

From: Leslie.C.Manace@kp.org [mailto:Leslie.C.Manace@kp.org]
Sent: Wednesday, June 30, 2010 7:03 PM
To: Genetic Testing Registry (NIH/OD/OSP)
Subject: RFI) on Genetic Testing Registry

1. Are there any types of genetic tests that should not be included in the GTR?

"recreational" ancestry testing i.e. by mitochondrial DNA, Y chromosome analysis
paternity testing

2. What are the potential uses of the GTR for (1) researchers, (2) patients/consumers, (3) health care providers, (4) clinical laboratory professionals, (5) payers, (6) genetic testing entities/data submitters, (7) policy makers, and (8) electronic health records?

- 1) central reference for labs focusing on particular loci/diseases
- 2) information
- 3) resource for availability of testing, databases for interpreting results
- 4) labs meeting CLIA/CAP standards
- 5) appropriate labs to be reimbursed for performing tests
- 7) increasing use of genetic testing

3. What data elements are critical to include for use by (1) researchers, (2) patients/consumers, (3) health care providers, (4) clinical laboratory professionals, (5) payers, (6) genetic testing entities/data submitters, (7) policy makers, and (8) electronic health records?

3) CLIA certification and number, contact/shipping information, specific testing offered (full sequencing vs. mutation panel, dosage testing), cost, turnaround time, what is included in report, availability of genetic counselor/director to contact

4. What are the potential benefits and risks associated with facilitating public access to information about the:

- a. Availability and accessibility of genetic tests?

benefit of more interest in personal and family medical history; risk of increasing needless testing with associated healthcare cost, adverse psychological outcome, improper interpretation and follow up care recommendations (with possibility for more needless evaluations/procedures increasing overall healthcare spending)

- b. Scientific basis and validity of genetic tests?

more informed public as opposed to a lack of overarching government body review of available tests

- c. Utility of genetic tests?

5. What is the best way to distinguish between data fields left blank because of an absence of data/evidence and those left blank for other reasons? How important is this distinction for enhancing transparency, including for the purpose of identifying research opportunities?

all fields should be completed, even with "not available" or "more information/evidence needed"

6. To adequately and accurately describe a genetic test, which of the following

data elements should be included in the GTR? Are there other data elements that should be added? What information is necessary to represent adequately each data element?

Contact information, Laboratory certifications, Name of the test, Intended use of the test, Recommended patient population, Limitations of the test, Test methodology, Analyte(s), Specimen requirements, Accessibility, Performance characteristics (Analytical sensitivity, Analytical specificity, Accuracy, Precision, Reportable range of test results, Reference range), Clinical validity (Clinical sensitivity, Clinical specificity, Positive and negative predictive value, Prevalence, Penetration, Modifiers), Utility (Benefits, Harms, Added value), Cost

7. What types of information might be difficult for test providers to submit and why?

a large part of the clinical validity and utility, due to lack of evidence (that research is just now happening)

8. What are the advantages and disadvantages of collecting and providing information on the molecular basis of genetic tests, such as detailed information about what the test detects and the specific methods employed?

9. In addition to the data elements, would it be helpful to reference other resources, and if so, which ones (e.g., published studies, recommendations from expert panels such as the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children, U.S. Preventive Services Task Force, or Evaluation of Genomic Applications in Practice and Prevention Working Group)? definitely! with hyperlink to EGAPP, papers, or other documents to make reviewing these easy

10. As the GTR is being designed, what are the important processes to consider to make the submission of data as easy as possible for the data provider (e.g., the capability of linking to information that has been submitted to other agencies, such as the Food and Drug Administration and the Centers for Medicare and Medicaid Services, or a master file of data common to particular tests)?

11. Which potential benefits and risks would be most likely to affect the decisions of researchers, test developers, and manufacturers on whether to submit data to the GTR, and what factors will best encourage submission of complete and accurate data?

allow for "insufficient evidence" to be supplied in fields such as utility added value

12. What are the most effective methods to ensure continued stakeholder input into the maintenance of the GTR?

simple BRIEF user feedback surveys

13. For what purpose(s) would you use the Registry to support your professional efforts?

uniform registry where key questions (cost, technique) can be quickly compared

14. Are there any other issues that NIH should consider in the development of the GTR?

quick "101" pop out windows for the public to click on to define terms, explain methods

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From: Leslie.C.Manace@kp.org [mailto:Leslie.C.Manace@kp.org]
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To: Genetic Testing Registry (NIH/OD/OSP)
Subject: RE: RFI) on Genetic Testing Registry

One more thought I forgot to include: it would greatly enhance efficiency of our practice if the testing requisition forms are UNIFORM (standardized) and EASILY COMPLETABLE and SUBMITTABLE ONLINE!

Best,

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