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Call for Action from Genetic Alliance

Registry of Genetic Tests: A Critical Stepping Stone to Improving the Genetic Testing System

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For years stakeholders in the genetics and healthcare community have been vocal about the need for changes in the genetic testing system, but there has been little consensus regarding what those changes should be. While debate has ensued, the field has continued to evolve in the number, types, and indications for genetic tests. Now more than ever it is time for change. We are at a juncture where it is critical for health, medicine, government, and industry to get this right, so we must work together to create policy and transform systems, allowing for continued growth and innovation while ensuring the safety and quality of tests to improve health.

The Genetic Information Nondiscrimination Act (GINA), signed into law in 2008, addresses a major barrier in the genetic testing system—fear of discrimination based on genetic information—and provides an on-ramp to move forward into better health. Genetic testing, innovation, and technology give us the tools to transform health through genetics. But without improvements to the system as a whole, these tools cannot be utilized as intended, nor to their full potential, and we will all suffer the consequences.

Benefits for individuals, families, and communities are the ultimate goal of the genetic testing system, and these benefits range from informational to life saving. We are all consumers; we all have been or will be patients; and we can all benefit from improved health through genetics. In evaluating this system and thinking about change, we must keep these important end goals in sight and be mindful that there are people waiting for tests and treatments that will save their lives.

Within the genetic testing system, there are questions about science, access, reimbursement, coverage, and oversight. Informed decisions must be made on the basis of analytic and clinical validity, clinical utility, and individual usefulness, as well as an understanding of oversight, regulation, and reimbursement. Accurate, reliable, and validated information must be available to individuals and providers as they make decisions about testing and the information gained through the testing process.

Although there is much work to be done in crafting a better system, a key and rather simple step is to develop a publically available, mandatory registry of genetic tests and laboratories performing genetic tests. Current lists are voluntary, so there is no comprehensive information available about tests that can be used by payers, providers, patients, and consumers for comparisons and evaluations. The Centers for Medicare & Medicaid Services (CMS) claims to have a list of laboratories, along with their Clinical Laboratory Improvement Act approval status, but this is not publically accessible in a useful format. How can oversight occur without knowledge of what tests are available, where tests are conducted, and by whom? How can patients, providers, and payers make informed decisions without knowing what options exist? A registry that includes all tests across the risk continuum and comprehensive standardized information in a format appropriate for the public would enable truly informed decision making regarding genetic testing. Indeed, a registry has been called for by Genetics and Public Policy Center and the Coalition for 21st Century Medicine.

Test performance characteristics and reference information, including analytical validity and clinical validity, should be available through the registry. It is anticipated that making this information available will in turn increase confidence in and improve the proper utilization of genetic tests, both those ordered by providers and those available directly to consumers. The testing landscape currently includes a wide range of institutions and companies offering testing services, with a broad range of interests. This diversity results in a spectrum of quality and intentions that include standard-setting good actors as well as bad actors who ultimately harm the system. Yet, a system that includes a mandatory registry and enables informed decision making has the potential to organically weed out the bad actors, through mechanisms such as adverse event reporting to track the harms of testing, and help ensure high quality tests.

To maintain credibility and independence, the registry should be housed at and managed by a federal regulatory body, such as the Food and Drug Administration (FDA), or by the National Institutes of Health (NIH). Existing voluntary registries for both clinical and research testing could be used as a model or baseline for a national mandatory registry. Innovative, forward-thinking technology solutions should be considered during creation of the registry, and novel partnerships among all stakeholders should inform its development through an open and transparent process. Education regarding basic genetics and the testing process; professional society recommendations and guidelines, information for patients and providers on risk or diagnosis; and referral networks for specialists, researchers, and disease-specific organizations could

all be built into or linked with the registry to maximize its potential benefits and facilitate its integration into the genetic testing and healthcare systems. This integrated, linked, layered registry would provide a gateway for access to services and information, as well as facilitate important connections for those who receive a diagnosis through the testing process.

A mandatory registry would enable a forward-looking oversight system that is flexible and nuanced. It would undergird a transformed system that is open, transparent, and coordinated with all stakeholders and agencies, and that balances safety, innovation, ethical and social issues, viability, and the risks and benefits of improved health. How can it be that the human genome has been sequenced through innovation and collaboration, yet the systems currently in place lack the capacity to maximize the benefits that are the inevitable result of such an achievement?

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