Microarray SNP Testing

Description:

Microarray SNP (single nucleotide polymorphism) 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 identify 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 array and therefore is 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.

Indications:

- Autism Spectrum Disorder
- Global Developmental Delay
- Intellectual Disability
- Multiple Congenital Anomalies

Methodology:

Microarray analysis is performed using a SNP based microarray chip with approximately 850,000 markers. This test is designed to detect chromosomal imbalances throughout the human genome including deletions, duplications, aneuploidy, and regions 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 Genetics and Genomics Diagnostic Laboratory at Cincinnati Children's. 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.

Turn-Around Time:

14-21 days

Results:

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

CPT Code:

- Microarray SNP - **81229**

Cincinnati Children's®
changing the outcome together

Genetics and Genomics Diagnostic Laboratory
CLIA#: 36D0656333
Phone: (513) 636-4474
Fax: (513) 636-4373
Email: LabGeneticCounselors@cchmc.org
www.cincinnatichildrens.org/genetics

Specimen:

Please note that two (2) different tubes are required

- 3mLs whole blood in lavender top (EDTA) tube **AND**
- 3mLs whole blood in a green top (Sodium Heparin) tube

Shipping Instructions:

Please enclose a test requisition with samples. **All information on the requisition must be completed before samples can be processed.**

Place samples in Styrofoam mailer and ship at room temperature for overnight delivery to arrive Monday through Saturday.

Cincinnati Children's
Genetics and Genomics Diagnostic Laboratory
3333 Burnet Ave.
TCHRF 1042
Cincinnati, OH 45229-3039

For samples that arrive on **Saturday**, please call the laboratory at 513-636-4474 to inform and include **"DOCK 5"** after "TCHRF 1042" in the address