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Sent: Sunday, June 27, 2010 5:25 PM
To: Genetic Testing Registry (NIH/OD/OSP)
Subject: Comments on RFI NOT-OD-10-101: The NIH Plan to Develop the Genetic Testing Registry

To Whom it May Concern:

We, 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 the law, appreciate this opportunity to comment on the recent proposal for development of a Genetic Testing Registry (GTR) by the National Institutes of Health. For decades our group has engaged scientists and the general public in discussions of the scientific and social issues related to genetic testing. As this technology becomes more prevalent, including through direct promotion of tests to consumers, we feel that there is an increasing 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. Our letter addresses the registry's potential for providing undeserved legitimacy to some genetic tests and arguing for additional levels of control, including peer-review, in the admissibility of genetic tests to the registry. We hope that our comments will prove useful in the ongoing discussions on the design, maintenance, and impact of this registry.

A comprehensive database of genetic tests housed by the NIH will be influential with a wide audience of health care professionals, private and public providers of genetic tests, as well as with the general public. With such broad potential impact, we are concerned that although the registry appears to be a step in the right direction towards standardizing the genetic testing industry, as it is currently proposed it will give a false impression to consumers of standardization and regulation where none actually exists. This concern is underscored by the statement taken from the GTR documents that: "submitters will be solely responsible for the quality of the data they provide to the GTR" and that there will be no NIH review of the submitted information. It is likely that a genetic test placed in the registry, along with the registry's connection to the NIH, will provide the appearance of legitimacy to any test found in the GTR. To develop a more specific discussion of our concerns, we ask the question: Should the GTR employ review standards that apply to either test providers and/or specific genetic tests, thereby imposing quality control measures on the admissibility of entries to the registry?

Considering whether or not standards should be applied to genetic testing providers, we examined GeneTests, the current registry of genetic tests operating through the NCBI. GeneTests provides genetic testing information as well as test availability from a diverse list of publically or privately operated clinics and laboratories. Voluntary registration of laboratories or clinics is required in order to submit tests and this registration requires certain information about the lab or clinic to be submitted. We expect that a similar process will be developed by the GTR and hope that this is a step at which reasonable oversight on reliability of the responsible laboratories and the analytical validity of the tests could be applied to test providers. Additionally, we hope that there is also a mechanism in place to encourage or mandate providers of health-related genetic tests to register with the GTR. One of our concerns relates to direct-to-consumer (DTC) providers of

genetic tests. At the present time, and largely dependent upon state-based regulatory considerations, some of these companies claim that the service they are providing consumers is merely educational and does not qualify as medical information, despite offering testing results that could reasonably be interpreted to have potential health implications. It is our view that health related tests offered by DTC companies need to be appropriately categorized as providing medical information and included in the GTR. Otherwise, these tests will not be subject to the requirements in place for the registry when these are the tests at the forefront of public awareness and access and arguably most in need of standardization.

Another area in which we believe that oversight is needed concerns the admissibility of specific genetic tests to the GTR. GeneTests currently lists over 2,000 genetic diseases for which testing information may be available. We expect that this number will grow with the addition of tests using the results of genetic association studies. DTC genetic testing companies have utilized these studies to develop many of their offered tests. As one example, the DTC company 23andMe, Inc. provides risk assessments for complex disorders such as type II diabetes mellitus; "recreational genetics" tests for traits such as earwax type; and, behavioral phenotypes such as "avoidance of error" and "obsessive compulsive disorder".

Criteria that have already been developed for establishing meaningful genetic associations (Chanock, S. J. et al. 2007. Nature 447 (7): pp. 665-660) should be applied in determining the admissibility of tests to the GTR. The two behavioral phenotypes mentioned above are offered to consumers despite being based on single studies with fewer than 100 participants and do not satisfy these published standards. Additionally, we question whether behavioral phenotypes should be included in the GTR as a general case. Claims of finding genes that appear to play a role in homosexuality, criminality and other complex human behaviors have been made and received high media attention. Yet, nearly all of these studies were subsequently retracted or failed the test of reproducibility. Although these may be extreme examples of the commercialization of dubious scientific findings, other tests offered by 23andMe, Inc. are supported by a single finding in the scientific literature or are offered as tests even if the initial association has been refuted by one or more contrary studies. This practice of offering questionable tests is not unique to 23andMe, Inc., and is our argument why criteria for the admissibility of genetic tests to the GTR need to be discussed and developed in the design phase of this registry.

Hosting a list of the currently available tests, with descriptions of validity and utility compiled by those who have a vested interest in promoting the tests, on an NIH site suggests that the government endorses these tests and has verified the information provided by the testing companies. The information on the GTR would benefit from quality control by a peer-reviewed process that applies standards for tests across all companies. Further, to make the GTR a legitimate resource to consumers, the GTR must propose standardized definitions of accuracy, validity, and utility and provide supporting evidence for each genetic test as well as substantive NIH feedback on where tests meet or fail to meet these standards. With these measures in place the incentive for companies to submit tests to the GTR would truly be a scientific vetting of the genetic tests in a way that would legitimately boost the reputability of the clinics, laboratories or companies that provide these genetic tests.

Sincerely,

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