Testing Children for Genetic Predispositions: Is it in Their Best Interest?

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Researchers summoned a Baltimore County woman to an office at the Johns Hopkins School of Public Health last spring to tell her the bad news. They had found a genetic threat lurking in her 7-year-old son's DNA—a mutant gene that almost always triggers a rare form of colon cancer. It was the same illness that led surgeons to remove her colon in 1979. While the boy, Michael, now 8, is still perfectly healthy, without surgery he is almost certain to develop cancer by age 40.

This genetic fortune-telling was no parlor trick. It was the product of astonishing advances in recent decades in understanding how genes build and regulate our bodies. And as scientists pinpoint new genes and learn to forecast the onset of more inherited disorders, millions of people are likely to demand their medical prognosis.[1]

Testing healthy newborns and children for genetic DNA abnormalities that will not manifest disease symptoms for many years, if ever, is now rarely done. However, the entry of such tests into the marketplace is raising the specter of their widespread use. Tests that predict the likelihood of cancer are attracting the greatest attention, and they may be the first such tests to be administered on a large-scale basis.[2] Currently, tests are available for predisposition to breast cancer, colon cancer, melanoma, and thyroid cancer.[3]

Marketing these tests to the general population is highly controversial, with proponents arguing that people have a right to know if they or their children are at increased risk and that it would be unethical to deny them that information. Much of the advocacy for widespread use of the tests comes from the biotechnology companies offering them.[4] For example, a recent news article about marketing these genetic tests described the question of testing children as "delicate." Yet it also reported, in a letter to dermatologists on testing for melanoma predisposition, that the manufacturer of the tests believed "[e]arly screening with this easy and painless test is particularly useful when testing children."[5]

Some advocates of testing children go so far as to state that a geneticist has a medical and legal duty to advise parents about presymptomatic testing procedures for some (even late-onset) diseases and either to administer the procedure or to refer the child to a colleague for administration (presuming the child meets certain pre-administration criteria).[6] Others describe the effort to keep the tests from patients as medical paternalism.[7] Opponents have characterized the initiative to market the genetic tests as "alarming," arguing that this area of genetic testing is still in the research phase and that the tests should not be marketed now.[8] Others argue that, due to the uncertain psychological consequences for children of predictive testing, such testing should not generally be done at this time or should be restricted.[9]

We support those who express caution and urge restraint[10] in conducting predictive genetic tests on children, and we suggest policy recommendations to safeguard children's interests from possible negative effects of such testing. Many of our suggestions are consistent with the recently published joint statement of the American Society of Human Genetics (ASHG) and the American College of Medical Genetics (ACMG) on genetic testing of children and adolescents.[11]

We specifically address: (1) the role of physicians in

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informing parents of healthy children about the availability of predictive genetic tests; (2) the role of physicians and parents in providing such tests to children; and (3) the role of physicians, parents, and other public and private entities, such as registries, in following up on tested children when new developments would benefit the child.

Throughout, we use the term *genetic disease* to refer to that rare group of disorders in which an abnormality (or abnormalities) in the genetic code (DNA) of an individual is associated with a near certainty of developing disease. In contrast, we use *genetic predisposition* to refer to an abnormality in the genetic code of an individual that results in an increased risk of developing a disease.

Current testing of newborns and children

Little genetic testing of children is currently performed, other than newborn screening for a small number of treatable genetic diseases that are expected to cause symptoms if not treated during infancy. When testing is performed on older children, it is generally limited to those few individuals who either are suspected of having a genetic disease or are in families at high risk for having a genetic disease. Several attempts have been made to categorize reasons to do genetic testing on infants and children.[12] Using the work of Wertz et al. and Fost as a starting point, we employ a comprehensive list of seven categories: (1) testing of immediate benefit to the infant or minor, including newborn screening, disease testing for a symptomatic condition in the child, or presymptomatic testing for which treatment during childhood is available and beneficial; (2) reproductive-associated testing and counseling for older adolescents that is mainly genetic carrier rather than genetic disease testing; (3) testing for the benefit of other family members' reproductive decision making, where it may be necessary to test both affected and unaffected family members to understand the inheritance of a genetic disease; (4) research-related testing that is generally conducted under informed consent protocols approved by institutional review boards; (5) testing by insurance companies for the purpose of excluding individuals from coverage; (6) presymptomatic testing to predict a child's future risk of developing a genetic disease or of having a genetic predisposition for which no current treatment or effective prevention exists; and (7) testing for carrier status at an age when the child cannot procreate.

Predictive testing

Our primary focus is category (6)—testing for presymptomatic genetic diseases or predispositions. While a relatively recent survey indicates that many British geneticists and pediatricians would presymptomatically test children for a genetic condition at the request of the parents and with little immediate benefit to the child,[13] many ethicists and professional genetics societies agree that testing children for genetic diseases, predispositions, or carrier status is only appropriate when a clear and timely benefit to the minor exists.[14] And the debate, in large part, has focused on determining what constitutes a clear and timely benefit and who would benefit from the information. While many professionals argue that there is insufficient benefit to the child to warrant widespread screening or even high-risk family testing for most genetic diseases during childhood,[15] some argue that the information may be beneficial to the child's family. Whether this benefit would outweigh the negative aspects of being identified as having a disease, such as being treated differently by one's parents (no college fund or other long-term plans), has significant ramifications for the appropriateness of predictive testing.

Controversies over predictive testing have arisen both in the context of screening newborns and testing infants and older children. As basic principles for newborn screening, the Institute of Medicine recommends that such screening take place only when "(1) there is a clear indication of benefit to the newborn, (2) a system is in place to confirm the diagnosis, and (3) treatment and follow-up are available for affected infants."[16] Though these guidelines are relatively straightforward for some diseases such as phenylketonuria (PKU), the difficulty in defining a benefit to the newborn is illustrated by experimental screening programs for alpha$_1$-antitrypsin deficiency, cystic fibrosis (CF), and Duchenne's muscular dystrophy.

Alpha$_1$-antitrypsin deficiency is a genetic enzyme deficiency, common in individuals of Scandinavian ancestry, that results in a high risk of developing adult-onset emphysema.[17] The deficiency results in early-onset emphysema in 80 percent of individuals with a severe enzyme deficiency. For individuals with the predisposition, emphysema occurs earlier in those who smoke or who are exposed to occupational or residential environments with high levels of particulates. In 1972, Sweden initiated a nationwide experimental newborn screening program for this condition. As part of the program, families were (1) told whether their child had the genetic deficiency, (2) counseled to protect their child from smoking or environments with high levels of particulates, and (3) followed to determine the psychological impact of the information. The government assumed that the benefits of the screening program would outweigh the negative implications of identifying a child as having a genetic disease. Surprisingly, the follow-up studies found that more than half of the families suffered severe negative psychological consequences, and that some negative effects were still present on follow-up at five to seven years.[18] As a result of early feedback on these negative effects, the Swedish government discontinued the program in 1974.[19]

Screening newborns for CF has also created contro-

versy. CF is a treatable but incurable autosomal recessive genetic disease that results in thick secretions in the lungs and pancreas and that leads to chronic pulmonary and digestive disease. Children often manifest symptoms within the first few years of life. Treatment has extended life expectancy to between twenty and thirty years of age, and has improved the quality of life for individuals affected by the disease. Various newborn CF screening tests have been available since 1968.[20] Early on, those offering the test assumed that identifying those who will ultimately develop the disease might result in better treatment outcomes for the patient and planning benefits for the family. Currently, however, only Colorado runs a routine CF screening program. Wisconsin established an experimental newborn screening program in 1985. The first five years of the Wisconsin program provided no evidence that presymptomatic detection had any clinical benefit, and a number of psychological and ethical issues were encountered. Researchers concluded it was premature to offer population-wide screening until the benefits and risks had been clearly determined.[21] Many people think (even in cases where there is a familial risk for the disease) that early detection has no value and may, in fact, cause the family significant psychological stress prior to the time when the individual might become symptomatic.[22]

Finally, screening for Duchenne's muscular dystrophy has provoked some controversy. Duchenne's causes an incurable and untreatable muscle-wasting that does not begin until at least two to four years of age and leads to death in the mid-teens. The primary justification for presymptomatic detection of it is to provide the family with reproductive options for future children. Screening of newborns for Duchenne's by measurement of blood spot creatine kinase activity has been possible since the mid-1970s.[23] While a voluntary screening program has been offered in Wales since 1990,[24] no routine newborn population screening for this condition is performed in the United States. This is largely because of the perception that the psychological burden of learning that your apparently healthy child will some day develop a lethal incurable genetic disease outweighs possible future family planning benefits.

These newborn screening controversies illustrate the difficulties in determining what constitutes a clear and timely benefit, in particular the difficulties of weighing psychological benefits and burdens.

Psychological impact

Concerns about testing children solely for predictive purposes have focused largely on the potential psychological implications to the child, especially in cases of predisposition to an incurable disease.[25] Wertz et al. have summarized some of the psychological and emotional consequences of such testing. They argue that in requesting testing, parents typically think only of the benefits of a negative test result and not of the potentially damaging effects of a positive result:

> "Planning for the future," perhaps the most frequently given reason for testing, may become "restricting the future" (and also the present) by shifting family resources away from a child with a positive diagnosis.... In families with a chronically ill child, there is less socialization to future roles for all the children, including those who are "healthy." Parents are less likely to say "When you grow up..." or "When you have children of your own..." to any of their children, because they cannot say these words to the ill child.... "Alleviation of anxiety," another reason commonly given by parents for predictive genetic testing, does not necessarily benefit the children. A positive diagnosis may create serious risks of stigmatization, loss of self-esteem, and discrimination [by] family or by institutional third parties such as employers or insurers. Testing may disrupt parent-child or sibling-sibling bonds, may lead to scapegoating a child with a positive result or to continued anxiety over a child despite a negative result....[26]

Balancing possible psychological harms and benefits makes the determination of what constitutes a clear and timely benefit particularly evasive. This difficulty is illustrated by testing for a very rare genetic familial cancer known as the Li-Fraumeni syndrome. In 1991, the National Cancer Institute and the National Center for Human Genome Research held two workshops to consider predictive testing for Li-Fraumeni syndrome families. Participants, who included experts in clinical medicine, laboratory science, epidemiology and biostatistics, medical ethics, law, psychology, and cancer control, made the following recommendation:

> Cancers occur with high frequency among children in Li-Fraumeni families, and testing these children (rather than delaying it until young adulthood) is recommended, with the goal of reducing cancer morbidity and mortality. As children mature, it is appropriate to consider their assent or dissent to testing as well as their parents' permission. Parents and investigators should develop a plan on the timing and person(s) to convey test results to children.[27]

The recommendation is apparently based on the unsupported optimism that knowledge of disease status will lead to a benefit to the child from increased cancer surveillance and early disease treatment. Unfortunately, the majority of cancers seen in Li-Fraumeni syndrome are incurable, and no evidence indicates that knowing a child carries the mu-

tant gene is of any benefit. This is reflected in Fost's more recent discussion of the syndrome:

> There is less information [as compared to, for instance, CF] on the benefit of identifying children with Li-Fraumeni syndrome, a genetic predisposition to cancer that exposes affected individuals to a 30 percent risk of cancer by age 30 years.... Although it is plausible ... that early screening, detection, and treatment of cancers in such individuals will result in better outcomes, that has not been shown. These theoretical benefits must be weighed against the risk of the possibly incapacitating psychological trauma associated with growing up under a sword of Damocles.[28]

Identifying a child with a genetic predisposition may lead to the "vulnerable child syndrome" in which parents become overprotective and unnecessarily restrict a child's activities.[29] Also, those who test negative have been shown to experience "survivor guilt."[30] Given these concerns, the International Huntington's Disease Association and the World Federation of Neurology have issued policy statements recommending that minors not be tested for Huntington's disease,[31] and the National Kidney Foundation has recommended that minors not be tested for the gene for adult polycystic kidney disease except in specific circumstances where preventive measures are applicable for stroke.[32]

Also, in their joint statement, ASHG and ACMG recommended that "[t]imely medical benefit to the child should be the primary justification for genetic testing in children and adolescents," and that if the medical benefits "are uncertain" or will not accrue until a later time, genetic testing should generally be deferred.[33]

Balancing burdens and benefits

As additional genetic tests become available, the controversy over what constitutes a clear and timely benefit will continue. Those diseases that threaten immediate harm to the patient unless the patient is treated and a treatment exists, present the easy cases. The more difficult cases involve conditions where there is incomplete penetrance, no treatment, but some preventive intervention that *may* reduce the likelihood that the patient will develop the condition [34] Such preventive measures may include a restricted diet, avoiding exposure to certain environmental aggravators (such as smoke or sunlight), preventive drug therapy (such as tamoxifen for breast cancer),[35] or even preventive surgery (such as a mastectomy when breast cancer is likely[36] or a colostomy when colon cancer is predicted). These cases are problematic in large part because of the uncertainty associated with manifesting the disease[37] and the uncertainty of the effectiveness of the intervention. Regarding the latter issue, the National Advisory Council for Human Genome Research recommends that presymptomatically testing either high-risk families or the general population for predisposition to specific cancers *not* be conducted until a number of questions have been addressed. Among them is the effectiveness of current interventions to prevent cancer morbidity and mortality in high-risk families and in the general population.[38]

The decision by a physician to offer or perform a presymptomatic genetic test and by a parent to have their child tested will require weighing the possible benefits of the test with its impact on the child's quality of life. While a child's parents are generally in the best position to make this decision, we are concerned about situations where no preventive interventions or treatments are available and parents still want the test performed.

The psychological and emotional value of the test information is likely to be different for parents than for their child. The difference, we argue, requires greater input from the medical profession in setting guidelines on when it is appropriate to inform families about predictive genetic tests, when it might be appropriate to perform them, and what to tell parents in obtaining their informed consent for testing their child.

Should physicians disclose the availability of presymptomatic genetic tests for children?

The controversy surrounding presymptomatically testing children for genetic disease may soon cause anxiety for some physicians as to whether they must or should disclose the availability of genetic tests to parents of healthy children. Physicians may be concerned about potential liability for failure to inform parents about such tests. Some have contributed to this concern by arguing that physicians may have a legal duty to disclose the availability of these tests.[39] They rely erroneously on case law on prenatal testing and wrongful birth. In these cases, parents have successfully claimed that had they known about the test, they would have consented to it; and, if it had indicated that their fetus had a serious genetic condition, they would have terminated the pregnancy. Instead, the physician's failure to inform them of the test resulted in their having a child with a severe genetic abnormality that could have been detected.[40]

The problem with this analogy is that, in the prenatal context, parents could use the information to make a decision to terminate the pregnancy. In the context of testing children for a genetic predisposition for which the parent can do nothing to alter the likely manifestation of the disease, the physician would be not be legally liable. Liability would only attach when a beneficial intervention exists and failure to test or to test in a timely manner would result in harm to the child.

Thus, for a genetic disease for which we have no effective preventive intervention or treatment, a physician would have no legal duty and should not fear liability for failure to inform parents of a genetic test. We argue that this is the case both when there is no family history or probable cause for believing the child has a genetic predisposition and when there is a family history of the disease. While courts have made a distinction between informing parents about a test for a genetic disease when there is and is not a family history of the disease,[41] these cases are based on the prenatal testing paradigm, in which parents have the option of terminating the pregnancy.

We argue further that, as a policy matter, a physician should not be obligated to disclose the availability of the tests under these circumstances. Where no family history of a genetic condition exists, requiring disclosure of all available tests would take considerable time on the part of a physician or other health professional for no likely benefit to the child or parents. Disclosure in this circumstance arguably wastes resources, and it has the potential for psychological harm. To discourage testing under these circumstances, we recommend that appropriate professional societies, such as the American Academy of Pediatrics (AAP), ASHG, and ACMG, issue a strong statement in support of *nondisclosure* where no available preventive intervention or treatment would benefit the child. Where there is a family history of a specific disease, the National Institutes of Health (NIH) should establish consensus panels to develop guidelines as to when disclosure of a predictive test is appropriate; and the relevant professional societies should adopt them. Such guidelines might include the appropriate age for testing as well as the age and circumstances when a preventive intervention, such as colonoscopy for colon cancer, is or is not appropriate.

Should physicians perform predictive genetic testing on children?

With the increased availability of tests for genetic predispositions, physicians will undoubtedly encounter parents who have read about the tests and request one or more of them for their child. Do physicians have a legal duty to perform such tests? More importantly, *should* physicians perform such tests?

As to the first question, physicians are under no legal obligation to provide the test. They are free not to provide a treatment or diagnostic test to a patient under most circumstances. In some cases, if the treatment or diagnostic test is considered part of standard medical care, a physician would need to inform a patient of it but would not be required to provide it. In some jurisdictions, under certain circumstances, they may have a legal obligation to refer the patient to another provider who would provide the test or treatment.[42] In a recent article, Clayton ably dispels the myth that parents have a constitutional right to demand medical treatment or testing for their child, as well as the belief held by some physicians that they will be liable under tort law for failure to provide the tests.[43] Her persuasive analysis should provide comfort to physicians who refuse to test children for genetic predispositions.

As to whether a physician *should* provide such tests for genetic dispositions, we argue generally that such tests should not be provided but that a distinction may be made between cases in which there is and is not a family history of a genetic disease. If there is no family history or other risk factors, to test a healthy child for predisposition to genetic diseases is simply a fishing expedition with no foundation. If there is a family history, we concur with the ASHG–ACMG position that the decision to test should be made by a physician in discussion with the child and the child's parents.

In very young children, testing should be delayed in most cases until the child can understand the implications of the test. For example, where a mother and her three-year-old daughter visit a pediatrician for the first time and a medical history of the child reveals that the mother's mother and sister both had breast cancer, the physician should inform the mother about the availability of the test for herself, and might suggest that, if interested in it, she talk to her internist who can refer her to a geneticist. With respect to her three year old, the physician should simply state that, at some time in the future (when the child is sufficiently mature to understand the information), the child's mother might want to talk to her daughter about the family history and the test and to let the daughter decide if she would like to have the test done and, if so, when.

In some cases, however, parents may persistently demand a test for their child. Some have argued that to deny the parents the test is medical paternalism and flies in the face of our general deference to parents regarding medical decision making for their children.[44] The law clearly gives parents this authority and assumes that parents will act in their child's best interests when making such decisions. Very seldom, in fact, are parents denied the right to make medical decisions for their children, and, when denied, the cases usually involve questions of parental abuse or neglect. But cases deferring to parental decision making are not analogous to the case of genetic testing. Virtually all cases of deference to parental decision making involve circumstances in which a medical professional is recommending a course of treatment for a child, for example, surgery or chemotherapy, and the parents refuse it. Although parental decision making can be taken away when failure to provide the treatment would threaten the child's life, in virtually all other cases the parents have the right to decide not to consent to a proposed treatment.

This stands in stark contrast to cases in which the parent wants a treatment or procedure for the child that is not recommended by the physician. An example might be a

common parental request to have the child's blood type determined. Pediatricians generally will not draw a child's blood simply to satisfy the parents' curiosity—he/she must have a medical reason to perform the test. Genetic tests may be somewhat more complex, and physicians may need guidance in determining where or under what circumstances a test should or might be provided. As with disclosure where there is a family history of a genetic disease, we encourage NIH to continue to bring together consensus panels for specific diseases and to develop guidelines as to when it is appropriate to perform a genetic test on a child. Such guidelines have been largely successful in limiting the performance of amniocentesis on pregnant women and in restricting the testing of testing individuals for CF carrier status. We recommend that these guidelines not be rigid, however, so that physicians have some latitude in deciding whether testing is warranted in a particular case. This flexibility should also allow a physician to converse with a child's parents or a child (if sufficiently mature) regarding the desire for the testing.

Although parents generally know their child best and care most about the child's welfare, we believe that physicians and health care providers have an obligation to provide them with sufficient information to make a true informed decision about the benefits and risks associated with testing for a genetic predisposition. If parents, despite a statement from their child's physician that the physician does not generally perform predictive genetic testing, want the test performed, we recommend that the physician refer them to an appropriately trained genetic counselor or another physician who can objectively explain the risks and benefits of such testing. If they still desire the testing, the health care provider must obtain their informed consent.

Informed consent for genetic testing: current law and practice

Informed consent requires a physician to obtain consent from a patient to perform a specific procedure or to take a certain course of action. When children are involved, consent is usually required from the child's parents. In general, the law provides that a physician disclose to a patient the nature of the proposed treatment or intervention, the risks and benefits associated with it, as well as the alternatives and their associated risks and benefits.[45] All material or significant risks must be disclosed. This requirement has generated questions, however, as to who determines what is material or significant. In some jurisdictions, the determination is made by the "reasonable physician"; in others, by the "reasonable patient."

A slight majority of states have adopted a physician-centered approach wherein the physician discloses risks that are customarily disclosed by physicians or what a reasonable physician would disclose under the same or similar circumstances.[46] Criticism of this "professional" approach has focused on the argument "that the standard of disclosure exercised by the medical community bears no inherent relationship to the amount of knowledge any particular patient might require to make an informed choice, and that the use of the professional standard bypasses an investigation into the actual importance the undisclosed information might have for the patient."[47] Defendants of this approach argue that "to adopt a standard based on the patient's need for information would result in requiring doctors to go over with every patient every possible aspect of any proposed treatment."[48]

The applicable standard will have significant implications for disclosure by physicians about the risks of genetic tests. Although the physical risks are generally minimal (a needlestick for obtaining a blood sample), social, emotional, psychological, and economic consequences may arise from a positive test result, such as loss of health insurance, social stigmatization, and job loss. Research protocols for testing for some genetic predispositions currently include disclosure of such risks. For example, at one research center where families at high risk for breast cancer are being tested, risks discussed include "jeopardizing insurance coverage, particularly for women who may seek costly preventive surgical options based on the information, the psychological impact of obtaining carrier status information, and the possible impact on family dynamics."[49]

These types of economic and psychological risks have been raised in the context of HIV testing. Many have argued, for instance, that informed consent for an HIV test should include the possible social consequences of a positive test—discrimination in housing, schooling, employment, and insurance. In some states, such as Maryland, prescribed consent forms for HIV testing, prepared by the state health department, include these risk factors.[50]

Others have argued that the potential psychological and economic impact and the potential stigmatization associated with a positive result from certain genetic tests are similar to those that result from a positive HIV test, and that physicians should be required to inform those to be tested of these risks.[51] While these risks may be perceived as material by a patient or his/her parents, they are less likely to be deemed relevant under a physician-centered approach to informed consent. A physician's disregard for the psychological impact of a test for a genetic predisposition is illustrated by the statement of a cancer researcher who favors marketing genetic tests for determining cancer predisposition. When interviewed by a *New York Times* reporter about concerns related to such testing, the researcher stated that "[i]nformation is neutral. We always have this question: Is it good or bad? We don't know until we use it. This is diagnostics, not a drug. The medical risks are zero. We're really talking about psychological issues."[52] Because of this attitude on the part of some physi-

cians, in the case of genetic testing of children, we argue that a patient-centered approach to informed consent is more appropriate.

Difficult issues

The child's role in obtaining informed consent

Testing children for genetic predispositions raises new issues in terms of informed consent and counseling, including the role of the child in the decision. As regards children's participation in decisions to be tested, Wertz et al. argue generally that genetic testing that confers no benefits on the child should be deferred until adulthood. The statement by ASHG and ACMG concurs with this position. However, both authorities concede that presymptomatic testing may benefit some minors by enabling them to make plans for the future. Under these circumstances, Wertz et al. argue that the minor should actually initiate the request for testing and that "[t]horough counseling of both the minor and family (including siblings) should precede testing, to assess the inner strength of all concerned."[53] They further argue that no ethical justification ordinarily exists for testing before the age of eleven or twelve years, absent proven medical benefits.[54]

Legally, the role of the child in the decision to test will clearly be at issue for adolescents. Currently, genetic testing of this age group is only rarely performed—usually only in a reproductive context—but it may be more common as genetic predisposition testing becomes available. Historically, consent for medical treatment of a minor has been obtained from the minor's parents or legal guardian. More recently, state statutes have created exceptions through which adolescents may consent to medical treatment on their own if they are considered "emancipated minors" or "mature minors." The definition of *emancipated* varies by state, but generally includes minors who are married, who have children, or who are financially independent from their parents and living away from home.[55] To qualify as mature, the minor must be competent to assess the risks and benefits of a medical procedure and to make a reasoned decision based on that assessment.[56] It is the physician who must assess the maturity of the minor, often a difficult call.[57] Moreover, statutes in some states allow minors to consent to certain types of medical procedures even if they are not emancipated or mature. These include treatment of venereal disease, care related to pregnancy, treatment for drug or alcohol abuse, mental health treatment, and contraception.

These statutes address situations where adolescents want tests or procedures that their parents do not want them to have.[58] Although this is unlikely in the context of genetic testing, it could occur where an adolescent at risk for Huntington's disease is contemplating marriage and wants to be tested for the gene. His parents may not want him to be tested because the test results may tell the parents about their own likelihood of developing the disease. It is possible that a genetic test for a minor would fall under some state statutes authorizing minor consent to reproductive counseling.[59] This would obviate the need for parental consent in such circumstances.

While some adolescents may request testing, a much more likely scenario is an adolescent's refusal to consent to a test that his parents wish him to get. Most of the literature in this area, informed by the research setting, is consistent in its call for the assent of the minor.

In the context of testing adolescents, a question that may arise is the physician's obligation to inform the adolescent of the test result. In testing for the breast cancer gene, researchers at at least one research center made a decision not to reveal test results to individuals younger than eighteen years. This decision was based on the following factors:

> (1) there are no known medical benefits to a minor of knowing that he or she is a gene carrier (we are unaware of any breast cancer cases occurring at this age or younger that can be attributed to the gene under study); (2) it is difficult to determine the emotional maturity required to receive results; and (3) there are no nationally established guidelines for screening children for late-onset disorders.[60]

While several courts have held that physicians have a duty to inform patients about medical test results even if those results would upset the patient,[61] disclosure to a minor is likely to be more complex. In the nonresearch context, physicians who test minors for genetic conditions at the request of the minors' parents will need to make an independent assessment of whether or when the minor is mature enough to understand the implications of the test results. If the physician finds the minor is sufficiently mature to understand the implications, he would arguably have a duty to inform the minor of the findings, absent an explicit statement by the minor that she did not want to know them.

Uncertainty and limited treatment options

Other difficult issues that may arise in informed consent for genetic testing of children include: (1) the value placed on uncertainty; (2) the fact that tests may not always be informative; (3) the probabilistic nature of test results; and (4) the fact that limited prevention or treatment options may exist.

While a number of writers have discussed the virtues of certainty in knowing one's predisposition, the benefits of uncertainty to all groups, but most especially to children, have not been established. In their recent statement,

ASHG and ACMG argue that a significant psychological benefit of genetic testing is the resolution of uncertainty. The statement cites a study, by Wiggins et al.[62] of adults *who decided to be tested for Huntington's disease*, suggesting that uncertainty may be even more stressful than knowing one will ultimately get the disease. While this may be true for some individuals, their conclusion about the benefits of uncertainty must be limited to those who *desire* testing. Most adults at risk for Huntington's disease, however, have chosen *not* to be tested. Studies have found that while many (two-thirds) of those at risk for Huntington's disease indicated prior to the availability of a genetic test that they would want to be tested, only a small percentage of those at risk have actually taken the test.[63] A recent study by Fanos and Johnson,[64] on testing for CF carrier status of siblings of individuals with CF, also found that many at risk prefer not to know whether they are carriers. The authors concluded that remaining unaware of carrier status may serve significant psychological functions for individuals at risk. Thus, it seems premature to conclude that individuals, particularly children, will benefit from knowing whether they are at risk of developing a serious genetic condition.

More research is needed on how this information will affect children and whether certain types of children, that is, children with specific personality traits or from certain types of families, would benefit from or be harmed by it. Parents making a determination to test a child for a genetic predisposition may also be overvaluing the benefit of that information. The information, for example, will not provide parents with the certainty that their child will get the disease—the child will simply have an increased risk. Nor will it tell them that their child will not get the disease. Individuals without a genetic predisposition to a disease, for example, breast cancer, may still develop it.

With respect to breast cancer, however, Lerman and Croyle argue that "[t]he incomplete penetrance of the major breast cancer–related genes affords a major opportunity—the potential for prevention or early detection in as yet unaffected gene carriers."[65] On the other hand, incomplete penetrance means that some who carry the gene (BCRA1) will not manifest the disease and thus may undertake unnecessary preventive measures, such as mastectomies. This possibility illustrates why it is important that, when obtaining informed consent, an effort must be made to explain to parents that a test result is not conclusive in terms of a child's getting the disease—it simply indicates that the child's probability of getting the disease is greater than the norm.

Communicating test results and counseling about future preventive options

As indicated, the best a genetic test can offer is an estimate of the risk an individual faces. Conveying the meaning of risk is problematic.[66] No satisfactory guidelines have been established for communicating information about genetic diseases for which there is incomplete penetrance. In the research setting, counselors are experimenting with different protocols. In the center described above, where high-risk family members were tested for BRCA1, each patient was seen by a team consisting of at least three members of one of the following disciplines: genetics, oncology, genetic counseling, or oncology nursing. The team gave each patient information about "methods of linkage analysis, the individual's test results, the implications for future risks of developing breast and/or ovarian cancer, medical intervention and screening options, and psychosocial counseling needs."[67] Furthermore, information and individual counseling services were offered to each family member to help ensure confidentiality for those who sought it. Women who had the gene were informed of their lifetime risk of developing breast cancer (85 percent) and of an increased risk of developing ovarian cancer (25 to 85 percent). Counseling for male carriers focused on the 50 percent risk of having passed a BRCA1 mutation on to each of their children.[68]

For women identified as BRCA1 carriers who had not yet developed breast cancer, "breast self-examination, annual examinations by a specialist, and mammography were discussed. These women were informed that these procedures were designed for early detection of breast cancer, not for prevention."[69] In addition, prophylactic options including bilateral mastectomy and oophorectomy were discussed, along with the risks associated with these options.[70]

The matter of counseling about preventive options can be a sensitive one for genetic counselors, especially when the prevention itself has risks and significant physical and psychological burdens, for example, mastectomy, and the condition for which the preventive measure is proposed is not a certainty.[71] This issue would be extremely sensitive were testing to be performed on adolescent girls in the early stages of breast development and could be devastating to their developing their sense of identity.

When preventive approaches carry few risks, however, for example, not smoking, physicians and counselors can be more directive, especially with minors. Andrews states that "some physicians are advising parents of children with a genetic propensity toward skin cancer to move to an area with a rainy climate." She also argues that as genetic research progresses, health care providers may be liable for not talking to patients about preventive strategies such as diet, job, and climate.[72]

Follow up—whose responsibility?

Physicians

When a physician performs a test for a genetic predisposi-

tion on a young child solely for predictive purposes and a number of years later therapies or new preventive strategies which were not known at the time of testing become available, some have argued that the physician has an obligation to follow up with these patients. No legal obligation of this sort currently exists, and we argue, as a matter of public policy, that such a burden should not be placed on physicians. Those who advocate this legal duty for physicians[73] generally refer to *Tresemer v. Barke*.[74] In that case, a physician was found to have a duty to warn his patient of the dangerous side-effects of a Dalkon Shield intrauterine device when, subsequent to its insertion, the physician obtained knowledge of its hazards. The court reasoned that "the general duty of care should be extended to include the duty to avoid further danger to a person, even when the original danger was created by innocent conduct."[75] But the California Court of Appeals also listed a number of factors that were appropriate to its decision as to whether to impose on a physician a duty to warn a patient about new information acquired about the dangers associated with a medical device. These factors included

> the foreseeability of harm to the plaintiff, the degree of certainty that the plaintiff suffered injury, the closeness of the connection between the defendant's conduct and the injury suffered, the moral blame attached to the defendant's conduct, the policy of preventing future harm, the extent of the burden to the defendant and [the] consequences to the community of imposing a duty to exercise care with resulting liability for breach, and the availability, cost and prevalence of insurance for the risk involved.[76]

Given these considerations, it is highly unlikely that a duty would be imposed on a physician, who provides a genetic test but has no ongoing physician-patient relationship, to re-contact the patient years after the test was done. For the physician who provides a genetic test and its result, the physician's actions have not created a risk to the patient; rather the physician has simply provided the patient with information about a preexisting risk. In this way, the genetic testing scenario is quite different from a physician who gives a patient a drug or implants a medical device where the drug or device creates a risk to the patient.

The physician's responsibility may also be tied to the nature of the physician-patient relationship. For example, a physician's duty to inform a patient of danger associated with a treatment previously given is abrogated when the physician-patient relationship ends. In *Fleischman v. Richardson-Merrell, Inc.*,[77] a physician was found not to owe a former patient a duty to warn about the side-effects of a drug that he had prescribed for her many months prior to his discovery of its side-effects. The patient was treated by the physician on July 15, 1960, for an abnormally high cholesterol level, at which time he prescribed the drug. The patient spoke to the physician only once thereafter—one week later—at which time he checked her condition. He did not treat or see the patient between July 1960 and December 1961. However, the patient continued to use the drug until December 1961 by purchasing it under its trade name. In that month, the first announcement was made to the medical profession of the dangerous propensity of the drug. The court held that "[t]he physician had no duty to continue to follow her progress after the prescription expired because she left his treatment and he had no knowledge she was continuing to use the drug, or reason to know of it."[78]

In jurisdictions that adopt this perspective, it is unlikely that a follow-up duty would be imposed on a physician who performs a genetic test but does not see the patient again. An argument can be made, however, that the genetic testing situation is distinct from the case where a physician has reason to believe his/her patient has stopped taking a potentially harmful drug and is no longer at risk. With a genetic test, the patient continues to be at risk of developing the disease, and thus any new information about the test or genetic condition would be relevant to the patient. But, again, the nature of the two acts on the part of the physician are distinct—giving a patient a drug has inherent physical risks, while giving a patient a genetic test provides that patient with information about a preexisting risk.

A number of policy arguments have been made for not placing this duty on the physician once the physician-patient relationship ends. First, putting the onus on physicians is an unfair burden. For the physician who does the genetic testing, it may be impossible for him/her to contact a former patient, especially given the mobility of families in today's society.[79] Second, the potential exists for a significant expansion of the requirement. What is the rationale of not imposing the same duty on physicians to re-contact other former patients when new treatments or preventive interventions for their conditions become available—for example, a new surgical procedure for heart disease or a new diet to reduce blood pressure. For these reasons, a strong policy argument can be made to limit a physician's duty to contact only those patients with whom the physician has an ongoing relationship.

While we do not think a physician should have a legal duty to follow up, physicians who test a young child must also inform parents of their need to stay in contact with a knowledgeable pediatrician or geneticist regarding their child's disease. We advocate that parents be assisted in their efforts to follow new developments in the treatment of their child's disease by the establishment of mechanisms like voluntary registries or toll free numbers, where individuals can find out the most up-to-date information on their child's disease.

Genetic registries

Disease registries have existed in the United States and other countries for several decades.[80] They have been most often used for specific cancers and tumors, but they have also been established for victims of stroke, psychiatric disorders, tuberculosis and other infectious diseases, birth defects, chronic renal disease, blindness, heart disease, occupational diseases, and trauma. More recently, they have been set up for individuals needing organ and bone marrow transplants. The registries have been established primarily by private entities—academic institutions or foundations for purposes of research—and most are voluntary in nature. In some cases, government agencies have established registries to collect epidemiological information for planning purposes or for identifying individuals in need of health or other services. An example of the first type of registry is the cancer registry established by the Centers for Disease Control (CDC).[81] An example of the second type includes registries of genetic diseases such as PKU for newborn screening programs.

One of the most weighty arguments against genetic registries in general, but specifically mandatory registries, is the potential infringement on the confidentiality of information provided to one's physician. This is especially troublesome given the stigma that may be associated with some genetic conditions and predispositions, and the potential loss of various types of insurance or employment. But even more forceful is the argument that genetic information contains information not only about the person tested but also about their parents, siblings, and children or future children. Some have argued against registries for this very reason. Others have argued for heightened privacy rules for a specialized type of registry—DNA data banks.[82] Given these concerns, the appropriateness of mandatory reporting to a genetic registry is highly questionable. Moreover, it is likely to be politically infeasible.

Recent congressional debate over the development of a national system to track the immunization status of children, in the Comprehensive Child Immunization Act of 1993,[83] illustrates this latter concern. The act would have authorized the secretary of Health and Human Services, in consultation with state public health officials, to establish state registry systems to monitor the immunization status of all children. The act was introduced in response to the nation's "alarmingly low" preschool immunization rate.[84] The legislation authorized optional grants to states to develop immunization registries and to supply aggregate state data to the CDC to guide federal efforts to improve immunization rates.

The registry provisions contained in the bill, as originally introduced, provoked concern about the protection of parental rights, privacy, and the potential use of registry data for purposes other than immunizations. During debate on the bill, hundreds of parents wrote to their legislators in opposition.[85] Although the bill was amended to address some of their concerns, the registry provisions did not survive passage.[86]

While any effort to create a national or even a system of mandatory statewide registries for purposes of genetic follow up would likely meet at least as much (if not more) opposition as that generated by the vaccine registry proposal, voluntary registries may be appropriate for specific genetic diseases. Such diseases would include those considered life-threatening or those that have the potential to affect seriously the quality of a child's life. Such registries should include guarantees of confidentiality and should be responsible for contacting registrants with beneficial information about their condition.

Safeguards protecting children—a recommendation

Given concerns about testing children for genetic predispositions, we urge adoption of safeguards to ensure that the tests are administered consistent with the child's best interests. While an argument can be made that safeguards should be in place for all tested, children are particularly vulnerable to the potential negative effects of predisposition testing. The most compelling argument for this is the impact a positive test result may have on how a child will be treated by his parents, family, and, potentially, society. Few empirical studies have been done on this issue (other than the Swedish experience with alpha$_1$-antitrypsin deficiency),[87] but caution in this type of testing is now warranted, and professional societies must play a role in encouraging physicians to exercise restraint in this area. Specifically, relevant professional societies such as AAP and ASHG should issue guidelines providing that physicians not generally disclose the availability of or perform genetic tests on children solely for predictive purposes where there is no history of the disease in the child's family or any immediate treatment or surveillance benefit to the child. We support ASHG's and ACMG's statement on testing children, but we also encourage NIH to continue to establish consensus panels to determine when testing is appropriate for children for *specific* genetic diseases as well as when specific preventive or treatment interventions may be appropriate. The relevant professional societies should adopt these guidelines and urge physicians to do likewise. As pointed out by Wertz et al., "professional guidelines offer greater flexibility than legal regulations in implementing standards" and would also serve as a counterbalance to pressures from biotechnology companies to test and to fears of malpractice suits.[88]

Where the guidelines indicate that testing might be appropriate and the parents want that genetic testing performed, we recommend that the child's physician, if not qualified himself, refer the child and his/her parents to a genetic counselor or knowledgeable physician to discuss

the test risks (including the psychological risks and the potential impact on family dynamics). We urge that counselors discuss with parents the risk of overvaluing information that can be obtained from genetic tests and not discount the subtle ways in which this information might psychologically harm a child by virtue of treatment by family, friends, and school systems. Finally, counselors should help parents to think through how they will use the information and at what point and under what conditions they will tell their child about a positive test result. The family, after meeting with the counselor, may still desire the test, but the additional counseling and discussion should clarify the issues and make parents more knowledgeable about the risks of the tests.

In addition, all parents seeking genetic testing of a child should give written consent to the procedure. State departments of public health (or comparable agencies) should consider designing model consent forms that list the potential risks and benefits of the proposed test, including the psychological risks and the impact on family dynamics. Forms prepared by some state health departments for HIV testing may serve as models. We also recommend, in all cases where a child tests positive for a genetic predisposition, that the child (if sufficiently mature) and his parents be provided the opportunity to meet with a genetic counselor or knowledgeable physician to explain the results. If necessary, psychological counseling should also be made available to the family.

As regards follow-up, physicians who offer or perform a predictive genetic test on a child have an obligation to tell the family that they should check back periodically with the physician to determine whether any new developments might benefit the child. Physicians should also know about possible resources for the family, including toll-free numbers, family support groups, or disease registries, that could assist them in keeping up-to-date regarding their child's condition. Finally, for certain life-threatening conditions or conditions that have the potential to effect the quality of a child's life significantly, registries should be established by national public health agencies or private disease associations. These registries would be voluntary, would track cases of genetic predispositions, and would inform registrants of new developments that could benefit them.

Conclusion

The availability of more and more genetic tests will create unique dilemmas for parents, their children, and their health care providers. We advise caution in the administration of these tests to children when such testing is solely for predictive purposes. Professional associations must take a strong stand on this issue and should provide physicians with guidance as to when they should disclose the availability of tests to families as well as to when it would be appropriate to perform such tests. If a predictive test is appropriate for a child, based on established guidelines, safeguards must be in place to ensure accurate and informed decision making by parents and child (if sufficiently mature). Finally, if predictive testing is done on a young child, resources should be made available, through government funding or private agencies, to assist parents in keeping informed about their child's condition and any beneficial interventions. In addition to the establishment of registries for some conditions, public health education and information dissemination strategies should be implemented to ensure that when new information is available, it reaches a large segment of the population. This way, parents of children like Michael, with the predisposing gene for colon cancer, will bring their children in to see a physician when a treatment or cure for colon cancer is available.

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References

1. D. Birch, "Genetic Fortune-Telling," *Baltimore Sun*, May 9, 1995, at A1.

2. Tests are also predicted to be available for predispositions to "complex conditions and behaviors," such as "mental illness, Alzheimer's disease, hyperactivity, heart disease, ... and susceptibility to alcoholism, addiction, and even violence." Some have described those persons who test positive for these conditions as the "pre-symptomatically ill" or the "person 'at-risk'." R.C. Dreyfuss and D. Nelkin, "The Jurisprudence of Genetics," *Vanderbilt Law Review*, 45 (1992): at 318.

3. According to newspaper reports, the tests cost approximately $800 for the first family member and $250 for each additional member. They are being offered under research protocols to cancer families by at least one biotechnology company. See G. Kolata, "Tests to Assess Risks for Cancer Raising Questions," *New York Times*, Mar. 27, 1995, at A1.

4. See M.R. Natowicz and J.S. Alper, "Genetic Screening: Triumphs, Problems, and Controversies," *Journal of Public Health Policy*, 12 (1991): at 485.

5. Kolata, *supra* note 3, at A9.

6. See, for example, N.F. Sharpe, letter, "Pre-Symptomatic Testing for Huntington Disease: Is There a Duty to Test Those Under the Age of Eighteen Years?," *American Journal of Medical Genetics*, 46 (1993): 250–53.

7. M.Z. Pelias, "Duty to Disclose in Medical Genetics: A Legal Perspective," *American Journal of Medical Genetics*, 39 (1991): at 350.

8. Kolata, *supra* note 3.

9. See, for example, N. Fost, "Genetic Diagnosis and Treatment: Ethical Considerations," *American Journal of Diseases of Children*, 147 (1993): 1190–95; D.C. Wertz et al., "Genetic Testing for Children and Adolescents: Who Decides?," *JAMA*,

272 (1994): 875–82; and P.S. Harper and A. Clarke, "Viewpoint: Should We Test Children for 'Adult' Genetic Disease?," *Lancet*, 335 (1990): 1205–06.

10. *Id.*

11. American Society of Human Genetics and American College of Medical Genetics, "Points to Consider: Ethical, Legal, and Psychosocial Implications of Genetic Testing in Children and Adolescents," *American Journal of Human Genetics*, 57 (1995): 1233–41.

12. Wertz et al., *supra* note 9; and Fost, *supra* note 9.

13. *Id.*

14. The Committee on Assessing Genetic Risks, which was appointed by the Institute of Medicine, recommends that "[c]hildren should generally be tested only for genetic disorders for which there exists an effective curative or preventive treatment that must be instituted early in life to achieve maximum benefit." See Institute of Medicine, L.B. Andrews et al., eds., *Assessing Genetic Risks: Implications for Health and Social Policy* (Washington, D.C.: National Academy Press, 1994): at 10. Detailed statements on the testing of children have been prepared by the Working Party of the Clinical Genetics Society in the United Kingdom and, more recently, by ASHG and ACMG, see *supra* note 11. The Working Party makes several recommendations, including the following:

> (1) The predictive genetic testing of children is clearly appropriate where onset of the condition regularly occurs in childhood or there are useful medical interventions that can be offered (for example, diet, medication, surveillance for complications). (2) In contrast, the working party believes that predictive testing for an adult onset disorder should generally not be undertaken if the child is healthy and there are no medical interventions established as useful that can be offered in the event of a positive test result.

A. Clarke, Working Party of the Clinical Genetics Society, "The Genetic Testing of Children," *Journal of Medical Genetics*, 31 (1994): at 785.

15. Clarke, *supra* note 14; but see Harper and Clarke, *supra* note 9.

16. Institute of Medicine, *supra* note 14, at 5.

17. T.F. McNeil et al., "Psychological Effects of Screening for Somatic Risk: The Swedish Alpha$_1$-Antitrypsin Experience," *Thorax*, 43 (1988): 505–07.

18. T. Thelin et al., "Psychological Consequences of Neonatal Screening for Alpha$_1$-Antitrypsin Deficiency (ATD)," *Acta Paediatrica Scandinavica*, 74 (1985): 787–93.

19. *Id.*

20. W.H. Schutt and T.E. Isles, "Protein in Meconium from Meconium Ileus," *Archives of Diseases of Children*, 43 (1968): 178–81.

21. P.M. Farrell and E.H. Mischler, "Newborn Screening for Cystic Fibrosis," *Advances in Pediatrics*, 39 (1992): at 66.

22. N.A. Holtzman, "What Drives Neonatal Screening Programs?," *N. Engl. J. Med.*, 325 (1991): 802–04.

23. D.M. Bradley et al., "Experience with Screening Newborns for Duchenne Muscular Dystrophy in Wales," *British Medical Journal*, 306 (1993): 357–61.

24. *Id.*

25. Wertz et al., *supra* note 9; and Clarke, *supra* note 14.

26. Wertz et al., *supra* note 9, at 878, citing J.H. Fanos, *Developmental Consequences for Adulthood of Early Sibling Loss* (Ann Arbor: University of Michigan Microfilms, 1987), and J. Dunn, *Sisters and Brothers* (Cambridge: Harvard University Press, 1985).

27. F.P. Li et al., "Recommendations on Predictive Testing for Germ Line p53 Mutations Among Cancer-Prone Individuals," *Journal of the National Cancer Institute*, 84 (1992): at 1157–58.

28. Fost, *supra* note 9, at 1193.

29. *Id.* citing A. Tluczek et al., "Parents' Knowledge of Neonatal Screening and Response to False-Positive Cystic Fibrosis Screening," *Journal of Developmental and Behavioral Pediatrics*, 13 (1992): 181–86.

30. D. Ball et al., "Predictive Testing of Adults and Children," in A. Clarke, ed., *Genetic Counselling: Practice and Principles* (London: Routledge, 1994): at 70.

31. *Id. at* 74.

32. Wertz et al., *supra* note 9, at 876, citing A. Gabow et al., "Gene Testing in Autosomal Dominant Adult Polycystic Kidney Disease: Results of a National Kidney Foundation Workshop," *American Journal of Kidney Diseases*, 13 (1989): 85–87. Also, according to Biesecker et al., the long-term outcome of polycystic kidney disease is not altered by early identification. The impetus for testing individuals for the disease comes primarily from "efforts to identify unaffected living related renal transplant donors at an age where renal ultrasound will not accurately identify all pre-symptomatic carriers; this can result in the identification of carriers for a disease in which early clinical intervention does not alter the course." The National Kidney Foundation only endorses testing presymptomatically for the disease as part of evaluation for renal transplant donation. B.B. Biesecker et al., "Genetic Counseling for Families with Inherited Susceptibility to Breast and Ovarian Cancer," *JAMA*, 269 (1993): at 1971.

33. American Society of Human Genetics, *supra* note 11.

34. Those in the public health field have made the argument that knowledge of a patient's genetic predispositions will be central to preventive medicine. Collins, director of the Human Genome Project at the National Institutes of Health, believes that information gained from genetic testing will play a crucial role in prevention:

> It's very possible that in the future a physician will give an 18-year-old patient a physical exam that includes a test of his or her DNA for hundreds of diseases with known genetic components. Family histories are useful, but genetic exams will give the doctor a much more precise tool to assess risks and give advice. The physician will be able to tell the patient whether the risk is high, low, or average for a given condition and to make life-style recommendations based upon known risks. There will be personalized schemes for a new kind of preventive medicine....

D. Breo, "Altered Fates—Counseling Families with Inherited Breast Cancer," *JAMA*, 269 (1993): at 2021.

Some argue that certain genetic tests may even replace other costly and inconvenient preventive screening tests currently in use. For example, the genetic test for colon cancer may free some patients from an annual colonoscopy, which can cost as much as $1,000 each per test. S. Brownlee et al., "Tinkering with Destiny," *U.S. News & World Report*, Aug. 22, 1994, at 61.

35. Currently, high-risk women may enroll in a tamoxifen chemo-prevention trial. This long-term randomized trial will compare the effects of tamoxifen with those of placebo in 16,000 women at increased risk for breast cancer. C. Lerman and R. Croyle, "Psychological Issues in Genetic Testing for Breast Cancer Susceptibility," *Archives of Internal Medicine*, 154 (1994): 609–17.

36. Lerman and Croyle point out that "[w]omen at high risk for breast cancer are increasingly seeking counseling about prophylactic mastectomy (PM), and PM has been advocated for selected members of HBC [hereditary breast cancer] families.... The efficacy of PM has yet to be established in controlled trials." In terms of psychological benefits, they argue that "[p]rophy-

lactic mastectomy may provide important psychological benefits, especially for women in high-risk families. For example, long-term uncertainty and worry might be reduced, as would dependence on screening and self-examination. Prophylactic mastectomy may also reduce the likelihood of experiencing the distress associated with false-positive mammography results." *Id.* at 613.

37. In the case of colon cancer, for example, only about 65 percent of those with the gene will develop the disease. "The Year in Genes," *Discover*, Jan. 1994, at 92. For those with the breast cancer gene, the odds are somewhat higher—an 85 percent lifetime risk of developing the disease. Biesecker et al., *supra* note 32, at 1970.

38. National Advisory Council for Human Genome Research, "Statement on Use of DNA Testing for Pre-Symptomatic Identification of Cancer Risk," *JAMA*, 271 (1994): 785; and "Statement of the American Society of Human Genetics on Genetic Testing for Breast and Ovarian Cancer Predisposition," *American Journal of Human Genetics*, 55 (1994): i–iv.

39. See, for example, Pelias, *supra* note 7; and Sharpe, *supra* note 6.

40. See, for example, *Berman v. Allan*, 404 A.2d 8, 15 (N.J. 1979) (court held that parents of a congenitally defective child had a valid claim for compensation for their mental and emotional suffering over the birth of the child when the claim was based on the failure of the expectant mother's doctors to inform the parents of the availability of the diagnostic procedure known as amniocentesis); see also *Phillips v. United States*, 566 F. Supp. 1 (D.S.C. 1981); and *Becker v. Schwartz*, 386 N.E.2d 807 (N.Y. 1978). These cases are often referred to as wrongful birth cases.

41. See, for example, *Munro v. Regents of the University of Cal.*, 263 Cal. Rptr. 878, 882 (Cal. Ct. App. 1989) (doctor did not commit medical malpractice in failing to check whether pregnant woman and her husband were carriers of Tay-Sachs disease, even though the couple's child was later born with Tay-Sachs, when defendants submitted expert evidence that the couple did not meet the profile characteristics necessary to warrant performing a Tay-Sachs carrier screening test); see also *Roth v. Group Health Ass'n, Inc.*, Dkt. No. 88-1005 (D.D.C., settled June 12, 1989) (woman who knew she had a genetic defect in her family sued her doctor and HMO for failing to perform amniocentesis because she was under the age of thirty-five; when her child was born with the defect, she filed a malpractice suit and received a $925,000 settlement).

42. See, for example, Md. Code Ann., Health-Gen. § 5-613(a) (1994) (if a patient or his agent or surrogate requests that everything be done for a seriously ill patient, including CPR, and the treating physician believes that CPR would be medically ineffective, the physician must inform the patient of the option to transfer the patient to another provider and must assist in that process).

43. E.W. Clayton, "Removing the Shadow of the Law from the Debate about Genetic Testing of Children," *American Journal of Medical Genetics*, 57 (1995): at 630–32.

44. Pelias, *supra* note 7.

45. To be successful in a case based on lack of informed consent, a plaintiff must show that (1) he consented to the test or treatment without the physician revealing all relevant risks, (2) he would not have consented to the test or treatment had he had complete information about the test or treatment, (3) the unrevealed risk did, in fact, materialize, and (4) the plaintiff suffered injury as a result. See, for example, *Nickell v. Gonzalez*, No. C-830460 (Ohio Ct. App., filed Feb. 22, 1984).

46. L.B. Frantz, "Modern Status of Views as to General Measure of Physician's Duty to Inform Patient of Risks of Proposed Treatment," *American Law Review 3d*, 88 (1978): at 1012–13; (Supp. 1995): at 78. Cases that measure the physician's duty to inform of risks by the customary disclosure practice sometimes specify that the custom must be that of physicians practicing in the community, in the locality, or in the area. *Id.*

47. *Id.* at 1016–20; (Supp. 1995): at 78-79.

48. *Id.*

49. Biesecker et al., *supra* note 32, at 1972.

50. See, for example, Maryland Department of Health and Mental Hygiene, "Informed Consent and Agreement to HIV Testing" (Form 95-2) ("If my test is positive, I may experience emotional discomfort and, if my test result becomes known to the community, I may experience discrimination in work, personal relationships, and insurance.")

51. See, for example, Wertz et al., *supra* note 9, at 878: "Minors who request testing should be informed, before testing, that third parties such as employers, insurers, and schools may be able to coerce their consent for access to test results by withholding employment, insurance, or school admission."

52. Kolata, *supra* note 3, at A9.

53. Wertz et al., *supra* note 9, at 879.

54. *Id.*

55. See, for example, Minn. Stat. Ann. §§ 144.341, 144.342 (West 1989).

56. See, for example, Miss. Code Ann. § 41-41-3(h) (1993).

57. Some states have enacted legislation protecting from liability those physicians who make a good faith effort to determine that a child/adolescent is mature enough to make a decision. See Miss. Code Ann. § 41-41-5 (1993); N.C. Gen. Stat. § 90-21.4 (1994); and Minn. Stat. Ann. § 144.345 (West 1989).

58. Such cases are likely to be quite rare. Very little research has been done on adolescents' interest in genetic testing for certain conditions. Nonetheless, one study of high school students' attitudes toward carrier screening for CF found that, although the students were quite receptive to the concepts of carrier screening and prenatal diagnosis, their attitudes changed considerably when confronted with the reality that they might be CF carriers. Levels of indecision increased markedly, and very few students (5/101) wished to be informed of their AF508 carrier status. A. Page et al., Abstract, "Attitudes of High School Students Toward Carrier Screening for Cystic Fibrosis," *American Journal of Human Genetics*, 51 (1992): A17.

59. See, for example, Cal. Civ. Code § 34.5 (West 1975).

60. Biesecker et al., *supra* note 32, at 1973. Researchers went on to say that this issue remains unsettled in protocols for testing Li-Fraumeni family members for the presence of p53 mutations, "despite clinical manifestations in children and adolescents." *Id.*

61. See, for example, *Dowling v. Mutual Life Ins. Co.*, 168 So. 2d 107, 118 (La. Ct. App. 1964) (duty of physician to inform patient that a test revealed the possibility of tuberculosis, despite evidence that the patient was a very apprehensive person, quite fearful of heart or lung trouble); and *Ray v. Wagner*, 176 N.W.2d 101, 104 (Minn. 1970) (duty of physician to take whatever steps possible to notify patient that results of a pap smear test indicated a possibility that she was suffering from cervical cancer).

62. S. Wiggins et al., "Psychological Consequences of Predictive Testing for Huntington's Disease," *N. Engl. J. Med.*, 327 (1992): 1401–05.

63. C.M. Benjamin et al., "Proceed with Care: Direct Predictive Testing for Huntington's Disease," *American Journal of Human Genetics*, 55 (1994): 606–17.

64. J.H. Fanos and J.P. Johnson, "Barriers to Carrier Testing

for Adult Cystic Fibrosis Sibs: The Importance of Not Knowing," *American Journal of Medical Genetics*, 59 (1995): 185–91.

65. Lerman and Croyle, *supra* note 35, at 610. However, the authors also state that

> [w]hether breast cancer morbidity and mortality can be reduced through the adoption of prevention and surveillance practices remains an open question. If current prevention trials provide conclusive evidence of efficacy, a more prescriptive approach to counseling high-risk women would be suggested. This approach emphasizes personal risk, as well as options and advice for possible breast cancer prevention and early detection. This is in sharp contrast to genetic counseling for Huntington disease, which emphasizes existential issues surrounding inevitable premature death and employs a nondirective counseling approach to protect a woman's privacy in making reproductive decisions.

Id.

66. "Even when people comprehend the numbers (and many cannot), they find uncertainty psychologically troubling. When confronted with a 50 percent risk, many patients conclude their chances are either zero or 100 percent...." Brownlee et al., *supra* note 34, at 63, quoting Biesecker, head of genetic counseling at the National Center for Human Genome Research.

67. Biesecker et al., *supra* note 32, at 1972.

68. *Id.*

69. *Id.*

70. *Id.* Along with these options, women were informed

> that age of onset of ovarian cancer in BRCA1 mutation carriers may be later than the age of onset of breast cancer, although significant uncertainty exists. This may offer a window of opportunity for younger women identified as carriers to complete childbearing prior to making a decision about having their ovaries removed. Recommendations were made for follow-up with surgical oncologists, plastic surgeons, and gynecologists for further discussion of surgical options.... Team members attempted to review options for screening versus prophylactic surgery without making specific recommendations.

Id. at 1973.

71. Health care providers have been held liable for encouraging a "preventive" course of action without assuring the accuracy of a test. See *Avery v. County of Burke*, 660 F.2d 111, 116 (4th Cir. 1981) (court held that fifteen-year-old pregnant woman inaccurately diagnosed with sickle cell trait had a valid cause of action against the clinic for violation of her civil rights by wrongfully causing her sterilization). This case is discussed in L.B. Andrews, "DNA Testing, Banking, and Individual Rights," in B.M. Knoppers and C.M. Laberge, eds., *Genetic Screening: From Newborns to DNA Typing* (New York: Excerpta Medica, 1990): at 217.

72. Andrews, *supra* note 71, at 223.

73. *Id.* at 223–26; and Pelias, *supra* note 7, at 353; see also G.J. Annas, "Privacy Rules for DNA Databanks Protecting Coded 'Future Diaries'," *JAMA*, 270 (1993): at 2350 (stating that "[t]here is legal precedent for holding physicians responsible to recontact patients when the physician learns of a new danger related to previous treatment").

74. *Tresemer v. Brake*, 150 Cal. Rptr. 384 (Cal. Ct. App. 1978).

75. A.G. Nadel, "Duty of Medical Practitioners to Warn Patient of Subsequently Discovered Danger from Treatment Previously Given," *American Law Review 4th*, 12 (1982): 41–45; (Supp. 1995): 4–5.

76. 150 Cal. Rptr. at 392.

77. *Fleischman v. Richardson-Merrell, Inc.*, 226 A.2d 843 (N.J. Super. Ct. App. Div. 1967).

78. *Id.* at 845.

79. See, for example, M.J. Khoury et al., "Residential Mobility During Pregnancy: Implications for Environmental Teratogens," *Journal of Clinical Epidemiology*, 41 (1988): 15–20 (in the Maryland State Birth Defects Registry Data for 1984, 20 percent of mothers changed their address between pregnancy conception and delivery; this was a cross-sectional socioeconomic group ascertained in order to locate children born with birth defects).

80. This is in contrast to DNA data banks, which may actually store a DNA sample or have on file a complex genetic profile of an individual. DNA data banks have been established in a number of states, primarily for individuals convicted of sex offenses, but in some states for other serious felonies. See, for example, statutes in Arizona, California, Colorado, Florida, Iowa, Minnesota, Nevada, South Dakota, Virginia, and Washington. These statutes are cited in E.D. Shapiro and M.L. Weinberg, "DNA Data Banking: The Dangerous Erosion of Privacy," *Cleveland State Law Review*, 38 (1990): at 473.

81. In 1992, Congress passed the Cancer Registries Amendment Act (42 U.S.C. § 280e (Supp. 1994)). It authorized the CDC to provide funds to states and territories to enhance existing cancer registries, to plan and implement registries where they do not exist, to develop model legislation and regulations for states to enhance viability of registry operations, and to set standards for completeness, timeliness, and quality of data received. The act requires that each applicant provide assurances that it will obtain state authorization to operate a statewide cancer registry, including regulations that "assure complete reporting of cancer cases ... to the statewide cancer registry by hospitals ... physicians, surgeons, and all other health care practitioners diagnosing or providing treatment for cancer patients." 42 U.S.C. § 280e(c)(2)(D)(i–ii). Regulations must also address "the protection of the confidentiality of all cancer case data, including a prohibition [of] ... the identification of ... an individual cancer patient." 42 U.S.C. § 280e(i)(2)(D)(v).

82. Annas, *supra* note 73, at 2349.

83. Comprehensive Child Immunization Act of 1993, S. 732, 103d Cong., 1st Sess. (1993).

84. 139 *Congressional Record* S15083 (daily ed. Nov. 4, 1993) (statement of Senator Kassebaum). Senator Kassebaum also stated that "[f]ewer than 60 percent of 2-year-olds in most states are fully immunized, and in some cities, fewer than 10 percent are fully immunized."

85. *Id.* (statement of Senator Helms).

86. Although federal legislation on this topic did not pass, several state legislatures have enacted legislation establishing an immunization registry designed to track every child's immunization record via a data base. See Tenn. Code Ann. § 37-10-401(c) (1995); Conn. Gen. Stat. Ann. § 19a-7h(a) (West Supp. 1995); and 1995 N.C. Sess. Laws 324.

87. McNeil et al., *supra* note 17; and Thelin et al., *supra* note 18.

88. Wertz et al., *supra* note 9, at 880.