Tay–Sachs Disease Carrier Testing Protocol*

Proceed with:
AJPO / Ashkenazi Jewish Mutation Analysis Panel Without Cystic Fibrosis (CF)

Includes:
- Hexosaminidase A and Total, Leukocytes/Ashkenazi Jewish (enzyme quantitation for Tay-Sachs disease)
- Targeted molecular analysis for:
  - Tay-Sachs disease
  - Canavan disease
  - Familial dysautonomia
  - Gaucher disease
  - Bloom syndrome
  - Fanconi anemia type C
  - Niemann-Pick disease types A and B

Is comprehensive Ashkenazi Jewish carrier screening desired?

Yes

No

Test for Tay-Sachs disease only

NAGR / Hexosaminidase A and Total, Leukocytes/Molecular Reflex

Hexosaminidase A enzyme quantitation is indeterminate or indicates carrier

Molecular testing for 5 common Ashkenazi Jewish mutations and 2 pseudodeficiency alleles

Mutation detected—carrier

No mutations detected

Pseudodeficiency mutation detected—noncarrier

Test partner for Sandhoff carrier status: Contact Biochemical Genetics Laboratory (800-533-1710) for testing options

Hexosaminidase A enzyme quantitation indicates noncarrier

Hexosaminidase A enzyme quantitation indicates possible Sandhoff carrier

STOP

Recommended testing partner; order NAGR / Hexosaminidase A and Total, Leukocytes/Molecular Reflex

Molecular testing does not rule out all possible mutations:
- Consider HEXAZ / Tay-Sachs Disease, HEXA Gene, Full Gene Analysis
- Consider testing partner, order NAGR

STOP

* Interpretive report provided for all tests in this algorithm

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