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National Society of Genetic Counselors

July 7, 2010

NIH GTR RFI Comments
National Institutes of Health
Office of Science Policy
6705 Rockledge Dr., Room 750
Bethesda, MD 20892

Dear GTR Staff,

The National Society of Genetic Counselors (NSGC) is writing to provide input to the National Institutes of Health (NIH) in light of the March 18, 2010 announcement for the creation of the Genetic Testing Registry (GTR).

The project has a worthy goal, and we are supportive of enhancing access to information about the availability, validity, and utility of genetic tests. There has been considerable previous work conducted through the NIH and GeneTests. We encourage the NIH to continue utilizing the GeneTests staff, genetic counselors, and the NSGC for expertise moving forward with the GTR. We believe that GeneTests can be used as a foundation to be improved upon during the development of the GTR.

In additions to the comments below, we want to inform the NIH that many members of the NSGC have contacted the NSGC leadership to express confusion and concern about the reasoning behind creating a new registry instead of adding additional data elements to GeneTests. The NSGC recommends additional communication beyond the online GTR FAQs to clarify the objectives behind the change, and the NSGC can help facilitate this communication to our members, who are a primary audience for GeneTests and in all likelihood the GTR.

We respectfully request the NIH to consider the comments and recommendations of the NSGC. We make some general recommendations first followed by responses to some of the specific questions outlined in the NIH's request for feedback. We responded to those that had the most relevance for genetic counselors.

General recommendations:

- Continue integration of GeneReviews with the GTR as it was with GeneTests.

The NSGC understands that the GTR does not intend to replace or retire GeneReviews. However, the integration of the highly readable GeneReviews with a laboratory registry such as the GTR has great value to providers. As the NIH is aware, GeneReviews provides a peer-reviewed, detailed overview of genetic disorders that is organized in a manner that is convenient and clinically relevant for practicing genetics professionals.

- The NSGC recommends the inclusion of an NSGC-appointed representative on the GTR's advisory board.

The NSGC understands that the NIH has an advisory group that includes genetics counselors. Genetic counselors are a valuable asset to the advisory board because they have considerable experience in knowing what data elements are important to include and how to present

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information in an understandable, relevant, and organized manner to various audiences. We recommend an NSGC representative be included on the panel to represent the interests of genetic counselors, a core audience for the GTR.

- Consider the impact of the number of data elements on the usability of the site.

More information is not always better. The increased transparency and availability of information must be weighed against an increasingly cumbersome interface. The NSGC understands that the NIH plans to beta-test a site and suggests that representatives of NSGC take part. One potential solution to consider is to require diagnostic laboratories to house some data elements on their own sites, which would have the benefits of both maintaining usability and preventing the misinterpretation that the NIH is endorsing testing.

- Consider the feasibility of peer-review of claims.

The goal set out by the HHS Secretary's Advisory Committee for Genetics, Health, and Society was for a mandatory registry The NSGC is concerned that tests that have little or no clinical validity will gain credibility simply by being listed on the GTR. We appreciate the potential debate that could ensue for some tests but we also fear tests that appear on the registry will automatically become valid in the eyes of the public and non-genetics professionals. If formal peer-review is not deemed feasible, then the NSGC would recommend creating the ability for external comments by providers and other laboratories.

- Focus on a key audience instead of attempting to reach multiple audiences:

We do not believe it is possible to create a resource that will be equally valued by all of the intended audiences. The needs of consumers, researchers, providers, payers and policy makers have some areas of overlap but also some critical differences. For example, clinicians may value and correctly interpret accuracy and analytical sensitivity/specificity, but consumers may misinterpret these data elements as clinical validity. In fact, even the needs of genetics professionals' versus non-genetics professionals are very different. Genetics providers will likely use this service similarly to how they have used GeneTests previously; as a resource for genetic test availability and application. Non-genetics providers and the public may use a registry to infer validity. The specifics of the data provided (listed in the NIH's feedback question #6) will not be useful to most non-genetics professionals and consumers, as they do not have sufficient training to interpret and assess them. Therefore, the NSGC recommends that the NIH develop the GTR with genetics professionals as the primary audience and other health care providers as a secondary audience.

Response to Specific Questions in RFI

2) (1) Researchers may use this site as they currently use Genetests, i.e. as a means of advertising studies for which they are actively recruiting participants. Providing the inclusion and exclusion criteria in addition to the existing data elements already present on GeneTests would save people time in identifying appropriate studies.(2) As stated above, consumers would likely use the GTR to determine test availability but may also infer validity if they believe that the NIH is "endorsing" the test. (3) See comments above and below. (4) and (6)Clinical laboratory professionals and genetic testing labs may use a registry similar to how they use GeneTests now. It serves as a marketing channel as well as a means to determine competition. (5) Payers could find a resource like this helpful if it addresses clinical validity in addition to analytical validity, as

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they are looking for guidance on which genetic tests they should cover. However, it should be recognized that a registry will not be sufficient for payers' purposes because it cannot assess the circumstances under which testing should be offered and covered on a case-by-case basis. (7) Policymakers may find a registry helpful because it could create some oversight of the genetic testing industry without imposing barriers to innovation and translation of genetic research into clinical practice. As a voluntary registry, not all laboratories will choose to be listed because they are not able to provide all of the requested data but could continue to market their tests. (8) A standard identifier for each test would make the EHR more robust if the records could support such a data element.

4) (a) Providers, consumers, test developers, researchers, and manufacturers would likely view the benefits of the GTR similarly to GeneTests, which is primarily as a communication tool about the availability of genetic testing. (b) & (c) See comments above. This information is useful for healthcare providers, but the public and even non-genetics professionals may have insufficient training to appropriately interpret some of these elements. Therefore, the NSGC recommends refocusing on genetics professionals as the primary audience.

5) The distinction between data fields left blank because of the absence of data/evidence and those left blank for other reasons is important. The option of "data not available" should be included.

6) (a) Contact information should be included for the tests listed. The NSGC also recommends that the laboratory be able to provide contact information for genetic counselors they employ. (b) CLIA certifications are important to include. (c) The common and commercial name for a test are relevant to include. The GTR could link to marketing materials on a laboratory's website but take care not to suggest an endorsement of the information presented. Also, issues such as unique product identifiers would be extremely helpful for comparing tests; however, as the NIH is aware, there are no current regulations that mandate such standards. The registry may be limited by the lack of industry and data standards. (d) Given the rapidly changing regulatory environment, the NSGC recommends waiting to assess the usefulness of including regulatory clearances until there have been conclusive decisions made. Inclusion of this information at this time would create confusion due to eligibility for regulatory clearance, different types of regulatory approvals, and the inclusion of non-US tests, etc. (e) Many genetic tests will fit into more than one category, and this is helpful for genetics professionals to know. For example, cystic fibrosis testing may be utilized for carrier screening or diagnostic testing. (f). Genetics professionals would find it helpful to know if a test was studied in a particular population and is therefore appropriate for that population. However, laboratories may be tempted to make the categories broad, so this is an example of why peer review or external commentary would be helpful to prevent misinformation. (g) See (f). (h) test methodology is very helpful for genetics professionals to understand, as it may determine the order in which testing is performed. For example, disorders that can result from both deletions and point mutations would require two different methodologies to assess the mutation status of the patient. (j) Helpful information to have for ordering purposes. (k) One of the primary advantages of GeneTests is that most laboratories list their services on the site. Therefore, genetics professionals have one place to go to find most laboratories offering the testing. (l) As stated earlier, the regulatory environment is changing rapidly, so the NSGC would recommend waiting until decisions have been made about regulation before deciding whether to include this data element. However, in the current regulatory environment, genetics professionals would probably find it helpful to know who can access this test. The ability to provide external comments would be helpful here because others could comment on whether the level of accessibility is appropriate. (m) Performance characteristics are helpful to have available and increase transparency. This would allow genetics professionals to distinguish between different

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laboratories. However, as stated above, too many data elements may make the interface cumbersome, so developers of the GTR should consider that trade-off. (n) As stated above, the NSGC greatly appreciates the effort the NIH is undertaking to increase transparency around the clinical validity of genetic testing. However, there are significant challenges to assessing clinical validity for genetic testing: it may be difficult to reach clinical significance for rare diseases, defining the population being studied may be difficult, etc .(o) As stated above, with a voluntary registry, utility is a risky factor to include because laboratories can make unsubstantiated claims or not acknowledge risks. Genetics professionals would likely disregard this section because it could be biased. Peer-review and/or ability to make external comments would help address this issue. (p) Pricing could be confusing because the price a patient would pay would vary depending on the payer, the provider and the institutional contracts. However, including the list price would give some transparency that might draw down costs due to competition.

14) Additional data elements to consider might be patent status of a test or gene.

The NSGC appreciates the opportunity to provide comments. We look forward to collaborating with the NIH to ensure the GTR is a valuable tool for genetics professionals.

Sincerely,

Elizabeth Kearney

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President