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)
  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

Is comprehensive Ashkenazi Jewish carrier screening desired?

Test for Tay-Sachs disease only

Hexosaminidase A enzyme quantitation is indeterminate or indicates carrier
Molecular testing for 5 common Ashkenazi Jewish mutations and 2 pseudodeficiency alleles

Mutation detected—carrier
Recommends testing partner; order NAGR / Hexosaminidase A and Total, Leukocytes/Molecular Reflex

No mutations detected
Molecular testing does not rule out all possible mutations:
- Consider HEXAZ / Tay-Sachs Disease, HEXA Gene, Full Gene Analysis
- Consider testing partner, order NAGR

Pseudodeficiency mutation detected—noncarrier

Hexosaminidase A enzyme quantitation indicates noncarrier

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

Hexosaminidase A enzyme quantitation indicates possible Sandhoff carrier

NAGR / Hexosaminidase A and Total, Leukocytes/Molecular Reflex

STOP

*Interpretive report provided for all tests in this algorithm