

CCI Genetics Unit submission form

Referring physician: _____ Department: _____

Centre: _____ Institution: _____

Email: _____ Phone: _____

Postal address: _____ Country: _____

Personal Information

First name: _____ Last name: _____

Gender: _____ D.O.B: _____ Nationality: _____

Diagnosis: _____ Age of onset: _____

Symptoms: _____ Consanguinity: _____

Genetic testing (Please select)

 Single EXON sequencing for panel confirmation Gene: _____ Exon: _____ Variant: _____
 (Turnaround time 2-4 weeks) [Cost per request]

 Single GENE sequencing for suspected genotype
 (Turnaround time 6-8 weeks) [Cost per request]

 Genes available:
 (Please select)

-
- AICDA
-
- IKZF1
-
- IL17A
-
- RAG1
-
- TNFRSF13B
-
-
- CTLA4
-
- IL10RA
-
- NFKBIA
-
- RAG2
-
- TNFRSF13C
-
-
- ICOS
-
- IL10RB
-
- PDCD1
-
- SH2D1A
-
- XIAP

Targeted gene PANEL

(Turnaround time 4 months) [Cost per request]

Please mark any of the panels below

 Primary Immunodeficiencies (PID)

ADA	AICDA	AIRE	ATM	BACH2	BANK1	BCL11B	BCL2L11	BLK	BLNK	BTK	CAMLG
CARD11	CARD9	CD19	CD40	CD40LG	CD79A	CD79B	CD81	CECR1	CFTR	CLEC7A	CR2
CTLA4	CXCR4	CYBA	CYBB	DCLRE1C	DKC1	DOCK8	EBF1	FANCA	FANCE	FASLG	FCGR3B
FOXP3	GATA2	ICOS	IFIH1	IGHM	IKBKB	IKBKG	IKZF1	IKZF3	IL12A	IL12RB1	IL17A
IL17F	IL17RA	IL17RC	IL21	IL21R	IL23A	IL23R	IL2RG	IL6	INPP5D	IRAK4	IRF2BP2
IRF3	IRF8	LRBA	MAGT1	MBL2	MCM4	MDC1	MRE11A	MS4A1	MYC	MYO9A	NCF1
NCF2	NCF4	NEIL1	NFKB1	NFKB2	NFKBIA	NHEJ1	NLR4	NLRP12	NLRP2	NLRP3	ORAI1
PGM3	PIK3CD	PIK3R1	PIK3R4	PLCG2	PMS2	POLE	PRKCD	PRKDC	PRRC2A	PTPN22	PTPRC
RAC2	RAG1	RAG2	RNASEH2A	RNASEH2B	RNASEH2C	RTEL1	SAMD9L	SAMHD1	SATB1	SEC61A1	SEC61A2
SEC61B	SEC61G	SH2D1A	SH3BP2	SKIV2L	SPINK5	STAT1	STAT3	STIM1	STXB2P2	TAP2	TCF3
TINF2	TLR3	TNFAIP3	TNFRSF11A	TNFRSF13B	TNFRSF13C	TNFSF12	TNFSF13	TNFSF13B	TNFSF15	TNIP1	TRAF3IP2
TRNT1	TWIST1	TYK2	UNG	VAV1	WAS	XIAP	ZNF341				

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 OR Inflammatory Bowel Disease (IBD)

ABCB1	ACP5	ADCY7	AGER	AGRN	ANKZF1	APEX1	ASIC2	ATG16L1	ATG16L2	BPI	BSN
BTNL2	C2CD4D	CALCOCO2	CARD8	CARD9	CD14	CD19	CD209	CD226	CD24	CD40	CFTR
CLDN10	CLEC2D	CNR1	COG6	COL7A1	CSF2RB	CTDSP1	CTLA4	CYBA	CYBB	DEFB4A	DLG1
DLG5	DMBT1	DNAH12	DNMT1	DUOX2	EED	EPCAM	FAAH	FCGR2A	FCGR2B	FERMT1	FOS
FOXP3	FUT2	FUT3	G6PC3	GAL3ST2	GLI1	HEATR3	HNFA4	HNRNPD	HSPA1L	ICOSLG	IFIH1
IFNA10	IFNA4	IKZF1	IL10	IL10RA	IL10RB	IL16	IL17A	IL17REL	IL23R	IL2RA	IL4R
INO80	IRF5	IRGM	ITGB2	JAK2	KRT18	KRT8	LACC1	LIG4	LMTK3	LNPEP	LRBA
LRP6	LRRK2	MBL2	MPO	MSH5	MST1	MST1R	MUC2	MUC3A	MXRA8	MYO5B	MYO9A
MYO9B	NCF1	NCF2	NCF4	NLRP3	NOD1	NOD2	NR112	PIK3R4	PGLYRP1	PGLYRP2	PGLYRP4
PRDM1	PTGER4	PTPN22	SERPINA1	SFTPD	SKIV2L	SLC22A4	SLC22A5	SLC26A6	SLC39A4	SMAD3	STK4
TGFBR1	TGFBR2	TLR4	TLR5	TLR9	TNFSF15	TRIM11	TRIM22	TTC37	TYK2	VWA1	WAS
XIAP	ZNF365	ZNF831									

 Whole EXOME sequencing

(Turnaround time up to 3 months) [Cost per request]

Functional testing

Validation of a genetic defect affecting protein function is also offered, as a research test.

Protein detection

By Flow cytometry:

IL17RA CD19 CD27 DOCK8 IL-21

By Western Blot:

SEC61A1 ZNF341

Protein expression upon stimulation

CTLA-4 LRBA pSTAT3 pSTAT1

Others

CTLA-4 (transendocytosis assay) CTLA-4 (expression)

Th17 (in-vitro expansion)

Each functional testing requires different specifications, for further inquiries please contact Jessica Rojas (jessica.rojas@uniklinik-freiburg.de).

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General guidelines

- ▶ Please write in a legible manner on the spaces allocated for 'Referring physician' and 'Personal information'.
- ▶ Please mark with an 'X' on the assigned boxes whenever possible.
- ▶ Please fill in one individual form per sample sent.
- ▶ In case an additional sample from a healthy travel-control is being sent, please fill in at least the section 'Personal information' on a separate form.
- ▶ A signed consent form from both parents is needed if the patient is under 18 years of age.

Recommendations for shipment

- ▶ Please ensure that fresh blood samples will be kept at room temperature during the travel.
- ▶ Please package the EDTA blood tubes in a way that prevents breakage and/or leakage.
- ▶ Please take into account that samples should be delivered no later than Friday as our laboratory will not be manned on the weekend.

Specifications for samples sent for genetic testing

- ▶ Please send at least 10ml of fresh EDTA blood per sample for any of the genetic tests. Samples of relatives will be appreciated as they can later be used to validate variant segregation.
- ▶ In case you prefer to send genomic DNA (gDNA), please send at least 1µg of gDNA in a total volume of at least 30µl.

Please send samples to:

Genetic testing	Functional testing
UNIVERSITÄTSKLINIKUM FREIBURG Center for Chronic Immunodeficiency Katrin Hübscher (AG Grimbacher) Breisacher Str. 115, 1st floor 79106 Freiburg, Germany katrin.huebscher@uniklinik-freiburg.de +49 761-270-77733	UNIVERSITÄTSKLINIKUM FREIBURG Center for Chronic Immunodeficiency Jessica Rojas (AG Grimbacher) Breisacher Str. 115, 1st floor 79106 Freiburg, Germany jessica.rojas@uniklinik-freiburg.de +49 761-270-77733

Declaration

This is to certify the shipments contain samples which are for Research Purpose Only. The samples are derived from humans and they are not hazardous, not infectious, not HIV positive, not toxic, and not radioactive. No import license is required for this shipment.

Bill shall be sent to:

Name

Address

Please be aware that we will proceed the sample only after we received the payment.

I hereby confirm that the patient has signed our patient consent form and/or a local ethics consent form, allowing for the study of his/her condition.

Date

Signature and stamp of physician