

RETINA_{pro}

For the identification of the variants involved in the Retinopathies

RETINA Pro is a **capture-based kit** for the molecular profiling of the variants involved in the Retinopathies, through a protocol based on Next Generation Sequencing technologies (NGS).

Inherited retinopathies are among the most genetically heterogeneous ocular disorders, requiring accurate and reliable molecular analysis for diagnosis and clinical research.

The kit is validated for germline analysis (SNPs, indels, CNVs) of DNA extracted from tissues, blood or body fluids.

RETINA pro is a kit for the analysis of 109 genes

ABCA4	CACNA1F	CNNM4	GRM6	KLHL7	OPA1	PRPF3	PROM1
RIMS1	SAG	ZNF513	ADAM9	CACNA2D4	CRB1	GRK1	RHO
LCA5	OTX2	PRPF31	RLBP1	SEMA4A	ADAMTS18	CDHR1	RS1
CRX	GUCA1A	LRAT	PCARE	PRPF6	ROM1	SLC24A1	USH2A
AIPL1	CEP290	CYP4V2	GUCA1B	LRP5	PDE6A	PRPF8	OFD1
RP1	SNRNP200	ARL6	CERKL	DHDDS	GUCY2D	MAK	KCNV2
PDE6B	PRPH2	RP1L1	SPATA7	ATF6	CFAP418	EYS	GPR179
IDH3B	MERTK	PDE6C	RAX2	RP2	TOPORS	BBS1	CNGB3
CHM	FAM161A	IMPDH1	NDP	PDE6D	RBP3	RP9	CABP4
TRPM1	BEST1	CLRN1	FSCN2	IMPG2	NMNAT1	PDE6G	UNC119
RD3	RPE65	TSPAN12	C2ORF71	CNGA1	FZD4	INPP5E	RRH
NR2E3	PDE6H	RDH12	RPGR	TTC8	C8ORF37	CNGA3	RGR
GNAT1	IQCB1	NRL	PITPNM3	RDH5	RPGRIP1	TULP1	PRCD
CA4	CNGB1	GNAT2	KCNJ13	NYX			

From DNA to Final data in less than 2.5 days with less than 3 hours hands-on time.



From gDNA



PCR UDI



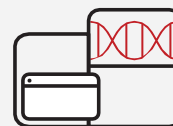
Hybridization



Enrichment PCR



Final library



NGS



to FINAL DATA

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Specifications

Sample type	DNA extracted from tissues, blood or body fluids
Input	Minimum of 50 ng DNA per sample
Panel size	About 310 Kb
Variant called	SNPS, indels, CNVs
Compatible Instruments	Illumina, Oxford Nanopore Technologies, Element Biosciences, MGI, GeneMind
Data Analysis	4eVAR

Cod. kit

Product	Cod.	Tests number
RETINA pro	RC3180, RDC3180	16
RETINA pro	RC3180, RDC3180	96

4eVAR: Comprehensive output files for analysis and reporting



- Quality metrics
- Alignment
- CNVs status
- Full variant table
- Genotype
- Coverage
- Mitochondrial analysis
- Customizable report

Data Analysis

Product	Cod.	Tests number
4eVAR	Series A6060-C3180-16	16
4eVAR	Series A6060-C3180-96	96

If you wish to learn more:



Contact us at: info@4bases.ch
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