John Doe
Sex Assigned at Birth: Male
Date of Birth: 01/01/2001
Sample ID: SM0004644
Sample Type: DATA
Collection Date: 05/23/2022
Received Date: 05/25/2022

Referring Lab: External
Phone: +1-555-555-5555
CLIA #: 05D1283911
CLIA #: 45D2013111

Requisition ID:
RQ-0000059

Report Number:
RPstg117

Report Date:
03/19/2025

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https://myome.com/r/?id=RPstg117

Test: Copy number analysis using whole genome sequencing

Indication for testing: Abnormal facial shape, Asymmetry of the position of the ears, Autism, Autistic behavior, Depressed nasal bridge, Hypertelorism, Plagiocephaly, Seizure

Positive	Pathogenic chr15q11.2 deletion
Positive	Pathogenic chr6q26 duplication

EVENT	CHROMOSOME REGION	CYTOBAND	SIZE (MB)	NUMBER OF GENES*	CLASSIFICATION
COPY NUMBER LOSS	chr15:22748502-23372000	chr15q11.2	0.62	6	Pathogenic
COPY NUMBER GAIN	chr6:162475207-163148693	chr6q26	0.67	6	Pathogenic

* Defined as gene-coding regions

ISCN Nomenclature: seq[GRCh37]15q11.2(22748502_23372000)x1, seq[GRCh37]6q26(162475207_163148693)x**INSERT_COPY_NUMBER_HERE**

INTERPRETATION

A pathogenic variant was identified on chromosome 15.

The individual harbors one copy (heterozygous) of a partial deletion of the chr15q11.2 region, which may be associated with 15q11.2 BP1–BP2 microdeletion syndrome. Germline pathogenic variations involving partial deletion of the chr15q11.2 region have been shown to be associated with 15q11.2 BP1–BP2 microdeletion syndrome, which manifests as developmental and language delay, neurobehavioral disturbances and psychiatric problems [GeneReviews. PMID: 20301323], [PubMed:25689425, PubMed:28387067]. 15q11.2 BP1–BP2 microdeletion syndrome, is inherited in an autosomal dominant manner, which means one copy of the altered gene in an individual are required for disease manifestation. However, incomplete penetrance and phenotypic variability, have also been reported in this region [PubMed:25689425].

MyOme, Inc.
1455 Adams Drive, #1150
Menlo Park CA 94025
Email: clinical@myome.com
Phone: +1-628-265-3818

Lab Director:
Brynn Levy M.Sc.(Med)., Ph.D., FACMG
CLIA #05D2203070 CAP #8939502

A pathogenic variant was identified on chromosome 6.

The individual harbors one copy (heterozygous) of a 'pathogenic' variant in the PRKN gene, which has been shown to be associated with parkin type of early-onset Parkinson disease (PARK-Parkin). Germline pathogenic variations in the PRKN gene have been shown to be associated with PARK-Parkin, which manifests as early onset Parkinson's disease (PD), characterized by bradykinesia, resting tremor, and rigidity [PubMed:20301651], [MedlinePlus]. PARK-Parkin, caused due to pathogenic variations in the PRKN gene, is inherited in an autosomal recessive manner, which means two copies of the altered gene in an individual are required for disease manifestation

NEXT STEPS

- These results should be interpreted in the context of this individual's clinical findings, family history, and other laboratory data. All genetic tests have limitations; please see limitations and other information for this test below.
- If this individual's clinical symptoms are not fully explained by the findings in this report additional diagnostic testing, such as whole exome sequencing or panel testing, may provide relevant clinical insight.
- Genetic counseling is recommended to discuss the significance of these results.
- Parental copy number analysis is recommended to determine whether the copy number change is inherited or a new event (*de novo*).

TEST METHODS

- Specimen receipt, accessioning, data analysis, and interpretation is performed by MyOme Inc., 1455 Adams Drive, Suite 1150, Menlo Park, CA 94025, CLIA# 05D2203070. Whole Genome Sequencing, excluding data analysis and interpretation, is performed by Broad Clinical Labs LLC, 27 Blue Sky Dr, Burlington, MA 01803, CLIA#22D2055652.
- Genomic DNA obtained from submitted samples was sequenced using Illumina technology. Reads were aligned to the NCBI GRCh37.p13 reference assembly
- Variants are interpreted and reported based on the standards and guidelines set forth by the American College of Medical Genetics and Genomics (ACMG). Classification categories include pathogenic (P), likely pathogenic (LP), variants of unknown significance (VUS), likely benign (LB) and benign (B). Reported variants only include those which are classified as P, LP, or VUS and overlap with the tested individual's indication for testing.
- All reported variants are confirmed by a secondary technology which may include arrays, MLPA, FISH, or PCR, depending on the nature of the copy number variant.

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TEST LIMITATIONS

- This test is designed to detect clinically relevant copy number variants (CNVs) ≥ 50 kilobases. Currently variants cannot be reliably called in certain regions not amenable to NGS and are not analyzed. This includes certain regions of high similarity on chromosome X and chromosome Y. CNVs in the following regions may not be detected for male patients (chrX: 60000-2699520; chrX:154930289-155260560; chrY:10000-2649520; chrY:59033286-59363566). CNVs involving pseudogenes may not be reliably detected or reported.
- Small pathogenic CNVs within the exon, some small intragenic deletions or duplications, as well as complex rearrangements may not be detected. This assay cannot discern between CNVs that are high copy number gains ≥4X.
- This test does not interrogate mitochondrial DNA.
- This analysis does not detect tandem repeats, balanced alterations (reciprocal translocations, Robertsonian translocations, inversions, and balanced insertions), point mutations, methylation abnormalities, genomic imbalances in segmentally duplicated regions and mosaicism; possible cases of mosaicism may be investigated at the discretion of the laboratory director. Areas of homozygosity (AOH) are not reported.
- A history of stem cell or bone marrow transplantation, or recent blood transfusion may impact the accuracy of the results.
- Like most tests, this test carries a risk of false negative or false positive results, which may be caused by, without limitation, sample contamination from biological or non-biological sources, specimen marking issues, rare genetic variants interfering with analysis, and other technical issues and limitations.

DISCLAIMERS

- This test was developed, and its performance characteristics were determined, by MyOme, Inc., a clinical laboratory certified under the Clinical Laboratory Improvement Amendments of 1988 (CLIA) and College of American Pathologist (CAP) accredited to perform high complexity clinical laboratory testing. This test has not been cleared or approved by the U.S. Food and Drug Administration (FDA).
- This test is a diagnostic test for copy number gains and losses ≥50 kilobases. This test provides no information for genomic variants smaller than 50 kilobases and it does not examine balanced rearrangements.
- Like most tests, this test carries a risk of false negative or false positive results. Testing is unavailable for samples damaged by human error, lost/destroyed due to weather, transit issues or other problems beyond the control of MyOme. Test results should always be interpreted by a clinician in the context of clinical and familial data with the availability of genetic counseling when appropriate. MyOme is not responsible for the content of third-party websites referenced in this report.
- The interpretation of CNVs is based on our current understanding of the genome. These interpretations may change over time as more information about these alterations becomes available. Possible diagnostic errors include CNV call errors, sample misidentification, and other sources.

REVIEWED BY

03/19/2025

Date

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