NEPHI screen

NEPHI screen is a kit for the analysis of the NF1, SPRED1 NF2, LZTR1 and SMARCB1 genes through a molecular protocol based on NGS technologies. The kit is validated for germline analysis (SNPs, indels,CNVs) of DNA extracted from body tissues (blood or other) and somatic analysis of DNA extracted from fresh tissue and protein.

NEPHI screen 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.

KEY FEATURES

- **REF: R2050-16 (16 test)**
- **REF: R2050-48 (48 test)**
- Genes targeted (N1): NF1, SPRED1
- Genes targeted (N2): NF2, LZTR1, SMARCB1
- Application N1: germline analysis
- Application N2: somatic analysis
- Number of pools N1: 3
- Number of pools N2: 3
- Panel size N1: 19 kb
- Panel size N2: 12.1 kb
- Input DNA: 20ng/reaction

SAMPLES/RUN*

	N1 Germline	N2 Somatic
MiSeq Nano Kit v2 (300-cycles)	25	3
Nano Kit v2 (500-cycles)	38	4
Micro Kit v2 (300-cycles)	>96	10
Kit v2 (300-cycles)	>96	38
Kit v2 (500-cycles)	>384	55
Kit v3 (600-cycles)	>384	90
MiniSeq Mid Output Kit (300-cycles)	>96	20
High Output Kit (300-cycles)	>384	60
iSeq 100 i1 kit (300-cycles)	>384	10
NextSeq 550 Mid-Output Kit	>96	30
High-Output Kit	>384	>384
Ion 314™ Chip	10	1
Ion 316™ Chip	50	5
Ion 318™ Chip/Ion 520™ Chip	>96	10
Ion 530™ Chip	>96	38
Ion PI™ Chip/Ion 540™ Chip	>384	>96

**the maximum number of samples per cartridge/chip estimated assuming an average depth of 300x for germline and 5000x for somatic analysis. The optimal number of samples must be empirically determined on local setups.*

INDEXES/BARCODES

For Illumina instruments	*For Ion Torrent instruments*
Index set A (6x4) REF: R5001	Barcode 1-16 REF: R4001
Index set B (6x4) REF: R5002	Barcode 17-32 REF: R4002

For ordering info please contact info@4bases.ch

4bases Portfolio

	Kit ID	Cod	Size (n° test)	CLINICAL APPLICATIONS	TARGETS	SAMPLES TYPE
Profiling of HotSpots somatic mutations (SNPs, indels) in cancer tissues	LUNG panel	R1000-16 R1000-48	16 48	NSCLC treatment	EGFR (exons 18, 19, 20, 21) KRAS (exons 2, 3, 4)	Tumor DNA tissue (fresh, frozen, FFPE, FNA, etc.) - Somatic analysis (SNPs, indels)
	COLON panel	R1010-16 R1010-48	16 48	mCRC treatment	KRAS (exons 2, 3, 4) NRAS (exons 2, 3, 4) BRAF (exon 15)	
	BENKit panel	R1020-16 R1020-48	16 48	MultiCancers treatment	KRAS (exons 2, 3, 4) NRAS (exons 2, 3, 4) BRAF (exon 11, 15) EGFR (exons 18, 19, 20, 21) PIK3CA (exons 10, 21)	
	THYRO-ID panel	R1030-16 R1030-48	16 48	Mutations profiling of Papillary Thyroid Carcinoma	KRAS (exons 2, 3, 4) NRAS (exons 2, 3, 4) HRAS (exons 2, 3) BRAF (exon 15) TP53 (exons 4, 5, 6, 7, 8, 9) NOTCH1 (exons 26, 27) PTEN (exons 5, 6, 7, 8) CDKN2A (exons 1, 2) EGFR (exons 18, 19, 20, 21) AKT1 (exon 1) CTNNB1 (exon 1) PIK3CA (exons 10, 21) TSHR (exons 6, 8, 9) hTERT (promoter)	
Full-gene sequencing (all CDS + flanking regions) for germline and/or somatic analysis	BRaCA screen	R2000-16 R2000-48	16 48	Hereditary and Somatic Variants profiling in Breast and Ovary cancer	BRCA1, BRCA2, TP53	Tumor DNA tissue (fresh, frozen, FFPE, FNA, etc.) or other (blood) - Somatic analysis (SNPs, indels) Germline analysis (SNPs, indels, CNVs)
	HECO screen	R2002-16 R2002-48	16 48	Germline Variants profiling in Hereditary nonpolyposis colorectal cancer (HNPCC)	APC, EPCAM, MLH1, MSH2, MSH6, MUTYH, PMS2, STK11	DNA from body tissues (blood or other) - Somatic analysis* (SNPs, indels) Germline analysis (SNPs, indels, CNVs)
	BRaVO screen	R2001-16 R2001-48	16 48	Germline Variants profiling in Hereditary Breast and Ovarian Cancer Syndrome (HBOC)	ATM, BRCA1, BRCA2, CDH1, CHEK2, PALB2, PTEN, TP53	
	HEVA screen	R2010-16 R2010-48	16 48	Hereditary Variants profiling in Breast and Ovary, Lynch Syndrome, and other cancer-related diseases	ATM, APC, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, EPCAM, MLH1, MSH2, MSH6, MUTYH, NBN, PALB2, PMS2, PTEN, RAD50, RAD51C, RAD51D, STK11, TP53	
	CFTR screen	R2030-16 R2030-48	16 48	Cystic fibrosis	CFTR	
	NEPHI screen	R2050-16 R2050-48	16 48	Neurofibromatosis (type 1, 2*) Noonan syndrome (type 1, 2)* Legius syndrome* Schwannomatosis*	NF1, SPRED1 NF2*, LZTR1*, SMARCB1*	
	IVF screen	R2040-16 R2040-48	16 48	Hereditary Variants profiling in genetic-related diseases	BDNF, BCHE, ATM, HBB, BLM, ASPA, CHM, GLA, MEFV, FANCC, G6PC, GALT, GBA, GCDH, GJB2, OTOF, PJVK (DFNB59), HFE2, FPN1, HFE, TFR2, ALDOB, RS1, GALC, GLB1, IDUA, SMPD1, NPC2, NPC1, NBN, FSHR, PAH, PKD2, PKHD1, GAA, M2/ANXA5, ELP1 (IKAP), DHCR7, EPB42, ANK1, HEXA, MPL, MTHFR, F5, F2, APoE, PAI1, TH, ATP7B, PEX1, AZFa, AZFb, AZFc, MUTYH, BRCA2, BRCA1, APC, SMN2, SMN1, F13A1, CFTR, DMD	
Extended screening of cDNA fusion transcripts	FUSION screen	R2020-16 R2020-48	16 48	Fusion transcripts and expression imbalances between the 3' and 5' regions of the genes related with Lung cancers	EML4, ALK, ROS1, RET	FFPE RNA samples