House Committee on Appropriations Subcommittee on Labor, Health & Human Services, Education, and Related Agencies

Written Public Testimony
Sharon F. Terry, President & CEO, Genetic Alliance

For release on delivery, expected:
March 18, 2009
10:00 AM

Chairman Obey, Representative Tiahrt, and members of the subcommittee, thank you very much for this opportunity to testify today on behalf of Genetic Alliance, and in fact, on behalf of all Americans who seek therapies and treatments for genetic diseases.

I did not choose this work as my career; the vocation was bestowed on me more than 14 years ago when my two children were diagnosed with a genetic disease called pseudoxanthoma elasticum (PXE). In my capacity as president and CEO of Genetic Alliance, I serve the 10,000 health related organizations in our network, of which 1,000 are dedicated to specific diseases.

Genetic Alliance was founded in 1986 as a support group for support groups, building capacity in those organizations. Today, our mission is to transform health using the tools and technologies born through the study of genetics and genomics. We actively engage all stakeholders to create novel partnerships, improve health systems, and revolutionize access to information to enable translation of research into services.

As a result, we are interested in all appropriations related to health, and we are aware that the commitment of this committee, the 111th Congress, in collaboration with the Administration of President Obama, is immense. We are grateful for Chairman Obey's significant contributions to important and meaningful Department of Health and Human Services (HHS) appropriations over the years. We ask that bold leadership continue to drive appropriations to reflect the extraordinary opportunities and challenges of health research and services today.

Our world is interconnected; we continually witness new organic linkages in global finance, social networking, and health and disease. To that end, we have several specific requests, and a final comment.

Health Information Technology

We ask that you focus a substantial amount of funding on health information technology to create a research to healthcare services continuum that leverages current technologies. All of the

current systems around research and services are built on cottage industry models and need to be brought into the 21st century commensurate with and exceeding the standards of other industries, such as the financial services industry. Further, privacy, confidentiality, and access can all be achieved with forward thinking solutions.

Strategic, Long-term Translational Research Plan

HHS, primarily through the National Institutes of Health (NIH), but in close collaboration with other agencies, should develop a strategic, long-term plan that includes new approaches and innovative translational tools to enhance the clinical adoption of discovery research. This will require strong leadership to catalyze unprecedented levels of collaboration and coordination. The Human Genome Project is a model for the execution of a large project requiring vision, planning, and collaboration. Furthermore, the recent passage of the Genetic Information Nondiscrimination Act, for which we applaud the US Congress, paves the way for needed advances in health through genetics and genomics.

We envision two projects of this type. The first is a cohort study, using robust health information technology and enrolling millions of Americans in a variety of studies, that enables large numbers of clinical trials. A large, national cohort study should be complemented by CDC surveillance for all diseases, along with the resources to initiate and manage essential services.

The second project involves increased and substantial funding for the Rare and Neglected Disease Initiative, first funded in this current fiscal year. There have been dramatic advances in understanding the causes of many rare and neglected diseases in recent years. The Human Genome Project has helped to define the molecular basis of many diseases that were known only by phenotype, or physical characteristics. This brings scientists to the point of being able to engage in target-based drug development. Through programs such as the NIH Roadmap Molecular Libraries initiative, scientists are gaining access to high throughput screening of chemical compound libraries, and are successfully identifying research probes for disease-related targets. By the end of 2008, the NIH Molecular Libraries screening network had identified 60 chemical probes with activity against the desired target. Some of these are potentially therapeutic. In one example, a small molecule compound has been identified and shown to cure schistosomiasis in an animal model. Schistosomiasis affects 250 million people worldwide.

Like the Genome Project, the Rare and Neglected Disease Initiative, in partnership with industry, advocacy, and academia, will develop novel technologies and ultimately new paradigms to develop drugs for diseases that offer little incentive for focused attention.

Regulatory Oversight

Appropriations must provide adequate funding for the Food and Drug Administration (FDA) and Centers for Medicare & Medicaid Services (CMS) to coordinate oversight and regulation of genetic and genomic testing, as the cornerstone of personalized medicine. FDA lacks the resources to address issues related to genetics and genomics. CMS should further seek new and creative ways, with full stakeholder participation, for coding, coverage, and reimbursement of genomic tests that will encourage innovation and not penalize or reduce reimbursement for established clinical laboratory tests.

A registry for genetic tests should be developed and maintained that includes the name of the laboratory performing a specific test, the name of the laboratory or manufacturer that developed the test, and information to support claims about the analytical validity and clinical validity of that specific test or test method. Submission of information to this registry should be mandatory for all advanced diagnostic assays. It is critical that an agency capable of integrating this registry with other databases—such as NIH or FDA—is given the necessary support to do so.

Oversight of clinical laboratory quality systems by the Clinical Laboratory Improvement Amendments (CLIA) program should be strengthened to assure that the information provided by advanced diagnostic testing is accurate, reliable, and timely. FDA and the Centers for Medicare & Medicaid Services (CMS) should avoid unnecessary duplication in oversight and reconcile any conflicts in regulation between the medical device rules and regulations under CLIA.

In the realm of rare diseases and orphan products, increased funding is needed to create systems that allow the FDA to be a leader in innovative oversight, which enables development of rare disease tests and therapeutics.

Services

The Health Resources and Services Administration (HRSA) should also receive funding commensurate with its sister agencies, as the focus on the continuum shifts from basic research to treatments and services. This should include a systems-based approach for newborn screening and follow-up, with adequate support for the various state programs. Further, HRSA should lead the nation in preparation for the issues that will arise as prenatal, newborn, and childhood screening—including direct-to-consumer—become widely available. The health literacy and education needs of the nation will increase, and adequate resources should be put toward health professional and consumer education to enable empowered decision-making.

The Social Security Administration is to be applauded for its Compassionate Allowances initiative, a way to expedite the processing of disability claims for applicants whose medical conditions are so severe that they obviously meet Social Security's standards. Resources need to be allocated to allow a more comprehensive rollout beyond the initial 50 conditions.

The Commons

We must take our advocacy, research, services, and policy to the next level and establish a networked approach that discovers treatments and manages disease. Until now, earmarking has been reflective of our collective understanding of the system and how to approach it. Yet when we look specifically at appropriations and funding as the energy to empower health systems, we recognize that earmarking is not an example of interconnectivity. Rather, it represents isolation, fragmentation, and segmented communication. Now is the time to work together to create a systemic response, not a partial response, in order to solve our health crises.

Disease advocacy organizations have worked together for many decades to drive transformation: for example, passage of the Genetic Information Nondiscrimination Act of 2008 and the Newborn Screening Saves Lives Act of 2008, and the lifting of the ban on federal funding for stem cells most recently.

These achievements teach us that working together toward a common goal is key to success. It is becoming increasingly clear that the transformation of basic science to services, which so many of us seek, will require deep and meaningful collaboration. This vision of a commons would necessarily include funds to build adequate infrastructure, provide resources, and create and support networks for all disease-specific interests to systematically address their needs—e.g. to electronically aggregate disease-specific natural history data, share methods for establishing best practices for standards of care, and build shared technology resources. We are seeking this on a federal level and ask that the funding be given to HHS agencies to catalyze this transformation.

We are aware that the collaboration we seek on the federal level must also take place in the nonprofit community. Many disease advocacy organizations move forward in an isolated manner to address their specific issues and needs. Historically, progress has been made in these disease-specific silos, and often the lessons learned are never shared with the community at large. This impedes the development of better health. Biology is systems based. Prior to the genomic age in which we work, perhaps it made sense to study diseases based on an organ, or location within the body. However, since sequencing the human genome, we know that there are gene families, pathways, and other more effective ways to understand disease. There are many examples of treatments and cures for diseases coming from an unexpected direction. We work to inspire the disease advocacy community to reflect the interactive, interconnected nature of science and seize the energy inherent in networks.

Congressional earmarks for specific diseases have contributed to this siloed effect, and have ultimately stifled progress for the greater good and the collective community. It is possible, given the systems structure of science, that they also stymie research on the very disease for which an earmark is sought. It is time to move away from earmarking as a solution.

Genetic Alliance strongly supports policy, systems, funding mechanisms, partnerships, and collaborations that benefit all stakeholders. This includes tools, technologies, and resources that are developed or designed for a specific cause, as long as those developments are freely available to all who can use, adapt, or benefit from their existence. Every effort must be made to disseminate success and to learn from failures. We acknowledge that the budget and appropriation process at any level must include prioritization and differentiation, but disease-specific earmarking should no longer be part of this process. There is not enough time, funding, or resources to study and develop treatments for each disease individually, yet there are millions of people waiting for our help.

Now is the time to strengthen our collaborations, as there have been significant advances in science, technology, knowledge of diseases, and processes for developing treatments. We must collectively share success and mine our failures in developing systems, practices, and initiatives to study diseases and get treatments to those in need. The NIH open access policy is a good example of the openness that must be supported, and we encourage its expansion to all federally funded research results.

We call for a culture shift in the relationship between advocacy, research, services, and policy. We are poised to synergize efforts to benefit all stakeholders.

Our long-term needs will no longer be best addressed by earmarking for one organization or one disease. We can go much further together. Let us step into the future as collaborators who build shared infrastructure and solutions that accelerate our work beyond what anyone can do alone.

We look forward to partnering with Congress and the Federal agencies to create this networked model for improved health for all.