

Introduction

The doctor looking after you, or your child, has decided to request a test called a microarray. This leaflet aims to explain what this test involves and the possible outcomes.

What are chromosomes and genes?

Each person has about 20,000 pairs of genes. They work like instructions that determine things such as the colour of our eyes, how many fingers and toes we have and how tall or short we are. Genes are so small that they cannot be seen, even under a powerful microscope. They are arranged along strands, rather like beads along a string. These strands are called chromosomes, and are inside most of the cells in our body.

Most cells in a person's body contain 23 pairs of chromosomes (46 in total), which can be seen down the microscope. To help tell which chromosome is which, each pair has been given a number. Pair number 1 is the largest; pair number 2 is the next largest and so on right down to pair number 22 which is the smallest. The remaining two chromosomes (X and Y) are called the sex chromosomes and they determine whether a person is male (XY) or female (XX).

Everyone has some small variations on their chromosomes, but not all of these cause problems. However, some people who have certain changes in the number or structure of their chromosomes may have a variety of problems, including developmental delay, learning and behavioural problems, heart defects, abnormalities of the fingers or toes and many others. Importantly, everyone has some small changes on their chromosomes, not all of them cause problems.

Chromosome tests

Karyotype – Chromosomes cannot be seen by the naked eye but can be seen under powerful microscopes. A "normal" result means that there are no obvious large pieces of chromosome material missing or extra and the chromosomes are arranged in the right order.

Microarray – This test uses techniques to look at the chromosomes in much finer detail, and can pick much smaller pieces of missing (deletion) or extra (duplication) chromosome than can be seen down a microscope.

What happens now?

A blood sample will be needed from your child and sent to our laboratory for microarray. Results will take approximately 12 weeks and the doctor will contact you once the results are received.

If the laboratory has a sample of DNA stored from a previous genetic test that your child has had, it may be possible to test this DNA sample. However a new blood sample may be required.



Results

There are three possible outcomes from the microarray.

- 1. The chromosomes may appear entirely normal. A normal microarray test does not rule out all genetic conditions as there may still be changes in individual genes, which cannot be detected by a microarray test.
- 2. The test may reveal a change which is highly likely to account for the problems that your child has. If we identify such a change then we can see you in clinic again to discuss this.
- 3. We could identify a change that we do not know the significance of in other words, we do not know if it may be the cause of your child's problems or not. In this case, we may ask for samples from both parents to see whether either of you have the same change as your child has in their DNA. This may help us to interpret the meaning of the genetic change, but some changes will remain uncertain.

There is also a very small chance that we could pick up something unrelated to your child's current problems which may have implications for their/the family's future health e.g. early onset cancer or heart disease. We would inform you of this result.

If you need more advice about any aspect of Microarray, you are welcome to contact:

Liverpool Centre for Genomic Medicine Liverpool Women's Hospital NHS Foundation Trust Crown Street Liverpool L8 7SS Telephone: 0151 802 5001 or 5008 Email: <u>lwft.clir</u>

Email: lwft.clingen@nhs.net

This leaflet can be made available in different formats on request. If you would like to make any suggestions or comments about the content of this leaflet, then please contact the Patient Experience Team on 0151 702 4353 or by email at pals@lwh.nhs.uk

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