

From: David Hyman [mailto:dhyman@thegeneticscenter.com]
Sent: Wednesday, June 09, 2010 4:16 PM
To: Genetic Testing Registry (NIH/OD/OSP)
Subject: Comments on NOT-OD-10-101 Notice to develop a Genetic Testing Registry

As a clinical geneticist in private practice, I regularly consult a genetic testing registry for assistance in providing optimal care to my patients. To date, this need has been admirably fulfilled by the GeneTests resource coordinated by Dr. Roberta Pagon. However, I understand that, in order to address needs of other constituencies, an expansion of the existing resource is being contemplated. I feel that this is a very important endeavor.

I propose that, what should be implemented is an *extension of the existing GeneTests resource*, to, as stated in the RFI, "...provide a centralized location for researchers, test developers, and manufacturers to voluntarily submit information about genetic tests such as their intended use, validity, and utility." GeneTests already provides a centralized location for information relevant to clinicians and laboratorians on availability of testing and the diseases for which testing is available. The existing system is well organized and integrated with GeneReviews (expert reviews in a standardized format) and OMIM (highly technical, and highly useful information on specific diseases and genes).

At present, GeneTests is organized by disease and by specific gene. It should be relatively easy for a developer of the GTR to work with Dr. Pagon and her colleagues at GeneTests to add functionality that will organize the existing data in a way to allow for the additional information on validity, utility, methodology to be retrieved by an interface that exists and is already familiar to most GTR stakeholders. Specific comments to the Request for Comments are as follows:

1. Genetic testing that is not related to a disease or condition referenced in the GeneReviews or OMIM databases should not be included. Or, to put it another way, if genetic tests meet reasonable criteria for inclusion in the GTR, there should be corresponding clinical and scientific references in GeneReviews and/or OMIM.
3. Information on cost of testing might be a useful addition for patients and health care providers. Additional information relevant to that constituency might include turnaround time, specific sample/sample transport requirements, and billing arrangements. Already, GeneTests provides contact information to the laboratory, from which much of this information can be accessed. However, it should be possible to more tightly integrate that information which is on the test providers' web sites with the GeneTests search functions.
6. I believe GeneTests already includes items a,b,c; adding d (regulatory information) will be helpful. Intended use of the test (e) is addressed in GeneReviews, and the GTR might add an additional option for searching by intended use. GeneReviews addresses 6e, 6f, 6g (issues that are really not amenable to simplification in a database format). Test methodology (6h) could be organized by GeneTests under more detailed categories than at present, which

should help some stakeholders. I see absolutely no need for 6(k), "availability;" this is information that will be in the database, so no submissions by individual test providers is necessary. [For instance, what if one provider asserts being a sole source, but the database contains other providers? How would this data inconsistency be identified and/or resolved without an extremely cumbersome process?] For most genetic testing, performance characteristics can be very difficult to determine. Rather than add any of the information in 6(m, n, o, and p), consideration should be given to enhance the standardized GeneReviews format for molecular testing (which currently includes Test Methods, specific types of mutations identified, and mutation detection frequency).

9. GeneReviews disease entries already reference other resources, and this is a "curated" set of references, rather than one provided by the individual test provider(s).

10. To make the data submission process simple, continuing to have test providers use GeneTests is the most straightforward method.

11. If submissions to the GTR will be "curated" by individuals responsible for updating GeneReviews and GeneTest, this will ensure that submissions are factually accurate and complete.

12. Already, GeneTests has an easy way to be contacted on the home page, and this should be the most effective method to "ensure continued stakeholder..."

In summary, the existing GeneReviews/GeneTests/OMIM database addresses a majority of the needs anticipated for the GTR. Adding the required additional functionality to these databases is far preferable to initiating a new, duplicative database, which could possibly be biased by commercial and economic considerations. I strongly urge the NIH to direct funds for the GTR to expand the scope of GeneTests and GeneReviews.

Sincerely,

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