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Medical Legal Issues in Prenatal Diagnosis

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Prenatal diagnostic testing offers parents the option of avoiding the physical suffering and emotional trauma that attends the birth of children who have severe, debilitating diseases, such as Tay-Sachs, Canavan, ornithine transcarbamylase deficiency, and Down syndrome. In addition, prenatal diagnosis can alert families and health care providers of the need to prepare for the delivery of a compromised child. Finally, in utero diagnostics increasingly help guide physicians and parents or present physicians and parents with opportunities for fetal therapy [1–5]. Although prenatal diagnostic testing encompasses a broad range of clinical diagnostic investigations for genetic and nongenetic conditions, the focus of this article is on the legal issues surrounding DNA-based prenatal testing for inherited conditions. Many principles discussed in this article are applicable to prenatal diagnosis performed for other reasons.

After the demonstration by Steele and Breg [6] in 1966 that chromosomes could be analyzed from cultured amniotic fluid cells, technical advances in cytogenetics, ultrasonography, clinical chemistry, biochemical genetics, and molecular diagnostics, together with legal changes stemming from the Supreme Court's decision in Roe v Wade [7], brought about dramatic growth in the implementation of prenatal diagnosis and screening in clinical obstetrics practice. Screening tests, most often using a pregnant woman's blood, enable more precise statements about risks of certain fetal diseases or defects. Ultrasonography or other imaging techniques may establish a fetal diagnosis or may reveal abnormal anatomy that generates an extensive differential diagnosis. Invasive procedures, like amniocentesis and chorionic villus sampling (CVS), offer definitive diagnoses and have low complication rates.

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Although testing for Down syndrome and other aneuploidies in women of advanced maternal age remains the most common indication for invasive prenatal testing, in utero diagnosis of a growing list of diseases and anomalies by analysis of fetal DNA is increasing in frequency [8]. The current introduction of array comparative genomic hybridization techniques is quickly expanding the diagnosis of small chromosomal abnormalities [9]. Future recommendations to offer widespread DNA-based carrier assessment and prenatal testing similar to those that have been published for cystic fibrosis [10] and fragile X syndrome [11] will likely accelerate the trend toward greater reliance on the use of molecular diagnostic techniques for prenatal diagnosis. Developments in technology have made clinical mutation detection from blood and other human tissues routine [12]. In the near future, noninvasive ways to accomplish DNA-based fetal genetic testing [13,14] will greatly enhance the importance of prenatal diagnostics to obstetricians, geneticists, primary care physicians, pathologists, and other clinical laboratory physicians.

The legal and ethical issues associated with prenatal diagnosis are complex and evolving. It will increasingly be necessary for practitioners to become informed about, and stay abreast of changes in, laws affecting the use of prenatal diagnosis. Key legal areas of concern in the United States include the requirements for informed consent and prenatal genetic counseling, definitions of negligent practice (particularly in relation to wrongful birth and wrongful life lawsuits), and genetic discrimination.

Informed consent

The modern concept of informed consent evolved from the law of "battery" [15]. Battery is defined as an intentional, nonconsensual, offensive touching of another person [16]. In part because of its historical roots, most physicians are familiar with the need to obtain informed consent before performing invasive diagnostic or therapeutic procedures. The requirement for informed consent extends to the provision of all medical care, including diagnostic laboratory testing. Battery has largely been supplanted by negligence (ie, medical malpractice) as a basis for litigation over the alleged failure of a physician to obtain informed consent. This is likely because most such claims are raised in conjunction with other allegations of substandard care, none of which reflects the deliberate intent to injure patient plaintiffs.

It is conceivable that a patient who was injured during CVS, for example, could rely on a battery theory if she alleges that the procedure was unnecessarily performed. A battery claim could also be raised by a patient who argues that the scope of diagnostic testing performed on her behalf exceeded that for which she gave consent. For example, if a patient who has a family history of Huntington disease underwent CVS for reasons unrelated to that disorder, and testing for Huntington Disease was mistakenly