

Submission re:

Request for Information (RFI) on the NIH Plan to Develop the Genetic Testing Registry  
Notice Number: NOT-OD-10-101

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I will comment on the following two questions:

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?

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?

As medical information becomes more computerized, interconnectivity of information resources becomes a very central issue, as we detail in a forthcoming chapter in the Swaiman “Pediatric Neurology: Principles and Practice” textbook (Segal and Leber 2010). In our SimulConsult decision support software, currently focused mostly on neurogenetics, we cover over 2,200 diagnoses and have over 3,400 links to resources about diseases, clinical findings, and lab tests. As a clinician gets advice to consider various tests, it is very helpful to get straightforward access to information about genetic testing that is available in a way that is not confusing to the clinician. The more than those running a Genetic Testing Registry are familiar with such workflow, the more helpful a GTR can be in optimizing clinical care.

The GeneTests listings have been quite helpful in this context, but our experience leads us to stress further data elements and organization that would make such resources more useful to clinicians.

- **Listing on each Web page the specific type of test done, because for some diseases there is more than one abnormal result that can be measured.** If a GTR Web page is devoted to a disease, it should give more than just the title of the disease and a list of labs testing for “the gene”, “the analyte”, or “the enzyme”. It should spell out which specific tests are offered. This is important because in some cases there is more than one abnormality in a disease – for example more than one gene leading to the condition or more than one analyte being abnormal. A clinician might arrive at a disease page by following a hyperlink from decision support software and be confused about which test is being offered and even confused about whether they have arrived at the correct Web page. Similarly, it is important to be able to search a GTR for the name of the gene and find the correct page.
- **A separate Web page for each type of test, not just for each disease, because for some genes abnormalities lead to more than one disease.** Certain genes are abnormal in more than one disease and, as a result, in diagnostic software abnormalities in the gene might be listed as a single finding present in several

diagnoses. Although it would be possible in diagnostic software to list multiple tips for such findings, leading to pages about the various relevant diseases, it would make more sense to centralize this information in the GTR and have a page with a listing for each gene for which tests are available, and if appropriate, links from there to pages organized by disease in which the gene is abnormal. With advances in gene sequencing it is likely that sequencing an entire gene will replace the hit-or-miss process of individual tests for known common variants, so the distinctions often made today of different types of tests on the same gene will become less important. For this reason, clinicians and decision support software are likely to be increasingly focused on the abnormal gene as a whole, and represent that as a single finding, making it important to have a URL collecting all the information about a particular gene and its abnormalities in various diseases.

- **A full list of all pages about gene tests.** For GeneReviews articles, it is possible to get a complete list of these disease narratives, but for GeneTests listings, there seems to be no way to get a full list other than by searching for entries starting with each letter of the alphabet and each number, a process with much duplication. It would be helpful to have a list of all pages about gene tests, assisting those preparing decision support software in connecting their lists of genes (as well as enzymes and analytes) to GTR information.

Capabilities of this sort would assist in the usefulness of a GTR as accessed from diagnostic decision support software.