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"Fitness" for Birth and Reproduction: Legal Implications of Genetic Screening*

GEORGE J. ANNAS†
BRIAN COYNE**

The introduction of accurate methods to screen for genetic defects in the adult, the newborn, and the fetus promises to increase man's control over his own destiny. If that promise is to be realized, however, careful planning will be needed to prevent the technology of screening from imposing its own ethic on man. The invention of the club enabled man to increase his ability to hunt for food, and simultaneously to brutalize his fellow man. In the same way, while advances in genetic screening could lead to an increase in self-autonomy for a few, they may also encourage the sterilization of those determined to be genetically "unfit" to reproduce and the abortion of those judged "unfit" to be born.

While one hopes that the new techniques will be responsibly used to prevent the genetic defects which now appear in over 200,000 infants annually in the United States, individual human rights of privacy, confidentiality, self-determination and procreation can probably be protected only by the legal process. The law is not likely to be effective, however, unless the lawyers who advise clients, the judges deciding cases, and the legislatures that pass laws understand the basic technology of genetic screening and its potential implications. Since the legal issues cannot be fully appreciated

†J.D., Harvard Law School; M.P.H., Harvard School of Public Health. Director, Center for Law and Health Sciences, Boston University School of Law, Boston, Massachusetts.

**Ph.D., University of Chicago. Milbank Memorial Fund Fellow in Health Services Research and Policy, Boston University School of Law, Boston, Massachusetts.

without some knowledge of the technology of screening, our discussion begins with a brief outline of this subject.

The Technology of Genetic Screening

The physical basis of human heredity is the genes that are passed on from parents to children.[1] Each gene encodes information specifying a single gene product. Each gene product, either directly or in combination with other gene products, determines a specific inherited trait such as blood group, eye color or Rh factor. Human genes are physically arranged in linear sequences along the chromosomes found in the nucleus of each human cell. The germ cells, ova in the female and sperm in the male, each contain a single set of chromosomes consisting of 22 autosomal[2] chromosomes, plus one X chromosome in ova and 22 autosomal chromosomes, plus one X or one Y chromosome in sperm.

Following fertilization of an ovum by a sperm, a human embryo is formed whose genetic complement consists of all 44 autosomal

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1. *See generally* any textbook on human genetics, *e.g.*, M. LEVITAN & A. MONTAGU, TEXTBOOK ON HUMAN GENETICS (1971). Both DNA (deoxyribonucleic acid) and RNA (ribonucleic acid) are linear sequences of nucleotides. A gene is defined operationally as a specific sequence of nucleotides (in DNA or RNA) which encodes the genetic information that specifies a single gene product. Acting as a template, the specific nucleotide sequence of the DNA is transcribed into an equivalent nucleotide sequence in RNA. There are four different nucleotides in RNA (adenine, guanine, cytosine, and uracil) and each sequence of three nucleotides is a code word specifying one of twenty different amino acids or, alternatively, encodes a punctuation mark in the genetic message such as "end here." The number of possible genetic code words is just the number of permutations of the four different nucleotides taken three at a time, *i.e.*, $4^3 = 64$. The genetic code is thus redundant, since the number of code words greatly exceeds the number necessary to code for all the amino acids plus punctuations. For instance, both guanine-adenine-guanine and guanine-adenine-adenine are code words for the amino acid glutamic acid. Translation of the genetic information in RNA consists in lining up in sequence the amino acids specified by each nucleotide triplet in the RNA (*i.e.*, the RNA stand serves as a template) and joining the amino acids together to form a polypeptide chain. One or more polypetide chains may later join together to become a protein molecule. For example, the protein globin in hemoglobin forms when two alpha and two beta polypeptide chains fold around each other to create a unique three-dimensional structure. The genetic disease sickle cell anemia results from a defect in the DNA gene that codes for the beta polypeptide chain. The defect consists in substitution of a single nucleotide (thymine for adenine) in the DNA gene, which upon translation of the genetic information into the beta polypeptide of globin results in the amino acid valine being substituted for glutamic acid in the amino acid residue number six position in the beta polypeptide chain. Although each beta chain consists of 116 amino acids and each alpha chain is a sequence of 121 amino acids, this single amino acid substitution has serious consequences for an individual unable to produce normal globin: his red blood cells assume a sickle shape and their ability to carry oxygen to his body tissues is substantially imparied. For further details concerning the molecular basis of heredity, see any modern text on biochemistry, *e.g.*, H.R. MAHLER & E.H. GORDES, BIOLOGICAL CHEMISTRY (1966)

2. The sex chromosomes in a normal individual are two in number, denoted XX in the female and XY in the male. All of the other 44 chromosomes are termed "autosomal chromosomes." An autosome is simply any chromosome other than a sex chromosome.

chromosomes (22 from the mother and 22 from the father) plus the X chromosome donated from the mother's ovum, plus either an X or Y chromosome from the father's sperm, whichever happened to have been present in the sperm. If the embryo receives an X chromosome from the father's sperm, the embryo will have two X chromosomes (XX), and will be female; if the sperm is carrying a Y chromosome the embryo will be male (XY). Since the X and Y chromosomes specify sex, they are termed the sex chromosomes.

With the exception of the germ cells, all other human cells (the somatic cells) contain 46 chromosomes consisting of 22 pairs of homologous chromosomes plus the two sex chromosomes. As the embryo develops, its cell population multiplies by cell division. At each cell division an identical complement of chromosomes is distributed to each daughter cell. Each chromosome contains upwards from one thousand genes, so that the full complement of human genes is on the order of tens of thousands. Not all genes are actively forming their gene products in all cells all of the time. Instead, various genes are selectively activated and repressed in different cells to create different cell types according to a carefully controlled program of embryological development. A wide variety of cell types, produced and maintained by selective gene repression and activation, forms the patterned arrays of cells that make up the tissues and organs of the human body.

A gene mutation is a physical change in a gene that causes the gene to specify an altered gene product or to fail to function at all. Mutations may result from an error in copying the gene preparatory to cell division, from radiation (ultraviolet light, X or gamma radiation), or from chemical mutagens. Most mutations are harmful and many are lethal. A mutation may involve deletion or substitution of genetic material at a single point (*point mutation*) or of a whole segment of a gene. On a grosser level, breaks and rearrangements of one or more chromosomes may occur leading to *chromosomal abnormalities.*

Dominant and Recessive Inheritance

The inheritance of a mutant gene may or may not lead to genetic disease depending upon the nature of the gene product the mutant gene specifies and whether the individual inherited one normal copy of the gene and one mutant copy, termed *heterozygous*, or inherited

two mutant copies (one on each of the two homologous chromosomes carrying the gene), termed *homozygous*. A mutant gene may be located on an autosome or on either of the sex chromosomes X or Y. Genes on the X chromosome are termed X-linked (sex-linked) and those on the Y chromosome are termed Y-linked. If a mutant gene is expressed in an individual who is heterozygous for that gene, the mutant genetic trait (the *phenotype*)[3] is a *dominant* trait. If the mutant gene is expressed only in the homozygous state, the genetic trait is termed *recessive*. Exceptions to the latter, however, are the X-linked recessive disorders in which only a single recessive gene in the male will give rise to a given trait or disease (*e.g.*, hemophilia).[4]

Autosomal Dominant Inheritance

Autosomal dominant genetic diseases include, among others, Huntington's chorea,[5] neurofibromatosis,[6] and Ehler-Danlos syndrome.[7] An individual with an autosomal dominant genetic disease is almost certainly heterozygous for the defective gene since a double dose of the defective gene is usually lethal. One or both of his parents must also have the disorder unless there has been a new mutation or illegitimacy. An affected parent will transmit the disease to approximately one half of his children without regard to the children's sex. An unaffected child can never transmit the disease.

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3. The genetic phenotype is the collection of observable properties (structural and functional) produced by the interaction between the organism's genetic potential (its genotype) and the environment in which it finds itself. Not all genes are expressed. Some gene products are totally non-functional and produce no observable characteristics under any circumstances. Other genes, however, may produce observable characteristics under favorable environmental influences. Alternative forms of the same gene at a particular gene locus on a chromosome are termed alleles. An individual's genotype might consist of a gene on a particular chromosome and its allele b on the homologous chromosome. If gene B is expressed and gene b is not, then B is the dominant allele and b is the recessive allele. For example, if B were the mutant gene that gives rise to Huntington's chorea, and if b is the normal allele, then an individual with genotype Bb will express phenotypically Huntington's chorea.

4. Hemophilia is due to a hereditary deficiency of blood coagulation Factor VIII and is characterized by spontaneous or traumatic hemorrhages, bleeding from the mouth, gums, lips, and tongue, and appearance of blood in the urine. In hemophilia A, there is a deficiency of antihemophilic factor A.

5. Huntington's chorea is a disorder of the central nervous system which usually develops in adult life into progressive chorea (involuntary and irregular, often twisting and writhing, movements of the extremities and the face), disturbed speech and dementia.

6. Neurofibromatosis is characterized by the presence of neurofibromas (*i.e.*, tumors that involve diffuse proliferation of peripheral nerve elements) in the skin or along the course of peripheral nerves.

7. Ehler-Danlos syndrome is a connective tissue disorder whose clinical manifestations include excessively soft joints that are subject to frequent dislocation, loose and fragile skin, easy bruising and calcified cysts.

Autosomal Recessive Inheritance

Autosomal recessive genetic diseases appear only in individuals who are homozygous for the defective gene, *i.e.*, a double dose of the defective gene is required for the disease to be expressed. Either both parents must be heterozygous (showing no obvious manifestation of the disease) or one parent is heterozygous and one parent is homozygous in whom the disease is also expressed. The first case will obviously occur much more often than the second. Although the heterozygous parents are usually clinically normal, some partial loss of enzymatic function may be detected in persons heterozygous for some metabolic disorders, and this fact is now being exploited in prenatal genetic screening for heterozygous parents. Persons heterozygous for recessive genetic disease are termed *carriers* and major research efforts are underway to develop accurate and efficient methods for detection of carriers. The list of hereditary disorders for which methods of carrier detection are available is continually growing and now includes, for example, hemophilia A,[8] muscular dystrophy (Duchenne),[9] phenylketonuria,[10] sickle cell anemia[11] and Tay-Sachs disease.[12]

Each parent who is heterozygous for a recessive mutant gene faces a 50 percent risk of transmitting that gene to his children. Thus, if a man and his wife are each heterozygous they face a 25 percent risk (½ x ½)that their children will be affected (homozygous), and a 50 percent risk that their children will be carriers like themselves (heterozygous). There is a 25 percent probability that any child of theirs would be entirely free from the defective gene.

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8. *See* note 4, *supra.*

9. Muscular dystrophy (Duchenne), also known as pseudohypertrophic infantile muscular dystrophy, is the most common form of the muscular dystrophies. This sex-linked recessive disorder begins in early childhood, affecting chiefly males, and is characterized by progressive weakness and atrophy of the thigh, hip and back muscles, with resulting waddling gait and forward curvature of the spine. Eventually the muscles of respiration, the heart and the esophagus also atrophy.

10. Phenylketonuria (PKU) is a hereditary metabolic disorder in which there is a deficiency of the enzyme phenylalanine hydroxylase, resulting in increased amounts of the amino acid phenylalanine in the blood and excess phenylpyruvic acid and other acids in urine. Clinical manifestations are mental retardation, eczema, and occasionally seizures.

11. *See* note 1, *supra.*

12. Tay-Sach's disease is a metabolic disorder which results in infantile idiocy, blindness, and early death. For a complete list of genetic disorders for which methods are available for detection of carriers, *see* A. MILUNSKY, THE PRENATAL DIAGNOSIS OF HEREDITARY DISORDERS (1973).

X-linked Inheritance

An X-linked genetic disease is either dominant or recessive, but in either case if a male inherits from his mother an X chromosome containing a defective gene, that gene will be expressed and he will be affected by the disease. This is because, unlike the female who has two X chromosomes, the male has only the one X chromosome, so that a second copy of the gene is not available in the normal state to produce a properly functioning gene product. The male's Y chromosome contains quite a different set of genes (those necessary for the development of the male sexual organs) from the X chromosome, and the Y chromosome cannot make up for the defective gene on the X chromosome. If the disease is recessive, women who are carriers (heterozygous, X-linked) transmit the mutant gene to half of their daughters, who will also be carriers, and to half of their sons, who will be affected. Males affected with an X-linked, recessive disease will transmit the mutant gene to all their daughters, who will be carriers, and to none of their sons.

In the case of X-linked, dominant disorders, all the daughters of an affected father will have the disorder, since the father transmits his mutant X chromosome solely to his daughters, but none of his sons will be affected unless the mother is also affected. The X-linked diseases include, for example, glucose-6-phosphate dehydrogenase deficiency,[13] various forms of muscular dystrophy, hemophilia,[14] retinitis pigmentosa[15] and Lesch-Nyhan syndrome.[16]

Polygenic Inheritance

Many genetic diseases and traits are controlled by multiple genes, and environmental factors may also be important determinants of the ultimate clinical picture. Among the diseases presently

13. Glucose-6-phosphate dehyrogenase deficiency is an inborn error of metabolism affecting the conversion of glucose-6-phosphate to 6-phosphogluconic acid, resulting clinically in hemolytic anemia.

14. *See* note 4, *supra.*

15. Retinitis pigmentosa involves the slow, progressive atrophy of the nerve elements of all lawyers of the retina, clumping of pigment and attenuation of the retinal arterioles. Onset may occur in childhood or later on, resulting in defective night vision, a constricted field of vision and eventual blindness.

16. Lesch-Nyhan syndrome is a disease due to deficiency of the enzyme hypoxanthine-guanine phosphoribosyltransferase. Clinical manifestations are mental retardation, spastic cerebral palsy, choreathetosis (a condition characterized by both choreiform and athetoid movements including involuntary, irregular, and often twisting and writhing movements of the extremities and face) and self-mutilating biting. A list of X-linked genetic disorders appears in R. GOODMAN, GENETIC DISORDERS OF MAN, 95 (1970).

regarded by man as polygenic in origin are atherosclerosis, essential hypertension and diabetes mellitus. Persons suffering from these disorders are probably often unaware of any hereditary basis of their disease and fail to obtain genetic counseling.

Chromosomal Abnormalities

Chromosomal abnormalities include cases of broken or rearranged chromosomes, extra or missing chromosomes or parts of chromosomes. A chromosomal abnormality may occur uniformly throughout the cells of an individual, or the individual may be a *mosaic* in whom the chromosome number or structure or both differ in different cells. The most common inherited chromosomal abnormality in man is Down's syndrome[17] (also termed mongolism) in which three copies of chromosome 21 are present instead of the normal two (hence the term trisomy 21). Among the more common chromosome abnormalities are the sex chromosome disorders XXX females, XYY males, Turner's syndrome[18] (XO, *i.e.*, the Y chromosome is missing), and Klinefelter's syndrome[19] (XXY). The incidence of Down's syndrome and of the sex chromosome disorders is heavily dependent upon the age of the mother. For example, trisomy 21 occurs in about 1:2300 births for mothers under the age of 20 years, 1:290 from 35 to 39 years, and 1:100 from 40 to 44 years, and increases to about 1:46 at 45 years of age and over.[20]

Detection of Genetic Disease in the Fetus

Once it has been determined that a husband or wife or both carry a defective gene and they have been counseled concerning the possibilities of their future offspring also becoming carriers or being affected with genetic disease, the couple must decide for themselves whether to risk having children. Until recently, unless

17. Down's syndrome is a constellation of congenital defects including mental retardation (whose severity varies among affected individuals) and atypical facial characteristics responsible for the older descriptive term mongoloid idiocy.

18. Turner's sydrome is characterized by short stature, undifferentiated (streak) gonads and variable abnormalities that may include webbing of the neck and cardiac defects. The phenotype is female.

19. Klinefelter's syndrome is characterized by the presence of small, abnormal testes and by an increase in urinary gonadotropins.

20. A. Milunsky, et al., *Prenatal Genetic Diagnosis*, 283 N. ENG. J. MED. 1370, 1371 (1970).

the genetic disease was dominant, the parents would not ordinarily know whether their child had the disease until after its birth. However, recent developments in fetoscopy,[21] radiography,[22] ultrasound,[23] and, most notably, in amniocentesis now allow detection of a number of genetic diseases and of some serious anatomical abnormalities in the fetus *in utero* prior to 20 weeks' gestation. This means that even if carriers decide to have children in spite of the risks, they may still choose to abort the fetus should antenatal diagnosis reveal genetic defects.

The fetus is enveloped by a delicate membrane called the amnion which is filled with amniotic fluid. Amniotic fluid is a complex mixture similar to extracellular fluid and includes the fetal urinary excretion of enzymes, amino acids and any abnormal metabolites. Chemical analysis of the amniotic fluid can, therefore, reveal certain genetic diseases in the fetus.[24] Of key significance is the fact that amniotic fluid also contains *cells of fetal origin,* derived mainly from fetal skin and amnion.[25] Once the fetal cells have been obtained by amniocentesis, biochemical and cytogenetic[26] studies upon the cells directly or upon tissue cultures seeded with the fetal cells may be performed to screen for biochemical defects and chromosomal abnormalities. *Amniocentesis* consists of inserting a needle through the mother's abdomen[27] and drawing out a sample

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21. Fetoscopy utilizes fiber optics to allow visualization of the fetus and may prove useful in the antenatal diagnosis of congenital malformations. The risks of fetoscopy are such that it should probably be reserved for high risk pregnant women who have already given birth to children with serious congenital malformations such as spina bifida (incomplete closure of the vertebral canal) and anencaphaly (absence of cerebrum and cerebellum and of the flat bones of the skull). Emery, *Antenatal Diagnosis: Limitations and Future Prospects,* in MEDICAL GENETICS TODAY 294 (Birth Defects Original Article Series, no. 10, D. Bergsma ed. 1974).

22. The fetus may be studied directly by radiography to detect skeletal abnormalities, and by amniography and fetography to detect soft tissue abnormalities. These methods are of proven value only in later pregnancy. Emery, *supra* note 21, at 290.

23. Diagnostic ultrasound is useful in locating the fetus and placenta, and in detecting gross structural abnormalities in the fetus. The procedure is apparently quite safe and often used in conjunction with amniocentesis to locate the placenta and to determine the presence of twins. Campbell, *The Prediction of Fetal Maturity by Ultrasonic Measurement of the Biparietal Diameter,* 76 J. OBSTET. GYNAEC. BRIT. COMM. 603 (1969); Hellman, et al., *Safety of Diagnostic Ultrasound in Obstetrics,* 1 LANCET 1133 (1970); Milunsky, et al., *supra* note 20.

24. Milunsky, et al., *supra* note 20 at 1370. Matalon and Dorfman, for example, have diagnosed Hurler's syndrome (lipochondrodystrophy) *in utero* by chemical analysis of amniotic fluid. *See generally* Milunksy, et al., *supra* note 20, at 1444-45.

25. *Id.*

26. Cytogenetics is a hybrid science in which the methods of cytology are employed in the study of chromosomes. The principal tool is the microscope which allows direct examination of the chromosomes and may reveal any gross structural abnormalities in the chromosomes, as well as extra or missing chromosomes.

27. The needle may be inserted transvaginally but this appears to increase the risk of abor-

of amniotic fluid. Amniocentesis cannot properly be performed earlier than 14-16 weeks gestation for lack of sufficient amniotic fluid, nor past the twentieth week.[28] The risks of amniocentesis include Rh sensitization,[29] hemorrhage, abortion, puncture and induced abnormalities of the fetus, and abdominal pain and peritonitis.[30] Total risks to both the pregnant woman and the fetus will probably amount to less than one percent.[31]

Where it is necessary to have a fairly large quantity of fetal cells for biochemical or cytogenetic analysis, the cells are cultured for up to eight weeks. Wherever possible, the use of uncultured cells will avoid such a long delay. Uncultured fetal cells are used mainly for sex prediction based on sex-chromatin and fluorescence studies.[32] In satisfactory preparations the accuracy of sex prediction can exceed 90 percent and serve, for example, as the basis for the selective abortion of female fetuses of males affected with an X-linked genetic disease.[33]

Detection of chromosomal abnormalities in fetal cells has become an established and accurate procedure. Both fetal sex and karyotype can be readily determined.[34] Chromosome studies on cultured amniotic fluid cells will ordinarily be undertaken where one of the parents is a mosaic,[35] or carries a translocation[36] which causes a severe physical or mental abnormality. More commonly, however, chromosome studies are undertaken because the mother is 35 or older, since the fetus carried by such a mother faces increased risk of trisomy 21 (Down's syndrome) and the sex chromosome abnormalities.[37]

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tion and microbial contamination of the sample. Milunsky, et al., *supra* note 20; Turnbull, et al., *Antenatal Diagnosis of Fetal Abnormality with Special Reference to Amniocentesis,* 66 PROC. ROY. SOC. MED. 1115 (1973).

28. Turnbull, et al., *supra* note 27.

29. Queenan, et al., *Role of Induced Abortion in Rhesus Immunization,* 1 LANCET 815 (1971).

30. Creasman, Laurence & Thiede, *Fetal Complications of Amniocentesis,* 204 J.A.M.A. 949 (1949); Nadler, *Prenatal Detection of Genetic Defects,* 74 J. PED. 132 (1969).

31. Turnbull, et al., *supra* note 29, at 1115.

32. Emery, *supra* note 23, at 291.

33. *Id.*

34. Hsu, et al., *Results and Pitfalls in Prenatal Cytogenetic Diagnosis,* 10 J. MED. GEN. 112 (1973); Milunsky, et al., *supra* note 20, at 1376.

35. A mosaic is an individual composed of cells of different genotypes. Mosaics are created spontaneously by gene or chromosomal mutation in somatic cells.

36. A translocation is the transfer of a broken portion of a chromosome to a different part of a homologous chromosome or to a nonhomologous chromosome. The result is that the translocated chromosome portion is no longer where it belongs.

37. Emery, *supra* note 21, at 291. Methods to detect chromosomal abnormalities have

Another important development in recent years has been the development of methods to detect the presence of inherited metabolic disorders in the fetus, using either uncultured or cultured fetal cells. The list of metabolic disorders now diagnosable *in utero,* while growing, is still quite limited.[38] Important inherited metabolic disorders not yet prenatally diagnosable include cystic fibrosis[39] (the most common hereditary metabolic disorder in western society)[40] and phenylketonuria.[41] Moreover, prenatal diagnosis is not yet practical for the common hematological disorders sickle cell anemia and glucose-6-phosphate dehydrogenase deficiency.[42] A prenatal test for sickle cell anemia is particularly desired since about one in 500 black infants in the United States has this painful and debilitating disease of the red blood cells.[43]

Screening Newborns for Inherited Metabolic Disorders

Among the states, Massachusetts seems to have the most extensive program of screening newborns for inherited metabolic disorders.[44]

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improved rapidly in recent years. New chromosome staining techniques now allow for the specific recognition of individual chromosomes. Chromosomes stained with quinacrine mustard, when viewed in the fluorescence miscroscope, reveal patterns of light and dark bands along the lengths of the chromosomes that allow for the ready identification of various chromosome translocations. The Giemsa stain is complementary to the quinacrine fluorescence method, revealing a different set of chromosomal details. Use of these methods depends upon locating cells in metaphase under the microscope (*i.e.*, cells in which the chromosomes are all lined up at the metaphase plate preparatory to reassortment into the daughter cells during cell division). Systems for automatic cytogenetic analysis that will take over the tedium of locating metaphase cells and perform a karyotype analysis are under development. Such systems will be required if chromosomal analyses on amniotic cells are to become routine for all pregnancies. A. MILUNSKY, THE PRENATAL DIAGNOSIS OF HEREDITARY DISORDERS. 32 (1973).

38. For a list of hereditary biochemical disorders diagnosable *in utero, see* Emery, *supra* note 21, at 292.

39. Brimblecombe, *Screening for Cystic Fibrosis,* 2 LANCET 1428 (1973).

40. Emery, *supra* note 21, at 292.

41. Milunsky, et al., *supra* note 22, at 1445.

42. Littlefield, et al., *Prenatal Genetic Diagnosis: Status and Problems,* in ETHICAL ISSUES IN HUMAN GENETICS: GENETIC COUNSELING & THE USE OF GENETIC KNOWLEDGE, 43, 48 (Hilton & Callahan eds. 1973).

43. *Id.*

44. The Massachusetts statute appears to give the state Department of Health great flexibility in determining which genetic screening programs will be undertaken. MASS. GEN. LAWS ANN. ch. 76, § 15B (Supp. 1974). Participation in the sickle cell anemia screening program is voluntary for most persons (§ 15B) but the commissioner of public health may require that children susceptible to the disease submit to a blood test (§ 15A). New York State, by contrasts, *requires* each infant to be tested within the first 28 days of life for PKU, homozygous sickle cell disease, branched chain ketonuria, galactosemia, homocystinuria, adenosine deminase deficiency, histidinemia, and such other diseases and conditions as the commissioner of public health may designate. N.Y. PUB. HEALTH LAW 2500(a) McKinney Supp. 1974).

Umbilical cord blood is tested at birth for galactosemia[45] and maternal phenylketonuria.[46] A blood sample is taken from the baby at 2-4 days of age in the hospital and tested for phenylketonuria, maple-syrup-urine disease,[47] homocystinuria,[48] tyrosinosis[49] and galactosemia.[50] Finally, a urine sample from the baby at 3-4 weeks of age is tested for other metabolic and renal transport disorders.[51] The routine screening of newborns is limited to metabolic disorders and does not include screening for chromosomal abnormalities since the costs would be prohibitive. The development of automated cytogenetic analysis may some day make feasible routine screening for chromosomal abnormalities in newborns.

Screening Children and Adults for Inherited Disorders

A child or adult may undergo testing for genetic disease either because he has been referred by his own physician to a genetic counseling unit or because he is a relative of a person who is a carrier or suffers from genetic disease. In addition, certain genetic disorders are especially prevalent among some ethnic groups. For instance, the hemoglobinopathies are most common among persons of African descent and Tay-Sachs disease is most common among Ashkenazic Jews.[52] Accordingly, campaigns have been undertaken to screen the North American Jewish population for Tay-Sachs disease, and some states have required the testing of school children for sickle cell anemia, although more recently some states have

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45. Galactosemia is an inborn error of metabolism due to absence of the enzyme 1-phosphate uridyl transferase resulting in inability to convert galactose into glucose. Clinical manifestations are a failure to thrive in infancy, jaundice, mental retardation, cataract formation, and involvement of liver and spleen.

46. Levy, *Newborn Screening for Metabolic Disorders,* 288 N. ENG. J. MED. 1299 (1973).

47. Maple-syrup-urine disease is a deficiency of the enzyme branched chain keto acid decarboxylase, which results in abnormally high concentrations of the amino acids valine, leucine, isoleucine, and the presence of the alloisoleucine in blood, urine, and cerebrospinal fluid. Clinical manifestations include the maple-syrup-like odor of the urine noted shortly after birth, and severe mental retardation, seizures and death if the severe infantile form of the disease is not treated by diet.

48. Homocystinuria is due to absence or deficiency of the liver enzyme cystathionine synthase. Clinical manifestations are mental retardation, enlarged liver, cardiovascular and skeletal disorders.

49. Tyrosinosis results from a deficiency of the enzyme p-hydroxylphenyl pyruvic oxidase in early life. Clinical manifestations are similar to PKU and include failure to thrive, seizures, gastrointestinal disturbances, and unless treated by diet, mental retardation.

50. Levy, *supra* note 46, at 1300.

51. *Id.*

52. R. Goodman, *supra* note 16, at 90.

repealed or amended these laws.[53] The screening of children and adults for genetic disease presents no special technical difficulties except to maintain high accuracy and keep costs down so that the methods used will be economically feasible for mass screening.

Legal Issues in Genetic Screening

There seems to be general agreement that the major goals of genetic screening are "to find persons with particular genotypes in order to fulfill such traditional medical objectives as the provision of care for people who are sick and the prevention of disease."[54] Screening may be done to provide reproductive information, for research, or to determine the incidence of various genetic disorders.[55] Because the legal issues depend on the circumstances under which genetic screening is done, this discussion will be divided according to the age of the screenee: prenatal, neonatal, and adult.

Prenatal Screening

As noted above, amniocentesis is usually performed around the fifteenth week of pregnancy, with results available around the twentieth week. Because no hospital routinely performs abortions after the twentieth or twenty-first week of pregnancy, there is no opportunity to rerun the test to confirm a diagnosis. Of course, as in any other diagnostic test, mistakes can be made. Specifically, one may obtain a false positive (a test that concludes that the fetus is affected, when in fact it is not), or a false negative (a test that concludes that the fetus is not affected, when in fact it is). The abortion of one false positive, a normal male fetus falsely diagnosed in a woman who had previously given birth to a Tay-Sachs daughter, has already been reported in the literature.[56] Other studies have revealed that a significant number of such errors is predictable, and that negligence in obtaining an inadequate sample of amniotic fluid, shipment difficulties, technical problems at the

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53. Curran, *The Questionable Virtues of Genetic Screening Laws,* 64 AM. J. PUB. HEALTH 1003 (1974).

54. NATIONAL ACADEMY OF SCIENCES, GENETIC SCREENING: PROCEDURAL GUIDANCE & RECOMMENDATIONS, 1 (1975).

55. *Id.*, at 2-4.

56. Kardon, et al., *Pitfalls in Prenatal Diagnosis Resulting from Chromosomal Mosaicism,* 80 J. OF PEDIATRICS 297 (1972).

laboratory, cell contaminations, and human error could all produce inaccurate test results.[57]

An example illustrates the possibilities. Assume that the genetic defect under study appears in one of every 50 subjects, and the test being used has been developed to a point where it has a sensitivity of 99 percent (*i.e.*, it correctly classifies 99 percent of all true positives), and a specificity of 99 percent (*i.e.*, it correctly classifies 99 percent of all true negatives). If this test were used prenatally, and a decision to abort or not to abort were made on the basis of the test results alone, the predicted results can be illustrated by the following fourfold table:

Test

Defect	*positive*	*negative*	
positive	true positive abort (99)	false negative don't abort (1)	100
negative	false postive abort (50)	true negative don't abort (4850)	4900
	149	4851	5000

Such a test would have a predictive value for a true positive of about 66 percent and a predictive value for a true negative of about 99.9 percent. The low incidence (1:50) of the genetic defect in this hypothetical situation means that the number of false negatives will be low, but at the expense of aborting a relatively large number of false positives, *i.e.*, while few defective babies will be born after testing, many healthy babies will be aborted. If the defect occurred in only one of every 100 subjects, and all other facts remained the same, such a test would yield as many false positives as true positives: for every defective fetus aborted, a healthy fetus would also be aborted. Such a test might nevertheless find acceptance in the medical profession for two reasons. One, there has been a traditional concern with letting false negatives "get

57. Friedman, *Legal Implications of Amniocentesis*, 123 U. PA. L. REV. 92, 103-104 (1974).

away" (*e.g.*, in cases of communicable diseases) and thus (especially if the treatment is not dangerous or expensive) one would rather have more false positives than false negatives. Two, as abortion "on demand" becomes more acceptable and commonplace to society, a false positive that is aborted may not seem as tragic to the medical profession.

Presently only about 60 of the more than 1600 known genetic defects can be prenatally diagnosed by amniocentesis. Since each of these requires its own "test" or interpretation of chromosome patterns, the fourfold table analysis can be applied to each of them separately. The following discussion of remedies presumes negligence in performance of some aspect of the test, although an action might lie for breach of contract if the physician guaranteed the accuracy of amniocentesis.

Remedies for Birth of a Defective Infant Following Amniocentesis

The threshold question is whether a court should permit any suit founded, as this one would be, on the concept of "wrongful life." The argument courts have favored to date is that no such action should be permitted since, in the words of an early leading commentator, "whether it would have been preferable for a child not to be born [is] a problem which admits of no solution,"[58] Like most "unanswerable" questions, however, it simply begs the relevant one: "Should courts permit compensation for injury suffered to an individual by the negligence of another, even though had there been no such negligent act the individual would not have been born?" So far the arguments against permitting recovery have not been persuasive.

In the leading case, for example, the New Jersey Supreme Court denied both the infant and his parents recovery when the child was born deaf and blind as a result of the rubella his mother suffered during her pregnancy. The court arrived at this conclusion even though her physician apparently knew of the possibility of this injury (estimating its chances at 1:4) and intentionally withheld this information from the mother. Its stated reason for denying the child's claim was that it was impossible to make the plaintiff child

58. Tedeschi, *On Tort Liability for "Wrongful Life"*, 1 ISRAEL L. REV. 513, 538 (1966).

"whole" again, since no one could measure the value of an impaired life versus no life at all.[59] The parents' claim was denied because they would have chosen abortion, a procedure the court found would violate "the preciousness of human life."[60] The former argument is not persuasive, since courts make similar determinations every day in cases involving personal injuries, wrongful death, and lately in analogous cases concerning negligent failures of sterilization procedures.[61] The policy against abortion is also undermined by the recent Supreme Court decisions stating that abortion can be a constitutionally protected right.[62]

What was probably really at stake (although the court seems to deny it) is a belief that life itself is always and under all circumstances a blessing.[63] It is, however, precisely because this premise is not accepted that a couple seeks amniocentesis in the first place, *i.e.*, their desire is to give birth only to a genetically normal child. If amniocentesis followed by abortion of affected fetuses is accepted as an appropriate decision, compensation for birth of an affected fetus due to medical negligence should also be accepted. Nothing in any of the other so-called "wrongful life" cases argues against this conclusion.[64]

A recent Texas case with circumstances similar to those in New Jersey came to a more reasonable conclusion. The Texas court said

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59. Gleitman v. Cosgrove, 49 N.J. 22, 227 A.2d 689 (1967).

60. *Id.*

61. *E.g.*, Custodio v. Bauer, 59 Cal Rptr. 463 (1967); and *see generally* Annot., *Medical Malpractice, and Measure and Element of Damages, in Connection with Sterilization or Birth Control Procedures,* 27 A.L.R. 3d 906.

62. Roe v. Wade, 93 S. Ct. 705 (1973), Doe v. Bolton, 93 S. Ct. 739 (1973).

63. *See* Capron, *Informed Decisionmaking in Genetic Counseling: A Dissent to the "Wrongful Life" Debate,* 1971 INDIANA L. J. 581, 595-602.

64. Steward v. Long Island College Hospital, 58 Misc. 2d 432, 296 N.Y.S.2d 41 (Sup. Ct. 1968), modified, 35 App. Div. 2d 531, 313 N.Y.S.2d 502 (1970), *aff'd,* 30 N.Y.2d 695, 283 N.E.2d 616, 332 N.Y.S.2d 640 (1972) (German measles case in which hospital committee had refused to permit abortion; court follows *Gleitman* saying recognition of right to recover must come from legislature); Zepeda v. Zepeda, 41 Ill. App. 2d 240, 190 N.E.2d 849 (1963), *cert. denied,* 379 U.S. 945 (1965) (illegitimate child's action against his father for illegitimacy dismissed because of the court's belief that granting relief would create too sweeping a precedent and therefore the decision was more properly one for the legislature); Williams v. State, 18 N.Y.2d 481, 223 N.E.2d 343, 276 N.Y.S.2d 885 (1956) (child born as a result of a sexual assault on a woman confined as a patient in a state mental institution; suit brought against the state dismissed because of difficulty in measuring damages for illegitimacy, and the potential far-reaching effects of granting relief); Pinkney v. Pinkney, 198 So. 2d (Fla. Dist. Ct. App. 1967) (an action by a daughter against her father for having caused her birth out of wedlock and for having shot her mother; action dismissed citing *Zepeda* as precedent). And *see generally,* Waltz & Thigpen, *Genetic Screening and Counseling: The Legal and Ethical Issues,* 68 NORTHWEST. U. L. REV. 696, 750-67 (1973), and Capron, *supra* note 63.

it was not necessary to determine the value of life with a defective body as opposed to no life at all. Instead, one could sue the negligent physician not for the amount of money it takes to raise a child, but for the amount of additional money the parents must spend solely because of the physical defects of the child. The decision to permit the parents to sue was based on the physician's failure to inform the mother of the potential risk to her fetus. In the words of the court:

> It is impossible for us to justify a policy which at once deprives the parents of information by which they could elect to terminate the pregnancy likely to produce a child with a defective body, a policy which in effect requires that the deficient embryo be carried to full gestation until the deficient child is born, and which policy then denies recovery from the tortfeasor of costs of treating and caring for the defects of the child.[65]

Any woman who employs a physician for prenatal care should have a right to have the physician fully inform her of any reason he has to believe that the fetus might be defective, and to further inform her of the existence of diagnostic tests that might identify precise genetic defects.[66] The physician incurs the duty of disclosure because it is precisely this kind of information that the woman employed the physician to learn in the first place, *i.e.*, to learn all she could to help her have a healthy child. If the physician fails to disclose this information he may be treating the patient under false pretenses, and accordingly her consent to that treatment may be invalid. The physician does not guarantee a healthy child, but the reasonable expectation of the patient is that she will be apprised of any information the physician has that the child might be defective and of the alternative ways to proceed, so that the patient can determine what action to take.

Remedies for the Abortion of a Healthy Fetus Following Amniocentesis

At least one physician has suggested to his colleagues in writing that an attempt to sidestep this issue is acceptable practice. In the words of Dr. Fritz Fuchs: "If [following abortion] the diagnosis cannot be upheld, and the patient demands to know, the obstetrician or whoever acts as the genetic counselor is in a delicate

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65. Jacobs v. Theimer, 519 S.W.2d 846, 849 (Tex. 1975). *Contra* Smith v. U.S., 392 F. Supp. 654 (D.C. Ohio 1975).

66. *Cf.* Friedman, *supra* note 57, at 146-48.

situation, unless he has made reservations for such a possibility in advance. He can tell the patient to attempt another pregnancy and, in my experience, most patients will follow this advice."[67]

Another pregnancy may well be advisable, but if negligence was involved in the faulty diagnosis so is malpractice. In such a suit assessing damages should present the court with little difficulty, since the case should be considered one for wrongful death for damages purposes. Many jurisdictions, for example, have recognized the rights of prospective parents to bring actions for fetal death negligently caused.[68] While recovery has been limited to viable fetuses, the main reason for this limitation appears to be the court's inability otherwise to determine the existence of proximate cause between the defendant's action and the death of the fetus.[69] With amniocentesis followed by abortion, of course, proving causation should not present a problem.

Failure to Perform Amniocentesis as Negligence

Under what circumstances does a physician have a legal duty to refer his patient to a specialist for the performance of amniocentesis? The general rule is that referral to a specialist is required when the physician knew or should have known that this particular pregnancy was beyond his own competence.[70] When the plaintiff is able to establish that her age or medical history indicated that amniocentesis should at least have been considered, the physician may be held liable for the birth of a defective fetus if it can be shown that amniocentesis would have detected the defect and led to an abortion.[71] The physician should not be permitted to argue that

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67. Fuchs, *Amniocentesis and Abortion: Methods and Risks,* 7 BIRTH DEFECTS 19 (April, 1971).

68. *E.g.,* Hale v. Manion, 189 Kan. 143, 368 P.2d 1 (1962); and cases collected in Annot., 15 A.L.R. 3d 992 (1967).

69. PROSSER, HANDBOOK OF THE LAW OF TORTS § 55 at 337-8 (4th ed., 1971); and *see* Mace v. Jung, 210 F. Supp. 706 (D. Alas. 1962).

70. G.J. ANNAS, RIGHTS OF HOSPITAL PATIENTS, ch. VIII (Consultation, Referral and Abandonment) (1975).

71. Waltz & Thigpen, *supra* note 64, at 757. Such cases have actually arisen involving both Down's syndrome and Tay Sachs. In the only jury case that the writers are aware of a verdict was returned in favor of the defendant physician who failed to advise a 42-year-old woman in 1971 of the existence of amniocentesis. At the time of trial the plaintiff's Down syndrome child was 3½ years old. Park v. Nissen, Cal. Super. Ct., Orange Co., Docket No. 190033 (Dec. 13, 1974). Facts summarized in 31 THE CITATION 38 (June 1, 1975). The Tay-Sachs case is apparently still in the courts. Schneck et al., letter to the editor, 292 NEW ENG. J. MED. 758 (1975).

he or she made an independent judgment that amniocentesis was not appropriate and therefore did not discuss it with the patient—the decision must be the patient's, not the doctor's. Further, no therapeutic privilege should protect the physician from nondisclosure in these circumstances since he is not treating the condition, but actually *preventing* the mother from making an informed decision concerning abortion, currently the only available "treatment" to prevent most genetic diseases.

A more novel, but not inconceivable, approach would hold the physician negligent for failing to routinely perform amniocentesis on all pregnant women with potentially affected fetuses, or for failing to refer these patients for such testing, even if such routine testing is not the standard of practice. The rationale would follow that enunciated in the recent case of *Helling v. Carey.*[72] There the supreme court of the state of Washington held that an ophthalmologist was negligent because he did not routinely perform a pressure test designed to detect glaucoma, even though the use of such a test was not standard medical practice among specialists and the incidence of glaucoma in patients the age of the plaintiff was approximately one in 25,000. The major rationale the court used was that the diagnostic test was safe, inexpensive, simple, and accurate, and could prevent or at least arrest blindness if routinely performed.[73] Therefore, the court reasoned that the public was entitled to the benefit of the test, and its non-use was itself evidence of negligence. A minority of the court would have found against the physicians on the basis of strict liability, on the theory that while they may not have been personally culpable they were in the best financial position to bear the loss and obtain insurance for such statistically predictable missed diagnoses.[74]

Since amniocentesis presently is relatively expensive, not easy, not 100 percent accurate, and carries significant risks to both the mother and fetus, the analogy is not presently persuasive. Nevertheless, as techniques improve in terms of accuracy and safety, one can envision application of this analogy to permit recovery for the birth of a defective child. A court would not have to

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72. 83 Wash. 2d 514, 519 P.2d 981 (1974).

73. *See* Annas, *The Case of the Simple, Harmless, Inexpensive, Conclusive Diagnotic Test,* 4 ORTHOPAEDIC REV. 67 (Feb. 1975).

74. *Id.*

go this far, however, to find a physician liable for not disclosing to an at-risk patient the existence of this diagnostic test. One possible rationale could be to find that the physician was treating the patient throughout the pregnancy and birth without her informed consent because he had withheld from her a vital piece of material information—the existence of a test to determine whether or not her fetus was affected by some specific defect.[75]

Neonatal Screening

As with amniocentesis, screening errors can also be made at birth. However, the consequences for the newborn are generally not death, but more likely improper or no treatment, with or without stigmatization. Two examples illustrate these problems.

In the initial enthusiasm that was created when a screening test for phenylketonuria (PKU) was developed, 41 states passed laws from 1963 to 1967 regarding such testing of newborns.[76] Most made testing mandatory for all newborns.[77] PKU is caused by an enzyme defect that allows phenylalanine to accumulate in the blood, which leads to irreversible mental retardation. It is thought by most that if children with an elevated blood level of phenylalanine are detected early enough and placed on a diet containing no phenylalanine, the effects of this disorder can be avoided.[78] Unfortunately, experience has demonstrated that the diet itself "is not without danger," and phenylalanine deficiency syndrome may cause listlessness, rash, bone changes, hypoglycemia, anemia, susceptibility to infection and death.[79] Many infants may have been subjected to the risks involved in this treatment unnecessarily because of the imprecision of the tests used to diagnose the disorder.[80]

In view of the facts that PKU accounts for fewer than one percent of the institutionalized retarded, is a relatively rare disorder, and has a controversial treatment of unknown success, it is not un-

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75. *Cf.* Milunsky & Reilly, *The "New" Genetics: Emerging Medicolegal Issues in the Prenatal Diagnosis of Hereditary Disorders,* 1 AM. J. LAW & MED. 71, 75-76 (1975).

76. Swazey, *Phenylketonuria: A Case Study in Biomedical Legislation,* 48 J. URBAN L. 883, 908-09 (1971).

77. *Id.* and *see Screening for Phenylketonuria* in NATIONAL ACADEMY OF SCIENCE, GENETIC SCREENING: PROGRAMS, PRINCIPLES & RESEARCH 19-93 (1975).

78. Swazey, *supra* note 76, at 899-900.

79. *Id.* at 900-901.

80. Parker, *Some Legal Aspects of Genetic Counseling,* 7 PROGRESS IN MED. GENETICS 217, 220 (1970).

reasonable to conclude that PKU screening laws must be considered "premature biomedical legislation."[81] This experience should teach that voluntary programs should be used as long as feasible to gain sufficient information about the efficacy of both the screening test and the intervention planned, and that mandatory statutes should not be enacted before there is at least a reasonable medical certainty that the measures they prescribe are both necessary for the public health and capable of achieving this legislative purpose. Knee-jerk reaction based on a laudable desire to help prevent mental retardation may create more problems than it solves, both by raising unreasonable expectations on the part of the public and by subjecting certain classes of persons to unproven testing and treatment.

Another problem, that of potential stigmatization, can be illustrated by screening the newborn for XYY. In 1962 it was suggested that this sex chromosome abnormality was associated with a predisposition to antisocial or criminal behavior.[82] Since then this theory has been widely debated and generally discredited, although no prospective study of the problem has yet been made.[83] In an attempt to do such a study, Dr. Stanley Walzer of Boston's Children's Hospital began in 1968 to screen all newborn males at the Boston Hospital for Women for XYY and to follow up those with this chromosome configuration, matching them with another male child with a normal XY sex pair.[84] Putting aside the controversy that later arose over the type of informed consent necessary from the mother both for the initial screening and the

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81. Swazey, *supra* note 76, at 927. At least two physicians have, however, been found negligent for failing to timely diagnose PKU. Naccarato v. Grob, 180 N.W.2d 788 (Mich. 1970), and current medical literature seems to support the treatment's efficacy. *See, e.g.*, Smith & Wolff, *Natural History of Phenylketonuria and Influence of Early Treatment*, LANCET 540 (Sept. 1974). Starfield & Holtzman, A Comparison of Effectiveness of Screening for Phenylketonuria in the United States, United Kingdom and Ireland, 293 N. ENG. J. MED. 118 (1975).

82. Court-Brown, 1962 LANCET 508; and *see* Jacobs, et al., *Aggressive Behavior in Mental Subnormality and the XYY Male*, 208 NATURE 1351 (1967), *and* Price & Whatmore, *Behavior Disorders and Pattern of Crime among XYY Males Identified at a Maximum Security Hospital*, 1967 BRIT. MED. J. 513.

83. *See e.g.*, SHAH, REPORT ON THE XYY CHROMOSOMAL ABNORMALITY, (U.S. Gov. Print. Office, 1970); Note, *XYY Syndrome and the Judicial System*, 6 N.C. CENT. L. J. 66 (1974).

84. *See* discussion in GENETICS AND THE LAW (Milunsky & Annas eds., in press). In June, 1975 Dr. Walzer decided to discontinue his research because "he had become worn down by harassment, unrelenting controversy and the threat of further opposition to his work by groups supporting children's rights." *N.Y. Times*, June 20, 1975 at 30, col. 1.

follow-up studies, a serious question of stigma can be raised.

With XYY thought by most of the scientific community to be no more disposed to violence or antisocial behavior than its "normal" counterpart, XY, why should parents have it suggested to them that their child is "abnormal" in this regard? Furthermore, what protections must be taken to insure that this information is not transmitted to the police or school authorities, who may pay more attention to this individual because of his chromosome makeup? Because of society's profound interest in and fascination with genetics and eliminating "defectives" or deviance, one must be extremely careful in classifying a certain genetic trait as potentially harmful to society, because the pressures to eliminate it (or its carriers) will be intense whether the thesis is correct or not. No matter what the literature, for example, XYY may be considered a "serious" enough "abnormality" to lead most couples who have undergone amniocentesis for another reason to choose an abortion for this reason alone even if the child would probably otherwise be normal. Premature publicity may make a reasoned discussion of the current state of knowledge concerning XYY extremely difficult. As will be discussed in the next section, the trend toward more genetic screening will lead to increased pressures for central registries and wider application of the information obtained. As genetic information is used for more and more purposes (and the probability of stigmatization based both on medical fact and popular beliefs increases), measures to insure both its accuracy and confidentiality become increasingly important.

Screening of Children and Adults

Screening in this group is almost always done as a prelude to genetic counseling concerning marriage or reproduction, although there have been examples of attempts to screen as a prerequisite for entry into grade school. Unlike newborn screening, these tests have almost always been voluntary. The major programs thus far have involved hereditary diseases concentrated in specific racial groups, Tay-Sachs in Ashkenazic (Eastern European origin) Jews and sickle cell disease in Blacks.

Tay-Sachs is a severe autosomal recessive condition characterized by blindness, severe mental retardation, and early death, usually before three years of age. Its incidence in Ashkenazic Jews

is about one in every 3,600 births. A number of large voluntary screening programs to identify carriers have been undertaken in the U.S. Since the disease has such horrible consequences to the child, and since it can be tested *in utero,* couples who are both carriers can still be assured of having healthy children provided the woman undergoes amniocentesis during pregnancy. Because of the high motivation not to have such a child, and the ability both to relieve anxiety in non-carriers and to provide an alternative for carriers, this type of screening has been generally well-accepted.[85]

Tay-Sachs is to be contrasted to sickle cell disease, a disorder of the blood in which the red cells assume a sickle or hollyleaf shape. Associated with the disease are hemolysis (breakdown of red cells) with resulting anemia, or occlusion of blood vessels which can lead to painful cycle cell crises. The disease may manifest itself in infancy or as late as the second year of life, and continues throughout one's life. Persons with sickle cell trait, however, rarely show any clinical symptoms.

About 7-9 percent of the U.S. Black population carry the trait, and about 0.3 percent suffer from the disease.[86] In 1970 a simple, inexpensive and relatively reliable test for sickle cell hemoglobin was made available.[87] Shortly thereafter, influenced by strong lobbying led by the Cynthia Foundation, a number of states, beginning with Massachusetts, passed laws mandating sickle cell screening at various times.[88] What followed was a reaction by the Black community on two fronts: the first pushing for more medical research into the problem; the second calling for screening on a purely voluntary basis.

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85. *E.g.,* Kaback, Zeiger & Reynolds, *Tay-Sachs Disease: A Model for the Control of Recessive Genetic Disorders,* PROCEEDINGS OF THE FOURTH INTERNATIONAL CONFERENCE, VIENNA, AUSTRIA, SEPT. 2-8, 1973, EXCERPTA MEDICA 248-262 (1974). An advisory committee of physicians in the city of Dayton, Ohio did, however decide to oppose a mass screening program for Tay-Sachs on concluding that screening the 1800 Ashkenazic Jews between the ages 16 and 45 to prevent the predicted birth of one child with the disease would not be worth the price in terms of increased anxiety in the 72 heterozygous carriers that would be identified in the screening program. Kuhr, *Letter to the Editor: Doubtful Benefits of Tay-Sachs Screening,* 292 NEW ENG. J. MED. 371 (1975). This decision has been subjected to criticism as an example of a "paternalistic attitude that new genetic knowledge imparted to the public may be more harmful than useful," and as "a disservice to the Ashkenazic Jewish community of Dayton." Schneck, Saifer & Volk, *Letter to the Editor: Benefits of Tay-Sachs Screening,* 292 NEW ENG. J. MED. 758 (1975).

86. Binder & Jones, *Prevalence and Awareness of Sickle Cell Hemoglobin in a Military Population,* 214 J.A.M.A. 909 (1970).

87. Loh, *A New Solubility Test for Rapid Detection of Hemoglobin S.,* 61 *J.* INDIANA STATE MED. ASSOC. 1651 (1968).

88. *See generally,* Reilly, *Sickle Cell Anemia,* J. LEGAL MED. 39 (Sept. 1973).

In 1972 the U.S. Congress, in response to a major address by former President Nixon, passed the National Sickle Cell Anemia Control Act. While $115 million was authorized under this act, no monies were actually appropriated, and what funding did occur came from other HEW sources. The act's express limitation on federal funding to voluntary programs, however, appears to have had an impact. Since that time more than half of the states with mandatory laws have replaced them with voluntary ones and made provisions for safeguarding confidentiality.[89] Such safeguards were deemed essential since this genetic information was sometimes used to deny employment and insurance. Moreover, the wisdom of these laws began to be questionable, since there was no method to detect sickle cell disease *in utero* through amniocentesis, and their only purpose seemed to be to warn couples who both had the trait not to have children (one in four of whom would have the disease), and to warn unmarried carriers not to marry carriers. Because this disease is concentrated in the Black population, the charge by that group that what was involved was an attempt to reduce Black reproduction cannot be easily dismissed. With this type of racial concentration, voluntariness in the program is especially important.[90] While none of these laws have been challenged as to their constitutionality, serious doubts could certainly be raised. The Kentucky law, for example, specifically applied only to those "of the Negro race."[91]

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89. Curran, *supra* note 53.

90. *See, e.g.*, Fort, et al., *Counseling the Patient with Sickle Cell Disease about Reproduction: Pregnancy Outcome Does Not Justify the Maternal Risk*, 111 AM. J. OBSTET. GYNECOL. 324-27 (1971). Based on a six percent maternal mortality rate and a 20 percent infant mortality rate among mothers with sickle cell disease these authors state: "We advocate primary sterilization, abortion if conception occurs, and sterilization for those that have completed pregnancies. Patients with sickle cell disease should be unhesitatingly thus counseled." While the authors might argue that such "counseling" is for the patient's own good, what is really at stake is an attempt by a group of physicians with specific beliefs, which could be viewed as racist or even genocidal, to impose those beliefs on a defined population of patients—regardless of the patient's own opinion concerning the appropriateness of taking such risks.

91. KENT. REV. STAT. § 402.310.

Sickle cell anemia is of relatively frequent occurence not only in Blacks but also in Meditteranean peoples and, generally, wherever falciparum malaria has been a problem. Motulsky, A. G., *Current Concepts on the Genetics of the Thalassemias*, 29 COLD SPRING HARBOR SYMP. QUANT. BIOL. 399 (1964).

The Thalassemias are a group of inherited disorders in which the synthesis of one or more of the polypeptide chains of hemoglobin is suppressed, either partially or totally. The clinical picture varies with the particular genetic defect from barely detectable to severely anemic. The most common form of Thalassemia is Cooley's anemia, also referred to as Mediterranean anemia of Thalassemia because of the high frequency in persons of Mediterranean

Confidentiality in Genetic Screening

No one should be screened without his or her prior, fully informed, consent. Since the genetic basis of disease is not well understood by the general population, special efforts are necessary to make the implications of a positive finding known to the individual prior to screening. The potential hazards of stigmatization, loss of employment or insurance, and family discord should all be openly emphasized, as well as the possibilities of a false-positive or false-negative finding. The procedure should be fully explained to the individual, and the individual should be granted complete access to all his or her medical records regarding the test and its interpretation. The individual should also have the right to refuse to permit *any* person other than his or her physician or the person in charge of the screening program to see or copy these records without his or her prior written permission, unless a third party with a legitimate interest in the data could in no way identify the patient from the record. As in amniocentesis, it seems apparent that the subject of the screening process has the right to all information obtained, except where he or she has made a voluntary and informed prior agreement restricting access to the findings.[92]

The much more difficult question that arises in this context is the right or duty of the physician-counselor to inform potentially affected relatives of the findings. If one of the parents is found to be a carrier of a serious genetic disease, can the physician inform potentially affected relatives? For example, if the mother is found to be a carrier of the Lesch-Nyhan syndrome through an affected son, can the physician inform the mother's sisters?[93] Assuming that the affected parents refuse to give the physician permission to notify their relatives who might also be affected, what should the physician do? The remedy of a suit for breach of confidence is inadequate protection for the patient, as it necessarily involves

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background. Cooley's anemia manifests itself as a severe anemia characterized by enlargement of liver and spleen, high white blood cell count, thickening of the diploe and long bones, and early mortality. For details, *see* GOODMAN, R. M., GENETIC DISORDERS OF MAN (1970) at 176. Demands for Thalassemia screening have been expressed in the United States by people of Mediterranean background. *See* Peason, H A., et al., *Screening for Thalassemia Trait by Electronic Measurement of Mean Corpuscular Volume,* 288 N. ENG. J. MED. 351 (1973).

92. *See generally* NATIONAL SCIENCE FOUNDATION, GENETIC SCREENING, *supra* note 77, at 251-64.

93. MILUNSKY, THE PRENATAL DIAGNOSIS OF HEREDITARY DISORDERS (1973) at 159.

public disclosure of the information the patient desires to be kept secret. Therefore the doctrine of informed consent prior to counseling (and screening), and again prior to disclosure to third parties, may help create an atmosphere that encourages strict observance of confidentiality, because the informed patient will probably not consent to screening and counseling in the first place if provisions for data confidentiality are known and perceived as being inadequate.

There is a long line of cases that *permits* a physician to disclose the diagnosis of a contagious disease to all who need this information in order to protect themselves from contracting the disease.[94] Statutes have also been enacted that *require* physicians to disclose confidential medical information to public authorities in certain circumstances (*e.g.*, some contagious diseases, gunshot wounds, child abuse, etc.), under the police power of the state to protect the health and safety of its citizens.[95] In such cases the physician has an affirmative duty to disclose what would otherwise be confidential medical information. While none of these seems to apply directly to the genetic counseling situation, a recent California case has indicated that the duty of a physician to warn potentially affected third parties of confidential medical information may be very broad indeed.

In enunciating a general rule based on a case in which a psychologist failed to warn a woman that his patient had threatened to kill her (the woman was, in fact, murdered by the patient), the court said: "Where a doctor . . . in the exercise of his professional skill and knowledge, determines, or should determine, that a warning is essential to avert danger arising from the medical or psychological condition of his patient, he incurs a legal obligation to give that warning."[96] The basis for this far-reaching and arguably unprecedented decision was the court's belief that we live in an "interdependent" and "risk-infested" society, and that members of such a society cannot tolerate being exposed to additional risks that physicians could eliminate by a simple act of communication.

94. Annas, *Law & Psychiatry: When Must the Doctor Warn Others of the Potential Dangerousness of His Patient's Condition?* 3 MEDICOLEGAL NEWS 173 (April, 1975).

95. G.J. ANNAS, THE RIGHTS OF HOSPITAL PATIENTS, ch. XI (Confidentiality and Privacy) (1975).

96. Tarasoff v. Regents of U. of Cal., 118 Cal. Rptr. 129, 529 F.2d 553 (1974).

It would be stretching this decision considerably to find a duty on the part of a genetic counselor to warn other family members that their *offspring* might be in danger because of a gene the family member *might* be carrying. However, in view of the public policy enunciated by this and other courts, a strong argument can certainly be made that such a disclosure would be *permissible* even if not required. Given this type of uncertainty in the law, and the fact that this situation can easily be anticipated as counseling becomes more widespread, it would seem appropriate that this be the subject of legislation. The policy of routine disclosure or of strict nondisclosure would be dictated by the view that the legislature took of the primary purpose of screening and counseling—the promotion of individual autonomy in making procreative decisions, or the minimization of certain genetic defects in the species. Unless and until such legislation is enacted, it is extremely important that the genetic counselor make clear, both verbally and in writing, the policy that he or she follows so that the patient can refuse to be screened or counseled if he or she is not in agreement with the policy. Such a policy is, of course, also necessary in deciding whether or not to obtain a complete listing of blood relatives as a routine part of the medical history.

The possibility of a central registry of genetic defects raises analogous problems of privacy and confidentiality, but with even more potential for abuse. The issues are similar to those involved in any centralized system of medical records keeping. First, any such system should be used to store identifiable patient data only with the fully informed authorization of the patient. Second, all information should be available for inspection and correction by the patient. Third, the information should be stored in such a way that gross data on incidence and prevalence can be made available to researchers and policy-makers without identifying any individual patient. Finally, access to the information by unauthorized third parties (*i.e.*, parties not authorized by the patient) should be strictly guarded against, with the central registry liable in tort for the foreseeable consequences of giving out information without authorization.[97]

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97. ANNAS, *supra* note 95.

Conclusion

It is a cliché to say that we stand on the frontier of the genetic era. Most of the techniques discussed in this article are products of the past decade. That any laws have been passed dealing with them is somewhat of a marvel. That almost no cases have come before the courts specifically on point should not be surprising. Nevertheless, the legal issues raised by genetic screening are not unique, and are susceptible to legal analysis by analogy to many of the more familiar problems in traditional medical decision-making. The challenge for the courts is to view genetic advances as steps in a continuum to enhance man's ability to control his life and that of his offspring. The challenge for state and federal legislatures is to carefully examine genetic advances and determine the appropriate governmental role in regulating and fostering their development in a manner which will enhance human rights. The challenge for the attorney counseling individual clients on these matters is to be knowledgeable enough to be able to recommend appropriate courses of action and appropriate experts to consult.

While genetic screening raises important racial, informational, scientific and constitutional problems in a novel context, the law can be an able guide to reasonable solutions.