The NIH Genetic Testing Registry

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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?