ARUP LABORATORIES | aruplab.com
500 Chipeta Way, Salt Lake City, Utah 84108-1221
phone: 801-583-2787, toll free: 800-522-2787
Jonathan R. Genzen, MD, PhD, Chief Medical Officer

PATIENT REPORT

Patient Age/Sex: Unknown

Specimen Collected: 27-Jan-26 15:22

Cytogenomic SNP Microarray - RAPID | **Received: 27-Jan-26 15:23** | **Report/Verified: 29-Jan-26 10:51**

Procedure	Result	Units	Reference Interval
Cytogenomic SNP Microarray	**Abnormal** * f1 i1		[Normal]

Result Footnote

f1: Cytogenomic SNP Microarray
Test Performed: Cytogenomic SNP Microarray - RAPID (CMA RAPID)
Specimen Type: Peripheral blood
Indication for Testing: Premature newborn, Cardiac defect

RESULT SUMMARY
Abnormal Microarray Result (Male)

Trisomy 18 (Edwards syndrome)

Classification: Pathogenic
Copy number change: 18p11.32q23 gain
Size: 77.9 Mb

RESULT DESCRIPTION
This analysis showed a gain of all probes on chromosome 18, indicating an additional copy (trisomy) of this chromosome.

INTERPRETATION
This result is consistent with a clinical diagnosis of trisomy 18 (Edwards syndrome). Features associated with trisomy 18 may include intrauterine growth restriction with low birth weight, multiple congenital anomalies involving the brain, spinal cord, heart, abdominal wall and kidneys, hypotonia at birth progressing to hypertonia in later infancy, feeding difficulties, and severe to profound developmental delay/intellectual disability. Other findings may include craniofacial dysmorphism, ear anomalies, clenched fists with overlapping fingers, and rocker-bottom or clubfeet. Trisomy 18 is also associated with high neonatal and infant mortality.

Recommendation:
Genetic counseling

Health care providers with questions may contact an ARUP genetic counselor at (800) 242-2787 ext. 2141.

References:
1) Cereda and Carey. The trisomy 18 syndrome. Orphanet J Rare Dis. 2012 Oct 23;7:81. PMID: 23088440.
2) Carey and Kosho. Perspectives on the care and advances in the management of children with trisomy 13 and 18. Am J Med Genet C Semin Med Genet. 2016 Sep;172(3):249-50. PMID: 27643592.
3) Andrews et al. Shared decision making and the pathways approach in the prenatal and postnatal management of the trisomy 13 and trisomy 18 syndromes. Am J Med Genet C Semin Med Genet. 2016 Sep;172(3):257-63. PMID: 27557275.
4) Jones et al. Smith's Recognizable Patterns of Human Malformations. 7th edition. Philadelphia, PA: Elsevier Saunders; 2013.
5) Gardner and Amor. Gardner and Sutherland's Chromosome Abnormalities and Genetic Counseling. 5th edition. New York, NY: Oxford; 2018.
6) The Trisomy 18 Foundation Support Group. (www.trisomy18.org)
7) The Support Organization for Trisomy 18, 13 and Related Disorders (SOFT). (www.trisomy.org)

Cytogenomic Nomenclature (ISCN):
arr (18)x3

*=Abnormal, #=Corrected, C=Critical, f=Result Footnote, H-High, i-Test Information, L-Low, t-Interpretive Text, @=Performing lab

Result Footnote

f1: Cytogenomic SNP Microarray

This result has been reviewed and approved by

A portion of this analysis was performed at the following location(s):

Test Information

i1: Cytogenomic SNP Microarray

INTERPRETIVE INFORMATION: CYTOGENOMIC SNP MICROARRAY - RAPID

Technical Information
- This assay was performed using the CytoScan(TM) HD Suite (Thermo Fisher Scientific) according to validated protocols within the Genomic Microarray Laboratory at ARUP Laboratories
- This assay is designed to detect alterations to DNA copy number state (gains and losses) as well as copy-neutral alterations (regions of homozygosity, ROH) that indicate an absence- or loss-of-heterozygosity (AOH or LOH)
- AOH may be present due to parental relatedness (consanguinity) or uniparental disomy (UPD)
- LOH may be present due to acquired UPD (segmental or whole chromosome)
- The detection sensitivity (resolution) for any particular genomic region may vary dependent upon the number of probes (markers), probe spacing, and thresholds for copy number and ROH determination
- The CytoScan HD array contains 2.67 million markers across the genome with average probe spacing of 1.15 kb, including 750,000 SNP probes and 1.9 million nonpolymorphic probes
- In general, the genome-wide resolution is approximately 25-50 kb for copy number changes and approximately 3 Mb for ROH (see reporting criteria)
- The limit of detection for mosaicism varies dependent upon the size and type of genomic imbalance. In general, genotype mixture due to mosaicism (distinct cell lines from the same individual) or chimerism (cell lines from different individuals) will be detected when present at greater than 20-30 percent in the sample
- Genomic coordinates correspond to the Genome Reference Consortium human genome build 37/human genome issue 19 (GRCh37/hg19)

Variant Classification and Reporting Criteria
- Copy number variant (CNV) analysis is performed in accordance with recommendations by the American College of Medical Genetics and Genomics (ACMG), using standard 5-tier CNV classification terminology: pathogenic, likely pathogenic, variant of uncertain significance (VUS), likely benign, and benign
- CNVs classified as pathogenic, likely pathogenic, or variant of uncertain significance are generally reported, based on information available at the time of review

Test Information

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- Known or expected pathogenic CNVs affecting genes with known clinical significance but which are unrelated to the indication for testing will generally be reported with opt-in
- Variants that do not fall within the standard 5-tier CNV classification categories may be reported with descriptive language specific to that variant
- In general, recurrent CNVs with established reduced penetrance will be reported
- Recessive disease risk may be reported based on review of the submitted clinical and phenotype information alongside concurrent sequencing data review and recurrent CNVs with established reduced penetrance will be reported
- For a list of databases used in CNV classification, please refer to ARUP Constitutional CNV Assertion Criteria, which can be found on ARUP's Genetics website at www.aruplab.com/genetics
- CNVs classified as likely benign or benign that are devoid of relevant gene content or reported as common findings in the general population, are generally not reported
- CNV reporting (size) criteria: losses greater than 50 kb and gains greater than 400 kb are generally reported, dependent on genomic content
- ROH are generally reported when a single terminal ROH is greater than 3 Mb and a single interstitial ROH is greater than 10-15 Mb (dependent upon chromosomal location and likelihood of imprinting disorder) or when total autosomal homozygosity is greater than 3 percent (only autosomal ROH greater than 3 Mb are considered for this estimate)

Limitations
This analysis cannot provide structural (positional) information associated with genomic imbalance. Therefore, additional cytogenetic testing by chromosome analysis or fluorescence in situ hybridization (FISH) may be recommended.

Certain genomic alterations may not or cannot be detected by this technology. These alterations may include, but are not limited to:
- CNVs below the limit of resolution of this platform
- Sequence-level variants (mutations) including point mutations and indels
- Low-level mosaicism (generally, less than 20-30 percent)
- Balanced chromosomal rearrangements (translocations, inversions, and insertions)
- Genomic imbalance in repetitive DNA regions (centromeres, telomeres, segmental duplications, and acrocentric chromosome short arms)

Data Sharing
In cooperation with the National Institutes of Health's effort to improve understanding of specific genetic variants, ARUP submits HIPAA-compliant, de-identified (cannot be traced back to the patient) genetic test results and health information to public databases. The confidentiality of each sample is maintained. If you prefer that your test result not be shared, call ARUP Laboratories at

Test Information

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800-242-2787 ext. 3301. Your de-identified information will not be disclosed to public databases after your request is received, but a separate request is required for each genetic test. Additionally, patients have the opportunity to participate in patient registries and research. To learn more, visit ARUP's Genetics website at www.aruplab.com/genetics.

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the U.S. Food and Drug Administration. This test was performed in a CLIA-certified laboratory and is intended for clinical purposes.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.