



2820 N Astor
Spokane, Washington 99207

September 15, 2011

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

Dear Dr. Patterson,

We read with interest the notice in the Federal Register, volume 76, number 144 regarding the intended Genetic Testing Registry (GTR). We have reviewed both this discussion and the document with screenshots of the proposed online form for test submission. As indicated in the Federal Register article, we have included feedback pertaining to several points.

We are a diagnostic testing laboratory that performs genetic testing on patient specimens. Our intended audience, should we list with a registry of genetic tests, is health care providers, specifically providers in specialties such as Genetics, Obstetrics, Neurology, and developmental specialists. The feedback we are providing on subsequent pages is specific to the needs of diagnostic testing laboratories and clinicians who are intended to be the end-user of such a registry.

Thank you for your consideration of our comments.

S. Annie Morton, MS, CGC
Justine Coppinger, MS, CGC
Nadia Altomare
Beth Torchia, PhD, FACMG
Blake C. Ballif, PhD
Jay Ellison, MD, PhD, FACMG
Lisa G. Shaffer, PhD

- 1) Evaluate whether the proposed collection of information is necessary for the proper performance of the function of the agency, including whether the information will have practical utility.
 - The benefits for a diagnostic testing laboratory to be listed in this registry are not clear.
 - Much of the information requested is not relevant to a health care provider ordering a genetic test.
 - It is not clear how the end user will search, view, and access this information and whether it will be relevant to their individual needs.
 - There is no evidence of safeguards within the system to ensure that data will be secure and used appropriately as defined by the Registry.
 - The amount of information that is being requested from laboratories is insurmountable for most diagnostic testing laboratories.
 - Instrumentation list and platform is of no clinical utility and allows for commercial bias, which is not useful or relevant in a diagnostic testing laboratory. The end user audience(s) must be more clearly defined and targeted.
 - For each targeted audience the data should be assessed and filtered for relevance. Different types of audiences will have specific needs, and if each type of user accesses the data in the same manner, the volume of data will be prohibitive of use for some types of users, such as clinicians and the public.
 - If this cannot be achieved, targeted audiences will not use this site, and diagnostic testing laboratories will not list with this site, defeating the purpose of the GTR to “Provide an information resource for the public, including researchers, health care providers and patients, to locate laboratories that offer particular tests”
 - Fields to be populated are not clearly defined.
 - This may lead to lack of uniformity between laboratories.
 - Each field that requires an entry should have information describing how the field should be completed to provide consistent entries between laboratories.
 - Fields to be populated are sometimes inappropriate.
 - The result of providing some of this information, for example, laboratory affiliations, may cause further division between laboratories and will be counterproductive toward collaborative efforts, which would defeat the goal of the GTR to “Facilitate genetic and genomic data-sharing for research and new scientific discoveries”.
 - The request for additional research information is inappropriate for diagnostic laboratories testing patient specimens.
 - Request for information about charging for parental testing is inappropriate because the fee for testing does not affect the clinical utility of the test. Simply asking whether parental testing is available is appropriate.
 - It seems redundant to enter specific information about gene locations when the NCBI maintains a browser that houses this data. The genes should automatically link to NCBI, negating the need to enter the information in the GTR. Similarly, it would be useful for conditions to link to GeneReviews and other NCBI resources.

- There does not seem to be utility to entering benign variants into this system, especially for laboratories performing diagnostic testing.
- 2) Evaluate the accuracy of the agency's estimate of the burden of the proposed collection of information, including the validity of the methodology and assumptions used.
- There is insufficient information provided to accurately determine the time/cost burden for entering diagnostic laboratory tests into this system. More information is needed about the ability to upload from GeneTests, the bulk upload feature, and specific options available for entering test information at various points in the process.
 - Because our laboratory currently utilizes the GeneTests directory, we have knowledge of the time burden to enter the minimal information into their system. Our laboratory has determined that the time and cost estimate proposed to enter laboratory tests into the GTR is a gross underestimate and prohibitive to diagnostic laboratories who want to list with this registry. Our estimate is as follows:
 - Assuming demographic laboratory and staff data is available to pull from GeneTests, we estimate this will take **1 hour** to complete the required fields and proof-read what has been transferred.
 - We estimate approximately **60 hours** of work will go into gathering data and putting together resources ahead of time so we have the appropriate information ready for data entry. A significant portion of this burden is interpreting how to enter the fields within the system because they are not clearly defined.
 - Assuming a bulk upload feature is available and usable, we estimate that each condition will require approximately 30 minutes for all fields that we deem relevant. Our laboratory currently tests for approximately 250 conditions, which translates to approximately **125 hours** to enter the conditions for which we test.
 - The hourly salary estimated by the GTR is under-estimated, as the individual entering data into this registry will need to be someone with a post-graduate degree to have enough knowledge to complete the fields. The hourly salary estimate provided should be doubled to take this into account, raising the **hourly salary to \$45.70**.
 - Based on our estimates, it will take approximately **45 minutes** per condition to load information into the GTR, while we estimate that it currently takes an average of **18 minutes** per condition to add listings to GeneTests. This is nearly tripling the time to enter data about laboratory testing with very little added clinical utility to the end user.
 - The time and cost burden is significantly underestimated and far outweighs the benefit to the laboratory or to the end user to list with such a registry. As a

diagnostic testing laboratory, we are likely to only fill out the minimally required fields if we choose to invest the effort to enter our testing in this registry.

- 3) Enhance the quality, utility, and clarity of the information to be collected.
 - See question #1 for suggestions.
 - In addition, there doesn't seem to be a lot of clinical utility to the majority of data requested with the exception of manufacturers and listing laboratories to gather information about their competitors. This does not benefit the public.
 - While many of the fields included in the GTR are not clinically relevant, there are several clinically useful pieces of information that have not been requested such as:
 - Turn-around time
 - Year or date testing was first implemented
 - Yearly test volume
 - Some type of peer review is absolutely necessary after laboratories submit their information to maintain integrity of the data submitted by laboratories. As currently planned, the only check on quality of the data is by the laboratories themselves. It is highly likely that the public will perceive that the data are vetted by the NIH. The necessary peer review should be carried out by experts from the medical genetics community. Without such review the data will be inferior to that currently provided by GeneTests.

- 4) Minimize the burden of the collection of information on those who are to respond, including the use of appropriate automated, electronic, mechanical, or other technological collection techniques or other forms of information technology.
 - As current users of GeneTests, we feel their system is very well developed and provides useful clinical information to its intended audience without bias. We feel that much of the supplementary information asked by the GTR introduces commercial bias and may create the potential for inappropriate competitive advantages. We recommend simply using the current GeneTests model and broadening the data fields to include test type/methodology used, proficiency testing details, the ability to add large panels and whole genome testing, and some test-specific details such as number of mutations tested, turn-around time, year or date testing was first implemented, and yearly test volume. This would maximize clinical benefit while maintaining an unbiased system.
 - Suggestions for mechanisms to minimize the burden include:
 - Download from GeneTests will be critical, but more information is needed to know how this will function. It will need to pull the maximum amount of information, however many of the fields in the GTR do not align well with GeneTests fields, therefore we have concern that this will not function well with the current GTR design.

- A bulk upload feature, which is mentioned but not described, may be useful, but more information is needed to know how this will function. This will be absolutely necessary for labs with a large number of test listings, however the questions included in the GTR will need to be answered regardless of whether the upload method is made more efficient, so this will help to minimize time expended to some degree, but will not alleviate the majority of the burden.
- It would be useful to have the ability to clone data already entered. For example, if a laboratory has two tests that would have nearly identical answers, it would be useful to enter the information in once, clone that test listing, and then make modifications to the listing that is already completed to make the listing appropriate for the second test.

5) Questions:

- If GeneTests is being replaced by GTR, will GeneReviews retain its current standards? What will be the impact on GeneReviews of the transition from GeneTests to GTR?
- Who specifically developed the questions asked by the GTR and who specifically will be maintaining the site?
- Is there some way to be added to a contact list so that we may be alerted when there are updates to the GTR development status, meetings, public comment periods, etc.?