

GENETIC DISORDER REPORT

Genetic Disorder: Mitochondrial genetic inheritance disorders
Subclass: Leigh syndrome

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

patient_age: 20
father_age: 45
mother_age: 45
gender: Female
genes_mother_side: Yes
inherited_father: Yes
maternal_gene: No
paternal_gene: No
blood_cell_count: 5000
white_blood_cell_count: 7000
respiratory_rate: 15
heart_rate: 72
parental_consent: Yes
follow_up: Medium
birth_effects: Yes
folic_acid_intake: Yes
blood_test_result: Abnormal
No_of_previous_abortion: 1

----- DESCRIPTION -----

Okay, let's break down Leigh Syndrome, focusing on the mitochondrial genetic inheritance aspect.

Leigh Syndrome: Explained

- * **Overview:** Leigh syndrome is a severe, progressive neurological disorder that typically appears in early childhood.
- * **Think of it like:** The central nervous system (brain and spinal cord) starts to "break down" or malfunction.
- * **Causes:** The primary cause lies in problems with the *mitochondria*, which are the "powerhouses" of the cell.
- * This malfunction in mitochondria can stem from genetic mutations (changes in DNA) that affect the mitochondria's ability to produce energy.
- * **Crucially:** A significant proportion of Leigh syndrome cases involve *mitochondrial DNA (mtDNA)* inheritance.
- * **Simplified:** Imagine your cells need fuel to run, and the mitochondria are the fuel factories. In Leigh syndrome, these factories malfunction.
- * **Symptoms:** Symptoms can vary a lot from person to person, but common ones include:
 - * **Early Development Problems:** Delayed motor skills (like sitting, crawling, walking), failure to thrive.
 - * **Movement Issues:** Muscle weakness, problems with coordination (ataxia), jerky movements (chorea).
 - * **Neurological Problems:** Seizures, vision problems (e.g., nystagmus - involuntary eye movements).
 - * **Respiratory Issues:** Breathing difficulties, which can sometimes be severe.
 - * **Gastrointestinal Issues:** Vomiting, diarrhea.
 - * **Heart Issues:** Hypertrophic cardiomyopathy (enlarged heart).
 - * **Important:** Symptoms can appear suddenly or gradually worsen over time. During stressful situations, symptoms may become more pronounced.
- * **Risk Factors:**
 - * **Family History:** The biggest risk factor is having a family history of mitochondrial disorders or Leigh syndrome.
 - * **Maternal Inheritance (for mtDNA-related cases):** If a mother carries a mutated mtDNA, there is a risk of passing it to her children.
 - * **Genetic Mutations:** Specific mutations in genes known to cause mitochondrial dysfunction are associated with Leigh syndrome.