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Sent: Friday, June 25, 2010 2:10 PM

To: Genetic Testing Registry (NIH/OD/OSP)

Cc: Ken Mitton

Subject: Comment submission regarding the NIH proposal for the GTR - Kenneth P. Mitton, PhD
- Eye Research Institute - Oakland University

Dr. Cathy Fomous,

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Dear Dr. Fomous,

My commentary is more of a practical suggestion to leverage the general scientific and clinical information that will be captured now and in the future from genetic testing. That is the link between specific mutations and phenotypes that could form a vital reference for physicians and genetic councillors when trying to anticipate the nature and progress their patient's disease. Changes to completely different target genes may result in the same clinical phenotype, and different mutations in the same gene can cause dramatically different phenotypes. Genes are linear objects in their primary sequence, as are the proteins they encode, and changes to different regions may render a condition that progresses over decades or over years, or vary in severity. How can one put this information from known cases into the hands of clinical staff, in a format that can summarize this experience and be easily updated?

OMIM, is an excellent database that summarizes information on genetic disease/differences, but it does not provide a visual summary. I would suggest that genome browsers such as the UCSC genome browser would provide a user friendly platform to keep curated tracks of mutation/phenotype correlations as they map along specific locations in exons, introns, coding regions of the gene affected. Imagine for example, finding a mutation in patient at the Rhodopsin gene, and being able to quickly view the Rhodopsin gene and see if the same mutation

was already reported or see the general phenotypes resulting in mutations around the same location as the new result. This data need not be identified, for HIPA concerns, but could present concise scientific observation to help clinical staff council their patients. Annotation tracks can be loaded into the genome browser for almost any purpose and viewed in their genomic structural context.

Each identified disease-associated base change/deletion could be simply mouse-clicked to view phenotype particulars, and be accessed from any computer platform or smart phone (iPhone) with a web-browser. In the office or in a hospital corridor. We currently use the genome browser to share genome related data (mouse and human) among research team members, and to make it more accessible to those without specialized software or bioinformatics experience, on their laptop or their iPhone.

While a goal of the GTA is to organize available testing resources and to determine the kinds of information to collect, I recommend a follow through to make the collective results of all this future testing data useful to clinicians the world over.

Sincerely

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