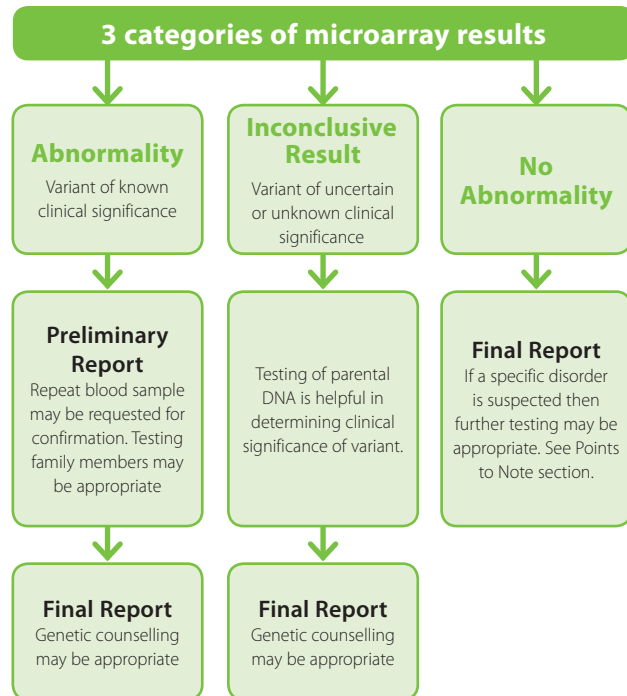


What to consider before testing

- This test has important implications for you and / or your children. Please read the information carefully and discuss any concerns or questions with your Doctor. You may discuss the option of further advice (referral for genetic counselling) if you feel you have further questions or concerns.
- You will need to sign a consent form that your Doctor will witness.

Microarray results and testing of parents

- The results will be classified into 3 categories as shown below.
- Parental blood samples can often clarify findings when the patient results are inconclusive.



Points to note

- NOT ALL genetic abnormalities can be identified by this test. Defects such as small DNA sequence changes or balanced chromosome rearrangements will not be detected.
- Your Doctor may recommend further testing for these other genetic abnormalities if appropriate.

Western Diagnostic Pathology is a member of the International Standard Cytogenomic Array Consortium (ISCA)

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Chromosome Microarray Analysis

A new technique revolutionizing
the field of genetic diagnosis
Information for Patients



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Chromosome Microarray Analysis

What is Chromosome Microarray Analysis (CMA) or Microarray?

- *Chromosome Microarray Analysis (Microarray) is a new technique for looking at an individual's genetic information in great detail. It is an extremely sensitive and sophisticated analysis.*
- *The traditional method for looking at an individual's genetic information is called karyotyping where chromosomes are examined down the microscope.*
- *Chromosomes are contained within the nucleus of our cells. Typically there are 46 chromosomes in each cell.*
- *Our chromosomes carry our genes, the basic units of heredity. Our genes are made up of DNA. There are an estimated 25,000 genes that influence our growth and development. Each gene occupies a specific location on a chromosome.*
- *The genetic information present in our genes varies slightly between individuals. This is what makes us unique. In some cases alterations can lead to the body not developing properly, and this can lead to genetic disease.*
- *The main advantage to microarray compared to karyotyping is that chromosome abnormalities not previously visible can be detected.*

Put Simply:

- *Think of our body like a large house*
- *Our cells like a room, eg the library*
- *The nucleus of our cells like a bookcase*
- *Our chromosomes like a set of 46 books*
- *Our genes like pages of books*
- *Our DNA like the sentences on the page*

Traditional chromosome karyotyping could tell us if there are any missing books or additional books in the set. In some cases it can even tell us if there are additional or missing chapters in a book.

The new Chromosome Microarray Analysis is so sensitive it is able to tell us if there are any extra or missing pages in the books.



What can be detected by microarray?

Microarray detects changes in the amount of DNA along the chromosomes.

It can detect a gain of DNA at a particular site on the chromosome, known as a duplication.

It can detect a loss of DNA, known as a deletion.

The gains (duplication) or losses (deletion) of DNA identified are termed copy number variants (CNVs).

Gains or losses of copies of a gene(s) MAY result in genetic disease or may simply be part of a person's unique genetic information. To clarify further, the laboratory may request samples from the tested individual's parents for comparison.

Our microarrays are targeted to detect more than 100 known genetic disorders caused by duplications or deletions of DNA.

Who should be tested?

Microarray is a useful test for patients suspected of having a genetic condition when previous tests (such as karyotyping and Fragile X testing) have not shown an abnormality.

Your Doctor will advise if this test may be helpful for you/ your child.