



July 31, 2010

**Via Electronic Mail Submission to: [GTR@od.nih.gov](mailto:GTR@od.nih.gov)**

Francis S. Collins, Ph.D., Director  
National Institutes of Health  
GTR RFI Comments  
Office of Science Policy  
Room 750  
6705 Rockledge Drive  
Bethesda Maryland 20892

**RE: National Institutes of Health Request for Information (RFI) on the National Institutes of Health Plan To Develop the Genetic Testing Registry**

Dear Dr. Collins:

On behalf of the Coalition for Twenty-First Century Medicine (C21CM), we are pleased to submit comments in response to the above-captioned notice requesting information on the National Institutes of Health's plans to develop a Genetic Testing Registry (GTR). The C21CM has long-supported the development of a genetic testing registry and has also called for such a registry to be mandatory under the authority of the Food and Drug Administration (FDA) working collaboratively with the NIH as part of a new regulatory policy for oversight of advanced diagnostic tests that is based upon evolving scientific methods and findings, including those to be reported to the GTR.

C21CM represents innovative diagnostic technology companies, clinical laboratories, researchers, physicians, venture capitalists, and patient advocacy groups who support the development of advanced diagnostic tests that improve the quality of healthcare for patients.

We applaud the NIH for announcing its work to develop the GTR, which will serve as a repository of information about genetic testing. We look forward to working with NIH as it proceeds with development and implementation of the GTR. In response to the above RFI, we offer the following outline for the content of the GTR (see Appendix), which we have developed as a regulatory proposal based upon the established [clinicaltrials.gov](http://clinicaltrials.gov) registry. As you know, [clinicaltrials.gov](http://clinicaltrials.gov) is a registry of federally and privately supported clinical trials conducted in the United States and around the world that is maintained as a service by the NIH.

Thank you for the opportunity to submit these comments in response to your request. We look forward to working with NIH as it proceeds with the design and implementation of the GTR.

The Coalition for Twenty-First Century Medicine

## APPENDIX: Content of a GTR

1. BASIC INFORMATION.—As applicable, information to identify the test, as follows:
  - A. The name of the clinical laboratory introducing the test.
  - B. The brand or commercial name of the test under which such clinical laboratory offers the test.
  - C. The non-proprietary or common name for the test.
  - D. The intended use of the test.
  - E. The type of specimen examined by the test.
  - F. The date on which the test was first introduced by the clinical laboratory.
  
2. DATA TO ESTABLISH PERFORMANCE SPECIFICATIONS.—As applicable, information used to establish the performance specification for the test, for each of the following performance characteristics:
  - A. Accuracy.
  - B. Precision.
  - C. Analytical sensitivity.
  - D. Analytical specificity to include interfering substances.
  - E. Reportable range of test results for the test system.
  - F. Reference intervals (normal values).
  - G. Other performance characteristic for test performance determined by the NIH to be relevant based upon stakeholder input
  
3. DATA TO SUPPORT CLINICAL VALIDITY.—For tests for which the intended use includes a claim of the clinical meaning or usefulness of the analytical result, information submitted to the GTR should include that sufficient to establish the clinical validity of the test, as follows:
  - A. Demographic characteristics of the patients who participated in any clinical study conducted to establish the clinical validity of the test, including the characteristics of patients whose specimens were collected for examination in the clinical study.
  - B. Primary and secondary endpoint measures included in any clinical study conducted to establish the clinical validity of the test, including the values for each measure and the results of scientifically appropriate tests of the statistical significant of such endpoint measures.
  - C. A contact person for obtaining scientific information about the clinical validity data.

4. EXPANDED REGISTRY DATA.—

IN GENERAL.—To provide more complete results information and to enhance patient access to and understanding of the results of clinical studies used to establish the clinical validity of genetic tests, based upon stakeholder input, the GTR should include the following expanded information:

A. A summary of the clinical study(ies) conducted to establish the clinical validity of the test, including the results, that is written in a non-technical manner that is calculated to be understood by the average person, if the NIH determines that such types of summary can be included in the GTR without being misleading or promotional.

B. A summary of the clinical study(ies) conducted to establish the clinical validity of the test, including the results, that is technical in nature, if the NIH determines that such types of summary can be included in the GTR without being misleading or promotional.

C. The full protocol or such information on the protocol for the study as may be necessary to help evaluate the results of the study.

D. Such other categories of information as the NIH determines appropriate based upon stakeholder input.

5. PROPRIETARY INFORMATION.—The NIH should establish a process to allow for submitters to the GTR to redact confidential information that the submitter determines is proprietary and confidential in nature.