

# GENETIC DISORDER REPORT

Genetic Disorder: Mitochondrial genetic inheritance disorders  
Subclass: Leigh syndrome

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

patient\_age: 85  
father\_age: 21  
mother\_age: 18  
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: 25  
parental\_consent: Yes  
follow\_up: High  
birth\_effects: Yes  
folic\_acid\_intake: Yes  
blood\_test\_result: Abnormal  
No\_of\_previous\_abortion: 9

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

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

**\*\*Leigh Syndrome: A Power Problem Inside Cells\*\***

**\*\*Overview:\*\***

Imagine your body's cells are like tiny power plants. These power plants, called mitochondria, create the

**\*\*Causes:\*\***

- \* **\*\*Faulty Mitochondria:\*\*** Leigh syndrome is caused by a problem with the mitochondria themselves.
- \* **\*\*Mitochondrial DNA (mtDNA) Inheritance:\*\*** This is passed down only from the mother. If the mother has a faulty mitochondria, there is a 50% chance of passing it to the child.
- \* **\*\*Nuclear DNA Inheritance:\*\*** This is more common. Mutations in genes in the cell's nucleus (the DNA in the nucleus) can cause Leigh syndrome.
- \* **\*\*The Result:\*\*** The genetic mutations impair the ability of the mitochondria to produce energy efficiently.

**\*\*Symptoms:\*\***

The symptoms of Leigh syndrome usually appear in infancy or early childhood, but can appear later in life.

- \* **\*\*Developmental Delays:\*\*** Slower to reach milestones like sitting, crawling, or walking. Loss of skills already learned.
- \* **\*\*Movement Problems:\*\*** Difficulty with coordination, muscle weakness, involuntary movements (like tremors or chorea).
- \* **\*\*Breathing Problems:\*\*** Irregular breathing patterns, sometimes with periods of rapid breathing or apnea.
- \* **\*\*Feeding Difficulties:\*\*** Trouble sucking, swallowing, or keeping food down, leading to poor weight gain.
- \* **\*\*Vision Problems:\*\*** Abnormal eye movements, vision loss.
- \* **\*\*Seizures:\*\***
- \* **\*\*Lactic Acidosis:\*\*** A buildup of lactic acid in the body, which can cause nausea, vomiting, fatigue, and coma.
- \* **\*\*Heart Problems:\*\*** (Less common)

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