Service Priorities and Programmes

Electronic Presentations

Convention ID: 904
Submitting author:
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Array CGH testing in Prenatal Diagnosis: A Promising New Service with Improved Diagnostic Yield and Shortened Reporting Time

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Keywords:
Comparative Genomic Hybridization
Prenatal diagnosis

Introduction

Array Comparative Genomic Hybridization (aCGH) with genome-wide coverage has proved to be valuable for postnatal and prenatal studies. Traditionally, prenatal diagnosis of chromosomal abnormalities has relied on conventional cytogenetics which required cell culture and chromosome analysis under microscope.

Objectives

To evaluate the use of aCGH for prenatal diagnosis in Hong Kong by comparing the (1) concordance of results and (2) reporting time against that of conventional cytogenetics.

Methodology

Two hundred and fifty-two prenatal samples (mostly amniotic fluid or chorionic villi) were analysed by both NimbleGen whole genome oligo array CGX-12 with 135K oligo probes/array as well as conventional karyotyping from January 2011 to December 2011 in Tsan Yuk Hospital Prenatal Diagnostic Laboratory. The proportion of cases detected with clinically significant copy number variations (CNVs) by aCGH were compared to abnormal karyotypes by conventional cytogenetics.

Result

Clinically significant CNVs were detected in 42 (16.7%) out of 252 samples. 12 abnormalities (4.8%) were not detected by conventional karyotyping. Among 106 samples with ultrasound abnormalities, clinically significant CNVs were detected in 36 (34%). 12 abnormalities (11.3%) were not detected by conventional karyotyping. These additionally diagnosed cases include complex unbalanced rearrangements and microdeletions which are clinically important and relevant for counselling on future reproductive risks. Array CGH also enhanced the characterization of the origin and genetic content of supernumerary marker chromosomes. The reporting time has shortened from 14 days for karyotyping to around 5 days for aCGH initial reporting. Conclusion: Array CGH is a promising prenatal diagnostic test with improved

diagnostic yield and shortened reporting time by 64%.