

**From:** Janice Rinsky [mailto:janice.rinsky@hsc.utah.edu]  
**Sent:** Thursday, June 03, 2010 5:01 PM  
**To:** Genetic Testing Registry (NIH/OD/OSP)  
**Subject:** Request for input-Genetic test registry

Hi,

I am a prenatal genetic counselor who frequently uses GENETESTS to find a laboratory that performs a particular test. It is an essential resource in my practice, especially when there is a test I need to order that I don't order very frequently. I, like many other genetic counselors, would have a much more difficult time if the information provided by GENETESTS were no longer available. Some of the essential components are:

What labs are offering specific testing, and how are they offering it?, IE-carrier testing, prenatal diagnosis, gene sequencing, etc. A link to the lab's website, as well as gene reviews on the condition in question is also very useful. Please make sure the new GTR includes these elements. I will comment below on what I feel qualified to comment on. I hope you find this useful in the development of the GTR.

1. Are there any types of genetic tests that should not be included in the GTR? I believe that if direct to consumer genetic tests are included, they should be put in a separate category, since they are ordered, and used in settings that are not clinical.
2. What are the potential uses of the GTR for (1) researchers-finding collaborators, (2) patients/consumers-Help them to find if a genetic test is available for their particular disorder, (3) health care providers-Finding a lab that will perform the test in question, having access to their contact and billing information, specimen requirements, sensitivity, and specificity. , (4) clinical laboratory professionals, (5) payers, (6) genetic testing entities/data submitters, (7) policy makers, and (8) electronic health records?
3. What data elements are critical to include for use by (1) researchers, (2) patients/consumers, (3) health care providers,-Same as currently present in gene tests. It would also be great to see CPT codes, billing information, sensitivity and specificity as required on their website link. (4) clinical laboratory professionals, (5) payers, (6) genetic testing entities/data submitters, (7) policy makers, and (8) electronic health records?
4. What are the potential benefits and risks associated with facilitating public access to information about the:
  - a. Availability and accessibility of genetic tests? Benefit-if they see a test is available, may argue with their insurance company to cover the cost.
  - b. Scientific basis and validity of genetic tests? Risk-very easy to misinterpret data.
  - c. Utility of genetic tests? Risk-very easy to misinterpret data or make assumptions about what a negative, or positive result might mean.
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? Instead of leaving a field blank, write in the explanation, ie-not available on a clinical basis, research trials being conducted, contact XYZ
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) Yes
  - b. Laboratory certifications (e.g., Federal or State certification of the laboratory that performs the test) Yes

- 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>ii</sup>)) [Yes](#)
  - d. Regulatory clearances (e.g., for tests reviewed by the Food and Drug Administration, the 510(k) or premarket approval (PMA) number) [Yes](#)
  - e. Intended use of the test (e.g., diagnosis, screening, drug response) [Yes](#)
  - f. Recommended patient population [Yes](#)
  - 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?) [Yes](#)
  - h. Test methodology [Yes](#)
  - i. Analyte(s)—What is being measured in the test (e.g., genetic sequence) [Yes](#)
  - j. Specimen requirements (e.g., blood, saliva, tissue samples, amniotic fluid) [Yes](#)
  - k. Availability (e.g., is the submitter the sole provider of the test or are there multiple providers?) [Yes](#)
  - l. Accessibility (e.g., accessible through a health provider, public health mandate, and/or direct-to-consumer) [Yes](#)
  - m. Performance characteristics<sup>i</sup> [Yes](#)
    - 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> [Yes](#)
    - 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 [Yes](#)
    - 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) [Yes](#)
7. What types of information might be difficult for test providers to submit and why? [Insurance coverage, availability, possibly accessibility](#)
  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? [This aids providers when making a choice about what test is most appropriate in each specific situation.](#)
  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)? [GENE REVIEWS, OMIM, link to published journal article \(?\)](#)

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)?
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? *Make it as easy as possible to submit data, have data that is in other sources (is-GENETESTS) automatically transfer to the new GTR.*
12. What are the most effective methods to ensure continued stakeholder input into the maintenance of the GTR? *Contact, automatically and quarterly, all entities that have input data asking them to review their information for updates, additions, etc. Make it easy to put in the updates. Make it free of charge.*
13. For what purpose(s) would you use the Registry to support your professional efforts? *The same as I do now.*
14. Are there any other issues that NIH should consider in the development of the GTR? *I believe there should be a method of screening entities that want to register with the GTR. There are some laboratories that make false claims, and the science used is questionable. I believe there should be some sort of peer review process, or genetics professional on board to review submissions. Perhaps require certain certifications by the lab, it's director, and proof by scientific and published data of what is being submitted. This way, if the lab claims certain sensitivities and specificities for the tests they are offering, there is valid information to back up their claim. Consumers using the GTR may assume that if a test is posted on an NIH sponsored website, it is automatically beneficial and the claims made by the lab are true. They may not understand PPR, specificity, sensitivity, etc. I often have patients call me asking if certain claims they read on a website that appear valid and professional are true, and often it is put up by a company that has little or no scientific data to back what is markets. (for example, stem cell therapy as a "cure" for Down syndrome).*

Thank you for taking the time to read and consider these suggestions. Please contact me if you have any questions.

Sincerely,

Janice

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