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

Physician: Doctor, Example

Patient: Patient, Example

DOB	[redacted]
Gender:	Female
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	00/00/0000 00:00

Rett Syndrome (MECP2), Full Gene Sequencing

ARUP test code 0051378

RETT FGS Specimen	Whole Blood

Rett Syndrome, Full Gene Sequencing

Negative

TEST PERFORMED - 0051378
TEST DESCRIPTION - Rett Syndrome (MECP2), Sequencing
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. This reduces, but does not eliminate, the possibility of Rett syndrome/neonatal encephalopathy as this assay does not detect all pathogenic MECP2 variants (e.g., large deletions/duplications, deep intronic variants or regulatory region variants).

RECOMMENDATIONS
Medical management should rely on clinical findings and family history. As not all pathogenic MECP2 variants are detectable by sequencing, MECP2 deletion/duplication analysis should be considered if clinical features of Rett syndrome are present (Rett Syndrome (MECP2), Deletion and Duplication; ARUP test code 0051618). A genetics 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.
Likely benign and benign variants are not included in this report.

This result has been reviewed and approved by Pinar Bayrak-Toydemir, M.D., Ph.D.

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

BACKGROUND INFORMATION: Rett Syndrome (MECP2), Full Gene Sequencing

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: 80 percent
METHODOLOGY: Bidirectional sequencing of the MECP2 coding regions (exons 1-4) and intron-exon boundaries.
ANALYTICAL SENSITIVITY: 99 percent
ANALYTICAL SPECIFICITY: 99 percent
LIMITATIONS: Deep intronic mutations and large deletions/duplications will not be identified. Diagnostic errors can occur due to rare sequence variations.

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 FGS Specimen	19-364-401205	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Rett Syndrome, Full Gene Sequencing	19-364-401205	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