

# PRENATAL CYTOGENETIC DIAGNOSIS BY ARRAY-BASED COPY NUMBER ANALYSIS

A Clinical Study



Patient Brochure

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Sponsored by the National Institute of Child Health and Human  
Development

You are being asked to consider participating in a research study because you are having prenatal diagnostic testing, such as a chorionic villus sampling (CVS) or an amniocentesis, for one of the following reasons:

- ❖ Advanced maternal age
- ❖ Family history of a genetic disorder
- ❖ Abnormal screening test results
- ❖ Your fetus has a finding on ultrasound that your doctor would like to further evaluate



Researchers are evaluating a new laboratory test called microarray analysis, which in some cases, may increase the information provided by the analysis of your CVS or amniocentesis. All microarray results will be confirmed by a known standardized test before being reported to you, the patient. The purpose of this document is to provide information about the study so that you can decide whether it is right for you. This information is meant to be used in addition to your discussion with a genetic counselor or genetic physician.



Microarray analysis is a method of looking at the structure and number of the chromosomes in our body. All cells of our body contain 46 chromosomes (23 pairs.) Chromosomes are the structures in our cells that carry our genetic information. The development of our body and brain is very dependent on having the correct number and proper structure of chromosomes.

When a chromosome, or even a small piece of a chromosome, is extra or missing, problems in development and/or birth defects may result. An example of a problem due to an extra copy of an entire chromosome is Down syndrome.

Cells from our body can be used to look at our chromosomes. The most common way to look at chromosomes is under a microscope in a test called a karyotype. In a karyotype, the number of chromosomes can be counted and the structure of chromosomes can be determined, but in a limited way. Another method, FISH testing, uses a microscope to look at very small areas on the chromosomes that cannot be seen on a karyotype test. However, only a limited number of areas can be evaluated under the microscope.

A simplified way of thinking about the microarray method is that all of the different chromosome tests, such as a karyotype and hundreds of FISH tests, combined into a single test. In this method, many pieces of DNA (the material that carries the genetic code that makes up our body), from specific parts of all of the chromosomes are

placed on a glass slide, called a microarray or DNA chip. The DNA from the person or fetus to be tested is matched up against normal DNA spots on the DNA chip. This test is able to determine whether the DNA from the tested person or fetus has any missing or extra copies of a chromosome or its parts.

The microarray test is currently offered as a diagnostic test on children or adults to determine if a missing or extra piece of DNA is causing their medical problems. Scientists and physicians can also use microarray analysis to evaluate a fetus using cells from an amniocentesis, or cells from a chorionic villus sampling (CVS). Results of preliminary studies on approximately 100 samples show that microarray will detect all of the conditions that a karyotype can detect, plus additional information caused by missing or extra pieces of material too small to be seen under the microscope, but capable of causing health problems for the child.



You are one of approximately 5,000 subjects to be asked to participate in this study. The microarray is a new diagnostic test, and therefore, we will be collecting information about the test that may help other couples in the future. If you choose to have microarray testing, we will ask your permission to collect medical information about you, your partner, your fetus, and your microarray test results that may be used to prepare future scientific publications. We will also ask your permission to keep and reuse your sample in a tissue repository at Columbia University for future research in this area.



- ❖ You are eligible to have microarray testing if you are pregnant and are presenting for an amniocentesis or chorionic villus sampling (CVS) as part of your clinical care of pregnancy.
  - ❖ There is no cost to you for this test.
  - ❖ You will not be paid for participation.
- ❖ Microarray testing is only being offered when both mother and father of the pregnancy are available to give blood for the study.
- ❖ If you choose to participate, you will still have all the testing your physician is recommending. You will also have the microarray test in addition to this testing.
  - ❖ The turnaround time for your standard test results will not be affected.
- ❖ Microarray may detect chromosomal abnormalities that would not otherwise be detected by routine testing, of which many of these are known to cause abnormalities. Some findings may be of unknown significance and their discovery could cause additional anxiety.
- ❖ If the results of this testing are normal, your fetus could still have a birth defect or mental retardation from causes that are not detected by the microarray test.