Genetic technology detects CHARGE syndrome in prenatal sample

December 6 2012

Brief Report in the December 6, 2012 issue of the *New England Journal of Medicine* (*NEJM*) focuses on findings from Cynthia Morton, PhD, director of Cytogenetics at Brigham and Women's Hospital and senior study author, and colleagues on a prenatal case study involving a new, balanced translocation (a genetic abnormality caused by chromosomal rearrangements) between chromosomes 6 and 8.

The researchers used customized whole-genome "jumping libraries" to identify the precise abnormal areas on the chromosomes and saw a disruption in the CHD7 gene, mutations in which are known to cause CHARGE syndrome (a rare disease that can lead to birth defects of the eyes, ears, heart and other organs). Clinicians diagnosed CHARGE syndrome in the child at birth.

Along with Dr. Morton's case study, two other studies involving 4,406 women undergoing prenatal diagnosis and 532 stillbirths, respectively, will appear in NEJM. The reports will appear with an accompanying editorial, "Application of Genomic Technology in Prenatal Diagnosis." In the editorial Lorraine Dugoff, MD, University of Pennsylvania, highlights how the reports show the power, complexity and some of the pitfalls of using new genomic technology in clinical practice.

Dr. Morton's major research interests are molecular and cytogenetic studies of uterine leiomyomata, hereditary hearing loss and cytogenetic approaches to gene discovery for developmental disorders.

Medical Xpress

Provided by Brigham and Women's Hospital

Citation: Genetic technology detects CHARGE syndrome in prenatal sample (2012, December 6) retrieved 4 May 2024 from https://medicalxpress.com/news/2012-12-genetic-technology-syndrome-prenatal-sample.html

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