

THALASSEMIA panel

For the screening variants causing alpha and beta Thalassemia



THALASSEMIA panel

THALASSEMIA Panel is a novel NGS Medical Device for the molecular profiling of variants causing alpha and beta Thalassemia.

Thalassemia is the name for a group of inherited characterized by abnormal hemoglobin production. THALASSEMIA panel is a kit for the analysis of the HBB, HBA1, HBA2, HBD, HBG1, HBG2, HS-40 and LCRB genes through a molecular protocol based on NGS technologies. The kit is validated for germline analysis (SNPs, indels) of DNA extracted from blood samples.

TECHNOLOGY

THALASSEMIA 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 THALASSEMIA 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.

Table 1: Main CNV detected:

Alpha globin deletions	Beta globin deletions
--SEA	Chinese
--FIL	Filipino
--THAI	Yunnanese
-(a)20.5	Taiwanese
--MED	SEA-HPFH
-(a)21.9	Hb-Lepore Boston
-(a)27.6	Hb-Lepore Baltimore
-(a)3.7	290bp-del
-(a)4.2	B619-del
`	δβSicilia

SAMPLE PER RUN

Instrument	Samples per run*
	Germline
iSeq 100 i1 kit (300-cycles)	28
MiniSeq Mid Output Kit (300-cycles)	96
MiniSeq High Output Kit (300-cycles)	168
MiSeq Nano Kit v2 (300-cycles)/(500-cycles)	6
MiSeq Micro Kit v2 (300-cycles)	28
MiSeq Kit v2 (300-cycles)/(500-cycles)	96
MiSeq Kit v3 (600-cycles)	192

*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
THALASSEMIA panel	H1090-16 (16 test)
THALASSEMIA panel	H1090-96 (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).

Table 2: List of target genes in THALASSEMIA panel

HBB	HBA1	HBA2	POLR3K	HS-40
HBZ	HBZP1	HBAP1	HBA2	HBA1
HBQ1	LUC7L	AXIN1	LCRB	HBG2
HBG1	HBD	HBB	3'HS1	

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.