Patient ID	Patient Name		Birth Date	Sex	Age
SA00170055	TESTINGRNV, SAMPLEREPORT-CMAP		1982-07-01	F	42
Order Number	Client Order Number	Ordering Physician	Report Notes		
SA00170055	SA00170055	CLIENT,CLIENT			
Account Information		Collected			
C7028846 DLMP Rochester		25 Aug 2024 09:41			

Chromosomal Microarray, Prenatal

Result Summary

MCR

22q11.2 microdeletion syndrome (DiGeorge/Velocardiofacial syndrome)

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 69 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:
PRODH, DGCR5, DGCR9, DGCR10, DGCR2, DGCR11, DGCR14, TSSK2, GSC2, LINC01311, SLC25A1, CLTCL1, HIRA, MRPL40, C22orf39, UFD1L, CDC45, CLDN5, LINC00895, SEPT5, SEPT5-GP1BB, GP1BB, TBX1, GNB1L, C22orf29, TXNRD2, COMT, MIR4761, ARVCF, TANGO2, MIR185, DGCR8, MIR3618, MIR1306, TRMT2A, RANBP1, MIR6816, ZDHHC8, CCDC188, LOC284865, LINC00896, RTN4R, MIR1286, DGCR6L, TMEM191B, PI4KAP1, RIMBP3, ZNF74, SCARF2, KLHL22, MED15, POM121L4P, TMEM191A, PI4KA, SERPIND1, SNAP29, CRKL, LOC101928891, AIFM3, LZTR1, THAP7, THAP7-AS1, TUBA3FP, P2RX6, SLC7A4, MIR649, P2RX6P, LRRC74B, BCRP2

Maternal cell contamination studies were also performed, and no evidence of maternal DNA was detected in this amniotic fluid specimen.

If additional cell cultures have not already been ordered, cultures from this specimen will be discarded 10 days after all cytogenetic test results have been reported. If further testing is desired, contact Mayo Clinic Laboratories at 800–533–1710.

This assay does not rule out balanced chromosome abnormalities, imbalances of chromosomal regions not represented by probes on the

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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array, or mosaicism. A normal result does not exclude the diagnosis of any of the disorders tested for on this array and does not guarantee a healthy pregnancy or outcome.

Reason for Referral

MCR

abnormal AFP, Tetralogy of Fallot

Specimen

MCR

Amniotic Fluid

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 1.0 megabase, duplications larger than 2.0 megabases, regions with isolated absence of heterozygosity (AOH) greater than 5 megabases (terminal) and 10 megabases (interstitial) on UPD- associated chromosomes, and genome-wide AOH involving greater than 5% 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. New York State mandates follow-up for all prenatal diagnostic results. Contact the laboratory at 800–533–1710 with any questions.

Released By

MCR

Erik C. Thorland, Ph.D.

Received: 26 Aug 2024 09:55 **Reported:** 26 Aug 2024 11:31

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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