

## GENETIC DISEASES EVALUATION CENTER

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### Test Name & Code:

- **Test Name:** Tay-Sachs Disease Genetic Test (*HEXA* Gene)
- **Test Code:** 4827

### Related Diseases & Genes:

- Tay-Sachs disease (OMIM #: 272800) || Gene: *HEXA* (OMIM #: 606869)
- "GM2-gangliosidosis, several forms" (OMIM #: 272800)

### Synonyms:

- Hexosaminidase A
- *HEXA* Mutations
- Hexosaminidase A Mutations

### Test Details:

- **Methodology:** DNA Sequencing
- **Performed:** Everyday
- **Reported:** 1 week

### Specimen Details:

- **Specimen Collection:** Peripheral blood
- **Container:** Tube with EDTA
- **Specimen Volume/Amount:** > 5 ml
- **Storage/Transport Condition:** Refrigerated
- **Unacceptable Specimens/Conditions:** Serum; Frozen specimens; Hemolyzed specimens; Icterus; Lipemia

### Testing Area(s):

- Inborn Errors of Metabolism

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