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Genetic Testing Oversight

A MAJOR IMPACT OF SEQUENCING THE HUMAN GENOME HAS BEEN THE ABILITY TO DETECT disease and the risk of disease through genetic testing. Today, there are genetic tests for more than 1000 diseases, and that number is increasing rapidly. Given the potential powerful health consequences of genetic test results, shouldn't someone be in charge of making sure that the tests are accurate and reliable? Amazingly, in the United States no one seems to be, despite a direct congressional mandate and a very clear public expectation that there be such oversight. How has such a failure come about, and what should be done to remedy the situation?

During the 1990s, in anticipation of the "genetic revolution" in medicine, numerous government and other advisory bodies recognized that the rules governing garden-variety laboratory tests were simply insufficient for the age of new genetics. They recommended that the Centers for Medicare and Medicaid Services (CMS), the agency within the U.S. Department of Health and Human Services (DHHS) that is responsible for the quality of clinical laboratories, beef up the standards for genetic testing laboratories. CMS was charged with adopting new regulations to guide a smooth translation of genetic testing from research to practice. Key among these recommendations was explicit enhancement of the accuracy and reliability of genetic testing under the Clinical Laboratory Improvement Amendments of 1988 (CLIA).

One would expect, then, that CMS has been active in ensuring that genetic testing laboratories are getting it right. After all, proficiency testing is mandatory for labs that perform diagnostic tests in microbiology, immunology, and clinical chemistry. In 2000, CMS announced that it would develop such tailored regulations for genetics. But nothing has happened for the past 6 years, leaving a system in place that still does not routinely evaluate the competence of genetic testing labs.

Things did look up when, in April 2006, DHHS placed the creation of genetic testing rules on its regulatory agenda, with a target date of November 2006. This announcement was received enthusiastically by diverse patient advocacy groups, health care provider organizations, industry, and genetic testing laboratories, which collectively urged expeditious action. Three months later, inexplicably, the government abruptly reversed course. CMS now asserts that creating regulations to ensure the quality of genetic testing laboratories lacks sufficient "criticality" to warrant rulemaking, and that existing CLIA regulations are adequate to protect the public health.

Existing regulations? A Senate hearing in July 2006 released a Government Accountability Office report that detailed fraudulent genetic tests offered over the Internet, and the failure of one of the laboratories doing the testing to deliver consistent results using the same DNA. A 2006 survey by the Genetics and Public Policy Center found that, in the United States, at least a third of genetic testing labs fail to perform proficiency assessments for some or all of their tests, and that analytic errors increase in direct proportion to the failure to perform proficiency testing. Draft guidelines for genetic testing quality released in 2006 by the International Organization for Economic Cooperation and Development similarly identified proficiency testing and lab quality as key to ensuring public health worldwide.

We know intuitively and empirically that errors in genetic testing can have tragic consequences. We need to forge and enforce rules—the right rules—to ensure the quality of genetic testing. Laboratories should be required to demonstrate that they can reliably perform the tests that they sell. And when they do poorly on proficiency testing, health care providers and the public have the right to know so that they can make wise health care decisions. That responsibility sits squarely with CMS. The Food and Drug Administration's (FDA's) jurisdiction also bears on genetic testing, and this year it has shown vigor in drafting guidelines for the safety of certain genetic tests. But the FDA's efforts cannot substitute for CMS doing its job to ensure laboratory quality.

At worst, genetic testing errors can kill; at best, they result in poorly spent health care dollars. Moreover, should the public begin to question the accuracy of genetic tests or insurers begin to question their validity, "personalized medicine" will be nothing more than a postscript on the pages of medical history. We need sensible regulation to secure the future of genetic medicine.

—Kathy L. Hudson

