



LABORATORY REPORT

Example Client, XYZ123
1234 Warde Road
Ann Arbor MI 48108

EXAMPLE, REPORT W
WX0000003827 M 07/08/1978 45 Y

Referral Testing

Collected: 09/18/2023 09:43 Received: 09/18/2023 09:43

Test Name Result Flag Ref-Ranges Units Site

Cytogenetics

Microarray CGH SEE BELOW QARL

ClariSure(R) Oligo-SNP Postnatal Chromosome Microarray

Specimen Type: Blood
Clinical Indication: Autistic Disorder

Result:

NORMAL MALE MICROARRAY RESULT

Interpretation:
No reportable copy number variants or regions of homozygosity were detected.

Recommendations:
Correlation with clinical findings and other laboratory results is recommended.

For more information, healthcare providers may call Quest Genomics Client Services at 866-GENEINFO (866-436-3463).

Nomenclature:
arr(X,Y)x1,(1-22)x2

Assay Information:
Method: Oligonucleotide-SNP (Affymetrix)
Resolution: 1.15 kb
Number of Probes: 2.67 million
Genome Assembly: GRCh37/hg 19 (Feb. 2009)

Benign copy number variations (CNVs) are relatively common; therefore, CNVs identified as known benign variants in the general population in publicly available databases (example: DGV; http://dgv.tcag.ca) will not be reported. Copy number alterations involving non-coding regions of the genome may not be reported. The performance of this test for detection of mosaicism has not been established. While this assay is validated and should have the capacity to detect a broad spectrum of specific disorders/syndromes, including aneuploidy and sub-telomeric deletions, not all of these diseases have been detected in our laboratory due to their rarity. This assay will only detect CNVs for genomic sequences that are represented on the array. This assay does not detect single nucleotide variants (SNVs) or insertions/deletions (indels) not covered by the platform, gains and losses below the level of resolution of the platform, balanced rearrangements (e.g.,

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translocations, inversions), or most epigenetic events. Only regions of allelic homozygosity (ROH) larger than our reporting threshold will be reported.

Any clinical concern for recessive disorders must be communicated by the ordering clinician for appropriate consideration. To investigate a hemizygous deletion for a specific autosomal recessive gene (disorder), please contact the Quest Diagnostics Genomic Client Services at the number below. Otherwise, carrier status will not be disclosed unless it is deemed appropriate by the director.

Limitation of variant analysis: The classification and interpretation of the variant(s) identified reflect the current state of our understanding at the time of this report. Variant classification and interpretation are subject to professional judgment, and may change for a variety of reasons, including but not limited to, updates in classification guidelines and availability of additional scientific and clinical information. This test result should be used in conjunction with the health care provider's clinical evaluation. Since the current classification of a CNV may change in the future, surveillance of the medical literature is strongly recommended prior to making any clinical decisions.

For questions regarding this testing and variant classification updates, please call Quest Diagnostics Genomics Client Services at 866-GENEINFO.

The oligo-SNP (oligonucleotide, single nucleotide polymorphism, Affymetrix CytoScan HD) assay uses a microarray containing over 2.67 million probes, including 1.9 million copy number probes and 750 thousand SNP probes. The overall average inter-probe distance is 1,150 base pairs. Settings for genome-wide screening are >200 kb for gains, >50 kb for losses, and a total of >10 Mb for autosomal ROH that are >5 Mb. These may be lower in cytogenetic relevant regions. For focused analysis of a particular region or segment please contact the laboratory.

This test was developed and its analytical performance characteristics have been determined by Quest Diagnostics Marlborough, MA. It has not been cleared or approved by FDA. This assay has been validated pursuant to the CLIA regulations and is used for clinical purposes.

Electronic Signature:
Patricia Minehart Miron, Ph.D., FACMG

Technical Services Performed At: Quest Diagnostics Marlborough, 200 Forest Street, Marlborough, MA, 01752.
Reading Location: Quest Diagnostics Marlborough, 200 Forest Street,

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Marlborough, MA, 01752.

CPT CODES
81229

The CPT codes provided are for information purposes only, and are based
on AMA guidelines without regard to specific payor requirements.
END OF REPORT
FINAL REPORT-ACC FINAL
AmeriPath Northeast, 1 Greenwich Place, Shelton, CT 06484. P(866)436-9632.
F(203)929-2344. Medical Director: Kamraan Z. Gill, M.D. CLIA
07D1035411, CT CL-0645

Reported Date: 09/18/2023 09:44 CGH

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