Prenatal testing: An introduction to array CGH

This leaflet offers more information about array CGH – a laboratory technique used to look for alterations in the chromosomes of your unborn baby. If you have any further questions or concerns, please contact the fetal medicine unit (details given at the end of this leaflet).

What are chromosomes?

Chromosomes are the structures in each cell of the body which carry genetic information (genes). Genes contain instructions to tell the body how to develop and function. Each cell has 46 chromosomes in 23 pairs. One half of each pair is inherited from each parent. Changes in the number or structure of chromosomes increase your child’s risk of physical and intellectual disability.

All women in the population are offered screening during pregnancy (combined test and ultrasound scans) to identify any potential problems. If screening identifies a problem, an invasive test is offered. The two types of invasive test available are called chorionic villus sampling (CVS) and amniocentesis, and are performed at different stages of pregnancy. If you agree to testing, your baby’s chromosomes will be analysed using a two-staged approach:

- The first technique, called QF-PCR, will check for the most common abnormalities, such as Down’s syndrome (trisomy 21), Edwards’ syndrome (trisomy 18) and Patau syndrome (trisomy 13). The results from these tests will usually be available within two to three working days.

- If the results of QF-PCR are normal but your baby’s scan reveals structural abnormalities, further chromosome analysis can be done using a technique called array CGH (also known as a microarray).

What is array CGH?

Array CGH (comparative genomic hybridisation) is a laboratory technique used to look for alterations in chromosomes which are too small to see down a microscope. This technique allows us to detect alterations that may cause a genetic condition.

What are the advantages of array CGH?

Array CGH is able to examine all 46 chromosomes in greater detail than other techniques are able to. This means that we can detect small pieces of additional or missing chromosome, which may help to explain abnormal ultrasound findings and contribute more precise information towards the diagnosis.

What are the disadvantages of array CGH?

Some genetic conditions are not caused by chromosome imbalances but by mistakes in individual genes. Array CGH cannot detect the tiny changes in individual genes. Additionally, array CGH will not detect all chromosome imbalances, as some are too small to be identified.

Occasionally, array CGH may detect a chromosome imbalance that we cannot fully interpret, making it difficult to know how, or if, this change is linked to the abnormalities highlighted by the ultrasound. In such cases, we may ask to test the parents of the unborn child to see if the imbalance is naturally inherited. However, uncertainty about the interpretation of the imbalance may remain.
What else can array CGH tell us?

Rarely, array CGH identifies chromosome changes that are unrelated to the ultrasound findings, but which may have implications for the future health of your baby or other family members. We will contact you about these incidental findings if we consider them to be relevant.

Examples of incidental findings include:

- a recognised imbalance that is associated with an increased risk of mild to moderate learning difficulties and/or autistic spectrum disorder. These types of imbalance are often carried by one of the healthy parents. It is not possible to predict how likely it is that the baby would have learning difficulties.

- an imbalance affecting a gene that is associated with a risk of developing breast, bowel or other forms of cancer. The imbalance may be inherited from a healthy parent, but may not. Carriers of these types of imbalances could benefit from screening to detect cancer early 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.

- an imbalance affecting a gene that is associated with an adult onset condition that is currently not treatable. The imbalance may be inherited from a healthy parent, but may not.

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When will I receive the results 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, who will arrange for a genetic counsellor to contact you.

Who can I contact for further information?

If you have more questions about array CGH, please contact the specialist midwives in the Fetal Medicine Unit on 020 8725 0080 (Monday to Friday, 9am to 5pm).

Additional 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

Patient Advice and Liaison Service (PALS)
PALS can offer you on-the-spot advice and information when you have comments or concerns about our services or the care you have received. You can visit the PALS office between 9am and 5pm, Monday to Friday in the main corridor between Grosvenor and Lanesborough Wing (near the lift foyer). Tel: 020 8725 2453    Email: pals@stgeorges.nhs.uk