

GENETIC DISORDER REPORT

Genetic Disorder: Mitochondrial genetic inheritance disorders

Subclass: Leigh syndrome

----- PATIENT DETAILS -----

patient_age: 15

father_age: 50

mother_age: 50

gender: Male

genes_mother_side: No

inherited_father: Yes

maternal_gene: No

paternal_gene: Yes

blood_cell_count: 5000

white_blood_cell_count: 7000

respiratory_rate: 18

heart_rate: 72

parental_consent: Yes

follow_up: Medium

birth_effects: Yes

folic_acid_intake: Yes

blood_test_result: Abnormal

No_of_previous_abortion: 1

----- DESCRIPTION -----

Okay, let's break down Leigh Syndrome, a type of mitochondrial disorder, in simple medical terms.

Leigh Syndrome: The Basics

- * **Overview:** Leigh syndrome is a severe neurological disorder that affects the central nervous system.
- * **Causes:** Leigh syndrome is caused by problems with the mitochondria. Mitochondria are like the powerhouses of our cells, providing energy for almost all cellular activities.
- * **Symptoms:** The symptoms of Leigh syndrome are varied and can appear at any point, but often start in early childhood. They include:
 - * **Developmental delays or regression:** Loss of previously acquired skills (like sitting, crawling, or walking).
 - * **Muscle problems:** Weakness, floppiness (hypotonia), stiffness (spasticity), or involuntary movements (dystonia).
 - * **Movement problems:** Difficulty coordinating movements, balance issues (ataxia).
 - * **Breathing problems:** Irregular breathing patterns, difficulty breathing.
 - * **Feeding difficulties:** Problems swallowing or sucking.
 - * **Vision problems:** Optic atrophy (damage to the optic nerve).
 - * **Seizures:** Uncontrolled electrical activity in the brain.
 - * **Lactic acidosis:** Buildup of lactic acid in the body, which can cause nausea, vomiting, fatigue, and muscle pain.
 - * **Failure to thrive:** In infants, this means not gaining weight or growing at the expected rate.
 - * **Heart problems:** Cardiomyopathy (weakening of the heart muscle)
- * **Risk Factors:**
 - * **Family history:** Having a family history of Leigh syndrome or other mitochondrial disorders is the most common risk factor.
 - * **Maternal inheritance:** If the mother carries a mitochondrial gene mutation, there is a high chance of passing it on to her children.
- * **Treatment:** Unfortunately, there is no cure for Leigh syndrome. Treatment focuses on managing symptoms and improving quality of life. It may include:
 - * **Medications:**
 - * To control seizures
 - * To reduce lactic acid levels
 - * To manage movement disorders
 - * Vitamins and supplements (like thiamine, coenzyme Q10, creatine, and leucine)