Genetic Disorder Prediction Report

Patient Age: 1

Gender: Male

Predicted Disorder: Genetic Disorder

Subclass: Tay-Sachs

Date: 26-12-2025

Description:

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

Causes
Tay-Sachs is inherited in an autosomal recessive manner, which means that a child must inherit two copi

Symptoms
Symptoms of Tay-Sachs typically appear in infants around 6 months of age and can include:
- Loss of motor skills (like sitting up or crawling)
- Increased startle response
- Seizures
- Vision and hearing loss
- Muscle weakness
- Cherry-red spot in the eye (visible during an eye exam)

As the disease progresses, children may experience severe cognitive and physical decline, and unfortuna

Diagnosis
To diagnose Tay-Sachs, doctors may perform:
- **Blood tests**: These can check for the levels of the Hex-A enzyme. A low level indicates Tay-Sachs.
- **Genetic testing**: This can confirm the presence of mutations in the HEXA gene.
- **Prenatal testing**: If there is a family history of Tay-Sachs, testing can be done during pregnancy to se

Treatment
Currently, there is no cure for Tay-Sachs disease. Treatment focuses on managing symptoms and provid
- Physical therapy to help maintain mobility
- Medications to control seizures
- Nutritional support
- Palliative care to improve quality of life

Follow-up Advice
If you or your family is affected by Tay-Sachs or if you are a carrier, it’s important to:
- Consult with a genetic counselor for personalized advice and support.
- Discuss family planning options if you are considering having children.
- Stay informed about ongoing research, as new treatments and therapies are being developed.

Remember, support groups and resources are available to help families cope with the challenges of Tay-