

GENETIC DISORDER REPORT

Genetic Disorder: Mitochondrial genetic inheritance disorders

Subclass: Leigh syndrome

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

patient_age: 45

father_age: 70

mother_age: 70

gender: Female

genes_mother_side: Yes

inherited_father: No

maternal_gene: Yes

paternal_gene: No

blood_cell_count: 5000

white_blood_cell_count: 6.7548

respiratory_rate: 15

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, a type of mitochondrial genetic inheritance disorder, in plain language.

Leigh Syndrome: In Simple Medical Terms

Overview:

- * Imagine your body's cells as tiny power plants. These power plants, called mitochondria, have their own DNA.
- * Leigh Syndrome is a severe neurological disorder that occurs when these mitochondrial power plants don't work properly.

Causes:

- * **Faulty Instructions:** Leigh Syndrome is caused by mutations (changes) in genes that control how mitochondria make energy.
- * **Mitochondrial DNA (mtDNA):** This is passed down only from the mother. If the mother has a mutation, it can affect her children.
- * **Nuclear DNA:** This is the DNA in the main part of the cell and is inherited from both parents.
- * **Energy Crisis:** Because of these gene mutations, the mitochondria are unable to produce enough energy for the body's cells.

Symptoms:

The symptoms of Leigh syndrome can vary widely, but common ones include:

- * **Developmental Delays:** Slower than usual progress in reaching developmental milestones like sitting up or walking.
- * **Movement Problems:** Muscle weakness (hypotonia), poor coordination (ataxia), difficulty swallowing (dysphagia).
- * **Breathing Issues:** Irregular breathing patterns, sometimes including periods of rapid breathing (hyperventilation).
- * **Vision Problems:** Optic atrophy (damage to the optic nerve) can lead to vision loss. Nystagmus (shaking of the eyes) is also common.
- * **Seizures:** Epileptic seizures can be a part of the syndrome.
- * **Feeding Difficulties:** Trouble sucking or swallowing, leading to poor weight gain.
- * **Heart Problems:** Cardiomyopathy (enlarged or weakened heart muscle).
- * **Lactic Acidosis:** A buildup of lactic acid in the body, which can cause nausea, vomiting, fatigue, and confusion.
- * **Regression:** Worsening of symptoms during times of stress, illness, or infection.