

# Genetic Testing Registry

**Clinical Laboratory Improvement  
Advisory Committee  
March 2, 2011**

Cathy Fomous, Ph.D.  
NIH Office of Biotechnology Activities

# Genetic Testing Registry

- ▶ Why develop a genetic testing registry?
- ▶ Development steps (January 2010-January 2011)
- ▶ Stakeholder comments
- ▶ Moving forward
- ▶ For more information

# The Call for a Test Registry



## U.S. System of Oversight of Genetic Testing: A Response to the Charge of the Secretary of Health and Human Services

Report of the Secretary's Advisory Committee  
on Genetics, Health, and Society

SACGHS recommended that HHS establish a test registry to increase the transparency of genetic testing.

Enhancing the transparency of information about genetic tests is a key prerequisite to improving oversight.

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.

Public Health  
Genomics

### Review

Public Health Genomics 2010;13:95-105  
DOI: [10.1186/108926699](https://doi.org/10.1186/108926699)

Received December 11, 2008  
Accepted after revision: April 11, 2009  
Published online: June 28, 2009

## Developing the Blueprint for a Genetic Testing Registry

G. Javitt S. Katsanis J. Scott K. Hudson

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GENETIC TESTING AND MOLECULAR BIOMARKERS  
Volume 13, Number 2, 2009  
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Pp. 153-154  
DOI: 10.1089/gmb.2009.1303

### Call for Action from Genetic Alliance

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

Kristi D. Zonno and Sharon F. Terry

# Congressional Interest

## House launches investigation into genetic tests

By [Rob Stein](#)

Washingon Post Staff Writer

Wednesday, May 19, 2010; 3:43 PM

A congressional committee Wednesday launched an investigation into genetic tests being sold directly to consumers.

The House Energy and Commerce Committee and its subcommittee on oversight and investigations sent letters to Pathway Genomics Corp. of San Diego, 23&Me Inc. of Mountain View, Calif., and Navigenics Inc. of Foster City, Calif., requesting information about their tests. The move was prompted after Pathway announced plans last week to sell its genetic test through drug stores nationwide for the first time "despite concern from the scientific community regarding the accuracy of test results," the letters stated.

Walgreens, the nation's largest drug store chain, announced it was postponing plans to sell the test, however, after the Food and Drug Administration

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May 19, 2010

Ms. Anne Wojcicki  
President  
23andMe, Inc.  
1390 Shorebird Way  
Mountain View, CA 94043

Dear Ms. Wojcicki:

The Committee on Energy and Commerce and its Subcommittee on Oversight and Investigations are examining personal genetic tests sold to consumers over the Internet. Recent press reports suggest that at least one genetic testing company is now seeking to sell these tests in retail locations, despite concern from the scientific community regarding the accuracy of test results.<sup>1</sup>

In order to assist the Committee with its examination of this issue, we ask that you provide the Committee with the following information and documents for the period from January 1, 2007, to the present:

1. A chart listing the conditions, diseases, consumer drug responses, and adverse reactions for which you test;

# FDA Activities

FDA activities related to genetic testing include

- ▶ May 2010 letter to Pathway Genomics—noted product appears to meet definition of device, requested clearance/approval #
- ▶ June 2010 letters to 5 DTC companies—requested companies to work with FDA to determine which claims required oversight
- ▶ June 2010 public meeting on array-based cytogenetic tests—gather information on questions related to review challenges
- ▶ July 2010 public meeting on oversight of laboratory-developed tests (LDTs)—gather stakeholder perspectives on LDT oversight
- ▶ March 2011 meeting of FDA Molecular and Clinical Genetics Panel of the Medical Devices Advisory Committee—discuss scientific issues concerning DTC genetic tests

# NIH Is Building a *Voluntary* Genetic Testing Registry



[NIH Office of the Director \(OD\)](#)

Embargoed for Release  
Thursday, March 18, 2010  
10 a.m. EDT

Contact:  
[NIH Office of Communications](#)  
301-496-5787 NIH Office of Communications

## NIH Announces Genetic Testing Registry

*Database to Fill Information Gaps and Serve as Research Resource*

To create a single public source of comprehensive information about genetic tests



The NEW ENGLAND JOURNAL of MEDICINE

Perspective  
JULY 22, 2010

## The Path to Personalized Medicine

Margaret A. Hamburg, M.D., and Francis S. Collins, M.D., Ph.D.

To improve research and public health through:

- Increasing transparency
- Increasing physician and researcher access to information

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### Introduction to the Genetic Testing Registry

About GTR  
News and Events  
Timeline  
Meetings

On March 18, 2010, the National Institutes of Health (NIH) announced its plan to develop the Genetic Testing Registry (GTR) to provide a centralized online resource for information about the availability and scientific basis of genetic tests. NIH expects that the GTR will be publicly available in 2011. For additional information, please refer to [About GTR](#) and the [FAQs](#).

# GTR Development Steps

January 2010

- ▶ Meeting with FDA and CMS
- ▶ GTR website, mailbox, and listserv created
- ▶ Announcement of GTR plan
- ▶ CDC, AHRQ, CMS, and FDA provided input on the draft RFI

June

- ▶ RFI public comment period
- ▶ Ongoing meetings with stakeholder groups

# GTR Policy Development Steps (continued)

August

- ▶ Analysis of RFI comments, planning of public stakeholder meeting
- ▶ Meeting of NIH clinical advisors to the GTR (October)
- ▶ Public stakeholder meeting (November 2) in conjunction with the ASHG annual meeting

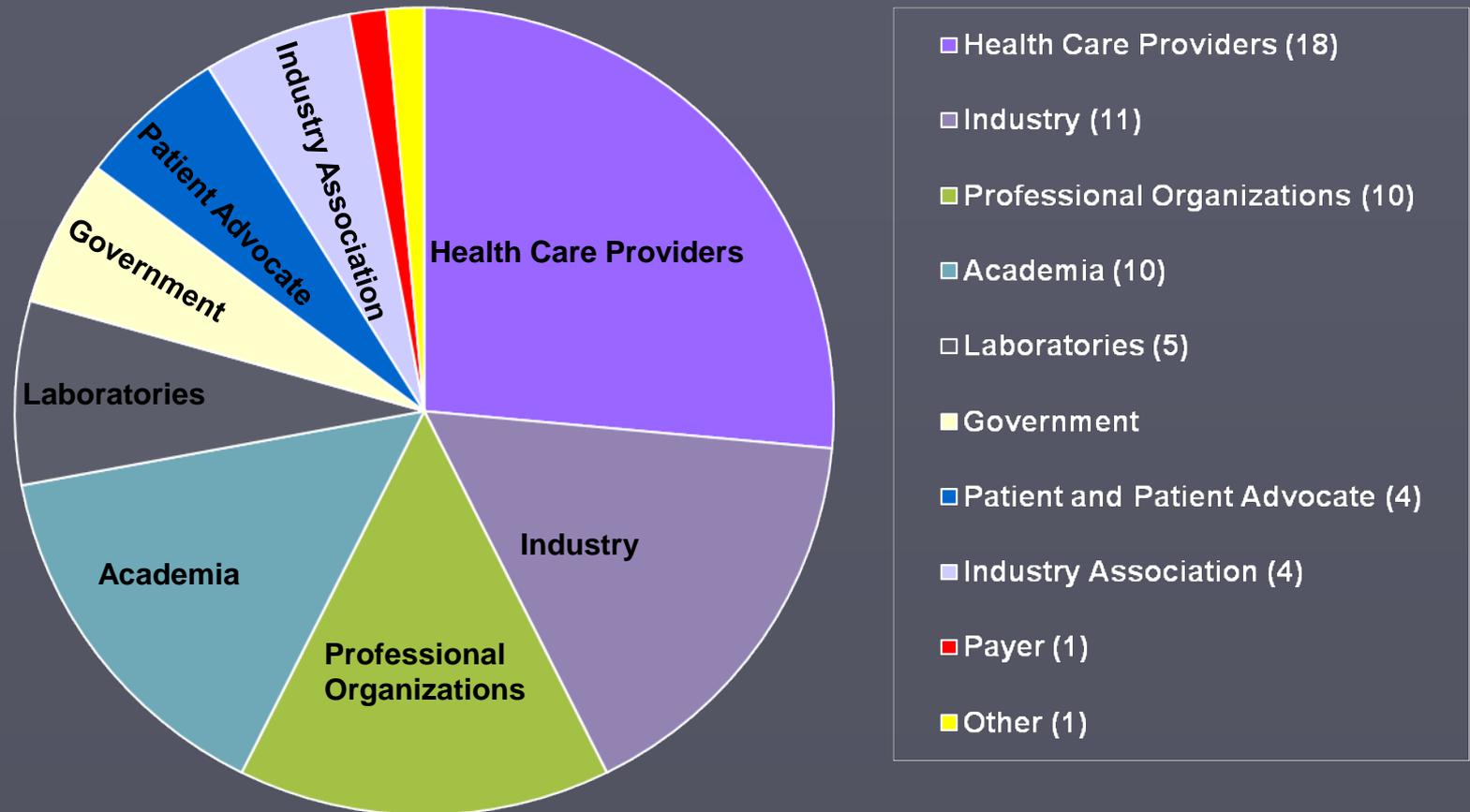
January 2011

- ▶ Meetings of the NCBI BSC Medical Genetics Working Group (November and December)
- ▶ Meeting of NIH clinical advisors to the GTR (January)

# GTR RFI Questions

- ▶ Critical data elements for various stakeholder groups?
- ▶ Potential benefits and risks of wider access to validity and utility information?
- ▶ Value of specific data elements?
- ▶ Information that will be difficult to provide?
- ▶ Advantages and disadvantages of capturing the molecular basis of the test?
- ▶ Information resources that should be provided?
- ▶ Processes to facilitate data submission?

# RFI Responses (n=68)



# RFI Responses – General Themes

- ▶ Overall, comments were supportive of the GTR concept
- ▶ General agreement with most of the proposed data elements
- ▶ Need for educational materials to define/explain data elements
- ▶ Potential uses of the GTR include
  - Determining test availability
  - Identifying laboratories to confirm research results
  - Facilitating research (e.g., identifying potential collaborators)
  - Learning about specimen requirements and test limitations

# RFI Responses – Concerns

- ▶ Critical to ensure accuracy of information in the GTR; users will assume test information on an NIH-sponsored website is accurate and valid
- ▶ Potential harm to patients if the information in GTR is inaccurate/incomplete or is misunderstood or misinterpreted (e.g., inappropriate testing, denial of insurance coverage)
- ▶ Some data elements (e.g., clinical utility, cost) will be difficult to provide, particularly for rare diseases

# RFI Responses – Concerns (continued)

- ▶ GTR could contribute to increased demand on the health care system and health care spending
- ▶ GTR should include only those tests with high sensitivity and specificity and well-established clinical validity
- ▶ Data submission will be time consuming, especially for small niche laboratories; challenging to keep data up to date

# Nov. 2 Stakeholder Meeting

## Focus Questions

1. If NIH adopts a phased approach to build the GTR, what criteria should be used to determine which genetic tests should be included in the first phase of the GTR, and what types of tests would meet these criteria?
2. Given that data submitters are unlikely to have clinical utility information, how is this data element best addressed in the GTR?
3. What are the benefits, risks, and challenges of including cost information in the GTR?
4. What safeguards can be put in place to prevent GTR users from misunderstanding, misinterpreting, or misusing the information in the Registry?
5. What mechanisms can be used to provide materials that explain the GTR's data elements to audiences with varying technical expertise?

# Nov. 2 Stakeholder Meeting: Comments

17 public comments (13 oral and 4 written)

- Professional organizations (7)
- Academia (3)
- Health care providers (2)
- Industry/Industry associations (2)
- Patient/Patient advocacy (2)
- Laboratory professional (1)

# Nov. 2 Stakeholder Meeting: Comments

General themes of responses to the 5 focus questions:

1. Phased approach: general agreement
  - Wide range of what to include in pilot
2. Inclusion of clinical utility: agreement that it is difficult
  - Provide guidance for submission, use existing resources
3. Inclusion of test price: strong divisions
  - Yes: better for patient
  - No: difficult for laboratories to provide
4. Safeguards against misuse:
  - Provide test limitations, use disclaimers
5. Mechanisms to explain data elements to different audiences:
  - Clarify intended audience, pilot test with end users

# The GTR is Coming into Focus

NIH will use a phased approach in building the GTR

- ▶ Initial phase will include
  - Single-gene tests for Mendelian disorders
  - Pharmacogenomic tests
  - Test panels
- ▶ Initial target audience is health care providers
- ▶ Some data elements proposed in the RFI may not be included, at least not in the initial phase
  - Test price
  - Turn-around time
  - CPT codes
  - Patent information

# Likely Phase I GTR Data Elements

- ▶ Laboratory and Personnel Information
  - Types of laboratory services, website URL, CLIA or other certification/licensure, contact information of laboratory personnel
- ▶ Test Information
  - Name and purpose of test
  - Test methodology and analytes
  - Analytic validity
  - Quality control and assurance (e.g., proficiency testing),
  - FDA review (e.g., cleared, approved, not required)
  - Clinical validity,
  - Clinical utility

# Next Steps

- ▶ Continue engagement with FDA, CMS, CDC, AHRQ
- ▶ Maintain dialog with stakeholders
- ▶ Gather feedback on proposed data elements
- ▶ Develop user interface and beta test
- ▶ Expected GTR launch in fall 2011
- ▶ Analyze usage after GTR launch

# GTR Website and Contact

- ▶ GTR website: <http://oba.od.nih.gov/gtr/gtr.html>
- ▶ GTR mailbox: [GTR@od.nih.gov](mailto:GTR@od.nih.gov)