Patient Name	Patient ID	Age	Gender	Order #
TESTINGRNV,CMAC	SA00052691	12	F	SA00052691
Ordering Phys CLIENT,CLIENT				**DOB** 01/01/2001
Client Order # SA00052691	**Account Information**			**Report Notes**
Collected 01/11/2013 08:00	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 01/24/2013 13:38				

Test	Flag	Results	Unit	Reference Value	Perform Site*
Chromosomal Microarray, Blood			REPORTED 01/16/2013 08:03		
Specimen		Blood			MCR
Specimen ID		1060293			MCR
Order Date		16 Jan 2013 07:44			MCR
Reason For Referral		autism			MCR
Method					MCR

Chromosomal microarray (CMA) analysis was performed using both copy number and single-nucleotide polymorphism (SNP) probes on a whole-genome array (Affymetrix CytoScan HD platform). The genome-wide functional resolution of this array is approximately 30 kilobases for deletions and 60 kilobases for duplications, with higher resolution in targeted genes and regions of known clinical significance. 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 500 kilobases, and interstitial absence of heterozygosity (AOH) larger than 10 megabases and terminal AOH larger than 5 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.

Results MCR

arr 22q11.21(17,041,724 19,835,417)x1 [hg19]

Interpretation MCR

The result is abnormal. An interstitial deletion of 250 oligonucleotide probes at 22q11.21, spanning approximately 2.8 megabases, was observed. Metaphase FISH studies using a probe within the deleted interval (HIRA, Abbott Molecular) confirmed this deletion. This deletion is consistent with a diagnosis of 22q11.2 deletion syndrome. Approximately 7% of these deletions are inherited and there is significant phenotypic variability. Parental FISH studies (CAT22, Test #81129) are suggested to determine if this deletion was inherited or de novo and may help establish recurrence risks. A genetic consultation may be of benefit. Manji et al., Genet Med 3:65 66, 2001; Kobrynski and Sullivan, Lancet 370:1443 1452, 2007.

DISCLAIMER: This test was developed and its performance characteristics determined by Laboratory Medicine and

Performing Site Legend on Last Page of Report

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Pathology, Mayo Clinic, Rochester, MN. It has not been cleared or approved by the U.S. Food and Drug Administration. 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.

As a participant in the ISCA (International Standards for Cytogenomic Arrays) Consortium, Mayo Clinic Cytogenetics Laboratory contributes submitted clinical information and test results for molecular cytogenetics tests to a HIPAA-compliant, de-identified public database hosted by the National Institute of Health. This is an international effort to improve diagnostic testing and our understanding of the relationships between genetic changes and clinical symptoms (for information about the database visit the consortium website at http://www.iscaconsortium.org). Confidentiality of each sample is maintained. Patients may request to opt-out of this scientific effort by: 1) checking the box below and faxing the report back to the laboratory, 2) calling the laboratory at 1-800-533-1710, extension 8-2952 and asking to speak with a laboratory genetic counselor. Please call with any questions.

[ ] Refusal for inclusion in these efforts may be indicated by checking this box and faxing the report to: 1-877-749-3332 (507-284-0043) Cytogenetics Laboratory, ATTN: Genetic Counselors Opt-Out

Consultant	Christina M Radtke	MCR
Released Date	16 Jan 2013 08:01	MCR

* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director: Franklin R. Cockerill, III, M.D.