

# GENETIC DISORDER REPORT

Genetic Disorder: Mitochondrial genetic inheritance disorders  
Subclass: Leigh syndrome

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

patient\_age: 30  
father\_age: 70  
mother\_age: 70  
gender: Female  
genes\_mother\_side: No  
inherited\_father: Yes  
maternal\_gene: No  
paternal\_gene: Yes  
blood\_cell\_count: 5.0888  
white\_blood\_cell\_count: 5000  
respiratory\_rate: 15  
heart\_rate: 73  
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, into simple medical terms:

**\*\*Leigh Syndrome: The Battery's Not Working Right\*\***

- \* **\*\*Overview:\*\*** Think of your cells like little cities, each needing power to function. Mitochondria are like the power plants of these cities.
- \* **\*\*Causes:\*\***
  - \* **\*\*Mitochondrial Problems:\*\*** The root cause is a problem with the mitochondria's ability to produce energy.
  - \* **\*\*Genetic Inheritance:\*\*** Leigh syndrome is most often inherited through genetic mutations.
    - \* **\*\*Mitochondrial DNA (mtDNA):\*\*** Sometimes, the problem is in the DNA within the mitochondria.
    - \* **\*\*Nuclear DNA:\*\*** More commonly, the problem is in the DNA inside the cell's nucleus (nuclear DNA).
  - \* **\*\*What Goes Wrong:\*\*** Because of these gene mutations, the mitochondria cannot produce enough energy.
- \* **\*\*Symptoms:\*\*** The symptoms of Leigh syndrome can vary widely, even among people in the same family.
  - \* **\*\*Neurological Problems:\*\***
    - \* Developmental delays or regression (losing skills they had already learned)
    - \* Muscle weakness
    - \* Movement problems (like difficulty walking, coordination issues)
    - \* Seizures
    - \* Problems with vision (like nystagmus - involuntary eye movements)
    - \* Swallowing and breathing difficulties
    - \* Intellectual disability
  - \* **\*\*Other Problems:\*\***
    - \* Vomiting
    - \* Diarrhea
    - \* Heart problems (cardiomyopathy)
    - \* Kidney problems
    - \* Failure to thrive (not gaining weight or growing properly)

\* **\*\*Risk Factors:\*\***