



September 14, 2011

Amy P. Patterson, MD
Associate Director for Science Policy, NIH
Office of Biotechnology Activities
6705 Rockledge Dr.
Suite 750
Bethesda, MD 20892

Sent electronically to gtr@od.nih.gov

Dear Dr. Patterson,

I would like to comment regarding the agency's estimate of the burden of the proposed collection of information required to register genetic tests with the Genetic Test Registry (GTR).

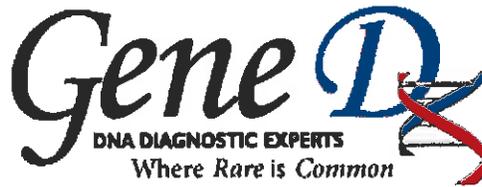
It is stated that the estimated average time (hours) per submission per respondent will be 0.5 hour to complete the minimal fields for each test and 2.5 hours to complete the optional fields for each test, coming to a total of 3.0 hours for each test that a laboratory submits to the GTR. It is also estimated that the average number of submissions per respondent is expected to be approximately 12 tests.

GeneDx is a clinical molecular diagnostic laboratory, located in Gaithersburg, MD. All of the tests we perform are currently listed on the GeneTests.org website. GeneDx has 474 individual test listings on GeneTests.org currently.

If my laboratory selects to list only the minimal fields for every test that is offered, that comes to 237 hours, or 5.9 full time weeks for one individual. As this work will have to be done by a genetic counselor (average hourly pay of \$38/hour), this will come at a cost of \$9000.

If my laboratory includes the optional fields for all of the 474 tests, the burden is 1422 hours, or 35.5 full time weeks, at a cost of \$54,000. The time to collect and organize the data required to complete the optional fields is not included in this estimate, but that time is also significant and may be an additional hour for each test.

I expect that those labs with few tests will have little trouble completing the entire set of required and optional data fields. I feel that if GeneDx does not submit the same data as those smaller labs we will have a competitive disadvantage when physicians compare laboratories as they choose a lab to which to send a patient's sample for testing. However, the burden and cost to submit comparable data will be prohibitive for GeneDx, with the result that the labs with smaller test menus will have an unfair competitive advantage.



Finally, and most importantly, the ultimate issue is that patient care may be compromised. It is likely that labs with larger test menus will choose to submit only those tests to the GTR for which there is competition between labs for samples. Those tests are the ones that generate the highest volume (and revenue) for the laboratory. If that happens, and the tests that are offered only for rare and ultra-rare disorders (and generate low volume/revenue) fall to the bottom of the priority list for submission to the GTR, patient care will be impacted. Physicians who depend on the GTR to identify laboratories for their patient's genetic testing needs will fail to find the tests for rare disorders listed there, and the patients will not get the testing they need for appropriate diagnosis and management.

Thank you for the opportunity to submit comments on the burden of proposed data collection for the GTR. I am available to discuss any issues pertaining to assuring that testing for patients with rare and ultra-rare disorders remains available.

Sincerely,

Sherri J. Bale, PhD, FACMG
Managing Director
GeneDx