

# GENETIC DISORDER REPORT

Genetic Disorder: Mitochondrial genetic inheritance disorders  
Subclass: Leigh syndrome

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

patient\_age: 20  
father\_age: 60  
mother\_age: 50  
gender: Female  
genes\_mother\_side: Yes  
inherited\_father: Yes  
maternal\_gene: Yes  
paternal\_gene: Yes  
blood\_cell\_count: 5000  
white\_blood\_cell\_count: 5.8213  
respiratory\_rate: 12  
heart\_rate: 75  
parental\_consent: None  
follow\_up: None  
birth\_effects: None  
folic\_acid\_intake: None  
blood\_test\_result: Not  
No\_of\_previous\_abortion: None

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

Okay, let's break down Leigh Syndrome in simple medical terms:

### **\*\*Leigh Syndrome: A Breakdown\*\***

- \* **\*\*Overview:\*\*** Leigh Syndrome is a severe, progressive neurological disorder that typically appears in early childhood.
- \* **\*\*Causes:\*\*** The root of Leigh Syndrome lies in the mitochondria, which are like the power plants of cells.
  - \* Think of it like a factory where the power plant is malfunctioning. If the power plant can't produce energy, the factory (cell) can't function properly.
- \* **\*\*Symptoms:\*\*** The symptoms of Leigh Syndrome can vary widely from person to person and often worsen over time.
  - \* Developmental delays (difficulty reaching milestones like sitting, crawling, or walking)\*
  - \* Muscle weakness or floppiness (hypotonia)\*
  - \* Movement problems (difficulty with coordination, tremors, seizures)\*
  - \* Breathing difficulties (irregular breathing, pauses in breathing)\*
  - \* Feeding problems (difficulty swallowing, vomiting)\*
  - \* Vision problems (nystagmus - involuntary eye movements, optic atrophy - damage to the optic nerve)\*
  - \* Lactic acidosis (build-up of lactic acid in the body, which can cause nausea, vomiting, and rapid breathing)\*
  - \* Irritability and/or lethargy\*
  - \* Failure to thrive (not gaining weight or growing as expected)\*
- \* **\*\*Risk Factors:\*\***
  - \* Family History: The biggest risk factor is having parents who carry the genetic mutations associated with Leigh Syndrome.
  - \* Consanguinity: In some cases, consanguinity (marriage between close relatives) increases the risk of genetic disorders.
- \* **\*\*Treatment:\*\*** Unfortunately, there's no cure for Leigh Syndrome. Treatment focuses on managing symptoms and slowing down the progression of the disease.
  - \* Medications: To control seizures, manage lactic acidosis, or address specific symptoms.\*
  - \* Physical therapy: To help with muscle weakness and movement problems.\*
  - \* Occupational therapy: To help with daily living skills.\*
  - \* Speech therapy: To help with feeding and communication difficulties.\*
  - \* Nutritional support: Special diets and supplements are sometimes used to help manage the condition.\*