Information for Patients

A Microarray Test
Also called a Detailed Chromosome Test or Array

A microarray is a special genetic test that looks in detail at a person's chromosomes to see if there are any extra or missing sections which might account for problems they have been experiencing.

What are chromosomes?

Our genetic information (DNA) includes around 20,000 individual instructions (called genes). Several hundred genes are packaged together into structures called chromosomes. We all have 23 pairs of chromosomes. They are numbered by size from 1 to 22, with pair number 1 being the biggest and pair number 22 the smallest. The final pair of chromosomes show the genetic sex of the person. People with one X and one Y chromosome are genetically male, whilst those with two X chromosomes are genetically female. We get one of each pair of chromosomes from our mother and one from our father. We pass on only one of each pair to our children.

When is an array test offered?

A microarray can be a useful test to try and find the cause of unexplained problems with learning, physical development or behaviour. It may also be used to help find a diagnosis for people with unexplained birth defects or medical problems, such as seizures.

What sample is needed?

To carry out a microarray, a small blood sample is usually taken for testing. If taking blood is very difficult, other samples, such as saliva, may occasionally be used.

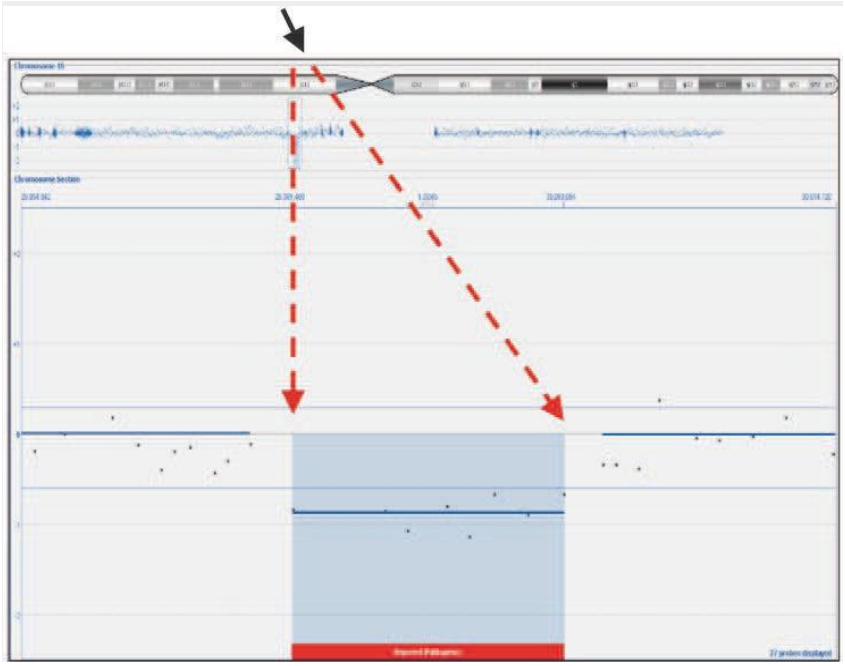
What does the test look for?

A microarray looks at thousands of points along each of a person's chromosomes in detail and compares them to a control sample. Scientists can then detect small missing or extra chromosome sections that cannot be seen down the microscope.

Where the amount of chromosome material is normal, the scientists can see a straight line on a computer read-out. If the patient has too much of a section of chromosome (a duplication), that section of the line is higher than usual and where there is too little chromosome material (a deletion), it is lower than usual. Sometimes the laboratory carries out extra tests to check the size or exact position of a duplication or deletion.

A picture of a genetic change found on microarray:

Missing chromosome section (deletion)



Why does it matter if there is a chromosome deletion or duplication?

Depending upon its size and position, a deletion or duplication can affect the number of copies of many important genes. Usually, if someone has a deletion they have only one copy, of all the genes within that section of chromosome, instead of the usual two copies. If someone has a duplication, they have three copies, of all the genes within that section of chromosome, instead of the usual two copies.

Deletions or duplications on the sex chromosomes (X and Y) can affect males and females differently. This is because males already only have one copy of each of the genes on the X chromosome, whilst females have two.

How many genes and which genes are involved in a deletion or duplication will change the symptoms that a person might experience. Other genetic and environmental factors can also affect how a person is affected.

How long will the test take?

Results of a microarray test will usually be back with your doctor or genetic counsellor in 6-8 weeks, but sometimes it can take longer than this to interpret the results.

Will the test result explain my child's difficulties?

The microarray test may or may not find the cause of your child's difficulties. Listed below are the types of result you may receive:

1. A normal result

If the test does not find any relevant missing or extra sections of chromosome, your child will receive a **normal result**. This does not mean that your child's problems are not genetic, as the test cannot detect every possible genetic cause. For example, sometimes the cause might be a tiny spelling mistake within a single genetic instruction (gene) which would not be picked up on the microarray test.

Following a normal microarray result, your doctor will consider other possible causes for your child's difficulties and may suggest further tests. This could mean a fresh sample of your child's blood is needed.

2. The cause or partial cause is found

In around one quarter of cases this test will find there is some extra (duplicated) or missing (deleted) chromosome material. If the chromosome deletion or duplication has been seen before, the scientists and/or your doctor will look to see whether it has been linked to similar problems to those your child has. They will then decide whether it is likely to be the **whole, or at least part of the reason for their difficulties**.

Some chromosome deletions and duplications are quite common, but do not always cause health problems. If one of these is thought to be the cause of some difficulties your child has experienced, other family members may also carry it but not have the same difficulties.

The microarray may find a very rare deletion or duplication, or one which hasn't been seen before. If this is the case, the scientists and/or your doctor will look carefully at what we know about all the genes within the duplicated or deleted section to see whether having extra or missing copies of those particular genes would be expected to cause the difficulties your child has experienced.

3. An uncertain result

Sometimes it is not possible to be certain whether a deletion or duplication is the cause of a child's difficulties or not. Further tests may be offered to try and clarify things, but this is not always possible. This type of change is called a **variant of unknown clinical significance**. With further information, and over time, we may discover more about these changes and be able to give a firmer answer.

4. An unexpected Result

Occasionally, an array finds a chromosome deletion or duplication, which does not directly link to a child's current difficulties but could have other health implications now or in the future. If this happens your doctor or genetic counsellor will explain this to you.

Who will give me the results?

The results will initially be explained by the doctor or genetic counsellor who arranged the test. Where possible, they will provide you with information about the deletion or duplication that has been found. They may suggest some extra blood tests for your child and/or both parents to help them interpret the results fully. For some results, they may also suggest extra health checks as a precaution.

Is an appointment in the Genetic Clinic necessary?

It is not always necessary to be referred to the Genetics Clinic for this test or to discuss the results, but it may be helpful if you have further questions or concerns about your child's result. If the microarray test has not found a cause for your child's difficulties, their doctor may refer them to the Genetics Clinic to decide whether further genetic tests might be helpful.

Parental Testing

If the laboratory needs extra information to help interpret your child's result, both parents may be asked to also give a blood sample. Even if this is not needed, parents can usually still choose to be tested for a deletion or duplication found in their child (although in some rare cases,

this is not possible for technical reasons). Your doctor or genetic counsellor can discuss this with you in the Genetics clinic.

When parents are tested, they do not usually have an array test to look in detail at all their chromosomes. Instead, they are usually offered a very specific test to just look for the particular deletion or duplication that has been found in their child.

What happens to the sample after testing?

It is routine practice for a Genetics laboratory to keep storing the DNA taken from a blood sample even after the result is reported. This is because we are constantly learning more about genetics and although the sample will not be tested for anything new without consent, it would be available for testing in the future, if needed. If you want further information about this, or you do not wish for the sample to be stored in this way, you should speak to the doctor or genetic counsellor who is ordering the test.

For more information

If you need more advice about any aspect of array tests, you are welcome to contact us at the **Manchester Centre for Genomic Medicine:**
Sixth Floor
Saint Mary's Hospital
Oxford Road
Manchester M13 9WL

Telephone: (0161) 276 6506 (Reception)
Facsimile: (0161) 276 6145
Website: www.mangen.co.uk

Department staffed Monday–Friday, 9.00 am to 5.00 pm.

Alternatively, the Charity Unique www.rarechromo.org/ provide excellent information and support for families affected by chromosome differences.