Tay–Sachs Disease Carrier Testing Protocol*

Is comprehensive Ashkenazi Jewish carrier screening desired?

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)

AND

- Targeted molecular analysis for:
  - Tay-Sachs disease
  - Canavan disease
  - Familial dysautonomia
  - Gaucher disease
  - Bloom syndrome
  - Fanconi anemia type C
  - Mucolipidosis IV
  - Niemann-Pick disease types A and B

French Canadian ancestry

Louisiana Cajun ancestry

Positive family history (known carriers or affected individuals)

Partner who is at risk to be a carrier

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

Recommend 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

Test partner for Sandhoff carrier status: Contact Biochemical Genetics Laboratory for testing options

Hexosaminidase A enzyme quantitation indicates noncarrier

Hexosaminidase A enzyme quantitation indicates possible Sandhoff carrier

STOP

STOP

STOP

* Interpretive report provided for all tests in this algorithm