What is microarray testing?

Microarray testing checks for chromosome imbalances in your baby. These imbalances may not involve the chromosomes that are investigated by the QF-PCR test.

Why have you been offered microarray testing?

Your family history or your ultrasound scan indicates that your baby has an increased chance of a chromosome imbalance. Microarray testing is used to find out if a chromosome imbalance is present in your baby.

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. There are usually two copies of each chromosome and these pairs of chromosomes are numbered from number 1, which is the largest pair, to number 22, which is the smallest pair. There is a 23rd pair, and in males this pair is made up of one X and one Y chromosome. In females this pair is made up of two X chromosomes.

A change in the number or structure of chromosomes can result in birth defects, learning problems, behaviour issues or other health problems. Even small changes may still affect growth and development. These small changes are sometimes called microdeletions (a tiny piece of chromosome is missing) and micro-duplications (a tiny piece of chromosome is doubled up).

What type of sample is needed?

The test is carried out on the cells obtained from an Amniocentesis or CVS (chorionic villus sample).

Some of the changes that can be detected by the microarray test may run in families and don't always cause problems to the baby. Therefore, blood samples from both parents are ideally taken at the same time as the amniotic fluid or CVS, to allow a better understanding of the significance of these changes.

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 very precisely.

An imbalance in the chromosomes may explain your 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 by this test, and some particular sorts of changes cannot be picked up by this test. These include balanced chromosome rearrangements, in which pieces of chromosomes have broken and rejoined in a balanced form.

Genetic conditions are not only caused by chromosome imbalances. So the test may not be able to give an explanation for your scan finding.

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 the reason for the test.

Rarely, an unexpected chromosome change can be identified which is unrelated to the reason for the test 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 2 weeks. If no imbalances are detected in the baby's chromosomes 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.

Additional information and support

ARC (Antenatal Results and Choices) is a charity that supports people through testing in pregnancy. They have a really helpful website.

www.arc-uk.org

Tel 0845 077 2290
or 0207 713 7486 via mobile
Email: info@arc-uk.org

Further Questions

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

Lothian

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0131 242 2659

Fife

Fetal Medicine midwife
07767 618365
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Prenatal Microarray Testing

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

South East of Scotland Clinical Genetic Service