

## GENETIC DISORDER REPORT

Genetic Disorder: Mitochondrial genetic inheritance disorders

Subclass: Leigh syndrome

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

patient\_age: 50

father\_age: 70

mother\_age: 72

gender: Female

genes\_mother\_side: Yes

inherited\_father: No

maternal\_gene: Yes

paternal\_gene: No

blood\_cell\_count: 5.54248

white\_blood\_cell\_count: 4.9467

respiratory\_rate: 40

heart\_rate: 72

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 related to Mitochondrial genetic inheritance disorders in simple

**\*\*Leigh Syndrome: Explained\*\***

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

- \* Imagine your body's cells are like little power plants, and mitochondria are the engines inside those p
- \* It's like a progressive power outage in the brain, leading to a gradual loss of mental and movement a
- \* It usually shows up in infancy or early childhood, but sometimes it can appear later.
- \* It is an inherited disorder, meaning that the parents can pass the damaged mitochondrial genes on t

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

- \* **\*\*Mitochondrial Malfunction:\*\*** The primary cause is a problem with the mitochondria's ability to creat
- \* **\*\*Genetic Inheritance:\*\*** Leigh syndrome can be caused by mutations in mitochondrial DNA (mtDNA).
- \* **\*\*Mitochondrial DNA (mtDNA):\*\*** mtDNA is passed down only from the mother. So, if a mother has
- \* **\*\*Nuclear DNA (nDNA):\*\*** nDNA is inherited from both parents. This form is more common. In thes
- \* In both cases, if the mitochondrial function is inhibited, the rest of the body suffers the consequences

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

The symptoms of Leigh syndrome can vary from person to person, but they commonly include:

- \* **\*\*Developmental Delays:\*\*** Slow or stalled development milestones like sitting, crawling, or walking.
- \* **\*\*Muscle Problems:\*\*** Weakness (hypotonia), floppiness, stiffness, or difficulty controlling movements.
- \* **\*\*Movement Problems:\*\*** Tremors, involuntary muscle contractions (dystonia), and difficulty with coo
- \* **\*\*Breathing Issues:\*\*** Irregular breathing patterns or episodes of apnea (stopping breathing).
- \* **\*\*Feeding Problems:\*\*** Difficulty swallowing, poor weight gain, and vomiting.
- \* **\*\*Vision Problems:\*\*** Abnormal eye movements (nystagmus) or vision loss.
- \* **\*\*Cognitive Issues:\*\*** Progressive intellectual disability in the later stages.