1-9-1996

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Genetic Testing in Children and Adolescents: Parental Authority, the Rights of Children, and Duties of Geneticists

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I. Introduction

During the past decade technological capabilities in molecular genetics have expanded exponentially.¹ Concomitant with technological growth developed concern about using new molecular technologies to generate personal genetic information about individuals and families who are coping with various genetic disorders. Geneticists began to scrutinize the relationships that arise in the context of genetic counseling, and, in particular, several geneticists and psychologists suggested that no children should ever be tested for the gene that causes Huntington disease,² even when testing is requested by their parents.³ This suggestion raised several issues that have direct bearing on decisions about genetic testing, including the nature of the disease and the availability of treatment, the roles of parents and the rights of children within the family, and the obligations of geneticists who participate in both testing and counseling.

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One potential effect of the policy suggested for Huntington disease was a realignment of the professional and personal relationships that arise when a patient or family seeks genetic evaluation and help. Geneticists and genetic counselors have traditionally acted from a perspective of providing information and counseling that will help patients and families understand and cope with their genetic problem on their own terms, within their own constellation of circumstances. If geneticists institute policies about who should or should not have access to genetic tests, decisions about testing will no longer rest with patients or families, and geneticists will place themselves squarely in the middle of families who are trying to deal with their own genetic legacies.

Geneticists are currently amassing a plethora of tests and information that may be of critical interest to individuals and families who have a stake in planning their own lives with a knowledge of their own genetic endowment. What is now incumbent on geneticists, genetic counselors, physicians, and other medical professionals is careful evaluation of their roles and influences in the lives of their patients. This consideration includes an examination of the changing nature of the physician-patient relationship as more sophisticated methods are developed for detecting and coping with the spectrum of genetic diseases. Also included in the examination of the practice of contemporary medical genetics is careful comparison of the relative positions of parents, children, and medical professionals in the changing world of medical genetics and genetic counseling.

II. The Physician-Patient Relationship

A physician-patient relationship is formed when a patient seeks medical care and the physician agrees to provide such care. Technically, the relationship is based on an implied contract, with duties and expectations attaching to both parties at the time the relationship is joined. The physician assumes a duty of providing due care to the patient, and expects remuneration and cooperation. The patient assumes a duty to assist in his medical management and expects the physician to provide appropriate care. This reciprocity is modified, however, by the fact that interactions between physicians and patients take place in a fiduciary interaction—one that is typified on one side by a person who has a duty, created by his position, to act primarily for the benefit of another person. The imbalance in the fiduciary relationship is based on the physician's special training and expertise in medicine and the patient's, or family's, relative lack of medical knowledge. Within the context of medical genetics, the fiduciary relationship relies on a genetics professional who has

both a broad knowledge of genetics and an ability to convey relevant information to people who seek it.

Beyond the continuing imbalance in the physician-patient relationship, however, is a substantive metamorphosis that has taken place during this century in the respective roles of the physician and patient in selecting an appropriate course of treatment. Prior to the twentieth century, medical knowledge was limited, with few if any options for patient care. However, the advent of anesthesia and the explosion of medical knowledge over the last 100 years have dramatically increased options for patients, and the physician-patient relationship has experienced a significant shift in the basis of decisional authority. A sequence of consistent judicial decisions has reinforced the right of the patient to make his own choices about which course of treatment to pursue. The shift in decisional authority has redefined the fiduciary duty of the professional, so that the duty of the physician has evolved from one of making the best decision for the patient to one of informing the patient about various options in order to permit the patient to make a fully informed decision. This new balance in the physician-patient relationship continues to acknowledge the physician's superior knowledge, but it also gives deference to patient autonomy for deciding among options for medical treatment, or indeed, for no treatment at all.

The case-law approach to the physician-patient relationship has broadened the concept of medical malpractice to include the "Doctrine of Informed Consent." From this doctrine is derived the principle that the patient, rather than the medical professional, is the primary decision maker in matters of medical care and treatment. To be sure, the physician may continue to exercise some control with respect to how, and how much, information is conveyed to the patient. But, nowadays, the ultimate power of decision rests with the patient—the person who must live with these decisions for a lifetime. In the context of medical genetics, patients and families who seek genetic counseling may reasonably expect to be fully informed about available tests and the implications of test results.

III. The Nature of the Disease

When researchers in Huntington disease suggested that no children be tested for the presence of the gene, they offered the rationale that testing children would be pointless because the disease has a late age of onset and no

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9. Mohr v Williams, 95 Minn 261, 269, 104 NW 12 (1905); Schloendorff v Society of New York Hospitals, 211 NY 125, 105 NE 92, 93 (1914); Canterbury v Spence, 464 F2d 772, 780 (DC Cir 1972).
10. Faden and Beauchamp, Informed Consent at 140 (cited in note 8).
presently available treatment.\textsuperscript{12} This argument against testing children for the Huntington gene does have some appeal: the child may gain nothing from presymptomatic testing, and may even be harmed by knowledge of inevitable debilitation. The argument continues with the assertion that the right to generate such devastating genetic information should rest with the child, who may later exercise that right after reaching adulthood, according to his own wishes. This line of reasoning has also been advanced by some genetics professionals who counsel about the dominant gene that causes familial breast cancer.\textsuperscript{13} What this argument fails to acknowledge, however, are the interests and obligations of parents, who are charged by society with providing for their children, and who may well be able to justify their quest for learning their children's genotypes even if the gene may not begin to work its damage until years later.\textsuperscript{14}

A somewhat different set of concerns arises in families coping with other types of genetic diseases, including one form of familial cancer, familial adenomatous polyposis of the colon (FAP).\textsuperscript{15} This disease appears in late childhood or adolescence and is amenable to surgical treatment. Psychological studies of children at risk for carrying the gene for FAP have revealed that children are intuitively aware of the risks and are often relieved to learn their genetic status, whether positive or negative.\textsuperscript{16} Children may even develop a unique, positive sense of "belonging" when they learn they are gene-positive,\textsuperscript{17} or alternatively, they may experience a unique, negative sense of "survivor guilt" if they are found to be gene-negative.\textsuperscript{18} Experience with FAP and other diseases that have a childhood onset and an option for treatment reveals that

\begin{itemize}
\item \textsuperscript{12} Bloch and Hayden, 46 Am J Hum Genetics at 2 (cited in note 3).
\item \textsuperscript{14} See, infra, notes 25-32 and accompanying text.
\item \textsuperscript{15} Familial adenomatous polyposis of the colon is caused by a dominant gene on chromosome 5. The appearance of benign polyps in the colon is a prelude to malignant transformations and bowel cancer. Children who are known to carry the gene can be monitored for the appearance of polyps and can avoid developing the cancer if the bowel is surgically removed. See generally, Thoene, \textit{Physicians' Guide to Rare Diseases} (cited in note 2).
\item \textsuperscript{17} Peterson and Boyd, 17 Monograph J Natl Cancer Inst at 69 (cited in note 16).
\item \textsuperscript{18} The sense of "belonging" relates to the child's closer identification with the affected parent and with siblings who are gene-positive. "Survivor guilt" may develop when a child who is gene-negative realizes that he or she may out-live siblings who are gene-positive. Dorothy C. Wertz, Joanna H. Fanos, and Philip R. Reilly, \textit{Genetic Testing for Children and Adolescents: Who Decides?}, 272 JAMA 875, 876 (1994).
\end{itemize}
reactions of individuals and families to information about individual status vary significantly among families, a finding that underscores the unique personal circumstances of each family and the variety of scenarios that are encountered in the counseling arena.

Additional levels of genetic concern have been recognized with identification of a genetic "predisposition" to develop a disease. A number of health problems are determined by the interaction between an individual's genotype and the environmental milieu. For example, a genetic predisposition to develop dangerously high blood cholesterol levels can be tempered by adopting a low-fat diet that may permit the individual to avoid the ravages of cardiovascular disease. Similarly, children who have the gene for familial hypertrophic cardiomyopathy can be treated with medications that may prevent cardiac arrhythmias and significantly reduce the risk of sudden death. When such clear benefits are so easily realized, professionals appear to be less concerned about possible negative effects of determining and revealing the genotypes of children.

Finally, carrier status for many recessive diseases is becoming increasingly feasible as the genes that cause these diseases are mapped and cloned. Some geneticists argue that testing for carrier status should not be available for children because determination of carrier status generally has no immediate medical implications for children and may even have a detrimental effect on relationships within the family. Such determinations could, however, have significance for adolescents, because of their increasing awareness of biological processes and their own emerging sexuality, even though the likelihood of their having affected children still remains small. Perhaps the most rational course to pursue for carrier detection in children is one of attention to the views of the children and their parents: children who have siblings with recessive diseases, for example, may have an extraordinary interest in early resolution of any uncertainty about their own genetic status and their chances of having children with the same problems, so that carrier testing at the request of the children and their parents may provide considerable benefit to several members of a family.

With respect to biological and medical effects, genetic diseases vary in their modes of inheritance, age of onset, and severity of expression. With respect to social factors, genetic diseases vary greatly in the burden to both the patient and the family—a burden that includes appropriate medical care, financial sup-

port and insurance issues,\textsuperscript{23} and the personal and interpersonal effects of the disease on individual family members, both affected and unaffected.\textsuperscript{24} Given such a patchwork of medical and social issues, the rational approach to decisions about testing children seems to lie both in appreciation of the disease process and in the unique, expressed interests of the members of each family. Such an individual approach, devoid of blanket professional judgments about making tests available or not, would honor the tradition of genetic counseling as well as the autonomy and individuality of each family: the issue is not the nature of the disease itself, but rather, the situation and expectations of the family that is coping with the difficult burden of genetic disease.\textsuperscript{25}

IV. Parental Authority

The family as the basic social unit of human beings has been remarkably stable over much of history. While some social arrangements do represent deviations from the "norm," the family unit has traditionally been headed by the father, who has occupied the position of provider and protector for his family. The mother, often with a nursing infant, was less mobile, and traditionally managed the home and looked after the children. Children customarily obeyed their parents and contributed their labor to the welfare of the family group.\textsuperscript{26} Throughout much of history, the dominant role of the man as head-of-household extended to his power of life and death over his spouse and children, and his power to sell them as property. The wife had few rights, and the children virtually none.\textsuperscript{27}

The recognition of both women and children as individual persons—the acknowledgment of their personhood in society—has progressed gradually. With the development of social conscience and awareness, both women and children have become persons in the eyes of society and the law, although persons with circumscribed rights. In the balance of roles among persons in the family, society has consistently supported, first, fathers, and now parents, as the persons who make decisions for their own families. On some occasions when states have assumed a decision-making posture with respect to the care and rearing of children, the judiciary has firmly but clearly reinforced the authority of parents to decide for their own children and families.

Support for parental authority has been supplied by the Supreme Court when challenges to state legislation have run afoul of constitutional guarantees.


\textsuperscript{24} Wertz, Fanos, and Reilly, 272 JAMA at 878 (cited in note 18).

\textsuperscript{25} American Society of Human Genetics, 27 Am J Hum Genetics at 240 (cited in note 4).

\textsuperscript{26} Vivian A Zelizer, Pricing the Priceless Child: The Changing Social Value of Children 58 (Basic Books 1985).

\textsuperscript{27} Mary Martin McLaughlin, Survivors and Surrogates: Children and Parents From the Ninth to the Thirteenth Centuries, in Lloyd deMause, ed, The History of Childhood 140 (Harper & Row 1974).
Three early cases raised questions about the authority of parents to make decisions about the education of their children. One decision held that a state that forced teachers to teach only English to grade school children deprived those teachers of a liberty interest guaranteed by the fourteenth amendment. The Court defined the “liberty” interest to include “the right of the individual to contract, to engage in any of the common occupations of life, to acquire useful knowledge, to marry, establish a home and bring up children.”

Two years later the Court invalidated an Oregon law that required all children between the ages of 8 and 16 to attend public school. In speaking for parents who preferred to send their children to private and parochial schools, the Court found a “liberty of parents and guardians to direct the upbringing and education of children under their control. . . . The child is not the mere creature of the State; those who nurture him and direct his destiny have the right, coupled with the high duty, to recognize and prepare him for additional obligations.”

In a third case based on decisions about educating children the Court supported the practice of Amish parents who removed their children from state schools at the end of eighth grade so that the children could finish their training within the Amish community. The Court found that this practice did not cause serious damage to the state’s interest in compulsory education, and that forcing Amish children to continue in state schools might constitute a violation of religious freedom guaranteed by the first amendment. These early cases serve as a constitutional foundation for parental authority within the family—a foundation that is strong enough to withstand the weight of state legislation that attempts to undermine the parental prerogative.

While Supreme Court decisions about education may seem peripheral to issues of genetic testing and the interests of geneticists, the Court has more recently considered the power of parents in making medical decisions for their own children. On several occasions in the early 1980’s, the parents of infants born with severe impairments elected to forego treatment for one problem when the infant’s overall diminished quality of life would remain unchanged. As these cases became familiar to the public, the United States Department of Health and Human Services ("DHHS") promulgated a regulation that permitted discontinuation of federal funds to any hospital that deprived an infant of treatment on the basis of its handicap. The Supreme Court found that DHHS had overstepped its legislative mandate and clearly stated that the difficult decisions about impaired newborns rest with the parents and physicians of these sadly compromised infants. The Court stated that “[s]tate law vests decisional responsibility in the parents, in the first instance.”

Thus, from the early cases about controlling a child's education to this recent case about treating handicapped newborns, the Supreme Court has consistently supported

parents as the persons charged with the awesome, sometimes terrible, duty to provide for and to make decisions for their own minor children.

On the other hand, the Supreme Court has also set limits on what parents may demand from the medical profession. In a case that addressed issues surrounding the commitment of children to state mental hospitals, the Court surveyed the hospital admission procedures and noted that each child received separate psychological and psychiatric evaluations before admission, assuring that parents could not use the hospitals as convenient places to house their obstreperous children. The Court noted:

[In the voluntary commitment setting . . . the parents . . . retain a substantial, if not dominant, role in the decision, absent a finding of neglect or abuse, and . . . the traditional presumption that the parents act in the best interests of their child should apply . . . [H]owever . . . the child's rights and the nature of the commitment decision are such that the parents cannot always have absolute and unreviewable discretion to decide whether to have a child institutionalized. They . . . retain plenary authority to seek such care for their children, subject to a physician's independent medical judgment.]

This case has some appeal to geneticists who refuse to provide genetic testing for children at the request of their parents on the grounds that testing will not provide a clear medical benefit for the child. However, the geneticist who adopts this position must account for the distinction between the commitment of a child to a mental hospital and a genetic test on a child at the request of his parents. Surely the physician's judgment is critical in the former decision, and in many other medical situations as well, but it is considerably more tenuous in the situation of genetic testing, if only because the parents may have well-founded reasons for knowing the genotypes of their own children: parents have interests in providing financial, medical, and environmental benefits for their children, as well as interests in planning their own lives and futures.

Parents in our society are expected to raise their children with care. Their task is one of heavy responsibility, often offset by personal satisfaction. Both society and the law give parents wide latitude in their roles as caretakers and decision-makers for their children. Both society and the law operate from the presumption that parents act in the best interests of their children. Reasonable persons may disagree with the opinions and decisions of some parents, but society continues to support the parental prerogative within the privacy of the family.

V. Rights of Children

Over much of human history children have enjoyed few if any personal rights. This century, however, has witnessed a surge of concern for young people and an expanding interest in their growth and maturation from infancy to adulthood. Children are no longer viewed as a source of labor. To the contrary, our recognition of children as small individuals has emerged side by side with recognition of the rights that vest in children as members of our social order. While children do not enjoy all the rights that society confers on adults, they do enjoy considerable moral and legal protection as persons deserving of respect and care. While professionals in genetic counseling are usually acutely aware of the dynamics in families that seek help with their genetic problems, the interests of children are continually the focus of special consideration.

The most fundamental right that society confers on children is the right to adequate care, including shelter, food, clothing, and health. To be sure, many children in our society do not receive adequate care—a home, a good diet, shoes and coats, or immunizations and antibiotics, but what they do receive is usually provided as a result of parental initiative or parental request, either in the private sector or in public assistance programs.

The idea that children are entitled to adequate care is incorporated into statutes governing child abuse and child neglect. Child abuse usually refers to physical abuse, while neglect refers to failure to provide the elements of adequate care. The statutes specify both criminal and civil penalties for parents who do not provide acceptable care for their children. Criminal prosecutions may include charges of assault and battery, or rape, or in extreme cases, manslaughter or murder, and penalties may include fines, imprisonment, or more. Civil penalties may include removing the child from the custody of the parent and placing the child in a more favorable, fostering environment. Conversely, an additional though unsuccessful source of legal redress has occasionally been sought by children who have been subjected to abuse or neglect: some victims have occasionally tried to sue their own parents for their injuries, but the courts have consistently refused to allow the children to

33. McLaughlin, Survivors and Surrogates at 140 (cited in note 27).
34. Zelizer, Pricing the Priceless Child at 7 (cited in note 26).
37. Parenthetically, the pivotal role of parents in providing adequate care for their children is emphasized by the government program of Aid to Families with Dependent Children. This program provides for children by giving money to the parent, with the expectation that the parent will use the money for the benefit of the children. See 42 USC §601 (1988 & Supp 1992).
pursue actions against their parents. In commenting about the parent-child relationship, the Oregon Supreme Court has noted that “[t]here are certain kinds of relationships which are not proper fodder for tort litigation and we believe this to be one of them.”39 The courts have steadfastly refused to allow children to sue their own parents in tort actions for any perceived injury.40

Additional although indirect legal protection for children is found in state statutes that require professionals to report instances of parental abuse, or suspected parental abuse. Failure to do so can result in criminal prosecutions and/or malpractice charges against physicians and other professionals.41 On the other hand, physicians who do report these problems in good faith are immune from prosecution for malpractice or defamation if they are mistaken in their suspicions. Children receive further legal protection in legislation that requires social agencies to act on behalf of children who are reported to be victims of abuse or neglect.42

In addition to protection from abuse and neglect, children have found additional protection from society in the development of health and immunization programs, public education and recreation programs, and “parenting” programs. On the practical side, these programs are immensely helpful in terms of personal and public health, in teaching children useful information and social and vocational skills, and in teaching new parents skills and methods for fulfilling their roles as parents. On the theoretical side, these programs are a clear acknowledgement of the personhood of children, deserving of nurture and respect.

However, even with the social progress in acknowledging the personhood and individuality of children and in reinforcing their rights, children still enjoy fewer rights than adults. One limitation of children is the fact that they are not autonomous persons. Personal autonomy in the legal sense vests only when a person reaches the statutory age of majority or is otherwise recognized as an adult in the eyes of the law.43 An example of minors who are legally regarded as adults are persons who have been emancipated, although even emancipated adolescents may continue to be restricted in their capacity to contract, for example, or in their capacity to sell or encumber property. So even emancipation may come with restricted autonomy, with full legal autonomy vesting only at the age of majority.

Within a typical family, the parents are the individuals who are autonomous, while the children are not. A family who seeks genetic evaluation will be under the care of the parents, who may exercise their autonomy in caring

40. Zepeda v Zepeda, 41 Ill App 2d 240, 190 NE2d 849 (1963), cert denied, 379 US 945 (1964); Lloyd v Howard, 566 S2d 424 (La 3d Cir 1990).
for and providing for their own children. To suggest that we owe a duty to children to protect their autonomy, or future autonomy, is to say that the not-yet-existent interests of children supersede the real and present interests of parents who are obligated to care for their families and who must plan their own lives. Parents have interests in exercising their own autonomy now, while their children have only a future prospect of becoming autonomous persons with the same rights as their parents. Finally, the exercise of parental autonomy should not be presumed to be only in the selfish interests of the parents: some parents may indeed be selfish, but parents are still the persons who must make decisions on behalf of their families.

The evolving rights of children have also expanded to judicial consideration for the welfare of children in a special standard of making decisions that are in the “best interests of the child.” While there is usually little doubt about where these interests lie, occasionally the task of defining the best interests of children is complex and difficult. In the case of Baby M in the 1980’s, for example, involving a contract for surrogacy, the Supreme Court of New Jersey found the best interests of the child to lie in shared custody between the couple who sought to become legal and social parents and the surrogate mother who had agreed to surrender the child at birth. This decision suspended the child between two vastly different sets of parents, households, and systems of values, and it raised lingering questions about definitions of the best interests of the child. In less complex circumstances, like parents’ requesting genetic testing for their family, the parents may indeed have questionable motives, but because they may also have admirably altruistic motives, the law presumes them to be acting in the best interests of the entire family.

The recognition of children as persons and individuals has also been affirmed by numerous acknowledgments that children should have a role in medical decisions. The courts in many states have been increasingly sympathetic to adolescents who independently seek treatment for sexually transmitted diseases and for pregnancy, and a body of rulings has been recognized as the “mature minor rules” that permit treatment of minors, in some circumstances, in the absence of parental permission. The extent of their participation should depend on their level of understanding and their general maturity, and the treatment must be reasonably simple. The caveat that the geneticist should heed is that the issue of a child’s consent, or an adolescent’s consent, must be weighed against the wishes of the parents and the gravity of the medical, or genetic, issue at hand.


VI. Duties of Geneticists

In the most elementary sense, a geneticist's duty toward the people who seek counseling derives from the physician-patient relationship that is formed when a patient or family seeks help. In the absence of a defined relationship, the geneticist owes no duty at all. But once the patient, or family, asks for help, and the geneticist agrees to contribute his or her expertise, the relationship is joined. The geneticist is then bound to help the patient, or family, understand their genetic situation and to adjust to it the best way they can.

One major concern in deliberations about genetic testing in children is identifying the patient, or patients, in the clinical and counseling setting. For the geneticist who trained in pediatrics, the initial answer may be that the patient is the child and that the duty is owed to the child. For the internist, on the other hand, the initial answer may be that the parents are the patients and that the duty is owed to them. An early definition of genetic counseling stresses that one goal of genetic counseling is to help patients to "choose the course of action which seems appropriate to them in view of their risk and their family goals and [to] act in accordance with that decision." Thus genetic counseling should help the family make the best possible adjustment to its genetic situation in view of its values and circumstances, and the counseling scenario often becomes more complex than the one-on-one interaction in many other physician-patient interactions. To deal with these more complex interactions, some observers suggest that "[t]raditional views about doctor-patient relationships may have to change. In genetics, the patient is really the family rather than the individual." The geneticist may thus have a duty to several persons, including both the parents and their children.

A second major concern in genetic testing in children is defining the extent of the duty of the genetics professional. If the goal of genetic counseling is to provide comprehensive help for the family, the geneticist must carefully examine the process of genetic counseling and the content of information that is conveyed. Geneticists who developed the protocol for counseling in presymptomatic testing for Huntington disease have constructed an exhaustive process, including several counseling sessions, both before and after genetic testing. This process serves to inform patients and to assess their understanding and reactions, and there is much to learn from the Huntington prototype. To provide counseling before testing is an excellent means of informing patients, or parents, about the many implications of the information they seek, including the fact that genetic information can have both positive

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46. See, supra, notes 6-7 and accompanying text.
47. American Society of Human Genetics, Genetic Counseling at 240 (cited in note 4).
and negative ramifications. Parents who seek to have their families tested can be helped to understand that genetic information about their children may—but not necessarily must—have a negative impact on each child individually and on family relationships in general. Parents should understand that some children want to know and indeed need to know about their own situation, and that some children feel a great sense of relief when they have hard information, one way or the other.\(^5\) Parents should also understand that some children do not want to know or need to know, and that some children may never want to know, even after they start making their own decisions. Finally, geneticists should respect the difficult decisions of parents, who may or may not reveal test results to their children, depending on what the parents think is best for their children and their family. The geneticist is obliged to present this information in a thorough manner and in understandable language, and to explore all options with the family. For the geneticist to fulfill his duty in the fiduciary relationship with the patient or family, both the process and the content of genetic counseling must be thoughtful and thorough. However, after the parents, and perhaps the children, are informed, what they decide in terms of pursuing testing or sharing information should be their own decisions.\(^5\)

Aside from considering the process and content of genetic counseling and genetic testing, geneticists should also be aware of the possible legal consequences associated with the birth of children who have predictable inherited disorders. After the decision of the United States Supreme Court extending the right to privacy to include a woman’s right to terminate a pregnancy\(^5\) and with the expansion of technologies in prenatal diagnosis,\(^5\) the options for ensuring the birth of healthy children increased dramatically. Parents have become increasingly aware of the possibility of having children with genetic disorders or other problems that can be predicted before conception or during pregnancy, and they have become increasingly insistent on their right to bear healthy children, within the limits of contemporary technology. Litigation in medical malpractice for wrongful birth has increasingly supported the claims of parents who assert injury resulting from the birth of children with predictable diseases or defects, and claims for wrongful life, brought by children born with predictable problems, have also been honored in a few cases.\(^5\) The

\(^{50}\) Indeed, one geneticist who has generally opposed genetic testing in children has observed that “children in families with Huntington disease often recognize much earlier [than the ages of 14 and 16 years] that something is wrong and thus might benefit from earlier information.” Michael Hayden, *Book Review*, 48 Am J Hum Genetics 171, 172 (1991) (reviewing Susan E. Folstein, *Huntington’s Disease: A Disorders of Families* (Johns Hopkins 1989)).


\(^{52}\) *Roe v Wade*, 410 US 113 (1973).


\(^{54}\) Mary Z. Pelias and Margery W. Shaw, *Medicolegal Aspects of Prenatal Diagnosis*,
geneticist who counsels cautiously and thoroughly may well avoid the inconvenience associated with allegations of wrongful birth and wrongful life as well as the damage associated with adverse judgments of medical negligence.

The duties of geneticists have thus become more rigorous as technological capacities in medical genetics have expanded. The scope of genetic counseling has come to include concern for all members of families that are coping with difficult genetic problems, and the geneticist has become increasingly aware of the psychosocial implications of genetic information. While few geneticists have formal training in psychology or sociology, they have developed an increasing sensitivity to the effects of genetic information in the lives of patients and families who seek information and help. Most geneticists have practiced and still practice from a view of non-directive counseling that subordinates personal value judgments of the professional to the judgments that patients make in view of their own circumstances.

VII. Conclusions

Potential applications of new knowledge and technologies in medical genetics have fostered a new examination of how professional geneticists should now practice genetic counseling and how the geneticist should interact with persons and families who seek help with their genetic problems. Newly developed powers to elucidate individual genotypes underscore the need for respecting the prerogative of parents to seek genetic information about their families, but only with caution and concern for the welfare of children whose lives may be significantly altered by knowledge of their most personal information. Striking a balance between parental concerns and interests, on the one hand, and the welfare of children, on the other, has expanded the role of the geneticist as counselor to individuals and to families. As geneticists continue to explore the implications of new knowledge and technologies, they will best serve patients and families when they avoid adopting across-the-board rules and policies and continue to treat each counseling situation as the unique set of personal circumstances that it is. In the difficult world of genetic testing in children, the geneticist will continue to convey difficult information to individuals and to families within their ability to comprehend, and will help these persons make the best possible adjustment to their situation. The geneticist who continues to be forthright and thorough will continue to help both parents and children as they adjust to their own, personal genetic situations.


APPENDIX: CASE SCENARIOS

1. HUNTINGTON DISEASE

The Disease: Huntington disease (HD) is an autosomal dominant neurodegenerative disorder characterized by premature neuronal cell death. Initial symptoms include clumsiness and forgetfulness. Personality changes are common and may be associated with depression or symptomatic schizophrenia. The average age of onset is 38 years, although symptoms may appear at any time between the first and seventh decade of life. The disease may have an earlier onset when the gene is inherited from the father than when the gene is inherited from the mother. Symptoms and debilitation progress to death, usually within 20 years of onset. Presymptomatic testing is available for determining individual genetic status. Because of concern for suicide and other adverse psychosocial reactions, individuals who seek presymptomatic testing are required to participate in several counseling sessions, both before and after testing, in order to assess the patient's mental and emotional status and to evaluate the patient's support system. There is no treatment for this disease.56

The Family: The Wilson family has a 20-year relationship with the genetic counseling clinic because of multiple family members affected with HD. Several individuals in the family, mostly minors, want more information about testing. Of the 15 family members who attend a counseling meeting, some are symptomatic, some are at risk, and the remainder are caregivers. Several of the seven adolescents readily indicate that they would commit suicide if they have the gene. Jimmy and Timmy, 14-year-old identical twins, appear to have early motor symptoms of the disease, and Jimmy has been referred for neurological evaluation because of behavior problems in school, including fighting and poor impulse control. Patrice, the 15-year-old sister of the twins, offers several sound reasons for her request for testing; she does not express suicidal intentions, appears otherwise mature in her reasoning, and shows no signs of having the disease. Lotty, an 18-year-old second cousin, with a tattoo on her left arm, a pierced nose, and partially shaved head, has a history of poorly controlled diabetes and has just learned that she is pregnant; she wants to be tested for the HD gene. She has wrecked 3 cars while showing off to friends and is in an abusive relationship with a 32-year-old male crack addict. She wants to end the relationship but is afraid for her life, although she admits that she has considered suicide during the past 3 months.

The Counseling Issues: Although this family has a clear general understanding of the consequences of having the gene for HD, each person who seeks testing, or whose parents request testing, should be evaluated individually. Determining the genotype of Jimmy will determine the genotype of his twin and may help to explain their symptoms and confirm a suspected diagnosis. While such information could aid in dealing with Jimmy’s behavior problems, it could

56. See, supra, notes 2-3 and accompanying text.
also push both twins toward suicide. Although Patrice is younger than the age of 18 recommended for testing, she appears capable of dealing with the outcome of testing and will probably be tested if her counseling sessions confirm her stability. While Lotty is of adult age, she is emotionally troubled and should be encouraged to resolve her immediate problems before pursuing testing for the HD gene.

2. TAY-SACHS DISEASE

The Disease: Tay-Sachs disease (TSD) is a progressive neurodegenerative disorder that is inherited in an autosomal recessive manner. For an individual to be affected, he or she must receive a copy of the abnormal gene from each parent. Individuals who have only one copy of the abnormal recessive gene are carriers and are unaffected by the disease. If both parents are carriers, they have a 25% chance of having an affected child, a 50% chance of having a carrier child, and a 25% chance of having a child who has no gene for TSD. Children born with two copies of the abnormal gene develop normally for about 6 months and then begin to show developmental regression, including loss of motor milestones and progressive paralysis, dementia, and blindness. Death usually occurs in the second or third year of life. The gene that causes TSD is found most commonly in persons of Ashkenazi Jewish descent; carrier frequency in this population is estimated at 1 in every 30 persons. Carrier testing and prenatal diagnosis are available and are considered reliable. There is no treatment for this disease.

The Family: Paula Hecht is a 53-year-old Ashkenazi Jewish woman who has requested carrier testing for her 13-year-old daughter, Sharon. Mrs. Hecht's family history reveals that her first child, born during her first marriage, died of TDS in 1960. Her second husband, Mr. Hecht, is not a carrier of the gene for TSD, and they have two sons and a daughter, Sharon. It becomes clear that Mrs. Hecht had a very difficult time dealing with the death of her first child and the ensuing dissolution of her first marriage, caused in part when her in-laws blamed her for the death of their first grandchild. Her in-laws correctly insisted that the child would have been healthy if Mrs. Hecht had not been a carrier, and they also asserted that nothing like this had ever happened in their family. Mrs. Hecht clearly feels ashamed of being a carrier and wants to spare her daughter the heartache of losing a child to TSD. Mrs. Hecht suggests that if Sharon is a carrier, she should avoid dating young men who have lost relatives to TSD. Sharon is clearly aggravated with her mother and is not interested in carrier testing or counseling. She appears to be an intelligent though rather immature individual.

The Counseling Issues: Testing a minor is only one issue in this scenario. Mrs. Hecht should first recognize that she hopes to alleviate her own guilt by having her daughter tested, that no “fault” attaches to her for the death of her

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57. Thoene, Rare Diseases at 242 (cited in note 2).
first child, and that both parents contributed equally to the first child's genotype. Resolving Mrs. Hecht's turmoil might be approached with information that every person carries several deleterious recessive genes, and that most persons discover their carrier status only after the birth of a child with a genetic disease. Sharon's avoidance of dating boys who have lost relatives to TSD will not eliminate the possibility of dating carriers because most carriers have no affected relatives. Further, Sharon's subsequent choices about having children may render any question of carrier status moot. Alternatively, she could later elect carrier testing or prenatal diagnosis, as she matures and makes her own choices.

3. SICKLE CELL ANEMIA

_The Disease:_ Sickle cell anemia (SCA) is an autosomal recessive disorder of the beta chain of hemoglobin, the oxygen-carrying protein in red blood cells. When blood oxygen level is dramatically decreased, as it is during strenuous physical exercise, hemoglobin molecules polymerize and distort the smooth, circular shape of red blood cells to pointed "sickle" shapes. The irregular shape then causes clogging of small blood vessels with oxygen deprivation and pain in surrounding tissues. Sickled red cells are eventually destroyed by the spleen, which becomes enlarged because it is overworked, and decreased red cell count results in anemia. The most common causes of hospitalization in children with SCA are bacterial infections and vaso-occlusive crises, and infection is the most common cause of death. Penicillin prophylaxis is the recommended treatment for avoiding infections. Age of onset varies, even within families, and older affected siblings may not be diagnosed until the disease is diagnosed in younger brothers or sisters. There is presently no cure for SCA, although treatment with hydroxyurea appears to decrease the frequency of crises.58

_The Family:_ Maryland is one of 2 states that require parental consent for newborn screening. Mr. and Mrs. Walker live in Baltimore and have just had a baby son, Justin, whom they refuse to have tested for SCA. Their older son, Stephen, was diagnosed with SCA two years ago at the age of 5, and his disease has been mild. The Walkers offer the following reasons for refusing to have Justin tested: (1) Stephen's late diagnosis and mild disease indicates that Justin, if affected, will also have a mild course; (2) Stephen's affected status reduces the chance that Justin will be affected; (3) the heelstick to obtain a blood sample could cause Justin extra discomfort.

_The Counseling Issues:_ The family's understanding of the genetics and medical basis of SCA needs clarification, and they should be urged to consent to testing. Extreme variability in expression of the disease implies that Justin could well have a severe course of the disease. Affected children benefit from very early penicillin prophylaxis even though they may not have problems during the first 6 months of life. Stephen's affected status in no way modifies the 25% chance that Justin is affected (or the 75% chance that he is unaffected): chance has no

58. See, generally, Thoene, _Rare Diseases_ (cited in note 2).
memory, and each conception is a "new roll of the dice." Hospitalization for bacterial infections and vaso-occlusive crises, which may be life-threatening, is far more uncomfortable than a heelstick for a sample of blood. Knowledge of Justin's medical status will have a significant effect on the course of his medical care and his ultimate well-being.

4. FAMILIAL ADENOMATOUS POLYPOSIS

The Disease: Familial adenomatous polyposis of the colon (FAP) is an autosomal dominant disease. Individuals who have one copy of this gene are affected with the disease, and each child of an affected person has a 50% chance of inheriting the disease gene from the affected parent. FAP is characterized by the appearance of adenomatous polyps in the large intestine and the transformation of polyps to malignant growths. The age of onset of malignant transformation varies, even within families, from childhood through the seventh decade, with an mean age of diagnosis at 40 years. Annual colonoscopy screening permits early detection and surgical removal of intestinal polyps and tumors, although tumors may develop at sites other than the colon. Testing family members can determine who carries the gene long before polyps appear.59

The Family: John Henry is a 12-year-old boy hospitalized for pneumonia. The medical student assigned to assess John has just finished a rotation in medical genetics and is conscientious about taking a thorough family history from John's father, Mr. Henry. John's parents are divorced, and John's mother, now Mrs. Duggan, age 30, has sole custody of her son and is not present in the hospital. Mr. Henry relates that his ex-wife's father, brother, paternal grandmother, and paternal uncle all died of colon cancer, though a specific diagnosis is unknown. When Mr. Henry later asks Mrs. Duggan about the disease, she refuses to discuss it. The attending physician requests a genetics consult, and the geneticist arrives while Mr. Henry and Mrs. Duggan are arguing. Mrs. Duggan does admit that many persons in her family have FAP, although she has no symptoms and is not screened regularly. Mrs. Duggan also relates that her sister lost her health insurance when she was found to have polyps— an event that Mrs. Duggan wants to avoid for herself and her son. And Mrs. Duggan states that she has not discussed FAP with her son because she does not want to cause him worry, although she has nightmares about the possibility that she and her son are affected. Mrs. Duggan also emphasizes her own continuing good health as a sign that her son could not have any problems until much later in life. Finally, Mrs. Duggan relates that a cousin died of bladder cancer, unrelated to FAP.

The Counseling Issues: Mrs. Duggan needs counseling to help her understand clearly the genetics and medical nature of FAP: her own risk of developing FAP is presently 50%, and her son's is 25%. If she has the gene for FAP, her son's risk is increased to 50%. The extremely variable age of onset of this disease

59. See, supra, notes 15-18 and accompanying text.
means that both she and her son might presently have polyps or malignant tumors. Her cousin’s bladder cancer may have been related to the gene for FAP, and both she and her son may have other tumors as well. Mrs. Duggan’s belief that she is sparing her son unnecessary worry is probably an illusion. John may well elect to be screened if given a choice, and Mr. Henry should be a part of any deliberations and decisions about testing. Mrs. Duggan’s concerns about insurance discrimination are valid and bode for postponing genetic testing, although both she and John should be screened for polyps on a regular basis.