

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. 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 Nanopore 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 Nanopore 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
	$\delta\beta$ Sicilia

SAMPLE PER RUN

Flowcell	Samples per flowcell 100x	Instrument	Flowcells per instrument*
FLO-MIN114	128	MinION	1
		GridION	Up to 5
FLO-PRO114M	640	PromethION 2\2 Solo	Up to 2
		PromethION 24	Up to 24
		PromethION 48	Up to 48

*Multiple flow cells can be used during the same run session, with different libraries, or same library increasing depth

ORDERING INFORMATION

Product	REF
THALASSEMIA panel	H1090-16 (16 test)
THALASSEMIA panel	H1090-96 (96 test)
BARCODE	
BARCODE ONT Set 16	R8001-16 (96 test)
BARCODE ONT Set 96	R8002-96 (96 test)
ADAPTER	
NANO ADAPTER	M1050-6

List of target genes in THALASSEMIA panel

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

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.