



Patient ID SA00170057	Patient Name TESTINGRNA, SAMPLEREPORT-CMAPC	Birth Date 1981-09-28	Sex F	Age 42
Order Number SA00170057	Client Order Number SA00170057	Ordering Physician CLIENT,CLIENT	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 25 Aug 2024 09:45		

Chromosomal Microarray, POC

Result Summary

MCR

Trisomy 13

Nomenclature

MCR

arr(13)x3

Sex:
XY

Interpretation

1 MCR

A chromosomal microarray profile consistent with trisomy 13 was observed. Autosomal trisomy accounts for approximately 30% of all miscarriages.

For any future pregnancy, the recurrence risk of a trisomic conception may be slightly increased compared to the usual maternal age-associated risk (Warburton et al., Am J Hum Genet. 75:376–385, 2004; De Souza et al., Am J Med Genet. 149A:2716–2722, 2009).

A genetic consultation may be of benefit.

Maternal cell contamination studies were also performed, and no evidence of maternal DNA was detected in this products of conception 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 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

miscarriage

Specimen

MCR

Products of Conception

Source

MCR

Fetal Tissue

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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Method

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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 09:56**Reported:** 26 Aug 2024 11:36**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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