

**Backgrounder on Human Gene Patents**  
**Prepared by the NSGC Public Policy Committee**  
**January 4, 2010**

**Overview: Patent Policy and Law**

The United States patent system is intended to preserve an inventor's right of ownership over a new and useful process, machine, manufacture, composition of matter, or improvement.<sup>(1)</sup> While preserving the right of ownership, the patent system is also intended to spur the introduction of innovation into the public marketplace. In exchange for the rights offered under the patent, an inventor agrees to release details of the patented subject to the public. This disclosure gives other inventors the opportunity to use the information as the building block for further innovations and improvements.

To be patentable, a subject must be novel and useful. A subject that was known, used, or published by others in the United States or a foreign country, or which is not substantially different from an existing subject is not considered novel. Another prong of the novelty test is that the subject must be "non-obvious," meaning that it is not something that would be easily apparent to someone with skill or expertise in the field related to the subject. To be useful, a subject must serve a purpose and be able to achieve the purpose stated in the patent application.  
(1)

Patents are granted by the United States Patent and Trademark Office (USPTO), an agency of the Department of Commerce. Once a patent is awarded, it is the responsibility of the patent holder, not the USPTO, to enforce the patent. If the patent holder identifies a party in violation of the patent, the patent holder may bring a suit in federal court. The patent holder can seek an injunction, ordering the other party to immediately cease activities in violation of the patent and/or seek financial damages for any profit lost due to the patent violation.

Patents last for 20 years from the time the patent application was filed. During this time, the patent holder has the exclusive right to bar others from "making, using, offering for sale, or selling or importing the invention."<sup>(1)</sup> The patent holder may issue licenses, allowing others to make, use, sell, or import the patented subject. The patent holder and licensee enter into an agreement that sets guidelines for permissible use of the subject under the patent, royalty fees for use of the subject, and other provisions.<sup>(1)</sup> A patent holder has the option to grant exclusive or non-exclusive licenses. A non-exclusive license means that the patent holder grants the same rights to all licensees.<sup>(2)</sup> Under an exclusive license, the patent holder grants a specific use or right to a licensee and promises not to extend that same use or right to other licensees. In order to enter into a license agreement, a licensee usually enters into negotiations with the patent holder to determine a royalty fee that the licensee must pay in exchange for use of the subject matter.<sup>(2)</sup> Once a patent expires, anyone is free to use the subject under any of the previously barred uses and the patent holder no longer holds the right to issue licenses. For example, generic versions of a pharmaceutical drug emerge once a patent expires, since other companies are now allowed to sell the formula originally protected under the patent.

## Background on Gene Patenting

The first gene patent was granted in 1982 to the University of California. Today, about 20 percent of human genes are patented. Genes, gene mutation sequences, and DNA sequences have all been awarded patents. Of the patent holders, about 60 percent are private research companies and 30 percent are universities. (5) For-profit organizations have generally been supportive of gene patents because without the ability to place licenses on gene sequences, they would not be able to collect the revenue required to offset initial research and development costs. (6) This revenue gives them the security to fund further research and genetic test development.

In 2001, the USPTO released guidance upholding gene patents in the language of the patent statute (35 U.S.C. §101). The USPTO maintained that patenting does not limit innovation; rather it encourages research by others since it requires the patent holder to publicly disclose the details of a gene sequence. Based on this disclosure, the idea is that other researchers benefit from the information as a starting point for a new use or improvement to a test. In its guidance, the USPTO assumed that it would be rare for a commercial patent holder to impose burdensome licensure rules on academic and non-profit research bodies. (7)

The issuance of licenses has been an issue in gene patenting. Some have supported gene patenting as long as non-exclusive licenses are granted and that royalty and other licensing fees are set at a financially reasonable level. (3) Those in favor of exclusive licensure state that it protects a licensee from direct competition.(4) Therefore, the licensee is more willing to financially invest in the research and development of a specific gene, knowing that they are not racing against the work of a competitor.

The Supreme Court has never directly addressed the legality of gene patents, but has ruled on several cases that address the patentability of subjects found in nature. If a case on gene patenting reaches the Supreme Court, it is expected that the Court will reference the following decisions.

- *Diamond v. Chakrabarty*: Upheld idea that anything in nature cannot be patented. However, if a natural substance or organism is isolated and purified by man into a form that is not found in nature, then it can be patented. (8)
  - Case involved a bacterium created in a laboratory. Although it is a living organism, it could not be found in nature; therefore, it could be patented.
  - 2001 USPTO guidance enforces idea that genetic sequences are isolated and purified into a form that is not found in nature; therefore, they are patentable.
- *LabCorp v. Metabolite*: Upheld patent for both an amino acid test and the natural correlation between the amino acid and Vitamin B. (9)
  - Dissenting opinion stated that the correlation was a description of a natural phenomenon and should not be patented.
  - Dissent supports the idea that patents encourage funding for research, but says that the Court should also have considered the harm to the public when researchers are forced to abandon projects because a subject is patented.

- *In re Bilski*: Federal Circuit Court decision requires that a patentable process rely on a particular machine or transformation of a material into a different state. (10)
  - This case involves mathematical-based business processes, but decision could invalidate many gene and genetic test patents, which do not meet the above criteria. (11)
  - The Supreme Court heard arguments in the *Bilski* case in October 2009, and will issue a ruling in spring 2010.

### **Implications – positive/negative and immediate/downstream**

While a fair amount has been written on this topic, it has primarily been in editorial and commentary format, which, although informed and thoughtful, does not document resulting benefits or harms to the general public. (11, 12) Efforts to document the consequences of human gene patenting have not substantiated significant or unremedial harms, although their authors urge ongoing monitoring of the situation or recommend improvements to current USPTO guidelines. (13, 14, 15)

Statements that patent protections for genes, mutations and DNA sequence are beneficial include:

1. Patents are economic drivers that bring advances and innovation into the marketplace.
2. Development of new tests and drugs requires massive investment which will happen only under a system, such as the patent system, that insures investors some opportunity for return on their investment.
3. “Genetic tests from companies with exclusive licensing rights are no more expensive or harder to access than those offered by various providers under non-exclusive license.” (14)
4. “Reports of researchers being blocked from access to patented DNA sequences or being sued for infringement are extremely rare, and workarounds are not difficult from a legal perspective” (14)
5. While access to patented technologies is necessary to advance research, the claim that patents impede or delay advances and innovation are primarily anecdotal, although there are some documented incidents of patents causing limited access to some specific genetic tests. (16)
6. The genetics community has been sensitized by several, high profile patent conflicts involving genetic testing (e.g. Canavan Disease) but these are more the exception than rule. (17, pg 20)

Statements that patent protections for genes, mutations and DNA sequences are not beneficial and might be harmful include:

1. Patents generate high fees for licensing and diagnostic testing development and this limits patient access to necessary information.
2. Exclusive licensing may inhibit patients from seeking a second opinion or from accessing testing through a laboratory with which their insurance plan does not cooperate.
3. Patient education literature developed by a company that holds the patent on a diagnostic or therapeutic may be biased, given the company’s commercial interest.
4. Exclusive licensing agreements inhibit biomedical and clinical research.

5. There is variation across industries and technologies as to the benefits of patent protections, yet there is insufficient analysis to say "...that patents induce additional research and development investment in the service industries and service functions of the manufacturing economy." (15)
6. The media's prominent coverage of the ACLU lawsuit against Myriad Genetics is, rightly or wrongly, setting a tone for acrimony and lack of trust by the general public.
7. In the past, research that has, in part, been funded through Federal (tax payer funded) grants have allowed some private industry claims to patent rights in spite of their use of publically funded sources.
8. Participants/subjects have participated in research that has lead to the development of a patentable gene or test for which they may not be compensated.

### **NSGC Priorities With Regard to Gene Patenting**

NSGC's broad priorities include but are not limited to:

1. Ensuring that patients have timely access to quality genetic services and testing.
2. Ensuring the trust and safety of the public with regard to genetic services.
3. Encouraging the research and innovation of genetic services.
4. Relevance to the NSGC Code of Ethics (18):
  - a. Section I, point 6: Acknowledge and disclose circumstances that may result in a real or perceived conflict of interest.
  - b. Section IV, point 6: Keep the public informed and educated about the impact on society of new technological and scientific advances and the possible changes in society that may result from the applications of these findings.
  - c. Section IV, point 8: Adhere to laws and regulations of society. However, when such laws are in conflict with the principles of the profession, genetic counselors work toward change that will benefit the public interest.

### **Rationale for NSGC Public Policy Statement on Gene Patents**

NSGC has a fundamental problem with the patenting of nucleic acid sequence data. The organization believes nucleic acid qualifies as matter that exists in nature, natural phenomena, which would fall outside of the scope of patentable subjects. We do not recognize the difference between the isolation of a genetic sequence found in nature, which is currently patentable matter, and the isolation of a naturally-occurring element or mineral, which cannot be patented under the original Patent Act.

NSGC does not have a fundamental issue with the patenting of biomedical innovations as a mechanism for bringing advances into the marketplace if the patents are awarded for truly innovative technology. Our primary concern lies in the exclusive licensure of the nucleic acid sequence itself.

Given that the USPTO currently awards patents on nucleic acid sequences, providing patent holders with the discretion to control access through licensure agreements, we have the following concerns about potential negative effects on the future of genetic and genomic medicine:

1. Progress in developing and offering multi-gene testing technologies will likely encounter increasing barriers and costs due to exclusive licensing arrangements allowed under current gene patenting practices.
2. The potential benefits and future applications of large-scale medical sequencing could be thwarted by the practice of “patent stacking,” involving multiple patents on a single sequence, requiring researchers to enter into licensing agreements with different patent holders.
3. As specific gene variations are revealed to be involved in more than one disease pathology, gene patents and exclusive licensure will complicate the ability for other researchers to develop and offer additional or alternative diagnostic testing for diseases other than the disease for which the patent is filed.

From a public policy standpoint, we believe the aforementioned concerns may potentially:

- Lead to significant limitations in genetic research, which are counter to the intent of the Patent Act;
- Stifle the development of innovative tests due to the inability of other researchers to access a sequence for other conditions; and
- Create exorbitant licensure costs that will be passed on to the consumer.

There are two fundamental arguments against gene patenting.

The first is a legal argument against the USPTO’s current interpretation of the patent statute. Under the statute, a subject cannot be patented if it naturally occurs in nature. This argument relies on complex legal arguments that are outside the scope of expertise of our profession.

The second argument focuses on the negative consequences for consumers of genetic services. Rather than arguing against existing statutory language and judicial interpretations of patentable matter, this argument focuses on our concern that continuing to award gene patents will increase exclusive licensure. Exclusive licensure creates the barriers to efficient research and innovation in personalized medicine<sup>1</sup>, potentially delaying the delivery of new tests and treatments to the public. Patents may not inherently act as barriers; rather it is the exclusive enforcement of such patents that may limit the public’s access to such innovation. This argument serves as the basis for the NSGC Public Policy Position Statement on Genetic Patents. We believe that these implications are a serious concern, where the risk of interfering with the public’s access to healthcare outweighs the exclusivity benefits enjoyed by the patent holders.

As genetic counselors whose mission is “to ensure the availability of quality genetic services,” we believe the NSGC statement provides a unique viewpoint from the perspective of the genetic counseling profession.

---

<sup>1</sup> “Personalized medicine” in the context of NSGC’s gene patent position statement is defined as “[the use of] new methods of molecular analysis to better manage a patient’s disease or predisposition toward a disease... Such approaches may include genetic screening programs that more precisely diagnose diseases and their sub-types, or help physicians select the type and dose of medication best suited to a certain group of patients.” (Personalized medicine 101. Personalized Medicine Coalition (2010): [http://www.personalizedmedicinecoalition.org/sciencepolicy/personalmed-101\\_overview.php](http://www.personalizedmedicinecoalition.org/sciencepolicy/personalmed-101_overview.php).)

## **Bibliography**

1. General information concerning patents. United States Patent and Trademark Office (2005): <http://www.uspto.gov/web/offices/pac/doc/general/index.html#faqs>.
2. Black's Law Dictionary (8<sup>th</sup> ed., 2004).
3. Our policy regarding gene patents, Navigenics (2009): [http://www.navigenics.com/visitor/what\\_we\\_offer/our\\_policies/gene\\_patents/](http://www.navigenics.com/visitor/what_we_offer/our_policies/gene_patents/).
4. Scherer, F.: The economics of human gene patents, *Academic Medicine* 77: 1348 (2002).
5. One-fifth of human genes have been patented, study reveals, *National Geographic* (2005): [http://news.nationalgeographic.com/news/2005/10/1013\\_051013\\_gene\\_patent.html](http://news.nationalgeographic.com/news/2005/10/1013_051013_gene_patent.html).
6. Gene patenting, American Medical Association (2009): <http://www.ama-assn.org/ama/pub/physician-resources/medical-science/genetics-molecular-medicine/related-policy-topics/gene-patenting.shtml>.
7. Utility examination guidelines, United States Patent and Trademark Office (2001): [http://frwebgate.access.gpo.gov/cgi-bin/getdoc.cgi?dbname=2001\\_register&docid=01-322-filed](http://frwebgate.access.gpo.gov/cgi-bin/getdoc.cgi?dbname=2001_register&docid=01-322-filed).
8. *Diamond v. Chakrabarty*, 447 U.S. 303 (1980).
9. *LabCorp v. Metabolite*, 548 U.S. 124 (2006).
10. *In re Bilski*, 545 F.3d 943 (Fed.Cir. 2008).
11. Smith, B.: Wrangling genes, *American Bar Association Journal*, July: 57-61 (2009).
12. Andrews, L. et al.: When patents threaten science. *Policy Forum. Science* 314:1395-1396 (2009).
13. Caulfield, T. et al.: Evidence and anecdotes: an analysis of human gene patenting controversies. *Nature Biotechnology* 24:1091-1094 (2006).
14. Editorial: Property rights, *Nature* 458:386 (2009).
15. SACGHS Public Consultation Draft Report on Gene Patents and Licensing Practices and Their Impact on Patient Access to Genetic Tests (2009).
16. A patent system for the 21<sup>st</sup> century. National Academy of Sciences (2004): <http://www.nap.edu/openbook.php?isbn=0309089107>.
17. Reaping the benefits of genomic and proteomic research: intellectual property rights, innovation, and public health. National Academy of Sciences (2006): [http://books.nap.edu/openbook.php?record\\_id=11487&page=R1](http://books.nap.edu/openbook.php?record_id=11487&page=R1).
18. National Society of Genetic Counselors Code of Ethics. National Society of Genetic Counselors (2006): <http://www.nsgc.org/about/codeEthics.cfm>.

### **Additional References:**

Cook-Deegan, R. et al.: The dangers of diagnostic monopolies. Nature 458:405-406 (2009).

ASHG Response to USPTO, March 22, 2000. [http://www.ashg.org/pages/statement\\_3222000.shtml](http://www.ashg.org/pages/statement_3222000.shtml).

Best practices for the licensing of genomic inventions: Federal Register, 69:67747-67748 (2004): [http://www.ott.nih.gov/policy/lic\\_gen.html](http://www.ott.nih.gov/policy/lic_gen.html).

Best practices for the licensing of genomic inventions: final notice: Federal Register, 70:18413-18415 (2005): [http://www.ott.nih.gov/policy/lic\\_gen.html](http://www.ott.nih.gov/policy/lic_gen.html).

Position statement on gene patents and accessibility of gene testing. American College of Medical Genetics (2009): [http://www.acmg.net/StaticContent/StaticPages/Gene\\_Patents.pdf](http://www.acmg.net/StaticContent/StaticPages/Gene_Patents.pdf).

Public consultation draft report on gene patents and licensing practices and their impact on patient access to genetic tests: Secretary's Advisory Committee on Genetics, Health and Society (2009): [http://oba.od.nih.gov/SACGHS/sacghs\\_documents.html#GHSDOC\\_011](http://oba.od.nih.gov/SACGHS/sacghs_documents.html#GHSDOC_011).