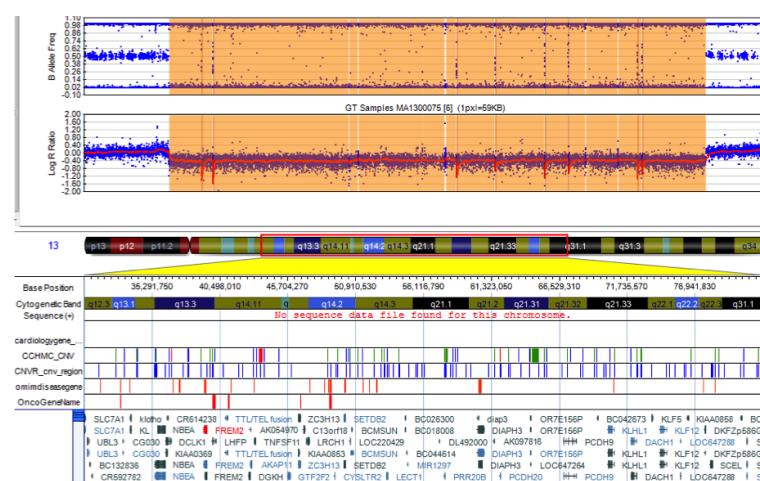


# Prenatal SNP Microarray



SNP (single nucleotide polymorphism) microarray is performed using a BeadChip platform for the most comprehensive coverage to detect cytogenetic imbalances smaller than what can be detected through routine chromosome analysis. This testing is designed to look for imbalances across the entire genome and can detect triploidy, aneuploidy, hundreds of common microdeletion/microduplication syndromes, subtelomeric deletions and duplications. Large regions of homozygosity can also be identified, indicating uniparental disomy or shared parental ancestry. The SNP based array can detect imbalances that may not be well described as well as further refine chromosomal breakpoints for previously identified chromosome imbalances. Testing will only detect the loss (deletion) or gain (duplication) of chromosomal regions represented on the arrays and is therefore not intended to replace conventional cytogenetic analysis. Balanced or low-level mosaic chromosome changes as well as tetraploidy may not be detected. Additionally, point mutations or single gene disorders will not be identified using this technology.

## Indication:

Prenatal Array testing could be offered when there is a suspicion of a chromosome imbalance. This is an ideal test for fetuses with abnormal ultrasound findings that are suggestive of a chromosome imbalance.

Prenatal array might also be offered for other indications when there is a suspicion of a suspected chromosome imbalance including advanced maternal age, abnormal screening results, or a family history of mental retardation or birth defects.

## Testing Methodology:

Microarray analysis is performed using a SNP based microarray chip with approximately 850,683 markers. This test is designed to identify chromosomal imbalances throughout the human genome including deletions, duplications and aneuploidy. Our laboratory has established criteria for reporting abnormalities based on size, gene content, and clinical significance.

## **Accuracy:**

The performance characteristics of this test have been developed and validated by the Cincinnati Children's Hospital Cytogenetics Laboratory. The vast majority of known microdeletion/duplication syndromes as well as many imbalances in regions that have not been previously characterized clinically can be detected using the SNP Microarray technology. However, conditions that may be caused by other genetic changes cannot be clinically ruled out based on a normal SNP Microarray result. If a specific genetic diagnosis is suspected, please contact the laboratory for additional testing. **See details at our website:** [www.cincinnatichildrens.org/cytogenetics](http://www.cincinnatichildrens.org/cytogenetics).

## **Specimen:**

**Please note: PARENTAL samples are requested to accompany the FETAL sample.**

### **Prenatal Microarray**

Available as a standalone test or as part of the Prenatal Reflex Test.

### **Amnio**

25 mLs amniotic fluid.

Smaller volumes may require additional culturing time. Consider sending additional fluid if special studies are ordered.

### **CVS**

40 mg branched, clean fetal villi.

Smaller volumes may require additional culturing time.

**NO formalin or freezing.**

### **Parental Sample**

#### **From EACH parent:**

3 mLs whole blood in lavender top (EDTA) tube AND  
3 mLs whole blood in green top (Sodium Heparin) tube  
OR one Oragene saliva kit (if needed, please call laboratory for supplies).

## **Turn Around Time:**

Direct Amnio: 7-10 days

Cultured Amnio and CVS: 14-21 days

For institutional billing: TAT begins at sample receipt

For insurance billing: TAT begins at precert approval

## **Cost & CPT Codes:**

Please call 1-866-450-4198 for current pricing and CPT codes, or with any other billing questions.

## **Results:**

Results will be reported to the referring physician and/or genetic counselor as specified on the requisition form.

## **Shipping Instructions:**

Please enclose the cytogenetic test requisition with sample. All information must be completed before sample can be processed.

Place samples in Styrofoam mailer and ship at room temperature by overnight Federal Express to arrive Monday through Friday. Saturday delivery is available. Please call for specific information and instructions for Saturday delivery.

Ship to:

Cytogenetics and Molecular Genetics Laboratories  
3333 Burnet Avenue NRB 1042  
Cincinnati, OH 45229  
513-636-4474