

What are the benefits of array CGH?

- It may help you and your doctor to monitor and watch for common health problems associated with your child's chromosome imbalance
- It may help to predict what to expect as your child gets older
- It may show which specific genes are included in your child's deletion or duplication. If the gene(s) has been associated with a particular feature or health problem it may help to guide management or treatment for your child
- It can help you to obtain specialist services for your child
- You can choose to join a support group to meet other parents facing similar challenges
- Parents and other family members can be tested to see if they are carriers of changes in their DNA that put them at risk of having more children with related chromosome changes

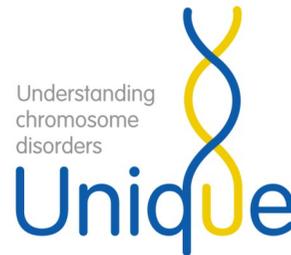
What are the limitations and problems of array CGH testing?

- Some chromosome or DNA changes cannot be detected by array CGH (for example very tiny changes to the DNA or rearrangements that do not result in any loss or gain of DNA material)
- Chromosome changes detected by array CGH are known as **copy number variants (CNVs)**. A CNV may be the cause of your child's problems; however, some CNVs are common in the general population and are completely harmless, and some may not have been seen before, but may nevertheless be unrelated to your child's problems. Parents may need to be tested in order to help interpret the results. The geneticist also will take a detailed family history, examine your child and review the medical information that is available to help to explain if the CNV is likely to be relevant
- Sometimes a CNV can be unrelated to your child's problems at the time of testing, but may have implications for his/her future health or development, or the health of other family members. Please discuss this with your genetics specialist
- Further information may become available about the significance of some CNVs as more children are tested by array CGH. Please contact your genetics specialist for advice on new developments

Families say ...

“ Our geneticist used an analogy which made things clearer for us. He said that previous test results were like an old-fashioned map of the world which showed just a wide overview (country level) and that doing an array is more like using Google earth which allows us to zoom in much more closely, even down to street level, to give a closer and clearer idea of which genes, if any, are missing or duplicated.

“ Without array CGH we wouldn't know what my son had. Now that we know, it has made us look forward and get on with our lives.



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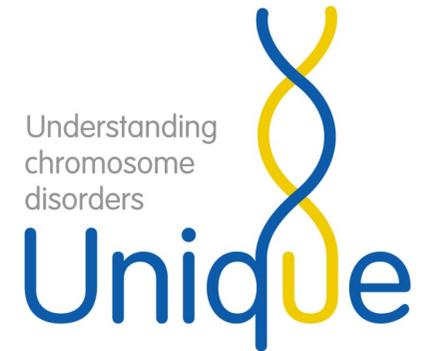
This leaflet is not a substitute for personal medical advice. Families should consult a medically qualified clinician in all matters relating to genetic diagnosis, management and health. The information is believed to be the best available at the time of publication. It was compiled by Unique and reviewed by Dr Shehla Mohammed and Professor Caroline Ogilvie, Guy's Hospital, London. UK and Professor Maj Hultén Professor of Reproductive Genetics, University of Warwick, UK and Chief Medical Advisor to Unique.
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Microarray-based comparative genomic hybridisation (array CGH)

rarechromo.org

What are chromosomes?

Chromosomes are the structures in each of the body's cells which carry the genetic information (DNA) that tells the body how to develop and function. They come in pairs, one from each parent, and are numbered 1 to 22 approximately from largest to smallest. Each person has another pair of chromosomes, called the sex chromosomes. Girls have two Xs (XX), whereas boys have an X and a Y chromosome (XY). Each chromosome has a short (p) arm and a long (q) arm.

Looking at chromosomes (chromosome analysis)

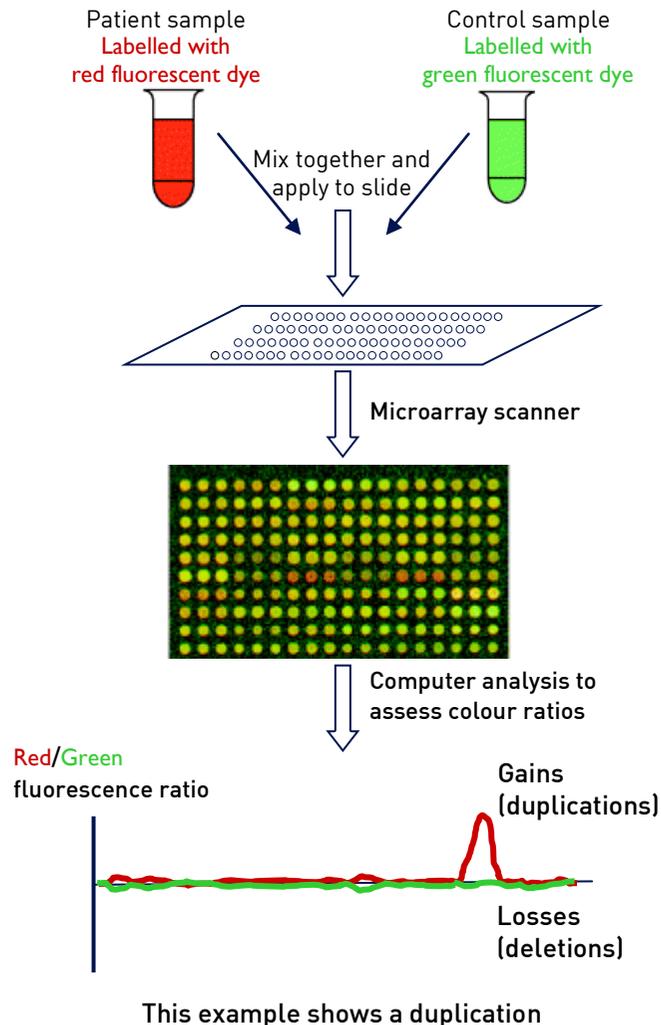
Chromosomes cannot be seen with the naked eye, but if you stain them and magnify them many hundreds of times under a microscope, you can see that each one has a distinctive pattern of light and dark bands. By looking at your child's chromosomes in this way, often referred to as karyotyping, it is possible, if the change is large enough, for a trained scientist to see if there is a chromosome imbalance (loss or gain of chromosome material) or if the chromosome is rearranged in any way. However, because the amount of material gained (duplicated) or lost (deleted) may be too small to see on a routine chromosome test, you may have been told that your child's chromosome analysis was normal. A new, more enhanced, test now available for looking at chromosomes is called a [microarray-based comparative genomic hybridisation](#) (or [array CGH](#), [aCGH](#)) test.

What is array CGH?

Array CGH is a significant advance in technology that allows detection of chromosome imbalances that are smaller than can be detected by the human eye through looking down the microscope. Karyotyping is only as good as the resolution of the microscope and is not able to detect subtle chromosome changes even by the most trained and experienced scientist. These smaller alterations are often called "submicroscopic" changes because they cannot be seen through the microscope. However, they can still disrupt the pattern of development. These very small changes are often called [microdeletions](#) and [microduplications](#). Array CGH is also sometimes called CGH array or simply microarray.

Array CGH compares your child's DNA with a control DNA sample and identifies differences between the two sets of DNA. In this way, deletions or duplications (imbalances) in your child's DNA can be identified. From this, the gene content of any such imbalance can be established.

How does array CGH work? The patient and control DNA are labelled with different coloured fluorescent dyes which act like coloured flags (e.g. red and green below), and applied to an array (a solid surface, usually a microscope slide, on to which tens of thousands of short stretches of DNA are spotted). The samples bind to the DNA on the slide. Where there is no change between the patient and control sample there will be equal binding and therefore equal amounts of red and green fluorescence. For regions where there is a duplication in the patient sample there will be more red fluorescence than green; conversely deletions will result in reduced red fluorescence and more green fluorescence.



What samples are needed for array CGH testing?

DNA for array CGH can be extracted from a blood or saliva sample from an adult or child. It can also be performed on DNA from prenatal samples, usually amniotic fluid or chorionic villi (placental tissue) or CVS from a pregnancy with ultrasound features suggestive of chromosome imbalance.

Why has array CGH been offered for our child?

Your doctor or geneticist may consider array CGH testing if your child has problems with learning, physical development, behaviour, autism or birth defects or medical concerns such as seizures. Recent studies have shown that around 20 per cent of children with unexplained learning and/or developmental disability will have chromosome changes that could not be detected by conventional chromosome analysis but can be detected through array CGH.

How will we be given the results?

The results are likely to be given to you by your geneticist or paediatrician who will talk you through your child's results. You will almost certainly then receive a follow-up letter. Alternatively, you may receive a preliminary result from the doctor doing the test and then be referred to geneticist for a more detailed explanation (if appropriate).

How long do the results take?

Results are usually available in 6-8 weeks. Testing a newborn baby with multiple problems is considered a priority and therefore results may be available slightly sooner. For prenatal testing, check with your local genetics centre.

What are the advantages of array CGH?

- All 46 chromosomes can be examined in a single test
- More sensitive and accurate than conventional karyotyping
- A diagnosis from array CGH may avoid your child having to undergo many other tests
- It can reveal which specific genes are included in the deletion or duplication
- It can be useful to further define breakpoints in imbalances that are already known