Genetic Alliance Public Comments to the Clinical Laboratory Improvement Advisory Committee September 6, 2007

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I represent Genetic Alliance, a coalition of more that 600 genetic disease advocacy organizations, representing more than 1000 genetic diseases affecting 25 million Americans. We are certain a genetics specialty for CLIA is critical.

In the early days of genetics research and services– even our organization, which celebrated its 20th anniversary last year –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.

What matters in all of this is access to quality genetic tests. 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 can also 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.

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.

We filed a citizens petition in September 2006 with the Genetics and Public Policy Center and Public Citizen. In addition to the petition, CMS received letters from nearly 100 organizations, including health care providers, patients, and industry, requesting that the agency create enhanced genetic testing regulations. The petition sought to increase the use of proficiency testing by requesting the creation of a "specialty" for genetic testing.

In a decision that places cost concerns above public health, the Centers for Medicare and Medicaid Services (CMS) has rejected a petition. Citing cost concerns, the agency told petitioners in a recent letter that it would not pursue the safety standards.

In response to this rejection, Kathy Hudson, Director of the Genetics and Public Policy Center said: "We believe that CMS has abdicated responsibility for ensuring the quality of genetic tests and has erroneously placed cost considerations above the public's health. The letter uses the word 'cost' repeatedly, but not once does it mention health and safety. We find this astounding for an agency charged with protecting patients by ensuring laboratory quality."

The petition called on CMS to exercise its authority under the Clinical Laboratory Improvement Amendments of 1988 (CLIA). As you are aware, the law requires the secretary of the Department of Health and Human Services (HHS) to set standards for laboratories to ensure their quality, including standards for "proficiency testing." Proficiency testing provides independent confirmation that the laboratory can get the right answer reliably when performing laboratory tests. However, CMS has not mandated participation in proficiency testing for genetic testing laboratories.

Genetic tests are currently available clinically for more than 1100 diseases. Genetic Alliance President and CEO, Sharon Terry is concerned that the results of these tests are being used to make life and death decisions. A patient who is going to decide whether to have a child, or undergo surgery, or take a particular drug really needs to know that the test used to make that decision gives the right answer.

In denying the petition for rulemaking, CMS stated that few proficiency testing programs exist and that the barriers to expanding the number were "technological and financial," rather than regulatory. In fact, the College of American Pathologists now offers proficiency testing for more than two dozen genetic tests, including for widely used tests such as cystic fibrosis and Factor V Leiden, but laboratories currently are not required to enroll in them. Mandatory requirements would result in more proficiency tests being developed, the petition argued.

To support its contention that developing new proficiency testing programs is too difficult, CMS points to its experience with Pap smears – stating that it took 17 years from the passage of CLIA to develop a nationwide proficiency testing program.

The third petitioner, Peter Lurie, deputy director of Public Citizen's Health Research Group, responded by saying: "The agency is using its own history of bureaucratic ineptitude as the basis for not moving forward on genetic tests. That's just absurd. The excuse 'it's too hard for us' is unacceptable when it comes to patient safety."

CMS's response also dismissed reports from three federal advisory groups over the past decade recommending that the agency strengthen genetic testing oversight, and ignored data on genetic testing laboratory quality gathered by the Genetics and Public Policy Center. The Center's 2006 survey of laboratory directors of genetic testing laboratories found that laboratories do not always enroll in available proficiency testing programs. Moreover, the survey found that laboratories that reported performing less proficiency testing also reported experiencing more "deficiencies" – or errors – at some stage of the testing process.

We ask that you, the advisory body for CLIA, to seriously consider the decision of CMS to refuse to create a genetics specialty for CLIA.

MORE INFORMATION:
Center for Medicare and Medicaid Services' full response to the Petition for Rulemaking:
http://geneticalliance.org/ws_display.asp?filter=cms.response.press.release

The Petition for Rulemaking:
http://www.dnapolicy.org/resources/Petition_For_Rulemaking_September_2006.pdf

Report - Public Health at Risk: Failures in Oversight of Genetic Testing Laboratories:
http://www.dnapolicy.org/pub.reports.php?action=detail&report_id=20

Survey of US genetic testing laboratories:
http://www.dnapolicy.org/resources/Nature_Biotechnology_September_2006_bw.pdf

Issue brief - Who regulates genetic tests?
http://www.dnapolicy.org/policy.issue.php?action=detail&issuebrief_id=10

Article - Senators Obama and Burr introduce genetics bill:
http://www.dnapolicy.org/news.enews.article.nocategory.php?action=detail&newsletter_id=21&article_id=83

Article - Senator Kennedy introduces the Laboratory Test Improvement Act:
http://www.dnapolicy.org/news.enews.article.nocategory.php?action=detail&newsletter_id=20&article_id=78

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