

GENETIC DISORDER REPORT

Genetic Disorder: Mitochondrial genetic inheritance disorders

Subclass: Leigh syndrome

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

patient_age: 45
father_age: 70
mother_age: 70
gender: Female
genes_mother_side: Yes
inherited_father: No
maternal_gene: Yes
paternal_gene: No
blood_cell_count: 5000
white_blood_cell_count: 6.7548
respiratory_rate: 15
heart_rate: 75
parental_consent: None
follow_up: None
birth_effects: None
folic_acid_intake: None
blood_test_result: Not
No_of_previous_abortion: None

----- DESCRIPTION -----

Okay, let's break down Leigh Syndrome, a type of mitochondrial genetic inheritance disorder, in plain language.

****Leigh Syndrome: In Simple Medical Terms****

****Overview:****

- * Imagine your body's cells as tiny power plants. These power plants, called mitochondria, have their own DNA.
- * Leigh Syndrome is a severe neurological disorder that occurs when these mitochondrial power plants malfunction.

****Causes:****

- * ****Faulty Instructions:**** Leigh Syndrome is caused by mutations (changes) in genes that control how mitochondria function.
- * ****Mitochondrial DNA (mtDNA):**** This is passed down only from the mother. If the mother has a mutation, all children will inherit it.
- * ****Nuclear DNA:**** This is the DNA in the main part of the cell and is inherited from both parents.
- * ****Energy Crisis:**** Because of these gene mutations, the mitochondria are unable to produce enough energy for the body's cells.

****Symptoms:****

The symptoms of Leigh syndrome can vary widely, but common ones include:

- * ****Developmental Delays:**** Slower than usual progress in reaching developmental milestones like sitting, walking, and talking.
- * ****Movement Problems:**** Muscle weakness (hypotonia), poor coordination (ataxia), difficulty swallowing.
- * ****Breathing Issues:**** Irregular breathing patterns, sometimes including periods of rapid breathing (hyperpnea) and periods of slow breathing (apnea).
- * ****Vision Problems:**** Optic atrophy (damage to the optic nerve) can lead to vision loss. Nystagmus (involuntary eye movements) is also common.
- * ****Seizures:**** Epileptic seizures can be a part of the syndrome.
- * ****Feeding Difficulties:**** Trouble sucking or swallowing, leading to poor weight gain.
- * ****Heart Problems:**** Cardiomyopathy (enlarged or weakened heart muscle).
- * ****Lactic Acidosis:**** A buildup of lactic acid in the body, which can cause nausea, vomiting, fatigue, and coma.
- * ****Regression:**** Worsening of symptoms during times of stress, illness, or infection.