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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	11/21/1980
Gender:	Female
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	01/01/2017 12:34

Tay-Sachs Disease (HEXA), 7 Variants

ARUP test code 0051428

Tay-Sachs Disease (HEXA), Specimen	Whole Blood	
Tay-Sachs Disease (HEXA), Allele 1	c.745C>T	*
Tay-Sachs Disease (HEXA), Allele 2	Negative	
Tay-Sachs Disease (HEXA), Interpretation	See Note	

Indication for testing: Carrier screening or diagnostic testing for Tay-Sachs disease.

Benign variant: This sample is negative for the five pathogenic variants tested in the HEXA gene; however, one copy of the HEXA pseudodeficiency allele, p.R249W (c.745C>T), was detected. This benign variant is commonly found in individuals who have been identified as Tay-Sachs carriers by enzymatic activity. Because this variant allows full beta-hexosaminidase A (HEX A) enzyme expression in vivo, it is considered benign. If this individual is asymptomatic and of Ashkenazi Jewish descent, his/her risk of being a carrier of Tay-Sachs disease is reduced from 1 in 30 to approximately 1 in 480. For non-Ashkenazi Jewish individuals, the recommended test for Tay-Sachs carrier screening is beta-hexosaminidase A (HEX A) enzymatic activity in leukocytes using synthetic substrates, which detects 99 percent of Tay-Sachs carriers.

This result has been reviewed and approved by Rong Mao, M.D.

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

BACKGROUND INFORMATION: Tay-Sachs Disease (HEXA), 7 Variants

CHARACTERISTICS: Tay-Sachs disease is a lysosomal storage disease that, in the most severe childhood-onset form, leads to a loss of motor skills beginning at 3- to 6-months of age and progresses to blindness, seizures, total incapacitation, and eventual death by 4 years of age. Adult-onset Tay-Sachs is a milder disease with later onset and slower progression. In adults, Tay-Sachs disease is associated with variable neurological findings, including progressive dystonia, spinocerebellar degeneration, motor neuron disease, and bipolar form of psychosis.
INCIDENCE: 1 in 3000 Ashkenazi Jewish individuals.
INHERITANCE: Autosomal recessive.
CAUSE: HEXA gene pathogenic variants.
VARIANTS TESTED: Four pathogenic 7.6kb del, c.1073+1G>A, p.Y427Ifs (c.1274_1277dup TATC), c.1421+1G>C; one mild pathogenic p.G269S (c.805G>A); and two pseudodeficiency alleles p.R247W (c.739C>T) and p.R249W (c.745C>T).
CLINICAL SENSITIVITY: 94 percent in Ashkenazi Jewish individuals, 59 percent in other ethnicities.
METHODOLOGY: Polymerase chain reaction (PCR) and fluorescence monitoring.
ANALYTICAL SENSITIVITY AND SPECIFICITY: Greater than 99 percent.
LIMITATIONS: HEXA variants other than those specified above will not be detected. 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
Tay-Sachs Disease (HEXA), Specimen	18-117-138378	4/27/2018 10:20:00 AM	5/1/2018 4:55:56 PM	5/5/2018 7:14:00 AM
Tay-Sachs Disease (HEXA), Allele 1	18-117-138378	4/27/2018 10:20:00 AM	5/1/2018 4:55:56 PM	5/5/2018 7:14:00 AM
Tay-Sachs Disease (HEXA), Allele 2	18-117-138378	4/27/2018 10:20:00 AM	5/1/2018 4:55:56 PM	5/5/2018 7:14:00 AM
Tay-Sachs Disease (HEXA), Interpretation	18-117-138378	4/27/2018 10:20:00 AM	5/1/2018 4:55:56 PM	5/5/2018 7:14:00 AM

END OF CHART

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