

Patient ID SA00170050	Patient Name TESTINGRNA, SAMPLEREPORT-CMACB	Birth Date 2018-03-15	Sex M	Age 6
Order Number SA00170050	Client Order Number SA00170050	Ordering Physician CLIENT,CLIENT	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 25 Aug 2024 09:33		

Chromosomal Microarray, Blood

Result Summary

22q11.21 deletion

MCR

Result

MCR

INTERPRETATION	CHANGE	REGION	SIZE
Pathogenic	Deletion	22q11.21	2.5 Mb

Nomenclature

MCR

arr[hg19] 22q11.21(18,916,842–21,465,662)x1

Sex:
XY

Interpretation

1 MCR

A 2.5 megabase deletion at 22q11.21 was observed. The deleted interval involves approximately 77 known genes. This deletion is consistent with a diagnosis of 22q11.2 microdeletion syndrome (DiGeorge/Velocardiofacial syndrome), which is associated with developmental delay, autism, cardiac abnormalities, dysmorphic facial features, and other congenital abnormalities. Approximately 7% of these deletions are inherited and there is significant phenotypic variability (Kobrynski and Sullivan, Lancet 370:1443–1452, 2007; McDonald-McGinn et al., GeneReviews. URL: www.ncbi.nlm.nih.gov/books/NBK1523/ accessed on 08/26/2024). Parental FISH studies, test CMAFF (CMA Familial Testing, FISH), could be performed to determine if this deletion was inherited or de novo and may help determine recurrence risks. A genetic consultation may be of benefit.

Genes in the 22q11.21 deletion:

LOC102724788, PRODH, DGCR5, DGCR5, DGCR5, DGCR2, DGCR11, ESS2, TSSK2, GSC2, LINC01311, SLC25A1, CLTCL1, HIRA, MRPL40, C22orf39, UFD1, CDC45, CLDN5, LINC00895, SEPTIN5, SEPT5-GP1BB, GP1BB, TBX1, GNB1L, RTL10, TXNRD2, COMT, MIR4761, ARVCF, TANGO2, MIR185, DGCR8, MIR3618, MIR1306, TRMT2A, MIR6816, RANBP1, SNORA77B, ZDHHC8, CCDC188, LOC284865, LINC00896, RTN4R, MIR1286, DGCR6L, FAM230G, LOC107987394, FAM230A, LOC107987389, GGTL3, TMEM191B, PI4KAP1, RIMBP3, FAM230J, ZNF74, SCARF2, KLHL22, MED15, POM121L4P, TMEM191A, PI4KA, SERPIND1, SNAP29, CRKL, LINC01637, AIFM3, LZTR1, THAP7, THAP7-AS1, TUBA3FP, P2RX6, SLC7A4, MIR649, P2RX6P, LRRC74B, BCRP2

This assay does not rule out balanced chromosome abnormalities, imbalances of chromosomal regions not represented by probes on the array, or mosaicism. A normal result does not exclude the diagnosis of any of the disorders tested for on this array.

Reason For Referral

MCR

heart defect, dysmorphic features

Performing Site Legend

Code	Laboratory	Address	Lab Director	CLIA Certificate
MCR	Mayo Clinic Laboratories - Rochester Main Campus	200 First Street SW, Rochester, MN 55905	Nikola Baumann Ph.D.	24D0404292



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Specimen

MCR

Blood

Method

MCR

Chromosomal microarray (CMA) analysis was performed using both copy number and single-nucleotide polymorphism (SNP) probes on a whole-genome array (Thermo Fisher Scientific) CytoScan HD Accel platform; 2.0 million copy number probes and 750,000 SNPs using in silico controls. The genome-wide functional resolution of this array is approximately 30 kilobases for deletions and 60 kilobases for duplications. All data was analyzed and reported using the February 2009 NCBI human genome build 37.1 (hg19). Deletions larger than 200 kilobases, duplications larger than 1.0 megabase, regions with isolated absence of heterozygosity (AOH) greater than 5–15 megabases (dependent on chromosome involved, position, and likelihood of UPD-associated phenotype), and genome-wide AOH involving greater than 2% of the autosomal DNA complement are generally reported. However, smaller changes with pathogenic potential will also be reported, while larger changes that are well-documented benign variants will not be reported. Copy number changes resulting in carrier status for autosomal recessive disorders may not be reported unless a concern for a specific disorder is communicated to the laboratory.

Additional Information

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An online research opportunity called GenomeConnect (genomeconnect.org) is available for the recipient of this genetic test. This patient registry collects de-identified genetic and health information to advance knowledge of genetic variants.

Released By

MCR

Erik C. Thorland, Ph.D.

Received: 26 Aug 2024 09:53

Reported: 26 Aug 2024 10:27

Laboratory Notes

- 1 This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the U.S. Food and Drug Administration.

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