Fact sheet **28**

CHROMOSOME MICROARRAY (CMA) TESTING DURING PREGNANCY

This fact sheet talks about a test in pregnancy called chromosome microarray (CMA). This test checks for extra or missing pieces of genetic material (DNA) in the cells of the baby.

IN SUMMARY

- If you are having a test in pregnancy such as a chorionic villus sampling (CVS) or amniocentesis, your doctor may suggest a CMA test that looks for extra or missing pieces of genetic material or DNA
- The information below will help you understand how this test works and the possible results it may provide
- Refer to the fact sheet on CMA for how this test may be applied on a sample taken from children and adults.

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 in the egg, and the other half from our father in the sperm.

Our DNA contains information (our genes) that is used by the cells to make our bodies grow and work properly. It is important that there is the right amount of DNA in each cell of our body. Too much or too little DNA can cause a condition affecting health, learning or development.

WHAT IS CMA TESTING DURING PREGNANCY?

Prenatal Testing Procedures

A sample of DNA can be taken during pregnancy using prenatal testing procedures called chorionic villus sampling (CVS) and amniocentesis.

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

CMA Testing

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

As there are usually two copies of each chromosome, there is generally also two copies of each section of DNA on each of the chromosomes.

Using a small sample of the DNA from a prenatal testing procedure, CMA testing will look for changes in the number of copies of the DNA segments in the baby. These are known as copy number variants. Some **copy number variants** can cause significant health concerns or learning disability.

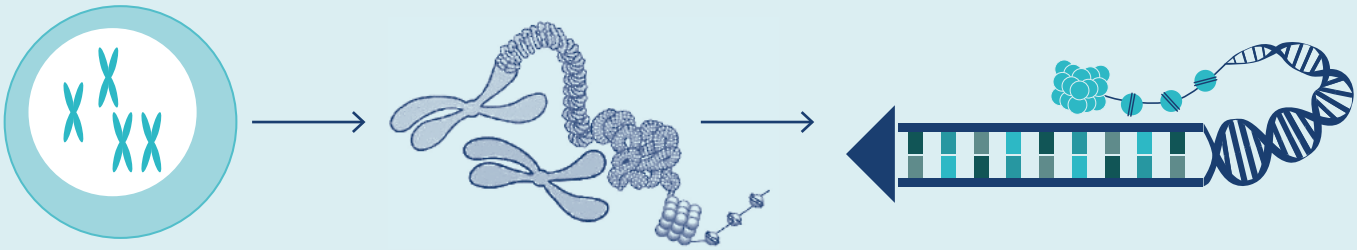
As well as finding smaller copy number variants, CMA can also find where there are missing or additional **whole** chromosomes such as Down syndrome, which happens when there is an extra copy of chromosome 21.

Sometimes CMA testing is done as part of screening for chromosomal differences in pregnancy where there are no known problems in the pregnancy. In other instances, CMA may be offered as a follow-up test when there is/may be a problem identified on pregnancy ultrasound. In this case, it is important for the health care team to consider CMA results together with what was found on ultrasound .

Content updated April 2021
SHPN: (HETI) 240948

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

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 important coded information or genes.



Cells
Cells contains our DNA packaged on chromosomes

Chromosomes
Each chromosome is made of DNA and contains many genes

Some genetic conditions occur when there is an extra or missing copy of an entire chromosome

DNA codes our genes
Each gene gives a specific message to the cells

Some genetic conditions occur when there is an extra or missing copy of a small section of DNA interrupting an important gene message

1.	**No copy number variant or chromosome imbalance is found.** This is the most common result.
2.	**A copy number variant is found that is unlikely to cause any health or developmental concerns.** These are sometimes called benign copy number variants and may be present in people who are healthy.
3.	**A copy number variant or chromosome imbalance is found that is linked with health or developmental concerns.** 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. This may help us understand the impact that a copy number variant could have on your baby. Sometimes waiting for more studies and data, may be needed to confirm our understanding of this. A local genetics service may assist.
4.	**A copy number variant or chromosome imbalance is found but its impact on health or development is unclear.** This type of result is called a **variant of uncertain significance**. Testing of parents will usually be offered to see if the variant is only present in the developing baby or whether it was inherited from a parent. The results might tell us: • If one of the parents has the same copy number variant and does not have any health or developmental conditions, 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 change in the baby and it happened during or soon after the baby's conception (a ***de novo* variant**). Depending on the situation, this may raise suspicion that this is the cause of a health condition. Even if present the effect that this variant may have on the baby's health and development, may still be uncertain until there is more evidence to say otherwise. Variants of uncertain significance that do not contain any genes are less likely to be harmful or have an impact on the baby (benign).

WHAT INFORMATION WILL THE CMA TEST PROVIDE?

There is no test in pregnancy that can rule out or find every possible health concern. There are several possible results that can be expected.

If a CMA test result predicts that your baby may have a health or developmental condition, you and your partner will be given as much information as possible about the condition and what this might mean for your pregnancy.

You can also talk about whether any more testing may help us to learn more about it.

Depending on the circumstances, if a CMA test does not indicate a clear cause for a genetic condition, other tests may be suggested.

Your doctor, midwife or genetics service can give you the opportunity to make an informed choice about whether or not you wish to have further testing and/or continue your pregnancy or not.