IVF screen

IVF screen is a kit for the analysis of 50+ 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). IVF 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: R2040-16 (16 test)**
- **REF: R2040-48 (48 test)**
- Genes targeted: 50+
- Application: germline analysis
- Number of pools: 3
- Panel size: 60.5 kb
- Input DNA: 20ng/reaction

Disease	Gene
Alzheimer And Parkinson Disease	BDNF
Alzheimer And Parkinson Disease	BCHE
Ataxia-telangiectasia	ATM
Beta-thalassemia	HBB
Bloom Syndrome	BLM
Canavan Disease	ASPA
Choroideremia	CHM
Fabry Disease	GLA
Familial Mediterranean Fever	MEFV
Fanconi Anemia	FANCC
Favism	G6PC
Galactosemia	GALT
Gaucher's Disease	GBA
Glutaric Acidemia, Type 1	GCDH
Hearing Loss, Dfnb1 And Dfnb9 Nonsyndromic	GJB2
Hearing Loss, Dfnb1 And Dfnb9 Nonsyndromic	OTOF
Hearing Loss, Dfnb1 And Dfnb9 Nonsyndromic	PJVK (DFNB59)
Hemochromatosis	HFE2
Hemochromatosis	FPN1
Hemochromatosis	HFE
Hemochromatosis	TFR2
Hereditary Fructose Intolerance	ALDOB
Juvenile Retinoschisis, X- Linked	RS1
Krabbe Disease	GALC
Mucopolysaccharidosis I, Or Hurler Syndrome	GLB1
Mucopolysaccharidosis I, Or Hurler Syndrome	IDUA
Mucopolysaccharidosis I, Or Hurler Syndrome	IDUA
Niemann-pick Disease	SMPD1
Niemann-pick Disease	NPC2
Niemann-pick Disease	NPC1
Nijmegen Breakage Syndrome	NBN
Ovarian Hyperstimulation Syndrome	FSHR
Phenylketonuria	PAH
Polycistic Kidney Disease Types I And Ii	PKD2
Polycistic Kidney Disease Types I And Ii	PKHD1
Pompe Disease	GAA
Pregnancy Loss	M2/ANXA5
Prothrombin Deficiency	Prothrobin (F2)

Disease	Gene
Riley-day Syndrome (Familial Dysautonomia	ELP1 (IKAP)
Smith-lemli-opitz Syndrome	DHCR7
Spherocytosis, Hereditary	EPB42
Spherocytosis, Hereditary	ANK1
Tay-sachs Disease	HEXA
Tay-sachs Pseudodeficiency	HEXA
Thrombocytopenia, Congenital Amegakaryocytic	MPL
Thrombophilic Mutations	MTHFR
Thrombophilic Mutations	FV-Leiden
Thrombophilic Mutations	Prothrobin (F2)
Thrombophilic Mutations	APoE
Thrombophilic Mutations	PAI
Tyrosine Hydroxylase Deficiency	TH
Wilson's Disease	ATP7B
Zellweger Syndrome Spectrum, Pex1-related	PEX1
Male Infertility	Yq11.21-sY81
Male Infertility	AZFa-sY84
Male Infertility	AZFa-sY84
Male Infertility	AZFa-sY625
Male Infertility	AZFa-M259
Male Infertility	Yq11.221-sY90
Male Infertility	AZFb-sY127
Male Infertility	AZFb-sY131
Male Infertility	AZFb-sY134
Male Infertility	AZFc-sY157 _(2)
Male Infertility	SRY-sY14
Male Infertility	AZFc-sY157 _(1)
Familial Adenomatous Polyposis	MUTYH
Breast-ovarian Cancer (Familial)	BRCA2
Breast-ovarian Cancer (Familial)	BRCA1
Familial Adenomatous Polyposis	APC
Spinal Muscular Atrophy	SMN2
Spinal Muscular Atrophy	SMN1
Factor XIII Deficiency	F13A1
Cystic Fibrosis	CFTR
Duchenne And Becker Muscular Dysthrophy	DMD

SAMPLES/RUN*

	Germline
MiSeq Nano Kit v2 (300-cycles)	3
Nano Kit v2 (500-cycles)	4
Micro Kit v2 (300-cycles)	10
Kit v2 (300-cycles)	40
Kit v2 (500-cycles)	60
Kit v3 (600-cycles)	>96
MiniSeq Mid Output Kit (300-cycles)	20
High Output Kit (300-cycles)	65
iSeq 100 i1 kit (300-cycles)	10
NextSeq 550 Mid-Output Kit	35
High-Output Kit	>384
Ion 314™ Chip	1
Ion 316™ Chip	5
Ion 318™ Chip/Ion 520™ Chip	10
Ion 530™ Chip	40
Ion PI™ Chip/Ion 540™ Chip	>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

Index set A (6x4) REF: R5001
Index set B (6x4) REF: R5002

For Ion Torrent instruments

Barcode 1-16 REF: R4001
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