

6 **GEN** Point of View

Successful Genetic Tests Are Predicated on Clinical Utility

Industry Desperately Needs Data Confirming that Usage Will Benefit Patients

Diane Allingham-Hawkins, Ph.D.

Over 1,200 genetic tests are now available on a clinical level with an additional 300 available on a research basis only, and the number is predicted to increase by 25% annually. Clearly, the business of genetic testing is booming. But are the test results reliable? Should insurers cover them? Should physicians trust them?

Genetic testing holds immense promise for diagnosis and treatment. Among medical professionals today, however, there is more skepticism than promise and for good reason.

Many of the genetic tests offered today have little or no evidence of clinically proven value. Moreover, the regulatory infrastructure to apply due diligence on the utility and validity of these tests is largely nonfunctional or nonexistent.

Of the tests currently available, only about a dozen are FDA-approved. The Clinical Laboratory Improvement Amendments (CLIA) set laboratory standards for quality assurance and accuracy. Even so, there is currently no genetics specialty under CLIA, and the general lab-testing standards don't adequately encompass many of the issues specific to genetic testing nor do they address clinical utility, which is defined as the ability of the genetic test to inform treatment or care decisions for patients or their family members.

The vast majority of tests marketed today are unregulated, and anyone can develop and bring a genetic test to market. Development methodologies and test components don't

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have to be disclosed and can vary significantly among laboratories. This leads to genetic test results that are often unreliable and difficult to interpret. More troubling still is the fact that genetic tests are increasingly marketed directly to consumers, bypassing physician oversight.

Exacerbating this already complex scenario is a lack of trained geneticists and genetic counselors to read and interpret test results. Most primary care physicians are not qualified to do so.

Acceptance

None of this is lost on the healthcare industry. Healthcare professionals want tools and techniques to better scrutinize genetic tests, results, and efficacy. Although genetic tests cost from \$100 to \$7,000 (or more), insurance companies will cover them—if they have hard evidence that proves their clinical validity and utility.

To gain acceptance of increased clinical usage, the industry must change how it develops and uses genetic tests. Firstly, there needs to be increased oversight of genetic testing. It's important that CLIA develop a genetics specialty that establishes, regulates, and enforces genetic testing procedures, protocols, etc. This would refine and clarify laboratory testing requirements and increase testing consistency across labs. Validation and accuracy standards would be documented, well known, and uniform.

For example, one standard that CLIA should implement is to require laboratories that conduct molecular genetic

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This Week's PODcast — *Clinical Trial Ethics in the Developing World*

GUEST Jonathan Kimmelman, Ph.D.

WHEN August 7, 2008

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TOPIC Carrying Out Early-Stage Clinical Trials in Developing Nations

Early-stage gene therapy clinical trials are recruiting patients from the developing world and providing medically deprived populations with access to interventions that show promise. However, the effects of these early studies

on humans are largely unknown.

According to bioethicists, the practice may be inconsistent with international ethics guidelines on justice.

Dr. Kimmelman discusses why researchers are increasingly looking to the developing world as a source of clinical trial participants and describes a number of key concerns about carrying out early-stage trials in developing nations. He offers some well thought out advice to companies or scientists planning to carry out early-stage trials in the developing world.

GEN Online Exclusives

This month there are three stories that appear in a more detailed version on our website. Are you wondering how the plethora of options for diagnostics at the approval stage came to be? Check

out **Regulatory Pathways for Molecular Dx** online. You can also eavesdrop on *GEN's Roundtable*, which appears in part on p. 26, by tuning into the related podcast. Finally, glean additional information about **Graduate Biotech Education** on the web. www.genengnews.com

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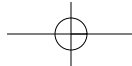
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Genetic Tests

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testing to participate in external proficiency testing programs for every test they perform to ensure the quality of testing. In addition, staff should be required to have specific genetic training and education credentials before being certified to create, conduct, or read tests.

In addition, there needs to be greater FDA oversight of genetic testing kits and reagents than currently exists. Likewise, the FDA must regulate home-brew tests by increasing developer accountability. Developers should be required to provide the FDA with empirical data about a test's validity, accuracy, and clinical utility.

Tests must meet certain prerequisites before developers can market the tests, including achieving acceptable levels of sensitivity and specificity, which would be test-dependent. Further, direct-to-consumer marketing itself must be regulated.

Fortunately, progress is being made. The Laboratory Test Improvement Act (S. 736), spearheaded by Sens. Edward Kennedy (D-Mass.) and Gordon Smith (R-Ore.), would amend the Food, Drug, and Cosmetic Act to require prescriptions for all laboratory-developed direct-to-consumer genetic tests. It also classifies tests as medical devices, which would put them under FDA regulation.

The Genomics and Personalized Medicine Act (S. 3822), introduced by Sens. Barack Obama (D-Ill.) and Richard Burr (R-NC), would require manufacturers of laboratory-developed genetic tests to submit analytical and clinical validity data to the Secretary of Health and Human

Services. The Secretary would make the data publicly available.

Some industry leaders aren't waiting for legislation. Medical analysts at Hayes are now evaluating genetic tests on points of analytical validity, clinical validity and utility, and ethical, legal, and social implications. Our firm's goal is provide a clear, objective report of the science behind the tests, clinical evidence supporting them,

and use of the tests in clinical practice.

These reports will give the industry the data required to determine whether usage and results benefit patients and whether tests are cost-effective for health plans and hospitals. The CDC's National Office of Public Health Genomics has made similar assessment efforts. However, the organization has produced only one report since it was created in 2004.

Industry reforms are needed to improve genetic testing and unlock its diagnostic and treatment potential. Increased regulation, unbiased assessment, and evidentiary proof of the clinical validity and utility of these tests are the path to this new era. **GEN**

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Letter to the Editor

As a response to the June 1 article, "Off-Label Promotion Under Scrutiny by DOJ," I would like to clarify a few important points.

As a company, **Jazz Pharmaceuticals** (www.jazzpharma.com) is committed to ethical and responsible marketing practices. As an outgrowth of our dedication to the best interest of patients, ethics and compliance have always been and will continue to be core values of our organization.

We feel it is important to note that no government action was ever brought against Jazz Pharmaceuticals. The lawsuit mentioned in the article was against Orphan Medical, a company acquired by Jazz Pharmaceuticals in June 2005, and the penalties are being paid by Orphan Medical. Jazz Pharmaceuticals did not become aware of the investigation until the spring of 2006. When the U.S. Attorney's office announced resolution of the matter, it noted our proactive activities to ensure compliance with all applicable laws and regulations governing the sale and marketing of pharmaceutical products, both before and after we became aware of the investigation.

Thank you,

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