

# GENETIC DISORDER REPORT

Genetic Disorder: Mitochondrial genetic inheritance disorders  
Subclass: Leigh syndrome

## ----- PATIENT DETAILS -----

patient\_age: 123  
father\_age: 175  
mother\_age: 125  
gender: Male  
genes\_mother\_side: Yes  
inherited\_father: Yes  
maternal\_gene: No  
paternal\_gene: No  
blood\_cell\_count: 4500  
white\_blood\_cell\_count: 6500  
respiratory\_rate: 18  
heart\_rate: 80  
parental\_consent: No  
follow\_up: High  
birth\_effects: Yes  
folic\_acid\_intake: Yes  
blood\_test\_result: Abnormal  
No\_of\_previous\_abortion: 8

## ----- DESCRIPTION -----

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

### **\*\*Leigh Syndrome: The Basics\*\***

- \* **\*\*Overview:\*\*** Leigh syndrome is a severe neurological disorder that typically appears in infancy or early childhood.
- \* **\*\*Causes:\*\*** Leigh syndrome is usually caused by genetic mutations that affect the mitochondria. Mitochondria are the powerhouses of the cell, responsible for producing energy.
- \* **\*\*Symptoms:\*\*** Symptoms can vary in severity and the age they start, but common ones include:
  - \* **\*\*Developmental Delays:\*\*** Slower than usual progress in reaching milestones like sitting, crawling, and walking.
  - \* **\*\*Muscle Problems:\*\*** Weakness (hypotonia), floppiness, or stiffness (spasticity).
  - \* **\*\*Movement Problems:\*\*** Difficulty with coordination, balance, and walking (ataxia).
  - \* **\*\*Breathing Problems:\*\*** Irregular breathing patterns or even episodes where breathing stops (apnea).
  - \* **\*\*Feeding Difficulties:\*\*** Problems with swallowing, leading to poor weight gain.
  - \* **\*\*Vision Problems:\*\*** Abnormal eye movements (nystagmus), optic atrophy (damage to the optic nerve).
  - \* **\*\*Seizures:\*\*** Uncontrolled electrical activity in the brain.
  - \* **\*\*Lactic Acidosis:\*\*** A buildup of lactic acid in the blood, which can cause vomiting, rapid breathing, and other complications.
- \* **\*\*Risk Factors:\*\***
  - \* **\*\*Family History:\*\*** Having a family history of mitochondrial disorders, especially if previous family members had similar symptoms.
  - \* **\*\*Carrier Status:\*\*** If a parent carries a genetic mutation that can cause Leigh syndrome, their child has a 50% chance of inheriting it.
- \* **\*\*Treatment:\*\*** Unfortunately, there is no cure for Leigh syndrome. Treatment focuses on managing symptoms and supporting the child's health.
  - \* **\*\*Medications:\*\***
    - \* **\*\*Vitamins and Supplements:\*\*** Some vitamins, like thiamine (B1), coenzyme Q10, and creatine, may help improve mitochondrial function.
    - \* **\*\*Sodium Bicarbonate or Citrate:\*\*** To help manage lactic acidosis.
    - \* **\*\*Anticonvulsants:\*\*** To control seizures.
  - \* **\*\*Physical Therapy:\*\*** To help maintain muscle strength and flexibility.
  - \* **\*\*Occupational Therapy:\*\*** To help with daily living skills.
  - \* **\*\*Speech Therapy:\*\*** To help with communication and feeding difficulties.