Prenatal Microarray Testing

What is Microarray testing?

Microarray testing allows the detection of chromosome imbalances which are too small to be seen by the routine chromosome tests offered during a pregnancy.

Why have you been offered Microarray testing?

Your serum screening results or your ultra sound have shown that there is an increased chance that your baby has a chromosome imbalance. Microarray testing is used to find out if a chromosome imbalance is present in your baby, which may explain these results.

What are chromosomes?

Chromosomes are the structures in each cell of our bodies that carry genetic information or genes. Genes contain instructions to tell the body how to develop and function. Each of our cells has 46 chromosomes these are made up of 23 pairs of chromosomes; we inherit one chromosome from each parent to make a pair.

The pairs are numbered from 1 to 22, from largest to smallest with a final pair of chromosomes called the sex chromosomes; X and Y. Girls have two X chromosomes and boys have one X and one Y chromosome.

Changes in the number or structure of chromosomes increases the risk of physical and intellectual disability. The routine chromosome test is called a Karyotype and uses a microscope to detect changes in chromosomes. This test is not able to detect smaller more subtle changes because they cannot be seen through the microscope. Small changes can still affect growth and development. These small changes are sometimes called microdeletions (tiny pieces of missing chromosome) and microduplications (tiny pieces of extra chromosome).

What are the advantages of Microarray Testing?

The main advantage of microarray testing is the ability to explore all 46 chromosomes in detail. This means we can detect extra or missing chromosomes or other chromosome changes much more precisely than with the routine tests.

An imbalance in the chromosomes may explain your serum screening result or ultrasound findings and allow more precise information to be given about the diagnosis. It may indicate if the condition is hereditary (can be passed from parent to child) and give information about the chance of the same condition affecting a future pregnancy.

What are the disadvantages of Microarray Testing?

Microarray testing will not detect all chromosome imbalances. Some are too small to be identified and some particular sorts of changes cannot be picked up by our test. Genetic conditions are not only caused by chromosome imbalances. So the test may not be able to give an explanation for your scan finding or serum screening result.

There may be difficulty in interpreting the results of the microarray test particularly if one of the parental blood samples is not available for comparison.

Microarray testing may detect chromosome changes called "variants of unknown significance". There is not enough information in the medical literature about these to be certain about how or if these changes are linked to serum screening results or ultrasound findings.

Rarely, an unexpected chromosome change can be identified which is unrelated to the serum screening or ultrasound findings but may have an impact on the future health of your baby or other family members.

How will you be given the results?

The results should be ready within two weeks. The specialist midwife will contact you with the results. If there are any chromosome imbalances found you will be offered an appointment with a Clinical Geneticist to discuss the results.

Further Questions?

If you have more questions about the microarray test then please contact the Specialist Midwives in prenatal diagnosis:

LOTHIAN
Fetal Medicine Unit, Royal Infirmary Edinburgh
0131 242 2659
FIFE
Fetal Medicine Midwives
07767 618365 / 07770 644387
BORDERS
Pregnancy Assessment Unit
01896 826735

Your local Genetic Service?

SE of Scotland Clinical Genetic Services
Western General Hospital, Crewe Rd,
Edinburgh, EH4 2XU
0131 537 1116
wgh.clinicalgenetics@luht.scot.nhs.uk

South East of Scotland Cytogenetic Service

NHS Lothian

This leaflet is written for people who are considering prenatal microarray testing

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