Evaluation of Array Comparative genomic Hybridisation in prenatal diagnosis of fetal anomalies: a multicentre cohort study with cost analysis and assessment of patient, health professional and commissioner preferences for array comparative genomic hybridisation

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Disclaimer: This report contains transcripts of interviews conducted in the course of the research and contains language that may offend some readers.

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Plain English summary

The EACH Study

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Plain English summary

One of the main aims of ultrasound scans during pregnancy is to detect fetal abnormalities, some of which are due to imbalances (gains or losses) of part or all of a chromosome. Each cell in the body contains 23 pairs of chromosomes and one of each pair is inherited from each parent. The chromosomes carry a person's genetic information stored as deoxyribonucleic acid. Babies with chromosomal imbalances have complex problems. Testing for chromosome imbalances involves tests (e.g. amniocentesis) that occasionally cause miscarriage. Major chromosome problems (e.g. Down syndrome) can be detected quickly by a polymerase chain reaction (PCR) test. Less common imbalances require the baby's cells to be grown and examined; this test (karyotyping) detects only large chromosomal imbalances. Chromosomal microarray (CMA) is a new genetic test that detects smaller chromosomal imbalances. Interpreting CMA is complex as not all imbalances cause problems. The purpose of the Evaluation of Array Comparative genomic Hybridisation (EACH) study was to help the NHS decide whether or not CMA should replace karyotyping.

Over 1100 women with a fetal abnormality detected on ultrasound and without a major chromosomal problem (as indicated by a normal PCR test) participated in the EACH study. CMA detected 3–4% more significant chromosome imbalances than karyotyping when the reason for testing was a major abnormality in one of the baby's organs, but not when there was increased fluid at the back of the baby's neck (nuchal translucency). The time from setting up the test to reporting the result of the test was 5 days less with CMA than karyotyping. Although the cost of CMA (£322) was slightly more than karyotyping, the cost for each extra chromosome imbalance detected (which was thought to explain the abnormal scan) was £9418. Interviews suggested that parents and health professionals found the CMA test acceptable despite the uncertainties it may introduce. The results suggest that CMA should replace karyotyping for the detection of chromosome imbalances in abnormal fetuses.

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