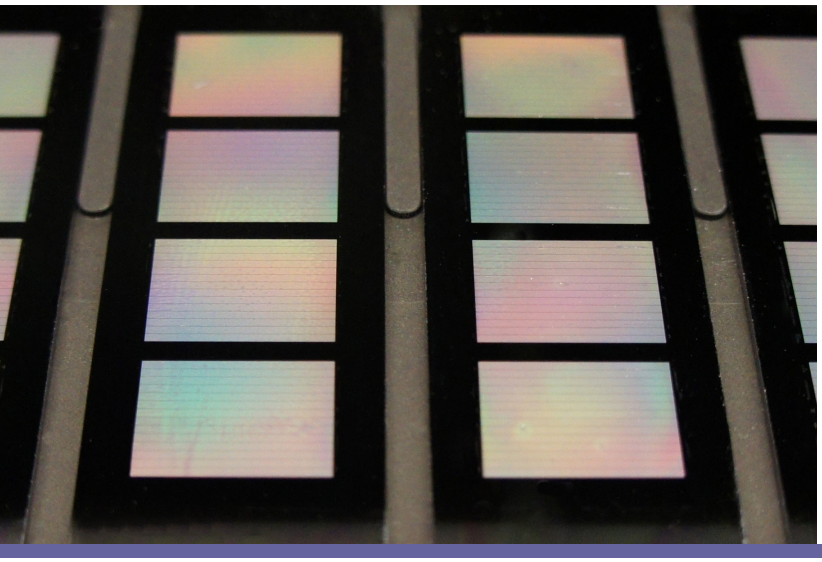
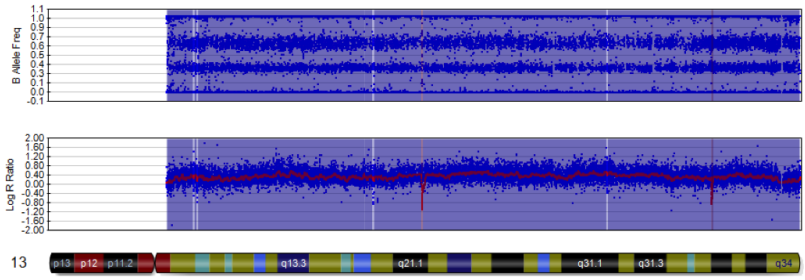
SNP Microarray Testing on Fetal Tissue/Products of Conception

Cincinnati Children's
Human Genetics





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.

Indication:

If chromosome analysis is normal or cannot be obtained because of a culture failure, microarray analysis is recommended. However, if you suspect a specific genetic condition or a single gene disorder, microarray testing may not be the appropriate next step.

SNP array testing is indicated when a chromosome imbalance is suspected such as fetal demise, miscarriage, fetal malformations, abnormal ultrasound findings, advanced maternal age, family history of chromosome abnormality, or abnormal maternal serum screening.

Testing Methodology:

Microarray analysis is performed using a SNP based microarray chip with approximately 951,117 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.

Cytogenetics Laboratory • CLIA#: 36D0900916 • Phone: (513) 636-4474 • Fax: (513) 636-4373
Email: cytogenetics@cchmc.org • www.cincinnatichildrens.org/cytogenetics

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:

25-50 mg of fetal tissue in sterile tube with sterile transport media (available from the laboratory).
If transport media is unavailable, the specimen may be placed in a sterile container on a piece of sterile gauze, moistened (not floating) with sterile saline or sterile water.
Send chorionic villi as well as fetal tissue, if available.

Do not place in formalin.

Indicate type of tissue and whether or not the specimen was obtained aseptically.

Label tube with mother's name, birth date, gestational age of pregnancy, 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

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