Client: ARUP Example Report Only
500 Chipeta Way
Salt Lake City, UT 84108
UNITED STATES

Physician: TEST,

## Patient: NEG EXAMPLE, RWGS REA

**DOB**

| | |
|---|---|
| **Sex:** | Female |
| **Patient Identifiers:** | 49771 |
| **Visit Number (FIN):** | 50126 |
| **Collection Date:** | 6/12/2023 07:40 |

## Whole Genome Reanalysis

ARUP test code 3005939

RWGS REA Int

Negative

TEST PERFORMED
Genome reanalysis was performed using the original rapid whole genome sequencing data from report date 08/05/2022, the current bioinformatics pipeline, updated population frequency data, and any new information provided about the patient's clinical findings. The overall result remains unchanged.

RESULT
Primary findings: Negative
Secondary findings: Negative

KEY CLINICAL FINDINGS
Abnormality of eye movement, abnormal cerebellar vermis morphology, abnormal head movements, cerebellar dysplasia, neurodevelopmental delay.

HPO terms used:
HP:0000496 (abnormality of eye movement), HP:0002334 (abnormal cerebellar vermis morphology), HP:0002457 (abnormal head movements), HP:0007033 (cerebellar dysplasia), HP:0012758 (neurodevelopmental delay).

INTERPRETATION
No variants were identified that are predicted to be causative for the patient's phenotype.

No secondary pathogenic variants were detected in the v3.1 list of genes that the American College of Medical Genetics and Genomics (ACMG) recommends reporting in all individuals undergoing genome sequencing (Miller, 2022). A list of ACMG genes is included in the additional technical information. These genes are evaluated only to the extent that standard genome sequencing allows. Single pathogenic variants in autosomal recessive ACMG genes are not reported.

RECOMMENDATIONS
Medical management and screening should rely on clinical findings. Genetic consultation is recommended. If after one year from report date clinical suspicion remains high for a genetic etiology, a reanalysis may be ordered, for a fee, though ARUP using these original sequencing data (Rapid Whole Genome Reanalysis, ARUP test 3005939).

REFERENCES
Miller DT, et al. ACMG SF v3.1 list for reporting of secondary findings in clinical exome and genome sequencing: a policy statement of the American College of Medical Genetics and Genomics (ACMG). Genet Med. 2022;24(7):1407-1414. PMID: 35802134.

H=High, L=Low, *=Abnormal, C=Critical

BACKGROUND INFORMATION: Whole Genome Reanalysis (Originally Tested at ARUP - No Specimen Required)
CHARACTERISTICS: Genome reanalysis may be performed when a previous genome analysis fails to determine the etiology for a suspected genetic condition. Rapid progress in the understanding of gene-disease relationships, in addition to improvements in variant-calling pipelines, underscores the utility of performing a bioinformatic-restricted reanalysis.

CLINICAL SENSITIVITY: Varies based on clinical symptoms, family history, inheritance pattern, and previous clinical evaluations

METHODOLOGY: A FastQ file of massively parallel sequencing (MPS) data from the original genome test was processed through our current variant calling and annotation pipeline. If the original sample(s) was available, Sanger sequencing was performed as necessary to confirm reported variants. Human genome build 19 (Hg19) was used for data analysis.

LIMITATIONS OF ANALYSIS: A negative result does not exclude a genetic diagnosis. The human genome cannot be completely analyzed as some genes have not been identified while others, due to technical limitations, cannot be sequenced or interpreted. Variants in intergenic or deep intronic regions will only be evaluated if an effect on gene expression is predicted via annotation software. Regulatory region variants and deep intronic variants will not be identified. Mitochondrial DNA is not analyzed. Chromosomal phase of identified variants may not be determined. Deletions/duplications/insertions of any size may not be detected by MPS. Diagnostic errors can occur due to rare sequence variations. Variants may not be identified due to technical limitations in the presence of pseudogenes, repetitive, or homologous regions. This assay is not designed to detect low-level somatic variants associated with disease. Please see Additional Technical Information located at http://ltd.aruplab.com/Tests/Pub/3005939 for more information.

LIMITATIONS FOR REPORTING AND INTERPRETATION: Only variants in genes suspected to be causative of the patient's symptoms are reported, with the exception of secondary pathogenic findings, if elected. Incorrect reporting of biological relationships among family members may affect result interpretation. Mode of inheritance, reduced penetrance, and genetic heterogeneity could reduce clinical sensitivity. Interpretation of this test result may be impacted if this patient has had an allogeneic stem cell transplantation.

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the U.S. Food and Drug Administration. This test was performed in a CLIA-certified laboratory and is intended for clinical purposes.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.

VERIFIED/REPORTED DATES

| Procedure | Accession | Collected | Received | Verified/Reported |
|---|---|---|---|---|
| RWGS REA Int | 23-163-100685 | 6/12/2023 7:40:00 AM | 6/12/2023 7:41:03 AM | 6/12/2023 7:43:00 AM |

**H=High, L=Low, *=Abnormal, C=Critical**

END OF CHART

H=High, L=Low, *=Abnormal, C=Critical