

GENETIC TESTING AND ETHICAL ISSUES-A CRITICAL ANALYSIS**Koyel Roy**

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ABSTRACT:

Genetic testing is poised to play an increasing role in the practice of obstetrics and gynecology. To assure patients of the highest quality of care, physicians should become familiar with the currently available array of genetic tests and the tests' limitations. Clinicians should be able to identify patients within their practices who are candidates for genetic testing. Candidates will include patients who are pregnant or considering pregnancy and are at risk for giving birth to affected children as well as gynecology patients who, for example, may have or be predisposed to certain types of cancer. The purpose of this Committee Opinion is to review some of the ethical issues related to genetic testing and provide guidelines for the appropriate use of genetic tests by obstetrician–gynecologists. Expert consultation and referral are likely to be needed when obstetrician–gynecologists are confronted with these issues.

Keywords-Genetic, clinics, ethical issues, gynecologists, obstetrician, cancer, pregnancy, expert.

Although ethical questions related to genetic testing have been recognized for some time, they have gained a greater urgency because of the rapid advances in the field as a result of the success of the Human Genome Project. That project—a 13-year multibillion-dollar program—was initiated in 1990 to identify all the estimated 20,000–25,000 genes and to make them accessible for further study. The project harnessed America's scientists in a quest for rapid completion of a high-priority mission but left a series of ethical challenges in its wake. When developing the authorizing legislation for the federally funded Human Genome Project, Congress recognized that ethical conundrums would result from the project's technical successes and included the need for the development of federally funded programs to address ethical, legal, and social issues. Accordingly, the U.S. Department of Energy and the National Institutes of Health earmarked portions of their budgets to examine the ethical, legal, and social issues surrounding the availability of genetic information.

The purpose of this Committee Opinion is to review some of the ethical issues related to genetic testing and provide guidelines for the appropriate use of genetic tests by obstetrician–gynecologists. It is important to note at the outset, given the increasing complexity of this field and the quickness with which it advances, that expert consultation and referral are likely to be needed when obstetrician–gynecologists are confronted with many of the issues detailed in this Committee Opinion.

The pace at which new information about genetic diseases is being developed and disseminated is astounding. Thus, the ethical obligations of clinicians start with the need to maintain competence in the face of this evolving science. Clinicians should be able to identify patients within their practices who are candidates for genetic testing. Candidates will include patients who are pregnant or considering pregnancy and are at risk for giving birth to affected children as well as gynecology patients who, for example, may have or be predisposed to certain types of cancer.

If a patient is being evaluated because of a diagnosis of cancer in a biologic relative and is found to have genetic susceptibility to cancer, she should be offered counseling and follow-up, with referral as appropriate, to ensure delivery of care consistent with current standards. In fact, genetic screening for any clinical purpose should be tied to the availability of intervention, including prenatal diagnosis, counseling, reproductive decision making, lifestyle changes, and enhanced phenotypic screening.

One of the pillars of professionalism is social justice, which would oblige physicians to “promote justice in the health care system, including the fair distribution of health care resources”¹. In the context of genetic testing, justice would require clinicians to press for resources, independent of an individual’s ability to pay, when they encounter barriers to health care for their patients who require care as a consequence of genetic testing and diagnosis.

Obstetrician–gynecologists also are ideally positioned to educate women. When they, or experts in genetics to whom they refer, counsel on genetics, they should provide accurate information and, if needed, emotional support for patients burdened by the results or consequences of genetic diagnoses, be they related to preconception or prenatal care, cancer risks, or other implications for health. Finally, clinicians should familiarize their patients with steps that can be taken to mitigate health risks associated with their genetic circumstance (eg, having a colonoscopy if there is a predisposition to colon cancer)².

It recently has been suggested that each person’s entire genome may be available for use by physicians for diagnostic and therapeutic purposes in the not-too-distant future³. Although that might seem like a medical panacea, the potential risks associated with wide-scale genetic testing are substantial. Many incidental findings will come to light, and yet, although those tested may be tempted to believe otherwise, genetic findings do not equate directly with either disease or health: “one hundred percent

¹ Medical professionalism in the new millennium: a physician charter. ABIM Foundation; American Board of Internal Medicine; ACP-ASIM Foundation. American College of Physicians–American Society of Internal Medicine; European Federation of Internal Medicine. *Ann Intern Med* 2002;136:243–6.

² Burke W, Press N. Ethical obligations and counseling challenges in cancer genetics. *J Natl Compr Canc Netw* 2006; 4:185–91.

³ Wade N. The quest for the \$1,000 human genome. *New York Times*, July 18, 2006. Available at: <http://www.nytimes.com/2006/07/18/science/18dna.html>. retrieved January 9, 2024.

accurate identification of such incidental pathologies will lead to iatrogenic pathology... the belief that genetics completely determines phenotypic outcome must be informed by an understanding that most genetic measurements only shift the probability of an outcome, which often depends on other environmental triggers and chance”⁴.

Informed Consent

Genetic Exceptionalism

Before the appropriate process for obtaining consent for genetic tests is considered, it is necessary to confront the broader question of whether the consequences of the results of those tests are substantively different from the consequences of other “medical” tests, for which specific consent is not always obtained. Some ethicists argue against what has been called the “exceptionalism” of genetic tests⁵. They maintain that many medical tests have consequences for patients that are similar to those of genetic tests. For example, there can be discrimination by insurance companies against individuals either with a genetic disease or with a disease that is not linked to any particular gene. Results of nongenetic tests, as well as genetic tests, can divulge information about family members (eg, tests for sexually transmitted diseases). Additionally, both genetic and nongenetic tests can provide information about a person’s medical future. As such, some authors have concluded that many genetic test results “may cause stigmatization, family discord and psychological distress. Regardless of whether a test is genetic, when this combination of characteristics is present...testing should be performed with particular caution and the highest standards of informed consent and privacy protection should be applied”⁶.

However, others argue that genetics should be treated as a unique class and be subject to a more rigorous process for consent. They base their belief on several factors. Genes, they argue, do not merely inform patients and their health care providers about the diagnosis of an extant illness. They also foretell the possibility (or in some cases the certainty) of a future disease, thus allowing “perfectly healthy” individuals to be subject to discrimination based on a predisposing gene. The DNA sample—which can be viewed as “a coded probabilistic medical record”—“makes genetic privacy unique and differentiates it from the privacy of medical records”⁷. Some believe that this information is even more sensitive given the uncertainties attached to genetic results (ie, the reliability of tests, the penetrance of genes, and the unavailability of efficacious interventions to reduce the consequences of genetic diseases). Additionally, the consequence of being found to carry a particular gene has resonance not only for the individual who is tested but also for family members.

Patients should be informed that genetic testing could reveal that they have, are at risk for, or are a carrier of a specific disease. The results of testing might have important consequences or require

⁴ Kohane IS, Masys DR, Altman RB. The incidentalome: a threat to genomic medicine [published erratum appears in JAMA 2006;296:1466]. JAMA 2006;296:212–5.

⁵ Gostin LO, Hodge JG. Genetic privacy and the law: an end to genetics exceptionalism. Jurimetrics 1999;40:21–58.

⁶ Green MJ, Botkin JR. “Genetic exceptionalism” in medicine: clarifying the differences between genetic and nongenetic tests. Ann Intern Med 2003;138:571–5.

⁷ Roche PA, Annas GJ. DNA testing, banking, and genetic privacy. N Engl J Med 2006;355:545–6.

difficult choices regarding their current or future health, insurance coverage, career, marriage, or reproductive options.

Role of the Obstetrician–Gynecologist

In addition to needing to ensure proper consent, the obstetrician–gynecologist who orders genetic tests should be aware of when it is appropriate to test, which particular test to order, and “what information the test can provide, the limitations of the test, how to interpret positive and negative results in light of the patient’s medical or family history, and the medical management options available”⁸. The health care provider ordering tests has a responsibility to use and interpret those tests correctly or to refer to someone with relevant expertise. Because completing all these tasks is particularly difficult when direct-to-consumer marketing of genetic tests is used, that marketing approach has significant limitations⁹. These enterprises receive compensation only if an individual, after counseling, chooses to undergo a test, bringing the standard of neutral counseling into question and further rendering the use of a market-driven approach to testing ethically problematic¹⁰. In the end, the physician plays an important role in providing adequate, neutral counseling; ensuring informed consent; and providing follow-up for genetic tests. Neutral counseling also may be compromised through the use of patient educational materials or counselors that are provided by a company that might profit from a patient’s decision to undergo testing.

Particular caution should be exercised when obtaining consent for collecting genetic material that may be stored and, therefore, can have future clinical or research applications. The American College of Medical Genetics (ACMG) recommends that when samples are obtained for clinical tests, counseling should address the anticipated use of samples, including whether their use will be restricted for the purpose for which they were collected and if and when they will be destroyed¹¹. When samples will be used for research or the development of diagnostic tests, the ACMG recommends that consent should include a description of the work (eg, its purposes, limitations, possible outcomes, and methods for communicating and maintaining confidentiality of results). There should be a discussion with the research participant about whether she wishes to give permission to use her samples without identifiers for other types of research, and she should be informed of the institution’s policy regarding recontacting participants in the future. Current and future use of samples for research should follow state and federal regulations governing protection of human participants in research¹². Two authors recently suggested that the “best consumer advice, given current law, is that one should not send a DNA sample to anyone who does not guarantee to destroy it on completion of the specified test”. Others argue for the creation of a repository of samples donated by genetic altruists to be used for many different types of research.

⁸ Genetics and Public Policy Center. Genetic testing practice guidelines: translating genetic discoveries into clinical care. Washington, DC: GPPC; 2006. Available at:http://www.dnapolicy.org/images/issuebriefpdfs/Professional_Guidelines_Issue_Brief.pdf. Retrieved January 9, 2024

⁹ Direct-to-consumer genetic testing. ACOG Committee Opinion No. 409. American College of Obstetricians and Gynecologists. *Obstet Gynecol* 2008;111:1493–4.

¹⁰ Wolfberg AJ. Genes on the Web—direct-to-consumer marketing of genetic testing. *N Engl J Med* 2006;355:543–5.

¹¹ Statement on storage and use of genetic materials. ACMG statement. American College of Medical Genetics Storage of Genetics Materials Committee. *Am J Hum Genet* 1995;57: 1499–500.

¹² Protection of human subjects. 46 C.F.R. § 46 (2007).

Genetic Testing in Children and Adolescents

Testing of children presents unique issues in counseling and consent. Although it is most commonly pediatricians or geneticists who are called on to test children for genetic diseases, obstetricians may be asked to test already born children of parents who, through the process of prenatal testing, have been found to be carriers of genetic diseases. In such cases, the physician should balance the rights of the parents to have information that can optimize the ongoing health care of their children against the rights of the children to have their best interests protected. There will be circumstances in which it can be determined that a child is at risk for an untoward clinical event in the future, but there may be no information about interventions that have the potential to reduce the likelihood of that event or the magnitude of its effect. In that circumstance, the benefits of testing a child are not always clear (eg, BRCA testing in a young child).

The American Society of Human Genetics (ASHG) and ACMG together have suggested, “Counseling and communication with the child and family about genetic testing should include the following components: 1) assessment of the significance of the potential benefits and harms of the test, 2) determination of the decision-making capacity of the child, and 3) advocacy on behalf of the interests of the child”¹³. These societies highlighted additional points about benefits and burdens that should be included in counseling, some of which follow:

Timely medical benefit to the child should be the primary justification for genetic testing in children and adolescents. If the medical benefits are uncertain or will be deferred to a later time, this justification for testing is less compelling.

If the medical or psychosocial benefits of a genetic test will not accrue until adulthood, as in the case of carrier status or adult-onset diseases, genetic testing generally should be deferred. Further consultation with other genetic services providers, pediatricians, psychologists, and ethics committees may be appropriate to evaluate these conditions.

Testing should be discouraged when the health care provider determines that potential harms of genetic testing in children and adolescents outweigh the potential benefits. A health care provider has no obligation to provide a medical service for a child or adolescent that is not in the best interest of the child or adolescent.

The ASHG and ACMG concluded, “Providers who receive requests for genetic testing in children must weigh the interests of children and those of their parents and families. The provider and the family both should consider the medical, psychosocial, and reproductive issues that bear on providing the best care

¹³ Points to consider: ethical, legal, and psychosocial implications of genetic testing in children and adolescents. American Society of Human Genetics Board of Directors, American College of Medical Genetics Board of Directors. *Am J Hum Genet* 1995;57:1233–41.

for children”.

Physicians (obstetricians and pediatricians) also have a responsibility to provide information to patients regarding newborn screening. The primacy of the child’s welfare should animate these discussions as well. More detail about this issue can be found elsewhere¹⁴.

Prenatal Genetic Testing

Genetic testing of the fetus offers both opportunities and ethical challenges. Preconception and prenatal genetic screening and testing are recommended for a limited number of severe child-onset diseases because such screening and testing provides individuals with the chance to pursue assisted reproductive technology in order to avoid conception of an affected child, to consider termination of a pregnancy, or to prepare for the birth of a chronically ill child. With advancing genetic technology, however, physicians may increasingly face requests for testing of fetuses for less severe child-onset conditions, adult-onset conditions, or genetically linked traits.

Principles regarding testing of children provide some guidance for when prenatal testing might be appropriate but this decision is significantly complicated by the various purposes that prenatal testing can have: to detect a fetal condition for pregnancy termination, to allow patients to prepare for the birth and care of a potentially affected child, or, more rarely, to detect and treat a fetal condition in utero. Furthermore, many times, a woman’s intentions regarding pregnancy termination evolve as genetic information becomes available to her. Therefore, testing the fetus for adult-onset disorders with no known therapeutic or preventive treatment (save prevention by pregnancy termination) should raise caution in a way similar to the manner in which testing of children can. In pregnancies likely to be carried to term, consideration should be given to whether, as in the case of testing children, the decision to test should be reserved for the child to make upon reaching adulthood. However, consideration also should be given to personal preference, that is, the interests individuals may have in terminating a pregnancy that may result in a life (such as life that will be affected by Huntington chorea) that they feel morally obliged or prefer not to bring into the world. Because these often are wrenching decisions for parents, referral to parent support networks (eg, National Down Syndrome Society, if that is the diagnosis of concern), counselors, social workers, or clergy may provide additional information and support¹⁵.

Genetic Data and the Family

In a large number of instances, when patients receive the results of genetic tests, they are party to information that directly concerns their biologic relatives as well. This familial quality of genetic information raises ethical quandaries for physicians, particularly related to their duty of confidentiality. In these circumstances, some have posited an ethical tension between obligations the clinician has to

¹⁴ Newborn screening. ACOG Committee Opinion No. 393. American College of Obstetricians and Gynecologists. *Obstet Gynecol* 2007;110:1497–500.

¹⁵ Invasive prenatal testing for aneuploidy. ACOG Practice Bulletin No. 88. American College of Obstetricians and Gynecologists. *Obstet Gynecol* 2007;110:1459–67.

protect the confidentiality of the individual who has consented to a test on the one hand and a physician's duty to protect the health of a different individual on the other hand. For example, a woman who discovers that she is a carrier of an X-linked recessive disease during the workup of an affected son might choose not to tell her pregnant sister about her carrier status because she does not believe in abortion and fears that her sister might consider an abortion¹⁶. In another example, a woman identified as a carrier of a gene predisposing individuals to cancer might not wish to share the information with relatives, some of whom might even be patients of the same physician who tested her, because such sharing would disclose her own status as a carrier.

In both the previously cited cases, information obtained with the consent of one individual could assist in the management of another. However, medical ethics as reflected in American Medical Association (AMA) policies recognizes a physician's duty to safeguard patient confidences in such cases (with a few notable exceptions, often mandated by law—for example, communicable diseases and gunshot and knife wounds should be reported as required by applicable statutes or ordinances)¹⁷. How assiduously that confidentiality needs to be guarded is the subject of some debate. Some have argued that genetic information should be subject to stringent safeguards because, even though there may be uncertainty about the ultimate biologic consequence of a given gene, the social consequences (discrimination and stigmatization) can be substantial¹⁸. The AMA's Council on Ethical and Judicial Affairs has argued that physicians do indeed have an obligation to pay almost unlimited obeisance to a patient's confidentiality save only for "certain circumstances which are ethically and legally justified because of overriding social considerations"¹⁹.

Conversely, there are those who argue against the withholding of important information from potentially affected family members²⁰. Those who subscribe to this belief feel that when information applies to family as much as to the proband, an obligation arises that extends from the physician to those potentially affected family members but no further. This view is consistent with court rulings in three states, which have held that a physician owes a duty to the patient's potentially affected family members²¹ 22 23 24. Two of these rulings addressed the question of how physicians must fulfill this duty and reached different conclusions. In one case, the court held that the physician can discharge the duty by informing the patient of the risk and is not required to inform the patient's child²². In another case, the

¹⁶ Minkoff H, Ecker J. Genetic testing and breach of patient confidentiality: law, ethics, and pragmatics. *Am J Obstet Gynecol* 2008;198:498.e1–498.e4.

¹⁷ American Medical Association. Principles of medical ethics. In: Code of medical ethics of the American Medical Association: current opinions with annotations. 2006-2007 ed. Chicago (IL): AMA; 2006. p. xv–lvii.

¹⁸ Annas GJ, Glantz LH, Roche PA. Drafting the Genetic Privacy Act: science, policy, and practical considerations. *J Law Med Ethics* 1995;23:360–6.

¹⁹ American Medical Association. Confidentiality. In: Code of medical ethics of the American Medical Association: current opinions with annotations. 2006-2007 ed. Chicago (IL): AMA; 2006. p. 136–50.

²⁰ Wachbroit R. Rethinking medical confidentiality: the impact of genetics. *Suffolk Univ Law Rev* 1993;27:1391–410.

²¹ Offit K, Groeger E, Turner S, Wadsworth EA, Weiser MA. The "duty to warn" a patient's family members about hereditary disease risks. *JAMA* 2004;292:1469–73.

²² *Pate v. Threlkel*, 661 So.2d 278 (Fla. 1995).

court did not decide how the physician could satisfy the duty to warn, other than requiring that “reasonable steps be taken to assure that the information reaches those likely to be affected or is made available for their benefit”²³. As these alternate decisions illustrate, the legal limits of privacy are evolving, emphasizing the need for patient communication and case-by-case evaluation.

Recommendations of Other Organizations

Organizations that promulgate guidelines for genetic care and counseling also have proposed different approaches to the disclosure of genetic information. The ASHG tailors its recommendations to the magnitude and immediacy of risk faced by kindred²⁴, encouraging voluntary disclosure by the proband but also articulating circumstances in which the proband’s refusal to do so should not preclude disclosure by the health care provider. According to the ASHG, disclosure is acceptable if “the harm is likely to occur, and is serious, immediate and foreseeable.” It adds that the at-risk relative must be identifiable and that there must be some extant intervention that can have a salutary effect on the course of the genetic disease. In summary, “the harm from failing to disclose should outweigh the risk from disclosure.” Although this suggestion to disclose seems unequivocal, it also posits circumstances for its exercise that are highly unlikely at the current time (ie, very few genetic diagnoses pose an immediate risk, let alone ones that can be substantively modified with an intervention²⁵).

The President’s Commission for the Study of Ethical Problems in Medicine and Biomedical and Behavioral Research also suggested circumstances in which a health care provider should disclose information in the absence of the proband’s permission to do so²⁶. The commission indicated that disclosure is required when four conditions are present: 1) efforts to elicit voluntary disclosure by the proband have failed, 2) there is a high probability that harm will occur if disclosure is not made, and intervention can avert that harm, 3) the harm would be serious, and 4) efforts are made to limit disclosed information to genetic information needed for diagnosis and treatment. Although the commission did not cite a requirement for an immediate risk, the requirements for a high probability of harm and for the availability of an efficacious intervention make it likely that adherence to these guidelines rarely will result in cases in which a patient’s rights of confidentiality are overridden in order to inform relatives at risk.

Role of the Obstetrician–Gynecologist

The best way for the obstetrician–gynecologist to avoid the challenging choice between involuntary disclosure and being passive in the face of risks to kindred is to anticipate the issue and raise it at the first genetic counseling session. At that session, the patient needs to be educated about the implications

²³ Safer v. Estate of Pack, 291 N.J. Super. 619, 677 A.2d 1188 (1996).

²⁴ Molloy v. Meier, 679 N.W.2d 711, 718 (Minn. 2004).

²⁵ Professional disclosure of familial genetic information. ASHG statement. The American Society of Human Genetics Social Issues Subcommittee on Familial Disclosure. Am J Hum Genet 1998;62:474–83.

²⁶ President’s Commission for the Study of Ethical Problems in Medicine and Biomedical and Behavioral Research. Screening and counseling for genetic conditions: a report on the ethical, social, and legal implications of genetic screening, counseling, and education programs. Washington, DC: U.S. Government Printing Office; 1983.

of findings for relatives and why voluntary disclosure would in many circumstances be encouraged (as well as the possibility that relatives might prefer not to know the results). Some bioethicists have even suggested that these sessions should be used as an opportunity for clinicians to articulate the circumstances under which they would consider disclosure obligatory, thus allowing patients to seek care elsewhere if they found the conditions for testing unacceptable (Macklin has referred to this as the “genetic Miranda warning”) ²⁷. Similarly, even if the health care provider would not disclose without consent under any circumstance, the initial counseling session could allow the health care provider to refer the patient elsewhere if they find they have an irreconcilable difference or have an objection of conscience in expectations about disclosure. Physicians also should make themselves available to assist patients at the time of disclosure if that will help assuage their patients’ concerns.

A particularly thorny issue related to the ownership of genetic information might be results that bear on paternity. It is possible that prenatal assessments and family testing might reveal that the husband, partner, or other putative father is not the biologic father. In 1994, the Committee on Assessing Genetic Risks of the Institute of Medicine recommended that in such situations the health care provider should inform a woman but should not disclose this information to her partner²⁸. The Institute of Medicine’s reason for withholding such information was that “genetic testing should not be used in ways that disrupt families.” Another reason may be that the physician–patient relationship exists solely with the woman. Others have disagreed with the Institute of Medicine’s recommendation²⁹. In some cases, it is not merely a matter of acting to protect families. For example, suppose a child is born with a disease that is caused by an autosomal recessive gene, and the husband does not carry the deleterious gene because he is not actually the father. If the physician were to maintain the charade of paternity, then the counseling given to both parents (ie, there is one chance in four that each subsequent child will have the same disease) would be false and might lead the husband to argue against more children or for unnecessary amniocenteses in all future pregnancies, or inappropriately lead to concern for others in his family.

Other circumstances exist in which the interests of a pregnant woman and family members might diverge. For example, if the husband’s father has Huntington chorea (an autosomal dominant trait), the pregnant woman might wish to test the fetus for the gene. If the father did not want to know his own status, a conflict would arise, pitting her right to know about her fetus against his right not to know about himself. Another example of conflict would be if problems arose during diagnostic linkage studies for prenatal or preclinical diagnosis in a family and some family members did not want to participate in the testing (eg, testing for thalassemia). It might then be impossible to make a diagnosis in the index case. Both ethical and legal precedents, however, argue that individuals cannot be forced to have such testing.

²⁷ Macklin R. Privacy and control of genetic information. In: Annas GJ, Elias S, editors. *Gene mapping: using law and ethics as guides*. New York (NY): Oxford University Press; 1992. p. 157–72.

²⁸ Institute of Medicine (US). *Assessing genetic risks: implications for health and social policy*. Washington, DC: National Academy Press; 1994.

²⁹ Ross LF. Disclosing misattributed paternity. *Bioethics* 1996;10:114–30.

Genetic Data and Insurers and Employers

Concerns about access to health and life insurance in the face of the discovery of a deleterious or predisposing gene is one of the most nettlesome issues facing health care providers who wish to use genetic testing to improve the health of their patients. In some ways, the importance of this issue is more pronounced in the United States because of the manner in which health care coverage is obtained. In countries with universal health care, individuals with the diagnosis of a predisposing gene need not fear the loss of access to health insurance.

In recognition of concerns related to genetic testing, in 1995, the Equal Employment Opportunity Commission issued guidelines stating that individuals who thought they had been discriminated against by an employer because of predictive genetic testing had the right to sue that employer. Additionally, the Health Insurance Portability and Accountability Act (HIPAA), enacted in 1996, prevented insurance companies from denying health care based on predictive testing for individuals transferring from one plan to another³⁰. Physicians should advocate for patients' ability to obtain health or life insurance uncompromised by the results of any genetic tests they might undergo.

Although there is scant evidence of widespread genetic discrimination, there is clear evidence that fear of that discrimination can drive patients away from needed testing or from participation in research and also may influence physicians' uses of genetic tests. In commenting on insurance and discrimination and considering needed protections and legislation, ACMG makes the following points: legislation must not impede the ability of individuals to maximize use of genetic information in their health care and employment decision making, and it must not limit the access of health care providers to genetic information needed to ensure that the care provided is beneficial and specific to the needs of the individual. Furthermore, the privacy of genetic information must be adequately protected. Protection against unfair discrimination on the basis of genetic risk for disease is achieved only by strategies that restrict use of genetic information in enrollment and rate-setting. Protected genetic information must include information based on evaluation, testing, and family histories of individuals and their family members³¹. Finally, as discussed before, it must be recognized that the confidentiality of these data has become difficult to guarantee in this era of electronic medical records.

Genetics and Assisted Reproductive Technology

There are at least two issues that relate to the intersection of genetics and assisted reproductive technology (ART). In the first instance, there is the need to consider whether all individuals, regardless of genotype, should have access to ART using their own gametes. In the past, individuals who were infected with deleterious viruses that have the potential to be passed to their children (eg, human immunodeficiency virus) were denied access to ART, in part because, before the advent of a variety of interventions, as many as one in four of their offspring would acquire an ultimately fatal infection, a risk similar to that if both parents are carriers for a serious autosomal recessive disease. Others have

³⁰ Fulda KG, Lykens K. Ethical issues in predictive genetic testing: a public health perspective. *J Med Ethics* 2006;32:143–7.

³¹ Hudson KL. Prohibiting genetic discrimination. *N Engl J Med* 2007;356:2021–3.

argued, however, that “procreative liberty should enjoy presumptive primacy when conflicts about its exercise arise because . . . [it] is central to personal identity, to dignity and to the meaning of one’s life”³². Such principles would support allowing prospective parents to be arbiters of the level of risk to which a child could be exposed.

Second is the question of the extent to which preimplantation genetics should be used in pursuit of the “genetically ideal” child. The American College of Obstetricians and Gynecologists (ACOG) already opposes all forms of sex selection not related to the diagnosis of sex-linked genetic conditions³³. In the near future, other potentially controversial genetic manipulations may be available. Complex genetic systems such as cognition and aging soon may be determinable and may be constituents of potentially desirable characteristics, such as intelligence or longevity. They could, therefore, be used or misused as parameters for prenatal diagnosis³⁴. Some have argued for a permissive approach, allowing parents to choose from a menu of possible children the one with the chance for the “best life.” That approach would allow selection for both disease-related genes (eg, eliminating carriers of BRCA genes) and no disease genes “even if this maintains or increases social inequality”³⁶. One author has referred to this as “procreative beneficence,” defining it as couples selecting, from the possible children they could have, the child who is expected to have the best life, or at least as good a life as the others, based on the relevant, available information³⁵. Conversely, in the United Kingdom, strict limits are set on the use of prenatal genetic diagnosis, and clinics must apply for a license for every new disease they want to include in screening. However, even in that country, the list of allowable preimplantation genetic diagnosis tests has been expanded recently to include susceptibilities for certain cancers³⁶.

Parents’ requests to select a certain genetic trait may pose even greater challenges for reproductive endocrinologists and embryologists when parents’ choices seem to be antithetical to the best interests of the future child. For example, deaf parents may prefer to select for an embryo that will yield a child who will also be deaf. Couples who have short stature due to skeletal dysplasia might feel they would prefer to have a child of similar stature. The technical ability to provide these choices is not far from reality, but the ethical roadmap that will offer direction to physicians is not as clearly laid out.

Conclusions

Genetic testing is poised to play a greater and greater role in the practice of obstetrics and gynecology. To assure patients of the highest quality of care, physicians should be familiar with the currently available array of genetic tests, as well as with their limitations. They also should be aware of the

³² Watson MS, Greene CL. Points to consider in preventing unfair discrimination based on genetic disease risk: a position statement of the American College of Medical Genetics. *Genet Med* 2001;3:436–7.

³³ Robertson JA. *Children of choice: freedom and the new reproductive technology*. Princeton (NJ): Princeton University Press; 1994.

³⁴ Sex selection. ACOG Committee Opinion No. 360. American College of Obstetricians and Gynecologists. *Obstet Gynecol* 2007;109:475–8.

³⁵ Henn W. Consumerism in prenatal diagnosis: a challenge for ethical guidelines. *J Med Ethics* 2000;26:444–6.

³⁶ Savulescu J. Procreative beneficence: why we should select the best children. *Bioethics* 2001;15:413–26.

untoward consequences their patients might sustain because of a genetic diagnosis. The physician should work to minimize those consequences. Genetic information is unique in being shared by a family. Physicians should inform their patients of that fact and help them to prepare for dealing with their results, including considering disclosure to their biologic family. If the genetic information could potentially benefit family members (eg, allow them to improve their own prognosis), physicians should guide their patients toward voluntary disclosure while assiduously guarding their right to confidentiality.

Recommendations

The ACOG Committees on Ethics and Genetics recommend the following guidelines:

Clinicians should be able to identify patients within their practices who are candidates for genetic testing and should maintain competence in the face of increasing genetic knowledge.

Obstetrician–gynecologists should recognize that geneticists and genetic counselors are an important part of the health care team and should consult with them and refer as needed.

Discussions with patients about the importance of genetic information for their kindred, as well as a recommendation that information be shared with potentially affected family members as appropriate, should be a standard part of genetic counseling.

Obstetrician–gynecologists should be aware that genetic information has the potential to lead to discrimination in the workplace and to affect an individual's insurability adversely. In addition to including this information in counseling materials, physicians should recognize that their obligation to professionalism includes a mandate to prevent discrimination. Steps that physicians can take to fulfill this obligation could include, among others, advocacy for legislation to ban genetic discrimination.

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