Genetic Testing Registry

Purpose and Scope

1. Will the Genetic Testing Registry include results from genetic testing?

No. The Genetic Testing Registry does not include data from individuals’ genetic test results.

2. What are the benefits of the Genetic Testing Registry?

The National Institutes of Health expects that the Genetic Testing Registry will benefit a large stakeholder community including health care providers, researchers, and test providers. Anticipated benefits include:

- Expanding publicly available information for clinicians, researchers, and others about test availability; indications for testing; and data about the accuracy, validity, and usefulness of a test.
- Identifying gaps in scientific knowledge.
- Enabling test providers to identify and connect with other providers to create collaborations, such as participating in quality assurance exchanges.
- Understanding trends in genetic testing over time, such as the uptake of next-generation sequencing by clinical testing labs.

3. Why did the National Institutes of Health develop the Genetic Testing Registry?

The National Institutes of Health (NIH) is the primary Federal agency for conducting and supporting basic, clinical, and translational medical research. Within NIH, the National Library of Medicine (NLM) and the National Center for Biotechnology Information (NCBI) support efforts to advance medicine and public health by sharing information and developing automated systems for storing and analyzing knowledge about molecular biology, biochemistry, and genetics. Currently, NLM and NCBI maintain a large number of public databases and resources that are heavily used by researchers, clinicians, consumers, and patients. The Genetic Testing Registry (GTR) is integrated with relevant NIH resources to assist each of these groups. Thus, NIH is a natural home for the GTR because of its role in advancing public health through science and its strong expertise in developing databases.
4. What is the intended audience for the Genetic Testing Registry?

The Genetic Testing Registry (GTR) is intended primarily for health care providers and researchers. Future phases may incorporate features designed for a user base that includes patients and the general public.

5. What is the scope of the Genetic Testing Registry?

The development and growth of the Genetic Testing Registry (GTR) has occurred in phases. Phase I focused on tests for heritable mutations, including biochemical and pharmacogenomic tests as well as tests using complex arrays and multiplex panels. As this phase continues to expand, the GTR has moved onto Phase II, which includes tests for somatic mutations and assays that use whole exome or whole genome sequencing.

6. What is the goal of the Genetic Testing Registry?

The overarching goal of the Genetic Testing Registry (GTR) is to advance public health and research into the genetic basis of health and disease. As such, the GTR has the following key functions:

- Encourage providers of genetic tests to enhance transparency by publicly sharing information about the scientific basis and utility of their tests;
- Provide an information resource for the public, particularly health care providers, to locate laboratories that offer particular tests; and
- Facilitate genetic and genomic data-sharing for research and new scientific discoveries.

7. What is the purpose of the Genetic Testing Registry?

The Genetic Testing Registry (GTR) is an online tool that enables health care providers, researchers, and others to navigate the rapidly changing landscape of genetic tests. GTR has detailed information about genetic tests for more than 3,800 inherited disorders—a number that has grown steadily and continues to rise. However, until the GTR, limited information about genetic tests was scattered across multiple databases. The GTR is a centralized, publicly available database that aims to provide comprehensive information
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about genetic tests.

Submitting Genetic Test Information

8. How should test submitters refer to their participation in the Genetic Testing Registry?

Test submitters may refer to the fact that information about their tests is available in the Genetic Testing Registry (GTR) and provide the relevant URL(s). Before providing test information to the GTR, submitters must agree to abide by a code of conduct, which stipulates that submitters make no explicit or implicit claims that their tests listed in the GTR, or other information submitted to the GTR, have been approved or endorsed by the National Institutes of Health (NIH), the Department of Health and Human Services, or the U.S. Government. If this stipulation is not honored, NIH reserves the right to take action, including, in its sole discretion, removing the submitter’s tests from the GTR.

9. What is the cost to participate in the Genetic Testing Registry?

There is no cost to submit information to the Genetic Testing Registry (GTR). Test providers can submit information about a single test or multiple tests, at no charge. It is also free to access information in the GTR.

10. How difficult will it be to submit data?

The National Center for Biotechnology Information (NCBI) has established an online submission system with several options for submission format. NCBI remains engaged with test providers in streamlining the submission system and has incorporated a number of time-saving features. NCBI will continue to work with test providers to ease data submission processes.

11. How will testing information be submitted?
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Information is collected and managed using an online submission system. Further information is provided in a tutorial about the GTR submission process.

12. What type of information can be submitted to the Genetic Testing Registry?

The Genetic Testing Registry (GTR) is designed to provide a wide range of information about genetic tests such as the purpose of the test and its limitations; the name, location, and credentials of laboratories providing the test; whether it is a clinical or research test; the testing method and what the test measures; analytical validity data, and evidence of clinical validity and clinical utility. The GTR will not include individual test results or other confidential or proprietary information.

13. Is participation in the Genetic Testing Registry mandatory?

No. Participation in the Genetic Testing Registry is not mandatory. Providers of genetic tests are encouraged to submit test information voluntarily.

14. How will submissions to the Genetic Testing Registry be referenced, and will they be linked to other National Institutes of Health resources?

Each test in the Genetic Testing Registry (GTR) is assigned a unique accession number, allowing for uniform reference to tests across various entities, including scientific publications and electronic health records. In addition, the GTR is integrated with other National Institutes of Health databases and resources.

15. Who can contribute test information to the Genetic Testing Registry?

Test providers who perform all or some portions of a genetic test can submit information to the Genetic Testing Registry. Providers of genetic tests include U.S. and non-U.S. clinical and research laboratories.
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16. What action will the National Institutes of Health (NIH) take if test submitters do not review submitted information at least once a year?

Test submitters agree to abide by a code of conduct, which stipulates that they will review and, if necessary, update the submitted information at least once a year. The NIH National Center for Biotechnology Information sends submitters an annual reminder of this obligation. If this stipulation is not honored by a test submitter, information that is more than one year old will be labeled as out of date. NIH reserves the right to take action, including removing out-of-date submissions from the GTR if the information is two years beyond the last annual review.

17. What action will the National Institutes of Health (NIH) take if it discovers that submitted test information is inaccurate or misleading?

Test submitters agree to abide by a code of conduct, which stipulates that they will uphold the integrity of the GTR through the submission of information that is accurate and not misleading. If this stipulation is not honored by a test submitter, NIH reserves the right to take action, including, in its sole discretion, requesting that the test submitter amend inaccurate or misleading information or, if a submitter fails to respond to this request, removing the relevant submission from the GTR.

18. What action will the National Institutes of Health (NIH) take if marketing materials state that NIH endorses a test?

Test submitters agree to abide by a code of conduct, which stipulates that submitters make no explicit or implicit claims that their tests listed in the GTR, or other information submitted to the GTR, have been approved or endorsed by NIH, the Department of Health and Human Services, or the U.S. Government. If this stipulation is not honored by a test submitter, NIH reserves the right to take action, including, in its sole discretion, requesting that the test submitter amend its marketing materials to delete any such claims or, if a submitter fails to respond to this request, removing the relevant submission from the GTR. Marketing materials may refer to the fact that test information is provided in the GTR.
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19. How will NIH assure the quality of the test information provided to the Genetic Testing Registry?

NIH does not verify the content of information submitted to the Genetic Testing Registry (GTR) by test providers. Submitters will be solely responsible for the content and quality of the data they provide to the GTR. However, NIH recognizes that it is critically important to ensure that GTR users understand that neither NIH, nor the government in general, endorses tests or has verified the accuracy of test information in the GTR. NIH implemented the following measures to ensure that these limitations are as clear as possible and also to hold submitters accountable for their responsibilities:

- The National Center for Biotechnology Information (NCBI) incorporates basic administrative checks for mistakes made by the submitter during the submission process.
- Submitters must agree to abide by the terms of a code of conduct and action will be taken, based on standard operating procedures, if submitters violate these terms.
- A prominent disclaimer on the GTR homepage clearly states that NIH does not independently verify information submitted to the GTR.
- NCBI assigns a unique accession number to each test, allowing uniform reference to tests in scientific publications; which facilitates third-party evaluations of GTR content. In addition, NCBI can link published reviews to tests referenced in publications.
- NCBI links to external resources such as professional practice guidelines and studies that support or refute claims made by submitters.
- GTR users can contact NCBI staff to report information that appears to be incorrect.

Stakeholder Involvement

20. How can I learn more about the Genetic Testing Registry and provide feedback?

The National Center for Biotechnology Information (NCBI) provides tutorials and information on how to use the Genetic Testing Registry (GTR). The Office of the Science Policy provides a collection of public comments, frequently asked questions, and other background information. Please use “Contact GTR” to provide comments or ask questions about the GTR.
21. How did stakeholder input help the National Institutes of Health shape the Genetic Testing Registry?

The National Institutes of Health (NIH) is appreciative of the thoughtful stakeholder feedback, which helped to define the scope of the Genetic Testing Registry (GTR), improve site navigation, enhance usability, and identify data elements important to potential GTR users as well as elements that would be difficult or burdensome for test developers to provide. Examples of changes based on stakeholder input are:

- NIH decided to use a phased approach in building the GTR, and stakeholder comments helped prioritize the types of tests that are included in the initial phase. Phase I includes single-gene tests for heritable mutations, including pharmacogenomic tests, and multiplex panels and arrays. Phase II includes tests for somatic mutations such as genetic variants in tumors and tests using whole-exome or whole-genome sequencing. Subsequent phases will add tests for infectious agents and direct-to-consumer genetic tests.
- NIH focused on health care providers as the intended audience for the initial phase of the GTR; future phases may expand the audience to include patients and the general public.
- NIH excluded certain data elements, at least from the initial phase of GTR, such as test price, turn-around time, and patent and licensing information.

22. Did the National Institutes of Health involve stakeholders during the development of the Genetic Testing Registry?

Yes, consulting with stakeholders—such as laboratory test developers, manufacturers, and health care providers—was a key step in the development of the Genetic Testing Registry (GTR), and continued stakeholder interaction will help optimize the GTR’s utility.

Stakeholder comments were solicited through the following means:

- Request for Information (RFI), Federal Register notice, June 11, 2010, 68 public comments.
- Public Stakeholder Meeting, November 2, 2010, 17 public comments and meeting discussion.
- Request for Comments, Federal Register notice, November 23, 2011, public comments.
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- Consultation with two GTR clinical advisory groups: four meetings with clinical experts from within NIH and six meetings of a medical genetics working group of the National Center for Biotechnology Information Board of Scientific Counselors in 2010-2011.
- Meetings with other government agencies that have an interest in genetic testing (e.g., the Food and Drug Administration and the Centers for Medicare & Medicaid Services), three meetings in 2010-2011.
- In-person meetings or teleconferences with stakeholder groups, 19 meetings in 2010-2011.
- Presentations and discussion at meetings of professional organizations, 7 meetings in 2010-2011.
- Comments submitted through “Contact GTR,” 95 comments received in 2010-2011.

23. Does the National Institutes of Health collaborate with other agencies in the Department of Health and Human Services that are interested in genetic test information?

Yes. NIH consults with the Food and Drug Administration and the Centers for Medicare & Medicaid Services (which enforces regulations based on the Clinical Laboratory Improvement Amendments) to streamline the process of data submission for test providers who may be required to provide similar information to these HHS agencies.

24. How was the Genetic Testing Registry designed?

The Genetic Testing Registry was designed by the National Institutes of Health (NIH) National Center for Biotechnology Information with extensive input from the stakeholder community, including clinicians and test providers, under the oversight of an NIH steering committee.

Paperwork Reduction Act

25. Does the Paperwork Reduction Act apply to the Genetic Testing Registry?

Yes. The Paperwork Reduction Act applies to the Genetic Testing Registry (GTR) because
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the National Institutes of Health, a federal agency, is collecting standardized information from 10 or more respondents (i.e., laboratories that provide genetic tests) within a 12-month period. The Paperwork Reduction Act requires two periods for the public to provide comments about the proposed information collection. You can read the public comments that were requested on July 27, 2011, and November 23, 2011.

26. What is the Paperwork Reduction Act?

The Paperwork Reduction Act (PRA) was passed by Congress in 1980 and requires that federal agencies receive Office of Management and Budget (OMB) clearance before requesting most types of information from the public (“information collections”). Specifically, PRA clearance is required when standardized information is collected from 10 or more respondents within a 12-month period. The OMB regulation that implements the PRA is at 5 CFR 1320. Additional PRA information is available from the Department of Health and Human Services.