

September 23, 2011

Amy P. Patterson, M.D.  
Associate Director for Science Policy  
National Institutes of Health

Dear Dr. Patterson,

I'm writing about the proposed Genetic Testing Registry (GTR). I'm a long time human genetics researcher and currently president of a private clinical DNA testing company, PreventionGenetics. PreventionGenetics offers sequencing tests for over 400 human disease genes (and this number is growing rapidly). We are fully CLIA-accredited. Our laboratory is one of the very best in terms of test quality. We do not offer any tests directly to consumers.

Like many in the human genetics and clinical DNA testing communities, I am leery about the replacement of GeneTests by the new Genetic Testing Registry (GTR). Dr. Bonnie Pagon has done a superb job of serving medical genetics through GeneTests. GeneTests provides a necessary function in medical genetics by connecting health care providers to testing labs. GeneTests not only helps the patients and providers, but it is an essential business tool for companies like PreventionGenetics. Our best means of advertising is through GeneTests. GTR therefore has important implications for our small sector of the nation's economy.

I have never heard any convincing arguments for transfer of the test-listing function from GeneTests to the Federal Government. It smacks of a power grab and an expansion of government at a time when this sort of activity is very unpopular with the public. It's unclear to me that the government can provide this service better than Dr. Pagon. Dr. Pagon was impartial and fully trusted by providers and labs. I am worried about delays, mistakes, and about control of the GTR by a small group of people in Washington. I think that decisions about DNA test quality and effectiveness should be made by patients and providers, not by the government. I see no reason to discontinue GeneTests. Why change something that is working very well?

However, if GTR proceeds, you will need to devise means of making data entry for the testing labs simple and efficient. PreventionGenetics has probably the longest menu of any testing lab in North America. The burden of submitting test information will be greatest for us and is on a vastly different scale than the small testing labs. Since I don't know the exact format for GTR, it is difficult for me to estimate the time and expense that will be required to submit our test information. However, based on 31 "minimal" and 85 "optional" fields, it seems that the current GTR is FAR too complex. 116 fields of data entry are FAR too many. The estimated time required for data submission (3 hours per test) could well be a GROSS underestimate. GTR should be substantially simplified before release.

In addition, the GTR data entry fields should be published many months before the registry opens so that we can organize our internal test information in the appropriate format. We also need an efficient means of transferring data about ALL of our tests in one step to the GTR database. One or more GTR staff members should work directly with us to accomplish this task. Manual entry of the same or similar information for each of our > 400 tests would be extraordinarily inefficient. I am concerned that GTR will have insufficient resources to function properly.

In summary,

1. I see no reason to switch from GeneTests to the GTR.
2. If it proceeds, GTR should be substantially simplified prior to release.

3. If it proceeds, GTR should have sufficient resources and informatics capabilities to efficiently allow clinical labs to enter test data.

Sincerely,  
James Weber

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