

RETI pro

For the screening of Retinopathies



RETI pro

RETI pro is a novel NGS medical device for the molecular profiling of the variants involved in the Retinopathies.

RETI solution

RETI pro is a kit for the analysis of 107 genes related to retinopathies, through a molecular protocol based on NGS technologies. The kit is validated for germline analysis (SNPs, indels, CNVs) of DNA extracted from blood or body tissues samples.

RETI pro kit contains all reagents required for the preparation of the capture of specifically designed probes and for the NGS analysis using Illumina.

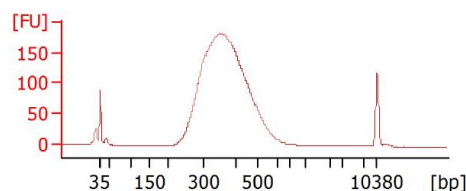
WORKFLOW

Library preparation follows a straightforward, capture-based protocol that can be completed in as little as 24 hours, with < 3 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.

Validation

To demonstrate assay capabilities, clinical samples were run in a clinical setting. DNA quality and quantity of the libraries prepared were verified using Qubit and Agilent Bioanalyzer.

Figure 1: RETI pro profiles examples



SAMPLE PER RUN

Instrument	Sample per run
MiSeq Micro Kit v2 (300-cycles)	8
MiSeq Kit v2 (300-cycles)	24
MiSeq Kit v2 (500-cycles)	24
MiSeq Kit v3 (600-cycles)	48
MiniSeq Mid Output Kit (300-cycles)	16
MiniSeq High Output Kit (300-cycles)	48
iSeq 100 i1 kit (300-cycles)	8
NextSeq 550 Mid-Output Kit	268
NextSeq 550High-Output Kit	268

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

Ordering Information

Product	REF
RETI pro	C3180-16 (16 test)
RETI pro	C3180-96 (96 test)
<i>For Illumina instruments</i>	
UDI Primers Set A (96 test)	7001
UDI Primers Set B (96 test)	7002
UDI Primers Set C (96 test)	7003
UDI Primers Set D (96 test)	7004
UDI Primers Set 16 (16 test)	7005

Table 1. List of target genes in RETI pro

ABCA4	CACNA1F	CNNM4	GRM6	KLHL7	OPA1	PRPF3	RIMS1	SAG	ZNF513
ADAM9	CACNA2D4	CRB1	GRK1	LCA5	OTX2	PRPF31	RLBP1	SEMA4A	
ADAMTS18	CDHR1	CRX	GUCA1A	LRAT	PCARE	PRPF6	ROM1	SLC24A1	
AIPL1	CEP290	CYP4V2	GUCA1B	LRP5	PDE6A	PRPF8	RP1	SNRNP200	
ARL6	CERKL	DHDDS	GUCY2D	MAK	PDE6B	PRPH2	RP11L1	SPATA7	
ATF6	CFAP418	EYS	IDH3B	MERTK	PDE6C	RAX2	RP2	TOPORS	
BBS1	CHM	FAM161A	IMPDH1	NDP	PDE6D	RBP3	RP9	TRPM1	
BEST1	CLRN1	FSCN2	IMPG2	NMNAT1	PDE6G	RD3	RPE65	TSPAN12	
C2ORF71	CNGA1	FZD4	INPP5E	NR2E3	PDE6H	RDH12	RPGR	TTC8	
C8ORF37	CNGA3	GNAT1	IQCB1	NRL	PITPNM3	RDH5	RPGRIP1	TULP1	
CA4	CNGB1	GNAT2	KCNJ13	NYX	PRCD	RGR	RRH	UNC119	
CABP4	CNGB3	GPR179	KCNV2	OFD1	PROM1	RHO	RS1	USH2A	