Prenatal Testing Chromosomal Microarray

Patient Information

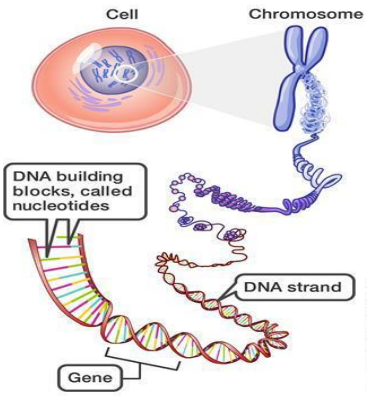
Prenatal Testing

Genetic testing of a baby's chromosomes can be performed during pregnancy (prenatal testing). The results can provide useful information when potential issues are identified through a screening test or an ultrasound scan.

One of these tests is a chromosomal microarray, which looks for abnormalities in the chromosomes.

What are Chromosomes?

Chromosomes are parcels of genetic material (DNA) packaged into genes. Genes contain the instructions for making the building blocks of our body. We have 46 chromosomes in 23 pairs.



Types of Chromosomal Abnormalities

Chromosomal disorders happen when there is:

- An extra copy of a whole chromosome (a trisomy) – For example, trisomy 21 (Down syndrome), trisomy 18 (Edward syndrome).
- Loss of a part of a chromosome (a deletion).
- Doubling of part of a chromosome (a duplication).
- Exchange of part of a chromosome with a part of another chromosome (a translocation).

Chromosomal disorders can result in:

- Physical changes in a baby – For example, heart defects, cleft lip and palate.
- Childhood learning disabilities.
- Medical problems requiring long-term follow-up and treatment.
- A shortened life expectancy.

The severity of a chromosomal disorder is determined by the amount of missing or extra DNA, and which genes are affected. Sometimes, these missing or extra pieces cause no problems.

What Does Testing Involve?

The test requires DNA, which is obtained from cells by a chorionic villus sampling (CVS) or amniocentesis procedure. For more information on these procedures, please refer to the booklet 'Antenatal Screening and Testing for Down Syndrome and Other Conditions' **(www.healthed.govt.nz/resource/antenatalscreening-and-testing-down-syndrome-and-otherconditions).**

How Does the Test Work?

A chromosomal microarray looks for chromosomal changes 50 times smaller than those seen using a microscope. The test compares DNA from the baby to a normal DNA sample, to see if there are any missing or extra pieces of chromosomes.

Missing or extra pieces of DNA can be passed down from a parent or can happen as a new event in a baby. Blood samples from the baby's parents are routinely requested to help determine which of these applies, and to help with interpreting the test results.

What are the Risks of Testing?

While a chromosomal microarray can detect small changes, not all genetic changes can be detected. These include tiny changes in individual genes which cannot be detected by this test.

A chromosomal microarray may identify changes that are difficult to interpret, and sometimes a blood sample from both parents is analysed for comparison. These changes may be reported as 'variants of uncertain significance'.

The test may show a finding which is not related to the ultrasound findings, but which may have other health implications for the baby or other family members (for example, an increased risk of a condition which only appears later in life).

If You Agree to Have the Test:

Your doctor will arrange the test for you. They will go through the consent process with you, explaining the contents of this leaflet. To proceed with testing, you will need to sign the consent form.

After a sample is taken and analysed in the laboratory, the results will be reported to your doctor within 2-3 weeks. Your doctor will explain the results to you, including any uncertain or unrelated findings. They will also refer you to other services if needed.

For more information, please contact your local Maternal-Fetal Medicine unit.

Canterbury Health Laboratories
www.chl.co.nz | 0800THELAB

GENETIC HEALTH SERVICE NEW ZEALAND
RATONGA HAUORA IRANGA AOTEAROA