Transparency, Openness, and Genetic Testing

Kristi D. Zonno and Sharon F. Terry

TRANSPARENCY IS A HALLMARK of The Obama Administration's Open Government Directive. It is a critical element in health reform overall, and in genetic testing particularly.

The Transparency Task Force of the Food and Drug Administration (FDA) held a meeting in June to obtain public input on how the FDA can increase transparency in information, activities, and decisions. As part of this meeting, multiple stakeholders, including Genetic Alliance, provided comments to the FDA Task Force. Themes of access to information, improved communication, balancing transparency and innovation for industry, seeking and valuing input from patients and consumers, and the need for consistency were the essence of comments from many participants.

Transparency and openness have the potential to transform the U.S. government and the work it does. This is critical in the realm of genetics and genomics if they are to transform how we understand human health. The Human Genome Project modeled the power of transparency of data and the paradigm changes that it causes, including a surge in innovation. Open access to powerful datasets and new technologies has led to dramatic advances in understanding the causes of diseases and establishment of a robust foundation to learn about the genetic basis of disease. With this comes the promise of developing lifesaving tests and treatments. Integrating these promises into health management requires proactive and informed decision making, and access to reliable information is instrumental to this process.

How do transparency and openness apply to the genetic testing and biomarkers? In this journal, we have previously discussed two key issues in the system: the need for a registry of genetic tests and the need for a measure of truth in genetic testing through infrastructure that ensures tests are accurate, reliable, and safe. Creation of a public registry for genetic tests will enable transparency and promote informed decision making for consumers, providers, and test developers. Those who have navigated the complexities of the genetic testing system understand these needs. A registry of genetic tests would greatly serve consumers, patients and families, and their healthcare providers as they seek understanding through genetic testing to answer questions such as:

- * What is the diagnosis or prognosis?
- * What is the risk of developing a condition?
- * What is the risk for my family members?
- * What is the risk of having a child with a specific condition?
- * What is the optimal treatment?
- * What is the predicted benefit of a specific intervention?

These are big questions. The answers to many of these questions can be life altering, and important decisions flow from their answers. The need for the registry is not only felt by patients and providers, as it will benefit all stakeholders, including researchers, industry, payers, and especially the federal agencies who are tasked to oversee various aspects of the genetic testing system.

A mandatory public laboratory test registry would include the myriad of genetic, genomic, and pharmacogenomic testing available to the U.S. market. At a minimum, the publicly accessible registry would include the name of the laboratory performing a specific test, the name of the laboratory or company that developed the test, and information to support the methods and claims about how useful the test is in obtaining correct results and improving clinical care. This will in turn strengthen oversight of clinical laboratory quality, currently tasked to the Centers for Medicare and Medicaid Services (CMS), to make sure that the information provided by genetic and other advanced diagnostic tests is accurate, reliable, and timely.

In developing the registry, we have a huge potential and grand opportunity in front of us. If this is done as a collaborative process across all health-related agencies that have a tie to genetics, and includes input from all stakeholders in the genetic testing system, we can make this a forward looking, forward thinking, comprehensive product. It's roles would relate to oversight, ensuring quality, providing education, and serving as a gateway to information and services for those considering testing and in follow-up to testing when results are delivered to patients and providers to answer those important questions previously mentioned. The registry should be overseen by the FDA and be managed by the National Center for Biotechnology Information (NCBI) to allow integration with all of the other genetic resources in the nation, including dbGaP, Genetests, PubMed, Clinicaltrials.gov, and others.

Additional transparency mechanisms communicated to the FDA include:

- * Immediate release of any and all FDA warning letters, including all untitled letters, which are issued but not consistently (or always) available to the public.
- * Information from public meetings should be archived and maintained so that it is easily accessible to the public.
- * Citizen's petitions should be made public and posted on the FDA website in a manner that is easily accessible to the public. Any responses or actions should be clearly evident.

Each of these elements will allow the genetic testing community to make the most timely and informed decisions in test development and deployment. Furthermore, as robust biomarkers are developed, many of which are critical to determining the efficacy of drugs as they are developed, transparency will be essential to optimize care delivery.

Through applying transparency and openness to the genetic testing system, and working with federal health-related agencies as they strive to apply these principles to their work, process, products, and services, we can collaboratively work to improve the health of all Americans.

Address correspondence to:

Kristi Zonno, M.S., C.G.C.

Director of Genetics and Health Policy

Genetic Alliance

4301 Connecticut Avenue, NW

Suite 404

Washington, DC 20008

E-mail: kzonno@geneticalliance.org

Sharon Terry, M.A.

President and CEO

Genetic Alliance

4301 Connecticut Avenue, NW

Suite 404

Washington, DC 20008

E-mail: sterry@geneticalliance.org

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