



Information for Parents about Microarray Analysis

Your doctor has requested a chromosomal microarray analysis for you / your child. This test is used to help identify a genetic cause for physical, behavioural or intellectual differences in children or adults.

Microarray analysis is a very detailed test looking for small changes in the amount of genetic information, which may be missed by a standard chromosome test, using a microscope.

The microarray result will be one of the following:

- **No clinically significant change i.e. a normal result**
No chromosome change has been detected. This result does not exclude all genetic conditions
- **Pathogenic change i.e. abnormal result**
A chromosome change has been detected which causes physical, behavioural or learning differences. This result may explain you /your child's physical, behavioural or intellectual differences.

Very rarely a chromosomal abnormality is found that has important health implications but is not related to the reason for referral e.g. the result may be relevant to genetic conditions that have an onset in later life or those that are associated with an increased risk of cancer (*Incidental finding*).

- **Change of uncertain/unknown significance**
A chromosome change has been detected, but there is limited information available about the effects of the chromosome change and therefore it is difficult to know whether the change is the cause you/ your child's differences or whether it may be associated with other problems.

A change may be associated with variability in the problems it can cause i.e. different people may be affected more or less severely, and it may not be possible to provide the exact risk of a change causing problems or how significant the problems will be.

In some cases samples from each parent may also be analysed to help to assess the importance of the change within the family. Interpretation of a finding may be more difficult if samples from both parents are not available.

Some additional possible outcomes of testing are:

- A change may be detected in a child, which may subsequently be found in either parent
- The test may reveal information about biological relationships
- The test may show that the child (and / or parents) is a carrier of a recessive genetic disorder. In this situation, genetic counselling will be required to ensure the family understand the implications of the results

Due to the rarity of some of the chromosome differences the laboratory participates in two international groups, which provide a resource about these various chromosome differences and the clinical features of individuals with those differences. The databases collect information about the microarray result, and the physical, intellectual or behavioural characteristics of the person with the abnormality. This information is submitted without the identifying information about the person. Participation in these databases is important for the assessment of the individual changes, and also for the future analysis of the test.



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Consent is given for:

1. Chromosomal Microarray (CMA)

Sample Type: Blood

Laboratory: Central and Southern Regional Genetic Service

2. Sample Storage

Sample Type: DNA and Fixed Cell Storage

Laboratory: Central and Southern Regional Genetic Service

3. Participation in Microarray Database Repository

Study: Decipher and ISCA

Location: Cambridge UK and USA

The information from this test may be used for other family / whanau (members) to benefit from genetic testing, if you do not wish to do this, please state below:

NOTE: Named test results will not be divulged without further consent.

I understand that this sample and/or any results will not be released to any other third party without my consent (or unless legally required).

I am aware that genetic testing may have insurance implications.

I have read and understood the information given to me and have had the opportunity to ask questions. I understand that I may withdraw or modify this consent at any stage, and that such withdrawal will not affect my further health care.

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| Signed: Patient / parent or guardian / next of kin | Date |
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| Signed: Medical Professional | Date |