

GENETIC DISORDER REPORT

Genetic Disorder: Mitochondrial genetic inheritance disorders

Subclass: Leigh syndrome

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

patient_age: 85

father_age: 21

mother_age: 18

gender: Male

genes_mother_side: Yes

inherited_father: Yes

maternal_gene: No

paternal_gene: No

blood_cell_count: 4500

white_blood_cell_count: 6500

respiratory_rate: 18

heart_rate: 25

parental_consent: Yes

follow_up: High

birth_effects: Yes

folic_acid_intake: Yes

blood_test_result: Abnormal

No_of_previous_abortion: 9

----- DESCRIPTION -----

Okay, let's break down Leigh syndrome in simple terms:

****Leigh Syndrome: A Power Problem Inside Cells****

****Overview:****

Imagine your body's cells are like tiny power plants. These power plants, called mitochondria, create the

****Causes:****

- * ****Faulty Mitochondria:**** Leigh syndrome is caused by a problem with the mitochondria themselves.
 - * ****Mitochondrial DNA (mtDNA) Inheritance:**** This is passed down only from the mother. If the mother has a mutation, it can affect all her children.
 - * ****Nuclear DNA Inheritance:**** This is more common. Mutations in genes in the cell's nucleus (the DNA outside the mitochondria) can also lead to problems.
- * ****The Result:**** The genetic mutations impair the ability of the mitochondria to produce energy efficiently, leading to various symptoms.

****Symptoms:****

The symptoms of Leigh syndrome usually appear in infancy or early childhood, but can appear later in life.

- * ****Developmental Delays:**** Slower to reach milestones like sitting, crawling, or walking. Loss of skills already learned.
- * ****Movement Problems:**** Difficulty with coordination, muscle weakness, involuntary movements (like shaking or stiffness).
- * ****Breathing Problems:**** Irregular breathing patterns, sometimes with periods of rapid breathing or pauses.
- * ****Feeding Difficulties:**** Trouble sucking, swallowing, or keeping food down, leading to poor weight gain and feeding管困难.
- * ****Vision Problems:**** Abnormal eye movements, vision loss.
- * ****Seizures:****
- * ****Lactic Acidosis:**** A buildup of lactic acid in the body, which can cause nausea, vomiting, fatigue, and confusion.
- * ****Heart Problems:**** (Less common)

****Risk Factors:****