CFTR Δ508 SEQUENCING

Test Description

The CFTR gene is located on the chromosome 7. It is responsible for regulating salt and water movement between cells. CFTR is a transporter gene. CFTR gene provides the information needed to transport sodium and chloride ions across the cell membrane. This in turn controls the flow of water in mucus, sweat, tears, saliva, and digestive enzymes. Its defect causes a wide range of physiological problems. ΔF508 is a class II (defective protein) CFTR mutation.
The CFTR protein when in the proper position opens channels in the cell membrane which release chloride ions out of respiratory epithelial cells. This causes osmosis to draw water out of the cell. The ΔF508 mutation can prevent the CFTR from moving into its proper position in the cell. This may result in increased water retention in the body.

Specimen Description

Sample quality is optimum for the test.
DNA concentration: 88.4 ng/µl

Clinical indication

Not available

RESULTS

The delta 508 Mutation in CFTR gene is not detected in the patient.

Gene	Variant	Zygosity	Inheritance	Result
CFTR [NM_000492.4]	c.1521_1523delCTT; [p.Phe508del [Exon 11]	Homozygous	Autosomal recessive	Absent

INTERPRETATION

Heterozygous Mutant (Carriers)
If two carriers of the gene mate, their offspring will have a 25% chance of having two copies of the mutation. Generally ΔF508 carriers are symptom free, however when combined with other mutations, varying degrees of CF-like symptoms can appear.

Homozygous Mutant (Affected)
Having a homozygous pair of genes with the ΔF508 mutation prevents the CFTR protein from obtaining its normal position in the cell membranes.
This causes increased water retention in cells, and a variety of effects on the body:
- Thicker mucous membranes in many parts of the body.
- Congenital Bilateral Absence of the Vas deferens (CBAVD) due to increased mucus thickness during fetal development.
- Pancreatic insufficiency, due to blockage of the pancreatic duct with mucus.
- This collection of symptoms is called cystic fibrosis

NOTE: ΔF508 is not the only mutation that causes CF.