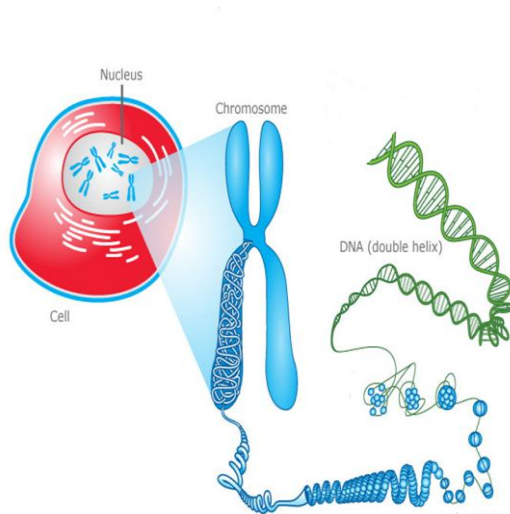


What is Chromosomal Microarray Analysis (CMA)?

CMA is an advanced genetic test that has replaced traditional chromosome testing (karyotype). CMA can detect whether there are extra or missing pieces of genetic material along any of our chromosomes.

Chromosomes are found within our cells and contain thousands of genes that tell our bodies how to grow and develop.

An individual who has extra or missing pieces of chromosomes may have problems with health, development, and/or learning.



What is the chance of finding a chromosome difference?

- The chance of having a pregnancy with Down syndrome, Trisomy 18, Trisomy 13, and some differences in sex chromosome number depends on the age of the pregnant woman, or the results of a prenatal screening test that takes the woman's age into account. A rapid test (QF-PCR) is first done on all CVS and amniocentesis samples to determine the total number of copies of chromosomes 21, 18, 13, X and Y.
- In addition to the likelihood of diagnosing one of the above conditions, **there is a 1-2% chance that CMA will detect a significant chromosome difference in the pregnancy.** This chance does *not* increase with the age of the pregnant woman. If there are ultrasound abnormalities in the pregnancy, the chance of detecting such a change may be higher; however a 'normal' ultrasound does not reduce this chance.
- **There is a 1% chance of an unclear CMA result**, called a "variant of unknown significance." This means that despite testing the parents, we do not know whether this finding may cause health and/or developmental concerns.

Other points to consider:

- A "normal" CMA result does *not* rule out all genetic conditions.
- We may have very little medical information about the types of problems that some chromosome differences can cause. The associated health issues can be extremely variable, ranging from mild to severe. In many cases, the severity of the health or developmental concerns cannot be predicted.
- If a chromosome difference is found, other family members may be offered testing.
- Although very rare, it is possible to find an unexpected chromosome difference associated with a higher chance of developing a specific adult-onset condition like breast cancer or dementia. This can have an impact on other family members.

Making a decision about having CVS or amniocentesis with CMA can be difficult. If you have further questions, speak to your genetic counsellor, or ask your health care provider about a referral for genetic counselling.

How are results reported?

In 2 separate parts:

1. **QF-PCR results** provide information regarding Down syndrome, Trisomy 18, Trisomy 13, or a difference in sex chromosome number (i.e. Turner syndrome, Klinefelter syndrome, Triple X syndrome, XYY), and are available before CMA is completed. The result is ready in 3-7 days after the procedure date.
2. **CMA results** are available in approximately 3 weeks from the date of the procedure.

In some cases, in order to interpret a result, a **preliminary CMA report may be issued requesting samples on both parents**. This will help the laboratory determine if the result is likely to be “benign,” as it is inherited from a healthy parent. A preliminary report does *not* necessarily indicate an abnormal result.

Summary of Key Points:

- **CMA detects more chromosome differences** capable of having an impact on a child’s health and/or development than previous traditional chromosome testing.
- A “normal” result **does not rule out all genetic conditions**.
- There is approximately a 1% chance of having an **unclear result**.
- **Unexpected findings** are rare but possible.
- With a normal chromosome number (46, XX or 46, XY) there is a remaining **1-2% chance of having a chromosome imbalance detectable only by CMA**.
- **Chromosome differences can have variable effects on an individual’s health**, ranging from mild to severe. In some cases, it may not be possible to predict the severity of a condition diagnosed prenatally.

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Prenatal Chromosomal Microarray (CMA) *via* Chorionic Villus Sampling (CVS) & Amniocentesis

If you are considering having a CVS or amniocentesis, it is important to understand the information these tests can provide.

