

February 16, 2012

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Office of Chief Economist  
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**RE: Docket No. PTO-P-2012-0003; Genetic Testing Study**

Dear Mr. Vishnubhakat:

The American Society for Clinical Pathology (ASCP) is pleased to submit the following comments regarding genetic diagnostic testing and intellectual property.

As a patient-centric organization, ASCP's mission is to protect patient safety while promoting advances in medicine. The impact of gene patenting on research and innovation has been and will continue to be the subject of intense debate, but in the final analysis, the policy that is ultimately in the best interest of patients is the one that should prevail. The lawsuit against Myriad Genetics represents a timely opportunity for the government to correct its initial approval of gene patents which are so problematic for good patient care. Ongoing advances in molecular diagnostics are creating a new frontier in healthcare, one that promises to revolutionize the practice of medicine, and continued and rapid improvement in molecular technology are leveling the playing field for laboratories that want to perform this type of testing. Soon we all will be able to offer patients care that is both personalized and selective, and in doing so, more fully realize our primary objective as health care providers – to deliver the right information and treatment to the right patient at the right time.

The practice of gene patenting fragments ownership of the genome, limiting research and the potential for us to offer patients the highest quality and most cost-effective diagnostics. Gene patents stifle the translational nature of this new technology. Ease of access to genomic discoveries is crucial if basic research is to be expeditiously translated into clinical laboratory tests that benefit patients in the emerging era of personalized and predictive medicine.

The Myriad case and others, as documented in the April 2010 report from the Secretary's Advisory Committee on Genetics, Health, and Society (SACGHS), demonstrate a number of harms to patients that result from gene patents. There are many nuances to the restrictions that the practice of gene patenting places on the delivery of patient care, access, quality, and cost.

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## **Gene Patents Restrict Patient Access to Diagnostics**

The most significant harm to patients is evident when an exclusive (or no license) is issued by a patent holder, thus limiting the performance of a given assay to a single laboratory. This type of circumstance severely limits patient access to testing, particularly when exclusive providers fail to contract with insurers such as state Medicaid programs, leaving patients without the option of a given genetic test should it be recommended by their physician.

Myriad's patents grant them the right to prevent clinicians or laboratories from independently looking at or interpreting a patient's BRCA1 and BRCA2 genes to determine the risk of breast and/or ovarian cancer. Myriad has chosen not to license their patents broadly, and as a result, women who fear that they may be at risk for breast and/or ovarian cancer are barred from obtaining a second opinion.<sup>1</sup> The gene testing monopoly that is created becomes analogous to having certain medical treatments only available at one medical center.

Roger Hubbard, ASCP Member and President and Chief Executive Officer of the Molecular Pathology Laboratory Network, Inc., states in his declaratory statement filed on behalf of ASCP in the lawsuit, "We receive an inquiry every few weeks from a hospital or laboratory asking us to analyze BRCA genes. In each instance, we tell the requesting entity that we cannot do that analysis as a result of the patents enforced by the defendant, Myriad. It is common knowledge in the genetics profession and among laboratories that have the ability to analyze genes that Myriad owns patents on the BRCA1 and BRCA2 genes and that Myriad vigorously enforces its patents. Our laboratory would do those analyses were it not for the patents."

Confirmatory testing by another laboratory is the equivalent to the time-honored practice of obtaining a second opinion from a clinician. Currently, gene patents deny many patients the ability to access a confirmatory test from a different laboratory. Genetic test results can have implications for major medical decisions, therefore the ability to obtain a confirmatory test from a second laboratory is a critical component of quality patient care. One of the plaintiffs in the Myriad complaint would have liked to have a second opinion in her BRCA1/BRCA2 genetic test results, but instead had to make major medical decisions based on the Myriad test results alone.<sup>2</sup>

Other types of access problems have arisen when a patent rights holder has cleared the market of other laboratories offering a genetic test provided by the patent rights holder. For example, there have been periods of time in the life of the BRCA1 and BRCA2 patents in which Myriad did not offer certain tests that were known to reveal additional mutations that increased the risk of breast and/or ovarian cancer. Likewise, Myriad prohibited anyone else from offering those tests to patients even though it knew that they could potentially provide women with essential

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<sup>1</sup> Association for Molecular Pathology v. USPTO, Case No. 09 Civ. 4515, (S.D.N.Y. Mar. 29, 2010)

<sup>2</sup> Association for Molecular Pathology v. USPTO, Case No. 09 Civ. 4515, (S.D.N.Y. Mar. 29, 2010)

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information about their risk of developing cancer. Eventually, Myriad began to offer this additional testing, but chose to package it separately from its standard test.<sup>3</sup>

An even more detrimental scenario occurred with genetic testing for long QT syndrome (LQTS), a serious condition affecting 1 in 3000 newborns that can cause sudden cardiac death. Mutations in 12 susceptibility genes account for some 75% of familial LQTS patients, and of that 75%, mutations in three genes account for most cases. The major LQTS susceptibility genes were discovered at the University of Utah funded in part by the National Institutes of Health (NIH), and the first LQTS gene patent was issued to the University of Utah in 1997. Genetic testing for LQTS is particularly important because knowing which gene (and which part of that gene) is mutated can have a direct bearing on decisions regarding preventative measures and pharmacological therapies.

GeneDx, a national reference laboratory that specializes in rare genetic disorders, developed and began to offer testing for a number of the LQTS susceptibility genes. Shortly thereafter, they received a “cease and desist” letter from DNA Sciences, a company that had obtained an exclusive license covering the LQTS susceptibility genes from the University of Utah. DNA Sciences, Inc. essentially cleared the market with their decision to fully enforce their patent license, yet at the time, they themselves were not actively offering LQTS gene testing.

This action had a potentially devastating impact on patients at risk for LQTS. For approximately 18 months, there was no test available to patients despite the fact that there were laboratories willing and able to offer that test.<sup>4</sup>

Exclusive licensing restricts a patient’s choices. It not only has the potential to clear the market of existing tests, but it also can prohibit laboratories from developing and offering assays presenting serious concerns surrounding quality.

### **Gene Patents Present Quality Concerns**

The most robust means of quality assurance are simply not available in the context of a single provider. Interlaboratory comparison remains the best way to develop gold standard proficiency testing (PT). Certainly, alternative types of PT exist, but none can match the advantages of peer comparison when assays are offered by multiple laboratories. Furthermore, because exclusive licensing eliminates competition, sole providers of assays have less incentive to improve their tests. When a laboratory is permitted to be a soul provider of a genetic test, clinicians and patients who are concerned about quality at the laboratory have no recourse in their choice of laboratories. Hence, it is the laboratory, rather than the physicians or patients that define the

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<sup>3</sup> Association for Molecular Pathology v. USPTO, Case No. 09 Civ. 4515, (S.D.N.Y. Mar. 29, 2010)

<sup>4</sup> Impact of gene patents and licensing practices on access to genetic testing for long QT syndrome. Angrist, Misha; Chandrasekharan, Subhashini; Heaney, Christopher; Cook-Deegan, Robert. *Genetics in Medicine*. 12(4):S111-S154, April 2010

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parameters of testing. In fact, there is concern by many that gene patenting could even be employed to monopolize an area of medical practice.

### **Gene Patents Curtail Our Ability to Deliver Cost-effective Care**

Patent-enabled exclusivity grants single providers carte blanche in determining market price. Gene patents have the potential to eclipse the need for competitive pricing, and our ability to offer cost-effective care for patients. Myriad, in their decision to fully enforce their patent protections, refused to license the technology to other laboratories which, could have engendered competition and lowered prices for consumers. Instead, Myriad kept all BRCA1 and BRCA2 testing in-house, enabling them to set the market price at just over \$3000 per test. Millions of women are potentially affected by either of these mutations, and for many of them this test at that price is simply cost-prohibitive.

Lisbeth Ceriani and Genae Girard, plaintiffs in the lawsuit against Myriad, were both diagnosed with breast cancer. Because of their relatively young age at diagnosis, their physicians recommended that they have the BRAC analysis to determine if their cancers were hereditary. The presence of a mutation for each of them means a significantly increased risk for ovarian cancer and would indicate removal of their ovaries before cancer can develop. Both Ceriani and Girard have health insurance that covers the testing. However since their insurance companies will not pay the full amount, Myriad refused to accept their insurance. As a result, neither woman (at the time of the filing of the lawsuit) had been tested.<sup>5</sup>

This scenario is by no means unique to Myriad. The SACGHS Task Force on Gene Patents and Licensing Practices reported that enforcement of patent rights creates access problems for patients when the exclusive-rights holder does not accept a particular insurance, including Medicaid or Medicare. Patients covered by these payers either forgo a needed test or pay out of pocket for it. When there are more test providers, there is a better chance for coverage. For example, Athena Diagnostics, which has exclusive rights to patents related to the hearing loss gene *GJB2*, has enforced its rights to narrow the market of other tests. Because Athena does not accept Medical, the California Medicaid program, access for MediCal patients has suffered.<sup>6</sup>

In summary, gene patents have significant patient care implications that are manifest in the areas of access, quality and cost. It is for these reasons that ASCP and many other pathology and clinical organizations support the elimination of gene patents based on the over-riding principles of patient centered advocacy i.e., acting first and always in the best interests of our patients.

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<sup>5</sup> Association for Molecular Pathology v. USPTO, Case No. 09 Civ. 4515, (S.D.N.Y. Mar. 29, 2010)

<sup>6</sup> SACGHS Task Force on Gene Patents and Licensing Practices Report. Available at [http://oba.od.nih.gov/oba/sacghs/reports/SACGHS\\_patents\\_report\\_2010.pdf](http://oba.od.nih.gov/oba/sacghs/reports/SACGHS_patents_report_2010.pdf) (Assessed February 10, 2012)

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Please feel free to contact me by email at [cba@UAB.edu](mailto:cba@UAB.edu), thorough my assistant, Margaret Dotzler, at 205-934-2219, or through Andrea Bennett, Senior Program Manager, ASCP Public Policy and Government Relations at 202-347-4450.

Sincerely,

A handwritten signature in black ink, appearing to read "C. Bruce Alexander". The signature is written in a cursive, flowing style.

C. Bruce Alexander, MD, FASCP  
President, ASCP