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Microarray analysis of Cell Free Fetal (CFF) DNA using amniotic fluid supernatant

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The application of array comparative genomic hybridization (CGH) in the prenatal diagnostic setting is an attractive alternative to karyotyping, providing a genome-wide screen for genomic imbalances at a higher resolution, allowing for the detection of all known recurrent microdeletion/ microduplication syndromes. Further advantages include a higher degree of automation, and with no cell culture required array CGH provides a more rapid result than karyotyping while overcoming the issue of culture failure. Success rates for amniotic fluid samples karyotype analysis in our laboratory are high (99%), however samples can be compromised by low volume, late gestation or blood/meconium. The aim of this study was to investigate the possibility of performe a prenatal Chromosome Microarray Analysis using cell free fetal (cff) DNA from stored amniotic fluid supernatant. Cell free fetal DNA was extracted from 10 amniotic fluid supernatant samples using QIAamp® DNA Blood Maxi Kit and run on Agilent 8x60k ISCA. The 10 samples had been previously characterized by routine Karyotype. The analysis included no abnormality (n=6), trisomy 21 (n=2), trisomy 18 (n=1) and a structural chromosome abnormality involving chromosome 18 (n=1). Aside from the identification of pathogenic copy number variations (CNVs), array CGH allowed a more precise characterization of imbalances. The use of cff DNA for CMA in amniotic fluid supernatant has been only recently reported in the literature (Madjunkova et al., 2014) and may be of benefit for compromised amniotic fluid samples. Keywords: Array CGH, prenatal cytogenetic, copy number changes. Projeto 10-560