Guy's and St Thomas' NHS Foundation Trust

Prenatal microarray

This leaflet provides information about prenatal microarray. It should be read before you sign a consent form agreeing to this test. If you have any further questions, please speak to a doctor or midwife caring for you.

What is prenatal array CGH?

Prenatal microarray is a detailed test used to identify chromosome changes.

What are chromosomes?

Chromosomes are structures which carry genes, and genes are instructions which tell the body how to develop and function. Each cell in the body has 46 chromosomes in 23 pairs. We inherit one of a 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 cause significant problems with physical and intellectual development.

If ultrasound scans have identified a possible problem during pregnancy, you will be offered a test to examine the baby's cells in order to check for any abnormalities in the chromosomes. A sample is obtained by two types of invasive tests during pregnancy; either a Chorionic Villus Sample (CVS) or an Amniocentesis. The first laboratory test (called a QF-PCR test) involves checking some of your baby's chromosomes for the most common abnormalities, such as Down syndrome (trisomy 21), Edwards's syndrome (trisomy 18) and Patau syndrome (trisomy 13). These results are usually available after 48 hours.

Why has microarray been offered to you?

If the result of the first laboratory test (QF-PCR) is normal and ultrasound scans have shown that your baby has an increased risk of too much or too little chromosomal material (we call this a chromosome change), you will be offered a microarray test. Microarray is a laboratory test that is used to see if the baby has a chromosome change which may explain the ultrasound findings. The results will be available in two weeks.

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What are the advantages of microarray?

The main advantage of microarray is that it can detect very small chromosome changes. These changes can be small deletions (tiny pieces of missing chromosome) and 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 microarray?

- Microarray does not detect all chromosome changes including very small changes in individual genes. Occasionally, the quality of the microarray result is not good enough to find some small changes, although most significant chromosome changes are seen.
- Sometimes the microarray result from the pregnancy can be difficult to interpret. In these cases, it can be helpful to test blood from the parents to see if the genetic change we have found in the pregnancy is inherited. This may help us decide if the genetic change is the cause of the ultrasound findings. Testing parents will also confirm their relationship to the pregnancy and may identify instances where one or both parents is not biologically related to the patient (for example in the case of sperm or egg donation).
- The microarray test may also detect changes called *'variants of unknown significance'* (VUS). This means there is not yet enough information available to know if these are significant or not. Where there is uncertainty, these variants may not be reported. This is because a VUS may not give any further information about how your baby will develop or how the pregnancy will progress. This is in line with national guidelines on 'Recommendations for the use of chromosome microarray in pregnancy' from the Royal College of Pathologists (June 2015 version 1 and any future updated versions).
- Microarray might detect chromosome changes which are considered 'low risk susceptibility factors' for some medical or developmental issues. Because the associated risk is very low for some of these changes, or there is not enough information available to understand their real impact, these will not be reported. This is in line with the national guidelines.

What else may microarray tell us?

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. This is called an *incidental finding*. For example, it may show your baby will have an increased risk of cancer later on in life. This chromosome change may or may not have been inherited from a healthy parent.

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 if there is an incidental finding this will be discussed with the geneticist.

A microarray might detect chromosome changes which are susceptibility factors or which have incomplete penetrance for some medical or developmental issues. This means that someone with a particular copy number change is more likely to have (or develop in future) a particular clinical feature, but that some people with that same copy number change will never have it. These changes can be difficult to understand; they can be inherited from a completely unaffected parent, or from a very mildly affected parent.

What are the possible outcomes of a prenatal microarray test?

Examples of findings that will be reported include:

- *'No abnormality detected'* – this means the test did not show a genetic change that explains the ultrasound scan findings. Please note, it does not mean that there is no genetic anomaly as there could still be a single gene or chromosomal change that the microarray has not detected or has not been reported as per national guidelines.
- *'Copy number variant of uncertain significance*'- this means that a chromosomal change has been found that may be clinically significant, but currently there is insufficient evidence to be completely sure. Further information from parents or published research may be needed.
- *'Likely pathogenic copy number variant'*- this means a copy number change was found that is likely to be significant but it is not possible to be completely sure that it has caused the patient's clinical features. This may include incomplete penetrance and susceptibility loci, as explained above.
- *'Pathogenic copy number variant'* – this is a significant result which explains the ultrasound scan findings and will be discussed in more detail with you, or an incidental finding as previously explained.

When will I receive the result of the investigation?

Results are usually available within two weeks. The specialist midwife will call you with the results once they are available. If any chromosome imbalances are detected, you will be offered an appointment with an obstetrician or clinical geneticist to discuss the findings further. If you feel that you would like further support, please inform the midwife or doctor involved in your care.

Useful sources of information

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

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

Contact us

If you have more questions about prenatal array CGH, please contact the specialist midwives in the **Fetal Medicine Unit** on **020 7188 8003** (Monday to Friday, 9am to 5pm).
If you have a significant medical problem out of hours, contact your GP in the first instance. If it is an emergency, call 999 for an ambulance.
For more information leaflets on conditions, procedures, treatments and services offered at our hospitals, please visit www.guysandstthomas.nhs.uk/leaflets

Pharmacy Medicines Helpline
If you have any questions or concerns about your medicines, please speak to the staff caring for you or call our helpline.
t: 020 7188 8748 9am to 5pm, Monday to Friday

Your comments and concerns
For advice, support or to raise a concern, contact our Patient Advice and Liaison Service (PALS).
To make a complaint, contact the complaints department.
t: 020 7188 8801 (PALS) **e:** pals@gstt.nhs.uk
t: 020 7188 3514 (complaints) **e:** complaints2@gstt.nhs.uk

Language and accessible support services
If you need an interpreter or information about your care in a different language or format, please get in touch:
t: 020 7188 8815 **e:** languagesupport@gstt.nhs.uk

NHS 111
Offers medical help and advice from fully trained advisers supported by experienced nurses and paramedics. Available over the phone 24 hours a day.
t: 111

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