

**Association for Molecular Pathology**  
*Promoting Clinical Practice, Basic Research, and Education in Molecular Pathology*  
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May 15, 2009

Steven Teutsch, MD, MPH  
Chair, The Secretary's Advisory Committee on Genetics, Health and Society  
National Institutes of Health (NIH) Office of Biotechnology Activities  
6705 Rockledge Drive, Suite 700  
Bethesda, MD 20892

Dear Dr. Teutsch:

The Association for Molecular Pathology (AMP) is pleased to have the opportunity to provide comments to the Secretary's Advisory Committee on Genetics, Health and Society on its Draft Report, *Gene Patents and Licensing Practices and Their Impact on Patient Access to Genetic Tests*.

AMP is an international medical professional association representing approximately 1,600 physicians, doctoral scientists, and medical technologists who perform laboratory testing based on knowledge derived from molecular biology, genetics, and genomics. Since the beginning of our organization we have dedicated ourselves to the development and implementation of molecular diagnostic testing, which includes genetic testing in all its definitions, in a manner consistent with the highest standards established by CLIA, the College of American Pathologists (CAP), the American College of Medical Genetics (ACMG), and FDA.

Our members populate the majority of clinical molecular diagnostic laboratories in the United States. They are frequently involved in the development of novel molecular tests, whether these are laboratory developed or commercially developed. Clinical laboratories cannot develop these important tests unless they have access to the broadest base of genomic discoveries. This access grows more critical as modern technology platforms allow for increasingly sophisticated tests that encompass a greater range and diversity of content from the genome.

AMP has long been concerned that the US Patent & Trademark Office has historically granted broad patents on genomic discoveries, including individual genes or mutations, and large numbers of new patent applications are continuously under review. In AMP's experience, patent holders and their exclusive licensees have frequently chosen to monopolize molecular testing by restricting other health care providers and facilities from developing or performing tests covered by these patents and licenses.

In 2008, AMP initiated a review of its own public policy positions on gene patents and the exclusive licensing of genetic discoveries, asking many of the same questions raised by the Secretary's Advisory Committee in the Draft Report. [see [http://www.amp.org/Gov/GenePatentPositionStatement\\_Final\\_Nov2008.pdf](http://www.amp.org/Gov/GenePatentPositionStatement_Final_Nov2008.pdf)] At the conclusion

of that process, AMP reaffirmed its belief that molecular test services constitute vital medical procedures. As such, they should be widely available to promote optimal patient care, medical education, and medical research. The research, development and practice of molecular testing are essential to medical practice, the education of physicians, researchers and health-care professionals, and the continued improvement of the quality of medical care.

AMP believes that attaching intellectual property rights to true acts of invention such as new therapeutics, diagnostics, or technology platforms is essential to encourage investment and reward innovation. A single gene or a sequence of the genome, however, is a product of nature and should not be patentable. Gene patents can serve as a disincentive to innovation in molecular testing because they deny access to a vital baseline of genomic information that cannot be “invented around.” Moreover, threat of enforcement from a patent holder and ensuing litigation costs lead to a chilling effect as clinical laboratories are reluctant to develop new tests that could directly benefit patients.

In addition to the concern about gene patents, AMP believes that exclusive licenses that confine molecular testing to a single provider are detrimental to the public interest by limiting patient access to testing, restricting medical practice and research, impeding the advancement of medical knowledge and enhancement of the public's health through informed clinical decision-making. Moreover, no governing standards currently exist that would prohibit the practice of granting exclusive licenses. Most patented discoveries of pathogen or human genes can be effectively translated into molecular tests provided they are licensed on a non-exclusive basis and licenses are easily obtainable, both in financial and practical terms.

AMP believes that while the Draft Report raises many key questions, it misses an opportunity to more definitively explore the negative impact on public health that derives from exclusive and restrictive licensing practices. We encourage the Secretary's Advisory Committee to consider additional case studies that demonstrate this point.

Spinal muscular atrophy (SMA), for example, refers to a group of autosomal recessive neuromuscular disorders characterized by degeneration of the anterior horn cells of the spinal cord, leading to symmetrical muscle weakness and atrophy. SMA is the second most common lethal, autosomal recessive disease in Caucasians after cystic fibrosis and has a carrier frequency of 1/40-1/60. Four types of SMA are recognized depending on the age of onset, the maximum muscular activity achieved, and survivorship: type I, severe infantile acute SMA, or Werdnig-Hoffman disease; type II, or infantile chronic SMA; type III, juvenile SMA, or Wohlfart-Kugelberg-Welander disease; and type IV, or adult-onset SMA. All types are caused by recessive mutations in the survival motor neuron (SMN1) gene. Currently, Athena Diagnostics holds the exclusive license and has sub-licensed its testing rights only to Genzyme. Yet the American College of Medical Genetics (Genet Med 2008;v10(11);pp840-842) has recommended SMA for population-based carrier screening (similar to cystic fibrosis). Given the number of births in the US each year, this will be very difficult to achieve with only two labs performing the test.

Another case covered in part by the Draft Report but one AMP believes should have greater examination concerns mutations in the connexin-26 and connexin-30 genes. These are the most common causes of congenital, nonsyndromic hearing loss, accounting for about half of the cases. As such, testing for them has become an important component of diagnostic work-ups of patients

with otherwise unexplained deafness or hearing loss, and has been proposed as an adjunct to newborn hearing screening which is currently conducted in most states. It is well documented that the sooner a baby is identified with hearing loss, the sooner he/she can be referred for definitive audiology evaluation and intervention (e.g., cochlear implant); and the sooner that occurs, the better the child's ultimate hearing and speech.

It is likely that connexin mutation testing will be routinely ordered as soon as a baby fails the first newborn hearing test, in order to avoid the delay that otherwise occurs in getting patients to return to the hospital for two repeat confirmatory hearing tests. In this way, the testing may become a matter of public health, just like newborn screening for PKU and galactosemia. Yet the exclusive license to these genes resides with Athena Diagnostics, and while some additional laboratories have been allowed to perform the test, many other laboratories have been forced to shut down their testing [including one that was in the midst of an NIH-funded grant to study the utility of newborn connexin tests]. If newborn DNA testing does become routine, it would be impossible for a single laboratory to provide satisfactory and timely service for such huge numbers of patients. Thus, this particular patent threatens not just individual patient care and access, but public health for the entire population.

In considering these issues, it's important to note that molecular tests are not static bodies of knowledge any more than our understanding of the human genome is static. Rather these tests are constantly evolving as our knowledge of the genome expands. Additional discovery in genomics should lead to the betterment of existing tests and the development of new tests provided those discoveries are available to laboratory professionals. In addition, access to underlying genes is essential to validate the results in existing tests, to in effect, gain a "second opinion" for patients.

After careful consideration and debate, AMP made four recommendations as part of its revised policy statement on intellectual property. Specifically, AMP recommended that

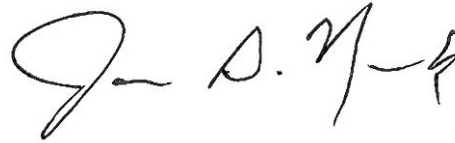
- The patenting of single genes, sequences of the genome, or correlations between genetic variations and biological states should be discontinued, either as a result of judicial review or through an act of Congress.
- Entities, including higher educational and research institutions, that currently hold gene patents, should not grant exclusive licenses to these patents.
- Improve patient access to innovative molecular tests. Financial terms for test licenses should be reasonable and "sole source" tests should be prohibited. License agreements should also be free of any terms that limit the number of tests that can be performed by a laboratory or regulate the technical performance or clinical uses of the test. License agreements should likewise be free of terms that inappropriately limit research related to testing or the public dissemination of the resulting research findings.
- Physicians, researchers, clinical laboratory directors, patient advocates, government officials, research funding agencies and other stakeholders should work cooperatively to develop alternative models to gene patents and exclusive licenses. These innovative models should increase patient access to health care and achieve greater benefit from the existing body of intellectual property linked to the human genome.

In this emerging era of personalized medicine, we believe these recommendations are in the best interests of the patients we serve. AMP's goal is to promote better access to innovative molecular

test services and to ensure the quality of those tests. Our hope is that the Secretary's Advisory Committee will also carefully weigh these recommendations and agree that they should be reflected in the Committee's final report.

Thank you for your attention and consideration of our comments. AMP hopes to continue to be a valuable resource to you as the Secretary's Advisory Committee addresses these critical issues. Please contact us if you need any clarification or further information.

Sincerely,

A handwritten signature in black ink, appearing to read "Jan A. Nowak". The signature is fluid and cursive, with a large initial "J" and a distinct "A." followed by "Nowak".

Jan A. Nowak, MD, PhD  
President