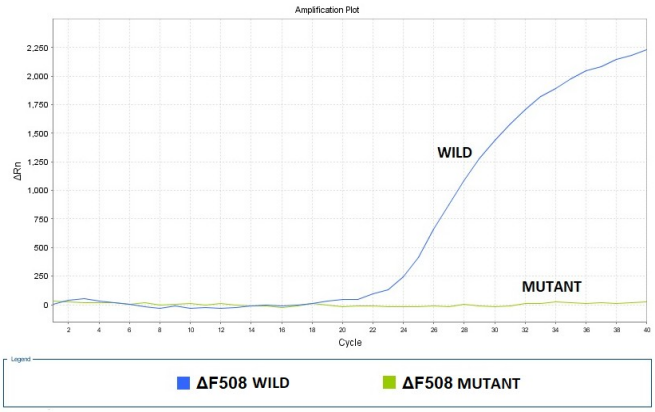
Patient Name	: Ms. SANCHALI	Reg No	: 0372402240075
Age and Sex	: 17 Yrs/F	Collection Center	: SPL-NP-003
Referring Doctor	: Self	Sample Drawn Date	: 24.02.2024
Referring Customer	: NA	Registration Date	: 24.02.2024
Sample type & Vial ID:	WB EDTA – 24465384	Report Date	: 13.03.2024

CYSTIC FIBROSIS - ΔF508 MUTATION ASSAY by real time PCR

TEST DESCRIPTION	RESULT
ΔF508 MUTATION	**Cystic fibrosis ΔF508 mutation NOT DETECTED.**



COMMENTS: Cystic fibrosis ΔF508 mutation is **NOT DETECTED**.
The specimen is a **NORMAL** phenotype.

METHOD: The test is performed by allele-specific real time PCR assay based on Taqman Chemistry. Normal specimen generate single signal corresponding to Wild allele. Mutant specimen generate single signal corresponding to mutant allele. Heterozygous specimen generates two signals which include one wild and one mutant allele.

GENETICS: The **CFTR** gene is located on the seventh chromosome. It is the section of DNA that is responsible for regulating salt and water movement between cells. CFTR is a transporter 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.

POSITIVE VARIANT INTERPRETATION: 1677delTA (c.1545_1546delTA) mutation classified as Class I mutation, caused by frame shift and nonsense mutations. The 1677delTA mutation is related to a severe clinical phenotype among homozygotes and possibly to an increased risk of meconium ileus. Manifestations of cystic fibrosis (CF) include meconium ileus, recurrent pulmonary infections, bronchiectasis, pancreatic insufficiency, biliary cirrhosis, and poor weight gain, among others.
A 2 bp deletion in exon 10 of the CFTR gene, - the 1677delTA mutation, causes a shift in the reading frame and generates a stop codon at position 515. 1677delTA has been reported as a pathogenic allele in homozygous state or in combination with other cystic fibrosis-causing alleles. This mutation has been identified in 3 families with high risk of CF, all from small ethnic group of Georgia.

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