



Archived Policy Statement

Press conference statement on the citizen's petition to the Centers for Medicare and Medicaid Services (CMS) requesting a genetics specialty under the Clinical Laboratory Improvement Amendments (CLIA)

September 26, 2006

By Sharon Terry,
President and CEO, Genetic Alliance

You have heard here about the issues regarding CMS & CLIA from a regulatory standpoint. I am a mother of two children with a genetic condition, pseudoxanthoma elasticum (PXE), and so I have spent the last ten years involved in genetic testing on the other side of the counter.

I am the president and CEO of Genetic Alliance, a coalition of more than 600 genetic disease advocacy organizations, representing more than 1000 genetic diseases affecting millions of Americans. In this position, I have seen and heard a great deal about why a genetic specialty for CLIA is critical. Our own sign on letter, sent to Mark McClellan earlier this year, resulted in an answer that indicated that CMS does not consider this a priority.

In the early days of genetic research and services— even our organization, which celebrates its 20th anniversary tomorrow —focused primarily on Mendelian disorders. More recently, research has turned to complex, multi-factorial conditions such as cancer, diabetes, and heart disease. In addition, personalized medicine is upon us, and scientists and clinicians can understand individual variation in therapeutic response and disease progression.

The bottom line in all of this, is all of us, the consumers of genetic information and services.

Here the rubber hits the road. Genetic tests provide information — information about whether someone has a disease, has an increased risk of developing a disease later in life, is at risk of passing a disease onto his or her offspring, is likely to suffer an adverse reaction to a medication, or is likely to benefit from a particular therapeutic intervention. An accurate test result also can help us make informed decisions about our health and health care. Even when no intervention is available, an accurate genetic test result has enormous value — for preparing for the effects of disease on the psychosocial aspects of life, and for allowing us to enroll in clinical trials, registries and participate in research.

Today I know about laboratory errors, not through a formal reporting system, since none exists, but through hundreds of stories Genetic Alliance has collected. These are the stories that those of us who need a genetic test – one day all of us – share with one another: tests with devastating consequences, causing death, disability, and significant anxiety for us.

A few examples of these kinds of stories, from published sources:

* An Ohio woman who knew she was a carrier of an X-linked genetic disorder underwent prenatal testing to determine whether her child would inherit the disorder. She was told she would have a girl who would not have the disorder.

Instead, she gave birth to a male child with serious disabilities caused by the disorder. The likely cause of this error was maternal cell contamination, in which the laboratory examined the mother's cells rather than those belonging to the fetus.[1]

* A Maryland couple who both were carriers of the cystic fibrosis gene and already had an affected child sought prenatal testing to determine whether their child would have the disease. The laboratory report indicated the fetus did not have cystic fibrosis. After the child was diagnosed with cystic fibrosis at three months of age, the laboratory issued an amended report indicating that the results had been positive for the cystic fibrosis mutation. Laboratory personnel admitted they had “misread the chromatograph” indicating the genetic mutation. [2]

* A young woman who had experienced several episodes of deep vein thrombosis (blood clots) was tested for the factor V Leiden genetic mutation, which is associated with an increased risk of blood clots. The laboratory indicated she had the mutation. Over the course of several years, two other laboratories reported that she was negative for the mutation. Based on the reports indicating she did not have the mutation, and seeking to conceive a child, she began to take a fertility drug known to increase the risk of blood clots. Two months later she experienced extensive blood clots. A fourth genetic test indicated she had the mutation. A case report reviewing this incident determined that the woman did in fact have the mutation and cited laboratory error (sample misidentification, test failure, incorrect interpretation, or clerical error) as a possible reason for the false-negative results by two of the four laboratories.[3]

* A Florida couple both tested negative for the genetic mutation that causes Tay-Sachs, a fatal childhood disease. Two copies of the mutation are required to cause the disease. The couple learned that the test results were incorrect for both parents when their son began exhibiting symptoms of Tay-Sachs shortly after birth. He died eight years later.[4]

* After a middle-aged man was diagnosed with a fatal adult-onset neurological disease caused by a dominant genetic mutation, three close relatives had genetic testing by a different laboratory. The laboratory, which had failed to use a sample from the affected relative for comparison, analyzed the relatives' DNA at the wrong location of the gene and issued a report to two of the relatives indicating they were negative for the mutation. Before releasing the third relative's results, the laboratory realized its error and notified the genetic counselor. The three relatives were informed of the error and decided to be re-tested. After much additional anxiety,

the two relatives again tested negative, while the third relative was found to have the mutation.[5]

So would a genetic specialty for CLIA mitigate all of these errors? No. But at present the bar is low, CMS has an obligation to raise it to minimize such errors. When one is in line for a genetic test, the last thing one wants to think about is the accuracy and reliability of the test. The burden of disease is substantial enough: we would like this easily remedied piece to pave the way toward the access to quality testing so essential to our health.

We ask CMS to give us greater assurance regarding the accuracy and reliability of the genetic tests we use to make profound medical and life decisions.

[1] *Shirmer v. Mt. Auburn Obstetrics and Gynecological Associates*, 844 N.E.2d 1160 (2006).

[2] *Hood v. Lab Corp. of Am.*, 2006 U.S. Dist. LEXIS 36464 (D.Md. 2006).

[3] Edward N. Libby et al., *False-Negative Factor V Leiden Genetic Testing in a Patient with Recurrent Deep Vein Thrombosis*, 81 *Am J. Hematology* 284 (2006).

[4] *Our Stories*, Matthew Forbes Romer Foundation, at <http://www.mfrfoundation.org/stories.php> (last visited Sept. 18, 2006).

[5] Jennie Feiger, *Protecting Patients While Managing Lab Errors*, 25 *Perspectives in Genetic Counseling* 4 (2003).