

GENETIC DISORDER REPORT

Genetic Disorder: Mitochondrial genetic inheritance disorders

Subclass: Leigh syndrome

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

patient_age: 30

father_age: 70

mother_age: 70

gender: Female

genes_mother_side: No

inherited_father: Yes

maternal_gene: No

paternal_gene: Yes

blood_cell_count: 5.0888

white_blood_cell_count: 5000

respiratory_rate: 15

heart_rate: 73

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, a type of mitochondrial disorder, into simple medical terms:

Leigh Syndrome: The Battery's Not Working Right

- * **Overview:** Think of your cells like little cities, each needing power to function. Mitochondria are like the power plants that produce energy.
- * **Causes:**
 - * **Mitochondrial Problems:** The root cause is a problem with the mitochondria's ability to produce energy.
 - * **Genetic Inheritance:** Leigh syndrome is most often inherited through genetic mutations.
 - * **Mitochondrial DNA (mtDNA):** Sometimes, the problem is in the DNA within the mitochondria.
 - * **Nuclear DNA:** More commonly, the problem is in the DNA inside the cell's nucleus (nuclear DNA).
 - * **What Goes Wrong:** Because of these gene mutations, the mitochondria cannot produce enough energy.
- * **Symptoms:** The symptoms of Leigh syndrome can vary widely, even among people in the same family.
 - * **Neurological Problems:**
 - * Developmental delays or regression (losing skills they had already learned)
 - * Muscle weakness
 - * Movement problems (like difficulty walking, coordination issues)
 - * Seizures
 - * Problems with vision (like nystagmus - involuntary eye movements)
 - * Swallowing and breathing difficulties
 - * Intellectual disability
 - * **Other Problems:**
 - * Vomiting
 - * Diarrhea
 - * Heart problems (cardiomyopathy)
 - * Kidney problems
 - * Failure to thrive (not gaining weight or growing properly)
- * **Risk Factors:**