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If you are unhappy with the advice you have been given by your GP, consultant, or another healthcare professional, you may ask for a second (or further) opinion.

The evidence used in the preparation of this leaflet is available on request. Please email: [patient.information@salisbury.nhs.uk](mailto:patient.information@salisbury.nhs.uk) if you would like a reference list.

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# **What is Array Comparative Genomic Hybridization (aCGH)?**

## **A guide for parents and carers**



### **The Wessex Regional Genetics Laboratory**

In the Wessex Regional Genetics Laboratory we test for genetic (inheritable) disorders. If we are told by your consultant which gene to look at we can look at specific genes that are known to cause a genetic condition in great detail (such as cystic fibrosis). However, often the specific genes that are causing the condition are not known and therefore we examine the whole of the genome (the DNA make-up of an individual) but in much less detail, looking for much larger gains or losses of whole genes rather than a change within a gene. In the past this has been carried out by analysing whole chromosomes down the microscope (known as conventional karyotyping). However, recent advances now make it possible to examine the entire genome in much greater detail. We can therefore detect all the chromosome imbalances that we could previously see down the microscope and also many smaller changes so that we can confirm a diagnosis that your consultant suspects or identify a genetic explanation for a congenital problem.

### **What is array-CGH (aCGH)?**

aCGH is a test that for many patients has now replaced an older chromosome test. The main aim of the aCGH test is to detect small genetic change. aCGH not only does this but can also give very accurate information

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on the size and possible consequences of the genetic changes found. Using aCGH, the laboratory is able to look at thousands of separate sections of your child's DNA (which contains the genes). All of these points are analysed with special computer software to see if sections of your child's DNA are either missing or present in extra copies. Both missing bits and extra copies of our genetic material may lead to clinical problems.

## **What happens when my child's blood is sent for aCGH testing?**

Blood is sent to the laboratory and DNA is extracted. It takes up to 4 weeks to get the results of the aCGH test. In three quarters of cases, the laboratory will not find a problem and will report this to your consultant. Your consultant may then want to look for other reasons for your child's health issues.

In approximately one quarter of cases, the aCGH test will reveal missing or extra genetic material and the laboratory will issue a report/preliminary report to your consultant. This report will give details of the genetic changes and in some cases will recommend further testing to help understand what has been found.

At this stage your doctor will ask both parents to give blood which will be sent to the laboratory for further testing. It is very important for the laboratory to receive blood samples from **both biological parents whenever possible** to allow them to fully interpret your child's genetic change. We understand that this is not always possible and if for any reason, you know that one parent will not be available for testing, please ask your consultant to let the laboratory know this as soon as possible. A further blood sample from your child may also be requested at this time if a different test is required for following up the child's imbalance in the parents.

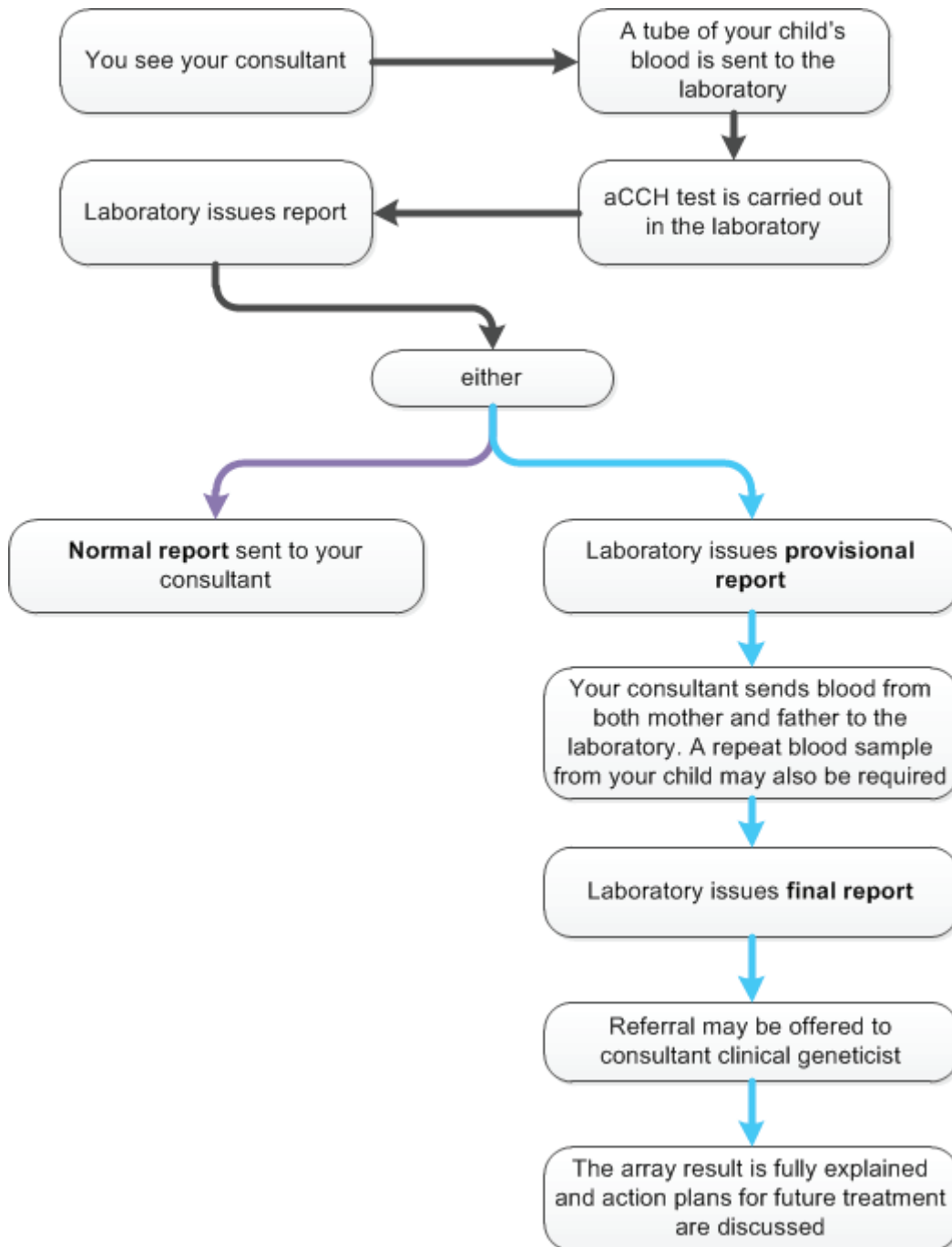
As soon as we have the blood sample from the parents (and your child, if requested) the next stage of testing begins. This involves examining both the mother's and father's DNA to see if either parent is carrying the same genetic change seen in your child. The blood tests on the parents will

only be looking for the change that was seen in your child.

If neither parent has the same genetic change as found in their child it is possible that the aCGH finding is the cause of your child's clinical problem. As mentioned above, aCGH is a way for the laboratory to look at the entire genetic make-up of a person in order to find missing or extra parts of the DNA. Many of the changes found by aCGH are seen fairly often and we know the sort of medical problems they can cause. However, some aCGH results are rare and may never have been seen before. In some cases, it is not possible to work out if a change that we have found will cause a problem or not, or what medical problems may be expected. Taking blood samples from the parents can sometimes help to clarify this. Sometimes the changes we find are within a normal range and do not indicate a problem.

When all the follow-up tests have been completed on the parent's blood, the laboratory will issue a final report. This will have the conclusions based on what has been found in your child's aCGH.

## An aCGH patient journey



Once the final report is issued you may be offered the opportunity to talk to a consultant clinical geneticist who will go over all the information with you and help you to understand what the results mean.

In our experience, aCGH will help your consultant to make a diagnosis

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in 1 in 6 cases. This is a major improvement compared to the diagnosis rate using standard chromosome testing, however we understand how frustrating this may be for those people for whom it does not give a diagnosis.

In very rare circumstances, the aCGH result may find a change in the DNA, which may be of clinical significance, but may not be directly related to the reason why your child's blood was sent for aCGH testing in the first place. Your consultant clinical geneticist will explain and discuss this with you in the very unlikely event that an unexpected result is found.

## Frequently asked questions

**Question:** How often will aCGH find something wrong with my child's genetic make up that will help to explain his/her clinical problems?

**Answer:** Many laboratories throughout the world including the Wessex Regional Genetics Laboratory (WRGL) here in Salisbury, are finding genetic causes for clinical problems using aCGH in around 15% (around 1 in 6) of all the patients referred for genetic testing. This is at least three times higher than the detection rate we were previously able to achieve.

**Question:** How often and for what reason does the laboratory also examine the parents' blood samples?

**Answer:** In approximately one quarter of cases, the aCGH test will show that your child has DNA (genetic material) either missing or in extra copies. Examining the parent's blood gives us essential information which helps us to understand the aCGH result. In some cases, the aCGH test result may show normal human variation and not therefore related to your child's clinical problems.

**Question:** Why is it so important for the parents to discuss the aCGH results not only with their consultant but also with a consultant clinical geneticist?

**Answer:** aCGH is a relatively new technology but is increasingly being used worldwide. The WRGL works closely with both the Wessex Clinical Genetics Service in Southampton (contact details at the end of the

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leaflet), and scientists across the world so that the very latest and most informed interpretation of your child's aCGH result can be given to you. The amount and quality of information is growing rapidly and so the consultant clinical geneticists are the best people to help (a) explain your child's aCGH result and (b) discuss what the results mean, not only for your child but potentially for you and other members of your family.

**Question:** How long will it take between taking blood from my child and our having the final aCGH result?

**Answer:** If the result is normal, it will be back with your consultant within 4 weeks. If we ask for parent's blood samples, the full result will usually be back with your consultant and the consultant clinical geneticist a month after the laboratory receives the parent's samples.

**Question:** is there a chance that aCGH might find missing or extra copies of genetic material in the parents' DNA and if so what could this mean?

**Answer:** aCGH may detect DNA that is either missing or present in extra copies in one of the parents. There are a number of possible explanations for this and your consultant clinical geneticist will discuss these fully with you.

**Question:** What happens next?

**Answer:** The 'Patient Journey' diagram on page 4 sums up what happens from the time the blood sample is taken to when you will know whether or not aCGH has helped in the diagnosis of your child's clinical problems. It is important to realise that a normal aCGH result also gives useful information for your consultant who may then consider other reasons for your child's health issues.



## Contact details and further information

Wessex Regional Genetics Laboratory

[www.wrql.org.uk](http://www.wrql.org.uk)

Wessex Clinical Genetics Service

Wessex Clinical Genetics Service

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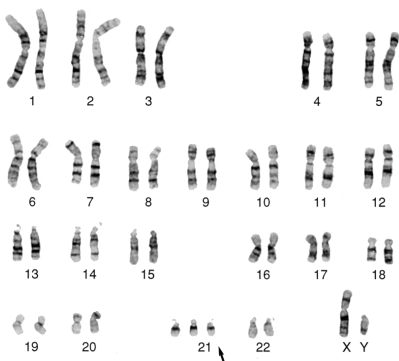
023 8120 6170

[www.wcgs.nhs.uk](http://www.wcgs.nhs.uk)

## UNIQUE (Rare Chromosome Disorder Support Group)

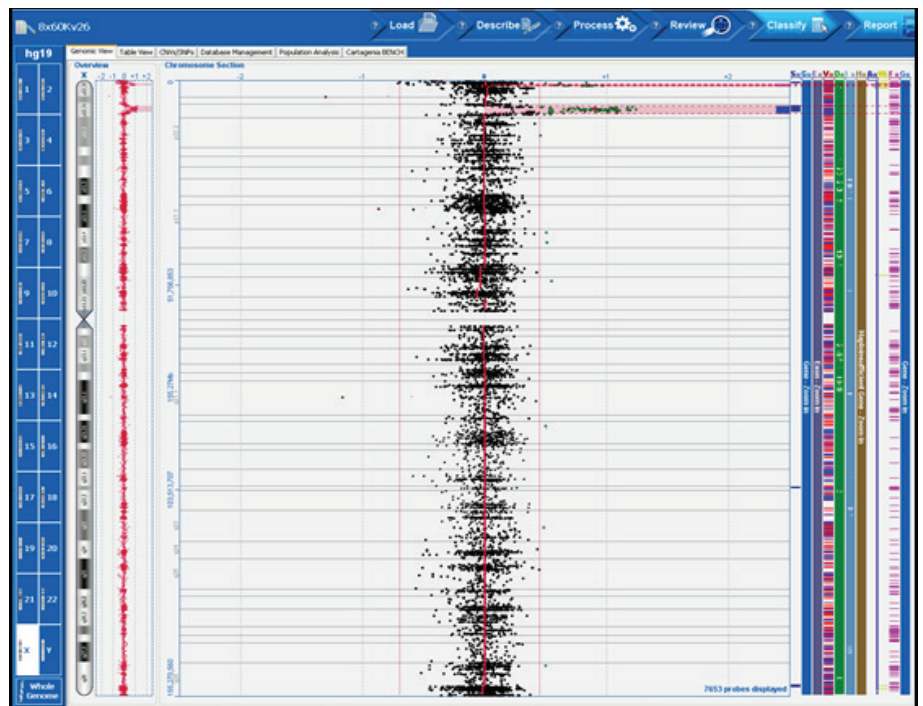
An organisation that offers support and information to all families affected by rare chromosome disorders.

[www.rarechromo.org](http://www.rarechromo.org)



Conventional karyotype test result

## aCGH result



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