Physician:
Physician NOT IN DATABASE,
BOX 356100

TEST, CASE
CASE #: TS-16-00035

Sex: F, DOB: 1/1/1980
Collected Date: 11/10/2016
Received Date: 11/10/2016
ASN/CSN:

Cytogenetics & Genomics Laboratory
University of Washington Medical Center
1959 NE Pacific St., NW 125, BOX 356100
Seattle, WA 98195-6100
Voice: (206) 598-4488
Fax: (206) 598-2610

Patient Name on specimen tested: Test, Case

Date Obtained: 11/10/2016 **Age:** 36 **Sex:** Female **Sample Type:** Molecular

Date Received: 11/10/2016 **Ref Phys:** Smith, MD, Jane M **Source:** Blood

Report Date: 11/18/2016 **Ref Fac:** City Medical Center **Test Type:** SNP Array

Report Status: Final **Other Phys:** Jones, MS, CGC, Carla

Clinical Indication: Autism spectrum disorder

ISCN Diagnosis: arr[GRCh37] (1-22,X)x2

Summary: Female, no clinically significant constitutional abnormalities detected by cytogenomic SNP microarray analysis.

Diagnosis and Comments: No clinically significant unbalanced constitutional genomic variants were detected in this peripheral blood sample by cytogenomic SNP microarray analysis, including deletions, duplications, and regions of copy number neutral absence of heterozygosity.

Benign genomic variants were not reported.

Recommendations:
1. Cytogenomic microarray results should be interpreted in the context of the patient's clinical and family history.

2. Genetic counseling can be helpful to patients in understanding their test results.

References:
UCSC genome browser: http://genome.ucsc.edu/
CCS SNP Array Evaluation Tool: http://firefly.ccs.miami.edu/cgi-bin/ROH/ROH_analysis_tool.cgi
DGV: http://dgv.tcag.ca/dgv/app/home
DECIPHER: https://decipher.sanger.ac.uk/
OMIM: http://omim.org/
dbVar: http://www.ncbi.nlm.nih.gov/dbvar/browse/
ClinGen: http://www.ncbi.nlm.nih.gov/projects/dbvar/clingen/
ExAC: http://exac.broadinstitute.org/

Test Limitations: The Illumina Infinium CytoSNP-850K BeadChip is used in this test for the sole purpose of identifying genomic chromosomal abnormalities. This microarray will detect aneuploidy as well as copy number gains (duplications or amplifications), copy number losses (deletions), and

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regions of copy number neutral absence or loss of heterozygosity (cnAOH or cnLOH) for the loci represented on the microarray. Analysis in our laboratory is limited to detecting copy number changes (deletions and duplications) that include at least 10 probes and regions of cnAOH that include at least 500 probes. We cannot exclude abnormalities below these levels of resolution. UPD is reported when telomeric regions of cnAOH are >5Mb in length and interstitial regions of cnAOH are >15 Mb in length. Possible identity by descent is reported when regions of cnAOH > 3 Mb comprise ≥1.5% of the autosomal genome. Deletions and duplications of ≥400 kb are reported, even if clinical significance is unclear, as per provider request. Smaller deletions or duplications in regions of known microdeletion/microduplication syndromes or in clinically relevant genes will also be reported. Benign genomic variants are not reported. For neoplasia arrays, likely constitutional variants of unclear clinical significance are not reported.

Cytogenomic microarray will not detect imbalances in the regions not represented on the microarray, low-level mosaicism (<20%), tetraploidy, balanced alterations (e. g. reciprocal translocations, Robertsonian translocations, inversions, balanced insertions), methylation anomalies and other epigenetic events, or point mutations that may be responsible for the clinical phenotype. The failure to detect an alteration at any locus does not exclude the diagnosis of any of the disorders represented on the microarray. The laboratory can assist the ordering provider in determining whether other types of testing, such as DNA sequencing for point mutations, are appropriate. This discussion should be considered in the context of the clinical phenotype.

The performance characteristics of this test have been validated by University of Washington Medicine Cytogenetics and Genomics Laboratory as required by CLIA '88 regulations. It has not been cleared or approved for specific uses by the U.S. Food and Drug Administration. Pursuant to the requirements of CLIA '88, this laboratory has established and verified the test's accuracy and precision. The laboratory is regulated under CLIA as qualified to perform high-complexity testing. This test is used for clinical purposes. It should not be regarded as investigational or for research.

Methods Summary: Genomic DNA extracted from the patient sample was amplified, fragmented, and hybridized to the Illumina Infinium CytoSNP-850K BeadChip (Chip ID: 123456789123_R01C01). This microarray contains 850,000 probes for SNP markers. Additional details of this microarray can be found at: http://www.illumina.com/content/dam/illumina-marketing/documents/products/datasheets/datasheet_CytoSNP850K_POP.pdf
After hybridization, the microarray was washed, labeled, stained, and scanned with an Illumina iScan. Allele and intensity ratio data of the fluorescent signals were generated. Microarray data were visualized and analyzed using Illumina BlueFuse Multi 4.2 to identify chromosomal copy number variants and regions of copy number neutral absence or loss of heterozygosity.

Procedures Performed:

81229 ZB149 Array CGH, Constitutional
G0452 Cytogenetics, Molecular, Complex Interpretation by an MD

Lab Director, MD
Cytogeneticist
Electronically signed 11/18/2016

In compliance with CMS regulations, the pathologist's signature on this report indicates that the case has been personally reviewed, and the diagnosis made or confirmed by the Attending Pathologist. Microscopic examination was used to arrive at the diagnosis unless indicated otherwise.

Kathleen Leppig MD
Pathologist
Electronically signed 11/10/2016

In compliance with CMS regulations, the pathologist's signature on this report indicates that the case has been personally reviewed, and the diagnosis made or confirmed by, the Attending Pathologist. Microscopic examination was used to arrive at the diagnosis unless indicated otherwise.