

Genetic Disorder Prediction Report

Patient Age: 20

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. This means that a child must inherit two copies

Symptoms

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

- Loss of motor skills (like crawling or sitting up)
- Increased startle response
- Weakness and decreased muscle tone
- Vision problems, such as a "cherry-red spot" in the eye
- Seizures
- Hearing loss

As the disease progresses, children may experience more severe neurological decline, leading to loss of

Diagnosis

Tay-Sachs can be diagnosed through:

- **Genetic Testing:** A blood test can identify mutations in the HEXA gene.
- **Enzyme Testing:** A blood sample can also measure the level of the Hex-A enzyme. Low levels indicate a diagnosis.
- **Prenatal Testing:** If there is a family history, testing can be done during pregnancy to determine if the fetus is affected.

Treatment

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

- Physical therapy to help maintain mobility
- Occupational therapy to assist with daily activities
- Medications to manage seizures or other symptoms
- Nutritional support to ensure proper feeding

Follow-up Advice

If your child has been diagnosed with Tay-Sachs, it's important to work closely with a healthcare team that

If you have any questions or concerns about Tay-Sachs or genetic testing, don't hesitate to reach out to your healthcare provider.