

Genetic Disorder Prediction Report

Patient Age: -90

Gender: Female

Predicted Disorder: Genetic Disorder

Subclass: Tay-Sachs

Date: 31-01-2026

Description:

Overview

Tay-Sachs disease is a rare genetic disorder that affects the nervous system. It is caused by a deficiency

Causes

Tay-Sachs is inherited in an autosomal recessive pattern, which means that a child must inherit two copies

Symptoms

Symptoms of Tay-Sachs usually appear in infants around 6 months of age and may include:

- Loss of motor skills (like sitting up or crawling)
- Increased startle response
- Weakness in muscles
- Difficulty with vision and hearing
- Cherry-red spot in the eye (visible during an eye exam)

As the disease progresses, children may experience seizures, paralysis, and eventually loss of life, typical

Diagnosis

To diagnose Tay-Sachs, doctors may perform:

- **Genetic Testing:** A blood test can check for mutations in the HEXA gene.
- **Enzyme Testing:** A blood sample can also be tested to measure the level of the Hex-A enzyme. Low
- **Family History:** Doctors will often ask about family history to assess the risk of the disorder.

Treatment

Currently, there is no cure for Tay-Sachs disease. Treatment focuses on managing symptoms and providing

- Physical therapy to help with muscle strength and coordination
- Occupational therapy to assist with daily activities
- Speech therapy to help with communication
- Medications to control seizures or other symptoms

Follow-up Advice

If you or your family member has been diagnosed with Tay-Sachs or is a carrier, it's important to:

- Work closely with a healthcare team that includes specialists in genetics, neurology, and palliative care.
- Consider genetic counseling to understand the risks for future pregnancies and explore options.
- Join support groups for families affected by Tay-Sachs, where you can share experiences and find emotional support.

Remember, while Tay-Sachs is a challenging condition, there are resources and support available.