The case for genetic testing oversight

Kathy L. Hudson, PhD, is Director of the US Genetics and Public Policy Center, and Associate Professor in Berman Bioethics Institute, Institute of Genomic Medicine, and in the Department of Pediatrics, Johns Hopkins University. Gail H. Javitt, JD, MPH, is the Center's Director for Law and Policy. Today

While some have warned of the emergence of 'genetic exceptionalism' - the fear that genetic medicine will be treated differently than, and regulated separately from, conventional medicine - the sad reality is that genetics has been ignored by US health oversight agencies such as the Centers for Medicare and Medicaid Services (CMS). Rather than receiving heightened or special attention, genetic testing has fallen between the cracks.

We have argued for some time that current oversight fails to ensure either the analytical validity (getting the 'right' answer reliably) or the clinical validity (verifying that the DNA variant has a proven connection to a health outcome) of genetic tests. A report by Congress's investigative arm, the Government Accountability Office (GAO), released at a Senate hearing in July, provided ample empirical evidence for our claims.

The GAO conducted an undercover 'sting' of companies selling so-called nutrigenomic tests directly to consumers, usually over the Internet. For example, they submitted a single DNA sample for testing under different assumed identities. Even though the DNA was identical, the genetic test results were not: clearly, the labs were unable to reliably reach the same conclusions about the same DNA. Instead, their diet and exercise advice appeared to be based almost entirely on the lifestyle profiles submitted along with the samples. In summarizing the situation, GAO concluded that, 'the results from all the tests GAO purchased mislead consumers by making predictions that are medically unproven and so ambiguous that they do not provide meaningful information to consumers'. These findings led the chairman of the Senate Special Committee on Aging, Senator Gordon Smith, to liken these tests to 'modern-day snake oil'.

Today, there are genetic tests clinically available for nearly 1000 diseases, with hundreds more in development. Genetic tests provide information -- information that can be used to diagnose disease, predict risk of future disease, and guide decisions about whether to undergo a medical procedure or take a particular drug or dosage of a drug. With so much riding on the results of genetic tests, it is imperative that they be accurate, reliable, and relevant to a patient's health. As we have argued here at the Genetics and Public Policy Center, and as the GAO report shows, current oversight of genetic testing fails to ensure this.

Responsibility for ensuring the analytic validity of genetic tests lies with the CMS, which implements the Clinical Laboratory Improvement Amendments of 1988 (CLIA). In 2000 the government indicated that it would strengthen regulation of genetic testing, bringing it in line with other types of tests considered to be high complexity. Six years later we are still waiting.

In November 2005, the Genetics and Public Policy Center called on CMS Administrator Mark McClellan to issue the proposed rule for a genetic testing specialty. Subsequently, nearly 100 groups, comprising patients, industry, health care providers, and women's health advocates, added their voices in calling for the proposed regulation. In fact, it is hard to find anyone opposing this change. In June, the agency indicated that they would

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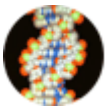

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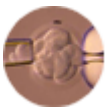

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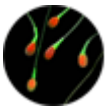

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hearing, however, CMS did an abrupt about-face: it failed to even mention the proposed rulemaking activity and instead insisted that existing regulations are adequate. After six years of 'we are working on it' CMS now appears to have changed its mind and will not pursue rulemaking for genetic testing specialty - a position contrary to every government advisory group, think tank, and nonprofit organization that's ever been asked to weigh in on this issue.

The notion that standards tailored for other complex testing - such as pathology, toxicology, and microbiology - could be used to ascertain a lab's ability to do genetic testing is preposterous. There are certain areas where genetic medicine is different, and this is one of them. Immediate action by CMS is urgently needed to create proficiency testing standards for genetic testing under CLIA.

Of course, fixing CLIA wouldn't solve problems with clinical validity or misleading claims such as those the GAO report highlighted, problems that prompted the Federal Trade Commission and Food amd Drug Administration (FDA) to issue warnings to consumers about at-home genetic tests on the day of the Senate hearing. That is because the CLIA statute is focused on laboratory quality, not the clinical validity of tests. Currently, there is no government agency with clear responsibility to ensure the clinical validity of most genetic tests.

Many laboratories are of extraordinarily high quality and offer only those tests for which there is broad scientific agreement regarding clinical validity. However, several reports, notably the GAO's, indicate that some laboratories are offering genetic tests to the public in the absence of sufficient evidence of their clinical validity. Frankly, there is no one minding the store. Moreover, because there is no requirement that laboratories disclose publicly the scientific basis for their tests, it is extremely difficult for consumers to determine whether a test is bogus or is based on real science. Ensuring that sufficient scientific evidence supports a test's relationship to health status is essential if health providers and the public are to make decisions informed by the best information possible.

We can only hope that the bright lights that the Senate hearing focused on these regulatory cracks can finally help overcome a decade of bureaucratic inertia.

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BioNews, c/o Progress Educational Trust, 140 Gray's Inn Road, London WC1X 8AX, UK
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