

HOTLINE: Effective February 22, 2022

New Test 3004486 Tay-Sachs Disease (HEXA) Sequencing and Deletion/Duplication HEXA NGS



Additional Technical Information



Patient History for Tay-Sachs Disease (*HEXA*) Testing

Methodology: Massively Parallel Sequencing

Performed: Varies **Reported:** 3 weeks

Specimen Required: Collect: Lavender or pink (EDTA) or yellow (ACD Solution A or B)

Specimen Preparation: Transport 3 mL whole blood. (Min: 3 mL)

Storage/Transport Temperature: Refrigerated.

<u>Unacceptable Conditions:</u> Serum or plasma; grossly hemolyzed or frozen specimens; saliva, buccal brush or swab, FFPE tissue.

Stability (collection to initiation of testing): Ambient: 72 hours; Refrigerated: 1 week; Frozen: Unacceptable

Reference Interval: By report

Interpretive Data:

Refer to report.

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.

Note: Gene Tested: HEXA (NM_000520)

The pathogenic 7.6kb deletion specific to French Canadian populations is detected by this assay at reduced sensitivity.

CPT Code(s): 81406, 81479

New York DOH approval pending. Call for status update.

HOTLINE NOTE: Refer to the Test Mix Addendum for interface build information.