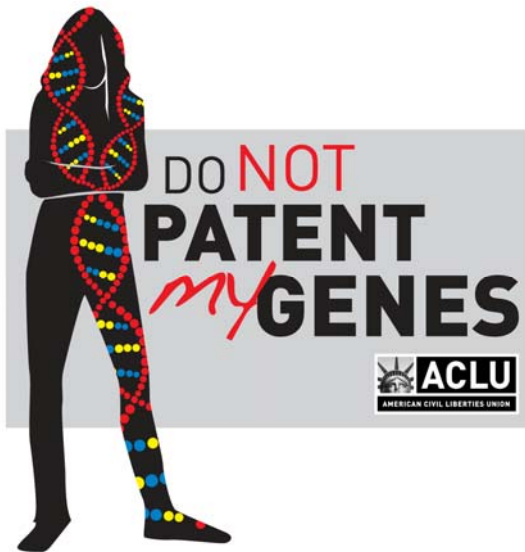


## Legal Challenge to Human Gene Patents

The U.S. Patent and Trademark Office (PTO) has granted thousands of patents on human genes; in fact, about 20 percent of our genes are patented. The PTO gives patent holders the exclusive rights to genetic sequences, their usage, and their chemical composition. Anyone who makes or uses a patented gene without permission of the patent holder – whether it be for commercial or noncommercial purposes – is committing patent infringement and can be sued by the patent holder for such infringement.



Because the PTO grants patents on the genes themselves, it essentially gives patent holders a monopoly over the patented genes and all of the information contained within them. Gene patent holders have the right to prevent anyone from studying, testing or even examining a gene. As a result, scientific research and genetic testing have been delayed, limited, or even shut down due to concerns about gene patents, and patients' options regarding their medical care have been restricted.

On May 12, 2009, the ACLU and the Public Patent Foundation filed a lawsuit challenging the PTO's practice of granting patents on human genes – specifically, the BRCA1 and BRCA2 genes, which are associated with breast and ovarian cancer. The lawsuit was filed on behalf of scientific organizations representing more than

150,000 geneticists, pathologists, and laboratory professionals, as well as individual researchers, breast cancer and women's health groups, genetic counselors, and individual women.

Mutations along the BRCA genes are responsible for most cases of hereditary breast and ovarian cancers. Genetic tests can detect these mutations and tell women if they are at increased risk of cancer, which in turn informs their decisions about screening, prevention, and treatment options.

The patents on the BRCA genes are particularly broad and offensive. The PTO has granted Myriad Genetics, a private biotechnology company based in Utah, patents on both the BRCA1 and BRCA2 genetic sequences, on mutations along those genes, on any methods for locating mutations on the genes, and on correlations between genetic mutations and susceptibility to breast and ovarian cancer. As a result, Myriad's lab is the only place in the country where full sequencing testing is performed for patients; others are prevented from conducting diagnostic testing or offering alternative tests. Myriad's monopoly on the BRCA genes makes it impossible for women to access other tests or get a second opinion about their results, and allows Myriad to charge a high rate for its test – over \$3,000.

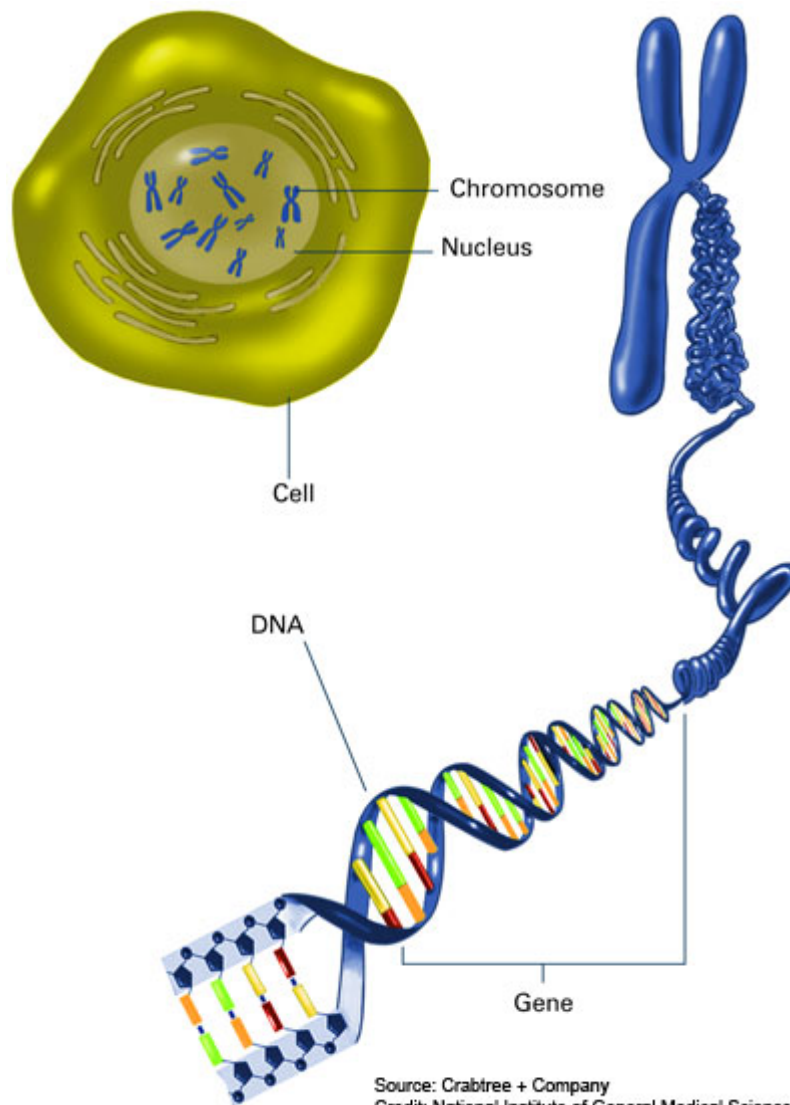
The lawsuit, *Association for Molecular Pathology, et al. v. United States Patent and Trademark Office, et al.*, was filed in the United States District Court for the Southern District of New York in Manhattan against the PTO as well as Myriad Genetics and the directors of the University of Utah Research Foundation, which hold the patents on the BRCA genes.

To learn more about the lawsuit and gene patents, visit [www.aclu.org/brca](http://www.aclu.org/brca).

## BRCA Genes and Patents – Frequently Asked Questions

### What is a gene?

Genes are the basic units of heredity in all living organisms. A gene is a segment of DNA, the molecules that contain instructions for the development and functioning of living organisms. It is estimated that humans have approximately 30,000 genes that make up our genome. DNA is found inside each cell's nucleus, and is organized into structures called chromosomes. Humans have 46 chromosomes – two sets of 23, with one set coming from each parent. The human genome can be thought of as a set of encyclopedias with 23 volumes, where each chromosome represents one volume. The DNA code is like the letters that are used to build the words, paragraphs, and pages of text in those volumes. Because genes vary in size, they can be thought of as a single paragraph or an entire chapter inside each volume.



### **What are the BRCA genes?**

The BRCA genes – BRCA1 and BRCA2 – are two genes that have been associated with hereditary forms of breast and ovarian cancer. Everyone has these genes. BRCA1 and BRCA2 are believed to be tumor suppressor genes, which means that when they are functioning normally, they suppress the growth of cancerous cells. Women who have certain mutations along these genes have an elevated lifetime risk of developing breast and ovarian cancer because their ability to suppress cancerous growth has been reduced. A woman with a BRCA mutation faces a 36 to 85 percent chance of contracting breast cancer and a 16 to 60 percent chance of ovarian cancer.<sup>1</sup> BRCA mutations are also linked to breast and prostate cancer in men.

### **What are gene patents?**

The U.S. Patent and Trademark Office (USPTO) grants patents on human genes, which means that the patent holders own the exclusive rights to those genetic sequences, their usage, and their chemical composition. Anyone who makes or uses a patented gene without permission of the patent holder – whether it be for commercial or noncommercial purposes – is committing patent infringement and can be sued by the patent holder for such infringement. Gene patents, like other patents, are granted for 20 years.

For example, Myriad Genetics, a private biotechnology company based in Utah, controls patents on the BRCA1 and BRCA2 genes. Because of its patents, Myriad has the right to prevent anyone else from testing, studying, or even looking at these genes. It also holds the exclusive rights to any mutations along those genes. No one is allowed to do anything with the BRCA genes without Myriad's permission.

### **How many genes are patented?**

A 2005 study found that 4,382 of the 23,688 human genes in the National Center for Biotechnology Information's gene database are explicitly claimed as intellectual property.<sup>2</sup> This means that nearly 20% of human genes are patented. In addition to the BRCA genes, genes associated with numerous diseases, both common and rare, are patented, including Alzheimer's disease, asthma, some forms of colon cancer, Canavan disease, hemochromatosis, some forms of muscular dystrophy, Long QT Syndrome, and many others.<sup>3,4,5,6,7</sup>

### **You mean companies can patent the *genetic test*, not the gene itself, right?**

No. Myriad has a patent on the BRCA1 and BRCA2 genes themselves. If Myriad had simply patented a test, then other scientists and laboratories could offer alternative testing on these genes. But because it has patented the genes themselves, Myriad has the right to control all research and testing on or involving the BRCA genes.

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<sup>1</sup> National Cancer Institute, "Genetic Testing for BRCA1 and BRCA2: It's Your Choice." Available at <http://www.cancer.gov/cancertopics/factsheet/risk/brca>.

<sup>2</sup> Kyle Jensen & Fiona Murray, "Intellectual Property Landscape of the Human Genome," *Science* 310(5746):239-240 (October 14, 2005).

<sup>3</sup> Ronald Kotulak, "Taking License with Your Genes: Biotech Firms Say They Need Protection," *Chicago Tribune*, September 12, 1999.

<sup>4</sup> US Patent No. 6,037,149 (issued March 14, 2000).

<sup>5</sup> Mildred K. Cho, et al., "Effects of Patents and Licenses on the Provision of Clinical Genetic Testing Services," *Journal of Molecular Diagnostics* 5(1):3-8 (February 2003).

<sup>6</sup> Arthur Allen, "Who Owns My Disease?" *Mother Jones* 26(6):52-59 (November/December 2001).

<sup>7</sup> Wendy Chung, "Statement to the House Judiciary Subcommittee on Courts, the Internet and Intellectual Property in Connection with a Hearing on Stifling or Stimulating – The Role of Gene Patents in Research and Genetic Testing," October 25, 2007.

### **How can someone patent part of the human body?**

Good question! The patent system was designed to grant certain rights to inventors for their inventions in order to reward and encourage human ingenuity. But genes are naturally-occurring parts of our bodies, not inventions. Researchers identify genes, they don't invent them. U.S. law recognizes this differentiation; there is long-standing legal precedent that "products of nature" are not patentable. You can't patent gold or other basic elements, for example.

Nevertheless, the USPTO has ignored this obvious discrepancy for roughly 20 years and has proceeded to issue gene patents on the basis that genetic sequences are "isolated and purified." But all this means is that the gene has been removed from the human body and the non-coding regions of the gene stripped away. These steps – simple enough for any graduate student in genetics or a related field to perform – do not make a gene patentable, any more than removing gold from a mountain makes gold patentable. This is why we are suing the USPTO, to get them to stop issuing such patents which are contrary to the law.

### **Didn't the Supreme Court resolve this issue in 1980 in *Diamond v. Chakrabarty*?**

No. In *Diamond v. Chakrabarty* a divided 5 to 4 Supreme Court held that a genetically modified bacterium that was genetically engineered to ingest oil (for use in oil spill cleanups) was not a "product of nature" and was patentable subject matter under the patent statute. The case did not deal with naturally occurring genes. It dealt with a man-made, genetically engineered micro-organism that the court described as not naturally occurring. The decision made clear that products and laws of nature are not patentable: "The laws of nature, physical phenomena, and abstract ideas have been held not patentable. Thus, a new mineral discovered in the earth or a new plant found in the wild is not patentable subject matter. . . . Such discoveries are 'manifestations of . . . nature, free to all men and reserved exclusively to none.'"

Further, in addition to claims on genes themselves, we are also challenging claims to correlating a mutated gene to having an increased risk of having a certain disease. This issue was nearly addressed by the Supreme Court in the 2006 case, *LabCorp v Metabolite*, where some of the current Justices said that, had the case been heard by the Supreme Court, they would have ruled that such claims are invalid for being unpatentable.

### **Are BRCA1 and BRCA2 the only genes associated with breast and ovarian cancer?**

No. There are more genes that are involved in breast and ovarian cancers, though perhaps not as strongly linked as the BRCA1 and BRCA2 genes. In addition, there are most likely additional cancer-related mutations along the BRCA1 and BRCA2 genes than those for which tests are currently conducted, including mutations that have not yet been identified. Nearly 2,000 distinct mutations and sequence variations have been found along BRCA1 and BRCA2.<sup>8</sup> However, due in part to the limitations that gene patents have placed on studying the two genes, the significance of many of these mutations is unknown.

### **How did Myriad come to have patents on the BRCA genes?**

Many researchers around the world were involved in identifying the BRCA1 and BRCA2 genes; most of them did not seek patents or did not enforce any patents obtained by their universities because they wanted research and testing to continue openly and unfettered by exclusive monopoly rights. Myriad

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<sup>8</sup> National Cancer Institute, "Genetics of Breast and Ovarian Cancer (PDQ): Major Genes: Mutations in BRCA1 and BRCA2." Available at <http://www.cancer.gov/cancertopics/pdq/genetics/breast-and-ovarian/HealthProfessional/page3>.

Genetics filed for a patent on the BRCA1 gene in 1994.<sup>9</sup> Myriad either bought out or fought off any other company that was providing tests or performing research on the BRCA1 gene. By 1999 Myriad was the only company offering a test for and performing research on the BRCA1 gene.<sup>10</sup> Myriad filed for a patent on the BRCA2 gene in 1995.<sup>11</sup> The USPTO then granted Myriad exclusive rights over this gene.

### **What does Myriad have patents for exactly?**

Myriad has patents on both the BRCA1 and BRCA2 genetic sequences, as well as any mutations along those genes. That means, if you were to take the gene you have in your body right now and remove it from all the other biological material that surrounds it, you would be committing patent infringement regardless of how or why you did it. Myriad also has patents on any methods for locating mutations, whether those methods are known now or not, and they also have patents on correlations between mutations and breast and ovarian cancer. The plaintiffs in this case, who are represented by the ACLU and the Public Patent Foundation at Benjamin N. Cardozo School of Law (PUBPAT), are not challenging any patent claims over specific tests for mutations.

### **Why are gene patents a civil liberties issue?**

Gene patenting is a civil liberties issue because gene patents undermine the free exchange of information and scientific freedom, bodily integrity, and women's health. In granting exclusive rights to gene patent holders, the U.S. government in essence gives those patent holders complete control over those genes and the information contained within them. This interferes with a person's right to know about his or her own genetic makeup and scientists' rights to study the human genome and develop new genetic tests. Granting a monopoly on fundamental pieces of knowledge infringes on First Amendment rights, which protect the freedom of scientific inquiry and the free exchange of knowledge and ideas.

### **What laws apply to human gene patents?**

The ACLU and PUBPAT believe that gene patents violate both existing patent law and the Constitution.

- Patent law has long held that products of nature and laws of nature are not patentable subject matter. The USPTO is failing to abide by this precedent when it grants patents on human genes. Human genes, even when removed from the body, are still products of nature, and their associations with diseases are laws of nature.
- The First Amendment protects the freedom of thought, academic inquiry, and the exchange of knowledge and ideas. Gene patents implicate the First Amendment because the very *thought* that there is a relationship between specific genetic mutations and diseases has been patented and because scientific inquiry is limited.
- The Patent Clause in Article 1, Section 8 of the U.S. Constitution gives Congress the power to award patents "to promote the Progress of Science and useful Arts, by securing for limited Times to Authors and Inventors the exclusive Right to their respective Writings and Discoveries." Human genes are not inventions, and awarding patents on them does not promote the progress of

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<sup>9</sup> Robert Dalpé, et al., "Watching the Race to Find the Breast Cancer Genes," *Science, Technology, & Human Values* 28(2):187-216 (Spring 2003).

<sup>10</sup> Shobita Parthasarathy, *Building Genetic Medicine: Breast Cancer, Technology, and the Comparative Politics of Health Care*, MIT Press, 2007.

<sup>11</sup> Robert Dalpé, et al., "Watching the Race to Find the Breast Cancer Genes," *Science, Technology, & Human Values* 28(2):187-216 (Spring 2003).

science. Instead, gene patents slow scientific advancement, because there is no way to invent around a gene – the gene is the basis for all subsequent research.

### **Why is the ACLU getting involved with patent law?**

The ACLU believes that genes and the relationship between genes and disease are products and laws of nature that should never have been considered as patentable subject matter. Along with PUBPAT, we are challenging gene patenting because we believe that the USPTO has violated patent law by granting patents over an area of intellectual activity that should not be covered by patents, and in doing so, is unreasonably restraining free speech and scientific research.

### **Whose rights are violated by gene patents?**

*Scientific researchers'* rights are violated because gene patents prevent them from freely engaging in research and exchanging information about the patented genes. If a researcher wants to study a gene that has been patented, s/he must obtain permission from the patent holder or risk being sued for violating the patent. The USPTO gives the patent holder the unrestricted authority to refuse research licenses, to charge high licensing fees, and to sue and/or shut down researchers who do not have a license.

→ *Using the patent power granted to it by the USPTO, Myriad has forced researchers to pay for Myriad's testing services if they want to tell genetic test results to women participating in their studies.*

*Clinical geneticists' and genetic counselors'* rights are violated because gene patents infringe on their freedom to provide their patients with information about their susceptibility for genetic diseases. Like researchers, clinicians must obtain licenses to conduct clinical testing, and as with research licenses, the USPTO gives the patent holder the authority to refuse to grant testing licenses, to charge high fees, and to shut down un-licensed testing. Genetic counselors are forced to refer their patients to the laboratory dictated by the patent holder and are restricted in the service they can provide their patients.

→ *Currently Myriad does not allow anyone else to conduct full sequencing testing on the BRCA genes, which means that, although there are many qualified geneticists who could conduct the test and provide patients with their results, no one but Myriad is allowed to do so.*

→ *Myriad has sent cease-and-desist letters to several laboratories in the United States to stop them from providing BRCA testing.*

*Individual patients'* rights are violated because gene patents impede access to medical information and care. The patent holder controls what information people can obtain about their own genes, how they may obtain this information, and from whom. The USPTO allows gene patent holders to control the price of diagnostic testing, whether the quality of their tests can be assessed, whether testing can be improved and alternative tests developed through research, and whether patients can get a second opinion.

→ *The monopoly the USPTO has granted to Myriad on BRCA allows Myriad to charge high rates for its testing – currently over \$3,000. Women who cannot afford this price and whose insurance companies do not cover the test have no access to BRCA testing. The USPTO has also given Myriad the power to decide that it will not contract with particular insurance providers. There are many qualified geneticists who could do the testing for less, but are not allowed to because of Myriad's enforcement of its patents.*

→ *Myriad does not allow anyone else to conduct full sequencing testing on the BRCA genes, so Myriad alone can examine a woman's genes to determine if she has any of the mutations that have so far been associated with a higher risk of hereditary breast cancer. There is no way for a woman to know if the test has been done properly, to verify that the results she has received from Myriad are accurate, or to undergo an alternative method of testing.*

- *The USPTO has allowed Myriad alone to determine which mutations on the BRCA genes to look for. For a period of time, Myriad's method of testing had a false negative rate that was estimated to be as high as 12%.<sup>12</sup> When Myriad decided to extend its testing to look for the mutations its standard test was missing, it chose to offer the new testing as a separate test, at an additional cost.*
- *The USPTO has given Myriad the sole power to determine what to do with the data it collects from people who are tested. Women who receive test results indicating that they have a "variant of uncertain significance" have no way to access further testing to find out if they are at elevated risk for cancer or to force Myriad to share their data with other researchers. African-Americans, Hispanics, and Asian-Americans are disproportionately likely to receive these ambiguous test results.*

*The American public's rights are violated because gene patents permit an unfair monopoly that limits the public's right to benefit from scientific breakthroughs that advance medical research. This monopoly has a chilling impact on other researchers' ability to conduct medical research, undermining advances towards better treatments, cures, and more accessible, affordable genetic testing. This blocks the public's right to know about, share, and benefit from research that helps lead to better medical care for cancer and other life-threatening diseases. Such a monopoly serves to profit one company at the expense of the public good.*

### **Can gene patent holders really prevent other scientists from doing research?**

Absolutely. While some patent holders have chosen not to enforce their patents, others have. Studies have demonstrated that research has been delayed, limited, or even stopped due to concerns about gene patents.<sup>13,14</sup> Researchers often avoid studying a patented gene to avoid the cost or difficulty of obtaining a license or over concerns that their license could be revoked. Furthermore, any time a researcher studying a patented gene finds something that could have commercial value, the patent holder can reach through their patent to claim the end-product. This has a chilling effect on research.

The impediment to research created by gene patents is especially harmful in light of the recent scientific understanding that disease is often caused by the complex interaction of many different genes. As more genes are patented, scientists who want to research these interactions will face increasingly higher licensing fees for patents owned by numerous entities.

Because Myriad has exclusive control over testing for the gene mutations, it also has a huge data set on women and the BRCA1 and BRCA2 genes. Myriad thus has the exclusive right to use – or not to use – this research data. Much more research on this data needs to be done regarding the meaning of various mutations, including whether some mutations are more prevalent in certain racial and ethnic minority groups. The fact that a single company controls all uses of this data interferes with this crucial research.

### **Aren't patents supposed to encourage research and innovation?**

Yes, the primary purpose of the patent system is to foster innovation. The USPTO awards the patent holder certain exclusive rights over his/her invention in return for sharing information about the

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<sup>12</sup> Tom Walsh, et al., "Spectrum of Mutations in BRCA1, BRCA2, CHEK2, and TP53 in Families at High Risk of Breast Cancer," *JAMA* 295(12):1379-1388 (March 22, 2006).

<sup>13</sup> Mildred K. Cho, et al., "Effects of Patents and Licenses on the Provision of Clinical Genetic Testing Services," *Journal of Molecular Diagnostics* 5(1):3-8 (February 2003).

<sup>14</sup> Jon F. Merz, et al., "Diagnostic Testing Fails the Test: The Pitfalls of Patents Are Illustrated by the Case of Haemochromatosis," *Nature* 415(6872):577-579 (February 7, 2002).

invention so that others can “invent around” it – that is, improve upon the original invention and design alternatives. This is best illustrated by an example:

When the first cell phone was invented in 1973, the inventor, Martin Cooper of Motorola, was able to patent his particular device. He was required to publish information about the device so that other inventors could learn from it and invent their own alternative devices. Hence the plethora of cell phone companies and options we have today.

But genes are different from cell phones and other things that are patented because they are not inventions, and other researchers cannot invent alternative genes. Even if patent-holders publish information about the genes they have identified, there is nothing to invent around – the genetic material contained in the gene *is* the information. Because this information is the foundation for future diagnostic tests and potential treatments, tying it up as intellectual property can inhibit, rather than stimulate, advances in biomedical research.

### **What about the argument that patents are a necessary incentive for research?**

People who support gene patents often argue that genetic investigation is like drug development and will not take place without the incentive of the patent system. But studies sponsored by the federal government have established that gene patents, unlike other patents, are not required to incentivize research.<sup>15</sup> The Human Genome Project sequenced the *entire human genetic sequence* and did not patent any of the genes it identified. More than five million dollars of federal tax money funded the pursuit of the BRCA1 gene specifically.<sup>16</sup> Overall, much of the world of science has progressed without any expectation of patents: Einstein’s equation,  $E=MC^2$ , and his theory of general relativity were developed without any patent incentives.

### **Who are the ACLU and PUBPAT suing?**

The first defendant is the U.S. Patent and Trademark Office (USPTO). That office has granted thousands of patents covering human genes – an estimated 20 percent of human genes are patented. We believe the USPTO should never have granted these patents in the first place, because patenting human genes is unconstitutional and unlawful. In order to invalidate gene patents, we have to challenge at least one specific gene patent. Since the patents on the genes that correlate with an increased risk of breast and/or ovarian cancer are among the most offensive, we are also suing Myriad Genetics because it controls the BRCA patents and aggressively enforces them. Myriad does not allow anyone else to conduct clinical full sequencing testing. In addition, these patents have particularly broad claims on the BRCA genes, covering mutations that had not yet even been identified.

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5/27/2009

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<sup>15</sup> Secretary’s Advisory Committee on Genetics, Health, and Society, “Public Consultation Draft Report on Gene Patents and Licensing Practices and Their Impact on Patient Access to Genetic Tests,” March 2009.

<sup>16</sup> Bryn Williams-Jones, “History of a Gene Patent: Tracing the Development and Application of Commercial BRCA Testing,” 10 *Health Law Journal* 123, 131 (2002).