

# GENETIC DISORDER REPORT

Genetic Disorder: Mitochondrial genetic inheritance disorders  
Subclass: Leigh syndrome

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

patient\_age: 12  
father\_age: 45  
mother\_age: 45  
gender: Female  
genes\_mother\_side: Yes  
inherited\_father: No  
maternal\_gene: Yes  
paternal\_gene: No  
blood\_cell\_count: 5.054  
white\_blood\_cell\_count: 8000  
respiratory\_rate: 20  
heart\_rate: 66  
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, a type of mitochondrial genetic inheritance disorder, in simple

### \*\*Leigh Syndrome: A Simplified Explanation\*\*

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

- \* Leigh Syndrome is a severe neurological (brain and nervous system) disorder that usually appears in early childhood.
- \* Think of it like this: The brain and muscles (and sometimes other organs) aren't getting enough "fuel" to work properly.
- \* It belongs to a broader group of diseases called mitochondrial disorders because the problem lies with the mitochondria.

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

- \* \*\*Mitochondria are the "powerhouses" of cells.\*\* They convert food into energy that the body can use.
- \* \*\*Leigh Syndrome happens when the mitochondria don't work right.\*\* They can't produce enough energy.
- \* \*\*Genetic mutations (changes) are the root cause.\*\* These mutations affect the genes that control the mitochondria.
- \* \*\*Mitochondrial Inheritance:\*\* This is how it gets passed down. Mitochondria (and their DNA) are inherited from the mother.

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

- \* Symptoms can vary quite a bit from person to person. Here are some common ones:
  - \* \*\*Developmental delays or regression:\*\* Babies might not reach milestones on time or might lose skills they've already learned.
  - \* \*\*Muscle problems:\*\* Weakness, floppiness (hypotonia), stiffness (spasticity), or involuntary movements.
  - \* \*\*Breathing problems:\*\* Irregular breathing, rapid breathing, or pauses in breathing (apnea).
  - \* \*\*Feeding difficulties:\*\* Problems sucking, swallowing, or digesting food, leading to poor weight gain.
  - \* \*\*Vision problems:\*\* Abnormal eye movements (nystagmus) or vision loss.
  - \* \*\*Seizures.
  - \* \*\*Lactic acidosis:\*\* A buildup of lactic acid in the body, which can cause nausea, vomiting, and

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

- \* \*\*Family history:\*\* Having a family member with Leigh Syndrome or another mitochondrial disorder.
- \* \*\*Maternal inheritance:\*\* If the mother carries a mitochondrial DNA mutation, the risk to her children is higher.

#### \* \*\*Treatment:\*\*