

# FH panel

For the genetic characterization of Familial Hypercholesterolemia



## FH panel

FH Panel is the NGS-bases kit for the molecular profiling of Familial Hypercholesterolemia (FH).

FH panel allow complete characterization of 9 genes involved in Familial Hypercholesterolemia and the identification of polygenic FH SNPs and SNPs for the prediction of statin response.

Familial hypercholesterolemia is a genetic disorder that affects about 1 in 250 people and increases the likelihood of having coronary heart disease at a younger age.

The importance of an NGS test is reflected in the timeliness of diagnosis and in the possibility of predicting the therapeutic path.

FH panel is a kit for the analysis of the FH gene through a molecular protocol based on NGS technologies. The kit is validated for germline analysis (SNPs, indels) of DNA extracted from body tissues (blood or others).

## TECHNOLOGY

FH panel kit contains all reagents required for the preparation of a specific bidirectional library of amplicons designed for the NGS analysis using Illumina sequencers.

## WORKFLOW

The FH 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 Illumina sequencers.

## SOFTWARE ANALYSIS

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

Table 1: List of target genes in FH panel

APOB	LDLR	PCSK9	LDLRAP1	STAP1
APOE	ABCG5	ABCG8	LIPA	

## SAMPLE PER RUN

Instrument	Samples per run*
	Germline
iSeq 100 i1 kit (300-cycles)	64
MiniSeq Mid Output Kit (300-cycles)	128
MiniSeq High Output Kit (300-cycles)	416
MiSeq Nano Kit v2 (300-cycles)/(500-cycles)	16
MiSeq Micro Kit v2 (300-cycles)	64
MiSeq Kit v2 (300-cycles)/(500-cycles)	260
MiSeq Kit v3 (600-cycles)	416

\*the estimated maximum number of samples per cartridge 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
FH panel	H1111-16 (16 test)
FH panel	H1111-96 (96 test)
Index	
Index Set 16	3006
Index Set 96	3007
Index Set 384	3009X

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