

# **Overview of Public Comments in Response to NIH's Request for Information on the Genetic Testing Registry**

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**NIH Public Meeting on the  
Genetic Testing Registry**

**November 2, 2010**

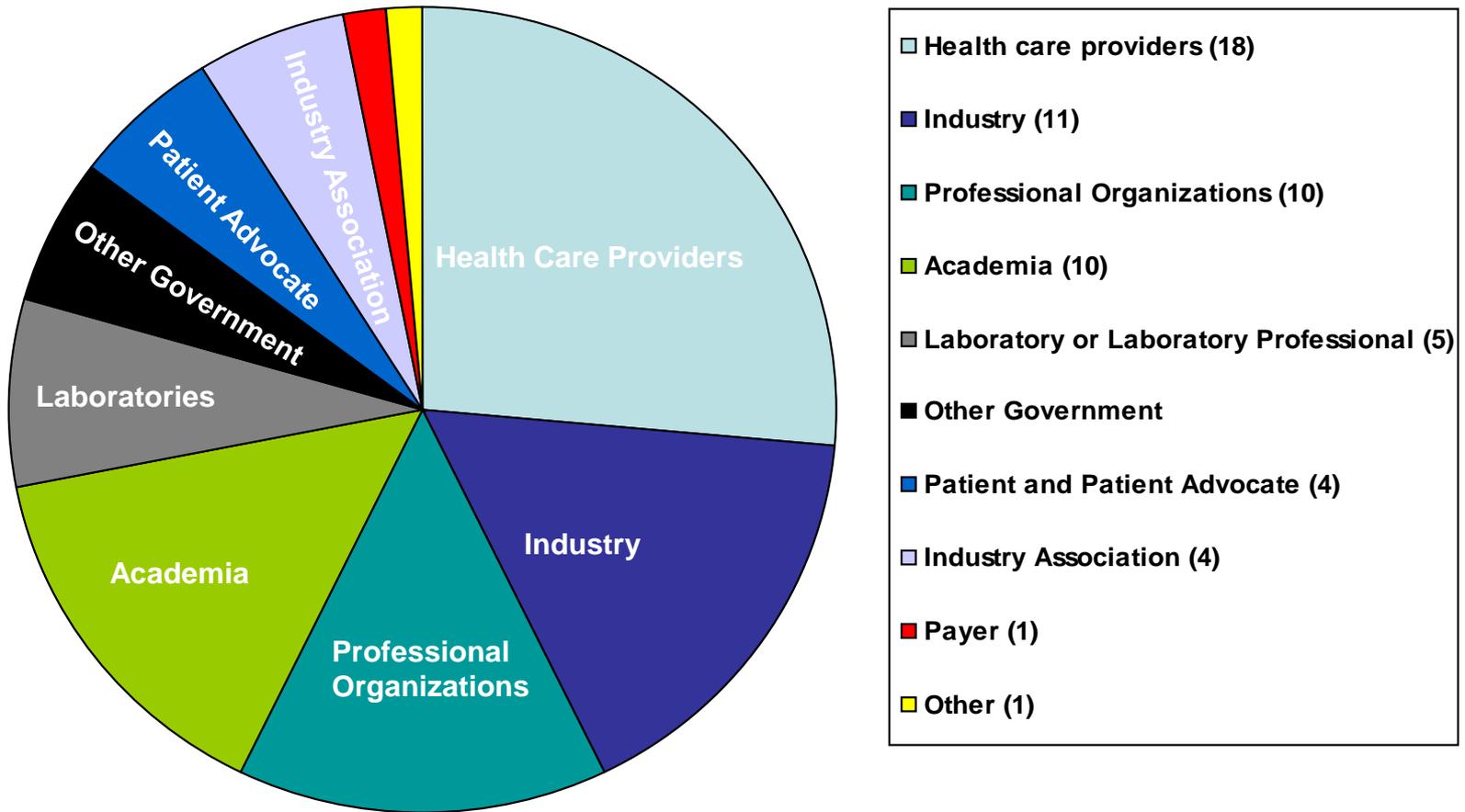
# Questions in the Request for Information (RFI)

- Scope of the registry?
- Potential uses?
- Critical data elements for various stakeholder groups?
- Potential benefits and risks of wider access to availability, validity, and utility information?
- Best way to distinguish between data fields left blank because of no known data vs submitter not providing information?
- Value of specific data elements?
- Information that will be difficult to provide?

# RFI Questions (continued)

- Advantages and disadvantages of capturing the molecular basis of the test?
- Information resources that should be provided?
- Processes to facilitate data submission?
- Potential benefits and risks that will affect data submission decisions?
- How to ensure continued stakeholder input?
- How will you use the GTR?
- Other issues?

# 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 each data element
- Potential uses of the GTR include
  - Determining test availability
  - Identifying laboratories to confirm research results
  - Learning about specimen requirements and test benefits/limitations
  - Facilitating research (e.g., identifying potential collaborators)

# 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
- Need for the GTR is not obvious when a voluntary registry hosted at NCBI (GeneTests) already exists

# 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

# Public 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 [i.e., price] 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?

# Written Comments on the Five Focus Questions

- Comment period closes November 12, 2010
- Submit written comments on to Cathy Fomous:
  - Email: [cfomous@od.nih.gov](mailto:cfomous@od.nih.gov)
  - Fax: 301-496-9839
  - Postal service: 6705 Rockledge Dr., Suite 705  
Bethesda, MD 20892

# Next Steps

- Analyze comments from public meeting
- Continue engagement with FDA, CMS, CDC, AHRQ
- Maintain dialogue with stakeholders
- Convene focus groups to provide feedback on GTR prototypes
- Develop registry and beta test
- Expected GTR launch in Spring 2011
- Analyze usage after GTR launch