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Genetic Testing and Counseling Primer for Elder Law and Special Needs Planning Attorneys

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I. Introduction

As Elder Law attorneys know, it is common to receive the following response when introducing themselves as Elder Law attorneys: “Oh, uh huh … what’s that?” However, over the past decade, this response has increasingly come to be replaced with the following response: “Oh good; I know someone who might need your help.”

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Similarly, genetic counselors usually need to explain what they do. The field of genetic counseling got its start in the 1970s. However, until recently, genetic testing was not readily available to the general population, and the profession remains somewhat unknown.

Looking back, both Elder Law and genetic counseling reflect the same development: advances in biology and medical science. In the bad old days, people generally got old, got sick, and died. Now, it is much more common for people to get old, get sick, and stay alive. Living longer typically involves new arrangements to handle various kinds of disability, dependency, and special medical care, hence the growing need for Elder Law attorneys. Meanwhile, stunning advances have been made in the science of genetics. Genetics is no longer only a topic for biology courses and science magazines; it can be used to great effect in individuals. Yet it is often complex to interpret, hence the need for genetic counselors.

Elder Law attorneys will find it useful to have some background in genetic testing: what it can and cannot do (at least as of now). Clients will likely increasingly mention or ask about genetic testing and expect their lawyers to have some familiarity with it. Elder law attorneys will sometimes tell their clients, “If you can tell me when you are going to get sick and when you are going to die, I can do perfect planning.” Clients are amused by this because it is obvious that no such information is available. However, genetic testing holds the promise of at least some predictability about disability and death and therefore an improvement in planning choices.

This article is not intended to be a scholarly presentation of any particular aspect of genetic testing. It is instead intended to provide some background on genetic testing and an overview of some of its more interesting implications. The discussion is confined largely to medical matters. The use of genetic tests has, of course, revolutionized criminal law, paternity suits, and anthropology, but this article will not attempt to cover these applications. Instead, it will focus on the public policy issues involving access to genetic testing.

II. Some Historical Context

Fifteen years ago, genetics received a big boost from a scientific race between the federal government and the private individual — and the private individual won. Craig Venter was the first person to sequence the human genome. He beat out the Human Genome Project, lead by Francis Collins, director of the National Institutes of Health. The Human Genome project was a huge international scientific research program and the world's largest collaborative biological effort. Venter, feeling that the big international effort was moving too slowly, used a controversial sequencing technique to reach his goal. In 2000, Venter and Collins jointly announced that Venter's company, Celera, and the Human Genome Project had both succeeded in mapping the human genome. This was three years before the completion of the Human Genome Project which took 13 years and cost almost $3 billion. A few years later,
in 2007, the first complete 6-billion-letter genome of an individual was published — using Craig Venter’s DNA. Most interestingly, it was found that a number of the sequences of the genome could be associated with such things as disease risk, physical traits, and even social behavior. These discoveries opened the door to the use of a person’s DNA in individualized medicine via genotyping.

Genotyping is the process of determining differences in genetic makeup of an individual by examining his or her DNA sequence. This sequence is then compared with a reference sequence to determine whether there are any mutations or changes in the genetic sequence that may be harmful. Mutations in the sequence or a change in the number of copies of a segment of DNA typically indicates an increased risk for disease. Mutations are often inherited from a parent, but they can also be de novo (occurring in a particular individual for the first time and not inherited). It is estimated that we all carry dozens, if not hundreds, of potentially injurious mutations, with at least a few that are classified as high risk. It is likely that some of these mutations are counterbalanced by either other genes or the environment (so-called “complex traits”). This is why not every individual with such mutations expresses an associated disease.

III. Three Uses of Genetic Testing

Genetic testing in the medical field (excluding pharmacological applications) can be loosely categorized into three applications: disease diagnosis, carrier status, and predictive testing.

A. Disease Diagnosis

The use of genetic testing as a diagnostic tool is most common when a child demonstrates medical or physical features of a genetically based condition. Genetic testing in this case is used to confirm or clarify a diagnosis. Diagnostic genetic testing in both children and adults can also be used to identify the cause of medical and/or physical problems when the cause is not readily apparent. Genetic testing for this purpose can involve the use of a wide panel of multiple genes. It may even involve whole exome or whole genome sequencing. Such diagnostic genetic testing may help physicians determine appropriate treatment and can often inform parents of the likelihood of a condition occurring in a future child.

B. Carrier Status

A second application of genetic testing is determining the carrier status of expectant couples and those planning a pregnancy. Carrier status testing identifies mutations carried by both parents that indicate a risk for their child to inherit two mutations and, therefore, develop a medical condition. Parents who each carry one mutation are typically not at risk, but the combination of the two in their child may cause serious medical problems. Carrier status testing is typically done by taking blood samples from the parents to identify their child’s risk for certain conditions, such as cystic fibrosis or Tay-Sachs disease.

Genetic testing during pregnancy is also performed to identify genetic conditions not


3 An exome is the part of the genome formed by exons, of which there are about 180,000 in the human genome, constituting about 1 percent of the total genome.
typically inherited, such as Down syndrome, trisomy 18, and spina bifida. This testing or screening for certain conditions can be invasive (such as amniocentesis) or noninvasive (such as drawing blood from the mother) and is typically used in tandem with ultrasound when a physical feature of a condition is identified in the fetus.

C. Predictive Testing

A third application of genetic testing is predictive testing, in which individuals are tested to determine their hereditary risk for disease, such as hereditary cancer or hereditary cardiomyopathy. Testing for Huntington's disease is an example of true predictive genetic testing. If testing determines that an individual carries a hereditary mutation in the HD gene, he or she is certain to develop Huntington's disease. The individual’s genotype therefore is a guarantee of developing the condition.

However, most hereditary mutations do not predict 100 percent risk for a condition. Instead, carrying a hereditary mutation increases the risk of developing the condition. Breast cancer risk in a woman with a BRCA1 mutation is a well-known example. A woman with a BRCA1 mutation has a higher risk of breast, ovarian, and other cancers, but she may never develop a cancer in her lifetime. Similarly with cardiomyopathy, an individual might have a hereditary predisposition but may never actually develop abnormalities of the heart muscle. These types of examples make it clear that there are other risk factors involved with developing disease. In the example of breast cancer risk with a BRCA1 mutation and cardiomyopathy risk with a MYH7 mutation, the risks are high enough that medical intervention is recommended, such as increased breast cancer screening through mammography and breast MRI or specialized cardiovascular screening tests. These tests are fairly easy to interpret given the amount of data on these genes and the specialized interventions available.

In all of these examples, a particular condition is definitely or highly likely to develop due to a single gene trait. This is Mendelian genetics (remember learning about Mendel’s peas?), in which we understand that a condition will develop because of one or two mutations in a single gene. This is in contrast to polygenetic traits, such as eye color, which are caused by several genes, and in contrast to multifactorial traits, which are caused by a combination of multiple genes and environmental effects. Multifactorial diseases include common conditions such as heart disease, stroke, dementia, and diabetes. These may cluster in families due to shared genetics, but risks for these conditions are also influenced by lifestyle factors.

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4 Trisomy 18 is a genetic condition caused by an extra number 18 chromosome. The extra chromosome leads to mental and physical defects and an extremely short lifespan.
5 Cardiomyopathy is a disease of abnormal heart muscle. Cardiomyopathy makes it harder for the heart to pump and deliver blood to the rest of the body.
6 MYH7 is a gene encoding a myosin heavy chain beta expressed primarily in the heart but also in skeletal muscles.
and a shared environment. Obviously, lifestyle factors such as smoking and diet are also relevant to disease risk and usually prevent the determination of simple one-to-one relationships between an individual’s genetic profile and disease. Finally, environmental pollutants also elevate disease risk regardless of genetic predisposition.

IV. Uncertainties in Interpretation of Test Results

Even with single gene traits, interpreting test results presents challenges. One challenge is the high likelihood of finding a change in a gene that has not yet been classified as either harmful or benign. These changes are referred to as “variants of uncertain significance.” They are typically small changes in the DNA sequencing that may or may not confer disease risk. Every individual has differences in his or her DNA as a result of his or her ethnic background. It is often difficult to determine whether a small change in the DNA sequencing is a true mutation (and therefore confers a risk of disease) or whether it is a polymorphism (a change that is common in the general population and does not typically indicate an increased risk of disease).

Yet another complicating factor is that genetic tests rarely look at the entire genome. This can be an issue when trying to understand multifactorial traits, because they by definition involve multiple genes (plus environmental effects). Some genetic tests look only at small segments of the genome that are thought to have disease significance. Many direct-to-consumer companies, such as 23andMe (discussed more later in this article), use this type of genetic testing. These single nucleotide DNA changes are aggregated to interpret the combined influence on disease risk. However, this is often not a complete assessment of genetic risk factors because all of the genes are not sequenced in their entirety and not all genes are tested. Therefore, the effect of a segment indicating a higher risk of an illness may be modified by another part of the genome that either has not been tested or is not currently understood.

Limiting the evaluation of disease risk to the results of genetic tests also risks missing a critical piece of medical information — the family. A family’s medical history — a report of the conditions or diseases family members have or have had — can offer vital information when assessing an individual’s disease risk. When a genetic test is ordered by a genetic testing provider, a family tree or pedigree is almost always taken. The family tree, in conjunction with genetic testing, allows a health care provider to assess the risk for a condition. For example, consider a 40-year-old man, “Mr. X,” who is interested in his risk for colon cancer, given that his father had colon cancer at age 55. He undergoes genetic testing without a risk assessment with a non–genetic testing provider and is told by the testing company that he has a low risk for colon cancer. He thinks he is now in the clear, when in fact his family history is still an indicator of risk because there are likely other genetic mutations involved in colon cancer risk that are not currently understood and therefore are not included in Mr. X’s genetic testing. The data gathered by looking at other families with similar cancer history indicates that Mr. X actually has about twice the risk for colon cancer as the general population or an approximately 10 percent lifetime risk. Given his family history, the National Comprehensive Cancer Network guidelines recommend earlier colon cancer screening using colonoscopy. In the case of Mr. X, this recommendation is to begin screening at age 40, or 10 years before his father’s diagnosis, instead of at age 50, the age at which it is recommended for the general population. Therefore, in this instance, Mr. X’s family history has a direct impact on his cancer risk and changes his recommended medical management, even though his genetic testing indicates a low risk for colon cancer.
In summary, other factors such as family history, lifestyle, and environment, in addition to an individual’s genotype, appear to play an important role in determining disease risk. It is difficult to determine how important these factors are in predicting disease risk when they are combined with the influence of our genetic code. As a result, even in the case of single gene disorders, the same mutation in the same family can have very different disease outcomes.

V. Specific Interventions

The fact that a person’s genes do not determine his or her destiny leaves open the possibility for interventions to lower the risks from potentially problematic genes. Interventions are possible when there are “actionable gene mutations.” These are mutations in genes that can be identified as causing or likely to cause a future illness and that the carrier can do something about. For example, one gene mutation can cause deadly blood clots, but this can be resolved by avoiding immobility for lengthy periods and/or taking anticoagulants such as warfarin (also known as Coumadin). So far, scientists have found about 200 such actionable gene mutations.

If such gene mutations can be identified early, before symptoms have been expressed, treatment is likely to be much more effective. This is the promise of “personalized medicine,” in which the treatment is targeted not only at the illness but also at the unique vulnerabilities of a particular individual.

VI. Genetic Counselors

Because the results of genetic tests are difficult to interpret, a separate medical specialty has arisen to address it: genetic counseling. The National Society of Genetic Counselors defines genetic counseling as “the process of helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease. This process integrates:

- Interpretation of family and medical histories to assess the chance of disease occurrence or recurrence.
- Education about inheritance, testing, management, prevention, resources and research.
- Counseling to promote informed choices and adaptation to the risk or condition.”

Genetic counselors are Master’s degree–level health care providers with education in human genetics and psychology. In the United States, they are certified by the American Board of Genetic Counseling. Genetic counselors work in a variety of settings, including university medical centers, private and public hospitals/medical facilities, diagnostic laboratories, pharmaceutical companies, not-for-profit organizations, and government organizations and agencies. Genetic counselors work in multiple specialty areas: prenatal medicine, cardiovascular disease, cancer, metabolic disease, neurology, pediatrics, infertility, pharmacogenetics, and genomic medicine.

Access to genetic counseling is increasing. Since 2006, the number of certified genetic counselors has increased 75 percent, with more than 4,000 practicing genetic counselors in the United States. Most genetic counselors report that they can see new patients in person.

10 Id.
within 1 or 2 weeks and that counseling can be provided by phone in some cases. Genetic counseling is typically covered by health insurance and, in some cases, the Affordable Care Act of 2010 mandates that it be covered without cost to the patient.

VII. Implications for Seniors and People with Disabilities

A. Seniors

Genetic testing is beginning to reveal information regarding susceptibilities to the diseases associated with old age: Alzheimer’s disease, Parkinson’s disease, diabetes, and cancer. Genetic test results showing a higher risk of such diseases can result in a cascade of consequences. Francis Collins, mentioned at the beginning of this article, responded to his test results thoughtfully by making lifestyle changes to reduce the probability that the increased genetic risk would be expressed in actual disease. It is important to note that, for some conditions, lifestyle factors’ influence on disease risk is understood; however, for many of the conditions that affect seniors, this influence is not yet known.

Other reactions to a high-risk test result may be more aggressive than diet and exercise changes. A well-publicized example is Angelina Jolie’s bilateral mastectomy. She was cancer-free but learned that she carries a BRCA1 mutation, which increases her lifetime risk for breast and ovarian cancer. She chose to undergo prophylactic mastectomy to reduce her breast cancer risk, whereas other women choose to increase breast cancer surveillance, such as undergoing more mammograms and breast MRIs. Both options are available to women who carry a BRCA1/2 mutation.

Will those found to be at elevated risk for more complex conditions such as Alzheimer’s disease or Parkinson’s disease make premature life choices, such as early retirement or marriage, based on perceived risk? Earlier in this article it is explained that an individual’s genotype rarely determines his or her medical destiny. For example, many people with a higher genetic risk for Alzheimer’s disease will not actually develop it, while many with no apparent higher genetic risk will. Is the risk that members of the general public will misunderstand and overreact to the results of a genetic test sufficient reason to prevent them from obtaining the information gleaned from such a test? Should we be ensuring that those undergoing genetic testing are aware of its benefits and limitations through individualized genetic counseling? This, of course, presents its own challenges of access and availability.

B. People with Disabilities

Some causes of disability are now detected by early genetic testing. Newborn screening for phenylketonuria (PKU) is a shining example of metabolic genetic testing that provides information before the onset of severe mental retardation and behavioral abnormalities. These disabilities result from a buildup of phenylalanine in the blood that is not correctly converted to the amino acid tyrosine in the body due to two mutations in the PAH gene. Testing for

11 Id.
13 Angelina Jolie, My Medical Choice, N.Y. Times (May 14, 2013), http://www.nytimes.com/2013/05/14/opinion/my-medical-choice.html?_r=0.
PKU enables successful medical intervention. Mental retardation can be prevented and the other symptoms mitigated by dietary intervention — feeding children foods with low levels of phenylalanine beginning at birth.

PKU testing is conducted throughout the United States and in many other countries around the world. In the United States, testing for PKU happens shortly after birth by testing a blood sample from the newborn. In the 1960s and 1970s, states began enacting laws that mandated PKU screening for newborns. This mandated screening was initially opposed by the American Medical Association, many state medical societies, and even some researchers in the field of human metabolism. However, most now agree that it is a successful example of low-cost metabolic testing (which in many states now includes screening for many similar metabolic disorders) that provides an opportunity for early disease identification and intervention.

VIII. Looking Into the Future

A. Science and Technology

If the past demonstrates a trend, it seems to be pretty safe to predict that the cost of genetic testing is going to continue to plunge in the near future. Indeed, Francis Collins of NIH predicts that a description of a person’s complete genome will be available for $1,000 in a few years (falling from about $350,000 a year or so ago). Furthermore, the identification of markers for disease risk, traits, and behavior will increase by leaps and bounds. What we know now is just the tip of the iceberg, soon to be more fully revealed.

As the cost of genetic testing goes down, and it becomes within the economic reach of people of average means, the ethical, social, and legal issues stemming from such testing will have to be more squarely addressed.

B. Federal Regulation

Most genetic tests are completely unregulated. In other words, they go to market without any independent analysis that confirms the claims of the seller. Regulation, such as it is, is split among three agencies of the federal government:

- The Food and Drug Administration (FDA) has the authority to regulate genetic tests but so far has only regulated the relatively small number of genetic tests that are sold to laboratories as kits.
- The Centers for Medicare & Medicaid Services (CMS) regulates clinical laboratories but does not confirm whether the genetic tests performed at them are clinically meaningful. CMS only requires that technicians have a certain level of training and proficiency and that lab procedures meet quality control standards.
- The Federal Trade Commission (FTC) regulates false and misleading advertising, and genetic testing has not been on its radar.

For years, expert panels, members of Congress, and other critics have complained about the lack of regulation and inaction by FDA regarding genetic testing. In 2010, FDA finally announced that it would expand its regulation to all genetic tests, but it had not taken any action as of January 13, 2015.14

C. Genetic Information Nondiscrimination Act

President George W. Bush signed the Genetic Information Nondiscrimination Act (GINA)\cite{15} into law in 2008. It passed through Congress with nearly unanimous bipartisan support. GINA bars health insurers and group health plans from charging higher premiums or denying coverage to a healthy individual based solely upon his or her genetic predisposition to an illness. It also bars employers from using an individual’s genetic information in making decisions about hiring, firing, promotion, and job placement.

Those arguing for GINA said that the Act was necessary to keep people from being discouraged from having their genes tested because of the possible adverse consequences. Proponents also said that promising personalized medicine based on an individual’s genotype would be retarded or prevented if people feared that their genetic information could be used to discriminate against them.

Opponents were largely drawn from the business community. They worried about GINA’s possible encouragement of litigation, burdensome recordkeeping and broader coverage, and higher costs of employer-provided health insurance coverage.

Critics point out that there are gaps in GINA’s protections. For example, it does not cover life insurance, disability insurance, or long-term-care insurance. A test showing that a person has an elevated risk of Alzheimer’s disease could prevent him or her from obtaining long-term-care insurance. Not only will the risk to the insurer of having to pay benefits increase, but the insurer will suspect — perhaps correctly — that applicants with such results are looking for insurance coverage solely because of their newly discovered genetic information. These are clear cases of “adverse selection” that insurers seek to avoid. However, claims of genetic discrimination in life insurance have been largely theoretical, and there are few examples of these types of claims. Further, almost half of the states have adopted nondiscrimination protections not provided by federal law.\cite{16}

D. FDA Versus 23andMe

When the genotyping of individuals became a practical reality, both economically and technically, a world of possibilities opened up. One such possibility was simply to tell people what their genotypes said about them. Of course, with the current state of technology and testing, it is impossible to unravel the implications of a person’s entire genome. However, when 23andMe was founded, genotyping was a well-established technology that allowed identification of variations in specific genetic base pairs called single nucleotide polymorphisms (SNPs). With varying degrees of certainty, these SNPs could be linked to different personal traits, disease risks, drug response, and status for carrying certain genetic variants.

In 2006, two entrepreneurs started a company, 23andMe, to do just that. They named the company for the 23 pairs of chromosomes that make up the DNA in a normal human cell and for the orientation of the company toward providing personalized information. The founders are Linda Avery and Anne Wojcicki. A year after the company was founded, Wojcicki married Sergey Brin, the co-founder of Google. Some press reports indicated that this association would ensure economic stability for the new company for the foreseeable future. Other substantial backers have since invested in the company; for example, Johnson & Johnson, Roche

\footnote{16}See Natl. Human Genome Research Inst., \textit{infra} n. 23.
Venture Fund, New Enterprise Associates, MPM Capital, and well-known venture capitalist Yuri Milner. The company continues to be privately held, and Wojcicki remains as CEO.

23andMe began offering personal genome testing kits directly to the consumer over the Internet in November 2007. All the consumer had to do was pay about $1,000, spit some saliva into a tube provided by the company, mail the tube back to the company, and read the results online a few weeks later. The price soon dropped to about $300 and dropped again in 2012 to about $100.

The actions of 23andMe were not without controversy. On the one hand, Francis Collins, director of NIH and a leader in the Human Genome Project, said on a 2012 PBS NOVA program that he had undergone genetic testing, found that he had a higher-than-normal risk of diabetes, and took immediate measures to reduce his weight, get more exercise, and change his diet. Others were much less enthusiastic about direct-to-consumer genetic testing. They worried (and still do) that the public will likely misunderstand the results, fail to seek or understand additional critical information (such as family history and personal lifestyle), and take hasty and imprudent actions as a result. There is limited data about whether direct-to-consumer SNP-based genetic testing will lead to adverse or improved health behaviors and beneficial or harmful psychological effects. A case study from two providers at Ohio State University’s Adult Genetics Clinic delves deeper into some of these potential issues.

Eventually, the axe fell on direct-to-consumer genetic testing. On November 22, 2013, FDA ordered 23andMe to stop marketing its Saliva Collection Kit and Personal Genome Service (PGS). As early as 2010, FDA had informed several genetic testing companies, including 23andMe, that it considered their genetic tests “medical devices” and therefore FDA approval was required to market them. Although 23andMe had apparently been trying to resolve its differences with FDA as early as 2008, its failure to communicate with the agency in 2013 (allegedly for 6 months) was the last straw. FDA wrote a scorching letter to 23andMe charging that the company had not demonstrated that it had “analytically or clinically validated PGS for its intended uses” and cautioning “FDA is concerned about the public health consequences of inaccurate results from the PGS device.” As a result, since December 2, 2013, 23andMe stopped all health-related advertising for its PGS.

23andMe continues to advertise and sell a genetic testing product, but as of December 5, 2013, purchasers have access only to raw (uninterpreted) genetic data, genealogical DNA test results related to ancestry, and database tools for finding relatives. Customers who bought the tests and received results before November 22, 2013, continue to have online access to health information such as the following:

- Disease Risks. Customers are informed whether they are at a higher, lower, or average risk of developing 122 illnesses, including Alzheimer’s and Parkinson’s disease, vari-

17 Email from Andy Kill, Media Relations, 23andMe, to author Gregory Wilcox (Jan. 26, 2015).
19 Cracking Your Genetic Code, supra n. 2.
ous cancers, glaucoma, and psoriasis. Information on a customer’s risk for particularly dreaded diseases, such as Alzheimer’s and Parkinson’s, is “locked.” In this case, the customer is required to read a report on the disease before obtaining his or her results.

- Carrier Status. Customers are informed whether they carry one of 53 genetic variants that could either cause them to have a genetic disease or to pass a genetic disease on to a future generation. Again, access to some reports is “locked” until a customer reads a report.

- Drug Response. Customers are informed about their likelihood having of an increased or reduced therapeutic response to 25 commonly prescribed drugs, such as warfarin (Coumadin), and an adverse response to such drugs as statins, floxacillin, and caffeine (though the company disclaims it is providing a predictive or diagnostic tool).

- Traits. Customers are informed about their traits, including obvious traits (such as hair and eye color), behavioral traits (such as smoking, drinking, and caffeine consumption), and traits such as lactose tolerance and the ability to taste bitterness in foods.

Since FDA issued its order, 23andMe has added disclaimers on its health pages still available to its earlier customers. 23andMe warns in the Health Overview section that the test results “have not been cleared by FDA” and states, “the information on this page is intended for research and educational purposes only, and is not for diagnostic use.”

E. State Regulation

States have been slow to enter the area of genetic testing. Indeed, no state has imposed restrictions or requirements on the sale of genetic testing directly to its residents, according to the National Human Genome Research Institute, a division of NIH. However, many states have enacted rules on topics related to genomics. The topics are, in order of the number of states that have enacted statues on them: health insurance nondiscrimination (48), privacy (38), employment nondiscrimination (35), research (25), other lines of insurance nondiscrimination (e.g., disability and long-term-care) (24), use of residual newborn screening specimens (11), and health insurance coverage (4). Readers can determine their state’s rules on these topics from the links in the Table of State Statutes Related to Genomics provided by the National Human Genome Research Institute.

F. Private and Public Choices

The impending flood of genetic information demands resolution of some of our most basic conflicting values. Is genetic information, similar to body weight and blood pressure, something that people should be able to determine and evaluate on their own, without either government regulation or the permission of experts? Although people can determine their

22 23andMe, Health Overview, The warning is made available only to those who have had their gene tested by 23andMe (including an author of this article, Gregory Wilcox), and who use the “health information” part of the company’s report of results. On file with author.

23 Natl. Human Genome Research Inst., Table of State Statutes Related to Genomics (last reviewed June 10, 2014), www.genome.gov/27552194..

24 Id.
body weight and blood pressure without the permission of a medical provider, physicians may still be the appropriate sources of advice on managing the information— but such advice is not legally required. Or is information from genetic testing more like information collected from an X-ray, urinalysis, or blood test, which requires interpretation by a trained specialist and therefore should only be provided by a medical provider? Or is making genetic information available so risky that it must be dispensed in a similar manner as drugs that can only be prescribed by licensed medical providers according to strict government regulations?

Influencing the answers to such questions are expectations about how much people will understand the test results and how they will respond to them. As described previously, genetic testing results can be complex and ambiguous. If people behave according to human nature, they may respond to complexity by oversimplifying and to ambiguity by jumping to unsupported conclusions.

Even if people fully understand the results of genetic testing, will they abuse or misuse the information they receive? For example, will a genetic report of a low risk of heart attack lead a person to eat more saturated fat and salty food? Will a person react to a perceived increased risk of a dreaded disease by taking drastic measures; for example, by making lifestyle choices, such as retiring or getting married, prematurely? Will a woman be led to having an abortion? The authors of another article published in *NAELA Journal* argue that the availability of prenatal genetic testing for mutations/chromosomal abnormalities that cause disability, such as Down syndrome, may result in markedly fewer disabled people in the future, because prospective parents will abort fetuses with genetic diseases.\(^{25}\) Authors Urbatsch and Fuller present data indicating that a significant percentage of women choose abortion when Down syndrome is diagnosed prenatally. They predict that if more women have access to noninvasive DNA testing techniques, which are more accurate than traditional marker testing, the overall number of abortions resulting from a positive test for Down syndrome will increase even if the percentage of women who choose abortion remains the same. This is complicated by the fact that current noninvasive DNA testing for women younger than 35 is currently being debated\(^ {26} \) and is not offered to all pregnant women. If this testing is offered to all pregnant women, this may affect the number of abortions resulting from a positive test for fetal chromosome abnormalities. In general, genetic testing will join the classic struggle: libertarians will worry about overreaching by the government, while regulators will worry about protecting the uninformed and undisciplined against misinformation and imprudence.

There is some basis for worries about misinformation and imprudence. Studies have shown that many health care providers are not prepared to use genetic tests in clinical practice\(^ {27} \) and that inaccurate medical management and misinterpretation of results occurs

\(^{25}\) Urbatsch & Fuller, *supra* n. 18.


when genetic testing is performed by providers with no specialized knowledge of genetics. Guidelines from some national professional organizations recommend that genetic professionals be involved in the genetic testing process. Perhaps as a result of this data and the potential for increased health care costs when tests are ordered by a non–genetic testing provider, at least one health insurance company now requires the involvement of a certified genetic counselor or similar professional when specific genetic tests are ordered. Other insurance companies may follow suit.

How much do we want to follow the ancient Greek advice “know thyself”? Not everyone wants to know that he or she is at risk of Alzheimer’s or Parkinson’s disease. Some cancer patients do not want to know their prognosis, arguing that it may discourage them from fighting the disease. After all, a person might die of something else before living long enough to encounter a predicted illness. Some argue, unless there is a medical therapy that goes along with the genetic prediction, why bother getting the genetic information in the first place? They argue that the benefits of knowing about a high disease risk are not necessarily worth the costs — such as a life lived differently and negative treatment by others.

Of course, for genetic conditions for which there are medical interventions, the importance of genetic testing and interpretation for the purpose of medical management is widely accepted. In these cases, many argue that knowledge is power and if increased screening or preventive medications can reduce the risk of developing a condition, people would absolutely want this information. If other body systems may be at risk, many individuals want to know to enable their health care providers to make better decisions about follow-up testing in case they have symptoms or an unclear test result. This information may enable a lower threshold for follow-up testing and earlier intervention and treatment.

And then there is the issue of privacy. Should a person’s genotype be his or her property? What then are his or her “property rights”? Should people be barred from using genetic information about another person? Currently, some insurers (health insurers) are barred by federal law from using a person’s genetic information, while other insurers (disability and long-term-care insurers) are not. What sense does this make? And what about personal versus family privacy concerns? Often the outcome of genetic testing of an individual affects other family members. Will parents want to know that they have genetic variants they could pass on to their children and make them vulnerable to disability or disease? What is the parents’ ethical duty to their children to determine whether such variants exist and prevent them from being passed on? Are parents to be condemned if they do not find out the risks of passing on dangerous genetic variants or if they do find out and fail to terminate a pregnancy? Also, what about the parents’ own parents and siblings? They too are often impacted by the results

31 Marieke J. H. Baars et al., supra n. 27; Karen Greendale & Reed E. Pyeritz, supra n. 28; K. L. Brierley et al., supra n. 29.
of genetic testing. Testing is often done to inform the family about why they are plagued with a particular condition.

Apart from their duty to their children and their other family members, do parents have any duty to society at large to protect it from the substantial costs of caring for people with a genetically based disability or disease? Are they required to refrain from sexual behavior for this purpose, or can they just terminate a pregnancy that turns out to be genetically problematic? Currently, a large percentage of the population fervently believes that it is wrong to terminate a pregnancy under any conditions, without apparent regard for the later costs to the parents and society. Accordingly, predictions that genetically based disabilities, such as Down syndrome, will decrease in the future may be premature.

IX. Conclusion

Genetic testing raises a host of medical, social, legal, ethical, and financial questions. It is not clear yet whether individuals will have unfettered pay-for-access to their genomes — or whether this is a good idea in the first place. What is certain is that the health care applications of genetic testing will continue to expand, the cost of genetic testing will decrease, and an individual health care consumer will more and more likely be offered genetic testing. The process of ordering genetic testing, understanding the utility of a particular test in determining disease risk, and the interpretation of results all afford opportunities for misunderstanding and misinformation.

In an ideal health care system, access to genetic information would be available along with comprehensive pretest counseling from licensed providers about the benefits and limitations of testing and post-test counseling about the implications of test results. Of course, we do not have an ideal health care system; therefore, compromises are inevitable. At this point the authors diverge in their viewpoints.

Rachel Koff: Direct-to-consumer testing is fraught with interpretation challenges and potential harmful psychological outcomes for consumers. While it may be interesting to learn how much Neanderthal DNA you carry or whether you have wet or dry earwax, the SNP-based testing for determining such factors does not currently have utility for medical use. FDA agrees, and genetic testing for medical use currently requires, that all genetic tests be ordered by a health care provider.

While direct-to-consumer testing has somewhat abated, genetic testing as a whole is expanding. This expansion should be done alongside increased education for the ordering medical providers about the limitations and ambiguities of test results. Further, medical providers should be encouraged to make referrals to genetic testing providers, such as a geneticist or genetic counselor, when a test presents uncertainty and when an actionable mutation is identified. Patients should also be encouraged to advocate for themselves, because the field is quickly evolving and some medical providers may not know about the applications of genetic testing and genetic testing's impact on medical management.

Government regulation of genetic testing on a laboratory level seems appropriate — making sure that laboratories are certified to use specific technologies appropriately and provide results with a small margin of error. With increasing health care costs, insurance company requirements for genetics professionals to order certain genetic tests seem reasonable. Anecdotal evidence indicates that the first foray into this requirement has helped patients get
proper pretest counseling and has stopped inappropriate tests from being ordered. Access to appropriate providers is increasing, and insurance companies are typically covering appointments. For testing for certain indications, appointments are being covered without cost to the consumer. This continuing growth of the genetics field will continue to assuage concerns about access to and availability of appropriate genetic testing providers.

Gregory Wilcox: Although there is an important role for government regulation of genetic testing providers (e.g., to ensure accuracy and require a balanced interpretation of results), access to genetic information should be freely available without permission from licensed medical providers. Certainly, analysis and interpretation of results by a genetic counselor is preferred. However, genetic counselors are still fairly rare and expensive; they simply will not be available to everyone seeking information about his or her genome. Certainly, also, there are dangers of misunderstanding and misuse of genetic testing results. However, there are dangers in allowing the public to freely access many over-the-counter drugs. Nevertheless, society has elected to accept the dangers of misunderstanding and misuse of these drugs in the interest of wider availability.

I prefer the “warning label” approach — where genetic test results are freely available without government requirements for professional permission or interpretation, but providers are required to fairly and reasonably disclose the ambiguity of the results, to issue warnings about oversimplification, and to advise consultation with genetic counselors for further interpretation before taking any action with important consequences. Consumers should be educated to understand that genetic testing results do not provide answers but instead raise questions.