ARUP LABORATORIES
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Jonathan R. Genzen, MD, PhD, Chief Medical Officer

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

Physician: Doctor, Example

Patient: Patient, Example

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

Tay-Sachs Disease (HEXA), 7 Variants

ARUP test code 0051428

Tay-Sachs Disease (HEXA), Specimen	Whole Blood
Tay-Sachs Disease (HEXA), Allele 1	c.739C>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.R247W (c.739C>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. If a diagnosis of Tay-Sachs disease is suspected, HEXA sequencing and deletion/duplication analysis (ARUP test code 3004486) should be considered.

This result has been reviewed and approved by [redacted]

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.

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US 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
Tay-Sachs Disease (HEXA), Specimen	24-033-113087	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Tay-Sachs Disease (HEXA), Allele 1	24-033-113087	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Tay-Sachs Disease (HEXA), Allele 2	24-033-113087	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Tay-Sachs Disease (HEXA), Interpretation	24-033-113087	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