



Genetic Testing Reference Materials: Ensuring the Quality of Genetic Testing

ICORD Meeting

May 21, 2008

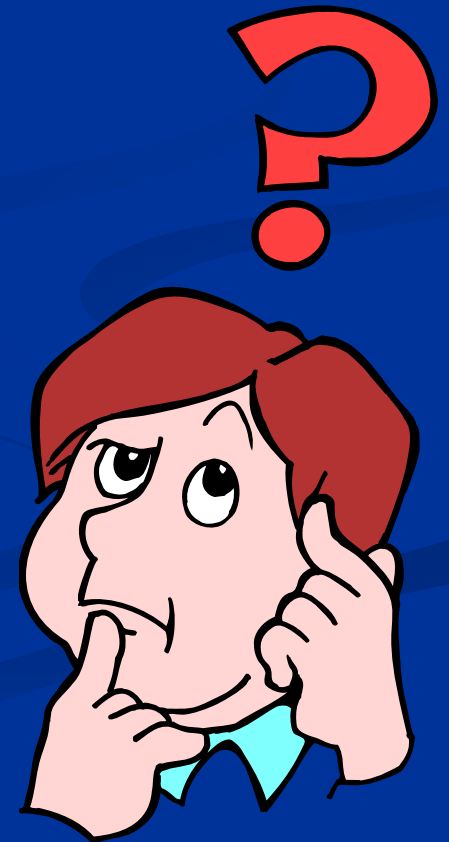
Lisa Kalman, PhD
Coordinator, GeT-RM
CDC

LKalman@cdc.gov





How does the genetic testing laboratory know that a test is working properly and will give accurate results?





Quality Control



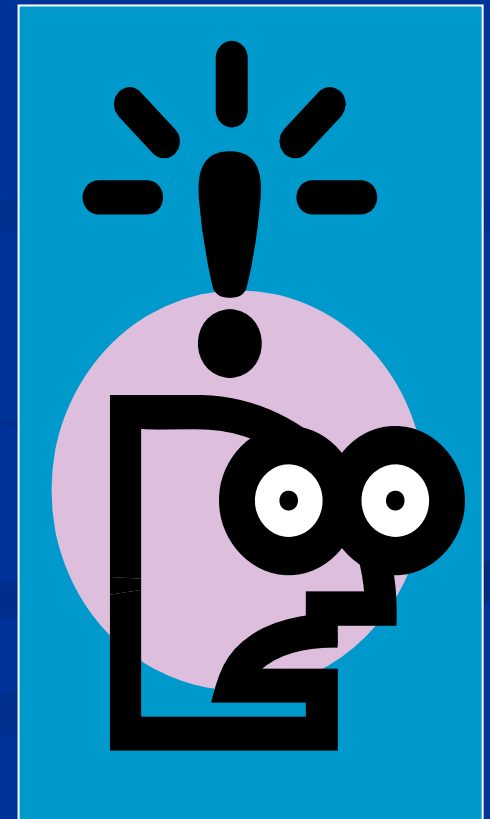
Reference Materials (Quality Controls) are used for:

- Daily quality control
- Comparing results between laboratories
- Proficiency testing
- Test calibration
- Test development and validation



Reference Materials for Genetic Testing

Reference materials are **NOT** publicly available for most genetic tests!





Genetic Testing Reference Material Program (GeT-RM)

A CDC-based program (in collaboration with the genetic testing community) to improve the availability of reference materials for genetic testing





Roles of GeT-RM

- Information exchange about reference materials for genetic testing
- Monitor reference material needs
- Facilitate submission, development and characterization of reference materials



GeT-RM Reference Material Projects

Genomic DNA Characterization

Completed:

- Fragile X
- Huntington Disease
- Cystic Fibrosis
- Ashkenazi Jewish Panel (9 disorders)

In Progress:

- Newborn Screening disorders
- MEN2A
- Alpha 1 antitrypsin
 - MTHFR
 - BRCA1/2 (Ashkenazi alleles)



GeT-RM Reference Material Projects

Genomic DNA Characterization

Future???:

- Duchenne/Becker Muscular Dystrophy
- Newborn Screening
- Hemoglobinopathies
- Pharmacogenetics

- Other Suggestions?????



Continued Reference Material Development...

Big Problem...

- **Limited access to patient blood samples!**
- **Lack of available cell lines!!!!!!**





GeT-RM is expanding into other areas of molecular testing -

- ✓ **Molecular Oncology**
- ✓ **Biochemical Genetic Tests**



**THANK
YOU**



GeT-RM Website:

<http://wwwn.cdc.gov/dls/genetics/rmmaterials/default.aspx>