

CFTR panel

Hereditary variants profiling in Cystic Fibrosis



CFTR panel

4bases CFTR panel kit is a CE-IVD kit for the identification of mutations in CFTR gene (Table 1) through a molecular protocol for Next Generation Sequencing (NGS) technologies on Nanopore platforms.

The kit is validated for germline analysis (SNVs, CNVs) of DNA extracted from blood samples and tissues (fresh, frozen or FFPE).

HIGHLIGHTS

- Coverage of the entire coding region and ± 25 bp beyond the flanking regions
- 1st and 2nd level analysis
- Coverage of all variants present in the CFTR2 database
- CNV analysis
- Poly-T and poly-TG detection
- All the known and unknown of the more than 22000 causative mutations in CFTR gene can be detected with a single test

TECHNOLOGY

CFTR panel kit contains all reagents needed for the amplification of CFTR gene and library multiplexing for NGS sequencing using Nanopore platforms.

WORKFLOW

The CFTR panel kit is part of an amplicon-based family solution of 4bases molecular technologies. Its accuracy and specificity in detecting genetic variants (i.e. SNVs, CNVs), coupled with its straightforward protocol (less than 3 hours hands-on-time), are the key of its reliability.

Thanks to 4bases proprietary analysis platform, 4eVAR, the clinical results are easily and efficiently accessible through a validated internal workflow.

SOFTWARE ANALYSIS

4eVAR is our proprietary analysis software. The analysis is designed based on the characteristics and technologies of the kit used, in order to increase accuracy of results, and to have the complete control on the entire process.

A complete report including 1st level analysis, SNVs and CNVs with ACMG and AMP classification for variant interpretation is given per each sample. Specific links for wider yet complete classification of detected variants are also provided, together with quality metrics per each sample in the run.

VALIDATION

The CFTR panel kit has been validated both internally and in clinical laboratories, and the whole protocol was tested on DNA standards and clinical samples (Figure 1).

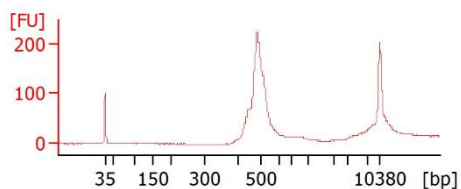


Figure 1. Agilent Bioanalyzer profile of a final CFTR panel library.

Table 1: Samples per CFTR panel sequencing run on Nanopore sequencers.

Flow Cell	Samples per Flow Cell (Coverage 100x)	Nanopore sequencer	Flow Cells per sequencer*
FLO-FLG114	4	Flongle	1
FLO-MIN114	48	MinION	1
		GridION	Up to 5
FLO-PRO114M	240	PromethION 2/P2 Solo	Up to 2
		PromethION 24	Up to 24
		PromethION 48	Up to 48

* Multiple Flow Cells can be used during the same run session, either with different libraries or with the same library to increase sequencing depth.

ORDERING INFORMATION **

	Product	REF
LIBRARY AMPLIFICATION REAGENTS	CFTR panel	H1060-16 (16 test)
	CFTR panel	H1060-96 (96 test)
MULTIPLEXING REAGENTS	BARCODE ONT Set 16	R8001-16 (16 test)
	BARCODE ONT Set 96	R8002-96 (96 test)
NANOPORE ADAPTERS	NANO ADAPTER	M1050-6 (6 sequencing run)
		M1050-24 (24 sequencing run)

** The reagents required for sequencing on Nanopore platforms have to be purchased separately: contact 4bases for further information.

CONTACT US

For more detailed information regarding validated solutions for automatic library preparation, please contact us at info@4bases.it.

For the target regions of CFTR gene (.bed file), please contact us at support@4bases.it.



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