

Information for Patients on Microarray Testing

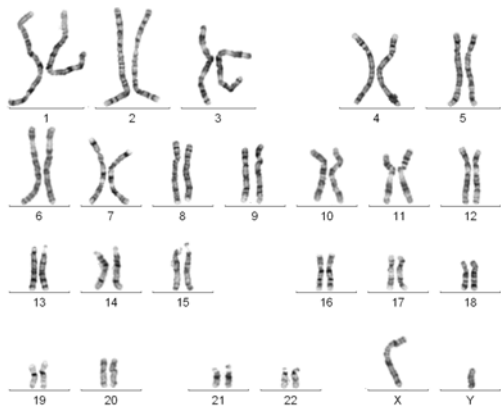
This leaflet provides information on Microarray testing and will help you understand how this test works and what information you will receive from it. Knowing this information might help you better understand the health problem that is affecting you/your child.

Background to Genetic testing

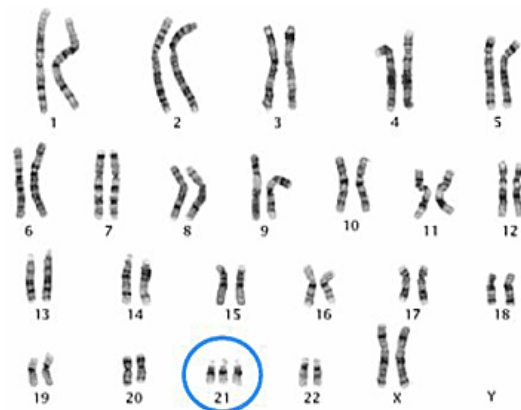
DNA is present in all the cells of our body and is arranged into 23 pairs of chromosomes. They are numbered in pairs from 1 to 22. Additionally, if the person is a male, there will be an X and a Y chromosome. If the person is female, there will be two copies of the X chromosome.

Chromosome testing (karyotyping) can tell if there is a gain (duplication) or loss (deletion) of a whole or part of a chromosome. Individuals with these abnormalities can be at a greater risk of developmental delay, birth defects and intellectual disability.

Chromosome Karyotype



Normal Male Karyotype: 46,XY



Down Syndrome Female Karyotype: 47,XX,+21

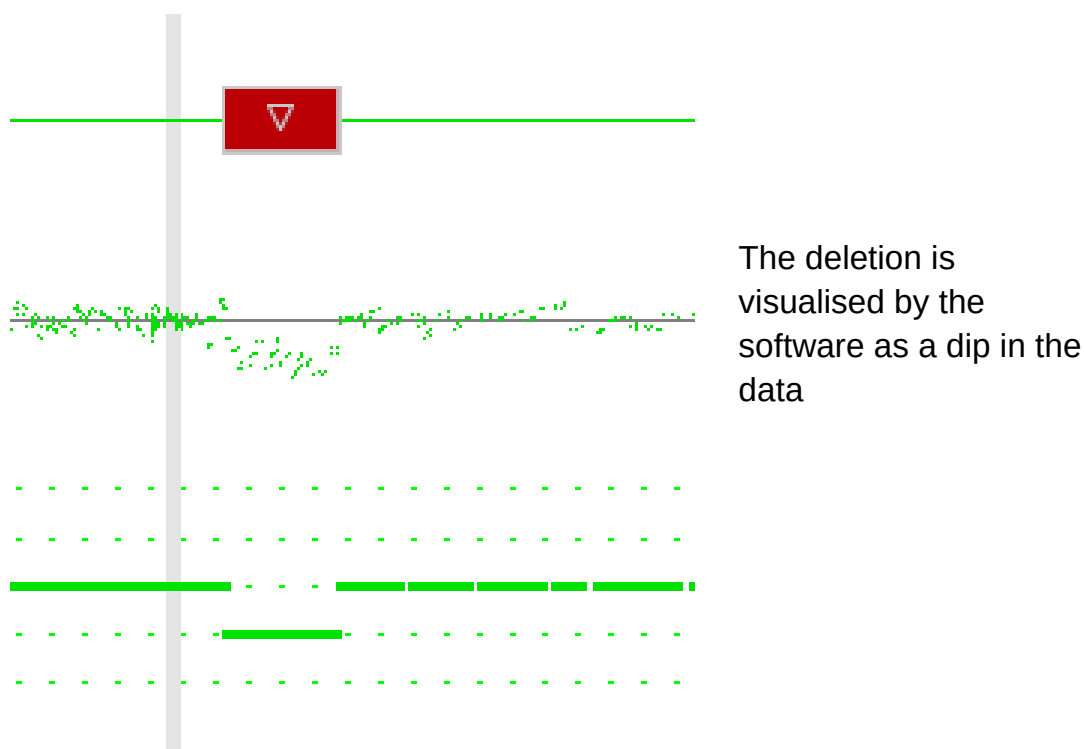
Note: There are 3 copies of chromosome 21 rather than the normal 2 copies.

What is a Molecular Karyotype (Microarray)?

A microarray can detect smaller losses or gains of DNA from chromosomes that may be missed by normal karyotyping. A microarray detects if there are small pieces of DNA missing (deletion) or additional pieces of DNA present (duplication). These changes are called copy number variants (CNVs). The test is not powerful enough to see very small changes within the genes, however it may detect whole/partial gene deletions or duplications.

The microarray is done by testing your/your child's DNA (usually from a blood sample) and will examine each chromosome for losses (deletions) or gains (duplications) of DNA. The image below shows how the microarray software visualises a deletion in a chromosome.

Microarray software Image with a deletion in chromosome 13q13.1



Why am I having this test done?

You or your child may have had a chromosome test done in the past, which gave a normal result. Microarray is a new and more powerful test, which can help to find a genetic cause for physical, behavioural or intellectual differences in children or adults. It is the first line genetic test ordered by a doctor when investigating an individual with unexplained developmental delay or intellectual disability, autism spectrum disorder and multiple birth defects. In about 15% of cases the microarray finds a change that may help explain these types of problems.

What will result from this test?

A microarray takes about 4-8 weeks to complete. The results of your/your child's tests will be given to you by the doctor who arranged the test. In some cases you/your child will be referred to Genetic Health Service NZ (<http://www.genetichealthservice.org.nz/>) where you will see a doctor/genetic counsellor who will help explain the results of the microarray.

The results from a microarray analysis can be divided into four main categories:

1. **Normal** – no copy number variants were detected. Not all genetic conditions are ruled out by a normal microarray analysis. In some cases, referral to the clinical genetics service will be needed to consider other genetic causes for your/your child's problems. This should be discussed with your doctor/your child's paediatrician.
2. **Pathogenic** copy number variant detected. A change that is associated with a previously described genetic condition and will provide an explanation for your/your child's differences. Referral to the clinical genetic service is usually helpful to explain the significance of the change.
3. **Variant of uncertain significance (VOUS)**. A change is detected but the importance of the change is unclear. It may not be possible to know with certainty whether the change is the cause of you/your child's differences. Referral to the clinical genetic service is strongly recommended to help understand the result. In some cases samples from each parent may also be tested to understand the importance of the change within the family.
4. **Variant with unanticipated implications**. Very rarely a change may be found that has important health implications for you or your child but is not linked to the problems you or your child currently have. For example, information may be given about genetic conditions that you/your child may not yet have symptoms of, those that are expected to have an onset in later life, or those that are associated with an increased risk of cancer.

If you have any questions about you or your child having a microarray, or about the results of this test, please discuss them with your doctor or your child's paediatrician. If the doctor arranging the test cannot answer the questions they can contact Genetic Health Service NZ for advice or refer you/your child to be seen in an outpatient clinic.

Contact and Information Details

<http://www.genetichealthservice.org.nz/> (Genetic Health Service NZ-Northern Hub)

Ph: (09) 307 4949 Ext. 25870

Toll Free: 0800 476 123

Email: GenSec@adhb.govt.nz

Note for clinicians: please refer to the LabPlus Test guide for specimen collection requirements and further information regarding diagnostic use and interpretation of Molecular Karyotyping (<http://testguide.adhb.govt.nz/EGuide>).