

Genetic Insights: quick reference guide for healthcare providers

This guide is intended to facilitate a discussion between a provider and their patient.

Genetic Insights test results: **Tay-Sachs disease carrier**

Key results

A pathogenic or likely pathogenic variant associated with being a carrier of Tay-Sachs disease was found in 1 copy of the *HEXA* gene. Carriers do not have the condition but are at higher risk of having children with the condition if the other biological parent is also a carrier.

Next steps

Clinical recommendations

Genetic Insights is a screening test and not intended for diagnosis or to replace routine carrier screening for family planning or during pregnancy.

*Follow-up genetic testing should be performed in a clinical setting, especially if biological children are planned. Comprehensive analysis in a clinical setting of the *HEXA* gene and/or enzymatic activity should be considered for the reproductive partners/gamete donors.*

Resources

Ready to order?

Check with your institution and/or patient's insurance about the preferred testing laboratory.

Blueprint Genetics® offers Tay-Sachs disease genetic testing. To confirm this test result, targeted variant testing for the variant identified is available. You can order a confirmation test here: [Blueprint Genetics/TVT](#)

Have questions?

Call **1.866.GENE.INFO** (1.866.436.3463) to speak to a specialized Quest genetic counselor or geneticist available to healthcare providers to discuss test selection and results.

Refer your patient to a genetic counselor.

Your patient can schedule a 1-on-1 remote genetic counseling session through their online Genetic Insights dashboard at no additional cost.

Genetic counselors can provide counseling on the implications of this test result and next steps for your patient.

To find a genetic counselor practicing in your patient's area for an in-person session, please visit FindAGeneticCounselor.NSGC.org

Patient conversation starters:

Patient conversation starters summarize the preceding information in plain language to support meaningful conversations between you and your patient.

FindAGeneticCounselor.NSGC.org

Your Genetic Insights test is a screening test. The next step is to have your result confirmed with a second genetic test, especially if you are considering having children. It's also important that you talk with a genetic counselor. Genetic counselors are experts in genetics and can help you understand this result and potential next steps.

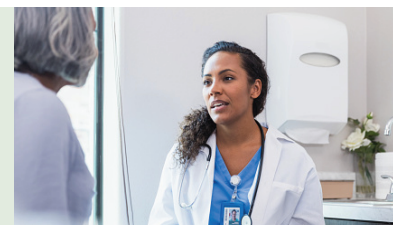
You can access a genetic counselor through your Genetic Insights dashboard at no additional cost to you, and we can discuss a referral to a local genetic counselor.

Additional resources

The following advocacy groups have additional information and resources about Tay-Sachs disease:

National Tay-Sachs & Allied Diseases Association: NTSAD.org

National Organization for Rare Disorders: RareDiseases.org/Tay-Sachs



Visit QuestDiagnostics.com/Genetic-Health-Screening for more information about this test.

What is Tay-Sachs disease?

Tay-Sachs disease is a rare genetic condition characterized by progressive death of nerve cells in the brain and spinal cord.¹ Symptoms typically appear in infancy and death most often occurs in childhood.¹ However, some DNA variants are associated with forms of the disease that present later in life with symptoms that may be less severe.¹

Tay-Sachs disease results from 2 variants, 1 in each copy of the *HEXA* gene.¹ People with a variant associated with Tay-Sachs disease in only 1 copy of *HEXA* are considered carriers and do not have the condition.

There is no cure for Tay-Sachs disease and treatments are generally supportive in nature.¹

What this result means for family members

Tay-Sachs disease is an inherited condition. If both biological parents are carriers, each child has a 1 in 4 chance of having Tay-Sachs disease.

If only one parent is a carrier, each child has a 1 in 2 chance of being a carrier and a 1 in 2 chance that they will not be carriers.

Parents and siblings of Tay-Sachs disease carriers may also be carriers.

Individuals with this result should discuss their carrier status with their reproductive partners as well as other family members who may also be carriers.

Comprehensive *HEXA* gene analysis and/or enzymatic activity testing for family members may be warranted, especially for family planning purposes.^{1,2}

References

1. Toro C, Shirvan L, Tiffit C. *HEXA* Disorders. March 11, 1999. Updated October 1, 2020. In Adam MP, Ardinger HH, Pagon RA, et al, editors. GeneReviews®. University of Washington, Seattle; 1993-2019. www.ncbi.nlm.nih.gov/books/NBK1218
2. The American College of Obstetricians and Gynecologists Committee Opinion. Carrier Screening for Genetic Conditions. Number 691, March 2017 (Reaffirmed 2023). www.acog.org/Clinical-Guidance-and-Publications/Committee-Opinions/Committee-on-Genetics/Carrier-Screening-for-Genetic-Conditions

This information is not a substitute for medical advice, diagnosis, or treatment. The diagnosis or treatment of any disease or condition may be based on personal history, family history, symptoms, a physical examination, laboratory test results, and other information considered important by a healthcare provider.

Individuals should talk with a healthcare provider about the meaning of genetic test results and before stopping, starting, or changing any medication or treatment.

Genetic Insights is a test developed and performed by Quest Diagnostics. The test results are not diagnostic and do not determine overall chances of developing a disease or health condition. The tests are not cleared or approved by the US Food and Drug Administration.

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Patient conversation starters:

Tay-Sachs disease is a rare genetic condition that primarily affects the function of the brain and spinal cord.

People with 1 DNA variant in the *HEXA* gene are considered carriers of Tay-Sachs disease.

Carriers do not have Tay-Sachs disease, but their biological children could have it if their other biological parent (or sperm/egg donor) is also a Tay-Sachs disease carrier.



Patient conversation starters:

Tay-Sachs disease runs in families. That means the DNA variants linked to it can be passed down from parents to their children. One of your biological parents is likely a Tay-Sachs disease carrier. Your siblings have a 1 in 2 (50%) chance of also being a Tay-Sachs disease carrier. Your biological children's chance of being a carrier or having Tay-Sachs disease depends on if their other parent is a carrier.

Sharing these results with your family members is important so family members can decide if they want to have genetic testing.