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Sent: Monday, July 26, 2010 4:23 PM
To: Genetic Testing Registry (NIH/OD/OSP)
Subject: Genetic Testing Registry

Dear GTR Staff;

I have reviewed the GTR request for information and wanted to take a moment to share my thoughts and concerns. I appreciate that you have specific questions that you'd like me to respond to however, I don't feel I have a significant understanding of how this new GTR will look/operate and have great reservations about how it may negatively impact current registries/review materials used clinically today. So rather than work through the fourteen questions listed within the request for information (RFI), let me skip directly to #14 - Are there any other issues that NIH should consider in the development of the GTR?

The overall vision of a centralized resource where individuals can access all relevant information about genetic tests is noble. However, creating such a resource through voluntary submissions of data from researchers, laboratory personnel and others without critical review to determine if their submissions are actually factual and valid causes me great concern. In addition, I worry that the NIH absorbing such a crucial clinical tool like GeneTests/Gene Reviews and merging or populating a new GTR with its data only to be altered by a voluntary submission of test information will yield this once vital clinical tool worthless.

I can't help but wonder if there isn't a different approach? Perhaps rather than centralizing everything about genetic tests into a resource with voluntary submissions, instead there could be a centralized portal that lists clinical tests and information that have been reviewed by geneticists (e.g., GeneTests/Gene Reviews), as well as listings of other tests (e.g. research, ancestry – a compilation of the voluntarily submitted information being described in the RFI). I recognize that sorting out the different categories of test types may be laborious and may appear to take away from the centralized and transparent objectives, but I offer this strategy as a way of preserving what is already a highly valued resource yet allowing the compilation of additional information and making that information easily available to all as well. Another strategy would be to begin building the GTR as described with voluntary information but not actually linking that resource with GeneTests/Gene Reviews until the data submitted are confirmed/validated and thorough reviews are developed by geneticists.

Finally, I believe that no matter what form the new GTR takes, it's value and overall use will depend significantly on how scientifically valid the information proves to be and how well such a site is updated and maintained. One of the strengths of GeneTests is in fact that the entire system is overseen by geneticists who work diligently make sure the information displayed is as accurate and up-to-date as possible. There must be a plan in

place for similar oversight and maintenance for a new GTR. Whether this function is performed by an advisory group or NIH staff, there must be representation from the clinical genetics community, not just academic, research or business focused input. I think the General Administrative Office's stealth report focusing on direct to consumer genetic testing companies released last week, has yet again suggested that when business interests are at heart, marketing takes priority over relaying factual information. An effective GTR cannot allow such practices yet I fear that is exactly what will happen if data are voluntarily submitted and there is not sufficient genetics oversight.

Again, I applaud the visionary leadership that NIH is pursuing as share the proposed goals of increased access to genetic testing information for all. I hope my comments will be helpful in considering a different, approach than has currently been suggested as I believe that transparency is great but accurate and scientifically valid information is equally important.

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