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Sent: Tuesday, June 22, 2010 9:58 AM  
To: Genetic Testing Registry (NIH/OD/OSP)  
Subject: Genetic testing registry

My comments and questions on the RFI on the NIH plan to develop the genetic testing registry are as follows:

The function of the GTR as outlined in the RFI overlap significantly with GeneTests. The new information in the GTR not present in Genetests will be the test-specific related information such as sensitivity, specificity, validity of the tests.

Genetests is a tremendous resource for the medical genetics community and is one that is used extensively by individuals in the field at all levels. My main concern is how the GTR will impact Genetests. Will the GTR replace Genetests? Why not build on Genetests instead of creating/replacing with something totally different? Genetests works and it is critically important that if anything replaces it, that it be just as effective and maintained and updated appropriately. My huge preference is for the GTR to be integrated into Genetests as I am concerned that the medical genetics community may lose a vital and critical resource. It is critically important to see who Genetests is important to and ensure that this usefulness is maintained.

GeneReviews is a component of Genetests. This has become an extremely important resource for the medical genetics community and one that we use extensively (again at all levels from attending MD's to training fellows to PhD's) for obtaining information on genetic disorders. It is also a very valuable teaching tool for our fellows. What will happen to GeneReviews? Will this be affected by the development of the GTR?

I am a DNA diagnostic lab director and while I agree that there is no central location with information on usefulness of a particular genetic test, with regards to sensitivity, specificity, clinical utility etc. I am not sure that a lot of this information can be clearly given for a lot of the testing that we do, as the testing is a lot more complex.

I am concerned that the different information that will be inputted into the GTR may result in it being too complicated and thereby affect the how useful it will be to the end user. Will there be too much 'granularity' in the information that will take away from the big picture that is so important in our field?

How will the information in the GTR be updated as new tests are developed by labs? To ensure uniformity of information it is important that there are specific individuals working for the GTR that input data.

Will there be genetic counselors and other medical genetics professionals (lab directors, medical geneticists) involved? To maintain the usefulness of this resource I think it is important for individuals who will be the end users to be involved as this develops and progresses and gets maintained.

What system will there be to provide input into the development of the GTR as it gets developed to ensure that it is maximally useful to the community?

Thank you for your consideration of these comments.

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