Direct to Consumer Genetic Testing: Think Before You Spit

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The combination of completion of the human genome project,¹ rapid advances in genetic testing and gene sequencing technology, and the internet has now allowed genetic screening to move out of the physician’s office and into the commercial marketplace.² Some direct-to-consumer marketing of genetic testing is no longer “medical” in several ways: it is not administered under the guidance of physicians; it is invariably unaccompanied by any professional counseling as to what the sequencing data means, and it is no longer promoted only by trained health care personnel or medical institutions.³

Taking genetic testing public has also resulted in genetic testing no longer being confined to the simple evaluation of serum or amniotic fluid for specific chromosomal abnormalities in order to detect the presence of a specific disease such as Tay-Sachs disease.⁴ Nor is the goal confined to the screening of higher-risk patient populations to determine whether there is a greater chance of developing a malignancy which might be prevented with early intervention.⁵ The latest commercial trend is the genetic “spit party.”⁶ For a fee ranging from $400 to $2500 or more, apparently healthy individuals can provide a sample of their saliva to a sponsor⁷ who then sends the fluid to a commercial laboratory to test the entire genome for thousands of variants of select portions of genetic material (known as “snips”). The goal is as much social as medical. Many participants are not simply interested in whether they have a “predisposition” to, or elevated risk for, developing any of a number of major diseases such as Parkinson’s disease, Huntington’s chorea or coronary artery disease.⁸ Just as important is a “determination” of one’s genetic risk for more benign social traits such as baldness or intelligence, or a predisposition to depression.⁹ And getting this genetic information is as

¹ Elizabeth Pennisi, Human Genome: Reaching Their Goal Early, Sequencing Labs Celebrate, 300 SCIENCE 409 (2003).
⁴ The American College of Obstetricians and Gynecologists, ACOG Committee Opinion, Prenatal and Preconceptional Carrier Screening for Genetic Diseases in Individuals of Eastern European Jewish Descent, Number 298, August 2004.
⁷ There are three for-profit non-medical companies offering this service at the moment in the U.S. They are 23andMe, Decode Genetics, and Navigenetics. None of the activities of these companies is regulated by FDA.
⁸ Schulman, supra note 6.
simple as logging on to one’s account at the start-up company acting as your genetic custodian.\textsuperscript{10}

Of course, the genetic information you get is only what you pay for. At the present time a full sequence of one’s own genetic code is a luxury item costing hundreds of thousands of dollars,\textsuperscript{11} and thus far only four complete human genome sequences have been announced by scientists around the world.\textsuperscript{12} A group of scientists at Harvard, the “PGP 10,” have agreed to forfeit privacy concerns and allow their entire genomes to be posted publicly,\textsuperscript{13} for the sake of research and to encourage others to do so once the price falls to an acceptable level. Importantly, “prospective participants are advised to consult with first-degree relatives but, except for identical twins, their consent is not required.”\textsuperscript{14}

It’s all a new business, well on its way to becoming a big business. Unfortunately, the commercial marketing is more targeted that the genetic testing being done.\textsuperscript{15} The lack of genetic counseling accompanying these commercial non-medical activities is particularly worrisome, but may be less of an issue because consumers of these services are interested in both social and medical information. Nevertheless, not enough people are thinking about whether the genetic information they are receiving is accurate enough in any meaningful sense to be truly predictive of the “trait” they are looking for.\textsuperscript{16} Worse still, there are serious questions remaining about where their genetic information could end up and who actually owns their genetic data. The long-term consequences of such testing, whether additional, unanticipated and unwanted testing will pop up, or whether this information should be treated differently than other medical information\textsuperscript{17} are also glossed over or remain unanswered by the individuals or organizations sponsoring these “spit parties.”

Unfortunately, the business side of direct to consumer genetic testing is not just confined to internet-based start-ups. Even academic medical centers may have potential conflicts of interest of which patients may not be aware. For example, Baylor College of Medicine in Houston, Texas has an ongoing prenatal genomic testing program offering extensive screening array of fetal DNA fragments.\textsuperscript{18} The medical school derives revenue from this genetic testing, and also owns Spectral Genomics, a company which markets arrays to the

\textsuperscript{10} Id. “Logging onto my account at 23andMe, the start-up company that is now my genetic custodian, I typed my search into the ‘Genome Explorer’ and hit return. I was, in essence, Googling my own DNA.”
\textsuperscript{12} Id.
\textsuperscript{13} Id.
\textsuperscript{14} Id.
\textsuperscript{17} Mark A. Rothstein, Genetic Exceptionalism and Legislative Pragmatism, 35(2) J. L. MED. ETHICS 59 (2007).
\textsuperscript{18} Bridget M. Kuehn, Prenatal Genome Testing Sparks Debate, 300 JAMA 1637 (2008).
public similar to those Baylor uses in its research.\textsuperscript{19} Even though an academic institution such as Baylor has the enormous advantage of being able to provide expert genetic counseling to patients, on a more clinical level it is not clear whether consumers understand that testing often identifies variants of unknown clinical significance, and that many of the potentially harmful variants have not been well-studied and are rare.\textsuperscript{20} Prenatal testing of an unborn child’s genome from fetal blood obtained either by amniocentesis or chorionic villus sampling may result in unnecessary termination of pregnancies by anxious parents lacking skilled genetic counseling to interpret ambiguous genetic information.\textsuperscript{21} Statements that “95% of the time you are dealing with pretty black-and-white information” will not be reassuring to anxious parents vulnerable to fears that the genetic information “might” indicate a serious condition even if it is explained to them that the information is of questionable clinical significance.\textsuperscript{22}

One could seriously question the ethics of testing for harmful or potentially harmful genetic variants in a clinical, as opposed to purely research, setting when physicians have no substantive information about the clinical prognosis of unborn children with such variants. Bio-ethicists and health law attorneys are understandably concerned about all of this and other problems of genetic screening.\textsuperscript{23} The fact is that there are serious legal, regulatory, and quality control issues which desperately need to be addressed.\textsuperscript{24}

\textbf{Regulatory Concerns}

Direct to consumer advertising of genetic testing (BRCA analysis) for breast and ovarian cancer risk by established medical genetics companies such as Myriad Laboratories in Salt Lake City has already resulted in subpoenas for information from the company\textsuperscript{25} and concerns that a test useful for less than 5% of the female population of the U.S. is being fraudulently promoted to millions of women for whom the test has little or no possible value.\textsuperscript{26} These issues surfaced despite the fact that: (1) much of the advertising and promotional material by this company for BRCA gene cancer risk testing is clearly educational in nature; (2) Myriad is a scientific company which has some of the most renowned genetic testing scientists in the United States on its staff; and (3) their sequencing equipment is state of the art and their staff, facilities, and equipment testing information are all readily accessible in the public domain.

\textsuperscript{19} Id.  
\textsuperscript{20} Id.  
\textsuperscript{21} Id.  
\textsuperscript{22} Id.  
\textsuperscript{26} Id.
For non-scientific genetic testing companies such as 23andMe who simply have customers interested in the chance to decode their genes, the problems of validating the quality of the genetic testing equipment, the qualifications of the scientists they employ, and the accuracy of the test results themselves are much greater. Also, the quality of the genetic information given to consumers is a much greater unknown.

Additionally, because private genetic testing kits such as those being promoted directly to consumers by companies such as 23andMe are only manufactured and used exclusively “in house,” they are not directly regulated by the United States Food and Drug Administration (FDA). Because these tests are not marketed commercially and never leave the laboratory where they are manufactured, they are classified as “home brew” diagnostic tests. Though approved for marketing by FDA as class I medical devices, FDA lacks direct regulatory control over activities such as determinations of safety, efficacy, accuracy/reproducibility of genetic testing, as well as quality control and standardized good manufacturing practice in order to protect consumers. This “home brew” exception constitutes a large hole in the federal regulatory matrix for genetic testing devices. “Home brews” are regulated by the Department of Health and Human Services under the 1988 Clinical Laboratory Improvement Amendments (CLIA).

Significant concerns have been voiced about the ability of CLIA to adequately monitor laboratories performing genetic testing, ensure that the test results are accurate enough and that the testing equipment reliable enough. A Congressional bill to transfer regulatory oversight over such laboratory facilities to the FDA was introduced by Senators Kennedy and Obama in 2006, but the bill failed to pass. In theory, FDA might be better suited than the Center for Medicare and Medicaid Services (CMS) to regulate all of the issues involved in genetic testing since it already regulates commercial testing devices and kits as medical devices, and has more medical staff and experience. Unfortunately, there is no way currently for consumers to know the accuracy of any of the genetic information they are paying significant sums of money to obtain.

Where Things Are Headed

The passage of the Genetic Information Nondiscrimination Act of 2008 (GINA) will hopefully ensure that individual genetic information, however obtained, will not be used

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27 Schulman, supra note 5.
28 The term “home brew” refers to reagents which are manufactured and used within the same facility yet made available to the public. These companies use their own reagents, and use them only in-house, and in the past used to sell the testing service to primary care physicians though this has changed in the past several years.
29 CLINICAL LABORATORY IMPROVEMENT AMENDMENTS (CLIA), PUBLIC LAW 100-578 (1988).
by insurers and employers in health insurance and employment decisions. This legislation goes a long way to protecting against some corporate abuse of otherwise private personal genetic information, but there are significant limitations on the protections GINA offers. GINA does not apply to life insurance, disability insurance, long-term care insurance, or other uses of genetic information.\footnote{Id.} Nor do GINA’s health insurance provisions apply to people who are symptomatic or to “non-genetic” predictive testing and information (also known as epigenetics).\footnote{Id.} None of the quality assurance and scientific legitimacy issues posed by the commercialization of genetic screening are addressed, or solved, by this piece of legislation either. The lack of any meaningful quality control over home-brew genetic testing remains a fairly large hole in the federal regulatory matrix governing medical products and protecting consumers even if some, but not all, of the consumer concerns about the privacy of their genetic information are met by GINA.