PHOENIX CHILDREN'S Hospital

Clinical Genomics Laboratory

Test Information Sheet

Phone: (602) 933-3500 Fax: 602-933-8961

Email: dgGenomicsLab@phoenixchildrens.com

Chromosomal Microarray

CPT CODE(S):	81229x1
TURNAROUND TIME:	14 days
PREFERRED SPECIMEN:	800µL of whole blood in EDTA (lavender top), or Sodium Heparin (dark green top)[1]
ALTERNATIVE SPECIMEN:	Oragene Saliva Tube (OGR-500 or OGR-575); Oragene Buccal Swab (OCD-100)

CMA is designed to look for imbalances in chromosomal material that are smaller than what can be detected through routine chromosome analysis. This testing can detect imbalances such as triploidy, aneuploidies, common microdeletions and duplications, and subtelomeric deletions and duplications. SNP based analysis also allows for qualitative information such as large regions of homozygosity. CMA has been recommended by the American College of Medical Genetics (ACMG), the American Academy of Neurology (AAN), the Child Neurology Society (CNS), and the American Academy of Pediatrics (AAP) as a first-line genetic test for individuals with these conditions.

Chromosomal Microarray Analysis (CMA) has the highest diagnostic yield of any single clinically available test for children with:

- Developmental delays
- Intellectual disability
- Autism spectrum disorders
- Multiple congenital anomalies

Testing Methodology:

CMA is performed using the CytoScan HD array. DNA isolated from peripheral blood/saliva is fragmented by restriction enzymes, amplified and hybridized to the array after biotin labeling. After hybridization, arrays are washed, scanned and the resulting data are analyzed using Chromosomal Analysis Suite software. Cytoscan HD array contains more than 2.6 million markers covering all RefSeq and OMIM™ genes. About 750,000 of these markers are single nucleotide polymorphic that can detect absence of heterozygosity (AOH) and the remaining 1.9 million markers are non-polymorphic to detect copy number changes. This platform has greater than 99% sensitivity in detecting copy number changes greater than 50 kb and AOH of greater than 5 Mb.

Limitations:

Benign copy number changes are not reported. Copy number changes <300 Kb are not reported unless they are known to be clinically significant. This technology is not designed to identify point mutations, single nucleotide polymorphisms or balanced rearrangements. This test has not been validated for detecting mosaicism. A normal SNP microarray result may not rule out a genetic diagnosis as other genetic changes may be present that cannot be detected through this methodology.

Results:

Results will be reported to the referring physician and/or genetic counselor as specified on the electronic or written requisition.

Shipping and Billing Information:

Please enclose completed requisition form and signed consent document with sample. All information must be completed before samples can be processed. Ship at room temperature by insulated carrier overnight. Do not heat or freeze.

Ship to:

ATTN: Clinical Genomics Laboratory
Phoenix Children's Hospital
1919 E. Thomas Rd.
Phoenix, AZ 85016

[1] Wait at least 2 weeks after a packed cell/platelet transfusion, and at least 4 weeks after a whole blood transfusion prior to blood draw for testing