Bristol Genetics Laboratory

North Bristol NHS Trust

Array Comparative Genomic Hybridisation (CGH)

Exceptional healthcare, personally delivered

What is array CGH?

Array CGH is a new test that is now offered to all patients referred with learning disability and to some patients with other physical problems. Using array CGH, the laboratory is able to look at thousands of separate sections of your child's DNA (which contains the genes). This shows whether sections of DNA are either missing or present in extra copies. Missing pieces (loss) or extra copies (gain) of genetic material may lead to clinical problems such as learning disability and some physical problems.

The main benefit of array CGH is that it is able to detect much smaller genetic changes than was previously possible. Array CGH can also provide very accurate information on the size and the possible effects of the genetic changes found.

Array CGH testing on your child will only be done with your consent.

What are genes?

Genes are the blueprint of the body. When one or more genes are lost or gained the blueprint is changed. This can lead to clinical problems from birth such as learning difficulties and some physical problems.

Genes are made of a chemical called DNA and contained inside larger structures called chromosomes. Most people have 23 pairs of chromosomes (46 in total). One of each pair comes from the mother and the other from the father. Chromosomes are numbered 1 (the largest pair) to 22 (the smallest pair). The 23rd pair (X and Y) are called sex chromosomes because they determine whether a person is male or female. Each chromosome has a "q" arm (which is long) and a "p" arm (which is short). The genes are arranged in groups clusters along these arms (like beads on a string).

What happens when my child's blood is sent for array CGH testing?

Two tubes from the same blood test are sent to the laboratory. DNA is extracted from one tube and the laboratory stores cells from the other. This is in case more tests are needed after the array CGH result is known.

The array test is technically quite complex and in a small number of cases may simply fail to work, in which case a repeat sample may be asked for.

The array CGH test normally takes four to six weeks to report. In three quarters of cases the laboratory will not find a problem and will issue a normal report to your doctor. Your doctor may then want to begin other tests.

What does a normal result mean?

A normal result means that the laboratory has not found a problem with your child's DNA. Having a 'normal' result will not always rule out a particular problem.

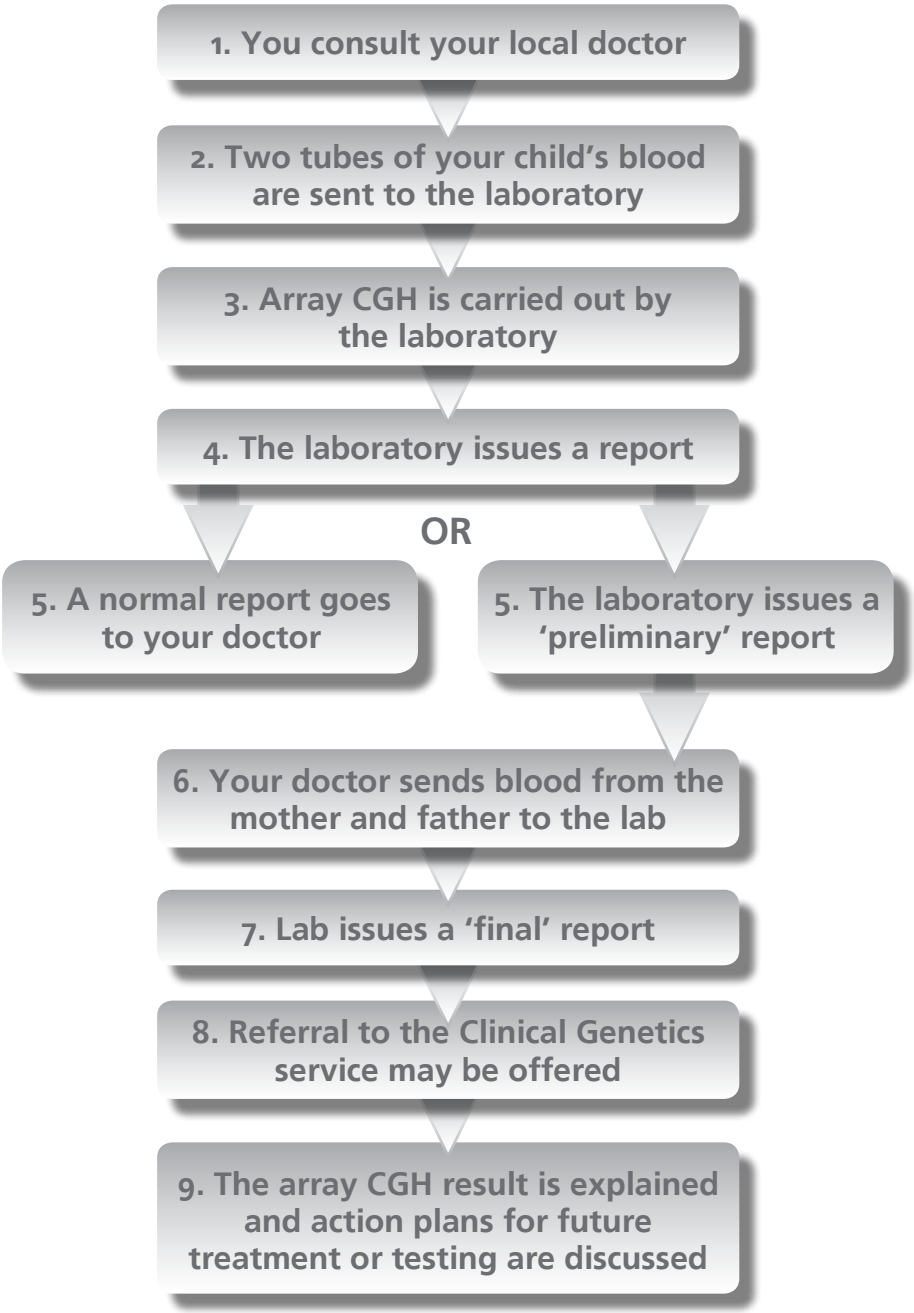
A normal array CGH result also provides useful information. After a normal result your doctor may begin further tests.

What happens if array CGH finds something?

In around one quarter of cases, array CGH will reveal missing or extra genetic material.

Many of the changes found by array CGH have a well known pattern of medical problems. Your doctor will discuss the results with you to help you to fully understand them. You may also be offered a consultation at your local Clinical Genetics service.

Some array CGH results are very rare or may never have been seen before. In these cases, it may not be possible to work out if a particular array CGH result will cause a problem, or what medical problems may be expected.


1. You consult your local doctor
2. Two tubes of your child's blood are sent to the laboratory
3. Array CGH is carried out by the laboratory
4. The laboratory issues a report
OR
5. A normal report goes to your doctor
5. The laboratory issues a 'preliminary' report
6. Your doctor sends blood from the mother and father to the lab
7. Lab issues a 'final' report
8. Referral to the Clinical Genetics service may be offered
9. The array CGH result is explained and action plans for future treatment or testing are discussed

At this stage the laboratory will issue a "Preliminary Report" to your doctor. This report will give details of the genetic change and will ask for samples from both parents. Testing both mother and father to look for the same genetic change can sometimes help understand this type of uncertain result.

Testing parents

If an uncertain result is found, your doctor will ask both parents to give a blood sample. This will be sent to the laboratory for further testing to help them to understand your child's results.

It is very important for the laboratory to receive blood samples from both parents. If you know that one parent will not be available for testing, please tell your doctor as soon as possible.

The mother's and father's samples will be tested to see if either parent is carrying the genetic change seen in your child. The blood tests on the parents will only be looking for the change seen in their child.

If the genetic change seen in your child is not found in the mother or father then it is likely that this change is the cause of your child's clinical problems.

Final Report

When the parental tests have been completed, the laboratory will issue a "Final Report". This is based upon what has been found in your child's array CGH and the results of parental testing. This will be discussed with you to help you to fully understand the results.

It is sometimes found that your child's genetic change is normal human variation and is not the cause of your child's clinical problems.

Once the final report has been issued you may be offered a consultation at your local Clinical Genetics service.

Unexpected findings

In very rare cases, the array CGH test may find a genetic change which may be clinically important but may not be directly related to your child's current problems. In the very unlikely event that an unexpected result is found, this will be fully explained during a consultation with your local Clinical Genetics service.

What happens to the sample after testing?

The blood samples and stored cells are routinely discarded after testing is completed. DNA is very stable and is stored indefinitely by the laboratory (or until it runs out).

Genetics is a rapidly developing field. It is possible to test DNA for a number of genetic conditions, with more tests becoming available each year. Storing DNA lets us to carry out any relevant tests in the future without taking a further sample.

However, if requested, we will make sure that the DNA is discarded after testing.

References and further information

The Bristol Genetics Laboratory www.nbt.nhs.uk [Last Accessed March 2012]

The rare chromosome disorders charity Unique www.rarechromo.org [Last Accessed March 2012]. Unique offers support and information to all families affected by rare chromosome disorders

Microarray-based comparative genomic hybridisation (array CGH) information leaflet produced by Unique (http://www.rarechromo.org/forum/DisordersLeaflets.asp)

Miller D et al (2010). Consensus statement: chromosomal microarray is a first-tier clinical diagnostic test for individuals with developmental disabilities or congenital anomalies. Am J Hum Genet 2010;14;86(5):749-64

NHS Constitution. Information on your rights and responsibilities. Available at **www.nhs.uk/aboutnhs/constitution**

How to contact us:

See your appointment letter for the number to phone with any queries you may have

www.nbt.nhs.uk/genetics

If you or the individual you are caring for need support reading this leaflet please ask a member of staff for advice.