Hexosaminidase A enzyme activity is indeterminate or indicates carrier for Tay-Sachs disease (TSD) and Sandhoff disease excluded. MUGS is performed.

2 disease-causing variants identified:
- Diagnostic for TSD
- MUGS cancelled
- Consider molecular confirmation and/or referral to Genetics Specialist

0-1 disease-causing variants identified:
- Reduced enzyme consistent with TSD suggests the presence of a variant that is undetectable by this method
- TSD confirmed

Hexosaminidase A enzyme activity is normal:

Order:
- NAGS / Hexosaminidase A and Total Hexosaminidase Serum
- MUGS / Hexosaminidase A (MUGS), Serum

MUGS performed

Hexosaminidase A enzyme activity indicates affected with Tay-Sachs disease (TSD):
- Diagnostic for TSD
- MUGS cancelled
- Consider molecular confirmation and/or referral to Genetics Specialist

Hexosaminidase A enzyme activity is indeterminate or indicates carrier for Tay-Sachs disease (TSD):
- If clinical suspicion remains, consider AB variant molecular testing, order CGPH (GM2A Gene List ID: IEMCP-V46XB3)
- Consider alternative diagnosis
- Consider referral to Genetics Specialist

Hexosaminidase A enzyme activity is normal:
- Diagnostic for SD
- MUGS cancelled
- Consider molecular diagnosis and/or referral to Genetics Specialist

Hexosaminidase A enzyme activity indicates possible Sandhoff carrier:
- SD confirmed
- Enzyme activity consistent with SD suggests the presence of a variant that is undetectable by this method

Most common forms of TSD excluded:

Infantile onset:
- Increased startle reflex
- Developmental regression
- Progressive neurodegeneration
- Cherry red spot

Childhood/Adult onset:
- Motor neuron disease
- Behavior changes and/or psychiatric manifestations
- Gait and speech disturbances
- Movement disorders

0-1 disease-causing variants identified:
- SD confirmed
- Enzyme activity consistent with SD suggests the presence of a variant that is undetectable by this method

2 disease-causing variants identified:
- Diagnostic for TSD
- Consider molecular diagnosis and/or referral to Genetics Specialist

HEXBZ / Sandhoff Disease, HEXB Gene, Full Gene Analysis, Varies

NAGS performed

Order:
- NAGS / Hexosaminidase A and Total Hexosaminidase Serum
- MUGS / Hexosaminidase A (MUGS), Serum

MUGS is performed

Sandhoff disease excluded


If clinical suspicion remains, consider AB variant molecular testing, order CGPH (GM2A Gene List ID: IEMCP-V46XB3)
- Consider alternative diagnosis
- Consider referral to Genetics Specialist

*Interpretive comments provided with all reports