

**From:** Rousseau, Julie [mailto:jrousseau@chla.usc.edu]  
**Sent:** Friday, June 04, 2010 11:43 AM  
**To:** Genetic Testing Registry (NIH/OD/OSP)  
**Subject:** Comments

As a clinical genetic counselor, I am both intrigued by and apprehensive about this registry. I do believe that having a centralized reference for clinicians and researchers is highly valuable, in the way that we have been using GeneTests for years. I hope that the NIH will continue to support GeneReviews and the clinic directory as these are very helpful to clinicians, particularly those who are not trained in genetics and who are increasingly ordering genetic tests. I believe that the registry should include sensitivity, specificity, etc, as well as the clinical utility, much in the way that GeneReviews currently does, particularly if the anticipated audience includes clinicians not trained in genetics. Finally, clinical staff is crucial to implementation of this registry in order to verify the accuracy of the information provided by the laboratories. Without this component, the registry has the potential to become more of a free advertisement than a compilation of validated laboratories. Also, as methodologies, known genes, etc, change over time, this registry should be updated regularly.

Below, I have answered some of the questions posed on the website:

1. Are there any types of genetic tests that should not be included in the GTR? No
2. What are the potential uses of the GTR for (1) researchers, (2) patients/consumers, (3) health care providers, (4) clinical laboratory professionals, (5) payers, (6) genetic testing entities/data submitters, (7) policy makers, and (8) electronic health records? For clinicians and researchers, this allows for identification of laboratories performing specific tests to facilitate ordering. For clinical lab professionals, it could provide the opportunity to receive more samples as more providers will be aware of the tests offered and also offer the opportunity to determine which tests to bring in-house as there are some tests performed by many labs and some performed only by one or two, making it more cost effective (potentially) to bring the latter in-house. For payers, being able to confirm that the reason a sample is being submitted to a particular out-of-network laboratory is because it is the only laboratory performing that test may allow for more confidence in their decisions and for fewer denials and appeals.
3. What data elements are critical to include for use by (1) researchers, (2) patients/consumers, (3) health care providers, (4) clinical laboratory professionals, (5) payers, (6) genetic testing entities/data submitters, (7) policy makers, and (8) electronic health records? The specific methodology used. The availability of tiered/sequential testing. Sensitivity and specificity. Samples types accepted.
4. What are the potential benefits and risks associated with facilitating public access to information about the:
  - a. Availability and accessibility of genetic tests? May encourage non-genetics clinicians and the general public to refer/self-refer to genetics clinicians more readily as they understand that more is available now than previously.

- b. Scientific basis and validity of genetic tests? Critical as this hopefully will document clearly where the data are generated and whether or not they are evidence-based.
  - c. Utility of genetic tests? Perhaps this will educate non-genetics clinicians and even the general public about the utility of genetic tests as the complexity is generally not well understood.
5. What is the best way to distinguish between data fields left blank because of an absence of data/evidence and those left blank for other reasons? How important is this distinction for enhancing transparency, including for the purpose of identifying research opportunities? If no data entered, should have an explanation of why.
6. To adequately and accurately describe a genetic test, which of the following data elements should be included in the GTR? Are there other data elements that should be added? What information is necessary to represent adequately each data element?
- a. Contact information (e.g., location, name of the laboratory director, and contact information for the laboratory performing the test) – add genetic counselors' names and contact information
  - b. Laboratory certifications (e.g., Federal or State certification of the laboratory that performs the test)
  - c. Name of the test (e.g., common test name, commercial name, marketing materials about the test and/or genetic testing entity, standard identifier (e.g. CPT codes, LOINC<sup>1</sup>))
  - d. Regulatory clearances (e.g., for tests reviewed by the Food and Drug Administration, the 510(k) or premarket approval (PMA) number)
  - e. Intended use of the test (e.g., diagnosis, screening, drug response)
  - f. Recommended patient population – this is a clinical decision and laboratories often recommend testing a larger population than appropriate. If this is included, it should be written by clinical staff of the GTR or a link should be provided to GeneReviews.
  - g. Limitations of the test (e.g., is the test validated only for certain subpopulations or limited to particular uses such as screening but not diagnostic testing?)
  - h. Test methodology
  - i. Analyte(s)—What is being measured in the test (e.g., genetic sequence)
  - j. Specimen requirements (e.g., blood, saliva, tissue samples, amniotic fluid)
  - k. Availability (e.g., is the submitter the sole provider of the test or are there multiple providers?) - this should not be provided by the laboratory, but verified by the clinical staff of the GTR
  - l. Accessibility (e.g., accessible through a health provider, public health mandate, and/or direct-to-consumer)
  - m. Performance characteristics<sup>1</sup>
    - i. Analytical sensitivity

- ii. Analytical specificity
  - iii. Accuracy
  - iv. Precision
  - v. Reportable range of test results
  - vi. Reference range
  - vii. Method used for proficiency testing (e.g., formal PT program, alternative assessment) and score
  - n. Clinical validity<sup>i</sup>
    - i. Clinical sensitivity
    - ii. Clinical specificity
    - iii. Positive and negative predictive value
    - iv. Prevalence
    - v. Penetrance
    - vi. Modifiers
  - o. Utility (e.g., clinical and/or personal utility) or outcomes – this is a clinical determination. Should not be provided by the laboratory, but by clinical staff at the GTR or by linking to GeneReviews
    - i. Benefits
    - ii. Harms
    - iii. Added value, compared with current management without genetic testing
  - p. Cost (e.g., price of the test, health insurance coverage) – health insurance coverage is highly variable; therefore, generalizations are impossible. If cost is to be included, frequent updates will be required to ensure accuracy – may be more efficient to link to laboratory's website for this information.
7. What types of information might be difficult for test providers to submit and why? – information about clinical utility, when the test is appropriate, insurance coverage information.
8. What are the advantages and disadvantages of collecting and providing information on the molecular basis of genetic tests, such as detailed information about what the test detects and the specific methods employed? – advantages are that there may be a greater understanding on the part of non-genetics clinicians. Disadvantages are that this may continue to be poorly understood by and therefore provide for increased confusion on the part of non-genetics clinicians.

9. In addition to the data elements, would it be helpful to reference other resources, and if so, which ones (e.g., published studies, recommendations from expert panels such as the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children, U.S. Preventive Services Task Force, or Evaluation of Genomic Applications in Practice and Prevention Working Group)? - Very helpful to reference resources (published studies; recommendations from expert panels; society policy statements, esp NSGC, ACMG)

10. As the GTR is being designed, what are the important processes to consider to make the submission of data as easy as possible for the data provider (e.g., the capability of linking to information that has been submitted to other agencies, such as the Food and Drug Administration and the Centers for Medicare and Medicaid Services, or a master file of data common to particular tests)? – I defer to the laboratory personnel

11. Which potential benefits and risks would be most likely to affect the decisions of researchers, test developers, and manufacturers on whether to submit data to the GTR, and what factors will best encourage submission of complete and accurate data? – if this truly replaces GeneTests in utility, not being listed in the GTR will lead to a great reduction in samples

12. What are the most effective methods to ensure continued stakeholder input into the maintenance of the GTR? – provide as good or better support than is currently available through GeneTests

13. For what purpose(s) would you use the Registry to support your professional efforts? – assuming accuracy and review by clinical staff at GTR, I would use the registry to determine to which lab to send a sample for a patient.

14. Are there any other issues that NIH should consider in the development of the GTR? Readability, accessibility, and accuracy are critical.

I look forward to seeing how this develops and hope that it proves to be as useful a resource as it has the potential to be.

Julie A. Rousseau, MS CGC

Certified Genetic Counselor

Childrens Hospital Los Angeles

Division of Medical Genetics

Phone: (323) 361-4743

Fax: (323) 361-1172

Pager: (213) 203-1392

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