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PERSPECTIVE

Beast of Burden? Comments on the NIH Genetic Testing Registry

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The Genetic Testing Registry (GTR) is a voluntary registry of genetic tests initiated and created by the National Institutes of Health (NIH) (Field *et al*, 2011). Given the proliferation of tests for many genetic conditions, the NIH recognized that a centralized public database for information about these tests was needed. NIH launched the GTR project "to advance public health and research in the genetic basis of health and disease" (NIH, 2011). The key objectives are to enhance transparency, provide an informative resource for the public, and facilitate data-sharing. The NIH has actively engaged stakeholders throughout the process, some of whom have expressed concerns about the registry—particularly how to convey exactly what the information provided represents. After approximately a year of work on the GTR, where does it stand?

Request for Comments

To solicit feedback on the developing GTR, the NIH posted a Request for Comments on July 27, 2011. The NIH estimated the burden for submitting tests to the GTR, where the submission form has 31 minimal fields and 85 optional fields. The NIH projected the annual burden to be 6.1 hours to complete all minimal fields and 36.6 hours for all fields, with a projected average of 12.2 tests per respondent. Submitters unfamiliar with the optional information may need more time to complete the form, and a bulk upload feature could be used to decrease the time burden. The NIH also proposed a mean annual cost of $836.30 per respondent, using national wage statistics to calculate the cost of laboratory technicians entering the data (NIH, 2011).

As part of the effort to enhance the GTR, the NIH asked for feedback on the following: (1) Whether the proposed collection of information is necessary and has practical utility; (2) whether the agency's estimate of the burden of the proposed collection of information is accurate; (3) whether there are ways to enhance the quality, utility, and clarity of the information to be collected; and (4) whether there are ways to minimize the burden of the collection of information on those who are to respond (NIH, 2011).

Responses

The responses to the Request for Comments came from 12 individuals or groups representing a range of stakeholders, including clinicians, testing laboratories, and health advocacy organizations. This is a very low number of respondents, given the tens of thousands of clinicians, testing laboratories, and advocacy organizations in the United States, and the comments received may not represent all concerns of patients, clinicians, and laboratories. Nearly all respondents believed the GTR (or at least the concept) was generally a positive and necessary initiative, and many of the responses constructively focused on areas for improvement or concern. Many noted that the GTR would be a useful tool for a variety of stakeholders that could increase transparency. Furthermore, many respondents supported the bulk upload feature, although some requested more information on how this would be implemented.

An area of concern raised by nearly every respondent was the NIH's calculation of burden. Most respondents felt that the statistics provided in the Request for Comments underestimated the costs, both financially and the number of hours required for submitting entries to the GTR. Some noted that the number of tests was underestimated because many labs offer multiple tests for the same condition. As a comparison, entries in GeneTests (www.ncbi.nlm.nih.gov/sites/GeneTests/), the registry the NIH uses to determine the number of tests, are indexed by condition only. Additionally, others suggested that next-generation sequencing methods could greatly increase the number of available tests. Increasing the number of submitted tests would probably raise the estimated financial burden.

Many respondents also suggested that the pay rate used by the NIH was underestimated. Although the Request for Comments used national wage statistics for laboratory technicians, many argued that submitters will be required to have a greater knowledge of the tests—mostly likely a genetic counselor or laboratory director. These employees will have significantly higher wages, and this could also increase the cost of submission. Some respondents suggested that the actual burden expected by submitting groups would be insurmountable.

In addition to costs of submission, many respondents were also concerned about the lack of oversight in data submission and about the possibility that the GTR would misrepresent the validity and utility of tests. Without proper oversight, some anticipated that testing companies might abuse the registry as a marketing tool. To prevent this, respondents proposed a regulatory body to review each entry or suggested

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that each test submission should require associated peer-reviewed literature. Respondents also suggested that information provided through the GTR be acknowledged as generalized in order to address potential misrepresentation of the submitted tests. Although information about utility and validity is important, respondents explained that both elements are fundamentally linked to the particular context of an individual patient.

Future Directions

The responses indicate that the GTR has promise and potential to help clinicians and patients and that possibly more refinement may be necessary. Because of the overwhelming concern about the true burden of submitting entries, perhaps a beta launch with a "coalition of the willing" might be useful in gathering the true burden of data submission. This could establish a baseline of cost for small and large labs, for both esoteric testing as well as reference labs. Then burden can be realistically assessed and decisions made about making the process more expedient, if necessary. Finally, the balance between crowd-sourcing the entries with ever-increasing quality—such as a wiki-style resource—versus a labor-intensive oversight process is important. Perhaps the community will police the resource, although that did not happen with GeneTests, which takes all comers, and the listings do not seem to be challenged. Increasing intelligent linking to the resource will help, allowing peer-reviewed literature and other databases to validate or invalidate entries. From the public's perspective, transparency of the data afforded by the GTR is a welcome addition to the laboratory testing arena, allowing the beginnings of a more open and dynamic ecosystem of genetic testing.

References

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