

GENETIC DISORDER REPORT

Genetic Disorder: Mitochondrial genetic inheritance disorders

Subclass: Cystic fibrosis

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

patient_age: 11

father_age: 41

mother_age: 51

gender: Male

genes_mother_side: Yes

inherited_father: Yes

maternal_gene: No

paternal_gene: No

blood_cell_count: 5.1179092424933112

white_blood_cell_count: 60872167523839368

respiratory_rate: 15

heart_rate: 88

parental_consent: None

follow_up: None

birth_effects: None

folic_acid_intake: None

blood_test_result: Yes

No_of_previous_abortion: None

----- DESCRIPTION -----

Okay, let's break down Cystic Fibrosis (CF) in simple medical terms. It's important to note that CF is NOT a mitochondrial disorder.

Disorder: Cystic Fibrosis (CF)

Overview:

- * Cystic Fibrosis is a genetic disease that primarily affects the lungs, pancreas, liver, intestines, sinuses, and sweat glands.
- * It causes the body to produce abnormally thick and sticky mucus. This mucus clogs the lungs, leading to frequent respiratory infections.

Causes:

- * CF is caused by a mutation (a change) in a gene called the CFTR (Cystic Fibrosis Transmembrane Conductance Regulator).
- * This gene provides instructions for making a protein that controls the movement of salt and water in and out of cells.
- * When the CFTR gene is mutated, the protein doesn't work properly, leading to the thick, sticky mucus.
- * **Inheritance:** You must inherit TWO copies of the mutated CFTR gene – one from each parent – to have CF.

Symptoms:

Symptoms can vary from person to person, but commonly include:

- * **Lung Problems:**
 - * Persistent cough with thick mucus
 - * Wheezing and shortness of breath
 - * Frequent lung infections (pneumonia, bronchitis)
 - * Nasal Congestion and Sinus Infections
- * **Digestive Problems:**
 - * Very salty-tasting skin
 - * Poor growth and weight gain, despite a normal appetite
 - * Diarrhea and constipation