

Genetic Disorder Prediction Report

Patient Age: 11

Gender: Male

Predicted Disorder: Genetic Disorder

Subclass: Leigh syndrome

Date: 26-12-2025

Description:

Overview

Leigh syndrome is a rare genetic disorder that affects the brain and nervous system. It usually appears in

Causes

Leigh syndrome is caused by mutations (changes) in the genes that are important for energy production in

Symptoms

Symptoms of Leigh syndrome can vary but often include:

- Delayed development or loss of previously acquired skills (like walking or talking)
- Muscle weakness and poor muscle tone
- Difficulty with movement and coordination
- Seizures
- Breathing problems
- Problems with feeding and swallowing
- Abnormalities in the heart or other organs

Symptoms usually start in the first year of life, but they can sometimes appear later.

Diagnosis

To diagnose Leigh syndrome, doctors will typically:

1. ****Take a Medical History****: They will ask about the child's symptoms and family history.
2. ****Perform a Physical Examination****: This helps assess the child's development and physical condition.
3. ****Order Tests****: These may include:
 - Blood tests to check for metabolic issues.
 - MRI scans of the brain to look for characteristic changes.
 - Genetic testing to identify specific mutations.

Treatment

There is currently no cure for Leigh syndrome, but treatment focuses on managing symptoms and improving quality of life.

- Physical therapy to help with movement and coordination.
- Occupational therapy to assist with daily activities.
- Medications to control seizures or other symptoms.
- Nutritional support, such as special diets or feeding tubes if necessary.

In some cases, supplements like thiamine (vitamin B1) or other vitamins may be recommended, but this will depend on the specific needs of the individual.

Follow-up Advice

If your child has been diagnosed with Leigh syndrome, regular follow-up with a healthcare team is important to monitor their condition and adjust treatment as needed.

Additionally, consider joining support groups for families affected by Leigh syndrome. Connecting with other families can provide emotional support and practical advice.