Genetic Disorder Prediction Report

Patient Age: 11

Gender: Male

Predicted Disorder: Genetic Disorder

Subclass: Tay-Sachs

Date: 26-12-2025

Description:

Tay-Sachs Disease: A Patient-Friendly Overview

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
- 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 more severe neurological decline, leading to signific

Diagnosis:
Tay-Sachs is diagnosed through a combination of:
- **Family history:** Understanding if there are any known cases in the family.
- **Blood tests:** These tests can measure the levels of the Hex-A enzyme. Low levels indicate Tay-Sach
- **Genetic testing:** This can confirm the presence of mutations in the HEXA gene.

Treatment:
Currently, there is no cure for Tay-Sachs disease. Treatment focuses on managing symptoms and provid
- Physical therapy to help maintain mobility
- Occupational therapy to assist with daily activities
- Medications to manage seizures
- Nutritional support

Follow-Up Advice:
If you or your family member has been diagnosed with Tay-Sachs or is a carrier, it’s important to:
- Stay in touch with a healthcare provider who specializes in genetic disorders.
- Consider genetic counseling for family planning and understanding the risks for future pregnancies.
- Join support groups or organizations that provide resources and connect families affected by Tay-Sachs

Remember, while Tay-Sachs is a serious condition, having a supportive healthcare team and community