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Sent: Monday, September 26, 2011 8:18 AM
To: Genetic Testing Registry (NIH/OD/OSP)
Cc: Beckwith, Jonathan Roger; MJCarson@bridgew.edu; Ingeller@alum.bu.edu;
mnthomps@fas.harvard.edu
Subject: Attention: Dr. Patterson, Comments on proposed collection of information for the GTR

Dear Dr. Patterson:

We write this letter in response to the call for public comments on the practical utility of the proposed collection of information for the Genetic Testing Registry (GTR). This correspondence reflects discussions of the Genetics and Society Working Group (GSWG), a multidisciplinary group of scientists and professionals trained in a variety of disciplines, including genetics, sociology, ethics, and law. For decades we have engaged scientists and the general public in discussions of scientific and social issues related to genetic testing. As this technology becomes more prevalent, including through direct promotion of tests to consumers, we believe that there is a need to provide easy-to-access information on the availability of tests, accurate representation of the science that these tests are based upon, and recognition of the potential and realized effects of genetic testing on the individual and the society. This letter addresses our concerns with the practical utility of the proposed collection of information and offers recommendations to improve the quality, clarity, and utility of the collected information.

Among genetic tests, validity is a key concern. Therefore, information on test validity must be, to the extent possible, be provided in a useful manner. To that end, there should be an expansion of data entry guides in the category of validity so that people using the database can understand test validity and so that transparency on issues of test validity is promoted. Validity information should be a significant part of the collected and posted information, and should include findings from tests of clinical and analytical validity and clear indications where validity has not been tested. Further, where third parties have done work that supports or fails to replicate provider tests of validity, this information should be included. Because a provider may not be aware of or may choose not to report information from such third party validations, we recommend an entry category where third parties could directly enter additional published information in a way that would cross reference to the GTR entries and appear in the user interface.

Clarity and ready access to all collected information should be a priority of formatting the information for presentation to users of the Registry. The demonstration entry provided for comment shows only what the test submitter will see, but not how this information will appear to the ultimate consumer/user of the GTR. It is important that the user be able to differentiate between optional information that was not entered by the test submitter and an indication that a test of clinical validity failed or was poor. For example, what will it mean if a test submitter fills out the form for a test and doesn't fill in the box about clinical validity? Does it mean it was not tested or does it mean they choose not to supply information from this test? By making data entry fields such as "assay limitations", "clinical validity" and "clinical utility" part of the minimal required entry (as opposed to optional) and by requiring providers to select "not done" from a pull-down menu if such metrics have not been explored, a distinction could be made between a test where the provider has not asked a particular question and a test where the provider wishes to withhold unfavorable information about a particular test. This will allow users to best utilize the information for test-test comparison and promote informed decision-making.

It is essential to create and provide a User's Guide for the GTR to help users navigate the test information and promote such informed decision-making. The guide should provide information on how to interpret things like "What does FDA Approval mean/signify?" "What is clinical validity?" "What is analytical validity?" "What does CLIA certified mean?" "What is the significance of non-replication?" "What does it mean if the company or institution that fills out the form for a test and doesn't fill in the box for clinical

validity?" It is clear that the NIH wishes to have submitters be solely responsible for the content and quality of the data provided to the GTR. However, it is the responsibility of the NIH to provide the user with information on how to interpret information on metrics such as accuracy, validity, and replication in the context of the scientific process. Even if the NIH will not review tests and/or GTR entries, a Users Guide could offer a mechanism for providing unbiased information on what criteria have been posed for establishing if a genetic association is meaningful (e.g., Chanock, S. J. et al. 2007. Nature 447 (7): pp. 665-660). We expect that, at least for FDA-approved tests, there will be review by that agency initiated by the proprietors of those tests and those using the GTR should be apprised of that information as well. Guidelines for interpreting that information must be provided to avoid having the GTR exist as a manufacturer's opportunity for test promotion and unchecked advertisement.

We hope that these comments will prove useful in ongoing discussions of how to best capitalize on the opportunity to responsibly provide consumers with accurate and useful information that this registry presents.

Sincerely,

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