Client: Example Client ABC123
123 Test Drive
Salt Lake City, UT 84108
UNITED STATES

Physician: Doctor, Example

Patient: Patient, Example

DOB	1/24/1998
Gender:	Female
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	00/00/0000 00:00

Rett Syndrome (MECP2), Sequencing and Deletion/Duplication

ARUP test code 0051614

Rett Syndrome (MECP2) Seq, DelDup Spcm	Whole Blood
Rett Syndrome (MECP2) Seq, DelDup Int	Negative

Section 79-1 of New York State Civil Rights Law requires informed consent be obtained from patients (or their legal guardians) prior to pursuing genetic testing. These forms must be kept on file by the ordering physician. Consent forms for genetic testing are available at www.aruplab.com. Incidental findings are not reported unless clinically significant but are available upon request.

TEST PERFORMED - 0051614

TEST DESCRIPTION - Rett Syndrome (MECP2), Sequencing and Deletion/Duplication

INDICATION FOR TEST - Confirm Diagnosis

RESULT

No pathogenic variants were detected in the MECP2 gene.

INTERPRETATION

No pathogenic MECP2 gene variants were detected in the coding region or intron/exon boundaries by sequencing and deletion/duplication analysis. This greatly reduces, but does not eliminate, the possibility of Rett syndrome/neonatal encephalopathy as this test does not detect all pathogenic MECP2 variants (e.g., deep intronic or regulatory region variants).

RECOMMENDATIONS

Medical management of this individual should rely on clinical findings and family history. Genetic consultation is recommended.

COMMENTS

Reference Sequences: GenBank # NM_001110792.1 (MECP2 exon 1) and NM_004992.3 (MECP2 exon 2-4)
Nucleotide numbering begins at the "A" of the ATG initiation codon.

This result has been reviewed and approved by [redacted]

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

BACKGROUND INFORMATION: Rett Syndrome (MECP2), Sequencing and Deletion/Duplication

CHARACTERISTICS: Classic Rett syndrome is a progressive neurodevelopmental disorder characterized by normal development until 6-18 months of age followed by rapid developmental regression, deceleration of head growth, loss of speech and acquired motor skills, and seizures; purposeful use of the hands is replaced by repetitive stereotyped hand movements. MECP2-Related disorders include Rett-like syndrome, severe congenital encephalopathy, or mild to severe mental retardation.
INCIDENCE: 1 in 10,000.
INHERITANCE: X-linked dominant; most cases are sporadic.
CAUSE: Methyl-CpG-Binding Protein 2 (MECP2) gene mutations.
CLINICAL SENSITIVITY: Up to 95 percent.
METHODOLOGY: Bidirectional sequencing of the MECP2 coding regions (exons 1-4) and intron-exon boundaries; Multiplex Ligation-dependent Probe Amplification (MLPA) to detect large deletions/duplications in the MECP2 coding regions (exons 1-4).
ANALYTICAL SENSITIVITY: 99 percent for sequencing and 90 percent for MLPA.
ANALYTICAL SPECIFICITY: 99 percent for sequencing and 98 percent for
MLPA
LIMITATIONS: Breakpoints of large deletions/duplications cannot be determined; deep intronic mutations will not be detected. Diagnostics errors can occur due to rare sequence variations.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online at www.aruplab.com.

Test developed and characteristics determined by ARUP Laboratories. See Compliance Statement C: aruplab.com/CS

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Rett Syndrome (MECP2) Seq, DelDup Spcm	20-272-145393	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Rett Syndrome (MECP2) Seq, DelDup Int	20-272-145393	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00

END OF CHART

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