would use predictive testing. Yet, once the test was available, only 13% took advantage of it.” Sonia M. Suter, *Whose Genes Are These Anyway? Familial Conflicts Over Access to Genetic Information*, 91 MICH. L. REV. 1854, 1866 n.71 (1993).

28 *See, e.g.*, Marlene Huggins et al., *Predictive Testing for Huntington's Disease in Canada: Adverse Effects and Unexpected Results in Those Receiving a Decreased Risk*, 42 AM. J. MED. GEN. 508 (1992).

29 Steven Hersch et al., *The Neurogenetics Genie: Testing for the Huntington's Disease Mutation*, 44 NEUROLOGY 1369, 1370 (1994). So concerned are many patients about this risk that they either will get testing anonymously or will choose to pay in cash to avoid having to notify insurers of the findings. Anne Chalfant, *Genetic Testing: What Does it Mean for Nurses?*, NURSEWEEK, Aug. 3, 1998, at <http://www.nurseweek.com/features/98-8/genes.html>.

30 *See, e.g.*, Steven Hersch et al., *supra* note 29; INT'L HUNTINGTON'S ASS'N & WORLD FED'N OF NEUROLOGY RESEARCH GROUP ON HUNTINGTON'S CHOREA, *Guidelines for the*

*Molecular Genetics*

*Predictive Test in Huntington's Disease*, 44 NEUROLOGY 1533 (1994) [hereinafter Guidelines].

31 Guidelines, *supra* note 30, at 1533.

32 *Id.* at 1534 (Guideline 3).

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psychological appraisal . . . to establish a baseline evaluation of each individual.”<sup>33</sup>

Finally, in addition to disclosing the typical genetic counseling information—such as

information about the disease, the patient’s risk, the testing options, the limitations

and accuracy of the test and possible testing outcomes<sup>34</sup>—providers should also

discuss the “social and psychological implications.”<sup>35</sup> In other words, patients should

be informed about much more than just the medical implications. They should also

be informed of “the possible result on the individual’s affective state, future medical

status, family members, career and financial planning, and family planning.”<sup>36</sup>

After the more recent identification of genes associated with inherited forms of

breast cancer,<sup>37</sup> geneticists and ethicists again encouraged more elaborate genetic

counseling than is typical with prenatal testing. For example, a consensus statement

of professionals recommended that the content of informed consent for BRCA1/2

testing include not only the purpose, practical aspects and limitations of the test, but

also the psychological and social implications.<sup>38</sup> The recommended disclosure

regarding psychological risks is rather comprehensive. Patients should be advised of

potential adverse responses in those found to be mutation carriers such

as anxiety depression, anger, and feelings of vulnerability. In addition,

participants may experience guilt over the possibility of having passed

the mutation to offspring. Patients who find they are not mutation

carriers may also experience guilt if other close family members are

found to carry the mutation—so-called survivor guilt. Those who are

not mutation carriers may experience regrets if they have made major life decisions such as prophylactic surgery, based on their perception of risk prior to testing. Individuals who decline testing or who do not receive definitive results may experience persistent anxiety over their risk status.<sup>39</sup>

Counselors are also advised to inform patients that “genetic testing for cancer susceptibility may limit their ability to obtain health, life, or disability insurance, may lead to limitations in coverage, or may result in higher premiums for insurance products”<sup>40</sup> or “may pose a risk to their present or future employment.”<sup>41</sup>

<sup>33</sup> *Id.* at 1535 (Guideline 6.2). Some centers will not offer HD testing to patients who are clinically depressed.

<sup>34</sup> Hersch et al., *supra* note 29, at 1371-72. *See also* Guidelines, *supra* note 30, at 1534-35 (guidelines 5.1.1, 5.2.1, 5.2.2, 5.2.3).

<sup>35</sup> Guidelines, *supra* note 30, at 1534 (Guideline 5.1.1).

<sup>36</sup> Hersch et al., *supra* note 29, at 1371.

<sup>37</sup> The BRCA1 gene was isolated at the end of 1994 and the BRCA2 gene at the end of 1995.

Gina Kolata, *Scientists Speedily Locate a Gene that Causes Breast Cancer; Better Screening Is Seen*, N.Y. TIMES, Dec. 21, 1995 at A15. Studies suggest that the cumulative risk of breast cancer by age seventy in BRCA1 carriers ranges from 35-87%. Cumulative risks for ovarian cancer by age seventy in BRCA1 or BRCA2 carriers range from 16-44%. Robert J. Pokorski & Ulrike Ohlmer, *Use of Markov Model to Estimate Long-Term Insured Lives' Mortality Risk Associated with BRCA1 and BRCA1 Mutations*, 4 N. AM. ACTUARIAL J. 130, 131 (2001).

<sup>38</sup> *See, e.g.*, Gail Geller et al., *Consensus Statement: Genetic Testing for Susceptibility to Adult-Onset Cancer: The Process and Content of Informed Consent*, 277 JAMA 1467, 1470-72 (1997).

<sup>39</sup> *Id.* at 1471.

<sup>40</sup> *Id.*

41 *Id.* at 1472. The consensus statement acknowledges how difficult it is to determine the precise risk of discrimination and also to document it. It mentions that while existing legislation may protect against some forms of discrimination, it does not cover all individuals and has not been assessed for effectiveness. *Id.* (discussing the Health Insurance Portability and Accountability Act of 1996, which prohibits the denial of insurance for preexisting conditions, and the Americans with Disabilities Act of 1990, which prohibits employment discrimination against those with a disability,

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These guidelines and recommendations reflect an acknowledgment that knowledge is not always inherently beneficial. Indeed, the concerns raised by susceptibility testing cautioned against the cultural view that knowledge is power. Researchers began to warn of the potential “toxicity” of genetic information from late-onset testing given the psychosocial risks it posed. Dr. Francis Collins, director of the National Genome Research Institute compared DNA testing to the introduction of a new drug. Just as new drugs should undergo trials to assess their toxicity, so should genetic tests, some of which “can actually end up hurting people.”<sup>42</sup> These concerns were heightened by the possibility of the emergence of a high-profit industry offering testing for susceptibility to cancer and other common diseases.<sup>43</sup> Profit incentives, many feared, would reduce or even eliminate adequate informed consent. Even worse, tests might be offered indiscriminately, even to those for whom such testing would not be clinically appropriate or informative.<sup>44</sup> For all of these reasons, leaders in genetics and ethics were legitimately concerned about the new form of genetic tests, which can redefine currently healthy people as “at-risk” for conditions for which there may be no known treatment or cure. In short, the new tests are fraught with serious emotional and social risks.

The same healthy skepticism and circumspection regarding susceptibility testing should apply to prenatal testing, which also produces knowledge that some may find

toxic. Just as late-onset testing can provide information about serious medical risks about which little can be done, prenatal testing can provide information about fetal anomalies, for which no treatment is available. As I will discuss below, prenatal testing can lead to psychological stress and anxiety for some patients, just as late-onset testing can. In a culture that values knowledge for its own sake, patients with inadequate counseling might embark on prenatal testing without full awareness of the distress that can be associated with this kind of information gathering, particularly when no real therapeutic options exist.

Although scholars have discussed the psychosocial issues surrounding prenatal testing,<sup>45</sup> the guidelines and recommendations in this area tend to be far less specific about what should be disclosed in the counseling process than the recommendations for late-onset testing.<sup>46</sup> The real failing, however, is not in the recommendations for (with a record of a disability or who are perceived as disabled).

<sup>42</sup> Paul Cotton, *Prognosis, Diagnosis, or Who Knows? Time to Learn What Gene Tests Mean*, 273 JAMA 93, 95 (1995); *see also* Nowak, *supra* note 19, at 464 (quoting Francis Collins: “Genetic testing should be considered in the same way as a new drug. It can have efficacy, and it can have toxicity.”)

<sup>43</sup> *See generally* Michael J. Malinowski & Robin J.R. Blatt, *The Commercialization of Genetic Testing Services: The FDA, Market Forces, and Biological Tarot Cards*, 71 TUL. L. REV. 1211 (1997).

<sup>44</sup> *See* Nowak, *supra* note 19, at 464 (discussing the usefulness of test results for a patient with an incurable disease and the effect that a positive result in this situation has on the patient). Another concern has been the lack of quality controls for such testing. Published accounts of testing errors describe the kind of damage such errors can cause. Nancy Seeger, for example, who had a family history of breast cancer, decided to undergo genetic testing. The laboratory report indicated that she had one of the breast cancer genes, presenting “a lifetime risk of breast cancer ‘as high as 85 percent’” and a lifetime risk for ovarian cancer of 50%. The report assured her that the results had been

“confirmed independently.” After undergoing prophylactic surgery to remove her ovaries (no reliable screening is available for ovarian cancer) and while contemplating bilateral breast removal, however, she received shocking news. She did not, in fact, have the mutation. Anne Underwood, *When “Knowledge” Does Damage*, NEWSWEEK, Apr. 10, 2000, at 62. Although laboratory error is possible with any laboratory test, the lack of quality controls in late-onset genetic testing heightens concerns about just this sort of scenario.

45 See ASSESSING GENETIC RISKS, *supra* note 3, at 79-80, 83.

46 In 1991, for example, an NIH Workshop on Reproductive Genetic Testing noted that, in addition to providing benefits, prenatal testing has the “potential to increase anxiety; place excessive  
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prenatal testing counseling, but in the increasing routinization of prenatal testing and social norms that pressure women to undergo prenatal testing. As we enter a new era of genetics in which more and more genes are identified, extra care should be taken to prepare patients for the emotional and social costs of prenatal testing and to give women the opportunity to accept or reject it. Prenatal testing issues will only get more complex. Indeed, as it becomes possible to test pregnancies for increasing numbers of late-onset conditions, the issues will multiply as concerns of late-onset testing are added to those of prenatal testing.<sup>47</sup>

Part II of this Article will describe the routinization that has occurred with prenatal testing and the role that genetics and medical professionals, the public and the legal community have played in this evolution. Part III will describe the costs of such routinization. In particular, it will show how it has impoverished informed consent by muting many of the issues at stake in prenatal testing. As a result, patients may not be fully prepared for the psychological ramifications of obtaining genetic information. At best, patients will not make the best decisions for themselves, and at worst, they will not feel they have true choice with respect to prenatal testing. Part

III also describes the larger societal costs. Not only does the routinization of prenatal testing threaten the disabled community, it also tends to ignore the moral element of responsibility, blame, and guilt on a woman for her pregnancy outcome; interfere with maternal-infant bonding; and disrupt relationships between a woman, family members and her community.”

#### ASSESSING

GENETIC RISKS, *supra* note 3, at 83 (citing NAT'L INST. OF HEALTH WORKSHOP ON REPROD.

#### GENETIC

TESTING. Reproductive Genetic Testing: Impact on Women. Bethesda, MD, Nov., 1991). Ultimately, however, the participants recommended merely that “standards of care for reproductive genetic services should emphasize genetic information, education and counseling rather than testing procedures alone.” *Id.* at 84. Similarly, the American Society of Human Genetics (ASHG) issued a policy statement in 1987 regarding maternal serum alpha-fetoprotein (MSAFP), a form of prenatal screening. *See infra* text accompanying notes 117-118 (describing the use and indications of MSAFP screening).

It recommended that “[counseling for MSAFP screening] should be nondirective and should begin early in pregnancy so that [the patient’s] decision is informed and unhurried.” AM. SOC’Y OF HUM.

GENETICS, *American Society of Human Genetics Policy Statement for Maternal Serum Alpha-Fetoprotein Screening Programs and Quality Control of Laboratories Performing Maternal Serum and Amniotic Fluid Alpha-Fetoprotein Assays*, 40 AM. J. HUM. GENET. 75 (1987), available at

<http://www.faseb.org/genetics/ashg/policy/pol-01.htm> (last visited Apr. 30, 2002). It also urged that

patients be “fully informed about the procedure and its implications,” though it did not elaborate on what patients should be told. *Id.* Similarly, the Code of Ethics adopted by the National Society of

Genetic Counselors urges genetic counselors to “strive to [e]nable their clients to make informed independent decisions, free of coercion, by providing or illuminating the necessary facts and clarifying the alternatives and anticipated consequences,” though it does not specify what those “anticipated

consequences” are. Cara Dunne & Catherine Warren, *Lethal Autonomy: The Malfunction of the Informed Consent Mechanism within the Context of Prenatal Diagnosis of Genetic Variants*, 14 ISSUES

L. & MED. 165, 190 (1998) (citing NAT'L SOC'Y OF GENETIC COUNSELORS, *A Voice, a Resource and*

*an Educational Environment for the Genetic Counseling Profession*, 1991, distributed at the 1997 Annual Genetics Conference, in Baltimore, MD, American Society of Genetics Counselors (on file with Cara Dunne)). Perhaps the group that comes closest to advocating a counseling process that resembles the recommendations for late-onset testing is the Committee on Assessing Genetic Risks. It recommends that informed consent for reproductive testing include, among other things, "consideration of all possible outcomes, including the possibility that one option might be termination of the pregnancy; [and] . . . knowledge of the potential need for and availability of psychosocial counseling." ASSESSING GENETIC RISKS, *supra* note 3, at 104. The committee does not, however, explicitly advocate

that counseling include a discussion of the nature of psychosocial effects that may result from prenatal testing or screening.

47 The guidelines for HD testing discourage prenatal HD testing unless the couple would terminate, stating that "[t]he couple requesting antenatal testing must be clearly informed that if they intend to complete the pregnancy if the fetus is a carrier of the gene defect, there is no valid reason for performing the test." Guidelines, *supra* note 30, at 1535 (Guideline 7.2). This recommendation expresses a level of directiveness that would rarely be seen in prenatal genetic counseling. The appropriateness of prenatal testing for late-onset conditions, such as HD and inherited forms of breast cancer, is a complex and controversial subject beyond the scope of this Article.

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these social norms and choices. As a result, society is poorly equipped to debate the ethical concerns raised by emerging forms of reproductive technologies.

#### II. THE ROUTINIZATION OF REPRODUCTIVE GENETIC TESTING

Although attitudes of patients and practitioners vary with respect to reproductive testing, most are in favor of it. For pregnant women of any age, prenatal *screening*—

identifying those at high risk for abnormalities, as opposed to making definitive diagnoses—has become virtually routine.<sup>48</sup> For women of advanced maternal age, particularly well-educated, middle- to upper-class women, amniocentesis has become, if not the norm, a choice of the vast majority.<sup>49</sup> Assumptions that women will undergo prenatal testing or screening are evident in some of the widely-read books on pregnancy.<sup>50</sup> As Barbara Rothman describes so eloquently in *The Tentative Pregnancy*,<sup>51</sup> prenatal testing has become part of social norms and expectations. Its routinization is reflected in attitudes of medical professionals, the patients offered prenatal testing and those they consult.<sup>52</sup> So accepted a part of pregnancy is it that women who reject such testing must be prepared with explanations and justifications.<sup>53</sup> People ask “why not?” No one asks “why?” We all know why, and many would make the same decision.

My professional and personal experiences are consistent with Rothman’s observations. As a genetic counselor, I observed that most, though not all, patients to whom we offered genetic screening or testing accepted it, often with seemingly little difficulty. Now as a law professor who teaches and writes about the ethical and legal aspects of genetic testing, I witness the same attitudes among my students when we discuss prenatal testing. A fair number of law students, most of whom have not yet faced questions about reproductive testing, often express the view of some patients: “Aren’t you supposed to have prenatal testing, especially if you’re older?” And finally, as I gained a new facet to my identity (the pregnant woman of advanced maternal age) I was struck by how many people, no matter how familiar, raised the subject of prenatal testing, and even more by the way they phrased their query. Most did not ask *whether* I underwent amniocentesis or CVS, but instead *what kind of testing* I chose. The assumption among most was that I was tested.<sup>54</sup> In short, my

<sup>48</sup> As above, the same claim could be made with respect to prenatal sonograms. *See supra* note

2.

49 BARBARA KATZ ROTHMAN, *THE TENTATIVE PREGNANCY: HOW AMNIOCENTESIS CHANGES*

*THE EXPERIENCE OF MOTHERHOOD* 16 (1986) (“Like maternity blazers, prenatal diagnosis is a hallmark of the professional woman’s pregnancy.”).

50 *The Girlfriend’s Guide to Pregnancy*, for example, states quite matter-of-factly that “[i]f you are age thirty-five or older, you will probably have a genetic test that actually samples some of the pregnancy matter or the amniotic fluid.” VICKI IOVINE, *THE GIRLFRIEND’S GUIDE TO PREGNANCY : OR*

*EVERYTHING YOUR DOCTOR WON’T TELL YOU* 59 (1995). *What to Expect When You’re Expecting*

notes that prenatal diagnosis is generally recommended for women with specific characteristics, including being over age thirty-five, although it is more attentive to some of the complexities involved, recognizing that because “of inherent risks, small as they are, prenatal diagnosis isn’t for everyone.”

ARLENE EISENBERG ET AL., *WHAT TO EXPECT WHEN YOU’RE EXPECTING* 42 (2d ed. 1996).

This may

be the most widely-read book on pregnancy. It is self-described as “America’s Pregnancy Bestseller” and has sold over nine million copies. *Id.*

51 ROTHMAN, *supra* note 49, at 16.

52 *Id.* at 45, 51-52 (For many patients, amniocentesis is already an “expected, accepted” part of the pregnancy experience.).

53 *Id.* at 63, 67.

54 How many of these questions were inspired by my former career as a genetic counselor, I cannot say. But I know that many asked me this question without full awareness of my professional past. Many seemed to raise the question merely because I was a pregnant woman over thirty-five.

professional and personal experiences suggest that reproductive testing for “older” mothers, and genetic screening for all pregnant women, has become almost as routine as forsaking alcohol and caffeine during pregnancy.

Why has it become the norm and what are the implications of such a trend?

Understanding fully how genetic counseling has become so routinized is complex and one would need to do some careful empirical research to reach definitive answers.

But I feel comfortable speculating based on existing data and the benefit of three perspectives: the genetic counselor, the professor of law and medicine and, more recently, the pregnant woman. One explanation is quite simply that the technology is available. It can be done, so it is offered, and it is accepted. But this does not fully answer *why* the offer is accepted.

Several complicated, intertwined reasons explain the routinization. Norms within the profession of genetic counseling, medicine and society play a role.

Although genetic counselors do not actively promote prenatal testing because nondirectiveness explicitly prohibits advice about whether or not to undergo testing, many of the values that shape the profession indirectly promote prenatal testing or screening. The routinization is intensified because amniocentesis, CVS and, especially, prenatal *screening* are now performed by obstetricians, instead of just genetics specialists. Non-genetic health professionals are less steeped in the culture of nondirectiveness than genetic counselors and are therefore more likely to promote any technology that provides additional information about the medical “condition” of pregnancy. Financial incentives to encourage testing only exacerbate this trend. In addition, the strong value our society places on knowledge, information and technology reinforces the public’s acceptance and expectation of prenatal testing. In part, the strong interest in information reflects a desire to gain some sense of control

over a process so fundamentally out of one's control. Finally, the legal community has indirectly helped routinize prenatal testing and screening through the threat of malpractice litigation and some specific legal measures.

#### A. GENETIC COUNSELORS AND OTHER HEALTH PROFESSIONALS

The role of genetic professionals in the routinization of reproductive testing is complex. On the one hand, the profession's commitment to nondirectiveness would suggest that genetic professionals are merely responding to social demands and desires, rather than shaping them. By refusing to direct clients toward a particular decision, counselors seem removed from influences that routinize prenatal testing. However, although nondirectiveness advocates neutrality,<sup>55</sup> genetic counselors are far from neutral about many aspects of genetic counseling.<sup>56</sup> In other words, many deeply held values shape their approach to genetic counseling. As we shall see, these values can subtly promote prenatal testing.

For example, genetic counselors are not neutral about the *value of nondirectiveness*. Even as the profession's understanding of the term evolves and the notion has come under recent attack,<sup>57</sup> most genetic counselors defend the approach vigorously. Counselors defend nondirectiveness on several grounds: 1) the duty to protect patient autonomy; 2) the inability to know what the "best" decision is for someone else; 3) the conviction that nondirectiveness is good for patients; and 4) less

<sup>55</sup> Kessler, *supra* note 21, at 176; Suter, *supra* note 22.

<sup>56</sup> Biesecker, *supra* note 23, at 145. *See generally* Sonia M. Suter, *Value Neutrality and Nondirectiveness: Comments on "Future Directions in Genetic Counseling"*, 8 KENNEDY INST.

#### ETHICS

J. 161 (1998).

<sup>57</sup> *See infra* notes 67-68.

frequently stated, the patient's responsibility to make decisions.<sup>58</sup> So strongly do genetic counselors feel about these rationales that they can be quite directive about *how* patients should approach decision making, even if they try not to direct the decision itself.<sup>59</sup>

The fact that counselors try to influence the process, as opposed to the decision, however, can subtly and unintentionally influence the decision itself.<sup>60</sup> The genetic counselor's deep and abiding interest in protecting patient autonomy can sometimes translate into a belief that the counselee has a "right and *responsibility*" to decide for herself. Many genetic counselors are uncomfortable with patients who want to abdicate the role of decision making.<sup>61</sup> By trying to urge patients to decide for themselves, counselors might be understood as urging information gathering. In addition to valuing nondirectiveness, genetic counselors deeply value access to and delivery of information. Indeed, trading information is central to genetic counseling and its *raison d'être*—to equip people to make informed decisions compatible with their beliefs, values and lifestyles. Without this exchange of information, no counseling could occur. Genetic counseling begins with the counselor obtaining as much information as possible from the patient, including family history, medical history, pregnancy history and reason for referral (such as advanced maternal age, family history of a genetic disease, etc.).<sup>62</sup> These various sources of information shape the discussion regarding the patient's risks and the available testing options.

The central part of genetic counseling, however, is the *delivery* of information, which can be quite comprehensive. In prenatal testing counseling, the genetic counselor begins by explaining that all pregnancies have a 3-5% population risk of birth defects, regardless of family history. She then describes the patient's particular pregnancy risk—for example, a 1/270 chance of Down syndrome, a 1/4 risk of cystic

fibrosis or a 1/25 chance of being a carrier for Tay Sachs.<sup>63</sup> A discussion of the various testing alternatives—carrier testing, prenatal screening or prenatal testing (amniocentesis or CVS)—follows. Finally, the counselor describes the information that such tests can provide, their limitations—not all conditions can be identified—and their risks. For example, amniocentesis and CVS pose a small risk of

58 Suter, *supra* note 22.

59 *Id.*; ROTHMAN, *supra* note 49, at 46 (noting that genetic counselors try to control the decision making process, even though they try not to control the decision itself).

60 ROTHMAN, *supra* note 49, at 46 (questioning whether one can influence the decision making process without also influencing the decision itself).

61 A situation in which a couple repeatedly requested that genetic counselors decide whether they should have prenatal testing for muscular dystrophy illustrates this perspective nicely. The counselors believed the couple's behavior demonstrated their fear of accepting "responsibility for their actions" and was an attempt to place the responsibility with the genetic counselors. ELEANOR GORDON

APPELBAUM & STEPHEN K. FIRESTEIN, *A GENETIC COUNSELING CASEBOOK* 78-79 (1983).

They

defended their refusal to decide for the patient by arguing that "no one has the right to make decisions which will affect the lives and feelings of other people." *Id.* Ironically, the genetic counselors supported the couple's decision to follow the advice of the rabbi. In essence, they supported the couple's refusal to decide as long as they themselves were not the one's who decided for the couple.

62 See Michael J. Malinowski, *Coming Into Being: Law, Ethics, and the Practice of Prenatal Genetic Screening*, 45 *HASTINGS L.J.* 1435, 1460 (1994).

63 This part of the counseling session can often resemble a mini-biology class with numerous diagrams illustrating meiosis (cell division that results in the final egg and sperm cells), chromosomes, nondisjunction (the error in meiosis that can result in the fetus having too many or too few

chromosomes) and, when applicable, patterns of inheritance to illustrate the risk to the pregnancy of a particular inherited disease.

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miscarriage to the fetus.<sup>64</sup> As one can see, even the most routine prenatal counseling visit entails a great deal of information exchange.

Conveyance of medical information is so crucial in genetic counseling that it can indirectly influence the genetic counseling process. Patients may sense the value genetic counselors place on medical information itself, as well as the value they place on genetics technology and research, which is the very source of the information geneticists offer to patients.<sup>65</sup> Advances in the field have made and continue to make knowable information about the fetus that only decades ago was unknowable.

Genetic counselors are therefore not neutral about science, technology and research and certainly not neutral about the availability of access to genetic testing. Patients may perceive and interpret these values as evidence that counselors favor prenatal testing.<sup>66</sup>

This commitment to science and technology and access to genetic testing has led some commentators to question the alleged neutrality toward genetic testing decisions themselves. Some argue that genetic counselors, by virtue of their chosen profession, must have a clear bias in favor of genetic testing.<sup>67</sup> Some even suggest that the offer of genetic tests reflects the profession's unspoken support of *termination* of affected pregnancies.<sup>68</sup> These claims are too strong to describe the entire class of genetic counselors and are inconsistent with the frequently articulated view of most genetic counselors that no objectively "right" decisions exist.<sup>69</sup>

Nevertheless, evidence suggests that some counselors, typically physician genetic counselors, hold those views.<sup>70</sup>

Whether or not genetic counselors actually believe genetic testing or termination

of affected pregnancies is the right decision for everyone, the majority of genetic counselors have such preferences for *themselves*. My anecdotal experience, confirmed by empirical data, indicates that even if they are not in a high-risk

64 See *supra* text accompanying notes 16-18.

65 See Gwen Anderson, *Nondirectiveness in Prenatal Genetics: Patients Read Between the Lines*, 6 NURSING ETHICS 126, 129 (1999) (“In genetics, clinicians and researchers believe that knowledge and genetic science are moral goods.”).

66 Even when counselors try not to express views, patients tend to read between the lines, often interpreting the counselor’s reticence in surprising ways. See Susan Michie et al., *Nondirectiveness in Genetic Counseling: An Empirical Study*, 60 AM. J. HUM. GEN. 40 (1997) (observing that patients often

felt “steered by the counselor,” regardless of the rated level of directiveness of the counselor).

67 BOSK, *supra* note 20, at 153; Angus Clarke, *Is Non-Directive Genetic Counselling Possible?*, 338 LANCET 998, 998-1000 (1991) (noting a “tendency for molecular genetic and other fetal diagnostic tests to be adopted as a matter of course once they become technologically feasible,” and contending that the “offer of prenatal testing implies a recommendation to accept that offer”).

68 Clarke, *supra* note 67, at 1000 (“I contend that an offer of prenatal diagnosis implies a recommendation to accept that offer, which in turn entails a tacit recommendation to terminate a pregnancy if it is found to show any abnormality.”).

69 APPELBAUM & FIRESTEIN, *supra* note 61, at 8 (stating that the counselor cannot know what the “best” decision is for the expectant couple and their family); PATRICIA T., *DEALING WITH DILEMMA: A HANDBOOK FOR GENETIC COUNSELORS* 102, 104 (1997) (Despite the counselor’s expertise about genetic matters, “it would be a mistake to conclude that they also know what a ‘better’ or ‘right’ decision would be for a particular family. . . . There is no one standard for judging . . . the correctness of a decision.”). See also BOSK, *supra* note 20, at 84 (In defense of nondirectiveness, one genetic counselor asked rhetorically, “how would anyone of us know how we would respond?”).

70 See ANDERSON, *supra* note 65, at 128 (observing that “scientists, healthcare professionals and society believe . . . as a source of knowledge for preventing or curing disease, genetic technology is morally good”), 129-30 (noting that genetic professionals believe genetic testing leads to an increased quality of life for fetuses, other siblings, parents and society). A study in 1973 found that 85% of genetic counselors, most of whom were physicians, believed that genetic counseling should achieve disease prevention. ROTHMAN, *supra* note 49, at 41 (citing James R. Sorenson, *Counselors: A Self Portrait*, in GENETIC COUNSELING VOL. 1, NO.5 (1973)).

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category, genetic counselors are especially likely to undergo prenatal testing and perhaps even to terminate affected pregnancies.<sup>71</sup> These personal biases in favor of testing and termination may inadvertently and subtly be conveyed to patients, even among genetic counselors committed to nondirectiveness.<sup>72</sup>

For many genetic counselors, the value of genetic testing is not always primarily linked to decisions about pregnancy termination. Many genetic professionals believe that the information from prenatal testing offers value to patients, quite apart from considerations of pregnancy termination. Some counselors believe that prenatal testing offers an antidote to the anxiety that pervades pregnancy since most couples, even high-risk couples, are likely to receive reassuring information. Even for the unlucky few who will receive unfortunate news and who would not terminate the pregnancy, the information can be valuable, in the view of many counselors, because it can help couples prepare emotionally and practically for their future disabled child.<sup>73</sup> Because the emphasis of counseling is adjustment to information, counselors tend to believe that the more advanced warning and preparation there is, the better off a patient or couple will be. Physicians in particular value such information as it can help them prepare for the difficulties associated with delivering a child with complications.<sup>74</sup>

Finally, whatever role genetic counselors have played in the routinization of prenatal testing, the fact that much of it has moved out of genetic centers to obstetricians' offices reflects how well accepted the procedure has become.<sup>75</sup> This transition occurred not only because genetic centers could not keep up with demand, but also because prenatal testing is becoming institutionalized as services are provided *en masse*.<sup>76</sup> Prenatal testing has now quite literally entered the domain of routine prenatal care.

This trend furthers the routinization of prenatal testing for a few reasons. The new providers of amniocentesis do not share the same commitment to nondirectiveness. Physician genetic counselors are more directive than masters-level genetic counselors,<sup>77</sup> and non-geneticist physicians are even more so.<sup>78</sup> As a result, 71 "Most of the counselors [Rothman interviewed] would have [amniocentesis] themselves. More than half would have, or want their daughters to have, an amniocentesis even at 25, a 'low risk age.' . . . Most would have abortions for most abnormalities, half said they would abort for *any* abnormality." ROTHMAN, *supra* note 49, at 46.

72 A number of commentators have questioned the ability to achieve neutrality in the counseling process, particularly when the counselors have personal biases. *See, e.g.,* Karen G. Gervais, *Objectivity, Value Neutrality, and Nondirectiveness, in Genetic Counseling, in PRESCRIBING OUR FUTURE: ETHICAL CHALLENGES IN GENETIC COUNSELING* 119, 127 (Dianne M. Bartels et al. eds.,

1993) (questioning the "concept of objectivity and the fact/value distinction" on which the normative modeling in genetic counseling is based); Dunne & Warren, *supra* note 46, at 188-89, 193 (1998) (describing the ways that counselors can be directive by failing to include information about "the human aspects of illness" associated with conditions for which prenatal testing was offered); Dorothy C. Wertz & John C. Fletcher, *Attitudes of Genetic Counselors: A Multinational Survey*, 42 *AM. J. HUM. GENETICS* 592, 600 (1988) (expressing skepticism that counselors can always neutrally support any

decision that the patient makes given the moral convictions that geneticists have).

73 See ROTHMAN, *supra* note 49, at 81 (noting that preparation and reassurance are the “bases on which amniocentesis is urged on women who are quite sure that they will not abort”).

74 I have heard this justification from obstetricians countless times when discussing prenatal testing. *See infra* note 172.

75 ASSESSING GENETIC RISKS, *supra* note 3, at 168.

76 *Id.*

77 See Deborah F. Pencarinha, *Ethical Issues in Genetic Counseling: A Comparison of M.S. Counselor and Medical Geneticist Perspectives*, 1 J. GEN. COUNSELING 19 (1992).

78 Because the commitment to autonomy (reflected in the norm of nondirectiveness) is greater in genetics than any other discipline, see *infra* text accompanying note 178, it is not surprising that

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many patients choose prenatal testing in part because their physician has *recommended* or even *encouraged* it—something masters-level genetic counselors usually would not do directly. Physicians tend to make such recommendations partly because “the idea that one would not want information is so counter to the medical profession’s world view.”<sup>79</sup> While genetic counselors tend to accept a broad range of reasons for rejecting prenatal testing, non-geneticist physicians are more inclined to encourage it, unless the patient has a history of infertility or miscarriage.<sup>80</sup> Genetic counseling may be less extensive in this setting as well, particularly with the time pressures of managed care, the shortage of genetic counselors and insurance reimbursement practices. Insurance companies often reimburse for diagnostic tests at far higher levels than for pretest counseling—if they reimburse for counseling at all.<sup>81</sup> As a result, fiscal concerns might lead practitioners to spend less time talking and more time pushing testing. All of these factors indirectly pressure patients to undergo prenatal testing.

As we have seen, numerous attitudes and circumstances within healthcare contribute to the routinization of prenatal testing and screening. Despite their commitment to nondirectiveness, genetic counselors may subtly promote prenatal testing because of values they hold dear. In addition, the movement of prenatal testing from genetics centers into obstetricians' offices as well as economic realities further routinize prenatal testing.

## B. PATIENTS AND SOCIETY

Whatever values genetic and other healthcare professionals bring to prenatal testing, patients come with their own. Patients' values reflect a combination of individual perspectives and social norms. While a great deal of variation exists among patients, some trends can be observed. Some women reject prenatal testing because they know they would not terminate a pregnancy for moral, religious or personal reasons. Others reject such testing because of the risk of miscarriage. But most women at increased risk of chromosomal or other detectable conditions (in other words, those for whom it is medically indicated) choose prenatal testing or screening.<sup>82</sup> Several factors contribute to this trend. Just as medical professionals are not neutral about the value of information, neither is our society, which views the gathering of information as a sign of responsible behavior and good decision making.<sup>83</sup> In short, knowledge is power. In the context of prenatal testing, as we shall see, patients tend to believe that getting information about the fetus is not only the right thing to do, but a form of reassurance and a way to get a sense of control over the potentially overwhelming experience of reproduction.

physicians who were not trained in genetics would be more directive than non-geneticist physicians.

<sup>79</sup> ROTHMAN, *supra* note 49, at 82. *See also* Anderson, *supra* note 65, at 129 (describing "the moral imperative to know" among healthcare providers).

<sup>80</sup> ROTHMAN, *supra* note 49, at 63; *see also* Anderson, *supra* note 65, at 129-30 (observing the

medical profession's view that genetic testing will further the well-being of fetus, siblings, parents and society).

81 Elizabeth B. Cooper, *Testing for Genetic Traits: The Need for a New Legal Doctrine of Informed Consent*, 58 MD. L. REV. 346, 360 (1999).

82 See *supra* text accompanying note 49.

83 Anderson, *supra* note 65, at 129 (noting the public's acceptance of the "moral imperative to know").

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### 1. "Doing What's Best"

Both my anecdotal observations as a genetic counselor and empirical studies suggest that our culture's value of knowledge generally and the ideal of being a good parent motivate many patients to undergo genetic screening or testing. A common refrain among the majority who choose prenatal testing is that they want to do everything they can for this pregnancy and future child.<sup>84</sup> One study that examined the reasons women accepted a prenatal screening test found that many women took the test "just to be safe," "to make sure my baby would be the healthiest it could be," or "to do anything that would help me or my baby."<sup>85</sup> Some women found empowerment in knowledge: "The more you know about your child, the better off you are." Others asked, "How could it hurt to know?"<sup>86</sup> I heard similar rationales countless times. Implicit, or at times explicit, is the idea that getting as much information as possible is inherently good for all concerned, including the fetus. Reflecting (and perhaps also shaping) these norms are some of the popular books on pregnancy. *The Girlfriend's Guide to Pregnancy* strongly advocates information gathering. It urges the reader not to "let fear [of amniocentesis] prevent [her] from getting the information [her] doctor thinks [she] should have."<sup>87</sup> When it comes to prenatal *screening*, the author is unequivocal: "it is a valuable warning

flare, so get the test.”<sup>88</sup> Some pregnancy books present information about amniocentesis in a manner that suggests it protects the fetus. *Your Pregnancy Week by Week* discusses amniocentesis in a section of the chapter entitled “How Your Actions Affect Your Baby’s Development.”<sup>89</sup> This same section heading is used in other chapters to discuss the harmful effects of smoking and alcohol consumption. In describing amniocentesis under such a heading, the book suggests that prenatal testing “constitutes maternal *good* behavior.”<sup>90</sup> *What to Expect When You’re Expecting* places a boxed insert entitled “Reducing the Risk in Any Pregnancy,” which includes advice on such things as smoking, alcohol and weight gain, in the middle of its discussion of amniocentesis.<sup>91</sup> “The completely unsupported inference here is that genetic disorders can be prevented by behavioral changes.”<sup>92</sup> “Good” mothers, these books imply, protect their babies by forsaking alcohol and undergoing prenatal tests. Usually unspoken, as Part III will discuss, is the link between prenatal testing and termination and how that relates to being a good parent. Lurking in these statements seems to be an undeveloped belief in the empowerment of knowledge and its relationship to conscientious parenting.

If women or couples link the notion of good parenting to pregnancy termination, it may be based on a perceived duty to prevent suffering in the unborn child. Some women may feel guilty or blameworthy for bringing a child with a genetic defect into

<sup>84</sup> See, e.g., ROTHMAN, *supra* note 49, at 59 (reporting one woman’s reasons: “I have done all I can do that is medically feasible and advisable, at my age, to ensure that any baby I have will be fine.”).

<sup>85</sup> Nancy Anne Press & Carole H. Browner, *Collective Silences, Collective Fictions, in WOMEN AND PRENATAL TESTING: FACING THE CHALLENGES OF GENETIC TECHNOLOGY* 201, 213 (Karen H.

Rothenberg & Elizabeth J. Thomson eds., 1994).

86 *Id.* at 213.

87 IOVINE, *supra* note 50, at 86.

88 *Id.* at 90.

89 GLADE B. CURTIS & JUDITH SCHULER, YOUR PREGNANCY WEEK BY WEEK 163 (4th ed. 2000).

90 HELENA MICHIE & NAOMI R. CAHN, CONFINEMENTS: FERTILITY AND INFERTILITY IN CONTEMPORARY CULTURE 84 (1997). *See also* CURTIS & SCHULER, *supra* note 89, at 163-64.

91 EISENBERG ET AL., *supra* note 50, at 50.

92 MICHIE & CAHN, *supra* note 90, at 84.

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the world.<sup>93</sup> Social attitudes only reinforce these views. When a television anchorwoman with ectrodactyly—a mild genetic condition that fused the bones of her hands—chose to continue a pregnancy with a fetus that inherited the condition, many accused her of being immoral or irresponsible.<sup>94</sup> Even healthcare professionals sometimes blame women for the birth of a child with a genetic condition, particularly if the woman refused testing.<sup>95</sup> Studies have shown that many physicians pressure women to terminate affected pregnancies, even when the defects are not severe.<sup>96</sup> In short, social norms influence the belief that “good” parents undergo prenatal testing.

## 2. Control and Reassurance

In addition to trying to be “good” parents, women and/or couples often pursue genetic testing for reassurance and a sense of control. Pregnancy is a significant emotional, biological and life-altering process. Women become deeply attached to the baby they have not yet met. Not surprisingly, many worry about what could potentially go wrong. The enormity of what is happening and the deep emotional connection women feel to their pregnancy can lead to a powerful sense of loss of control and need for reassurance. For many, knowledge offers an antidote,<sup>97</sup> perhaps

more illusory than real, to the overwhelming emotions that can coincide with pregnancy.

Several studies have shown that one of the main reasons patients seek prenatal testing is for reassurance.<sup>98</sup> I observed an extreme example of this motivation in one case. The patient, a woman in her early thirties, came to our clinic because she had been exposed to chemotherapy and radiation treatment for breast cancer a few years before her pregnancy. Although no data suggested that the prior treatment increased her risks above the 3-5% population risk of birth defects, we had to acknowledge that few studies had been conducted. We offered her an amniocentesis, primarily for reassurance, even though it was not technically medically indicated. We emphasized, however, that the test could only rule out chromosomal abnormalities and spine defects, as well as any structural abnormalities that might be observed on an ultrasound. It could not, however, detect conditions that worried her, such as most

<sup>93</sup> Lori B. Andrews, *Prenatal Screening and the Culture of Motherhood*, 47 HASTINGS L.J. 967, 981 (1996) (citing Donna G. Olsen, *Parental Adjustment to a Child with Genetic Disease: One Parent's Reflections*, 23 J. OBSTETRIC GYNECOLOGIC NEONATAL NURSING 516, 516 (1994)). In addition, some women experience guilt simply for having passed on the genetic condition to their child. *Id.* at 980-81.

<sup>94</sup> *Id.* at 981-82.

<sup>95</sup> *Id.* at 982.

<sup>96</sup> *Id.* at 990 n.117.

<sup>97</sup> People may seek knowledge simply because uncertainty makes them uneasy. Not knowing may make an individual feel more out of control than knowing, even if knowledge may potentially bring bad news.

<sup>98</sup> See J. M. Green, *Claiming or Harming? A Critical Review of Psychological Effects of Fetal Diagnosis on Pregnant Women*, Galton Institute Occasional Papers, Second Series, No. 2; Theresa

Marteau et al., *The Impact of Prenatal Screening and Diagnostic Testing upon the Cognitions, Emotions and Behaviour of Pregnant Women*, 33 J. PSYCHOSOMATIC RESEARCH 7 (1989). For a discussion of the variations in women's perception of the amniocentesis experience see Caroline C. Nielsen, *An Encounter with Modern Medical Technology: Women's Experiences with Amniocentesis*, 6 WOMEN & HEALTH 109 (1981). Not surprisingly, the pregnancy books also describe reassurance as a primary reason for prenatal testing. EISENBERG ET AL., *supra* note 50, at 42 (stressing the value of prenatal testing even for those who would not consider an abortion because "[f]or the vast majority of expectant parents the best reason for prenatal diagnosis is the reassurance it almost always brings"); IOVINE, *supra* note 50, at 59, 85, 87 (describing AFP screening and ultrasounds as tests that can offer reassurance).

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forms of mental retardation, deafness and blindness. A few days later when she came in for the amniocentesis, as if she had forgotten the prior discussion, she asked if the test would rule out mental retardation, deafness and blindness. Even after hearing again about the limits of the test, she apparently could not absorb that information. When I called her with the good news that the baby had forty-six chromosomes and normal alfa-fetoprotein (AFP) levels<sup>99</sup>, she asked again about mental retardation, blindness and deafness. Although she was an intelligent woman, her need for reassurance outweighed her ability to grasp the information.

Although some level of worry or concern about the fetus' well-being is an inevitable experience for most pregnant women, the existence of prenatal testing may actually create the need for certain forms of reassurance. For example, until prenatal testing for Down syndrome and other chromosomal abnormalities became available, the need for reassurance through amniocentesis did not exist.<sup>100</sup> Prior to such technology, Down syndrome and chromosomal abnormalities were not standard pregnancy concerns.<sup>101</sup> As we develop the means to test for more conditions, we will

add to the list of concerns about which pregnant women will want reassurance. For example, soon pregnant women will worry about cystic fibrosis, a recessive inherited condition that most commonly affects people of Northern European descent. Rare in the general population—roughly 30,000 children and adults have the disease—it is the most common inherited disorder among Caucasians.<sup>102</sup> Although the gene was identified over ten years ago, the American College of Obstetrics and Gynecology (ACOG) recently decided that carrier testing should be offered to “every Caucasian—or the partner of a Caucasian—who is pregnant or considering having a baby.”<sup>103</sup> As this becomes the standard of care, more Caucasian women will be concerned about their 1/29 risk of being a carrier.<sup>104</sup>

The desire for control that motivates some couples to undergo prenatal testing may extend beyond a need for reassurance. It may also be tied to fantasies about one’s future child and hopes for a kind of perfection. The desire to do what is best for one’s child may be linked to a wish to have the best child possible and to “the

99 See *infra* text accompanying notes 117-118.

100 Rothman describes not only how the existence of amniocentesis creates anxiety, but also how it destroyed traditional means of reassurance for many women, such as feeling fetal movement. Most women who undergo amniocentesis, she found, do not find fetal movement reassuring, whereas those who do not have amniocentesis found quickening reassuring. See ROTHMAN, *supra* note 49, at 108-10.

101 Of course, the fact that women are increasingly delaying childbearing means that more and more women are in the “high risk” group with respect to Down syndrome.

102 Luran Neergaard, *Gene Tests Offered for Cystic Fibrosis*, DESERET NEWS, Oct. 3, 2001, at A11.

103 *Cystic Fibrosis: Widespread Testing Begins this Month*, AM. HEALTH LINE, Oct. 2, 2001 (quoting the American College of Obstetricians and Gynecologists); Neergaard, *supra* note 102. The

test will also be available to non-Caucasians, who have a lower incidence of carrying the disease gene.

*Id.* This test is not 100% accurate, however, raising concerns about the need for adequate counseling.

104 *Id.* The risk to the fetus is much smaller. If the woman and her partner are at “high risk”

(i.e., have a 1/29 chance of being a carrier), the fetus has a risk of 1/3364 ( $1/29 \times 1/29 \times 1/4$ ) of being affected. This is not high compared to risks that we all face, though it is considered high in the world of inherited disorders. Of course, if both partners are carriers, the fetus has a 1/4 chance of being affected.

In my experience, although many people are aware that cystic fibrosis exists, most do not understand the nature of the disease, let alone that it is not only inherited but one of the most common inherited diseases. Even those who know it is inherited are often quite surprised to discover that the risks are as high as 1/29 for Caucasians. Thus, even if some women were concerned about cystic fibrosis before these recommendations were issued, I suspect that the vast majority will now add it to their list of pregnancy concerns.

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premium we place on ‘normalcy.’”<sup>105</sup> Though a powerful distinction exists between the eugenics era and contemporary society (the availability of prenatal testing is quite different from laws mandating sterilization of “imbeciles”), some of the underlying impulses are related—discomfort with imperfection in ourselves, our children and society.<sup>106</sup> A few women will admit their discomfort with disabilities.<sup>107</sup> Even more may describe concerns about the societal costs of bringing disabled children into the world.<sup>108</sup> Outside pressures exacerbate these attitudes. The genetics literature describes health maintenance organizations’ (HMOs’) considering the withdrawal of coverage for pregnancy or pediatric care if parents did not terminate affected pregnancies.<sup>109</sup>

Most women who choose prenatal testing, however, rarely describe discomfort with disabilities or concerns about the societal costs of genetic anomalies. Some may

be uncomfortable expressing such views to themselves, let alone others. Other women may have motivations, such as a desire to prevent suffering they believe a disabled child will experience.<sup>110</sup> Whatever the motivation, the undeniable, though frequently unvoiced reality is that prenatal testing offers a kind of quality control over our future children and society.<sup>111</sup> For some, it reflects an unwillingness to let things unfold naturally and a desire to control the process of reproduction, but it happens silently and without transparency. As I shall discuss in Part III, providers and patients are both reluctant to draw out these connections explicitly, reflecting the unease we all feel with such attitudes, even as they shape the decisions of many.

### C. THE LAW

As we have seen, the routinization of genetic testing reflects biases within the medical community as well as social norms. The law, however, looms large in this process, both directly and indirectly. The legal doctrine of informed consent shapes

105 Malinowski, *supra* note 62, at 1453. Factors such as desired number of children or stage of life when embarking on parenthood might influence one's desire for "normalcy." One might argue that the desire for perfection would be high among Western couples given the low birth rates. Such couples may take greater pains to achieve the "ideal" child if they plan on having only one or two children. See Ruth R. Faden et al., *Prenatal Screening and Pregnant Women's Attitudes toward the Abortion of Defective Fetuses*, 77 AM. J. PUB. HEALTH 288, 289 (1987) (noting that those whose ideal number of children was smaller "often were likely to view abortion as justified" when amniocentesis identified a neural tube defect). Similarly, women who delay childbearing may try harder to have the "perfect" child, since they have fewer "chances" than women who start earlier. On the other hand, one might argue that couples with fewer children may be less concerned with "perfection" because they have more energy and resources for a special-needs child than couples with several children. Additionally, women who delay child-bearing may be less interested in prenatal testing because they may be less willing to risk losing a pregnancy when they have less time to bear children.

106 Garland E. Allen, *Is A New Eugenics Afoot?*, 294 SCIENCE 59, 61 (2001).

107 ROTHMAN, *supra* note 49, at 60-61 (describing a woman who expressed particularly strong feelings of disgust regarding Down syndrome, using such adjectives as “idiot” and “mutant being.”).

108 *Id.* at 59-60 (discussing the benefits to society of using technology in this manner).

109 David T. Morris, *Cost Containment and Reproductive Autonomy: Prenatal Genetic Screening and the American Health Security Act of 1993*, 20 AM. J.L. & MED. 295, 308-10 (1994). One HMO tried to secure agreements that couples would terminate an affected pregnancy in exchange for the provision of prenatal testing. *Id.* In the most famous anecdote, an HMO refused to pay for the healthcare costs of a fetus diagnosed with cystic fibrosis. Only after public outcry did the HMO reverse its decision. Andrews, *supra* note 93, at 986.

110 *See* Malinowski, *supra* note 58, at 1472-74 (describing a couple’s plan to decide whether to terminate a pregnancy based on the severity of the condition and the child’s potential quality of life because they “could [not] watch a child suffer through life.”)

111 Rothman describes this way of looking at childbearing as a kind of commodification of children, a view of children as “products of conception.” *Id.* at 2.

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many aspects of the genetic counseling process. To be sure, the ethical norms of genetic counseling, quite apart from concerns about liability, inspire a commitment to informed consent. As mentioned earlier, the purpose of genetic counseling is to help patients make fully informed decisions. But the threat of malpractice motivates the profession to document the process with painstaking care, to develop detailed informed consent forms and to provide patients or physicians with detailed letters describing the counseling session.

Wrongful birth lawsuits,<sup>112</sup> a cousin of informed consent actions, are also legal specters in the genetic counseling world. Genetic professionals can potentially be held liable for birth defects in a patient’s child if they did not meet the standard of

care in offering prenatal testing to a high-risk patient, failed to diagnose a detectable defect through prenatal testing, or failed to inform the patient about a detected defect.<sup>113</sup> The causation element of these lawsuits directly links prenatal testing to the choice of termination: in order to succeed, the plaintiff must establish that she would have terminated the pregnancy, had she learned of the birth defect in time.<sup>114</sup> Because the legal standard of care in medical malpractice actions has traditionally been defined by medical professionals,<sup>115</sup> the prospect of wrongful birth lawsuits should not, in theory, affect the practice of genetic counselors. The existence of these suits, however, may create inadvertent pressures to push testing. If a provider persuades a patient to undergo testing, she reduces the chance of wrongful birth liability.<sup>116</sup> Indeed, liability concerns have in at least one important instance altered the standard of care. The best example is the profession's decision to offer maternal serum alpha-fetoprotein (MSAFP) screening to all pregnant women. MSAFP screening was first used to identify pregnancies at increased risk for neural tube defects, such as spina bifida or anencephaly.<sup>117</sup> The screening test measures a protein produced by the fetal kidneys, AFP. AFP is measurable in both amniotic fluid and the mother's serum. Tests can be performed in the second trimester to

<sup>112</sup> Three jurisdictions, California, New Jersey and Washington, also recognize wrongful life lawsuits. 2 BARRY FURROW, ET AL., *HEALTH LAW* § 19-5, at 473 (1995). In these lawsuits, the plaintiff is the child born with the birth defect. In essence, the plaintiff claims that she has been damaged by being born with the birth defect. Implicit is the idea that the plaintiff would have been better off never having been born. *See id.* at 462.

<sup>113</sup> Ellen Wright Clayton, *What the Law Says about Genetic Testing and What it Doesn't*, in *WOMEN AND PRENATAL TESTING: FACING THE CHALLENGES OF GENETIC TECHNOLOGY* 131, 139

(Karen H. Rothenberg & Elizabeth J. Thomson eds., 1994).

114 *Id.* at 140.

115 FURROW ET AL., *supra* note 112, § 6-2, at 362.

116 Malpractice claims for wrongful birth are more prevalent than claims for failure to inform of the risk of miscarriage from amniocentesis, probably because providers of amniocentesis are so careful to discuss and provide consent forms that describe the risks of the procedure. *See, e.g.*, *Bedel v. Univ. of Cinn. Hosp.*, 669 N.E.2d 9 (Ohio App. 10 Dist. 1995) (affirming the dismissal of an informed consent action alleging the physician's failure to discuss the risks of the miscarriage that followed amniocentesis on the grounds that the patient was verbally informed of the risk and signed three consent forms). Similarly, causes of action for incorrectly diagnosing a fetal abnormality that resulted in the patient's decision to terminate what was, in fact, a normal pregnancy are rare. *See, e.g.*, *Martinez v. Long Island Jewish Hillside Med. Ctr.*, 512 N.E.2d 538 (N.Y. 1987) (reversing the lower court's dismissal of a claim for emotional distress when physician erroneously diagnosed brain abnormalities, resulting in pregnancy termination; emphasizing, however, the unusual circumstances of the case: the plaintiff suffered emotional distress not because of what happened to the fetus, but because of the psychological injury from agreeing to an act she believed to be a sin except in unusual circumstances).

117 Studies later found an association between low MSAFP levels and increased risks of Down syndrome in women under thirty-five years of age. Sherman Elias et al., *Carrier Screening for Cystic Fibrosis: A Case Study in Setting Standards of Medical Practice*, in *GENE MAPPING: USING LAW AND*

*ETHICS AS GUIDES* 186, 196 (George J. Annas & Sherman Elias eds., 1992).

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measure MSAFP levels. When a fetus has an open neural tube defect, higher amounts of AFP will circulate in the amniotic fluid and maternal serum. As a result, elevated MSAFP levels are an indication of pregnancies at increased risk for neural tube defects.<sup>118</sup>

MSAFP screening, however, presents numerous challenges. First, it is a *screening* test (i.e., it cannot diagnose defects, it can only identify high-risk pregnancies). MSAFP levels can be elevated for reasons other than neural tube defects, such as erroneous estimations of gestational age, multiple pregnancies, increased risk of premature delivery, fetal death and other abnormalities.<sup>119</sup> If an ultrasound provides no explanation for elevated levels, an amniocentesis is recommended. Only about 5-10% of those who undergo amniocentesis will have elevated amniotic AFP levels, most of which are associated with neural tube defects.<sup>120</sup> Although 70-90% of fetuses with neural tube defects can be identified through MSAFP screening, the vast majority of pregnancies with positive (elevated) MSAFP levels will not have a fetal abnormality. In other words, the false positive rate is quite high.<sup>121</sup>

In the early 1980s, ACOG stated that “routine maternal serum AFP screening of all [pregnant women] is of uncertain value.”<sup>122</sup> The organization was concerned that, without widespread availability of high-quality laboratory, counseling, ultrasound and amniocentesis services, “MSAFP could simply increase cost and parental anxiety . . . and possibly lead to unnecessary abortions.”<sup>123</sup> In other words, ACOG argued that MSAFP screening should not become the standard of care until “a coordinated system of care that . . . provide[s] a safeguard essential for ensuring prompt, accurate diagnoses and appropriate follow-through services” exists.<sup>124</sup>

Nevertheless, in 1985, before such a system was in place, ACOG’s Department of Professional Liability issued an “Alert” entitled “Professional Liability Implications of AFP Tests.”<sup>125</sup> In a climate of heavy malpractice litigation in obstetrics, the Alert declared that it was “*imperative* that every prenatal patient be advised of the availability of this test and that [member physicians’] discussion about the test and the patient’s decision with respect to the test be documented in the

patient's chart.”<sup>126</sup> As a result, MSAFP screening for Down syndrome became the

118 *Id.* at 195-96.

119 *Id.* at 196.

120 *Id.*

121 MSAFP screening, when conducted alone for Down syndrome, had worse statistics. The test could identify only 30% of such pregnancies, though the vast majority of low MSAFP levels were not associated with Down syndrome or other abnormalities. Personal communication with Suzanne Diment, R.N., Henry Ford Hospital (April 19, 2002). Better screening tests for Down syndrome have replaced simple AFP screening. The triple screen test, which analyzes AFP, human chorionic gonadotropin and unconjugated estriol, is typically used. Newer tests include the quadruple screen, which combines analysis of inhibin A (a hormone) with the triple screen test, and the integrated screen, which combines the quadruple test with ultrasound analysis. These newer screening tests identify more cases of Down syndrome and have lower false positive rates than MSAFP screening alone. *See* Brody, *supra* note 16, at F6. For example, depending on cut-off levels, the false positive rate with the quadruple test is around 4-6%, with a detection rate of 76% (91% for women over thirty-five years of age). Personal communication with Suzanne L. Diment, R.N., Henry Ford Hospital (April 19, 2002).

122 Elias et al., *supra* note 117, at 196-97 (citing AMERICAN COLLEGE OF OBSTETRICIANS AND GYNECOLOGISTS (“ACOG”), Technical bulletin No. 67, *Prenatal Detection of Neural Tube Defects*, ACOG, Washington, D.C., 1982).

123 *Id.* at 196.

124 *Id.* (emphasis omitted).

125 *Id.* at 197.

126 *Id.* (emphasis added).

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standard of care,<sup>127</sup> not for medical reasons, but in response to liability concerns.<sup>128</sup>

Although the MSAFP story is not representative of how the standard of care is

generally devised within the genetics/obstetrics community, it powerfully illustrates the way the law can shape medical practice.<sup>129</sup> By the early 1990s, approximately 65% of women in prenatal care had had MSAFP screening.<sup>130</sup>

The MSAFP story has another legal twist that has affected patient and provider attitudes. In response to a national goal of encouraging at least 90% of pregnant women to be offered “screening and counseling on prenatal detection of fetal abnormalities,” California legally mandated that all healthcare providers *offer* MSAFP screening to all pregnant women.<sup>131</sup> As one excellent study has shown, this legal obligation profoundly influenced the way in which providers offered, described and discussed MSAFP screening.<sup>132</sup> Since the clearest evidence of compliance is to have a patient take the test, the legal mandate prompted many healthcare professionals, at a minimum, to *encourage* MSAFP screening. Several practitioners viewed the mandate as “tantamount to a requirement that they test all their pregnant patients.”<sup>133</sup> Some tried to make it difficult for women to refuse by telling them to take the test. Others raised the topic several times when patients refused so they could document the repeated refusals,<sup>134</sup> undoubtedly creating some pressure to accept the test. In addition, medical centers instituted various mechanisms to

<sup>127</sup> Interestingly, although the Food and Drug Administration approved the use of MSAFP screening for neural tube defects in 1983, it never approved the use of MSAFP screening for Down syndrome. *ASSESSING GENETIC RISKS*, *supra* note 3, at 79.

<sup>128</sup> Ironically, by encouraging providers to adopt AFP screening as part of the standard of care, ACOG actually made wrongful birth claims based on a failure to offer MSAFP screening much more viable. Some have speculated that the MSAFP story was the legal liability department’s reaction to a current case at the time, *Helling v. Carey*, 519 P.2d 981 (1974), in which the Supreme Court of Washington defined the legal standard of care, not based on the medical standard, but instead based on the “reasonable prudence” test. Elias et al., *supra* note 117, at 197-98. In *Helling*, the defendant

physicians, who had complied with the medical standard of care not to screen for glaucoma in patients under forty, were found to have deviated from the “reasonable prudence” standard by failing to screen a patient under forty. Though *Helling* is one of only a few cases that reject customary medical practice, some have identified a general trend toward a general negligence standard, rather than a medical-based standard, of care in medical malpractice. See Philip G. Peters, Jr., *The Quiet Demise of Deference to Custom: Malpractice Law at the Millennium*, 57 WASH. & LEE. L. REV. 163 (2000).

129 Ultimately, ACOG’s scientific committees did recommend that MSAFP screening be routinely offered to all pregnant women. Elias et al., *supra* note 117, at 198. When the cystic fibrosis gene was identified, genetic professionals raised concerns that fear of medical liability would drive practitioners to offer widespread cystic fibrosis screening to couples planning pregnancy, even at a time when the testing offered only limited information. See Benjamin S. Wilfond & Kathleen Nolan, *National Policy Development for the Clinical Application of Genetic Diagnosis Technologies: Lessons from Cystic Fibrosis*, 270 JAMA 2948, 2949 (1993). Cautioned by the MSAFP story, the profession had studiously avoided adopting carrier testing for cystic fibrosis as part of the standard of care until evidence suggested the test was sufficiently informative. Only now, more than ten years after the gene’s identification, has the profession been persuaded that the test is ready for widespread availability. See *supra* text accompanying note 103.

130 Press & Browner, *supra* note 85, at 202.

131 CAL. CODE REGS. Tit. 17, §. 6527 (2002).

Clinicians shall provide or cause to be provided to all pregnant women in their care before the 140th day of gestation, or before the 126th day from conception, as estimated by medical history or clinical testing, information regarding the use and availability of prenatal screening for birth defects of the fetus. This information shall be in a format to be provided or approved by the Department [of Health] and shall be given at the first prenatal visit and discussed with each pregnant woman.

*Id.*

132 Press & Browner, *supra* note 85, at 202.

133 *Id.* at 205.

134 *Id.* at 205.

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encourage healthcare providers to push AFP screening.<sup>135</sup>

In addition, the study found that providers limited the discussion of AFP screening to no more than two minutes and described it as a “simple blood test.”<sup>136</sup> Little, if anything, was done to emphasize that the test was voluntary. Instead, a great deal was done to routinize it. Providers tended to say almost nothing about the purpose of the test.<sup>137</sup> In forty observed sessions, they offered only the most general descriptions of the conditions for which screening was done. None of the providers discussed the decisions a woman might confront in the event of a positive diagnosis, and pregnancy termination was discussed only twice.<sup>138</sup>

In short, so little time was devoted to AFP screening and patients were told so little about its purpose that patients rarely had questions, even when given the chance to voice them.<sup>139</sup> In response to the legal mandate, providers seemed more interested in *persuading*, rather than *informing*, patients. Not surprisingly, the study found very high rates of acceptance when compared with the national average.<sup>140</sup> If the goal was getting people to take the test, the program succeeded. If the goal was ensuring fully informed, voluntary decision making, in which patients are made aware and given a chance to consider the personal implications of their choice, the program seemed a dismal failure.<sup>141</sup>

When providers have biases that favor prenatal testing, when social norms promote such testing and when the law directly or indirectly encourages testing, it should come as no surprise that prenatal testing and screening have become so much a part of the modern experience of pregnancy. Is this routinization merely a

reflection of contemporary attitudes toward knowledge and control over reproduction or does it also present costs to patients and society? The next section argues that the side effects of routinization pose problems for patients and society in numerous ways.

### III. THE COSTS OF ROUTINIZATION

To avoid being misunderstood as opposing prenatal testing, let me emphasize that I believe that the widespread *availability* of genetic testing and screening is a good thing. Indeed, it is welcomed by many women, couples and healthcare providers. If patients are to have true choice and autonomy in reproductive decision making, then such tests must be offered and made available to all.<sup>142</sup> My concern

135 Centers used such measures as circulating memos stamped “Think AFP” and encouraging providers to contact women who missed AFP appointments to discuss their reservations. *Id.*

136 *Id.* at 206.

137 *Id.* at 206-07.

138 *Id.* California also developed an AFP booklet to be given to all prenatal patients. The booklet was similarly vague about the conditions AFP screening might detect. Moreover, nothing in the booklet indicated that the majority of these conditions cannot be treated except through pregnancy termination. It only stated that if the fetus has a birth defect, “different options will be discussed” and services are available to “support whatever decision the woman makes.” *Id.* at 208-09.

139 *Id.* at 206.

140 *Id.* at 202, 205. The study found acceptance rates of 85% compared with the national acceptance rate of about 65%. *Id.* at 216 n.10.

141 The authors provide clear evidence of just how poorly informed the patients actually were. None surveyed could adequately explain the conditions screened for. Less than one-third even recognized the term neural tube defect and, of those, only two-thirds had an accurate idea of what the term meant. Sixty percent recognized the term spina bifida, but only half of them could define the condition. Fewer than half of the surveyed patients knew what would happen next if the AFP test result

was positive, and more than one-third believed that the state required pregnant women to take the test.  
*Id.* at 209-10.

142 A key element to true choice with respect to prenatal testing is making the technology

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instead is with the *manner* in which prenatal testing is offered. The routinization of prenatal testing and screening is problematic on several levels: for healthcare professionals, pregnant women and society as a whole. For pregnant women, the routinization reduces, rather than expands, choice. In addition, it impoverishes the informed consent process: it lessens the possibility of choosing *not* to know by obscuring the crucial issues that underlie decision making in this area, such as what it means to terminate affected pregnancies. Finally, this routinization allows society to avoid openly discussing the meaning of prenatal testing and its moral implications, handicapping our ability to debate the value of new genetic technologies.

#### A. TRUE CHOICE OR THE ILLUSION OF CHOICE?

Perhaps the most striking effect of the routinization of prenatal testing and screening is that it has made the goal of promoting patient choice more a hope than a reality. With this routinization, true choice often gives way to the *illusion* of choice. As Ruth Hubbard has observed, when “‘choices’ become available, they all too rapidly become compulsions to ‘choose’ the socially endorsed alternative.”<sup>143</sup> Even when women understand conceptually that they have a choice, social norms and beliefs about what is best for their child may make choice illusive. According to a study of women who were offered AFP screening at an HMO in California, many women felt it would be wrong to refuse such testing because they thought it was for the benefit of their child.<sup>144</sup> If prenatal testing reflects good parenting and good judgment, how can one refuse it?

In some cases, patients actually believe they have no choice with regard to

prenatal testing or screening. I recall patients frequently explaining that they had been referred to our center because their physicians told them they “had to” have an amniocentesis.<sup>145</sup> Even if their physicians had only recommended prenatal testing, it is easy to see how they might interpret a physician’s advice as a mandate in light of social attitudes about prenatal testing. The perception that one has no choice is even stronger and more prevalent with respect to prenatal *screening*. A study in Finland found that half of the women interviewed believed that AFP screening was routine or “self-evident.” Only one-quarter felt that they actively participated in the decision whether to undergo prenatal screening.<sup>146</sup> In addition, in California, where healthcare providers have a legal obligation to offer MSAFP screening, more than one-third of women interviewed believed (or suspected) the state *required* pregnant women to undergo screening. That everyone does not have equal access to healthcare generally and genetic testing specifically is not news: it is estimated that over forty million Americans are uninsured. Milt Freudenheim, *Coalition Forms to Reverse Trend of Fast-Rising Ranks of Uninsured Americans*, N.Y. TIMES, Feb. 9, 2002, at A12. *See also* Clayton, *supra* note 97, at 134-38 (describing problems of access to genetic services). The problem of unequal access to genetic technology, important though it is, is beyond the scope of this Article.

143 Ruth Hubbard, *Some Legal and Policy Implications of Recent Advances in Prenatal Diagnosis and Fetal Therapy*, 7 WOMEN’S RIGHTS L. REP. 210 (1982).

144 ROTHMAN, *supra* note 49, at 11 (“In gaining the choice to control the quality of our children, we may rapidly lose the choice not to control the quality, the choice of simply accepting them as they are.”); Nancy Press & C.H. Browner, *Why Women Say Yes to Prenatal Diagnosis*, 45 SOC. SCI. & MED.

979 (1997) (reporting that a study of AFP screening at an HMO in California showed that most women felt obligated to undergo prenatal screening for the benefit of their child and because it was wrong to refuse testing).

145 See Anderson, *supra* note 65, at 128 (noting that in this study, “[patients’] initial reaction was to assume that it is a medically necessary procedure”).

146 Paivi Santalahti et al., *Women’s Decision-Making in Prenatal Screening*, 46 SOC. SCI. & MED. 1067, 1069-70 (1998).

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take the test.<sup>147</sup>

The combination of the manner and setting in which prenatal testing or screening is offered and social attitudes about the value of knowledge not only routinizes testing, but also makes choice largely illusory. This result is deeply problematic because, without a sense of choice, patients may not have contemplated larger questions raised by prenatal testing and screening such as: How much information do they want about the fetus? What will they do with the information? How will the information and process of testing affect their experience of pregnancy? Patients who believe they have no choice may find themselves unexpectedly on a path of decision making for which they are completely unprepared.

#### B. FAILED INFORMED CONSENT: LACK OF PREPARATION FOR DIFFICULT DECISIONS AND EMOTIONAL ISSUES

The lack of preparation for decisions that prenatal testing may present arises both because choice is illusory and because patients and many providers tend to divorce the process of testing and screening from some of its larger implications. The more routine a test becomes, the less patients and providers focus on such issues. The result can be that many women are unprepared for anxiety and distress when they are suddenly faced with difficult choices. Some may even discover that they would not have undergone testing had they understood its larger implications. For some of these women the information from prenatal testing can be toxic—it can cause them more harm than good. The failure of providers to tie important considerations to the

decision to begin prenatal testing is quite simply a failure of informed consent. In particular, it falls far short of the genetic counseling model of informed consent, which strives to encourage decisions consistent with one's goals and life plans. But, as some scholars have suggested, this failure may sometimes be collective,<sup>148</sup> patients may be complicit in refusing to press the larger questions. This section will explore the issues that are often divorced from testing decisions.

### 1. The Purpose of Testing: Prenatal Testing and Termination

As noted above, some patients have a clear sense of the personal value of prenatal testing; for them the benefits clearly outweigh the limitations. Others have as strong a sense of its lack of value, finding that the scales tip the opposite way. A good many patients, however, often fail to articulate why they choose prenatal testing. When I was counseling I was struck by how rarely patients developed their claims that prenatal testing was best for the fetus. It was not clear whether patients had fully explored the questions that immediately followed. What made prenatal testing best for the fetus in *this* context? Had they connected pregnancy termination with doing what was best, and if so, how? Did they believe that a child with Down syndrome, for example, would suffer so severely that non-existence was preferable to existence; had they even considered that question? How much did they know about Down syndrome children and their experiences? How much did their statements reflect their views of what is best for them or society? Did they see a complicated connection between their lifestyle and the child's happiness; i.e., were they concerned that their lifestyle would prevent them from giving a disabled child all that he or she would need to have a fulfilling existence or that a disabled child would disrupt their lifestyle or their ability to care for other family members? And how would a choice

147 Press & Browner, *supra* note 85, at 209-10.

148 *Id.* at 214.

to terminate or not terminate a pregnancy affect them emotionally?

When patients told me only that they wanted prenatal testing to do what was best for their child, all of those questions leapt to mind. Yet I confess, I held my tongue. Committed to nondirectiveness as I was, such questions seemed like challenges.<sup>149</sup> Since leaving genetic counseling and thinking more critically about nondirectiveness, however, I believe I should have raised all of those questions if my goal was to help them choose what was best for them. Those kinds of questions press precisely to the heart of what genetic counseling is supposed to achieve—decision making based on full consideration of one's values, moral views, preferences and life-plans.<sup>150</sup>

Yet often counseling focuses on the scientific information without directly introducing some of the more sensitive issues of prenatal testing, such as the possibility of pregnancy termination. Indeed, the biggest “elephant in the room” during discussions of prenatal testing, ironically, is termination. Not only do patients avoid the issue, providers also tend to minimize it. In the context of prenatal screening, this tendency is greatest for two reasons. First, patients are one step further removed from the possibility of facing such a choice.<sup>151</sup> Second, the test is most likely to be offered by non-genetic counselors, who tend to spend less time discussing testing generally. One study, for example, demonstrated that healthcare providers in California almost never mentioned the possibility of pregnancy termination when discussing MSAFP screening.<sup>152</sup>

Even genetic counselors who offer more comprehensive information are uncomfortable emphasizing termination. In her research, Rothman observed that genetic counselors often obscured the issue. Some did not discuss termination unless it was raised, and “almost no counselors discuss[ed] how the abortion would be done

in a pre-amino session, ‘unless [patients] ask[ed].’”<sup>153</sup> Although genetic counselors are trained to inform patients that most conditions detectable through prenatal testing are untreatable and to discuss the option of pregnancy termination, Rothman’s description rings true. Genetic counselors do not tend to dwell on this issue, largely because most patients will not face those heart-wrenching choices. Most will be reassured. Genetic counselors may believe that an overemphasis on termination will give the wrong impression about the degree of risk that couples face. Perhaps they worry about creating anxiety. For many genetic counselors, the focus is on providing testing to offer reassurance or preparedness.

While most couples will ultimately receive good news and reassurance from prenatal screening or testing, a small percentage will not. It is this percentage who

149 By contrast, I have no problem raising such questions in my law school classes. The norms of law school teaching entail pressing students to think about the full implications of their statements to see how far an argument goes and what underlies it. The emphasis on nurturing and support in genetic counseling and, especially, neutrality can make the goal of helping patients clarify their values and true preferences difficult because pressing questions like these could be viewed as critical, judgmental, directive and overly challenging.

150 As noted above, see *supra* note 23, I am skeptical that the rigid adherence to neutrality that many advocate in defending nondirectiveness can achieve that goal. Certainly not all counselors interpret nondirectiveness so rigidly. See Malinowski, *supra* note 62, at 1468 (describing the minority view among genetic counselors that it is not nondirective to express one’s view that a “moral and ethical judgment [is] okay for one situation but not for another”).

151 If patients have an abnormal screening result, they would then have the option of diagnostic testing, which might indicate that everything is all right.

152 Press & Browner, *supra* note 85, at 207.

153 ROTHMAN, *supra* note 49, at 39 (describing her “distinct impression that the client had no

idea what [prenatal testing] was all about,” in several instances, even when the client was well educated and middle class).

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will face difficult choices and struggle emotionally with distressing news. If the purpose of testing is to provide information of value to a patient, and if for some patients part of the value is having the option to terminate the pregnancy, providers should be direct about this option and its emotional costs. On the other hand, prenatal testing information may not be valuable to everyone. For those for whom termination is not an option, learning that the fetus has an untreatable anomaly might cause more harm than good. Although some might want the information to prepare for the birth of the child, others might feel it would cause too much stress during the pregnancy. Such patients can avoid what is for them potentially “toxic” knowledge, if they understand that termination is the only way to eliminate the condition for which prenatal testing is offered. Counselors should therefore be willing to discuss the relationship of pregnancy termination to prenatal testing and to urge patients to consider whether termination is a viable option and what it would mean to them. To relegate termination to the fringes of the discussion is to ignore the reality that it is one of the possible reasons for prenatal testing, in addition to reassurance and preparedness. Worse, it leaves patients who will face these hard questions unprepared.

If the connection between prenatal testing and termination is given too little explicit attention, the psychological ramifications of choosing to terminate a pregnancy for fetal abnormalities—therapeutic abortions, as they are sometimes called—are given virtually none. This omission is surprising given evidence that emotional distress, depression and marital stress are reactions in a significant portion of women and couples who have chosen to have “therapeutic abortions,”<sup>154</sup> which are

thought to be more emotionally difficult than abortions for personal reasons, i.e.,

“social” abortions.<sup>155</sup>

154 Aliza Kolker & B. Meredith Burke, *Grieving the Wanted Child: Ramifications of Abortion After Prenatal Diagnosis of Abnormality*, 14 HEALTH CARE FOR WOMEN INT’L 513 (1993) (describing grief, mourning and marital problems in women who went through therapeutic abortions); M.C.A. White-Van Mourik, *The Psychosocial Sequelae of a Second-Trimester Termination of Pregnancy for Fetal Abnormality*, 12 PRENATAL DIAGNOSIS 189, 192-95 (1992) (noting that eighteen months after therapeutic abortion, 20% of women were still depressed and 71% observed a change in their relationships; 12% separated). One self-described “semi-anecdotal” study found signs of emotional distress in twelve women interviewed three to forty-nine months after having a therapeutic abortion. P. Donnai et al., *Attitudes of Patients after “Genetic” Termination of Pregnancy*, 282 BR. MED. J. 621, 622 (1981). Seven made good emotional recovery, three fair and two disturbing and distressing recovery. *Id.* Another study of families who chose therapeutic abortions found depression was prevalent in most couples who chose therapeutic abortions. Only 2/13 of women and 4/11 of men failed to mention depression in describing their reactions. Bruce D. Blumberg et al., *The Psychological Sequelae of Abortion Performed for a Genetic Indication*, 122 AM. J. OBSTETRICS & GYNECOLOGY 799, 805-06 (1975).

155 Blumberg et al., *supra* note 154, at 805. Another study asserts that early social termination of unwanted pregnancies is not that hard, though they provide little empirical data to support that sweeping claim. P. Donnai et al., *supra* note 154, at 622.

Numerous factors explain why therapeutic abortions are more difficult than social abortions. One difference is in the timing. Social abortions tend to be conducted in the first trimester, whereas—until CVS became more widely used—therapeutic abortions were conducted in the second trimester, after amniocentesis results were available. Abortions in the second trimester are more difficult than earlier abortions both emotionally and physically. Women undergoing second trimester abortions have often

felt fetal movement, which can create a powerful emotional bond with the fetus; many may have also bonded with the fetus through ultrasound images; and their pregnancy has become evident to others, making the termination decision potentially more public. Pryde et al., *infra* note 163, at 503. Fantasies about the fetus and the future child's sex, appearance and talents are more likely in the second than first trimester, in part, because of the emotional bonding that has occurred. Blumberg et al., *supra* note 154, at 806. Moreover, first trimester abortions are safer, easier to perform and less emotionally and physically demanding. Blumberg et al., *supra* note 154, at 807; Pryde et al., *infra* note 163, at 503. Therapeutic abortions also end what is often a desired or otherwise wanted pregnancy, resulting in a

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In spite of the emotional trauma of therapeutic abortions, one study found that “77 per cent of the families . . . reported that they would again opt for amniocentesis and, if indicated, selective abortion in any future pregnancy.”<sup>156</sup> This means, however, that roughly one quarter of the study population would not necessarily have made the same choice. Surely then, the risk of emotional distress is both prevalent and powerful enough to warrant disclosing it to patients. Yet as one study has observed, although families “received excellent counseling regarding the genetic and technical facets of amniocentesis,” counseling did not include a discussion of “the psychological aspects of the [termination] procedure.”<sup>157</sup> The authors rightly conclude that failure to educate patients about these risks “precludes a fully informed decision.”<sup>158</sup> In light of the recent concern about the potential anxiety and stress associated with late-onset testing,<sup>159</sup> it is surprising that so little attention has been given to the emotional costs of therapeutic abortions. The reluctance to address the emotional aspects of termination is more evidence of the discomfort professionals experience in linking prenatal testing to termination.

Although one arguably cannot fully know how one will react to the terrible news that their fetus has an untreatable abnormality, a fully informed decision should

contemplate that scenario. In offering testing to patients, providers should therefore ask patients to consider the value of this information to them. Would learning about a birth defect prior to delivery be helpful or not? Are they undergoing testing primarily to be reassured? If so, are they prepared for the possibility, though slim, of getting bad news? And do they realize that normal chromosomes and AFP do not guarantee a healthy baby? Is pregnancy termination an option they would consider and, if so, what would it mean to them? And finally, how does the value of information to them balance against the small, but real, risk of miscarriage from undergoing amniocentesis and CVS?<sup>160</sup> Although these questions may be difficult, their consideration is crucial if the goal is to ensure truly informed decisions.

## 2. The Implications of Prenatal Screening

Although prenatal screening is one step removed from diagnostic testing, it deep sense of loss. M. Di Gusto et al., *Psychological Aspects of Therapeutic Abortion After Early Prenatal Diagnosis*, 18 CLINICAL & EXPERIMENTAL OBSTETRICS & GYNECOLOGY 169, 172 (1991)

(reporting that 42% of pregnancies in which fetal abnormalities were found were planned and 36% were desired; only 22% were completely by chance, but by the time of diagnosis had become acceptable). In contrast, social abortions, by definition, are based on the undesirability of pregnancy. Terminating pregnancies because of genetic defects also carries for many a sense of guilt for having passed, albeit unintentionally, a genetic trait to the fetus—couples may feel they have “caused” the abnormality. Blumberg et al., *supra* note 154, at 806.

<sup>156</sup> Blumberg et al., *supra* note 154, at 807.

<sup>157</sup> *Id.* at 807-08.

<sup>158</sup> *Id.* at 808. The authors also advise easier termination, earlier termination and post-abortion support. *Id.*

<sup>159</sup> See Huggins et al., *supra* note 28, at 514-15.

160 The medical profession has tended to view this balance in rather simplistic terms; it compares the numerical risk of abnormality with the risk of complications. Indeed, it is precisely because the risk of miscarriage was lower than the risk of Down syndrome in women over thirty-five that amniocentesis was routinely offered to those women. Asking individuals to compare the risks of procedural complications with the risk of having an affected fetus, however, has always struck me as an “apples and oranges” kind of comparison. This pure number comparison assumes that couples weight a pregnancy loss equally to having a child with a birth defect. The comparison, however, depends on much more than the statistical risk. It depends on one’s personal valuation of obtaining the knowledge and not losing the pregnancy. If anything is personal, surely it is the weight a woman or couple would assign to either negative outcome.

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raises many of the issues discussed above. In addition, for many women, it can create anxiety. Not only do patients who choose prenatal screening take one step down the path of gathering fetal information and all that that implies, but they also face the complications of a screening, as opposed to diagnostic, test. Because the goal of screening tests is to identify as many high risk patients as possible, they pose a substantial risk of false positives—abnormal screening results that do not correspond to fetal anomalies.<sup>161</sup> The fact that providers tend to view prenatal screening as routine and to gloss over its implications, emotional risks and limitations, means that patients often experience unexpected anxiety when they undergo prenatal screening.

A positive (abnormal) screening result can cause anxiety in a few ways. The mere fact of having an “abnormal” test result, not surprisingly, can cause significant distress.<sup>162</sup> For women who do not understand the distinction between screening and diagnostic tests,<sup>163</sup> the anxiety levels may be even higher because they may misinterpret the positive result as a definitive diagnosis of Down syndrome or spina

bifida. Some may not have fully understood that if prenatal screening results are positive, then further testing, such as amniocentesis, may be the only way to determine whether the abnormality actually exists. If they are reluctant to undergo prenatal diagnostic testing, then they may face a very difficult choice about whether to have an amniocentesis or continue the pregnancy with a high risk. If they ultimately reject diagnostic testing, the rest of the pregnancy will be fraught with tension, anxiety and worry.<sup>164</sup> For women who would not choose diagnostic testing, all of the anxiety might have been avoided by refusing the screening test.<sup>165</sup> For

<sup>161</sup> See *supra* text accompanying *supra* note 119. The risk of false positives with newer screening tests is lower. See sources cited *supra* note 121. How patients react to these new tests has been less well explored than their reactions to MSAFP screening.

<sup>162</sup> Theresa M. Marteau et al., *The Psychological Effects of False-Positive Results in Prenatal Screening for Fetal Abnormality: A Prospective Study*, 12 *PRENATAL DIAGNOSIS* 205, 211 (1992).

Receiving an abnormal AFP result on a routine screening test is associated with extremely high levels of maternal anxiety, as high as patients with a diagnosis of generalized anxiety disorder and higher than patients the night before major surgery. This high level of distress is reflected in increased worry about the baby's health and a more negative attitude towards both the baby and the pregnancy.

*Id.* (citations omitted).

<sup>163</sup> See Peter G. Pryde et al., *Prenatal Diagnosis: Choices Women Make About Pursuing Testing and Acting on Abnormal Results*, 36 *CLINICAL OBSTETRICS & GYNECOLOGY* 496, 499 (1993)

(noting

that whether this poor understanding is the result of "policies of 'universal screening,' in which an inadequate effort has been made on the part of prenatal care providers to educate and provide informed consent at this level, or whether it is a counseling psychology issue is not yet known").

[T]here are data to suggest that a majority of women choose to be screened despite a

remarkable rate of participants demonstrating, in exit interviews, a poor understanding of the [screening] program. Many patients appear to have a poor grasp specifically about the concept of screening, the value and limitations of the information being sought, the meaning of a positive test, and the potential pregnancy decisions that might be faced in the rather common event of a positive screen result.

*Id.*

164 “Extremely high levels of anxiety have been found at the time of the test result and some weeks later in women who receive an abnormal result on first, but not subsequent testing” Theresa M. Marteau et al., *Anxiety, Knowledge and Satisfaction in Women Receiving False Positive Results on Routine Prenatal Screening: A Randomized Controlled Trial*, 14 J. PSYCHOSOMATIC OBSTETRICS

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GYNAECOLOGY 185, 187 (1993) (citing four studies with such findings). One study found that women who chose diagnostic testing after receiving a positive AFP screening result experienced less anxiety than those who do not. Moreover, the anxiety of those who did not choose diagnostic testing extended into the post-partum period. Marteau et al., *supra* note 162, at 211.

165 I must emphasize that this is not to suggest that the screening test is inappropriate, but rather that it is not appropriate for all people. Some people, if they fully understand the distinction between THE ROUTINIZATION OF PRENATAL TESTING 261

them, the knowledge from prenatal screening can be more harmful than beneficial. If uninformed of the implications of a screening test, however, these women find themselves in a tough emotional bind—should they take a test they would not have chosen or continue the pregnancy filled with anxiety?<sup>166</sup>

The majority of women with false positives, however, do choose diagnostic testing.<sup>167</sup> One might expect that normal amniocentesis results would reduce their anxiety and stress. And indeed, one study has found that to be true.<sup>168</sup> My own experience and that of other studies, however, indicates that many women remain

anxious even after receiving normal amniocentesis results or conversely, that subsequent normal results are not completely reassuring after having had a positive screening result.<sup>169</sup> I came to this conclusion incidentally, and somewhat anecdotally, when doing follow-up on patients from our clinic who had positive (abnormal) MSAFP results.<sup>170</sup> The majority had chosen amniocentesis and received normal results. Most had delivered normal healthy babies. A notable percentage,<sup>171</sup> much to my surprise, described a continued sense of anxiety and unease during the rest of the pregnancy. Some were still angry about their experience even after having delivered a normal, healthy baby.

The women I spoke with described a new sense of unease or anxiety that remained for the duration of the pregnancy, even when diagnostic results were normal. Many felt that the low or high AFP value had to mean that something was wrong, even if it was not detected through diagnostic testing. Some women seemed not to have fully understood the concept of a screening test and the idea of false positives, which contributed to their stress. Finally, some women may have learned that abnormally high screening results can indicate an increased risk of other screening and testing, might opt for no screening in the first place, because they know they would not undergo further prenatal testing. For these people, screening is particularly problematic if a positive result occurs. Others may not know how they would react if faced with a positive result, and they may therefore choose to undergo screening and deal with the issues if they arise.

<sup>166</sup> Research suggests that stress can have negative effects on pregnancy outcomes. One study, for example, has found that the stress of working during pregnancy can increase the risk of preeclampsia (pregnancy-related hypertension), which can cause complications in late pregnancy. *See* Jenny Hope, *Working While Pregnant "May Harm Mother and Baby"*, DAILY MAIL, Apr. 18, 2002, at 23. Another study, however, found that psychological stress at work was not related to preterm, low birthweight delivery, unless the working women did not want to remain in the work force. C.J. Homer

et al., *Work-Related Psychosocial Stress and Risk of Preterm, Low Birthweight Delivery*, 80 AM. J. PUBLIC HEALTH 173 (1990) (stating that “[p]ersonal motivation to work, as well as the physical effort of work, should be considered in evaluating the impact of a job’s psychologic characteristics on pregnancy outcome”). How these data bear on the pregnancy risks of anxiety caused by prenatal testing or screening is an empirical question worth exploring.

167 Marteau et al., *supra* note 162, at 211 (finding that eighteen out of twenty-six women with abnormal results underwent amniocentesis).

168 Barbara K. Burton et al., *The Psychological Impact of False Positive Elevations of Maternal Serum Alpha-Fetoprotein*, 151 AM. J. OBSTETRICS & GYNECOLOGY 77 (1985). The authors suggest,

however, that they may have used unreliable measures of pregnancy attitudes. *Id.* at 81-82.

169 Marteau et al., *supra* note 162, at 206 (citing other studies that have found this phenomenon when people receive false positives for other screening tests and later receive normal test results).

170 *But see* Pryde et al., *supra* note 163, at 500 (“Our own data suggest that, although anxiety is clearly increased in such women with results over baseline, it is only slightly greater than that experienced by women seeking counseling for advanced maternal age. After counseling it appears to be commensurate with comparable actual genetic risks. We speculated that careful education regarding the nature of the MSAFP screening program is the most critical factor modifying women’s perceived risks (and thereby, levels of anxiety) under these circumstances.”).

171 Unfortunately because the purpose of this study was to follow clinical outcomes, not psychological responses, we did not obtain precise statistics of psychological reactions. My descriptions are based on qualitative impressions from conversations I had with many women.

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complications, prolonging their initial unease.<sup>172</sup>

Empirical studies are largely consistent with my experience. One study found that, although most women who had diagnostic testing following positive AFP results

were reassured when the amniocentesis results were normal,<sup>173</sup> some women maintained high anxiety levels.<sup>174</sup> In addition, women who had had initial positive screening results experienced higher anxiety, even after having received normal results from subsequent testing, than those who had had initial normal results.<sup>175</sup> Unlike my observations, however, this study found no evidence that positive screening results had any effects later in pregnancy or during the post-partum period.<sup>176</sup>

Some of this anxiety could be avoided through careful, thorough genetic counseling, in which patients are informed of the distinction between screening and diagnostic tests and the possible need for further testing. Yet, because prenatal screening has become so routinized, such discussions are increasingly infrequent, leaving many women unprepared for the emotional costs and difficult dilemmas they may face. Campaigns, such as California's mandatory screening program, only intensify this problem. Even women who fully understand all of this information intellectually may face increased anxiety, believing that "something must be wrong."<sup>177</sup> As a result, the risk of anxiety should be part of the content of informed consent.

The failure to discuss these risks undercuts the goals of genetic counseling—to help patients make decisions consistent with their values, beliefs, circumstances and life plans. Genetic counselors have been described as more committed to patient autonomy than most other healthcare providers.<sup>178</sup> The focus on informing patients about the technical aspects of prenatal testing and genetic diseases as well as the commitment to nondirectiveness reflect a desire to help patients make decisions that are "intentional, substantially noncontrolled, and based on substantial

<sup>172</sup> See Joseph Green, *Prenatal Screening and Diagnosis: Some Psychological and Social Issues*, 97 BR. J. OBSTETRICS & GYNAECOLOGY 1074, 1075 (1990).

[I]n the case of AFP screening, the accumulation of data indicating that pregnancies with abnormal levels are at increased risk for a wide range of perinatal complications will only increase the fears of women with ‘false’ positive results, particularly when it is unlikely that this fore-knowledge can be used in any useful way. . . . The evidence linking anxiety in pregnancy with poor obstetric outcome should lead us to question whether we wish to create anxiety in . . . pregnant women in order to detect one neural tube defect, which arguably would, in any case, have been detected by a routine ultrasound scan.

*Id.* Some obstetricians, however, encourage MSAFP screening *because* elevated MSAFP levels are associated with an increased risk of fetal anomalies and they can monitor these pregnancies for negative outcomes. Personal communication with Christopher Grover, M.D. (June 4, 2002).

173 Of 372 women in the study, twenty-six had abnormal results. None of the women with abnormal screening results was found to have an abnormality on further testing and none had a child with Down syndrome or spina bifida. Marteau et al., *supra* note 162, at 208.

174 *Id.* at 213.

175 *Id.* at 206.

176 *Id.* at 209. Interestingly, a follow-up study by the authors found that women who received abnormal AFP screening results did not show any rise in anxiety either when they received the results or later. Marteau et al., *supra* note 164, at 192 (2d study). They offered several possible explanations: 1) clinical practice had changed since their prior studies or as a result of the more recent study, 2) the act of filling out the questionnaires reduced anxiety, 3) women didn’t fill out the questionnaires at the height of their anxiety and 4) the more anxious women did not complete the questionnaires. *Id.* at 194.

177 Marteau et al., *supra* note 162, at 213 (“[W]omen may believe that their initial abnormal result is indicative of some underlying problem: ‘no smoke without fire.’”).

178 Jeffrey R. Botkin, *Prenatal Screening: Professional Standards and the Limits of Prenatal Choice*, 75 OBSTETRICS & GYNECOLOGY 875 (1990) (“There is perhaps no area of medicine with a stronger commitment to patient autonomy than reproductive genetics.”).

understanding.”<sup>179</sup> One of the reasons genetic counselors advocate nondirectiveness is because of the nature of the decisions being made. Patients seeking reproductive genetic counseling are not typical patients who seek treatment of a disease. Instead, they face reproductive decisions that can affect life plans or evoke strong emotional responses with potential long-term effects on the individual and family.<sup>180</sup>

Although much of the literature on genetic counseling discusses the importance of these psychological elements, the reality, particularly as prenatal testing has become routinized, is that they have not become part of the informed consent process. If patients are truly to make decisions that are harmonious with personal values and preferences, the informed consent process<sup>181</sup> must prepare patients for the possible psychological and social ramifications of deciding to undergo genetic testing, including the anxieties that might arise and the range of difficult decisions patients may face. Only then can patients decide whether the knowledge from prenatal testing will be empowering or toxic for them.

This model of informed consent parallels the recommendations for late-onset testing, which urge disclosure of not only the technical aspects of the testing, but also the possible psychological reactions one may experience in the process. Some might criticize such a model on the grounds that disclosure of all of the risks and implications of prenatal screening will create anxiety in most or many women who are offered testing. Only an empirical study can answer that question. My sense is that if it does create anxiety, it would create lower levels of anxiety, though perhaps in more women. If the sole goal of genetic counseling is to reassure and reduce anxiety, then one would want to know whether the overall anxiety—measured by magnitude and incidence—is greater with or without these discussions. But reducing anxiety is not the sole goal. Instead, the goal is to help patients reach these important

decisions in ways that are consistent with their values and life plans. That task requires some difficult introspection, which may not be fully free of anxiety, but which one hopes will ultimately lead to the best decision for each patient. It is for these reasons that the same caution regarding the potential toxicity of knowledge from late-onset testing should exist with respect to prenatal testing.

### C. WHAT KIND OF KNOWLEDGE COUNTS?

In addition to impoverishing informed consent, the routinization of prenatal testing or screening also supports and reinforces an attitude about what sources of knowledge and information count. It suggests that the medical data we can obtain from prenatal screening, amniocentesis, chorionic villus sampling or ultrasounds are the only valid sources of information about the pregnancy. This emphasis on scientific knowledge, however, both overstates its reliability and accuracy and undervalues intuition or “gut feelings.”<sup>182</sup> Prenatal screening tests, for example, are

179 Gail Geller et al., *Genetic Testing for Susceptibility to Adult-Onset Cancer: The Process and Content of Informed Consent*, 277 JAMA 1467, 1468 (1997) (quoting P.S. APPELBAUM ET AL., *INFORMED CONSENT: LEGAL THEORY AND CLINICAL PRACTICE* (1987)).

180 Andrews, *supra* note 93, at 978-80, 982-84; Kessler, *supra* note 21, at 169-70 (discussing the development of the role of genetic counseling toward a psychological focus on the individual and family).

181 Informed consent, to be meaningful, really should be understood as a “an ongoing *process* that takes place while the emotional, physical and information status of the patient is changing,” rather than a “discrete act that takes place in a circumscribed period of time.” Nancy Press & C.H. Browner, *Risk, Autonomy, and Responsibility: Informed Consent for Prenatal Testing*, HASTINGS CTR. REP. May-

June 1995, at S9, S10 (emphasis added).

182 By intuition or “gut feelings,” I refer to the inner sense one might have about one’s health or

not diagnostic by definition. As a result, they miss some affected pregnancies (false negatives) and identify as “at-risk” many non-affected pregnancies (false positives).

Ultrasounds are also not fully diagnostic, as miraculous as they are in revealing images from the womb. They can identify some abnormalities with high levels of accuracy, but a lack of precision is inherent: ultrasounds sometimes lead to false diagnoses or miss serious abnormalities.<sup>183</sup>

Even amniocentesis and CVS, highly accurate diagnostic tests, are not infallible.

Human error can lead to incorrect results. In addition, these tests can produce ambiguous results. <sup>184</sup> A rarely found chromosomal rearrangement might be detected, the prognosis for which is uncertain—the child might be completely unaffected or it might suffer some complications or abnormalities. Even relatively straightforward diagnoses provide only limited information. An amniocentesis result of 47, XY, +21, indicates only that the fetus is male and will have Down syndrome.<sup>185</sup> It cannot predict how severely diminished the child’s mental capacity will be or whether he will have heart defects, intestinal obstructions or other complications. And the couple who receives the good news that a child has all forty-six chromosomes with no rearrangements still faces a 3-5% baseline risk of birth defects. They cannot be reassured that their child will not have undetectable forms of mental retardation, blindness, deafness or susceptibility to serious childhood illnesses.

In short, prenatal diagnostic screening and testing offers some information, but far from the complete predictive picture many patients imagine. None of these medical procedures alone or in conjunction can offer the full scope of reassurance that many people want. Medical and genetic technologies are simply far from omniscient. In spite of these limitations, providers, patients and the public tend to rely on medical knowledge as the most potent knowledge one could have about the

fetus.

Contrast such attitudes with those regarding intuition. Our faith in medical technology not only overvalues its powers, but also tends to negate the value of intuition. Women often describe strong intuitions about their pregnancies. How accurate this heightened sense of perception is regarding their fetus and bodies is not clear. We know it is not infallible, but the reliance on technology as the sole source of information about the fetus leaves little room for discussion of these alternative sources of knowledge. There is a tendency in our culture, particularly among well-educated women, to feel uneasy expressing or relying on such intuitions because they do not believe it is valid “knowledge.”<sup>186</sup> Many feel a need to validate their intuition body—a judgment based on imponderable evidence or “knowing without knowing how you know.”

Kathleen Doheny, *Playing a Hunch: Intuitive and Analytical Thought Styles Can Coexist and Strengthen Each Other*, ST. LOUIS POST-DISPATCH, Feb. 10, 1991, at PD14. The validity of this kind of

knowledge has not been widely studied; “intuition is by its nature difficult to study because whatever is happening is happening on a subconscious level.” Barbara Brotman, *How Do You Know?: If You Don’t Even Need to Read This Story, You Might Want to Anyway*, CHI. TRIB., Sept. 22, 1996, at CN1.

Nevertheless, some scientists are attempting to understand this phenomenon through scientific analysis. See, e.g., Rob Stein, *With New Findings, Neuroscientists Have a Hunch Intuition Makes Sense*, WASH. POST, Feb. 28, 1997, at A14 (describing a study that identified a part of the brain that seems to be required for intuition).

183 AMERICAN INSTITUTE OF ULTRASOUND IN MEDICINE, STANDARDS FOR PERFORMANCE OF

THE ANTEPARTUM OBSTETRICAL ULTRASOUND EXAMINATION 2 (1994) (stating that following the

standards will improve the chances of detection of abnormalities, but also noting that it is impossible to

detect all anomalies).

184 See Malinowski, *supra* note 62, at 1463-64 (discussing the possibility of ambiguous results).

185 Louis J. Elsas II, *A Clinical Approach to Legal and Ethical Problems in Human Genetics*, 39 EMORY L.J. 811, 834 (1990).

186 Of course, not all women feel that way. Rothman describes some women who felt they had

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with scientific data.

I felt this tension quite strongly as I struggled with decisions about whether to undergo prenatal testing or screening. Trained in two “rational” disciplines, genetics and law, I knew that an amniocentesis or CVS would be more accurate in determining whether my fetus had a chromosomal abnormality than any intuition I might have. Yet I found it hard to ignore my deep sense that everything was okay, even though a part of me found it a bit silly to credit my intuition at all. After all, what value did a feeling (based on nothing perceptible or scientific) have, particularly in contrast to the power of the technology? I recalled plenty of cases in which women were “sure” the fetus had a problem and were pleasantly proven wrong, and just as many who were “sure” everything was okay and were sorrowfully disappointed.<sup>187</sup>

How could I, an educated woman, rely on anything but scientific data to assess the state of my fetus? And yet, I found it hard to ignore my intuition. Ultimately, I decided we simply do not know enough to conclude that scientific knowledge alone has value and that intuition has none. My sense, though difficult to prove, was that each has value and each has limits, probably to varying degrees. In the end, I honored my intuitions, ever aware of their limitations.<sup>188</sup>

As prenatal testing and screening become routine, even less space is left for the recognition, let alone discussion, of intuition. Social norms pressure women not only

to be willing, but to expect, to learn about the fetus only from scientific sources.

Something is lost in the pregnancy experience when there is no longer a place for or opportunity to explore the intuitions that women experience.

#### D. PRENATAL TESTING VERSUS LATE-ONSET TESTING

As we have seen, the routinization of prenatal testing is problematic on several levels. One of the most serious concerns is that it treats prenatal genetic information as necessary and inherently beneficial. Yet as many realized with respect to late-onset testing, genetic information is not of equal value to everyone. For some individuals it can potentially be toxic; the discovery that one is at risk for a serious condition with no known treatments may cause more harm than good if, for example, it leads to psychological distress. Yet the term “toxic” knowledge has rarely, if ever, been used in the reproductive context. Why not, when prenatal testing can also reveal information about untreatable conditions (albeit in the fetus rather than the patient herself)? One explanation is that because late-onset testing is relatively new,<sup>189</sup> a layer of caution surrounds it. Similarly, late-onset testing has not yet become routine. Much of it is still offered primarily in research settings or at “alternative sources of knowledge,” which didn’t require scientific verification. ROTHMAN, *supra* note 49, at 72. One of the patients Rothman interviewed stated, “this may sound odd, but I already felt quite sure he was fine.” *Id.* at 52. Although this woman ultimately relied on her intuition in deciding not to be tested, the preface to her statement reflects a recognition that such knowledge is not socially valid.

<sup>187</sup> Sometimes patients, particularly less-educated patients, offered rather mystical explanations for events that seemed utterly to discount proven medical science. A baby was born with a birth defect, one patient, told me, because someone had blown smoke toward the woman’s pregnant womb.

<sup>188</sup> It turns out that my intuition that all was well with my baby was right. I gave birth to a healthy baby boy. My sense, especially in the beginning of the pregnancy, that I was carrying a girl, however, turned out to be wrong. My anecdote of course does not resolve the question of the validity of

intuition, particularly because the odds were clearly in my favor to have a healthy child and merely 50/50 that I would have a girl.

189 *See supra* text accompanying notes 24-25.

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genetics clinics.<sup>190</sup> Prenatal testing has, in contrast, become routine, not only within genetics centers but especially in obstetricians' office. The time devoted to counseling has gradually decreased over the years.

Another explanation for the relative difference is that medical professionals may view the two types of testing as substantively different. One of the key limitations of many forms of late-onset testing, such as HD testing, is that medical treatments or preventive measures are limited or nonexistent. In the reproductive context, one cannot cure most of the detectable conditions, but the option of termination exists. To put it quite crudely, termination "eliminates" the condition and, as a result, the medical profession views it as a form of treatment.<sup>191</sup> Indeed, the term "therapeutic abortion" connotes therapy and treatment. The idea that something like treatment is available in the prenatal setting, however, minimizes the enormity of deciding to end a pregnancy and overstates the notion of "treatment." It medicalizes a complex personal and social decision and unintentionally conveys a sense that fetuses are easily expendable. This is particularly problematic in light of evidence of the emotional costs of terminating a pregnancy and the fact that, for some, termination is not a viable option. Until we can truly treat the detectable fetal anomalies, rather than merely terminate pregnancies, the differences between late-onset testing and prenatal testing do not seem great.<sup>192</sup>

Indeed, both forms of testing share some similarities. Each provides information people might find helpful in making major life decisions. Prenatal test results may bear on decisions whether to continue or terminate a pregnancy or to

place an affected child up for adoption. These decisions may in turn affect other life choices, such as whether to have more children or to continue in a particular career.<sup>193</sup> Similarly, learning whether one carries a gene for a late-onset condition might influence major life decisions, such as whether to pursue or continue certain careers, to marry or to have children.<sup>194</sup> In addition, the information from both forms of testing can potentially influence relationships. Parents who pursue prenatal testing

<sup>190</sup> In the process of searching for genes and new mutations for late-onset conditions, researchers often recruit volunteers to undergo genetic testing. *ASSESSING GENETIC RISKS, supra note*

*3*, at 157. While such studies are underway or shortly after a new gene has been identified, nongeneticist physicians, most of whom have little training in genetics, are unlikely to be aware of tests for these conditions, much less to offer them routinely to patients. This is generally a good thing. Given their lack of genetics education, such physicians are likely to have difficulty interpreting the often complex data. Unfortunately, over time some of the same pressures that have contributed to the routinization of prenatal testing may develop in this area. Some believe that pressures from the biotechnology industry, fears of legal liability and growing public awareness of genetic tests for late-onset conditions may pressure non-geneticist healthcare providers to offer late-onset tests to patients. Malinowski & Blatt, *supra note 44*, at 1246-47. Indeed, based on the concerns that late-onset testing is particularly complex, the Committee on Assessing Genetic Risks recommended that counseling for such testing “be provided in a specialized genetics center familiar with the genetics and psychosocial aspects of the disorder in the context of pilot studies.” *ASSESSING GENETIC RISKS, supra note 3*, at 177.

<sup>191</sup> Anderson, *supra note 65*, at 129-30 (describing the “moral imperative” of testing that the medical profession adopts, in the view that it is for the good of the fetus, family, and society).

<sup>192</sup> Just as the possible toxicity of information from late-onset testing is not a reason for everyone to avoid genetic testing, neither is the potential toxicity of information from prenatal testing a

reason for everyone to avoid prenatal testing. But it is a risk about which patients should be informed.

193 For example, choosing to continue a pregnancy with an affected fetus may require additional financial and emotional resources. As a result, some couples may have fewer children or choose less demanding careers. Conversely, it might inspire some couples to have more children, in the hopes of having an unaffected child.

194 Learning that you have the gene for HD might make you less inclined to pursue a career in neurosurgery for fear that the chorea would impair your surgical abilities, for example. Conversely, you might develop extra drive because the information makes you want to live your life as fully as possible.

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for a condition in an existing child may experience guilt with respect to the affected child. Their decision might suggest that the affected child would not have been born had her parents used prenatal testing when pregnant.<sup>195</sup> Patients who learn they have a gene for a late-onset condition may experience guilt because of the possibility they passed the gene to their children.<sup>196</sup> Those who do not have the gene may suffer from survivor guilt if other family members are affected.<sup>197</sup> In short, decisions about prenatal and late-onset testing both influence and depend on life plans, personal values and relationships.

Despite these similarities, one might draw distinctions between late-onset and prenatal testing based on the degree of risk patients face. Individuals with a family history of HD may face a 50% risk of carrying the gene and a nearly 100% chance of developing the disease, if the gene is present.<sup>198</sup> A woman whose mother and sisters carry the BRCA1 or BRCA2 gene also has a 50% chance of carrying the gene and a cumulative risk of 35-87% of developing breast cancer by age seventy, if she has the gene.<sup>199</sup> In contrast, a woman who will be thirty-five when she delivers her child has a 1/250 risk of having a child with Down syndrome.<sup>200</sup> Clearly, the prenatal risk is

lower. In a profession that tends to view relatively low risks as high,<sup>201</sup> however, it would seem odd to distinguish such forms of testing on that basis.<sup>202</sup> Geneticists believe these lower risks are sufficiently high to warrant prenatal testing, even if it involves a small risk of miscarriage. The risk may be lower than 50%, but it is very real in the genetics community. In short, the knowledge in the prenatal context is also potentially toxic, even if the likelihood of a positive result is lower than with late-onset testing.

Perhaps the most important reason to treat prenatal testing with the same caution as late-onset testing is that many pregnant women experience some psychological vulnerability with respect to the well-being of their fetus. The possible psychological stresses associated with prenatal testing or screening therefore seem especially potent—as important to such patients as the risk of anxiety from late-onset testing. The elaborate informed consent process of late-onset testing therefore seems equally important in the reproductive context.<sup>203</sup> Indeed, the need for such counseling is heightened in light of social norms that pressure women to undergo testing and in light of the routinization that leaves many patients unaware of the full implications of such testing.

195 See Suter, *supra* note 27, at 1865. Similar conflicts can exist when a sibling has the condition in question. See Andrews, *supra* note 93, at 978.

196 *Supra* text accompanying note 39.

197 *Id.*

198 Because HD is a monogenic, autosomal dominant condition, the presence of the gene is predictive of disease. In contrast, definitive prediction is not possible through a single genetic test for multifactorial conditions. ASSESSING GENETIC RISKS, *supra* note 3, at 86.

199 See sources cited *supra* note 37.

200 ASSESSING GENETIC RISKS, *supra* note 3, at 158.

201 In recent decades, genetic counselors have become increasingly intolerant of risk. In 1973, three-quarters of genetic counselors surveyed considered a one percent risk to be “very low” or “low.” By 1990, a survey of genetic counselors found that only one in six respondents considered it “low.” Only a risk of one in five hundred was viewed by all genetic counselors as not “high.” MICHIE & CAHN, *supra* note 90, at 81.

202 Genetic counselors tend to perceive risk as more significant than the rest of the population. Rothman reported that half of the counselors she interviewed found that 1/50 is a high or very high risk. Only 75% view 1/400 as a low or very low risk. ROTHMAN, *supra* note 49, at 43.

203 Even if the stresses associated with each are different in degree (a debatable point), the stresses are similar in quality, which argues for similar counseling approaches.

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#### E. SOCIAL IMPLICATIONS

Not only does the routinization of prenatal testing or screening reduce choice for patients, it also has troubling implications for society at large. The social norms that make prenatal testing not only acceptable, but expected, have ramifications today and in the future. One concern is that these norms negatively affect the disabled community. As prenatal testing or screening becomes routinized our society devalues, or could be perceived as devaluing, those with disabilities. Although not everyone in the disability movement rejects abortion for disability-based reasons,<sup>204</sup> many are concerned that the routinization of prenatal testing can stigmatize the disabled, particularly if reproductive testing is promoted as a way to eliminate costly conditions.<sup>205</sup> The disabled community is also concerned that prenatal testing intended to prevent suffering reflects ignorance about the true impact of disabilities. Whereas society tends to believe many, if not most, disabilities significantly lower quality of life, the disabled community rejects those views. Many disabled people believe their quality of life is good. Much of the disability movement has tried to

foster awareness that the challenges of being disabled lie less with the disability itself than with “architectural, technological, legal, or attitudinal” barriers in society.<sup>206</sup>

The routinization of prenatal testing and, in particular, the failure to probe the deeper reasons people pursue such testing, impoverish society’s understanding of the meaning of disability and the role of the disabled in our community.

This routinization also tends to prevent a broader debate in our society about the “moral nature of information itself.” As Rothman points out, “we do not even have a language of morality that allows us to talk about the moral nature of information.”<sup>207</sup>

Medical professionals and society at large have tended to value information for its own sake, without much consideration of its costs to society or its moral dimensions.

The informed consent process for prenatal testing tends to focus more on the technical aspects of information, rather than on its psychosocial and moral aspects.

In other words, it medicalizes the information and the decision making process. This is not surprising because nondirectiveness is grounded in a form of moral relativism.

Genetic counselors are therefore very reluctant to view these choices as moral because they fear it will imply they are judging patient choices.<sup>208</sup>

To mask the moral implications of prenatal testing, however, perpetuates the sense that information is not only inherently beneficial, but also amoral. My choice of the word “amoral,” as opposed to “immoral,” is quite deliberate. I am not making claims about the morality of decisions whether to test a fetus or to terminate an affected pregnancy. It is difficult to assess the morality of such decisions without

204 Some individuals in the disability community support any form of abortion on autonomy grounds. Some want to preserve the right since women with disabilities may be more likely to require abortions for medical reasons. Deborah Kaplan, *Prenatal Screening and Diagnosis: The Impact on Persons with Disabilities*, in *WOMEN AND PRENATAL TESTING: FACING THE CHALLENGES OF GENETIC*

TECHNOLOGY 49, 51 (Karen H. Rothenberg & Elizabeth J. Thomson eds., 1994).

205 Some elements of society may view the disabled as an unfortunate drain on public resources.

Andrews, *supra* note 93, at 993. The former Surgeon General, Jocelyn Elders, has described abortions as having a “positive, public health effect” because they reduce the number of children born with severe birth defects. Dunne & Warren, *supra* note 46, at 172.

206 Kaplan, *supra* note 204, at 52.

207 ROTHMAN, *supra* note 49, at 83.

208 See generally Sonia Suter, *Sex Selection, Nondirectiveness, and Equality*, 3 U. CHI. L. SCH.

ROUNDTABLE 473 (1996) (describing the discomfort genetic counselors have in dealing with decisions they view as immoral, such as sex selection). Genetic counselors may be reluctant to treat prenatal testing decisions as moral decisions to distance themselves from the politically and ethically loaded specter of abortion, which looms large on the horizon of their work.

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understanding the context and reasons for them. Nevertheless, these are moral decisions because they are based in part on moral reasons. To ignore the moral content of these decisions minimizes their personal import and handicaps society’s capacity to deal with the moral questions prenatal testing raises.

The ability to address the moral issues of reproductive testing is crucial as technology develops alternative ways to control the traits in our children: from genetic modification to treat disease, to genetic modification to alter traits, to cloning.

Although the public incorrectly believes these technologies are on the verge of success and widespread applicability, they will one day be upon us. These technologies offer the means of controlling the quality of our children without the need to terminate pregnancies, eliminating one of the more obvious moral issues surrounding prenatal disease or trait selection.

But the moral dimension of prenatal testing is not limited to the question of

termination. Deciding whether to eliminate or select characteristics in a future child, or even to discard an affected embryo, requires one to consider numerous moral issues such as parental obligations to the potential future child, existing children and others; parental interests in reproductive autonomy; the interests of the future child; the moral status of an embryo; society's interests in reducing disease; future generations' interests in the preservation of genetic diversity; and the impact on the disabled community. The less comfortable we are acknowledging the moral dimension of prenatal testing, the more difficult we will find ethical debates regarding future technologies. Given the routinization of prenatal testing today, society may well view such future parental choices as largely amoral and therefore unworthy of moral deliberation, especially once termination is removed from the equation.<sup>209</sup> As a result, people may be increasingly willing to engage in this kind of "quality control" without much moral deliberation.

The eugenics movement of the past, in which society tried to control reproduction, is unlikely to reappear today. However, a new, more subtle form of eugenics is currently emerging at the individual level. Terminating a pregnancy because of a negative trait can be a form of negative eugenics in that it eliminates undesirable traits.<sup>210</sup> Though autonomy, which supports the general right to terminate a pregnancy, also supports the right to make such choices, we should be willing to acknowledge the moral import of such choices.<sup>211</sup> In routinizing prenatal testing, we fail to do so and we create a kind of social pressure toward eugenic choices—selecting desirable traits in our children. As technology makes it possible to select or alter traits without pregnancy termination, such pressure will only increase. As Rothman states, "In gaining the choice to control the quality of our children, we may rapidly lose the choice not to control the quality, the choice of simply accepting them as they are."<sup>212</sup>

209 Reproductive cloning may be an exception; a majority of the public finds it distasteful and immoral. See Judy Holland, *Bush Finds Some Allies in War Against Cloning: He Calls on Trio of Injury Victims*, SAN ANTONIO EXPRESS-NEWS, Apr. 11, 2002, at 3A (noting that 77% of those surveyed are opposed to research on cloning humans).

210 See David E. W. Fenner, *Negative Eugenics and Ethical Decisions*, 17 J. MED. HUM. 17 (1996). Is terminating a pregnancy because the fetus has Down syndrome medical prevention or the elimination of undesirable traits? The line between preventing medical conditions and eliminating undesirable traits becomes particularly blurry when we describe conditions that are not life threatening, such as Down syndrome, dwarfism, deafness, etc. See *id.* at 19-20. Indeed, the distinction between the two is not static because social norms determine what we consider to be a disease.

211 Some of these choices may be morally sound depending on the moral justifications and context. Others may not be. As noted above, either way, they are moral decisions.

212 ROTHMAN, *supra* note 49, at 11.

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#### IV. CONCLUSION

I have argued that prenatal testing has become routinized as a result of the actions and attitudes of the legal community, the medical community and society at large. Trying to undo this routinization will be a difficult task given the complicated interplay of forces. The legal community's attempt to preserve choice through wrongful birth claims and mandating the offer of prenatal tests creates pressures to test. The movement of prenatal testing from genetics centers to obstetricians offices only furthers those pressures, especially when managed care and other financial factors create incentives for physicians to spend more time testing and less time talking. When all of these factors coexist with social norms that view prenatal testing as the responsible thing to do, it is no wonder that prenatal testing has become so

routine and that the option *not* to test has become more difficult.

We should be troubled by this trend because the decisions surrounding prenatal testing are difficult personal, moral decisions that require serious deliberation. Moreover, the information gleaned from such testing can be toxic to some, particularly if they are unaware of the full implications of prenatal testing and screening (the lack of treatment for most conditions, the potential stress associated with false positives, etc.) In order to make decisions consistent with their values and life plans, patients must be aware not only of the technical aspects of prenatal testing, but also the possible difficult choices they may have to make and the psychological ramifications of such choices. Patients must be encouraged to grapple with some very tough questions about their motivations for testing and the meaning of this information to them. Instead, however, the routinization of prenatal testing has impoverished informed consent and the decision making process.

With the recent completion of the mapping of the human genome, the range of possible prenatal tests will explode, further complicating the decisions patients face. The implications of prenatal testing will vary with the nature of the disease.<sup>213</sup> As choices expand and as technologies offer a broader range of reproductive options, we should hope that patients and society at large are equipped to consider the moral implications of these technologies. One way to move in that direction is to urge patients to begin these tough deliberations on a personal level as they consider whether prenatal testing for various conditions is appropriate for them. Let us resist the pressure to obtain knowledge for its own sake. Only the truly informed patient can decide whether knowledge from genetic testing will be empowering or toxic.

<sup>213</sup> For example, as tests become available for late-onset conditions, a new layer of complexity is added to the equation as patients grapple not only with issues of reproductive testing, but also with issues associated with late-onset testing.