

GENETIC DISORDER REPORT

Genetic Disorder: Mitochondrial genetic inheritance disorders

Subclass: Leigh syndrome

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

patient_age: 22

father_age: 60

mother_age: 55

gender: Male

genes_mother_side: Yes

inherited_father: Yes

maternal_gene: Yes

paternal_gene: No

blood_cell_count: 4500

white_blood_cell_count: 6500

respiratory_rate: 11

heart_rate: 88

parental_consent: No

follow_up: High

birth_effects: Yes

folic_acid_intake: Yes

blood_test_result: Slightly abnormal

No_of_previous_abortion: 1

----- DESCRIPTION -----

Okay, let's break down Leigh Syndrome, a type of mitochondrial disorder, in a way that's easy to understand.

Leigh Syndrome: Simple Medical Explanation

Overview:

Imagine your body's cells as tiny houses, and inside each house is a power plant called the mitochondria.

Causes:

* **Mitochondrial Dysfunction:** Leigh Syndrome is caused by problems with the mitochondria's ability to produce energy.

* **Genetic Inheritance:** The "faulty parts" are due to changes (mutations) in the genes that control how mitochondria work.

* **Mitochondrial DNA (mtDNA) Inheritance:** Mitochondria have their own DNA (mtDNA) separate from the rest of the cell.

* **Nuclear DNA Inheritance:** Most of the genes involved in mitochondrial function are actually located in the nucleus.

Symptoms:

Leigh Syndrome symptoms usually appear in infancy or early childhood, but can appear later. Because

* **Developmental Delays/Regression:** Loss of previously acquired skills like crawling, walking, or talking.

* **Movement Problems:** Difficulty with coordination, muscle weakness, stiff muscles (spasticity), jerking movements, or loss of balance.

* **Breathing Problems:** Irregular or shallow breathing, sometimes with periods of stopping breathing (apnea).

* **Feeding Difficulties:** Problems sucking, swallowing, or gaining weight.

* **Vision Problems:** Abnormal eye movements (nystagmus), optic atrophy (damage to the optic nerve), or sensitivity to light.

* **Brain Problems:** Irritability, lethargy (excessive tiredness), intellectual disability, or seizures.

* **Heart Problems:** A heart condition called cardiomyopathy.

* **Lactic Acidosis:** Build-up of lactic acid in the body, which can cause vomiting, abdominal pain, and

Risk Factors: