

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 Ion Torrent 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 Ion Torrent 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
Ion 316™ Chip/Ion 510™ Chip	18
Ion 318™ Chip/Ion 520™ Chip	35
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
THALASSEMIA panel	H1090-16 (16 test)
THALASSEMIA panel	H1090-96 (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).

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.