Test Information

Test Name: Chromosomal Microarray, Congenital, Blood

Performing Lab: Cytogenetics

Synonyms: Microarray
Copy Number Microarray
Array CGH
Constitutional Array
SNP Array
Whole Genome Array

Turnaround Time: 4-21 days

Additional Interpretation: This test has been identified as a first-tier postnatal test for individuals with multiple anomalies that are not specific to well-delineated genetic syndromes, including (but not limited to): developmental delay or intellectual disability, as well as autism spectrum disorders, as recommended by the American College of Medical Genetics and Genomics.

Given that chromosomal microarray testing provides a higher resolution than possible with conventional chromosomal testing, this analysis is also appropriate to evaluate individuals who have previously had a normal conventional cytogenetic study, since these individuals could have cryptic imbalances that would be undetectable using chromosomal banding (or targeted FISH) tests. The types of clinical findings that might be reported in this follow-up testing group include (but are not limited to) congenital anomalies, developmental delay or intellectual disability, and autism.

This test can also be used to assess regions of homozygosity related to uniparental disomy or identity by descent.

This test can result in the detection of copy number variants of uncertain significance. Additional studies of parental specimens can aid in the interpretation of these variants.

Although chromosomal microarray data is powerful for detecting even small imbalances, this method does not provide information about the structural

nature of the imbalance. Therefore, some abnormal results may also require further characterization by FISH or additional techniques to allow for the provision of appropriate genetic counseling.

Methodology: DNA is extracted from the patient's peripheral blood. After extraction, the DNA is digested, labeled and hybridized to the microarray. Following hybridization, the microarray is scanned and the intensity of signal is measured and compared to a reference data set. These data are used to determine copy number changes and regions showing a loss of heterozygosity.

CPT Code Information: 81229

Specimen & Collection Information: The specimen requirements are listed below.

Specimen Name: Whole Blood

Container type: These specimens can be collected into either (1) a lavender top tube (EDTA); (2) a dark green top tube (sodium heparin) or (3) a yellow top tube (acid-citrate-dextrose [ACD])

Specimen Volume: 3ml

Special Handling: Room temperature

Specimen Transportation and Storage Information: Deliver to the lab immediately for processing. Keep at room temperature. Do not spin.