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 using this technology. 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.

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, aneuploidy, and region of homozygosity. 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
**Specimen:**

*Please note that two different tubes are needed.*

3 mLs whole blood in lavender top (EDTA) tube
AND 3 mLs whole blood in a green top (Sodium Heparin) tube.
Label tube with patient’s name, birth date, and date of collection.

**Turn Around Time:**

14-21 days.

**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