Cystic Fibrosis Gene Sequencing-Sanger (Common Variants)

Patient		Sample		Clinician	
Name	Dummy	Sample Type	Amniotic Fluid	Name	Dr.XXX
Age	24 Years	Sample ID/ Specimen ID	XXX	Hospital	XXX
Gender	NA	Date and Time of Sample Collection	NA	Address	NA
Ethnicity	Unknown	Date and Time of Sample Received	NA	Clinical Information	Dummy, a 24 year old female with GA of 24 weeks 4 days. Her scan report shows foetal ascites and evidence of echogenic areas in the intervoxel region ? Meconium peritonitis.
Place	XXX	Date and Time of Sample Reported	NA		
Phone No./ Email ID	XXX XXX				

Clinical History

Dummy, a 24 year old female with GA of 24 weeks 4 days. Her scan report shows foetal ascites and evidence of echogenic areas in the interbowel region ? Meconium peritonitis

Results

-Negative-

(No Common pathogenic variants detected for the 8 common variants of *CFTR* gene)

Key Findings

Gene	Variant*	Exon/Intron	Zygosity	Inheritance	Result
CFTR (NM_000492.4)	c.1521_1523delTCT; p.Phe508del	Exon-11	Reference	Autosomal Recessive	**The individual does not carry copy of the tested variants.**
	c.1393-1G>A; p.?	Intron			
	c.1624G>T; p.Gly542Ter	Exon-12			
	c.1652G>A; p.Gly551Asp	Exon-12			
	c.1657C>T; p.Arg553Ter	Exon-12			
	c.350G>A; p.Arg117His	Exon-4			
	c.1792_1798delAAAACTA p.Lys598GlyfsX11	Exon-14			
	c.1029del p.Phe342_Cys343insTer	Exon-8			

Genetic test results are reported based on the recommendations of American College of Medical Genetics.

Note: No significant maternal cell contamination is detected in Af of Dummy.
Maternal cell contamination (MCC) was ruled out using STR markers.

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Gene Summary

This gene encodes a member of the ATP-binding cassette (ABC) transporter superfamily. The encoded protein functions as a chloride channel, making it unique among members of this protein family, and controls ion and water secretion and absorption in epithelial tissues. Channel activation is mediated by cycles of regulatory domain phosphorylation, ATP-binding by the nucleotide-binding domains, and ATP hydrolysis. Mutations in this gene cause cystic fibrosis, the most common lethal genetic disorder in populations of Northern European descent. The most frequently occurring mutation in cystic fibrosis, DeltaF508, results in impaired folding and trafficking of the encoded protein. Multiple pseudogenes have been identified in the human genome.

Test Description:

Cystic fibrosis is one of the most common, fatal genetic diseases and is caused by mutations in the cystic fibrosis transmembrane regulator (CFTR) gene. While hundreds of mutations have been discovered in CFTR, sequencing can be cost prohibitive when running large numbers of samples using Sanger sequencing. CFTR Panel provides a simple and affordable CFTR gene sequencing with dedicated analysis workflows for optimal variant calling. This panel analyzes all coding exons, intron-exon boundaries and UTRs of CFTR with 102 amplicons, including the >160 mutations from CFTR2.org research database.

Cystic Fibrosis (CF) is an autosomal recessive disorder impacting more than 70,000 children and adults worldwide.1 Defects in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene lead to disruption in normal protein expression causing dysregulation of ion chloride channels affecting respiratory, digestive, and reproductive systems. Cystic Fibrosis affects both males and females, and people of all racial groups; however, it is most prevalent in Caucasians of Northern European descent.

Recommendations:

- The results should be interpreted in the context of the patient's medical evaluation**.** Correlation of the genetic findings with the clinical condition of the patient is required to arrive at accurate diagnosis, prognosis or for therapeutic decisions.
- Large deletions/duplication that eliminates most or all of the coding sequence will also not be detected by this assay. In addition, mutations in other genes associated with *CFTR* gene will not be identified.

Test Methodology:

- *CFTR* are identified by analysis of the PCR products using automated DNA sanger-sequencing technique.
- There are several mutations in *CFTR* -insertion, deletion, du plication, point mutations. Among all of these, the eight common mutations are- c.1521_1523delTCT, c.1393-1G>A, c.1624G>T, c.1652G>A, c.1657C>T, c.350G>A, c.1792_1798delAAAACTA, c.1029del, account for mo s t of the mutations in *CFTR* mutated individuals.
- This test can be useful in the patients who have symptoms, or parental carrier testing in order to diagnose prenatal.

References:

1. Cystic Fibrosis Foundation website, What is Cystic Fibrosis? https://www.cff.org/What-is-CF/CF-Genetics/
2. Cystic Fibrosis Mutation Database http://www.genet.sickkids.on.ca/app
3. Hendrix et. al. Newborn Screening Quality Assurance Program for CFTR Mutation Detection and Gene Sequencing to Identify Cystic Fibrosis Journal of Inborn Errors of Metabolism & Screening. 2016, Volume 4.

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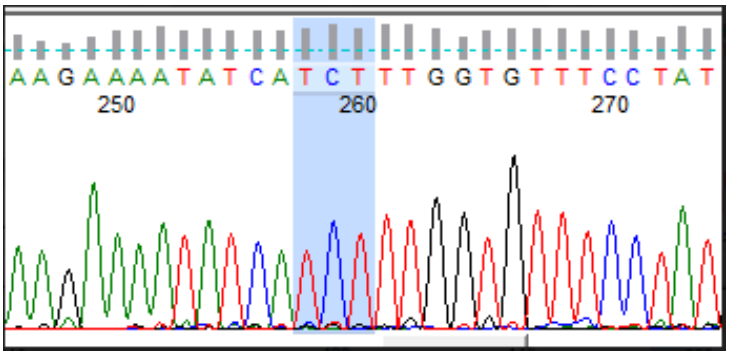
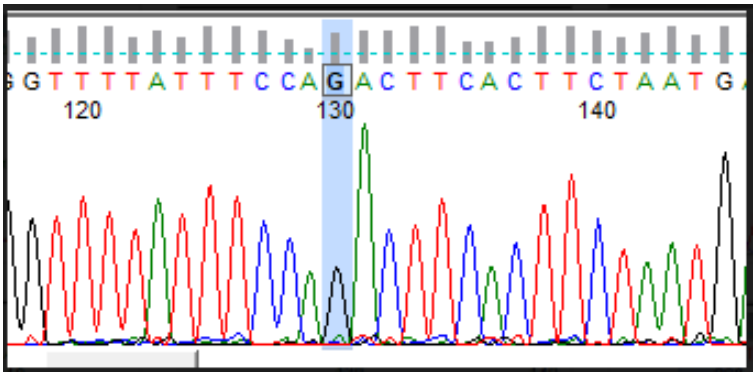
4. Grangeia A, Alves S, Goncalves L, Santos AC, Barros H, Barros A, Carvalho F, Moura C. Spectrum of CFTR gene sequence variants in a northern Portugal population. Pulmonology 2018;24(1):3-9.
5. Schwarz JM, Cooper DN, Schuelke M and Seelow D. Mutationtaster2: mutation prediction for the deep sequencing age. Nature Methods.2014; 11: 361-62.

Analysis Result:

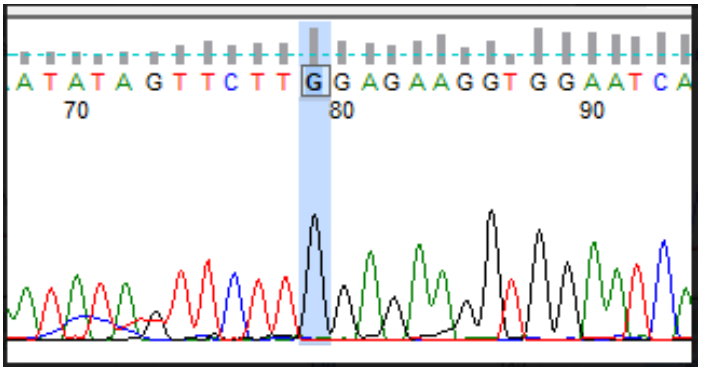
1. CFTR: c.1521_1523delTCT



2. CFTR:c.1393-1G>A



3. CFTR:c.1624G>T



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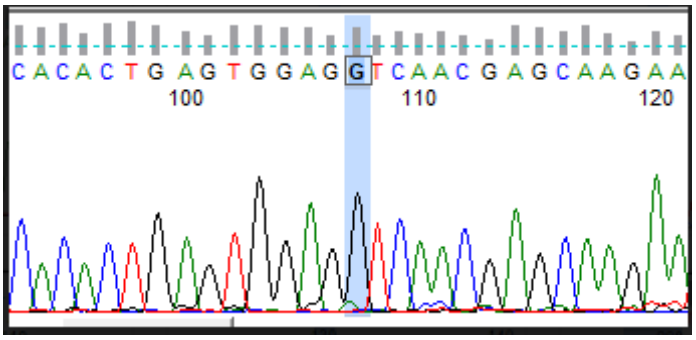
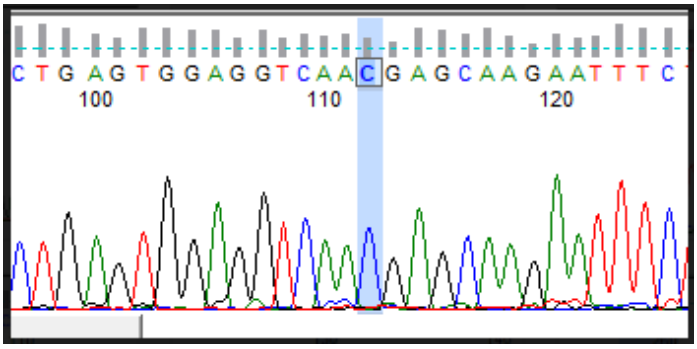
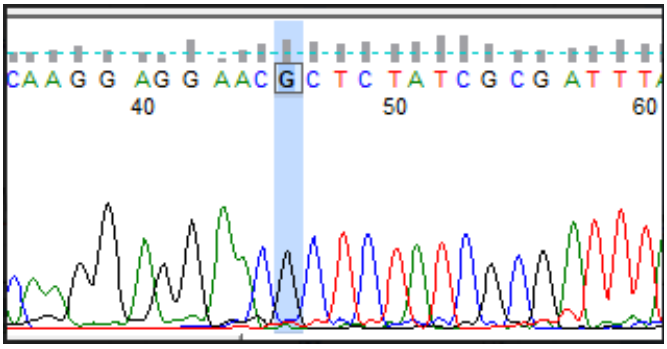
4. CFTR:c.1652G>A



5. CFTR:c.1657C>T



6. CFTR:c.350G>A



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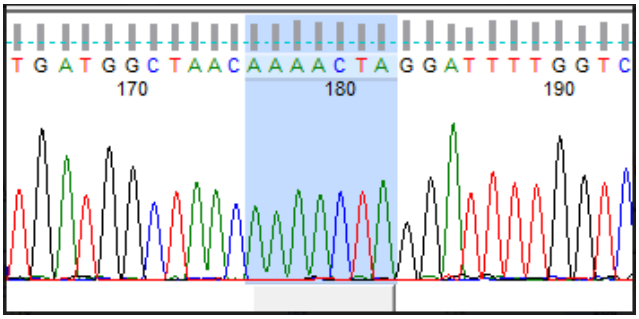
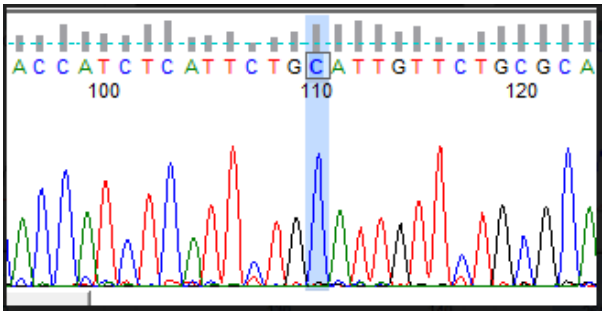
7. CFTR:c.1792_1798delAAAACTA



8. CFTR:c.1029del



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