

GENETIC DISORDER REPORT

Genetic Disorder: Mitochondrial genetic inheritance disorders

Subclass: Leigh syndrome

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

patient_age: 50

father_age: 70

mother_age: 72

gender: Female

genes_mother_side: Yes

inherited_father: No

maternal_gene: Yes

paternal_gene: No

blood_cell_count: 5.54248

white_blood_cell_count: 4.9467

respiratory_rate: 40

heart_rate: 72

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 related to Mitochondrial genetic inheritance disorders in simple terms.

Leigh Syndrome: Explained

Overview:

- * Imagine your body's cells are like little power plants, and mitochondria are the engines inside those power plants.
- * It's like a progressive power outage in the brain, leading to a gradual loss of mental and movement abilities.
- * It usually shows up in infancy or early childhood, but sometimes it can appear later.
- * It is an inherited disorder, meaning that the parents can pass the damaged mitochondrial genes on to their children.

Causes:

- * **Mitochondrial Malfunction:** The primary cause is a problem with the mitochondria's ability to create energy for the body's cells.
- * **Genetic Inheritance:** Leigh syndrome can be caused by mutations in mitochondrial DNA (mtDNA).
 - * **Mitochondrial DNA (mtDNA):** mtDNA is passed down only from the mother. So, if a mother has a mutation, all her children will inherit it.
 - * **Nuclear DNA (nDNA):** nDNA is inherited from both parents. This form is more common. In these cases, both parents may have a mutation.
- * In both cases, if the mitochondrial function is inhibited, the rest of the body suffers the consequences.

Symptoms:

The symptoms of Leigh syndrome can vary from person to person, but they commonly include:

- * **Developmental Delays:** Slow or stalled development milestones like sitting, crawling, or walking.
- * **Muscle Problems:** Weakness (hypotonia), floppiness, stiffness, or difficulty controlling movements.
- * **Movement Problems:** Tremors, involuntary muscle contractions (dystonia), and difficulty with coordination.
- * **Breathing Issues:** Irregular breathing patterns or episodes of apnea (stopping breathing).
- * **Feeding Problems:** Difficulty swallowing, poor weight gain, and vomiting.
- * **Vision Problems:** Abnormal eye movements (nystagmus) or vision loss.
- * **Seizures:** Uncontrolled electrical activity in the brain.