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
  - Mucolipidosis IV
  - Niemann-Pick disease types A and B

**Is comprehensive Ashkenazi Jewish carrier screening desired?**

**YES**

**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**

**STOP**

**NAGR / Hexosaminidase A and Total, Leukocytes/Molecular Reflex**

**Hexosaminidase A enzyme quantitation is indeterminate or indicates carrier**

**STOP**

**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**

**STOP**

**Mutation detected—carrier**

**Recommend 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**

**STOP**

**Mutation detected—carrier**

**No mutations detected**

**Pseudodeficiency mutation detected—noncarrier**

**STOP**

*Interpretive report provided for all tests in this algorithm*