

# GENETIC DISORDER REPORT

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Genetic Disorder: Mitochondrial genetic inheritance disorders

Subclass: Leigh syndrome

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

patient\_age: 20

father\_age: 54

mother\_age: 48

gender: Female

genes\_mother\_side: Yes

inherited\_father: Yes

maternal\_gene: Yes

paternal\_gene: Yes

blood\_cell\_count: 2000

white\_blood\_cell\_count: 7.8582

respiratory\_rate: 13

heart\_rate: 75

parental\_consent: None

follow\_up: None

birth\_effects: None

folic\_acid\_intake: None

blood\_test\_result: Yes

No\_of\_previous\_abortion: None

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

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

**\*\*Leigh Syndrome: In Simple Medical Terms\*\***

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

Leigh Syndrome is a severe, progressive brain disorder that typically appears in infancy or early childhood.

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

- \* **\*\*Mitochondrial Dysfunction:\*\*** The main problem is with the mitochondria, which are like tiny power plants in our cells.
- \* **\*\*Genetic Mutations:\*\*** The most common cause is a mutation (a change) in a gene that affects how mitochondria work.
- \* **\*\*Inheritance Patterns:\*\***
  - \* **\*\*Mitochondrial Inheritance:\*\*** Mitochondria have their own DNA. If the mother has a mutation in her mitochondrial DNA, it can be passed on to all of her children.
  - \* **\*\*Nuclear Inheritance:\*\*** Some mutations that cause Leigh Syndrome are in genes located in the cell nucleus. These can be passed on from either parent.
  - \* **\*\*Autosomal Recessive:\*\*** Both parents must carry one copy of the mutated gene for the child to be affected.
  - \* **\*\*X-linked:\*\*** The mutated gene is on the X chromosome. (More common in males).

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

The symptoms can vary, but often include:

- \* **\*\*Developmental Delays:\*\*** Slower than normal progress in reaching milestones like sitting, crawling, and walking.
- \* **\*\*Loss of Motor Skills:\*\*** Problems with movement, balance, and coordination. This can manifest as difficulty walking, climbing, or using fine motor skills.
- \* **\*\*Breathing Problems:\*\*** Irregular breathing patterns, sometimes with periods of rapid breathing (hyperventilation).
- \* **\*\*Feeding Difficulties:\*\*** Difficulty sucking, swallowing, or gaining weight. Vomiting is also common.
- \* **\*\*Vision Problems:\*\*** Nystagmus (involuntary eye movements), optic atrophy (damage to the optic nerve), and other visual impairments.
- \* **\*\*Seizures:\*\*** Can occur in some cases.
- \* **\*\*Lactic Acidosis:\*\*** A buildup of lactic acid in the blood, which can cause nausea, vomiting, fatigue, and muscle weakness.
- \* **\*\*Failure to Thrive:\*\*** Not gaining weight or growing as expected.