CFTR panel

For the diagnosis of hereditary variants in Cystic Fibrosis

CE IVD

CFTR panel

CFTR Panel is a CE-IVD kit for the molecular profiling of CFTR gene.
Mutations in the CFTR gene cause the CFTR protein to malfunction or not produce, leading to a buildup of thick mucus, which in turn leads to persistent lung infections and multi-organ complications.
More than 2200 causative mutations of Cystic Fibrosis have been described and with the CFTR panel kit it is possible to detect all variants, known and unknown, with a single test.

The importance of an NGS test is reflected in the timeliness of diagnosis in newborn screening, in the diagnosis of Cystic Fibrosis in asymptomatic patients, in the diagnosis of CFTR-related diseases and in the possibility of directing the therapeutic path. CFTR panel is a kit for the analysis of the CFTR gene through a molecular protocol based on NGS technologies. The kit is validated for germline analysis (SNPs, indels, CNVs) of DNA extracted from body tissues (blood or others).

TECHNOLOGY

CFTR panel kit contains all reagents required for the preparation of a specific bidirectional library of amplicons designed for the NGS analysis using Ion Torrent.

WORKFLOW

The CFTR panel kit is part of a DNA-to-variant solution that offers streamlined content, easy-to-perform library preparation, push-button sequencing systems, and simplified data analysis.

Library preparation follows a straightforward, PCR-based protocol that can be completed in as little as 5 hours, with < 1.5 hours hands-on time. Resulting libraries can be normalized, pooled, and then loaded on to a flow cell for sequencing.

Prepared libraries are sequenced on any compatible Ion Torrent sequencers.

SOFTWARE ANALYSIS

CFTR panel is part of an integrated solution with a dedicated and validated CE-IVD software.

The analysis includes a first level filter including all variants of the CFTR2 reference database for cystic fibrosis.

We support the implementation and customization of the analysis based on the needs of the laboratory.

SAMPLE PER RUN

Instrument	Samples per run*
	Germline
Ion 316™ Chip/Ion 510™ Chip	14
Ion 318™ Chip/Ion 520™ Chip	28
Ion 530™ Chip	96
Ion PI™ Chip/Ion 540™ Chip	384

**the estimated maximum number of samples per chip assumes a reading depth of 300x for the germline. The optimal number of samples can be empirically estimated on the local setup.*

ORDERING INFORMATION

Product**	REF
CFTR panel	H1060-16 (16 test)
CFTR panel	H1060-96 (96 test)
Barcode	
Barcode Set 1-16	6001
Barcode Set 17-32	6002
Barcode Set 33-48	R6003
Barcode Set 49-64	R6004

***the kit is also available in its version only for research use (RUO).*