

This fact sheet describes a test carried out during pregnancy called chromosome microarray (CMA). It checks for extra or missing pieces of genetic material (DNA) in the cells of the baby.

In Summary

- **Chromosome microarray (CMA) testing is a genetic test that has only become available during pregnancy in recent years**
- **If you are having a prenatal procedure such as a chorionic villus sampling (CVS) or amniocentesis, your doctor may suggest a CMA test that looks for extra or missing parts of chromosomes**
- **The information below will help you understand how this test works and the possible results it may provide.**

OUR DNA

In all the cells of our body our genetic material, or DNA, is packaged on string-like structures called chromosomes (see *Figure 28.1*). There are 46 chromosomes in each of our cells, made up of 23 pairs. Half of the chromosomes come from our mother and the other half from our father.

Our DNA contains 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 because having more or less DNA than usual can cause health problems or intellectual disability.

WHAT IS CMA TESTING DURING PREGNANCY?

Prenatal Testing Procedures

A sample of the baby's DNA can be obtained during pregnancy using prenatal testing procedures called chorionic villus sampling (CVS) and amniocentesis.

A fine needle guided by ultrasound is used to obtain cells that contain the DNA from the developing placenta or amniotic fluid around the baby. The baby's cells are then sent to the laboratory for CMA testing.

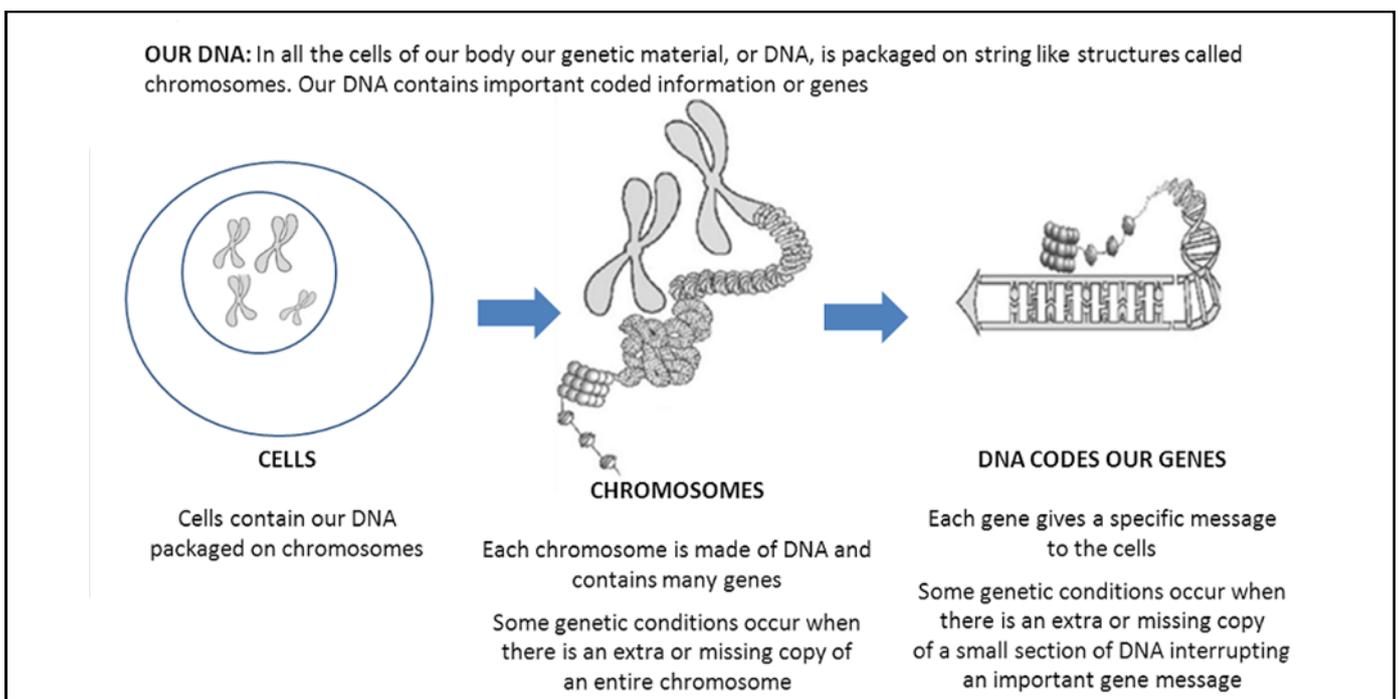


Figure 28.1: Diagram showing structure of chromosomes and genes within the chromosomes.

CMA Testing

Chromosome microarray (CMA) testing is a detailed genetic test that can detect extra or missing segments of chromosome material or DNA.

There are usually two copies of each chromosome, therefore there should be two copies of each segment of DNA along each of the chromosomes.

Using a small sample of the baby's DNA which is obtained using a prenatal testing procedure, CMA testing will look for small changes in the number of copies of the segments (known as a copy number variant). Some changes, or copy number variants, can cause significant health problems or intellectual disability.

CMA is different to other genetic tests as it can detect these small changes along each of the chromosomes.

As well as detecting these small changes, CMA can also detect where there are missing or additional whole chromosomes, such as Down syndrome which occurs when there is a whole extra copy of chromosome 21.

During pregnancy, a CMA test will examine the amount of DNA present in the baby and can detect:

- Whether there are missing or extra whole chromosomes (such as in Down syndrome), and
- Whether there are extra or missing segments of a chromosome (a copy number variant).

WHAT INFORMATION WILL THE CMA TEST PROVIDE?

There is no test during pregnancy which can rule out or detect every possible health concern. When a CMA test is carried out, there are several possible results that can be expected.

1.	<p><i>No copy number variant or chromosome imbalance is found.</i></p> <p>This is the most common result.</p>
2.	<p><i>A copy number variant is found which does not cause any health or developmental concerns.</i></p> <p>These are sometimes called benign copy number variants and are present in many people who are healthy.</p>
3.	<p><i>A copy number variant or chromosome imbalance is found that is associated with health or developmental problems.</i></p> <p>Sometimes additional CMA testing in both parents will be suggested in order to see if the variant is only present in the baby or was inherited from a healthy parent. Additional testing to confirm the result by another method may also be suggested. This will help us understand the impact that a copy number variant could have on your baby.</p>
4.	<p><i>A copy number variant or chromosome imbalance is found but its impact on health or development is unclear.</i></p> <p>This type of result is called a variant of unknown significance. Additional testing may be necessary to confirm this type of result by another method. Also, testing of parents will usually be requested to see if the variant or imbalance is only present in the developing baby or whether it was inherited from a parent.</p> <p>The results of this additional testing may show:</p> <ul style="list-style-type: none"> • If one of the parents has the same copy number variant/chromosome imbalance and is healthy, then the variant is unlikely to be of concern in the baby • If neither parent has the same copy number variant/chromosome imbalance, this means that it is a new chromosome change in the baby and it must have occurred during or soon after the baby's conception. The effect that a variant of unknown significance could have on the baby's health and development, even if it is present may be unclear.

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