MedicalXpress

Cystic fibrosis testing -- next steps

April 27 2009

Three reports describing advances in cystic fibrosis genetic testing appear in the May 2009 issue of The *Journal of Molecular Diagnostics*.

Cystic fibrosis is a hereditary disease that affects mucus secretions in the lungs, liver, pancreas, and intestines. Approximately 1 in 4000 children born in the United States is affected with cystic fibrosis. Cystic fibrosis is an autosomal recessive disease caused by mutations in the *CFTR* gene; cystic fibrosis patients must inherit a mutated gene from each parent.

Genetic screening for cystic fibrosis carrier mutations (one copy of a mutated gene) is universally recommended for the reproductive-age population. Current professional guidelines call for screening a panel of 23 common mutations in *CFTR*; however, many laboratories screen for an expanded panel of mutations. In the May 2009 issue of The *Journal of Molecular Diagnostics*, three articles describe improvements in cystic fibrosis genetic screening. In one article, Pratt et al describes a project coordinated by the Centers for Disease Control and Prevention's Genetic Testing Reference Material (GeT-RM) Program to develop a set of reference materials for the expanded cystic fibrosis panel of mutations. The public availability of these materials will help to ensure the accuracy of cystic fibrosis genetic testing. The reports by Schwartz et al and Hantash et al identify mutations that may lead to false screening results, either due to a large deletion in *CFTR* or because of mutations that interfere with laboratory screening methods.

Wayne Grody, of the UCLA School of Medicine, Los Angeles, CA, who is not affiliated with these studies, states "Taken together, these three

papers demonstrate how the widespread and thoughtful experience with [cystic fibrosis] mutation testing and screening continues to reveal new insights about the mutational alleles of the *CFTR* gene and further refinements in how best to detect them and assure appropriate quality control while doing so."

More information:

Hantash FM, Rebuyon A, Peng M, Redman JB, Sun W, Strom CM: Apparent homozygosity of a novel frame shift mutation in the CFTRA gene because of a large deletion. J Mol Diagn 2009, 253-256

Pratt VM, Caggana M, Bridges C, Buller AM, DiAntonio L, Highsmith WE, Holtegaard LM, Muralidharan K, Rohlfs EM, Tarleton J, Toji L, Barker SD, Kalman LV: Development of genomic reference materials for cystic fibrosis testing. J Mol Diagn 2009, 186-193

Schwartz KM, Pike-Buchanan LL, Muralidharan K, Redman JB, Wilson JA, Jarvis M, Cura MG, Pratt VM: Identification of cystic fibrosis (CF) variants by PCR/oligonucleotide ligation (OLA) assay. J Mol Diagn 2009, 211-215

Source: American Journal of Pathology (news : web)