Hexosaminidase A enzyme activity is indeterminate or indicates carrier for Tay-Sachs disease (TSD)

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

NAGS performed

Hexosaminidase A enzyme activity indicates affected with Tay-Sachs disease (TSD)
- Diagnostic for TSD
- MUGS cancelled
- Consider referral to Genetics

Hexosaminidase A enzyme activity is indeterminate or indicates carrier for Tay-Sachs disease (TSD)

Hexosaminidase A enzyme activity is normal

Hexosaminidase A enzyme activity indicates possible Sandhoff carrier

Sandhoff disease excluded

MUGS is performed

Hexosaminidase A enzyme activity indicates affected with Sandhoff disease (SD)
- Diagnostic for SD
- MUGS cancelled

If still clinically suspicious consider AB Variant (GM2A) molecular testing
Consider alternative diagnosis
Consider referral to Genetics

Yes

2 disease-causing mutations identified

TSD confirmed

No

0-1 disease-causing mutations identified

2 disease-causing mutations identified

HEXAZ / Tay-Sachs Disease, HEXA Gene, Full Gene Analysis
- Diagnostic for TSD
- Consider molecular diagnosis and/or referral to Genetics

0-1 disease-causing mutations identified

Reduced enzyme consistent with TSD suggests the presence of a mutation that is undetectable by this method

TSD confirmed

Most common forms of TSD excluded

Infantile onset
One or more of the following:
- Increased startle reflex
- Developmental regression
- Progressive neurodegeneration
- Cherry red spot

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

*For Carrier Testing, please see Tay–Sachs Disease Carrier Testing Protocol
**Interpretive comments provided with all reports