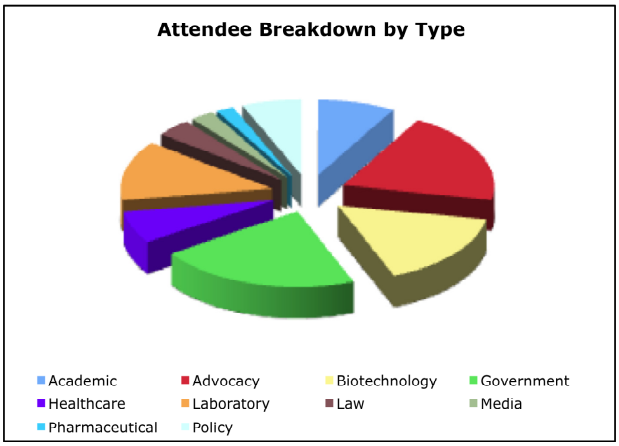
Introduction



Individuals and families affected by genetic conditions, numbering in the millions all over the globe, suffer a great deal. While many do not yet label themselves as affected, we all carry dozens of deleterious mutations, many of which contribute to the myriad of common conditions from which we suffer. Although the human genome sequence was finished in 2000— seven years before this Summit—we are not at a point of regular translation of basic science to improved human health. We must accelerate the pace of translation. To do this, we must understand the bottlenecks, discover the communication disconnects, and pave the way to increased collaboration leading to the discovery that is within our reach.

Genetic Alliance convened this Summit as 'open space'; a place where all stakeholders could come and freely offer their concerns, opinions, and resources. We sought a safe place for truth telling, and we are grateful for the many stakeholders who rose to the occasion.

We welcome and support innovation, and know that we must be disruptive to create the new systems necessary to create tests, therapies, and treatments. We also know that the lay public needs to understand the options available to them, and people need assurance that these options are safe and effective. We are certain that an informed public, for whom decision-making takes place in an atmosphere of transparency, enhances access to services. The new age is not only one of new technologies, but also one of entry into a community commons, where it becomes increasingly apparent that creative use of shared resources in novel partnerships will promote the solutions we all seek.

This meeting is dedicated to those who depend on our good senses, and hope for the breakthroughs we imagine.

Welcome & Context

Sharon Terry, President and CEO of Genetic Alliance, welcomed participants and set the tone for the two day Summit by reflecting upon its name, Eyes on the Prize: Truth Telling about Genetic Testing. The coveted prize is better health for all of us: for those we love as well as those we have never met, in the community, the country, and the world at large. Terry invited everyone to use the prize as a lens. While it is a challenge to abandon our individual agendas and conflicts of interest, doing so illuminates the path to real progress. Each sector of the genetic testing community—academia, advocacy, government, industry, clinicians, and patients—has a distinct perspective on the challenges and opportunities that accompany the path forward. However, that perspective too often translates into a narrow point of view.

By abandoning our turf and rising above our own perspectives, we can have dialogue about genetic testing that is more open, honest, comprehensive and fulfills our goal of "Truth Telling."

Terry asked each panelist to declare all conflicts of interest, whether for profit or nonprofit, commercial or personal. Panelists disclosed their employers, pertinent financial holdings and board memberships, and any other relevant personal interests. This structure created an avenue for open, honest dialogue and gave listeners a more developed understanding of speakers' perspectives. It also provided speakers with the opportunity to articulate and examine any biases before moving into the depth of the discussion. This helped all parties recognize questions that stem from self-interest, such as "What about MY disease? What about MY bottom line? What about MY institution's interests? What about MY intellectual property?" The intent was not to eradicate self-interest, but instead to be transparent about it.

This led to productive questions that addressed the heart of what matters: "How can we improve the public's health? How can we condense the timeline from research discovery to standard of care? How can we manage intellectual property and create a balance between sharing information through the public domain while maintaining financial incentives to support innovation?"

Terry laid out the goals of the Summit: to determine (1) consensus points within the genetic testing community, (2) hard questions that need further discussion, and (3) action steps.

The Department of Health and Human Services and Personalized Healthcare

Greg Downing, PhD, Program Director for Personalized Healthcare for the U.S. Department of Health and Human Services (HHS), provided an update on HHS's Personalized Healthcare Initiative, a high priority for HHS Secretary Michael Leavitt [note 1]. As defined by HHS, personalized healthcare consists of "medical practices that are targeted to individuals on the basis of their specific genetic code in order to provide a tailored approach."

Downing emphasized that there are many factors involved in bringing new genomic technologies into the marketplace, and consideration of these factors is impacting the direction of HHS. There is "a systems problem in overall healthcare," and HHS wants to transform the healthcare system so that it focuses on (1) prediction, (2) prevention, and (3) preemption. Ensuring that new technologies are accessible is of particular concern.

HHS thus seeks input from the community regarding analytical validity, clinical validity, and clinical utility. The Department is interested in the community's needs within the current framework as well as projected future needs.

Downing mentioned ongoing federal efforts outside HHS. The Food and Drug Administration (FDA) recently released a new guidance on analyte specific reagents (ASRs). Also, the Secretary's Advisory Committee on Genetics, Health, and Society (SACGHS) is working on a draft report that will provide more information on policies and practices for large genomic databases.