



Patient ID SA00128199	Patient Name TESTINGRNA, SAMPLEREPORT-CMAMT	Birth Date 1978-05-02	Gender F	Age 41
Order Number SA00128199	Client Order Number SA00128199	Ordering Physician CLIENT,CLIENT	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 06 Apr 2020 14:50		

Chromosomal Microarray, POC, FFPE

Result Summary

MCR

Trisomy 18

Nomenclature

MCR

arr(18)x3

Sex:
Female

Interpretation

1 MCR

A chromosomal microarray profile consistent with trisomy 18 was observed. Autosomal trisomy accounts for approximately 30% of all miscarriages.

This test does not exclude an unbalanced translocation. Parental chromosome studies, test CHRCB (Chromosomes, Congenital, Blood), are suggested to determine whether either parent carries a balanced rearrangement associated with increased recurrence 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 (MCC) studies were not performed because a peripheral blood specimen from the mother was not available for testing. Maternal cell contamination studies, test PPAP (Parental Prenatal Array Prep Test), will be performed if an additional maternal specimen collected in EDTA is provided, however, MCC is unlikely to impact this abnormal result.

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

miscarriage

Specimen

MCR

Tissue, Slides

Source

MCR

Chorionic villi

Performing Site Legend

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



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Tissue ID

MCR

20-ABC123

Method

MCR

Chromosomal microarray (CMA) analysis was performed using molecular inversion probes on a whole-genome array (Applied Biosystems (Affymetrix) OncoScan platform; approximately 328,000 probes in triplicate) using in silico controls. The genome-wide functional resolution of this array is approximately 500 kilobases for non-mosaic deletions and non-mosaic duplications. However, functional resolution of this array varies significantly dependent upon size of the abnormality, probe density in region, percentage of abnormal cells, and quality of the DNA obtained. Mosaic abnormalities, especially those that are represented in a minor fraction of the sample, may not be detected. All data was analyzed and reported using the February 2009 NCBI human genome build 37.1 (hg19). Deletions larger than 1.0 megabase, and duplications larger than 2.0 megabases 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. Regions with absence of heterozygosity (AOH) are generally not reported.

Released By

MCR

Erik C. Thorland, Ph.D.

Received: 07 Apr 2020 14:57

Reported: 07 Apr 2020 15:33

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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