PID pro

For screening of Primary Immunodeficiency Disorders

PID pro

PID pro is a kit for the identification of mutations in genes related to Primary Immunodeficiency Disorders (PIDs).

PIDs are a diverse group of genetically determined disorders affecting the immune system. PIDs exhibit a variety of symptoms depending on the part of the immune system that is affected.

5.2% of PIDs reported in the European Society for Immunodeficiency (ESID) registry are caused by deficiency of the complement system, a fundamental part of the innate immune system.

In severe PIDs, early diagnosis can prevent complications from infections and allows for referral to targeted treatment.

Neonatal Screening specifically represents one of the most effective tools for early diagnosis that can lead to an improvement in the prognosis and outcomes of the disease.

TECHNOLOGY

PID pro is a kit for analyzing genes related to PIDs through a molecular protocol based on Nex Generation Sequencing (NGS) technology. The kit is validated for the germline analysis (SNPs, indels, CNVs) of DNA samples extracted from blood or tissues.

PID pro kit contains all reagents required for the preparation and capture of specific probes designed for sequencing on Thermofisher platforms

WORKFLOW

Library preparation follows a simple capture-based protocol that can be completed in 1.5 days, with less than 5 hours of hands-on-time.

The resulting libraries can be normalized, merged and then loaded on to a chip for sequencing.

REFERENCES

- Bousfiha A. et al. Human Inborn Errors of Immunity: 2019 Update of the IUIS Phenotypical Classification. J Clin Immunol. 2020 Jan.
- Brodszki N. et al. European Society for Immunodeficiencies (ESID) and European Reference Network on Rare Primary Immunodeficiency, Autoinflammatory and Autoimmune Diseases (ERN RITA) Complement Guideline: Deficiencies, Diagnosis, and Management. J Clin Immunol 2020.
- Stray-Pedersen A. et al. Primary immunodeficiency diseases: Genomic approaches delineate heterogeneous Mendelian disorders. J Allergy Clin Immunol. Allergy Clin Immunol. 2018 Feb.
- Raje N. et al. Utility of next generation sequencing in clinical primary immunodeficiencie. Curr Allergy Asthma Rep. 2014.
- Bisgin A. et al. The Utility of Next-Generation Sequencing for Primary Immunodeficiency Disorders: Experience from a Clinical Diagnostic Laboratory. Biomed Res Int. 2018 May.

SAMPLE PER RUN

Instrument	Samples per run*
	Germline
Ion 530™ Chip	8
Ion PI™ Chip/Ion 540™ Chip	24
Ion 550™ Chip	72

**the estimated maximum number of samples per chip assumes a reading depth of 300x for the germline. The optimal number of samples can be empirically estimated on the local setup.*
The volume present in the kit is calculated to allow the subdivision into multiples of 8 analysis sessions. Dividing the kit in different ways decrease the total number of tests that can be performed.

ORDERING INFORMATION

Product	REF
PID pro	RC3190Y-16 (16 test)
PID pro	RC3190Y-96 (96 test)
Adapter	
Y ADAPTER	R9001-16 (16 test)
Y ADAPTER	R9001-96 (96 test)

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Table 1: PID pro genes list

ACD	ACP5	ACTB	ADA	ADA2	ADAM17	ADAR	AICDA	AIRE	AK2	AK7	ALG6	ANGPT1	ANKZF1	AP3B1
AP3D1	ARHGEF1	ARMC4*	ARPC1B	ASAH1	ATM*	ATP6AP1	B2M	BACH2	BCL10	BCL11B	BLM	BLNK	BLOC1S3	BLOC1S6
BTK	C11orf70	C17orf62	C1QA	C1QB	C1QC	C1S	C2	C3	C5	C6	C7	C8A	C8B	C9
CARD11	CARD14	CARD8	CARD9	CARMIL2	CASP10	CASP8	CBL	CCBE1	CCDC103	CCDC114	CCDC151	CCDC39	CCDC40	CCDC65
CD19	CD247	CD27	CD3D	CD3E	CD3G	CD40	CD40LG	CD46	CD55	CD59	CD79A	CD79B	CD81	CD8A
CDC42	CDCA7	CEBPE	CEP164	CFAP298	CFB	CFD	CFH*	CFI	CFP	CFTR*	CHD7	CIB1	CIITA	CLCN7
CLPB	COL7A1	COPA	CORO1A*	CR2	CSF2RA*	CSF2RB	CSF3R	CTC1	CTLA4	CTPS1	CTSC	CXCR2	CXCR4	CYBA
CYBB	CYP27A1	DCLRE1C	DDX58	DEF6	DGAT1	DIAPH1	DKC1	DNAAF1	DNAAF2	DNAAF3	DNAAF4	DNAAF5	DNAH1	DNAH11
DNAH5	DNAH8	DNAH9	DNAI1	DNAI2	DNAJB13	DNAJC21	DNAL1	DNASE1L3	DNASE2	DNMT3B	DOCK2	DOCK8	DRC1	DSG1
DTNBP1	DUOX2*	EFL1*	EIF2AK3	ELANE	EPG5	ERBIN	ERCC2	ERCC3	ERCC6L2	EXTL3	FADD	FANCA	FANCB	FANCE
FANCF	FANCI	FANCL*	FAS	FASLG	FAT4	FCHO1	FERMT1	FERMT3	FOXI3	FOXN1	FOXP3	FPR1	G6PC	G6PC3
G6PD	GAS8	GATA2	GFI1	GINS1	GTF2E2	GTF2H5	GUCY2C	HAX1	HELLS	HMOX1	HPS1	HPS3	HPS4	HPS5
HPS6	HTRA2	HYOU1	ICOS	ICOSLG	IFIH1	IFNAR1	IFNAR2	IFNGR1	IFNGR2	IGLL1	IKBKB	IL10	IL10RA	IL10RB
IL12B	IL12RB1	IL12RB2	IL17F	IL17RA	IL17RC	IL1RN	IL21	IL21R	IL23R	IL2RA	IL2RB	IL2RG	IL36RN	IL6R
IL6ST	IL7R	IRAK4	IRF2BP2	IRF4	IRF7	IRF8	IRF9	ISG15	ITCH	ITGAM	ITGB2	ITK	JAGN1	JAK1
JAK3	KDM6A	KMT2A	KMT2D	LAMTOR2	LAT	LCK	LCT	LIG1	LIG4	LIPA	LPIN2	LRBA	LRRC56	LRRC6
LRRC8A	LYN	LYST	MAGT1	MALT1	MAP3K14	MCIDAS	MCM4	MEFV	MKL1	MOGS	MPLKIP	MS4A1	MSH6*	MSN
MTHFD1	MVK	MYD88	MYO5B	MYSM1	NBAS	NBN	NCF2	NCF4	NCSTN	NEUROG3	NFAT5	NFE2L2	NFKB1	NFKB2
NFKBIA	NHEJ1	NHP2	NLRC4	NLRP1	NLRP12	NLRP3	NOD2	NOP10	NOTCH2*	NSMCE3	OAS1	OFD1	ORAI1	OSTM1
OTULIN	PARN	PAX1	PEPD	PGM3	PIH1D3	PIK3CD	PIK3R1	PLCG2	PMM2	PMS2*	PNP	POLA1	POLD1*	POLE
POLE2	POLR3A	POMP	PRF1	PRKCD	PRKDC	PSENEN	PSMA3	PSMB4	PSMB8	PSMG2	PSTPIP1	PTEN*	PTPRC*	RAB27A
RAC2	RAG1	RAG2	RANBP2*	RASGRP1	RBCK1	RELA	RELB	RFX5	RFXANK	RFXAP	RHOH	RIPK1	RMRP	RNASEH2A
RNASEH2B	RNASEH2C	RNF113A	RNF168	RNF31	RNU4ATAC	RORC	RPGR*	RPSA	RSPH1	RSPH3	RSPH4A	RSPH9	RTEL1	SAMD9
SAMD9L	SAMHD1	SAR1B	SCO2	SEC61A1	SEMA3E	SERPING1	SH2D1A	SH3BP2	SH3KBP1	SI*	SIAE	SKIV2L	SLC26A3	SLC29A3
SLC35C1	SLC37A4	SLC39A7	SLC46A1	SLC5A1	SLC7A7	SLC9A3*	SLX4	SMARCAL1	SMARCD2	SNX10	SP110	SPAG1	SPINK5	SPINT2
SPPL2A	SRP54	SRP72	STAT1	STAT2	STAT3	STAT4	STAT5B*	STIM1	STK4	STN1	STX11	STX3	STXBP2	TAOK2
TAP1	TAP2	TAPBP	TAZ	TBX1	TCF3	TCIRG1	TCN2	TERC	TERT	TFRC	TGFB1	TGFBR1	TGFBR2	THBD
TICAM1	TIMM50	TINF2	TLR3	TMC6	TMC8	TMEM173	TNFAIP3	TNFRSF11A	TNFRSF13B	TNFRSF13C	TNFRSF1A	TNFRSF4	TNFRSF6B	TNFRSF9
TNFSF11	TNFSF12	TONSL	TOP2B	TP63	TPP2	TRAF3	TRAF3IP2	TREX1	TRNT1	TTC37	TTC7A	TYK2	UNC13D	UNC45A
UNC93B1	UNG	USB1	VAV1	VPS13B	VPS45	WAS	WDR1	WIPF1	WRAP53	XIAP	ZAP70	ZBTB24	ZCCHC8	ZMYND10
ZNF341														