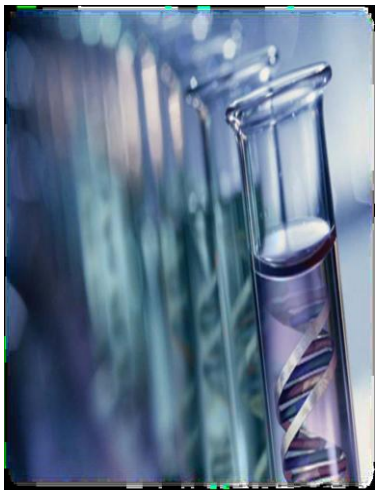
National Institutes of Health Genetic Testing Registry



Scientific advances over the past decade have greatly expanded our understanding of the genomic and genetic factors involved in health and disease. This knowledge has led to tremendous growth in the development and use of genetic tests, many of which are highly complex. Currently, however, there is no centralized source of information about the characteristics of these tests. A comprehensive, publicly available registry of genetic tests would be beneficial for several groups such as health care providers, researchers, laboratories, manufacturers, and policy makers. Calls for greater transparency of genetic testing have come from a number of quarters (e.g., the Secretary's Advisory Committee on Genetics, Health, and Society[1]).

The National Institutes of Health (NIH) responded by creating the Genetic Testing Registry (GTR, http://www.ncbi.nlm.nih.gov/gtr), a public database of test information submitted voluntarily by genetic test providers (e.g., laboratories conducting the test). The registry was developed with input from stakeholders such as laboratory test developers, health care providers, and industry groups. The intended audience for the GTR is health care providers and researchers. Enhancing access to detailed test information is important to enable informed decision-making by clinicians and to facilitate research. The GTR also may be of value to other groups such as clinical laboratory professionals, payers, policymakers, and regulators.

The GTR is designed to provide information about the purpose of the test and its limitations, whether it is a clinical or research test, the test methodology and analytes that are measured, analytic validity, clinical validity, and clinical utility as well as laboratory contacts, credentials, and certifications. The GTR will be developed in a phased manner, and Phase I will focus on tests for heritable mutations, including pharmacogenomic tests and tests using complex arrays and multiplex panels. Future phases will incorporate tests for somatic mutations and whole exome or whole genome sequencing assays.

The GTR will be of immediate value to clinicians by providing information about test availability and the accuracy, validity, and usefulness of a particular genetic test. It will also highlight evidence gaps where additional research is needed to understand the clinical validity and clinical utility of genetic tests. In addition, the GTR may facilitate collaborations such as laboratory participation in quality assurance exchanges. Also, given that the adequacy of genetic testing oversight has been an issue for more than two decades, the GTR's ability to enhance the transparency of this field will be of value to public policy makers.

The National Center for Biotechnology Information (NCBI), part of the National Library of Medicine at NIH, is responsible for developing and maintaining the Registry. The project is overseen by the NIH Office of the Director. For background information and public comments about the GTR see http://oba.od.nih.gov/gtr/gtr.html.

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[1] Secretary's Advisory Committee on Genetics, Health, and Society. U.S. System of Oversight of Genetic Testing: A Response to the Charge of the Secretary of Health and Human Services. 2008. See http://oba.od.nih.gov/oba/SACGHS/reports/SACGHS_oversight_report.pdf.