

GENETIC DISORDER REPORT

Genetic Disorder: Mitochondrial genetic inheritance disorders
Subclass: Leigh syndrome

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

patient_age: 20
father_age: 54
mother_age: 48
gender: Female
genes_mother_side: Yes
inherited_father: Yes
maternal_gene: Yes
paternal_gene: Yes
blood_cell_count: 2000
white_blood_cell_count: 7.8582
respiratory_rate: 13
heart_rate: 75
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, a type of mitochondrial disorder, in simple terms.

Leigh Syndrome: In Simple Medical Terms

Overview:

Leigh Syndrome is a severe, progressive brain disorder that typically appears in infancy or early childhood.

Causes:

- * **Mitochondrial Dysfunction:** The main problem is with the mitochondria, which are like tiny power plants in cells.
- * **Genetic Mutations:** The most common cause is a mutation (a change) in a gene that affects how mitochondria function.
- * **Inheritance Patterns:**
 - * **Mitochondrial Inheritance:** Mitochondria have their own DNA. If the mother has a mutation in her mitochondrial DNA, all her children will inherit it.
 - * **Nuclear Inheritance:** Some mutations that cause Leigh Syndrome are in genes located in the nucleus of the cell.
 - * **Autosomal Recessive:** Both parents must carry one copy of the mutated gene for the child to have the disorder.
 - * **X-linked:** The mutated gene is on the X chromosome. (More common in males).

Symptoms:

The symptoms can vary, but often include:

- * **Developmental Delays:** Slower than normal progress in reaching milestones like sitting, crawling, and walking.
- * **Loss of Motor Skills:** Problems with movement, balance, and coordination. This can manifest as clumsiness or falling.
- * **Breathing Problems:** Irregular breathing patterns, sometimes with periods of rapid breathing (hyperventilation) followed by slow breathing.
- * **Feeding Difficulties:** Difficulty sucking, swallowing, or gaining weight. Vomiting is also common.
- * **Vision Problems:** Nystagmus (involuntary eye movements), optic atrophy (damage to the optic nerve).
- * **Seizures:** Can occur in some cases.
- * **Lactic Acidosis:** A buildup of lactic acid in the blood, which can cause nausea, vomiting, fatigue, and confusion.
- * **Failure to Thrive:** Not gaining weight or growing as expected.