



## Service Priorities and Programmes Electronic Presentations

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### **Demonstration Trial on Replacement of Cytogenetics by Array Comparative Genomic Hybridization in Prenatal Diagnosis**

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#### **Introduction**

Chromosomal microarray has been increasingly used in prenatal diagnosis as first line test because of improved diagnostic yield and shorter reporting time compared to conventional cytogenetics. However this remained a self-finance test to women.

#### **Objectives**

To demonstrate the clinical acceptability on the use of array comparative genomic hybridization (aCGH) to replace cytogenetics in prenatal diagnosis. The impact on genetic counselling and laboratory resources shall be assessed.

#### **Methodology**

Women requiring invasive prenatal diagnosis by chorionic villus sampling or amniocentesis were offered the options of conventional cytogenetics or aCGH with standard pretest counselling by trained midwives. For those who opted for aCGH, rapid aneuploidy testing by quantitative fluorescent polymerase chain reaction (QF-PCR) was performed first to exclude common aneuploidies and triploidy. aCGH would be arranged for those with normal QF-PCR results. Conventional cytogenetics was reserved for those with abnormal aCGH.

#### **Result**

From November 2014 to January 2016, 130 women who required invasive prenatal tests at two obstetric units were offered aCGH instead of cytogenetic test at no cost to women. 29 did not proceed to aCGH testing because QF-PCR showed fetal

aneuploidy and 1 withdrew from the study. One hundred aCGH tests were performed. There were 83 (63.8%) samples with normal aCGH results and completed reporting with aCGH only at median reporting time of 6 calendar days (95% reported within 14 days). 47 (36.2%) samples required karyotyping in addition to aCGH, in view of fetal aneuploidy (28), abnormal aCGH results (11), withdrawal or reported while awaiting clarification of aCGH results (8). This translated into additional 8.5% (11/130) postnatal counselling of abnormal aCGH results, saving of nearly two-third of laboratory manpower on prenatal chromosomal study with 60% improvement of cytogenetics reporting time within the study period. This study showed that aCGH can be used post rapid aneuploidy QF-PCR testing to replace about two-third of the cytogenetic study for prenatal diagnosis, with acceptable pre and post-test counselling workload.