Prenatal Diagnosis

Prenatal array CGH

Information for parents
What is array CGH (comparative genome hybridisation)?

Array CGH (also known as microarray) allows us to detect chromosome imbalances which are too small to be seen by the routine chromosome tests offered during pregnancy.

Why have we been offered array CGH?

Ultrasound scans have shown that your baby may have an increased risk of a chromosome abnormality. Array CGH is used to find out if a chromosome imbalance is present in the baby, which may explain the ultrasound findings.

What are chromosomes?

Chromosomes are the structures in each cell of the body which carry genetic information or ‘genes’. Genes contain instructions which tell the body how to develop and function.

Each cell has 46 chromosomes in 23 pairs; 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 further pair of chromosomes called the sex chromosomes; X and Y. Girls have two Xs, whereas boys have an X and a Y chromosome.

Changes in the number or structure of chromosomes increases the risk of physical and intellectual disability.

The routine chromosome test offered during pregnancy is called a karyotype. This uses a microscope to look at and detect changes in chromosomes. This test is not able to detect smaller subtle chromosome changes, because these cannot be seen through the microscope.

Small chromosome changes can still disrupt growth and development. These small changes are often called microdeletions (when a tiny piece of chromosome is missing) and microduplications (when a tiny piece of chromosome is doubled up).
What are the advantages of array CGH?

The main advantage of array CGH is that it allows us to explore all 46 chromosomes in detail. This means we can detect extra or missing chromosomes and other chromosome changes much more precisely than with the routine chromosome tests.

An imbalance in the chromosomes may explain your ultrasound findings and allow us to give you more precise information about the diagnosis. It may show whether the condition is hereditary (can be passed on from parent to child) and can give information about the chance of the same condition affecting a future pregnancy.

What are the disadvantages of array CGH?

Array CGH will not detect all chromosome imbalances, as some are too small to be identified. Some changes cannot be picked up by our current test.

Genetic conditions are not only caused by chromosome imbalances, but may also be caused by changes in individual genes. Array CGH cannot detect the tiny changes in individual genes.

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

Array CGH may detect chromosome changes called ‘variants of unknown significance’. There is not yet enough information in medical literature about these, so it can be difficult to be certain about how or if these chromosome changes are linked to the ultrasound findings.

Rarely, an unexpected chromosome change can be identified which is unrelated to the ultrasound findings, but may have implications for the future health of your baby or the health of other family members. Please let the geneticist know whether you wish to know about these changes, or only wish to know about the results relevant to the ultrasound findings.
How will we be given the results?

The results will be available in about two weeks.

If no imbalances are detected in your baby’s chromosomes, the Specialist Midwife will contact you with the result.

If there are any chromosome imbalances identified you will be offered an appointment with the Clinical Geneticist, to discuss the result.

Further questions

If you have more questions about the array CGH test, please contact the Specialist Midwives in Prenatal Diagnosis.

Tel: 01865 221 716

If you have a specific requirement, need an interpreter, a document in Easy Read, another language, large print, Braille or audio version, please call 01865 221 473 or email PALS@ouh.nhs.uk

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