Ambry Genetics
A Konica Minolta Company

SNP Array Product Summary | August 2018

CHROMOSOMAL MICROARRAY ANALYSIS (CMA) FOR COPY NUMBER VARIATION DETECTION

When To Consider Testing

SNP Array should be considered for all individuals with syndromic or non-syndromic conditions that may be caused by gain or loss of genomic material (imbalance of genomic copy number). Indications include individuals with neurodevelopmental conditions (intellectual disability, developmental delay, and/or autism), dysmorphic features, multiple congenital anomalies, seizure disorders, as well as other health and developmental concerns.

Practice Guideline

American Academy of Neurology (AAN) | American Academy of Pediatrics (AAP) | American College of Medical Genetics and Genomics (ACMG)

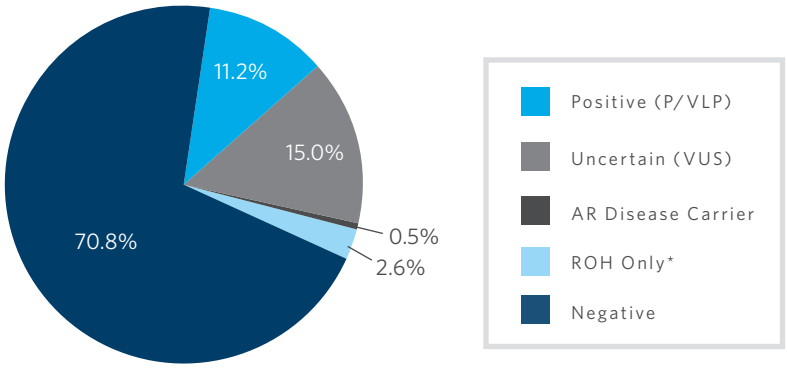
The AAN, AAP, and ACMG recommend chromosomal microarray (CMA) and fragile X DNA analysis as first-tier genetic tests in the evaluation of individuals with intellectual disability and/or autism spectrum disorder.

Michelson (2011) Neurology | Moeschler (2014) Pediatrics | Miller DT (2010) Am J Hum Genet

TEST DETAILS

- Genome-wide detection of copy number losses and gains at a much more detailed resolution than traditional chromosomal karyotype analysis
- Copy losses >100kb in size and copy gains >300kb in size are reported
- >1.9 million copy number probes and ~750,000 SNP probes
- Detection of copy number neutral regions of homozygosity (ROH); risk for autosomal recessive diseases and uniparental disomy (UPD)

Overall Detection Rates



** Additional ROH detected in 2.1% cases of pathogenic, likely pathogenic, VUS, or AR disease carrier CNV, for a total ROH detection rate of 4.7%*

Samples Submission

Multiple sample types accepted including blood, saliva, and saliva swab.

Parental Studies

No-cost parental analysis is available for copy number variants that are of uncertain clinical significance. Please contact Ambry for more details.

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