TRICORE REFERENCE LABORATORIES

CHROMOSOMAL MICROARRAY

TriCore Reference Laboratories now offers chromosomal microarray (CMA) testing for postnatal diagnosis of constitutional disorders. The American College of Medical Genetics and Genomics (ACMG) recommends chromosomal microarray as first-line testing in individuals with intellectual disability/developmental delay and congenital abnormalities when patient history and physical examination do not provide an obvious syndrome diagnosis. The diagnostic yield of the CMA assay in these individuals is 10-20%. In contrast, the diagnostic yield of G-banded karyotype is approximately 3%.

The prevalence of developmental disabilities in the United States is reported to be 13.87% across all racial, ethnic, and socioeconomic groups. Intellectual disability/developmental delay and congenital abnormalities both may be associated with copy number variations (deletions or duplications) in the genome too small to be detected by traditional G-banded karyotype. The chromosomal microarray assay utilizes more than 2.69 million markers for copy number analysis, including 749,000 SNP probes and 1.9 million non-polymorphic markers, spaced across the entire genome, with focused coverage of genes and regions of high relevance. CMA can detect variations in copy number as well as increased homozygosity that may be indicative of uniparental disomy or consanguinity, which may suggest an increased risk for an underlying autosomal recessive disorder. Confirmation of results with FISH, qPCR, G-banded karyotype, or other method may be required. Depending on results and clinical presentation, additional testing may be indicated.

Some abnormalities cannot be detected by CMA, including very small deletions/duplications, balanced rearrangements, mosaic copy number <20%, base pair mutations in genomic DNA, and mutations of mitochondrial DNA. The CMA assay is not approved for use in prenatal testing, population screening, or detecting acquired or somatic mutations. In patients with obvious chromosomal abnormalities such as Down syndrome, a family history of chromosomal abnormalities, or a history of miscarriages, G-banded karyotype is recommended as the first line test. Clinical genetic evaluation and counseling is recommended before ordering CMA testing.

For additional information, contact TriCore's molecular diagnostics laboratory at 505-938-8684 or cytogenetics laboratory at 505-938-8430.

For consultation, please contact:
Dr. Rama Gullapalli rgullapalli@salud.unm.edu
Dr. Sheldon Robinett sheldon.robinett@tricore.org
Dr. Qian-Yun Zhang qzhang@salud.unm.edu

CHROMOSOMAL MICROARRAY

Test Code..... CMASNP
CPT Code..... 81229
Methodology..... Genomic Microarray (Oligo-SNP Array)
Turnaround Time..... 7-14 days

COLLECTION	
Specimen	Whole Blood
Collection Container	Preferred: Green top tube (sodium heparin) Acceptable: Lavender top tube (EDTA)
Collection Amount	Preferred.....5.0 mL Minimum.....1.0 mL
Rejection Criteria	Frozen, clotted and specimens other than whole blood are unacceptable
PROCESSING	
Specimen Processing Instructions	Do not centrifuge Do not freeze
SHIPPING/TRANSPORT AND STORAGE	
Stabilities/Storage (*Collection to initiation of testing*)	**Temperature** **Stability** Ambient 2 days Refrigerated 5 days Frozen Unacceptable
Shipping Instructions	Ship refrigerated