This fact sheet describes a blood test which checks for extra or missing pieces of genetic material (DNA) in the cells of the body

In summary

- **Chromosome microarray (CMA) testing is a detailed genetic test that can detect extra or missing segments of genetic material or DNA**
- **The test can be done on a blood sample (in adults and children).**

OUR DNA

In all the cells of our body our genetic material, or DNA, is packaged on string like structures called chromosomes. Our DNA contains coded information (our genes) that is used by the cells to make our bodies grow and function properly. It is important that there is the correct amount of DNA information in each cell of our body, as having more or less DNA than usual, can cause health or developmental concerns.

There are usually 46 chromosomes in each of our cells, arranged in 23 pairs.

WHAT IS CMA TESTING?

Chromosome microarray (CMA) testing is a detailed genetic test that can detect extra or missing segments of genetic material or DNA. As there are usually two copies of each chromosome, there should be two copies of each segment of DNA.

Using a small sample of blood, a CMA test will examine the amount of DNA present and look for a variation in the number of copies of the segments. This is known as a **copy number variant.**

If the test finds a copy number variant, the laboratory will check which genes it contains. Understanding the genes that are present in extra copies, or are missing, can provide information about what the result means for that individual such as:

- The cause of the developmental or health concern
- How symptoms might develop over time
- The possibility of the condition affecting future children.

WHAT INFORMATION WILL THE CMA TEST PROVIDE?

There is a CMA Testing Guide for doctors and patients available at www.genetics.edu.au

As part of an investigation into a developmental or health concern about your child or yourself, your doctor or genetics specialist may suggest a chromosome microarray (CMA) test that looks for extra or missing DNA segments. The information below will help you understand how this test works and what information you may receive from it.

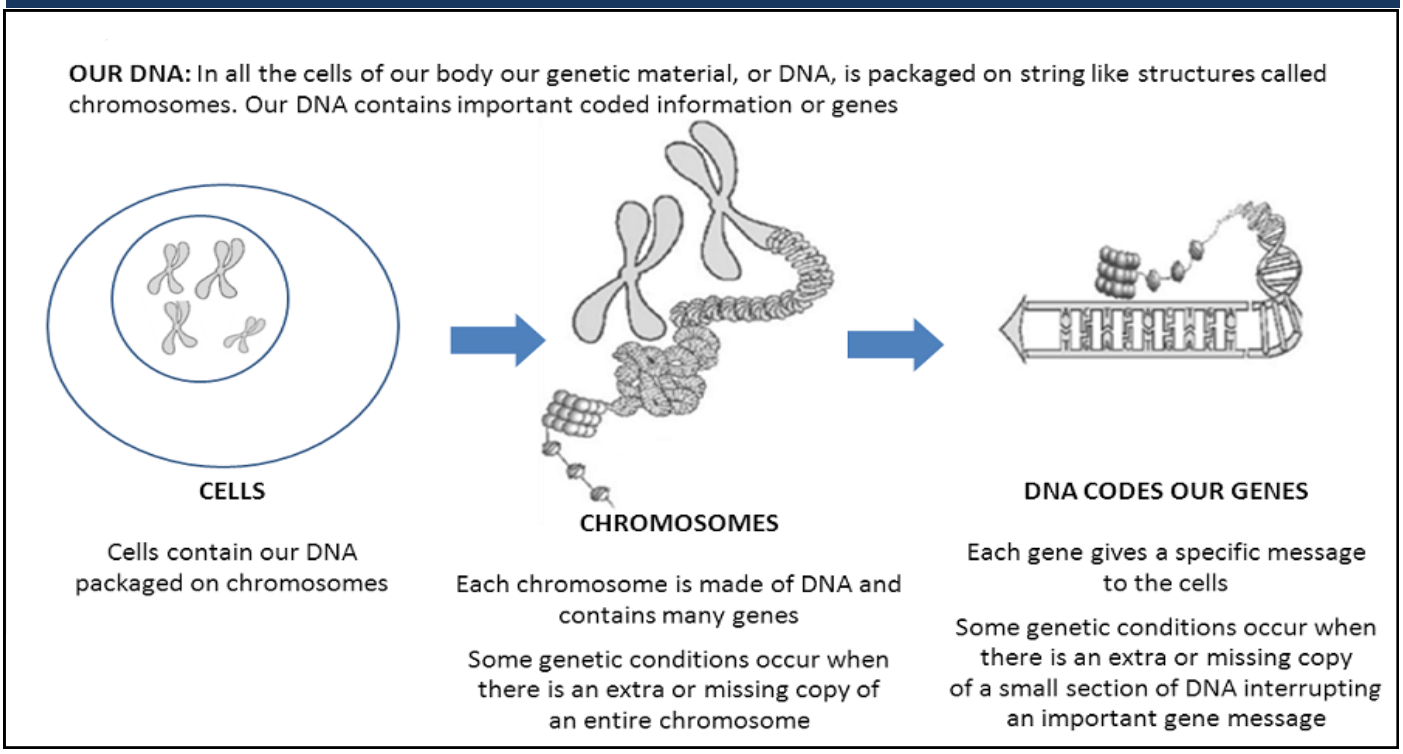
1. ***No copy number variant is found***
   The cause of the health or developmental concern remains unexplained.

   This is the most common result.

2. ***A copy number variant is found which does not usually cause any health or developmental concern***
   These are sometimes called **benign copy number variants**.

   The cause of the health or developmental concern remains unexplained.

3. ***A copy number variant is found that explains the health or developmental concern***
   Sometimes additional testing will be suggested to confirm the result by another method or testing of the parents to see if the variant is only present in the child or was inherited from a parent.



4. ***A copy number variant is found but its impact on health or development is unclear***
   This type of result is called a **variant of unknown significance**. There are a number of steps needed to further investigate this result. Additional testing will be necessary to confirm the result by another method. Also, testing of parents will usually be requested to see if the variant is only present in the child or was inherited from a parent.

The results of this additional testing will be:

- *One of the parents also has the copy number variant*
  - ◊ If the parent has the same condition as the child, the variant is likely to be the cause of the health or developmental concern
  - ◊ If the parent does not have the same condition as the child, the variant is less likely to be the cause of the health or developmental concern. However, further testing of other family members might be needed to confirm this interpretation of the finding
- *Neither of the parents has the copy number variant*

This means the variant must have occurred during or soon after the child's conception (a **de novo variant**). Two interpretations are possible:

- The variant is considered to be the cause of the developmental or health concerns being tested for

  OR
- We do not understand how the variant might relate to the developmental or health concerns being tested for. Therefore the result remains of uncertain significance.

5. ***A copy number variant is found that contains a gene or genes unrelated to the developmental or health concerns, but which could potentially cause other health problems in the future.***

   This will be a rare result. The genes located in the extra or missing DNA segment are known to be associated with other health or developmental concerns. This information could be significant for your child's, or your, future health.

   It is important to discuss what type of information you will receive from this test with your doctor or genetics clinic.