Patient ID	Patient Name		Birth Date	Gender	Age
SA00126160	TESTINGRNV, SAMPLEREPORT-CMAPT		1979-08-08	F	40
Order Number	Client Order Number	Ordering Physician	Report Notes		
SA00126160	SA00126160	CLIENT,CLIENT			
Account Information		Collected			
C7028846 DLMP Rochester		06 Feb 2020 09:03			

Chromosomal Microarray, Tumor, FFPE

Result Summary

MCR

See Interpretation

Result

MCR

CHANGE(CN)	REGION	GENOME COORDINATES	SIZE (Mb)
Loss	3q11.2q29	chr3:96192382–197852564	101.7
Loss	4p16.3p14	chr4:69403–38395949	38.3
Loss	8q24.11q24.12	chr8:117999278–120440816	2.4
Loss	8q24.21q24.3	chr8:131213569–146292734	15.1
Gain	10p15.3p11.1	chr10:126069–39146676	39.0
Loss	13q12.11q12.2	chr13:23092669–28654152	5.6
Loss	13q13.1q31.1	chr13:32447713–85350192	52.9
Gain	13q31.1q32.2	chr13:85382135–98308641	12.9
Gain	13q32.2q34	chr13:98317122–115103150	16.8
Loss	16q11.2q24.3	chr16:46461308–90158005	43.7
cnLOH	17p13.3p13.1	chr17:400958–10025298	9.6
Loss	Xq12	chrX:66767001–67677253	0.910
Loss	Xq12q13.3	chrX:67698615–75655848	8.0
Loss(x0)	Xq13.3q21.1	chrX:75672234–77361669	1.7
Loss	Xq21.1q28	chrX:77369299–155219364	77.9

Nomenclature

MCR

arr(1–22,XX)cx

Interpretation

1 MCR

The result is abnormal. A complex molecular karyotype was observed, including homozygous loss of Xq13.3q21.1 (including ATRX), gain of 10p15.3p11.1, loss of 13q13.1q31.1 (including RB1), and copy-neutral loss of heterozygosity (cnLOH) of 17p13.3p13.1 (including TP53). These abnormalities are consistent with the diagnosis of an IDH-mutant astrocytic glioma and mutations of TP53 and ATRX have been reported in these gliomas (Eckel-Passow et al. N Engl J Med 372:2499–2508;2015, The Cancer Genome Atlas Network. N Engl J Med 372:2481–2498;2015, Suzuki et al. Nat Genet 47:458–468;2015, Bai et al. Nature Genet 48:59;2016, Ceccarelli et al. Cell 164:550–563;2016).

The tumor also exhibited multiple other copy number alterations consistent with IDH-mutant astrocytic glioma, including loss of 3q11.2q29, loss of 4p16.3p14, loss of 8q24.11q24.12 (including EXT1), loss of 8q24.21q24.3, and gain of 13q32.2q34 (including TNFSF13B). See 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	William G. Morice M.D. Ph.D	24D0404292

Patient ID	Patient Name		Birth Date	Gender	Age
SA00126160	TESTINGRNV, SAMPLEREPORT-CMAPT		1979-08-08	F	40
Order Number	Client Order Number	Ordering Physician	Report Notes		
SA00126160	SA00126160	CLIENT,CLIENT			
Account Information		Collected			
C7028846 DLMP Rochester		06 Feb 2020 09:03			

Result section for a complete list of reported abnormalities.

Of note, 1p and 19q whole arm co-deletion was not observed.

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

Reason for Referral MCR

glioma

Specimen MCR

Tissue, Paraffin

Source MCR

Brain

Tissue ID MCR

SP-17–3984-H6

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 clonal abnormalities, especially those that are represented in a minor fraction of the sample, may not be detected. Deletions larger than 1.0 megabase, duplications larger than 2.0 megabases, and copy neutral loss of heterozygosity (cnLOH) larger than 10 megabases are generally reported. However, complex genomic alterations may be reported in aggregate, and well documented pathogenic constitutional and/or acquired abnormalities of any size may also be reported. All data were analyzed and reported using the February 2009 NCBI human genome build 37.1 (hg19). The genome coordinates described are best estimates and may not represent precise breakpoints, especially for abnormalities detected in a low percentage of cells.

Released By MCR

William R. Sukov, M.D.

Received: 07 Feb 2020 09:04 **Reported:** 11 Feb 2020 07:58

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.

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