CFTR panel

Hereditary variants profiling in Cystic Fibrosis

4bases

When Precision Matters

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 **Oxford Nanopore Technologies**' 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 both the entire coding region with ± 25 bp beyond the flanking regions, and deep intronic variants
- 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 **Oxford Nanopore Technologies**' 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.

Table 1: Samples per **CFTR panel** sequencing run on Oxford Nanopore Technologies' sequencers.

Flow Cell	Samples per Flow Cell (Coverage 100x)	Oxford Nanopore sequencer	Flow Cells per sequencer*
FLO-FLG114	2	Flongle	1
FLO-MIN114	32	MinION	1
		GridION	Up to 5
FLO-PRO114M	128	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 (GridION, PromethION), either with different libraries or with the same library to increase sequencing depth.

PLEASE NOTE: The volumes of the reagents in the kit are calculated to allow the subdivision into multiples of 8 samples per analysis session. Dividing the kit differently will decrease the total number of tests that can be performed. For different settings, please contact us at support@4bases.ch.

PLEASE NOTE: The number of multiplexable samples and coverage depend on starting gDNA degradation, presence of contaminants, external factors, library loading procedure.

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)
OXFORD NANOPORE ADAPTERS	NANO ADAPTER	**M1050-6** (6 sequencing runs)
		M1050-24 (24 sequencing runs)

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

PLEASE NOTE: Should you need to run more than/multiple of 96 samples in the same library, please contact us.

- For more detailed information regarding validated solutions for automatic library preparation, please contact us at **info@4bases.ch**
- For the target regions of *CFTR* genes (.bed file), please contact us at **support@4bases.ch**