Patient Information

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Prenatal Array CGH

Clinical Genetics and Fetal Medicine

This leaflet provides information about prenatal array CGH (comparative genomic hybridisation). It should be read before you sign a consent form agreeing to this test. If you have any further questions, please speak to your doctor or midwife.

What is a prenatal array CGH test?

Array CGH is sometimes called a microarray test. It is a detailed chromosome test that looks to see if your baby has missing genetic material (deletion) or extra genetic material (duplication). Having too much or too little genetic material can sometimes cause problems with physical and/or intellectual development. If ultrasound scans have identified a possible problem, you will be offered a test to examine your baby's chromosomes. A sample is obtained by either a Chorionic Villus Sample (CVS) or an Amniocentesis.

What are chromosomes?

Chromosomes are structures that contain our genetic material (genes). We have around 20,000 genes which provide instructions that tell our body how to develop and function. Every cell in the body has 46 chromosomes. These are arranged in 23 pairs. Females have two X chromosomes (XX) and males have an X and a Y chromosome (XY). The remaining chromosome pairs are numbered from 1-22. We inherit one of each chromosome from our mother and one of each chromosome from our father.

What tests will the laboratory do on the sample?

The first laboratory test is called QF-PCR. It tests for the most common chromosome abnormalities: Down syndrome (trisomy 21), Edwards syndrome (trisomy 18) and Patau syndrome (trisomy 13).

If the QF-PCR result is normal the laboratory will go on to perform the array CGH test.

What are the advantages of array CGH?

The main advantage of array CGH is that it can detect very small chromosome deletions and duplications. These are sometimes also called copy number variants (CNVs). Identifying a CNV may help to explain the ultrasound scan findings and may enable us to give you more precise information about what they could mean for your baby's development.

What are the limitations of array CGH?

Array CGH does not detect all genetic changes. It cannot detect very tiny deletions or duplications or changes within individual genes.

Sometimes the array CGH result can be difficult to interpret. In these cases, it can be helpful to test a blood sample from both parents to see if the copy number variant has been inherited. This may help to decide whether it explains the ultrasound scan findings.

Some copy number variants do not cause any problems and are just normal variations, i.e. part of what makes us different from one another. Sometimes it is very difficult to tell if a CNV is a normal variation or one which could cause abnormalities. When we cannot fully interpret a change it is called a 'variant of uncertain significance' (VUS). Variants of uncertain significance are not usually reported by the genetic laboratory. VUS will only be reported if it may be possible to confirm it is significant by doing further tests on the sample, or a further scan. This decision has been made in line with National Guidance[1].

Array CGH might detect a specific CNV associated with a small increased chance of developmental delay or behavioural problems. Most people with these specific changes do not have any problems. They are unlikely to be the cause of your baby's ultrasound scan findings. These specific changes will not be reported because the chance of them causing a problem is small. This decision also follows National Guidance[1].

What else may array CGH tell us?

Occasionally array CGH identifies a chromosome change which is not related to the ultrasound findings, but which may have implications for the future health of your baby and/or possibly for other family members. These are called 'incidental findings'. Not all incidental findings will be reported. They will be reported if screening or treatment is available to reduce the chance of health problems. If this happens, the result will be discussed with you and you may be referred to the Clinical Genetics department for further information.

What are the possible results of a prenatal array CGH test?

There are three possible outcomes for this test:

- **A normal chromosome result.** This means that the test did not identify a chromosome change that explains the ultrasound scan findings. This does not exclude a genetic cause for the ultrasound findings as there may still be an individual gene change or a chromosome change too small for the array CGH to detect.
- **A significant copy number variant.** This means that the test identified a significant chromosome change. This will be discussed with you and you may be referred to the Clinical Genetics department.
- **An uncertain result.** This means that a chromosome change has been found but the significance of the change is not yet clear, therefore, we cannot be certain about the effect of the change on your baby. Not all uncertain results will be reported. If an uncertain result is reported it will be discussed with you.

When will I receive the results?

Array CGH testing is performed at the Bristol Genetics Laboratory. The results are usually available within two weeks. A member of the fetal medicine team will usually telephone you with the results once they are available. If a significant copy number variant is detected, or a variant of uncertain significance is reported, you will be offered an appointment with an Obstetrician or Clinical Geneticist to discuss the result further. If you would like further support, please ask your doctor or midwife.

Useful sources of information

Antenatal Results and Choices (ARC)

ARC offers information and support to parents before, during and after antenatal screening.

- Tel: **0845 077 2290 / 020 7713 7486**
- Website: **www.arc-uk.org**

UNIQUE

UNIQUE offers support and information for parents of babies diagnosed with rare chromosome disorders.

- Tel: **01883 723356**
- Website: **www.rarechromo.org**

Contact Us

If you have more questions about prenatal array CGH, please contact the specialist midwives in the Fetal Medicine Team on:

Exeter ..... **01392 406533 / 406540**

North Devon ..... **01271 314037 / 322788**

Torbay **01803 615952**

Truro **01872 253092**

Plymouth **01752 439792**

[1]. Recommendations for the Use of Chromosome Microarray in Pregnancy, Royal College of Pathologists, June 2015