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Prenatal Chromosomal Microarray

INDIANA UNIVERSITY
DIAGNOSTIC GENETICS and GENOMICS

http://geneticslab.medicine.iu.edu/

Prenatal Chromosomal Microarray (CMA)

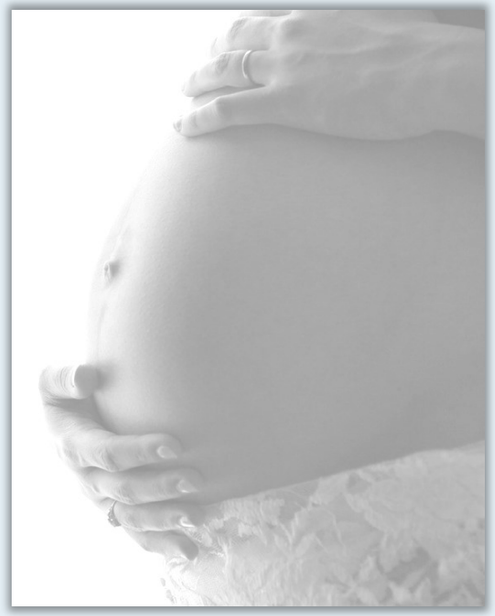
Having the proper amount of genetic material (DNA) in each cell of the body is important for normal growth and development. The DNA is located along the 23 pairs of chromosomes (46 total) in each cell. A basic karyotype analysis can only detect the number of chromosomes in each cell and large structural changes in these chromosomes. Chromosomal Microarray (CMA) analysis is an advanced method of looking at the structure and number of the chromosomes in our body. CMA is able to detect the large changes identified by karyotype, as well as smaller changes (called microdeletions and microduplications) that cannot be identified by karyotyping alone. CMA can also show whether a pair of chromosomes in the fetus came from just one parent (instead of both parents) and whether the genes originated from parents who are related. These events may help explain certain genetic disorders. For these reasons, the American College of Obstetricians and Gynecologists (ACOG) recommends that anyone who is considering prenatal diagnosis for chromosomal conditions be offered the option of CMA analysis.[1]

1. ACOG Practice Bulletin No. 163: Screening for Fetal Aneuploidy. American Obstetricians and Gynecologists. Obstet Gynecol. 2016; 127(5):e123-37.

Chromosome abnormalities may be associated with:

- Developmental delay
- Intellectual disability
- Autism spectrum disorders
- Short stature
- Other congenital abnormalities
- Embryonic and fetal death
- A history of miscarriage
- A family history of a chromosome abnormality, intellectual disability or birth defects.

In the event a chromosomal abnormality is identified, genetic counseling is highly recommended to explain the meaning of the result as well as discuss options for clinical management.



Prenatal Genetic Testing

Most pregnancies result in the birth of a healthy baby. About 3 out of 100 babies have some type of medical condition, such as a birth defect. Genetic testing during pregnancy can sometimes detect these types of medical conditions before birth; however, it is important to note that the majority of women who undergo genetic testing for their pregnancy will have normal results.

How is Prenatal CMA Testing Performed?

As with many prenatal genetic tests, CMA testing requires an amniocentesis or chorionic villus sampling (CVS) procedure. These are both ways to obtain cells that contain the fetus's chromosomes during pregnancy. Should you choose to have prenatal CMA testing, your doctor will discuss the details of the amniocentesis or CVS procedure with you. After the procedure, the sample will be sent to our laboratory where the CMA testing is performed. Most prenatal CMA results are ready 7-10 days after samples arrive in the lab. However, if the submitted sample is too small or suboptimal, analysis will be performed on cultured cells, which may require additional time for cell growth.

WHAT WILL MY RESULTS TELL ME?

What does a positive or abnormal CMA result mean?

A pathogenic or likely pathogenic result means that a deletion or duplication in the chromosomes has been detected that is known to be or likely to be associated with a genetic disease. The possible health effects of these chromosomal changes, if known, will be discussed with you by your doctor or genetic counselor.

What does an uncertain or "variant of uncertain significance" result mean?

An uncertain result means it is not clear if the finding is linked to the patient's phenotype or associated with disease. Some uncertain findings may be benign, with no impact on fetal development. Others may cause birth defects or developmental disabilities, the extent of which cannot be determined until after delivery. Uncertain results are often in regions of chromosomes where we do not have enough information to determine if two copies are necessary and only having one or having three copies would result in problems.

What does a negative or normal CMA result mean?

A negative result means that the CMA test did not detect a significant deletion or duplication in the chromosomes of the fetus. Most individuals have several very small deletions and duplications that are seen frequently and are felt to be normal. However, a normal result cannot rule out all genetic problems. Your doctor will discuss the results with you.

Why does the laboratory request blood samples from parents?

Occasionally, a finding is detected in the fetal sample that has unclear clinical consequences. Not all changes in chromosome structure cause problems in a pregnancy or a person. When a change in the chromosomes is detected in a fetus, samples from parents are evaluated to determine if one of the parents also has the change. If one parent has the change, the fetus is predicted to follow a course similar to that parent. Blood samples from parents are requested at the same time as the fetal sample to provide timely and comprehensive results to provide the best care and alleviate anxiety.

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