**The Genetic Testing Reference Materials Coordination Program (GeT-RM)-
A sustainable community process to improve availability of appropriate reference materials for genetic testing**

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Background: The expansion of molecular genetic testing in clinical and public health practice has increased the need for appropriate and characterized cell line/genomic DNA reference materials for quality assurance, test validation, proficiency testing, and development of new genetic tests. However, despite the growing test volume and the rapidly increasing number of tests being offered, the necessary reference materials are not available for the vast majority of tests.

Method: The U.S. Centers for Disease Control and Prevention (CDC), in collaboration with members of the genetic testing community, has developed a program to improve public availability of reference materials and facilitate information exchange and communication on reference materials development, contribution, characterization, distribution, and needs assessment. This CDC-based Genetic Testing Reference Materials Coordination Program (GeT-RM) provides continuing support and coordination to improve genetic testing reference material availability. The GeT-RM Program 1) facilitates the identification, procurement, characterization and availability of needed reference materials; 2) facilitates exchange of quality control and reference material-related information; and 3) explores collaborative efforts for ongoing needs monitoring and materials development.

Results: The GeT-RM has characterized reference materials for Huntington disease, Fragile X, disorders on the Ashkenazi Jewish Panel (Bloom syndrome, f. dysautonomia, Canavan disease, Niemann-Pick disease, Tay-Sachs disease, Gaucher disease, Glycogen storage disease type 1a, Fanconi anemia and mucolipidosis type IV). The GeT-RM is currently characterizing reference materials for cystic fibrosis and a number of other genetic tests, including many newborn screening disorders. The Get-RM program also collects genetic information from other sources about publicly available cell lines/DNA with clinically important mutations, including many pharmacogenetic polymorphisms, which may be useful as reference materials. This information is posted on the program website. To date, the GeT-RM has focused its efforts on DNA based testing for inherited genetic disorders. However, there is a similar lack of reference materials for other areas of genetics, including molecular oncology, molecular infectious disease testing and biochemical genetic testing. To address these needs, the GeT-RM, together with the genetics community, relevant professional organizations and government agencies is working to provide information about currently available reference materials and are considering mechanisms to address reference material needs for these areas.

Conclusions: The increased availability of characterized reference materials, which can be used for quality assurance, proficiency testing, test development and research, will help to improve the quality and accuracy of genetic testing. More information is available at the GeT-RM website, http://www.phppo.cdc.gov/dls/genetics/qcmaterials/default.aspx.