



Collaboration Education and
Test Translation Program

Secretary's Advisory Committee on Genetics, Health, and Society (SACGHS)

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<http://www.cettprogram.org>



Quality Testing Rare Genetic Diseases: Steering Committee

- ❖ CDC
- ❖ NIH-ORD
- ❖ EMORY
- ❖ HRSA
- ❖ ASHG
- ❖ ACMG
- ❖ SIMD
- ❖ Genetic Alliance



CETT Program Objectives

- ❖ Promote new genetic test development
- ❖ Translate from research to clinical practice
- ❖ Educate about each rare genetic disease; research opportunities & clinical impact
- ❖ Collect and Store clinical and genetic information



Previous Conferences

- ❖ **May 19–21, 2004 Atlanta, GA**
Promoting Quality Laboratory Testing for Rare Diseases: Key to Ensuring Quality Genetic Testing for Rare Diseases
<http://www.phppo.cdc.gov/dls/genetics/RareDiseaseConf.aspx>

- ❖ **September 26–27, 2005 Washington, DC**
Access to Quality Testing for Rare Diseases
<http://rarediseases.info.nih.gov/QTRD>

- ❖ **October 6-7, 2006 Atlanta, GA**
Quality, Access, and Sustainability of Biochemical Genetic Testing



Biochemical Genetic Testing - Recommendations

- ❖ **Extend CETT pilot program to support the translation and implementation of new BGT and those only available in non-USA laboratories**
- ❖ **A laboratory consortium should be established to monitor, determine, and coordinate tests that must be available to meet the standards of care.**
- ❖ **Training of both laboratory and clinical personnel should be encouraged in the area of BGT**
- ❖ **Guidelines should be developed to ensure the quality of testing, result interpretation, and diagnosis for inherited metabolic disorders and other genetic diseases**
- ❖ **Quality assurance measures should be enhanced**
- ❖ **International cooperation and coordination should be improved**
- ❖ **Information resources should be enhanced to provide easy-to-access, user-friendly information on BGT, testing services, and test strategies**



Outcomes of BGT Meeting

- ❖ **NIH ORD CETT program to support translation of BGT from research to practice.**
- ❖ **Gene Tests will provide specific information on BGT, either by expanding the current capacity or by setting up a companion site through subcontracting.**
- ❖ **ACMG and SIMD are committed to developing testing guidelines needed by users and providers of BGT services in collaboration with other professional organizations.**
- ❖ **CDC will prepare a report of this meeting and will post information on this meeting (background information, presentations and discussion, and meeting report)**
- ❖ **Steering Committee will review the recommendations, to refine roles and responsibilities and to engage efforts and additional collaborations as needed.**
- ❖ **Manuscript (s) will be prepared for peer-reviewed publication.**
- ❖ **A follow-up meeting to review development and formulate further recommendations.**



Rare Diseases Approved for Translations

Molecular Genetic Tests (10 approved and 8 in review):

- ❖ Cherubism (Toronto Sick Children)
- ❖ Cornelia de Lange Syndrome (U Chicago) **Clinically Available
- ❖ Primary Ciliary Dyskinesias (University of North Carolina – Chapel Hill)
- ❖ Infantile Neuroaxonal Dystrophy (Oregon Health and Science U)
- ❖ Joubert Syndrome (Prevention Genetics)
- ❖ Kallman Syndrome (Gene DX)
- ❖ Progressive Familial Intrahepatic Cholestasis (8 diseases/3 genes) (Baylor U-Mitochondrial Lab)
- ❖ X-Linked Periventricular nodular heterotopia (Harvard U)



Rare Diseases Approved for Translations (cont.)

Multiple Methodology Approach to testing:

- ❖ X-Linked Chondrodysplasia- molecular genetic testing in collaboration with biochemical genetic sterol analysis-clinical mass spectrometry (U Chicago)
- ❖ Silver Russel Syndrome-methylation (quantitative Taqman) assay and molecular genetic testing (Emory U)



Advocate Mentors

- ❖ Group of disease specific advocate leaders
- ❖ Resource to each collaborative group
- ❖ Assigned early in the process



Experience of CETT Program to Date (cont.)

- ❖ Need for templates of educational materials for understanding genetic test and rare diseases for clinicians and individuals and families
- ❖ Need for report forms to be interpretable to non-genetic clinicians (example language)
- ❖ Need for test results to be understandable and provide limitations of test



- ❖ Review Board - Three teams of five members, one each from:
 - Laboratory genetics
 - Medical genetics
 - Research
 - Primary care
 - Disease specific advocacy group



Applicants = Collaborative Group

- ❖ Clinical (CLIA-certified) laboratory
- ❖ Researcher (laboratory and/or clinician)
- ❖ Disease specific advocacy group



Procedures

- ❖ Review Scientific Evidence
- ❖ Proposed Methodology
- ❖ Impact on Healthcare
- ❖ Laboratory Qualifications
- ❖ Data Collection Plan
- ❖ Educational Materials Development and Dissemination Plans
- ❖ Evidence of Collaboration
- ❖ Funding/Commitment
- ❖ Web Site: <http://www.cettprogram.org/>



Potential Outcomes

- ❖ Improve understanding of CLIA and quality standards
- ❖ Improve dialogue among stakeholders: Clinical laboratories, reference laboratories, researchers, clinicians, disease specific advocates, oversight bodies, payers
- ❖ Collect genotype/clinical information:
 - improve test interpretations
 - genotype/phenotype correlations



Program Staff

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