

THALASSEMIA panel



For the screening variants causing alpha and beta Thalassemia

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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 solution

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.

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

TARGET REGIONS

Entire genes:

- HBB, HBA1, HBA2
- Alpha globin cluster related
 - POLR3K, HS-40, HBZ, HBZP1, HBAP1, HBA2, HBA1, HBQ1, LUC7L, AXIN1
- Beta globin cluster related
 - LCRB, HBG2, HBG1, HBD, HBB, 3'HS1

Table 1: Principal 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	Sample per run
MiSeq Nano Kit v2 (300-cycles)	6
MiSeq Nano Kit v2 (500-cycles)	6
MiSeq Micro Kit v2 (300-cycles)	28
MiSeq Kit v2 (300-cycles)	>96
MiSeq Kit v2 (500-cycles)	>96
MiSeq Kit v3 (600-cycles)	>96
MiniSeq Mid Output Kit (300-cycles)	57
MiniSeq High Output Kit (300-cycles)	>96
iSeq 100 i1 kit (300-cycles)	28
NextSeq 550 Mid-Output Kit	>384
NextSeq 550High-Output Kit	>384
Ion 314™ Chip	4
Ion 316™ Chip	18
Ion 318™ Chip/Ion 520™ Chip	35
Ion 530™ Chip	>96
Ion PI™ Chip/Ion 540™ Chip	>384

**the maximum number of samples per cartridge/chip estimated assuming an average depth of 300x for germline samples. The optimal number of samples must be empirically determined on local setups.*

Ordering Information

Product	REF
THALASSEMIA panel	H1090-16 (16 test)
THALASSEMIA panel	H1090-96 (96 test)
<i>For Illumina instruments**</i>	
Index Set series RUO - CE	3000
<i>For Ion Torrent instruments**</i>	
Barcode series RUO - CE	6000

***for the complete list of available indexes and barcodes, refer to Flyer_Index-Barcode*