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Lee Black

Jaques Simard

Bartha Maria Knoppers

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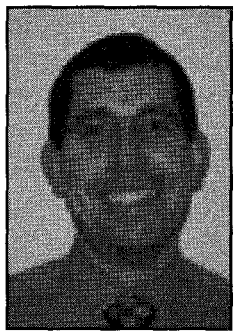
Genetic Testing, Physicians and the Law: Will the Tortoise Ever Catch Up with the Hare?

Lee Black, J.D., LL.M.,[*]
Jacques Simard, Ph.D.,[**]
Bartha Maria Knoppers, Ph.D.[***]



As science progresses and new treatments for (and genetic susceptibility to) disease are discovered, questions arise as to how physicians relate this information to patients. This is especially important in the field of cancer research, where the study of genetics has changed how researchers think about causes and therapies. The use of genetic testing to identify specific mutations known to increase risk of cancer now allows patients to know of their potential risk and to be proactive with prevention decisions. However, the rapid increase in the use of genetic testing creates difficulty for physicians who must counsel patients on whether to take genetic tests and must then explain what the results mean. Complicating this already complex situation is the desire of patients to receive testing based upon their own perceived risks rather than medically determined risks. As genetic tests become cheaper and more widespread and as potential patients continue to hear and read sensational stories about genetic disorders and testing, the question becomes: Will physicians face new liabilities based upon inaccurate or misleading results, or for disclosing (or failing to disclose) relevant results to family members of a patient.

I. THE GENETIC HARE

There is no doubt that the field of medical genetics is changing rapidly.

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[**] Cancer Genomics Laboratory, Centre Hospitalier Universitaire de Québec and Laval University, Quebec, Canada, Canada Research Chair in Oncogenetics.
[***] Director, Centre of Genomics and Policy, McGill University, Montreal, Quebec, Canada.

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However, science can be slow to follow up on early promises. In the early 1990s, scientists, along with the media, trumpeted the mapping of the human genome and its impact on medical science. It was expected that, within decades, the genetic foundation of susceptibility for a plethora of diseases would be discovered. While it is true that there have been many important discoveries—including that of the BRCA 1 and BRCA 2 (standing for BReast CAncer) genes and their mutations (hereinafter BRCA mutations)—two decades after the initiation of the Human Genome Project, much of the early promise remains unfulfilled.

We are left with the general knowledge that our genes might be the cause of, or at least a contributor to, many diseases, but we have no specific knowledge of what these genes and mutations are. In the field of hereditary breast and ovarian cancer (HBOC), some mutations, such as the BRCA mutations, are known, but researchers believe that many others remain to be discovered and that known mutations must be studied further to determine their effect on cancer development. Regardless, genetics and 'personalized medicine' (the use of an individual's genotype to guide treatment decisions) are regarded as the future of health care. The question remains whether physicians, their patients and the law are prepared for this future.

II. PHYSICIANS AND PATIENTS

The fields of genetics and genetic testing have exposed problems in how physicians communicate with patients. To begin, although there are guidelines and educational resources available for both, many physicians are not adequately trained to understand the consequences of specific genetic variations or to relay reliable information back to patients.[1] Indeed, it was recently noted that "only 11% of US and Canadian medical schools reported practical training in the use of medical genetics as part of their curricula."[2] As genetic testing becomes more commonplace, this lack of training will likely result in a misleading interpretation of results that will detrimentally affect both physicians and patients alike.

Secondly, although the general public's knowledge of the technical aspects and risk of genomics and genetic testing are low, their attitude is generally positive, which could lead to an increase in requests for testing without sufficient comprehension of what tests actually entail. Although obtaining a blood or cell sample is relatively low-risk, there are issues of

1. Keyan Salari, *The Dawning Era of Personalized Medicine Exposes a Gap in Medical Education*, 6 PLOS MED. (2009), *available at* http://www.plosmedicine.org/article/info%3Adoi%2F10.1371%2Fjournal.pmed.1000138; Joan L Bottorff et al., *The Educational Needs and Professional Roles of Canadian Physicians and Nurses Regarding Genetic Testing and Adult Onset Hereditary Disease*, 8 CMTY. GENET. 80 (2005).

2. *Id.*

privacy, consent, risk communication and possible psychosocial consequences that accompany genetic results. A positive result could lead to anxiety (whether the patient will develop the disease and when it could happen, etc.), as well as the recommendation in some instances for prophylactic surgery, such as a mastectomy or oophorectomy (removal of the ovaries), to lessen the risk of breast or ovarian cancer. If the result is either negative or non-conclusive, patients must be made to understand that it does not necessarily mean that there is no potential genetic source for cancer. An inappropriate interpretation of the risk assessment could lead to unnecessary mental anguish or surgery or could cause a physician or patient to fail to consider additional genetic risks. Legal action toward a physician might follow.

Finally, a physician might believe it necessary to inform a patient's relative of the result, if the patient refuses to inform a relative whose health might be implicated. Siblings, parents, children, cousins and other family members could be at risk of carrying the same mutation as the patient; for example, children of a BRCA 1 or 2 mutation carrier have a fifty percent chance of inheriting the same mutation. As genetic information is generally relevant to more than one person, the question for physicians is whether they have an obligation, or even the ability, to inform these third parties of potential danger.[3]

III. RISK COMMUNICATION AND LIABILITY

One challenge for physicians in the environment of genetic medicine is their ability to effectively communicate risk information to patients and others. Post-test communication of risk raises a number of potential liability issues for physicians. Recommending particular action, or inaction in the case of negative results, could lead patients to take drastic steps, such as prophylactic mastectomies for example, that would never be recommended for non-carriers. Similarly, playing down potential and unknown genetic risk in response to a negative test, especially for patients with a strong family history of breast cancer, could cause inattentiveness to other warning signs. Physicians could certainly be faced with liability in these circumstances under current law.[4]

Another area with potential consequences for physicians is disclosure of test results or genetic risk to family members of a patient who refuses to inform family members of the results. There are precedents for breaching

3. Mireille Lacroix et al., *Should Physicians Warn Patients' Relatives of Genetic Risks?*, 178 CAN. MED. ASSOC. J. 593 (2008).

4. Kathleen A. Mahoney, *Malpractice Claims Resulting from Negligent Preconception Genetic Testing: Do These Claims Present a Strain of Wrongful Birth or Wrongful Conception, and Does the Categorization Even Matter?*, 39 SUFFOLK U. L. REV. 773 (2006).

the otherwise sacrosanct confidentiality of medical information in both law and ethics. Ethically, physicians can disclose patient information for the protection of third parties in limited circumstances, or where required by law, such as for public health. Legally, courts have accepted an *obligation*, not just an option, to disclose confidential information when a third party is in imminent danger (the *Tarasoff* rule).[5] The question, however, is how well these exceptions to confidentiality apply to genetic information. Two American cases often cited regarding physician disclosure of genetic risk to family members have contrary results, with one holding that telling patients of the implication for their family is sufficient and the other requiring disclosure directly to family members.[6] Unfortunately, the law has not kept up with scientific advances and the unique legal questions that are raised within the field of genetics and genetic testing.

IV. THE SPEED OF TECHNOLOGY AND THE LEISURELY PACE OF LAW

Presently, there is little cohesive (or even coherent) legal guidance for physicians faced with communicating the results of genetic tests. Many of the current legal doctrines that ordinarily apply to medical issues miss important aspects of genetics or are even entirely inapplicable. The *Tarasoff* rule for breaching confidentiality, sometimes posited as a solution for communicating genetic risk to patients' family members, is inadequate.[7] Genetic risk is based on an inherent feature of a third party rather than a danger controlled or caused by the patient. Furthermore, science has not yet advanced far enough to quantify exact risks represented by genetic variations associated with increased cancer susceptibility; rather, risks are presented as a range of percentages. How can we determine the level of danger posed and whether that is sufficient to permit or require breaching confidentiality? Alternatively, the information gleaned from a genetic test is considered by many to be familial, so why shouldn't a physician be permitted to share it with others who might also have a need to know?

Statutory laws protecting medical information also fail to consider the context of genetics. For instance, the privacy rules of the Health Insurance Portability and Accountability Act (HIPAA) focus on the level of the potential danger to the third party and the imminence of the threat. This

5. Tarasoff v. Regents of the University of California, 551 P.2d 334 (Cal. 1976). Canada has adopted a similar rule, although it is more permissive than obligatory: the danger must clear, serious and imminent. However, the danger can be to an identifiable group *or* a specific individual. *Smith v. Jones*, [1999] S.C.R. 455.

6. Pate v. Threlkel, 661 So.2d 278 (Fla. 1995); Safer v. Estate of Pack, 677 A.2d 1188 (N.J. Super. Ct. App. Div. 1996).

7. The same is true in Canadian law. In the Province of Quebec, which is a civil law regime, the *Code of Ethics of Physicians* sets forth similar requirements for when confidentiality can be breached in instances of danger to third parties.

type of narrow focus is common in rules limiting disclosure but does not take into account the nature of the information being protected—in the case of genetic risk information, the fact that it applies to more than one person. In addition, if the results of a test were shared with a third party such as an insurer, would such a disclosure violate the rights of the parents of a patient, at least one of whom would be implicated as being a carrier of the same genetic mutation?

In addition, legal rules for liability based on failure to test, faulty results, or other issues leading to injury may be difficult to apply when test-based recommendations and interpretations change with new discoveries. Medical science rapidly shifts and common practice can quickly change, as illustrated by the recent modifications to mammography screening recommendations issued by the US Preventive Services Task Force.[8] Will physicians be faced with lawsuits for *not* following the previous recommendations, or will patients who undergo surgical procedures based on perceived risks seek liability when recommendations are later shown to be incorrect or inaccurate? Current medical malpractice law does not always address new science and is even more problematic when dealing with unknown or limited risks—lawyers like absolutes, but genetics is rarely decisive.

These issues have been discussed in the medical and legal spheres for decades, and yet most remain unresolved. The slow legal response to these important questions—and the failure of many physicians and most lawyers to understand the intricacies of genetic medicine—has left a black hole in the law.

V. IT IS TIME FOR SOME ACCELERATION

The current absence of guidance need not continue. There is much that can and should be done in both medicine and law. The field of genetics is changing how physicians ought to be incorporating genomics into their practices. For one, genetic education for physicians and other health professionals should become the standard, at least for those who might regularly come into contact with genetic issues. Genetic education should include both the science of genetics as well as the legal issues surrounding genetic information. Medical student education has already made advances in this direction, but older physicians should be offered additional opportunities to gain this knowledge. This is an essential first step—it will be much easier for physicians to communicate risk to patients and their families if they fully understand the implications of genetic information.

Physician education is only a small part of the solution. As those who

8. http://www.ahrq.gov/clinic/uspstf09/breastcancer/brcanrs.htm.

create law and policy, attorneys, judges, and legislators must also become better educated in the science of genetics and the consequences of related laws and policies. Lawmakers have made some strides regarding genetic information, such as the enactment of the Genetic Information Nondiscrimination Act of 2008. However, the focus on potential genetic discrimination by insurers and employers has come at the cost of other important issues in genetics, and general comfort with the status quo is not appropriate when it comes to genetics. As noted above, neither medical malpractice law nor the law of confidentiality (privacy *and* disclosure laws) are particularly well-suited to the arena of genetics. To continue with a system that reacts without sufficient understanding is not tenable, especially when scientific discoveries will continue exponentially regardless of the law.

Better understanding of communication issues regarding genetic risk by both physicians and the legal community will, in the end, help patients. The physician who recognizes when a patient should have genetic testing and a legal system that considers the information needs of patients and their relatives, as well as the intricacies of genetic medicine, would much better serve us than the present system. However, the current absence of legal guidance in many areas of potential liability has created roadblocks to effective education and communication, both between health professionals and between health professionals and patients. Considering that genetics has been and continues to be dubbed the future of medicine, it is vital that the tortoise that is the law catch up with these expectations.