

# GENETIC DISORDER REPORT

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Genetic Disorder: Mitochondrial genetic inheritance disorders

Subclass: Leigh syndrome

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

patient\_age: 39

father\_age: 65

mother\_age: 59

gender: Female

genes\_mother\_side: Yes

inherited\_father: Yes

maternal\_gene: Yes

paternal\_gene: Yes

blood\_cell\_count: 6.76619

white\_blood\_cell\_count: 4.64949

respiratory\_rate: 49

heart\_rate: 71

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 terms.

### \*\*Leigh Syndrome: A Problem with Energy Production in the Brain\*\*

\* \*\*Overview:\*\* Imagine your brain cells need tiny power plants (mitochondria) to function. Leigh syndrome is a disorder that affects these power plants.

\* \*\*Causes:\*\* Leigh syndrome is usually caused by a genetic mutation (a change) in the DNA that affects the mitochondria.

\* \*\*Mitochondrial DNA (mtDNA):\*\* This DNA is passed down **\*only from the mother\***. If the mother has a mutation, it can be passed on to her children.

\* \*\*Nuclear DNA:\*\* This DNA is found in the cell's nucleus and is inherited from both parents. Mutations in nuclear DNA can also cause Leigh syndrome.

Essentially, these mutations prevent the mitochondria from efficiently converting food and oxygen into energy.

\* \*\*Symptoms:\*\* Symptoms can vary, even within the same family, but commonly include:

\* \*\*Early signs:\*\* Often appear in infancy or early childhood. These might include poor sucking ability, feeding difficulties, and developmental delays.

\* \*\*Movement problems:\*\* Difficulty with coordination, balance, muscle weakness (hypotonia), and stiffness.

\* \*\*Developmental delays:\*\* Slower than normal development of motor skills (crawling, walking), speech, and cognitive abilities.

\* \*\*Respiratory problems:\*\* Breathing difficulties, including rapid breathing or periods of stopped breathing (apnea).

\* \*\*Seizures:\*\* Uncontrolled electrical activity in the brain.

\* \*\*Vision problems:\*\* Nystagmus (involuntary eye movements), optic atrophy (damage to the optic nerve), and visual field loss.

\* \*\*Heart problems:\*\* Cardiomyopathy (enlarged or weakened heart muscle).

\* \*\*Lactic acidosis:\*\* A buildup of lactic acid in the body, which can cause nausea, vomiting, abdominal pain, and fatigue.

\* \*\*Risk Factors:\*\* The main risk factor is having parents who carry a genetic mutation associated with Leigh syndrome.

\* \*\*Treatment:\*\* There's no cure for Leigh syndrome. Treatment focuses on managing symptoms and improving quality of life.

\* \*\*Nutritional support:\*\* Special diets, feeding tubes (if needed), and vitamin supplements (like thiamine and carnitine) can help reduce lactic acidosis and manage other specific symptoms.

\* \*\*Medications:\*\* Targeted medications can help manage specific symptoms like seizures or heart problems.