

The NIH Genetic Testing Registry

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CLIAC Meeting

March 2, 2011

Disclaimer

"The findings and conclusions in this presentation are those of the author and do not necessarily represent the official position of the Centers for Disease Control and Prevention/the Agency for Toxic Substances and Disease Registry (CDC/ATSDR)"

The NIH GTR

- Announced 3/18/2010 to provide a centralized online information resource on genetic tests
 - Availability
 - Scientific basis
- Perceived need to collect detailed information about 1600 genetic tests

Goal of the GTR

- “Advance public health and research into basis of health and disease”

http://oba.od.nih.gov/GTR/gtr_faqs.html#GTR_FAQ001

- Transparency about test availability and utility
- Identify locations of laboratories and test offerings
- Data-sharing for research and discoveries

Why NIH

- Conducts/supports basic, clinical, and translational medical research
- NIH includes a large number of public databases
 - National Library of Medicine (NLM)
 - National Center for Biotechnology Information (NCBI) = will create, design and maintain the GTR

Scope of the GTR

- Genetic test is “a test that involves an analysis of human chromosomes, deoxyribonucleic acid, ribonucleic acid, genes and/or gene products (e.g., enzymes and other types of proteins), which is predominantly used to detect heritable or somatic mutations, genotypes, or phenotypes related to disease and health.”

NCBI will seek

- Detailed information on molecular testing quality measures – analytical and clinical validity
- To provide more assay-specific performance characteristics
- Share this information with: consumers, patients, health care providers, third-party payers, providers of genetic tests, researchers

Caveats

- No standards for common data elements
- Submitters will be responsible for content and quality of data provided to the GTR
- No auditing by NIH to verify the submitters' claims about test performance
- Information may be misinterpreted or misapplied leading to adverse health decisions and public health risks

Question

- Does CLIAC wish to make a recommendation to the HHS Secretary regarding the NIH GTR?