

From: Maureen Bocian [mailto:mebocian@uci.edu]
Sent: Tuesday, June 08, 2010 12:44 PM
To: Genetic Testing Registry (NIH/OD/OSP)
Subject: RFI on the NIH Plan to Develop the Genetic Testing Registry

RE: RFI on the NIH Plan to Develop the Genetic Testing Registry

I am a medical geneticist, certified by the American Board of Medical Genetics in clinical genetics and clinical cytogenetics and practicing in an academic medical center.

I am very concerned about this plan in several respects:

(1) I am strongly opposed to any plan that might adversely affect GeneTests (GeneReviews) in any way. This is an extremely valuable resource, utilized daily not only by me but also by our entire professional staff and our trainees in medical genetics and genetic counseling. The principal investigator, editors, and editorial staff are outstanding, and this new Plan should not be designed to replace or interfere with GeneTests(GeneReviews) or the integration of links to genetic testing information into its Reviews

(2) Many or most high-quality genetic testing laboratories have much of the information in your Plan already listed on their websites. Some, such as GeneDx and the Greenwood Genetic Center, have outstanding information sheets that contain most of the important information, plus CPT codes (which you do not have in your Plan) in an easy-to-read, print-able format. GeneTests provides links to these websites in an easy-to-peruse format that allows us to select one or more labs in which we are interested. It seems to me that a better alternative to upsetting an extremely well-functioning system (GeneTests) could be to request each laboratory to include a basic set of the information from your Plan on its website. Then the information would be available through the GeneTests link to each lab's website without having a separate system.

Another possibility would be to have a link to your data sheets on the GeneTests website--but I still would have concerns about the false appearance of government-backed validity of the information in such a registry, when in reality there seems to be no plan to have expert review of the information provided.

(3) It appears that all submissions to the new Registry would be voluntary--under what oversight? Who will ensure that the information provided is not overstated or inaccurate or biased? Would being listed in such a Registry give the appearance of having been vetted by the NIH or other government agency, when in reality it would be nothing more than the information already available on the laboratory's commercial website?

(4) Item 6f - Recommended patient population: Who will ensure that companies don't recommend a larger-than-necessary patient population in order to boost their sales?

(5) Item 6k - Availability (e.g., is the submitter the sole provider of the test or are there multiple providers?): This already is easily discernable on the GeneTests screen, which is formatted to

include all laboratories that provide the test along with the general types of methodology (e.g., sequencing, mutation screening, etc.) they provide, as well as their locations (including non-U.S. laboratories, which are essential to include because there are many tests that are only available outside the U.S.)

(6) Item 6L - Accessibility (e.g., accessible through a health provider, public health mandate, and/or direct-to-consumer). This appears to give NIH-based validity to direct-to-consumer testing, on an equal level with many excellent clinical testing laboratories. I find this particularly distressing.....

Do you also plan to include (God help us) the laboratories that provide all kinds of highly questionable testing of genetic markers, amino acids, enzymes, vitamins, co-factors, and "serum porcelain levels"--generally ordered by chiropractors and fly-by-night, self-made "health professionals" or directly to consumers--and resulting in incomprehensible and inaccurate reports of diagnoses of genetic disorders and other health implications that are intended to lead to sales of their non-FDA-approved nutrient brews and other useless products (this rant is based on experiences with confused patients who have received such reports and then have come for genetic counseling based on the results)?

- (7) Item 6o - Utility (e.g., clinical and/or personal utility) or outcomes
1. Benefits
 2. Harms
 3. Added value, compared with current management without genetic testing

These questions are better answered by a panel of experienced medical geneticists and genetic counselors than by individual laboratories, many of whom do not have the expertise (or perhaps the inclination) to present these arguments in a well-informed, unbiased manner.

(7) Item 13 - For what purpose(s) would you use the Registry to support your professional efforts? I would use GeneTests first, and I would use the planned Registry only if it would provide additional data not available on GeneTests or on a laboratory's web site.

How would the American College of Medical Genetics--especially the Laboratory Quality Assurance Committee-- potentially be involved in such an effort?

Thank you for your kind attention.
Maureen Bocian, M.D.

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