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Genetic Testing Registry Public Meeting

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I would like to thank Drs. Fomous, Hudson and Ostell for the opportunity to share my thoughts about the Genetic Testing Registry and how to make it an effective tool for healthcare providers and consumers. These comments come from my personal experience in genetic test translation, quality assessment and healthcare provider education over the past ten years. Today I am not speaking for any government agency or professional society.

From 2005 until this summer I was one of the leaders of a program funded by the NIH Office of Rare Disease Research to increase the translation of genetic tests from research to clinical care. The Collaboration, Education and Test Translation or CETT Program was created by NIH-ORDR and multiple partners including the American Society of Human Genetics, American College of Medical Genetics, Centers for Disease Control and Prevention, Genetic Alliance and others. The primary goal of the program was to facilitate the development of clinical tests for rare genetic conditions. An important secondary goal was to develop a transparent way to insure these tests were ready for the clinical arena and to work with the research and laboratory communities to develop the best test that would meet the needs of the clinical and patient communities.

I believe there are two components of the CETT Program that could be adapted by the Genetic Testing Registry and would benefit the registry. In the CETT Program each test was evaluated by an external Review Board comprised of medical geneticists, non-genetic healthcare providers, researchers, laboratory experts, and patient advocates. Each member provided an important viewpoint in this review process. The group reviewed each proposed test for its scientific merit and potential impact on the patient. In almost all reviews the submitting laboratory was provided feedback that improved the test. Some of the lab directors were initially unsure of this process but the majority of them commented that the process was very helpful and the feedback provided was important. If done in the correct educational spirit this process works well.

The Genetic Testing Registry should consider developing an optional test review process utilizing a similar panel of experts. Once the laboratory community becomes familiar with the process they will likely find it helpful. As a healthcare provider and potential patient using genetic testing, I would like to know that a genetic test has been reviewed by an independent group. If a test has not been reviewed that does not mean I would not use the test, but I would probably want more information from the laboratory before ordering the test. I know the FDA is developing a review process and if that happens in a timely fashion I would encourage the FDA to use a similar process and include the research, medical, laboratory and patient communities in the test assessment process.

The second lesson from the CETT Program is a process to determine potential clinical utility. Almost all genetic tests offered today lack data to confirm clinical utility. If we require clinical utility before a test is released, many important genetic tests may never be developed. Also having a check-box in the registry about clinical utility would rarely be informative because it would be blank most of the time.

It is possible to ask about “potential clinical utility”. In the CETT Program we asked the applicants to develop two Diagnostic and Treatment Pathways, one without the test and one with the test. Did the test change the diagnostic or treatment pathway? The development of these pathways was discussed with the researchers and often the researchers could provide information to reinforce the impact of the test on the pathway. In some cases the test meant the patient could avoid invasive or risky testing procedures. In other cases the test provided additional information which could help with prognosis. And at a minimum the test often reduced the time to diagnosis or the “diagnostic odyssey”. This reduced the number of clinical visits and often reduced the total cost of a diagnosis.

The GTR should consider developing categories of “potential clinical utility” which could be applied to tests in the registry based on an assessment of diagnostic and treatment pathways. Examples might include:

- Avoidance of invasive testing
- Improved disease description
- Shortened diagnostic timeline
- Recurrence risk information

By engaging the research and laboratory communities in this discussion, the ability to collect additional information to “prove” clinical utility will likely occur. We found in the CETT Program that researchers often did not understand the type of information that needed to be collected to support clinical utility. When involved in the discussion, researchers were sometimes able to provide information or adapt their research to collect some of the needed information.

Collecting the information to support clinical utility will require the input of the research, laboratory, clinical and patient communities. It will not be possible for the laboratories to do this independently.

In closing, I believe the Genetic Testing Registry should consider creating review boards including researchers, geneticists, general healthcare providers and the patient community AND it should use a model of “potential clinical utility” based on diagnostic and treatment pathways.