Emerging Medical Liability Theories in Genomic Medicine

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In April 2003, the National Human Genome Research Institute, the Department of Energy and the International Human Genome Sequencing Consortium successfully completed the Human Genome Project (HGP). In the years following the HGP's successful completion, technological innovation in genomic science continues, creating capabilities to more accurately and efficiently identify human genetic variation. This innovation has led to broader use of genomic technology in traditional clinical treatment settings, while also leading patients to direct-toconsumer genetic laboratory kits. While it is widely believed that genomic technology will allow for more precise and efficient patient care, there are corresponding concerns about how the rapid advancements in genomics, coupled with consumer expectations about the use of genomics in their treatment, could result in an "onslaught of expensive malpractice lawsuits against physicians in coming years," see "Study Finds Docs Could Face Greater Malpractice Risk in Personalized Rx Era," by Turna Ray, Genomeweb (June 27, 2011).

The liability concerns for Pennsylvania health care providers regarding genomic testing technologies are varied. This article addresses the medical practices and specialties that are most affected by the advances in genetic testing technologies and primary drivers of observed genetic malpractice suits. It then examines potential emerging and novel areas of liability in genomic medicine before concluding with a brief section addressing practical tips.

COMMON USES OF GENOMIC TESTING TECHNOLOGY

A study published last year in the Food and Drug Law Journal identified and quantified categories of genomic testing giving rise to claims in malpractice litigation, see "Genomic Malpractice: An Emerging Tide or Gentle Ripple?" by Gary E. Marchant and Rachel A. Lindor, 73 Food and Drug L.J. 1 (2018). As a part of a comprehensive legal research study, Marchant and Lindor identified 202 medical malpractice cases. Within them, they found that the cases fell into the following medical disciplines:

- Prenatal: 125 cases.
- Newborn: 16 cases.
- Diagnosis: 22 cases.
- Susceptibility: 21 cases.
- Pharmacogenomics: 12 cases.

The large number of prenatal care genomic malpractice cases relative to the total number of cases makes it worth noting a few procedures where genetic testing is used. In the prenatal care context, noninvasive genetic testing collects the mother's blood to test for an abnormal number of chromosomes in fetal DNA found in the

blood. One form of this noninvasive testing—termed "sequential screening"—is able to detect Down syndrome (trisomy 21) and Edwards' syndrome (trisomy 18) at 11 to 13 weeks with 80 percent accuracy, see "Prenatal Genetic Diagnosis and Screening Services," Penn Medicine. Another form of prenatal genetic testing is called "cell-free DNA." Cell-free DNA is commonly used for pregnant women who have a high baseline risk (women over 35 or history of a baby with a genetic disorder).

There are other forms of genomic testing with which medical liability lawyers should be familiar. One is called pharmacogenomic testing, a recent technology involving "testing for inherited genomic variations affecting drug metabolism that can be used to predict individualized responses to medications and to prevent adverse drug reactions through individualized dosing regimens of certain medications." Another emerging technology in the genomic space is "predisposition testing." Predisposition testing allows providers to evaluate a patient for genes that predispose a patient to a particular condition. One specific application of predisposition testing has been in diagnosing the BRCA1 and BRCA2 gene variants in women, which, if present, increase a woman's risk of breast and ovarian cancer, see "BRCA Gene Test for Breast and Ovarian Cancer," Mayo Clinic, (Jan. 25, 2019).

LIABILITY RISKS IN PENNSYLVANIA

Reported cases suggest that the advent and acceptance of prenatal genetic testing technologies present fewer liability risks in Pennsylvania when compared to other state jurisdictions. The primary reason for this is that Pennsylvania completely proscribes both wrongful life and wrongful birth causes of action by statute (see 42 Pa. Stat. and Cons. Stat. Ann. Section 8305; as in Sernovitz v. Dershaw, 127 A.3d 783 (Pa. 2015)). This means a provider who fails to properly interpret a genetic test cannot be sued for medical malpractice when the claim is that, "but for" the diagnosis, the fetus would have been aborted. Thus, the most robust use of genetic testing, namely in prenatal care, has little bearing on medical liability in Pennsylvania as the statute bars these claims. But see *Ginsberg v. Quest Diagnostics*, 117 A.3d 200 (N.J. Super. 2015) (allowing wrongfulbirth and limited wrongful-life claims in New Jersey).

Yet, as observed, prenatal care is not the only form of genomic testing commonly used by providers. Within viable claims against providers stemming from genomic testing, according to Marchant and Lindor, five types of errors have been observed: failure to diagnose; failure to interpret; failure to return; failure to offer; and failure to treat.

The first three errors are fairly self-explanatory. Failure to diagnose is seen when a physician fails to use genomic testing to diagnose a genetic condition that results in an adverse outcome, as in Humana of *Kentucky v. McKee*, 834 S.W.2d 711, 719 (Ky. 1992) (finding hospital could be found negligent for failing to diagnose phenylketonuria). Failure to interpret occurs when a physician fails to properly read a genomic test and does not properly advise the patient on the results of the test. Failure to return is seen when a physician fails to issue test results to patients.

Failure to offer is seen when a genetic test is appropriate, but the physician fails to advise the patient about the test. There are some important notes in this regard. First, at least one court has found that the cost of a

procedure is not a defense for failing to advise a patient regarding the existence of a potentially beneficial genetic test, as in *Downey v. Dunnington*, 895 N.E.2d 271, 276 (Ill. App. 2008) (holding doctor had duty to recommend genetic testing for BRCA1 and BRCA2 mutations before prophylactic surgery). Second, while informed consent claims are not actionable for failure to offer (see, e.g., *Reed v. Campagnolo*, 630 A.2d 1145 (Md. 1993)), physicians should always document that a patient has been offered a particular genetic treatment (regardless of cost) and ensure the patient has signed an informed consent agreement indicating genomic testing has been offered.

Finally, failure to treat claims encompass situations where the genetic disorder is properly identified but not properly treated. A case based in this theory will largely proceed as a traditional medical negligence claim because the genetic testing component is presumably not the alleged source of negligence.

NOVEL FORMS OF LIABILITY

One novel claim in the context of genomic testing is a provider's duty of care to a third party regarding an adverse genetic condition. In *Polaski v. Whitson*, 49 Pa. D. & C.5th 73 (Lehigh C.P. 2015), a physician performed a 12-lead electrocardiogram as part of a comprehensive physical for Raymond James Polaski in 2010. Following this treatment, Polaski died as a result of hypertropic cardiomyopathy (HCM), a complex type of heart disease affecting the heart muscle. The physician did not diagnose Polaski as having HCM because he did not conduct a followup of the electrocardiogram with a genetic test that would have discovered this condition. Two years later, Polaski's son, Joseph Polaski, died as a result of HCM.

Joseph Polaski's estate sued his father's doctor, claiming the doctor owed Joseph a duty to warn of the HCM genetic heart condition, and that Raymond Polaski's doctor was negligent by failing to conduct the genetic test on his father. Raymond Polaski's doctor moved for summary judgment. The court denied the doctor's motion and found that a doctor may, in fact, hold a duty to a third-person for failing to perform a genetic test.

In another form of liability, cases from other jurisdictions have held a physician can be liable for negligent selection of a laboratory that provides substandard analysis of genetic material or fails to interpret test results, as in *Berman v. Laboratory Corporation of America*, 268 P.3d 68, (Okla. 2011). Pennsylvania courts would likely adopt the logic of sister state jurisdictions if presented with this issue.

OUTLOOK AND PRACTICAL TIPS

Health care provider liability risk related to conducting-or failing to conduct-genetic testing of patients is very much an evolving area of the law. Pennsylvania courts deciding the initial wave of cases on this topic will provide insight on best preventative practices. Nevertheless, providers may be able to limit exposure in a few respects. One, providers will benefit from staying up to date on routine genomic technologies, which will allow them to appropriately recommend, conduct and refer for testing. Second, providers must ensure they understand how to interpret genomic testing, and training programs on how to interpret should be used. Third, providers should offer useful genomic testing and document that genomic tests have been offered. Finally, providers should consider consults with genomic counselors. While this option may not be

available for all providers, especially outside of major academic centers, it nonetheless may be good practice to provide the patient with the option, even if the counselor is not readily available.

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