

GENETIC DISORDER REPORT

Genetic Disorder: Mitochondrial genetic inheritance disorders

Subclass: Leigh syndrome

----- PATIENT DETAILS -----

patient_age: 20

father_age: 60

mother_age: 50

gender: Female

genes_mother_side: Yes

inherited_father: Yes

maternal_gene: Yes

paternal_gene: Yes

blood_cell_count: 5000

white_blood_cell_count: 5.8213

respiratory_rate: 12

heart_rate: 75

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 in simple medical terms:

Leigh Syndrome: A Breakdown

* **Overview:** Leigh Syndrome is a severe, progressive neurological disorder that typically appears in

* **Causes:** The root of Leigh Syndrome lies in the mitochondria, which are like the power plants of our

* *Think of it like a factory where the power plant is malfunctioning. If the power plant can't produce

* **Symptoms:** The symptoms of Leigh Syndrome can vary widely from person to person and often w

* *Developmental delays (difficulty reaching milestones like sitting, crawling, or walking)*

* *Muscle weakness or floppiness (hypotonia)*

* *Movement problems (difficulty with coordination, tremors, seizures)*

* *Breathing difficulties (irregular breathing, pauses in breathing)*

* *Feeding problems (difficulty swallowing, vomiting)*

* *Vision problems (nystagmus - involuntary eye movements, optic atrophy - damage to the optic nerve)*

* *Lactic acidosis (build-up of lactic acid in the body, which can cause nausea, vomiting, and rapid breathing)*

* *Irritability and/or lethargy*

* *Failure to thrive (not gaining weight or growing as expected)*

* **Risk Factors:**

* *Family History: The biggest risk factor is having parents who carry the genetic mutations associated with Leigh Syndrome.*

* *Consanguinity: In some cases, consanguinity (marriage between close relatives) increases the risk of passing on the disorder.*

* **Treatment:** Unfortunately, there's no cure for Leigh Syndrome. Treatment focuses on managing the symptoms.

* *Medications: To control seizures, manage lactic acidosis, or address specific symptoms.*

* *Physical therapy: To help with muscle weakness and movement problems.*

* *Occupational therapy: To help with daily living skills.*

* *Speech therapy: To help with feeding and communication difficulties.*

* *Nutritional support: Special diets and supplements are sometimes used to help reduce the effects of the disorder.*