

Prenatal Chromosomal Microarray


T

A

A

G

C



Many couples face important decisions regarding the type of prenatal testing they wish to have for their current pregnancy. For women who are considering amniocentesis or chorionic villus sampling, Baylor Medical Genetics Laboratory is now offering Chromosome Microarray Analysis (abbreviated CMA). CMA utilizes the latest advances to provide expanded genetic testing for a large number of serious genetic disorders not detected by routine chromosome analysis. This brochure was developed to provide you with more information about this new prenatal test to help you decide if it is right for you.

What is Chromosome Microarray Analysis?

CMA is a relatively new test that is designed to look at the chromosomes in a more detailed way. It is based on the fact that too much or too little genetic material can cause problems in development. These gains or losses in DNA can lead to genetic conditions that present with significant disabilities. The new test uses a gene chip or microarray to analyze various areas of the human genome for abnormal regions that contain too many or too few copies of the genetic material.

How does the testing work?

The microarray, or gene chip, is like a map that is covered with hundreds of tiny dots consisting of DNA from known locations on each of the 46 chromosomes. The test looks for imbalances in the amount

of chromosomal material between DNA from a control and a patient's DNA, (or DNA from the fetus, present in the CVS or amniocentesis sample). When the patient sample and the control sample are compared and added to the microarray, doctors/scientists can determine if there are any differences in copy number. If a difference is found, the location of the difference and whether the difference is because of extra or missing material will often determine the type of disorder that the baby will have.

What does CMA test for?

The new test can accurately identify a number of chromosome disorders early in pregnancy that previous screens could not, including virtually all known microdeletion and microduplication syndromes.

Microdeletion syndromes are genetic diseases caused when a small amount of DNA is missing (deleted) from a chromosome. Microduplication syndromes are genetic diseases caused when a small amount of DNA is present in an extra copy (duplicated) on a chromosome. When a person is missing or has an extra copy of these pieces of DNA they can have mental and/or physical problems. Everyone has the same risk to have a child with one of these disorders, no matter what their age is.

Known genetic diseases detected by CMA include conditions characterized by mental retardation such as Angelman syndrome, Wolf-Hirschhorn syndrome, Williams syndrome, and Prader-Willi syndrome. It also detects a variety of gains or losses towards the ends (telomeres) of the chromosomes, which are important causes of many developmental disability syndromes. For more information on the conditions tested for by CMA please go to the Baylor Medical Genetic Laboratories website at www.bcmgeneticlabs.org.

A common belief of couples undergoing or considering amniocentesis or CVS is that the testing on the fetal sample includes all genetic tests. The reality is that these only test for chromosome number and basic structure, in a test called

a karyotype. When an amniocentesis is performed for genetic risks, the fetal sample is also routinely analyzed only for a fetal protein associated with open neural tube defects (spina bifida). Additional tests are added only if there is a specific indication, such as a family history of a specific disorder. However, most conditions that can affect the fetus occur for the first time in families and an increased risk usually can not be predicted. The new CMA test now offers the ability to expand the information gained from an amnio or CVS to over 140 genetic disorders for everybody, regardless of their family history, without any additional risk to the pregnancy.

What are the benefits of the testing?

The CMA is an expanded version of the prenatal testing currently available. This testing has the ability to detect abnormalities that are not detected by standard prenatal testing. If an abnormality is found, this information may be important for you in many ways. It may give you important information that could help you make decisions about your pregnancy or help guide the medical decisions for your baby after delivery. If an abnormality has already been detected in your fetus by ultrasound, the CMA test may help to determine the cause of the abnormality.

What are the limitations of the testing?

While the CMA test is very accurate, it is possible that your baby could have one of the medical conditions included in the CMA test, but the CMA test was unable to detect the condition. This is possible because many genetic syndromes have more than one cause. For some conditions included in the CMA test, 99% of the cases can be detected by the test, while for others, the detection rate may be 70% or less.

Because the CMA is a new and sensitive test, it is possible that the test will detect an abnormality for which there is very little medical information available

to predict the type of problems that may develop in the baby. As with any genetic test, results may also impact other family members and indicate the need for further testing.

Another consideration for families is the cost of the CMA testing. Since CMA is a new test, it may not be covered by your medical insurance.

How long do the results take?

The results of the CMA analysis take approximately 7-14 days for completion.

How do I learn more?

We hope this brochure answers some of your questions about the CMA testing offered by the Medical Genetics Laboratories. This testing must be done in conjunction with routine chromosome analysis and a signed consent must accompany the sample. Due to the complexity of the CMA testing, an appointment for genetic counseling is highly recommended prior to having the testing. The genetic counselor will not only discuss this testing in greater detail, but will also determine whether any other additional testing is appropriate and ensure that all of your concerns are addressed. The genetic counselor is also available by phone or in person to follow-up with you after the results of this testing become available. Further information, including consent forms and requisition forms are available at the Medical Genetics Laboratories website (www.bcmgeneticlabs.org).



Medical Genetics Laboratories

One Baylor Plaza, Nab 2015, Houston, Texas 77030

713-798-6555 • 1-800-411-gene (4363)

www.bcmgeneticlabs.org

BCM
Baylor College of Medicine