

## GENETIC DISORDER REPORT

Genetic Disorder: Mitochondrial genetic inheritance disorders

Subclass: Leigh syndrome

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

patient\_age: 15

father\_age: 50

mother\_age: 50

gender: Male

genes\_mother\_side: No

inherited\_father: Yes

maternal\_gene: No

paternal\_gene: Yes

blood\_cell\_count: 5000

white\_blood\_cell\_count: 7000

respiratory\_rate: 18

heart\_rate: 72

parental\_consent: Yes

follow\_up: Medium

birth\_effects: Yes

folic\_acid\_intake: Yes

blood\_test\_result: Abnormal

No\_of\_previous\_abortion: 1

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

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

#### **\*\*Leigh Syndrome: The Basics\*\***

\* **\*\*Overview:\*\*** Leigh syndrome is a severe neurological disorder that affects the central nervous system.

\* **\*\*Causes:\*\*** Leigh syndrome is caused by problems with the mitochondria. Mitochondria are like the

\* **\*\*Symptoms:\*\*** The symptoms of Leigh syndrome are varied and can appear at any point, but often s

\* **\*\*Developmental delays or regression:\*\*** Loss of previously acquired skills (like sitting, crawling, or

\* **\*\*Muscle problems:\*\*** Weakness, floppiness (hypotonia), stiffness (spasticity), or involuntary move

\* **\*\*Movement problems:\*\*** Difficulty coordinating movements, balance issues (ataxia).

\* **\*\*Breathing problems:\*\*** Irregular breathing patterns, difficulty breathing.

\* **\*\*Feeding difficulties:\*\*** Problems swallowing or sucking.

\* **\*\*Vision problems:\*\*** Optic atrophy (damage to the optic nerve).

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

\* **\*\*Lactic acidosis:\*\*** Buildup of lactic acid in the body, which can cause nausea, vomiting, fatigue, a

\* **\*\*Failure to thrive:\*\*** In infants, this means not gaining weight or growing at the expected rate.

\* **\*\*Heart problems:\*\*** Cardiomyopathy (weakening of the heart muscle)

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

\* **\*\*Family history:\*\*** Having a family history of Leigh syndrome or other mitochondrial disorders is th

\* **\*\*Maternal inheritance:\*\*** If the mother carries a mitochondrial gene mutation, there is a high chan

\* **\*\*Treatment:\*\*** Unfortunately, there is no cure for Leigh syndrome. Treatment focuses on managing s

\* **\*\*Medications:\*\***

\* To control seizures

\* To reduce lactic acid levels

\* To manage movement disorders

\* Mitochondrial supplements (like thiamine, coenzyme Q10, carnitine, and creatine)