

GENETIC DISORDER REPORT

Genetic Disorder: Mitochondrial genetic inheritance disorders

Subclass: Leigh syndrome

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

patient_age: 12

father_age: 45

mother_age: 45

gender: Female

genes_mother_side: Yes

inherited_father: No

maternal_gene: Yes

paternal_gene: No

blood_cell_count: 5.054

white_blood_cell_count: 8000

respiratory_rate: 20

heart_rate: 66

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

Leigh Syndrome: A Simplified Explanation

* **Overview:**

- * Leigh Syndrome is a severe neurological (brain and nervous system) disorder that usually appears in early childhood.
- * Think of it like this: The brain and muscles (and sometimes other organs) aren't getting enough "fuel" to work properly.
- * It belongs to a broader group of diseases called mitochondrial disorders because the problem lies in the mitochondria.
- * **Causes:**

- * **Mitochondria are the "powerhouses" of cells.** They convert food into energy that the body can use.
- * **Leigh Syndrome happens when the mitochondria don't work right.** They can't produce enough energy for the body's cells.
- * **Genetic mutations (changes) are the root cause.** These mutations affect the genes that control the mitochondria.
- * **Mitochondrial Inheritance:** This is how it gets passed down. Mitochondria (and their DNA) are inherited from the mother.

* **Symptoms:**

- * Symptoms can vary quite a bit from person to person. Here are some common ones:
 - * **Developmental delays or regression:** Babies might not reach milestones on time or might lose skills they already had.
 - * **Muscle problems:** Weakness, floppiness (hypotonia), stiffness (spasticity), or involuntary movements (dystonia).
 - * **Breathing problems:** Irregular breathing, rapid breathing, or pauses in breathing (apnea).
 - * **Feeding difficulties:** Problems sucking, swallowing, or digesting food, leading to poor weight gain.
 - * **Vision problems:** Abnormal eye movements (nystagmus) or vision loss.
 - * **Seizures:**
 - * **Lactic acidosis:** A buildup of lactic acid in the body, which can cause nausea, vomiting, and fatigue.

* **Risk Factors:**

- * **Family history:** Having a family member with Leigh Syndrome or another mitochondrial disorder.
- * **Maternal inheritance:** If the mother carries a mitochondrial DNA mutation, the risk to her children is higher.
- * **Environmental factors:** Some environmental factors, like certain medications or infections, can trigger symptoms in people with mitochondrial disorders.