

NIH – Genetic Test Registry - Request for Comments

1. Tests which include genetic technologies, but are not testing for germline or somatic genetic differences may be less relevant to include – e.g. PCR for HIV virus detection.

2. The primary use for all stakeholders will be the ability to identify laboratories that offer genetic testing for particular conditions or circumstances. In addition, specific uses for the following may include:
 - (2) patients/consumers - increased confidence in laboratories quality, or at least ability to compare laboratories.
 - (3) health care providers - better understanding of laboratory tests' limitations, possible test results, potential benefits to patients, who best to offer testing to.
 - (4) clinical laboratory professionals - increase focus on lab quality, educational information.
 - (5) payers - baseline information to determine if evidence of medical benefit exists to determine coverage
 - (6) genetic testing entities/data submitters - ability to compare their services to those of other laboratories
 - (7) policy makers – increased awareness of available tests and technologies that may require policy changes
 - (8) electronic health records – direct linkage to labs on GTR for ordering purposes.

3. Although the data elements listed below may be important to multiple types of users, those who might particularly benefit are listed by corresponding number:
 - Background/literature in support of the test (1,3,4,6)
 - Benefits/Limitations of the test (all)
 - Predictive value/discriminatory accuracy (1,3,4)
 - Methodology employed (1,4,6)
 - Available information on analytic validity, clinical validity, clinical utility (1, 3, 4, 5, 6, 7)
 - Possible test results and interpretation (all to varying degrees)
 - Impact on population health (prevalence, positive/negative predictive values) (1, 3, 4, 7)
 - Costs/billing/ordering information (2, 3, 4, 7)

4. a) Benefits: allow public to know what genetic tests are available, and potentially to compare and contrast labs in terms of quality, accessibility, cost, etc.

Risks: Potential for public to misinterpret information provided about test

b) Benefits: better ability to judge potential uses and usefulness of genetic test, and compare among labs

Risks: Potential to impact lab innovation/ expansion – business competition; potential for misunderstanding/misinterpretation depending on how information presented

c) Benefits: Ability to judge benefits vs. harms of test usage

Harms: Solid data on clinical utility (e.g. from random controlled trials) unlikely to be available for many tests which may still have substantial benefit

5. All data fields must be completed (forced choice of some response e.g. – “absence of data/evidence” or “other”)

6.
 - a. Website if available; contact for specific questions re: test use and interpretation – e.g. genetic counseling.

 - b. Yes. Also - qualifications/certifications of lab director and professionals providing consultative services

 - c. Yes. Also, gene/genes involved using standardized nomenclature, ICD-9 codes for related health conditions/symptoms.

 - d. Yes

 - e. Yes, with explanation

 - f. Yes, with variations/limitations

 - g. Yes, with possible uses/applications for which data remains limited

 - h. Yes, insofar as it does not appear to threaten the laboratories’ intellectual property rights.

 - i. Yes

 - j. Yes

 - k. No – I do not think this should be a requirement of the submitting laboratory to identify other providers.

 - l. Yes

 - m-o. Yes – all, to the extent they are available and can be concisely and adequately explained. May need to develop drop down menus with specific choices to facilitate consistency of entry and comparability.

 - p. Yes to cost of test, health insurance coverage too variable to request reporting.

Note: Much of the above will require provision of these items in separate educational materials (preferably separate formats for consumers and clinicians). These supplemental explanatory materials will be critical to the goal of the GTR to protect consumers and maximize benefits and minimize harms of genetic testing.

7. For many tests, there will be limitations to available data for clinical utility – though possible benefits/harms should be presented, and reasons for lack of data should be explained.
8. Advantages are ability to compare tests, disadvantages relate to threats to intellectual property, and commercial incentives.
9. A comprehensive resource link per disease and/or test would be very helpful to both consumers and healthcare providers. In addition to recommendations from expert panels, links to reliable consumer/support group information (e.g. Genetic Alliance) would be helpful, along with ways to access genetic service providers.
10. Ease of entry, drop down confined choices, appropriate and screened links - including ability to link to master file where appropriate.
11. Benefits would be “free advertising” – risks would be if data entry required in some way threatened labs ability to promote business model.
12. Continue to engage all stakeholders in the development process, particularly including consumers and healthcare providers in designing the information they need to make the GTR an effective and usable resource.
13. As a clinical genetic counselor, a counselor involved in public health genomics translation efforts, and a director of a genetic counseling training program – I would utilize a GTR in direct patient care, public health/policy efforts and educational activities.
14. Consider efforts to ensure translation of genetic testing for rare conditions from the research to the clinical setting by modeling after the successful Collaboration, Education and Test Translation (CETT) Program of the NIH-ORDR.

Cecelia Bellcross, PhD, MS, CGC
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