

GENETIC DISORDER REPORT

Genetic Disorder: Mitochondrial genetic inheritance disorders
Subclass: Leigh syndrome

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

patient_age: 22
father_age: 60
mother_age: 55
gender: Male
genes_mother_side: Yes
inherited_father: Yes
maternal_gene: Yes
paternal_gene: No
blood_cell_count: 4500
white_blood_cell_count: 6500
respiratory_rate: 11
heart_rate: 88
parental_consent: No
follow_up: High
birth_effects: Yes
folic_acid_intake: Yes
blood_test_result: Slightly abnormal
No_of_previous_abortion: 1

----- DESCRIPTION -----

Okay, let's break down Leigh Syndrome, a type of mitochondrial disorder, in a way that's easy to understand.

****Leigh Syndrome: Simple Medical Explanation****

****Overview:****

Imagine your body's cells as tiny houses, and inside each house is a power plant called the mitochondrion.

****Causes:****

- * ****Mitochondrial Dysfunction:**** Leigh Syndrome is caused by problems with the mitochondria's ability to produce energy.
- * ****Genetic Inheritance:**** The "faulty parts" are due to changes (mutations) in the genes that control mitochondrial function.
 - * ****Mitochondrial DNA (mtDNA) Inheritance:**** Mitochondria have their own DNA (mtDNA) separate from nuclear DNA.
 - * ****Nuclear DNA Inheritance:**** Most of the genes involved in mitochondrial function are actually located on nuclear DNA.

****Symptoms:****

Leigh Syndrome symptoms usually appear in infancy or early childhood, but can appear later. Because

- * ****Developmental Delays/Regression:**** Loss of previously acquired skills like crawling, walking, or talking.
- * ****Movement Problems:**** Difficulty with coordination, muscle weakness, stiff muscles (spasticity), jerky movements.
- * ****Breathing Problems:**** Irregular or shallow breathing, sometimes with periods of stopping breathing.
- * ****Feeding Difficulties:**** Problems sucking, swallowing, or gaining weight.
- * ****Vision Problems:**** Abnormal eye movements (nystagmus), optic atrophy (damage to the optic nerve).
- * ****Brain Problems:**** Irritability, lethargy (excessive tiredness), intellectual disability.
- * ****Heart Problems:**** A heart condition called cardiomyopathy.
- * ****Lactic Acidosis:**** Build-up of lactic acid in the body, which can cause vomiting, abdominal pain, and

****Risk Factors:****