Prenatal chromosome microarray: Information for parents

What is prenatal chromosome microarray?

Prenatal chromosome microarray (CMA) is a test used to pick up chromosome changes which are too small to be seen by the standard tests available in pregnancy.

What are chromosomes?

Chromosomes are the structures which carry genes. Genes are the body's instructions. Each cell in the body has 46 chromosomes in 23 pairs. We inherit one member of each chromosome pair from each parent. Girls have two X chromosomes (XX) and boys have an X and a Y chromosome (XY). The other chromosome pairs are numbered from 1 to 22. Having too much or too little chromosomal material can lead to physical and/or learning problems.

In order to diagnose a chromosome abnormality in an unborn baby it is necessary to examine the cells from the pregnancy. A sample of these cells can be obtained by either a chorionic villus sampling (CVS) or by an amniocentesis. Your doctor or midwife will be able to tell you more about these tests. Once the laboratory has the sample of cells, they will first do a test which involves checking for the three most common chromosome changes, including Down syndrome (trisomy 21), Edward syndrome (trisomy 18) and Patau syndrome (trisomy 13). These results are usually available after 48 hours.

Why has chromosome microarray (CMA) been offered to you?

If the results of the first test show that one of these three conditions is NOT responsible for the ultrasound findings which have led to the testing of the pregnancy, a second test of the same CVS or amniocentesis sample will be offered. This is the chromosome microarray (CMA). Chromosome microarray is a laboratory test that is used to see if the baby has other chromosome changes which may explain the ultrasound findings. The results will be ready in about two weeks.

What are the advantages of chromosome microarray?

The main advantage of microarray is that it can detect very small chromosome changes which cannot be seen by the standard chromosome test. These changes are called micro deletions (tiny pieces of missing chromosome) and micro duplications (tiny pieces of extra chromosome). A change in the chromosomes may explain the ultrasound findings and allow more precise information to be given about what this means for your baby.

What are the disadvantages and limitations of chromosome microarray?

- Chromosome microarray does not detect all chromosome changes as some are too small to be identified by this test.
- Some conditions are caused by changes in individual genes and chromosome microarray cannot detect tiny changes in individual genes.
- Sometimes results can be difficult to interpret unless a blood sample from both parents is available for comparison.
- Chromosome microarray may detect changes called 'variants of unknown significance'. This means there is not yet enough information available to know if these changes have caused the ultrasound findings, or not. Where there is uncertainty, these variants may not be reported because this result does not provide reliable information about how your baby will develop or how the pregnancy will progress.
- If your child has a chromosome microarray performed *after* they are born this may reveal a result not found by the testing in pregnancy, as we only report results in pregnancy that could have caused the ultrasound findings, or results that could have future health implications for your baby

What else may a chromosome microarray tell us?
Chromosome microarray may occasionally identify a chromosome change which is not related to the ultrasound findings but which may have implications for the future health of your baby and possibly for other family members. For example, it may show your baby will have an increased risk of cancer later on in life.

This chromosome change may have been inherited from a healthy parent, or may not. Carriers of these types of chromosome changes could benefit from screening to detect cancer earlier and improve the success of treatment. This may not be relevant at the time of the pregnancy but could be discussed at a later date. This is called an incidental finding.

What are the possible different outcomes of a chromosome microarray test?

1. The result is reported as normal as no chromosome change was found that could explain the ultrasound findings
2. A chromosome change is found which explains the ultrasound findings. This will be discussed in more detail with you.
3. A chromosome change is found, however the significance in relation to the ultrasound findings is unclear. Samples taken from you, and possibly the father of the baby, will be tested as well and the result will be discussed with you if the chromosome change could be related to the ultrasound findings.
4. A chromosome change is found which not the cause of the ultrasound findings is but which may have significance to the future health of the baby. This will be discussed in more detail with you.

Further questions
If you have more questions about the chromosome microarray test, please ask the doctors or midwives in the Fetal Medicine Unit.

Other services
Antenatal Results and Choices (ARC)
Offers information and support to parents before, during and after antenatal screening.
Tel: 0845 077 2290 / 020 7713 7486 **Web:** www.arc-uk.org

UNIQUE
UNIQUE offers support and information for parents of babies diagnosed with rare chromosome disorders. **Tel:** 0188 372 3356 **Web:** www.rarechromo.co.uk/html/home.asp