

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 our cells. * This malfunction in mitochondria can stem from genetic mutations (changes in DNA) that affect the mtDNA. * **Crucially:** A significant proportion of Leigh syndrome cases involve *mitochondrial DNA (mtDNA)* mutations. * **Simplified:** Imagine your cells need fuel to run, and the mitochondria are the fuel factories. In Leigh syndrome, these factories aren't working properly.
- * **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, and feeding difficulties.
 - * **Movement Issues:** Muscle weakness, problems with coordination (ataxia), jerky movements (dystonia).
 - * **Neurological Problems:** Seizures, vision problems (e.g., nystagmus - involuntary eye movement), and hearing loss.
 - * **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 worsen.
- * **Risk Factors:**
 - * **Family History:** The biggest risk factor is having a family history of mitochondrial disorders or other neurological conditions.
 - * **Maternal Inheritance (for mtDNA-related cases):** If a mother carries a mutated mtDNA, there is a high chance of passing it on to her children.
 - * **Genetic Mutations:** Specific mutations in genes known to cause mitochondrial dysfunction are also associated with Leigh syndrome.