

Prenatal Chromosomal Microarray Analysis

A new option for Prenatal Diagnosis

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Information Source for Medical Professionals

*Baylor was one of the first labs to offer array-CGH
for clinical application, and we remain a leader in the implementation
of new technology for array-CGH.*

Now Available *from the*
Medical Genetics Laboratories of
Baylor College of Medicine

**Prenatal Chromosomal
Microarray Analysis**

Prenatal Chromosomal Microarray Analysis (Prenatal CMA) is a new test that offers pregnant women and their doctors the capability to detect fetal genetic abnormalities that had previously been impossible. The unprecedented power of this new technology provides a rapid and comprehensive evaluation of fetal chromosomes at a level of resolution far superior to a standard karyotype. As a result, genetic abnormalities that would otherwise go undetected can now be identified by Prenatal CMA.

Women undergoing amniocentesis or CVS can now be offered a prenatal diagnosis for:

- *DiGeorge syndrome*
- *Williams syndrome*
- *Smith-Magenis syndrome*
- *Miller-Dieker syndrome*
- *Angelman syndrome*
- *Prader-Willi syndrome*
- *Telomere deletion syndromes*
- *More than 150 other genetic syndromes*

This new technology is revolutionizing the field of cytogenetics and the ability to detect genetic defects. It is immediately available for your patients.

Q&A for Medical Professionals

What does ACOG recommend?

Invasive diagnostic testing for aneuploidy should be available to all women, regardless of maternal age. Pretest counseling should include a discussion of the risks and benefits of invasive testing compared with screening tests; how many women will have a positive result (screen-positive rate) and, of those, how many will have a true positive result (detection rate); the detection rate of aneuploidies other than Down syndrome; and the type and prognosis of the aneuploidies likely to be missed by serum screening. Counseling should be provided by a practitioner familiar with these details.

What disorders can be detected?

Prenatal Chromosomal Microarray Analysis can detect over 150 known genetic disorders in a highly reliable and accurate manner. The capabilities far exceed the abilities of the older method, karyotype analysis. Microarray analysis will identify abnormalities that would often be undetectable by karyotype. See accompanying list of disorders.

Is such testing cost effective?

In 2004 in *Lancet* (PMID 14751700), Harris et al. state: "Prenatal diagnostic testing can be cost effective at any age or risk level. Current guidelines should be changed to offer testing to all pregnant women, not just those whose risk of carrying an affected fetus exceeds a specified threshold."

Do women want more options?

In 2004, CAUGHEY et al. in *ObstetGynecol* (PMID: 14990419) state: A substantial proportion of women of all ages indicate a desire to undergo and a willingness to pay for prenatal diagnostic testing. Variations in willingness to pay are correlated with both socioeconomic and attitudinal differences in addition to age. Guidelines regarding use of prenatal genetic diagnosis should be expanded to offer testing to all women, not just those deemed at increased risk.

In 2005 in *Gynecol Obstet Invest* (PMID 15711084), Kuppermann and Norton state: "In this paper, we summarize the evidence suggesting that this aspect of prenatal testing guidelines should be revisited, and that women should be allowed to make informed decisions regarding the use of invasive testing that are reflective of their own values and preferences."

If a woman at ordinary risk chooses to have amniocentesis, should the new CMA test be offered?

We believe that Prenatal CMA is an attractive option for any woman undergoing genetic amniocentesis or CVS. The availability of Prenatal CMA adds to the ability to detect serious abnormalities.

Q&A for Medical Professionals

What is the chance of finding a significant abnormality that would not be detected by karyotype (the current cytogenetic test)?

We are participating in an NIH funded inter-institutional study to address this question. Current estimates are that the probability of finding a serious abnormality that would be missed by karyotype is between 1 in 200 to 1 in 600.

How reliable are the findings of this new test?

Prenatal CMA is highly reliable due to the unique design of the test. It uses the latest molecular detection methods to analyze the DNA of the fetus. The test provides an objective, highly accurate result that is easy to interpret and eliminates the subjectivity and lower resolution of karyotype analysis.

What about the detection of findings of uncertain significance?

It is the lab policy that findings of uncertain significance such as a medium size deletion or duplication that is also present in a healthy parent will not be reported unless the patient specifically requests this information

through a physician or counselor. In over 300 cases where parents were informed of findings of uncertain significance, no family has chosen to terminate a pregnancy based on findings of uncertain significance.

If a patient chooses to have Prenatal CMA testing, should a karyotype or aneuploidy FISH still be performed?

It is quite rare for a karyotype to detect an abnormality of significance for the health of the fetus that would not be detected by Prenatal CMA. However, it is probably most conservative at present to perform both karyotype and CMA. This may change in the future. We believe that it is far better utilization of funds to use the dollars that might go for aneuploidy FISH towards payment for CMA.

What is the turn around time for Prenatal CMA?

If the sample comes directly to the BCM Laboratory, analysis is performed without cell culture and the turn around time is 5-9 days. If the direct analysis of amniotic fluid or CVS is not satisfactory, cells will be cultured to obtain sufficient DNA and turnaround time will be longer.

Will insurance pay for the testing?

Most insurance companies will pay for some portion of the cost for this new technology. Since it is a new test, insurance plans have varying levels of coverage.

Will the BCM Medical Genetics Laboratories bill insurance?

BCM Medical Genetics Laboratories will bill insurance. BCM requires a partial prepayment from the patient with sample submission.

Patients will be responsible for copayment, coinsurance, deductible and may be responsible for payment due to insurance coverage, limitations, exclusions or non-covered services.

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