

# How prenatal genetic testing protects patients—and you

Misinterpreted or incomplete genetic testing does not serve the patient—and is likely to spark a lawsuit

## CASE 1 ObGyn fails to scrutinize test results

A woman consulted board-certified ObGyns during the first trimester of her pregnancy and informed one of them that she had recently been given a diagnosis of measles, but was unsure whether the illness was German (rubella virus) measles. The physician examined her and ordered a rubella titer. The results, he noted, “were indicative of past infection.” Instead of performing further testing, he told the patient she “had nothing to worry about because she had become immune to German measles as a child.”<sup>1</sup>

Soon after birth, her child was diagnosed as suffering from congenital rubella syndrome, including eye lesions, heart disease, and auditory defects.<sup>1</sup>

Apparently, the physician had failed to consider whether the positive test result was indicative of current, rather than past, infection—and failed to perform further testing to determine whether the titer was falling, stable, or rising.

Is he legally liable for this omission?

**T**he physician in this case violated a simple tenet: If a test is ordered, the clinician must be able to interpret the results or order further testing to ensure that the patient is given valid advice. This physician’s error was compounded by the fact that the

patient had notified him of the potential for fetal injury.

Would he be held accountable in a court of law?

The answer to that question depends on:

- the state in which the case arose, and the laws in effect at the time of the incident
- the action initiated by the plaintiff and her attorneys, which may involve “wrongful birth,” “wrongful life,” or other allegation
- the time that transpires between the genetic test (or diagnostic test, such as a rubella titer) and the legal case—in other words, the statute of limitations. In some cases, this period encompasses the time elapsed between *omission* of a test (or birth of a child) and the filing of a legal claim.

In this article, I highlight three pertinent legal cases and the lessons to be learned. Because national legal case reporters generally publish opinions from appellate courts, they usually focus on determinations of law, with factual determinations and decisions on liability remanded back to the trial court.

For the purposes of this article, I assume that the facts in the judicial opinion are correct, but focus on legal principle.

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## Don't overlook the patient when planning prenatal tests

*Patient testing: ethical issues in selection and counseling. ACOG Committee Opinion No. 363. American College of Obstetricians and Gynecologists. Obstet Gynecol. 2007;109:1021-1023.*

**K**nowledge can be a dangerous thing. At the very least, a bid for genetic data requires careful consideration and preparation, particularly when the information may determine whether a woman continues or terminates her pregnancy.

In April, the American College of Obstetricians and Gynecologists published a Committee Opinion on ethical considerations in the selection and counseling of patients in regard to testing. Although the opinion concerns testing in general—rather than prenatal genetic testing specifically—a number of recommendations are applicable to both:

- Perform testing for the benefit of the patient, not simply because a third party—e.g., her partner, extended family, employer, insurer, or health-care provider—deems it to be necessary. Ensure that she gives consent before proceeding.
- Do not base testing decisions on assumptions about how the patient will respond. “Prejudgments about a patient’s wishes regarding fetal abnormalities, for example, should not preclude her being offered prenatal testing,” the opinion states.
- Tell the patient how the results will be communicated, and to whom.
- Provide or refer the patient for specific counseling whenever planned testing may have multiple medical or psychological implications.
- Respect the patient’s autonomy and involve her in decision-making. If she elects to forego a recommended test, document her refusal in the medical record—and include the reason.

### Detectable genetic disorders have increased exponentially

Almost any health-care provider encounters patients who have inheritable disease. Cases have arisen against an oncologist for failure to convey to the patient the potential for genetic transmission of colonic polyposis and against a surgeon for failing to warn a young breast cancer patient that she may carry genes that predispose her offspring to ovarian cancer. But the highest-risk category is reserved for the obstetrician, who must, on a daily basis, consider the need for appropriate screening and proper counseling about the availability of genetic testing that may enable the

patient to avoid passing catastrophic disease to her children.

This responsibility is complicated by a dramatic increase in the number of genetic tests. Few areas of medical practice have expanded as rapidly as prenatal genetic testing and counseling. Forty years ago, an obstetrician—or even a nurse—might have discussed the risks of pregnancy at an advanced age, as well as a few possible genetic abnormalities. Today, certified genetic counselors are employed by many clinics and even private practices, and more than 1,000 disorders—as many as 1,300, by some reports—can be detected by the proper test.

Although most of these disorders are uncommon, several dozen or so are more prevalent and, therefore, regularly tested for. They include cystic fibrosis, Down syndrome, and neural tube defects, as well as diseases associated with certain religious, racial, or ethnic groups, such as thalassemia (Mediterranean, Asian, or African heritage), Tay-Sachs (Eastern European or Ashkenazi Jewish descent), and sickle cell disease (African-American ancestry).

### Ethical complexity makes resolution difficult

Legal actions are bound to increase as technology evolves, particularly when a child is born with a genetically transmissible disorder that could have been identified with proper counseling and testing. These legal actions are particularly difficult to resolve because they involve complex ethical issues. For example, in “wrongful birth” actions, the parents often allege that they would not have conceived a child if they had known that it might be afflicted with a certain trait or illness, or that they would have terminated the pregnancy if they had learned of the disorder in a timely fashion.

In most of these cases, the plaintiffs are the parents. In a few states, however, successful actions have been brought by the child (“wrongful life”), who, some

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contend, essentially claims that nonexistence would be better than life with a serious congenital condition. They usually seek the extraordinary expense associated with the condition, as well as other damages.

Similar actions have been brought by children, and parents of children, with acquired conditions such as congenital rubella syndrome, as in the opening case. In that scenario, the parents' case was declined by attorneys because their state of residence did not allow actions for wrongful birth or wrongful life. The state courts subsequently changed the law to allow wrongful birth actions, but by the time the parents learned of the change in the law, the statute of limitations had expired. They therefore added the attorneys as defendants, alleging that they failed to recall the parents when the law changed, in time to file their lawsuit. By the time the case finally reached the state's supreme court, the law had changed again, and the child was allowed to proceed with a wrongful life action.

### **CASE 2 Physician mistakes patient's tissue for fetus's**

Before becoming pregnant, a woman experienced several medical problems that prompted her and her husband to seek genetic testing and counseling. Testing revealed that she had a balanced translocation of chromosomes 11 and 22.<sup>2</sup>

After conceiving, the woman underwent chorionic villus sampling, which indicated that the fetus was probably a female with the same chromosomal condition as the mother—therefore, she would develop normally. The mother also underwent several ultrasonographic tests to rule out fetal abnormalities and was told that imaging showed a normally developing fetus.

The woman went on to deliver a boy, and genetic testing revealed that he had trisomy 22, with severe, permanent disabilities.<sup>2</sup> The parents sued.

Were the woman's health-care providers liable?

This case prompts a number of questions:

- Did the physicians seek confirmation that the tissue studied was fetal?
- Did they counsel the parents about the possibility that maternal genetic material might be recovered during sampling? If so, did they consider that possibility when the specimen was identical to the mother's?
- How is it possible that, on subsequent ultrasonographic evaluations, the male sex of the fetus was not noted? If it was noted, did it register with the providers that the result was inconsistent with the genetic studies?

These questions were not addressed in the court opinion, but they could have had a significant impact on the final outcome of the case. Although the court allowed some aspects of this case to go forward, damages were limited to the costs of the pregnancy and—for reasons not disclosed—that claim was dismissed. If the delivery had been complicated, however, those costs could have been substantial.

Despite the dismissal, which was a favorable turn of events for the physicians, they almost certainly lost substantial work time and experienced the emotional roller-coaster ride that accompanies being sued—not to mention the revelation that they may have harmed a patient.

### **CASE 3 Physician fails to complete testing**

The mother of a developmentally delayed daughter delivered another child. Before she conceived, however, she and her husband consulted a physician (a pediatrician) to find out whether there was a genetic reason for the daughter's developmental delay and the mental retardation of another child, the girl's half-brother.<sup>3</sup> The physician wrote in her notes: "? Chromosomes + fragile X" to indicate testing planned for the daughter.

The testing was performed at a medical center and reported to be normal. The physician relayed this finding to the parents, but failed to mention that fragile X testing

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### **FAST TRACK**

**Any test planned in discussion with the patient must be carried out—unless the patient is notified otherwise**

## What's the standard of care in genetics? And where are the pitfalls?

Aubrey Milunsky, MD, DSc, DCH

The standard of expected care in clinical genetics is generally the same across most medical specialties. That standard is compounded in obstetrics, however, by critical considerations in three areas in the spectrum of care: future conception, ongoing pregnancy, and postnatal implications. Although guidelines and committee opinions on genetics-related care that are issued by professional societies, such as ACOG, are important in practice, they do not, in fact, set the actual standard of expected care—as those issuing bodies have emphasized in their statements.

Mostly, it is *expert opinion* that informs a jury about the standard of expected care in genetics. And, ultimately, any action (or inaction) taken by a jury is judged according to what the average, prudent physician would do under the circumstances in question.

### Errors tend to be basic

Medical negligence claims relative to genetics in obstetrical practice are usually *not* a complex matter of

failure—for example, failing to diagnose an isodicentric chromosome, misinterpreting a copy number genome variation, mistaking a gene polymorphism as a disease-causing mutation, and failing to order a microdeletion analysis when a gene sequence is reportedly normal. Rather, errors are usually basic and, invariably, not single; often they result in serious consequences. Space allows only a few, brief (and arbitrary) examples here of where pitfalls can be found:

- genetic counseling
- laboratory interactions
- group practice
- knowledge of practical genetics.

**Genetic counseling** Simple but critical matters in this area of office practice include taking a family history and not ignoring potential implications in that history of mental retardation, congenital malformations, genetic disorders, specific cancers, and ethnicity. These clues should alert you to offer diagnostic or carrier tests or to refer the patient for a genetics consultation.

Some OBs have the benefit of genetic counselors in their practice, but fail to realize that these members

### FAST TRACK

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had not been conducted. Because they had discussed fragile X testing during the initial consultation, the mother assumed it had been performed and was included in the normal results.<sup>3</sup>

The latest child was also developmentally delayed, and subsequent testing revealed that all three children were positive for fragile X syndrome. The parents sued, alleging that the physician was negligent for failing to perform the genetic test, and claiming that they would not have conceived another child if they had known of the daughter's fragile X status.

Are their claims reasonable?

Despite a state statute that prohibited wrongful birth and wrongful life actions that allege that the pregnancy would have been terminated if the parents had known of the genetic impairment, the state supreme court allowed this case to proceed because the issue was whether the mother would have avoided *concep-*

*tion*. The plaintiffs also argued that the claim did not exceed the statute of limitations. Their reasoning: Although the medical care had been provided more than 4 years earlier (the limitation period), the injury did not occur (i.e., action did not “accrue”) until conception of the child.

Perhaps most telling was the court's statement that, “[O]ur decision today is informed by the practical reality of the field of genetic testing and counseling; genetic testing and diagnosis [do] not affect only the patient. Both the patient and her family can benefit from accurate testing and diagnosis. And, conversely, both the patient and her family can be harmed by negligent testing and diagnosis.”<sup>3</sup>

### Establishing degree of harm is especially difficult

As you might imagine, allegations involving potential pregnancy termination

of the staff, albeit very valuable, are not the full and final repository of knowledge about genetic disease. Failure to offer a test, failure to refer for a genetics consultation, and failure to provide accurate information on risk and recurrence all invite litigation.

**Laboratory interactions** Errors by the office clinician abound in this area. They include failure to obtain the result of a lab test, to provide timely communication, to order the correct test, and to advise the lab about the specific indication for the test (such as a subtle translocation) and misinterpretation of a test result.

**Group practice** brings with it much benefit but significant risk, too. Assuming that one of your colleagues checked a lab result or communicated important information, without you—personally—having checked the record or obtained the result, can lead to catastrophe. Exercise great care, therefore, in overseeing how lab reports return to the office; who sees them; how, and by whom, they are filed; who is informed, and how, about an abnormal report; and what type of documentation the practice insists upon. Suffice it to say: This territory is replete with litigation.

**Knowledge of practical genetics** requires your continual learning. Failure to realize (or determine) that a disorder or defect is genetic inevitably raises problems. A lack of awareness of basic advances in medicine—such as preconception folic acid supplementation that provides 70% protection against a neural tube defect—may come to light only after a child is born with spina bifida.

### Seek support in your work

Obstetrical practice is exciting and gratifying, but it is ever more challenging in light of the dramatic and continuing advances in human genetics. It is impossible for a busy clinician to keep up with the massive torrent of new information on genetic disease. When possible, establishing linkage—even by telephone—with a clinical genetics group could provide you with the opportunity to practice within the standard of expected care, and without unnecessary anxiety.

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arouse intense emotions on both sides of the abortion debate. The determination of damages is equally contentious. Among the issues that may arise at the time of trial:

- How much is a parent harmed by the birth of a child with hereditary disease?
- Should damages be offset by the benefit of having any child, even if the child is not “normal”?
- How do you measure the value of a life with Down syndrome, for example, compared with no life at all?
- Should damages include the cost of raising the child, including additional medical expenses, which can be substantial?

These and many other serious questions have faced judges and legislators for more than 30 years, and are expected to remain controversial for the foreseeable future.

## Assigning blame for genetic disease is difficult

The arguments in cases involving genetic testing often differ from those made when a fetus sustains injury. In the latter, the injury may be caused by the provider—e.g., after administration of a cytotoxic drug during pregnancy or because of induction of labor at a seriously miscalculated gestational age. Legal actions associated with such injury are widely accepted.

In cases involving genetic counseling, however, the disease is not caused by the practitioner. Rather, the plaintiff alleges that the affected child was born because the provider failed to:

- properly counsel the patient not to conceive
- offer the parents the option of terminating the pregnancy or
- successfully prevent or terminate a pregnancy.

At present, quite a few states allow parents to proceed when the action is

### FAST TRACK

**Never assume that one of your colleagues checked a lab result or communicated critical information without verifying the facts yourself**

for what most courts and commentators call “wrongful conception”—i.e., when negligent preconception advice, or a lack of advice, leads to conception and birth of a normal, but unplanned child (this also applies to negligent contraception and sterilization procedures). Actions for wrongful birth are not as widely accepted. Some courts have allowed recovery of some damages for these actions, whereas others have prohibited them, based on state common law principles or existing statutes. And actions by the child—so-called wrongful life actions—are allowed in only a very few states (California, Washington, and New Jersey), and damages are limited even more strictly.<sup>4</sup>

It would be unwise to base one’s conduct on existing law, which is unpredictable. The law can change if a judge determines that a prior opinion was incorrect, a statute was unconstitutional, or a particular injury justifies compensation. Legislative bodies also can change with an election, and the statutes can be amended. The best strategy for you to deal with this uncertainty? Provide comprehensive prenatal care of the highest quality.

### Lessons learned

Among the lessons to be gleaned from these cases are:

- Be sure that you understand the limits of any test that you order, and convey those limits to the patient.
- When you inform a patient about a planned test, that test should be completed. If it is not, explain the failure to perform the test (or the lost result, etc.) to the patient so that she can act accordingly. In that regard, a laboratory or other facility can be held liable for a negligently performed test or, as was the case in one lawsuit, a sperm bank may be liable for failing to inform the recipient of donor sperm that the donor had a positive family history

of renal disease (i.e., autosomal-dominant polycystic kidney disease).<sup>5</sup>

- Be aware that, as the amount of information available in the popular literature and on the Web expands, patients will come to question their care more often. An example: A patient recently brought an action against her health-care provider because vitamins containing folate were discontinued shortly before she became pregnant, and the child was born with a neural tube defect.<sup>6</sup>
- Most court cases, including those cited in this article, are based on the credibility of witnesses recollecting events. Well-documented, legible records written contemporaneously with an event are of enormous benefit in cases involving sometimes widely divergent recollections. When health care meets appropriate standards and is well documented in the records, most plaintiffs’ attorneys will not undertake what can be protracted and expensive litigation. When they do, the health-care provider usually prevails.

The bottom line? You are more obligated than ever to continue your medical education—not so much to avoid lawsuits, although that is certainly a possible benefit—but to ensure that the care you provide remains current, injury is avoided, and the patient’s concerns are properly addressed. ■

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4. A recent summary by a state supreme court reveals the position of various states on wrongful life actions: 27 states have refused to recognize actions by the child by court ruling or statute, or both, while three states do allow them. Willis v Wu, 362 SC 146 (2004), 607 SE2d 63.
5. Johnson v Superior Court, Los Angeles County, 101 Cal App 4th 869 (2002).
6. Brown v US, 462 F3d 609 (6th Cir 2006).

### FAST TRACK

**“Wrongful life” actions are allowed in only three states**