Patient information	Result-/Return address	
		CCI - Advanced Diagnostics Unit
		Breisacher Str. 115 • 79106 Freiburg Telefon: 0761 270-71010 Telefax: 0761 270-9671070
آ	Name of clinic	cci.diagnostik@uniklinik-freiburg.de www.cci.uniklinik-freiburg.de
7	Adress	
ī	Phone Fax	Centrum für Chronische Immundefizienz
CCI immunodeficiency diagnostics Processing only after prior registration (note shipping conditions on page 2) Sample registration: +49 (0)761 270-71010, telephone Clinical information: +49 (0)761 270-77550 (ped.) / +49 (0)761 270-77640 (adult.) 1. Suspected diagnosis/main symptoms (please enclose summary/last doctor's letter)		
		_ Date of blood collection:
		Time of blood collection:
Cortisone > 0.5 mg/kg Immunosuppre	ssants:	
Statement after Rituximab Inone Statement after HSZT		
2. Cost carrier (please tick, if no information is provided, billing will be done via the sender)		
outpatient: GKV patient (enclose laboratory referral form 10BF or 10; only within Germany) inpatient/abroad: invoice to sender/clinic		
private/self-payers (invoice to patient, please pro	vide address) other:	
invoice recipient:		
3. Individual requirements (5 mL EDTA blood each: ¹ = control blood required, see page 2): ² = blood count required		
Lymphocyte Phenotyping	IBD Panel	
Lymphocyte populations with naïve and	Granulocyte function: ¹	$\Box IL-4 / IFNy^{1}$
activated T cells (T, B, NK cells, CD45RA/RO,	Oxidative Burst (CGD, IBD)	IL-17 / IFNγ ¹ (STAT1 GOF, HIES)
HLA-DR; Diff-BB required for assessment)'	XIAP in Lymphocytes ¹ (XLP2)	STAT1 phosphorylation ¹ (STAT1 GOF)
(TCR $\alpha\beta$ /γ δ , naïve, memory, TEMRA - CD57; only in	XIAP/NOD2 function test (XLP2, IBD) (with altered XIAP expression) ¹	STAT5 phosphorylation ¹ (X-SCID)
combination with LP)	IL10R function (only in infants,	STAT6 phosphorylation ¹ (X-SCID)
Cell pnenotyping (memory, switched, IgG+, IgA+, plasmablasts, transitional, CD21low)	after consultation) ¹ Regulatory T cells (FoxP3, IPEX) ¹	CARD11, MALT1, BCL10)
T cell repertoire (24 Vβ chains)	Special Cell Populations ¹	ADA2 Enzyme deficiency test ¹
SCID Panel (NG Screening)	Dendritic cells ¹	contact: Frau Dr. Grünert/Frau Dr. Schumann,
Stage 1 ² : Lymphocyte phenotype with naïve	(pDC, mDC; GATA2 Defect)	phone.: 0761 270-43680)
and activated T cells	$\square MAIT cells1 (V\alpha7.2/CD161; e.g. XLP2)$	Grapulacuta/Manacuta Eurotion
Stage 2 ^{1,2} : T-phänotyp., proliferation, ADA,	$\square \text{ NKT Cells' (V} \alpha 24/V \beta 11; e.g. \textbf{XLP1})$ $\square \text{ Pogulatory T colls^1 (Fox D2; IDEX)}$	
		\Box TLR function test ¹ (LPS Stimulation.
HLH Panel	Protein Detection ¹	IRAK-4, MyD88)
Fresh NK cell degranulation (FHL, CHS, GS) ¹	CD11b/CD181 ¹ (LAD Typ 1)	USV CER INAR TYPE STATE STATE
Sumulated INK Cell / CTL degranulation (IL2)		(VOV-GEF, IFINAK, ITKZ, STATT, STATZ)
	$\square \text{ GD40L OIT T CEIIS' (HIGM)}$ $\square \text{ SAP in Lymphocytes}^{1}(\textbf{XLP1})$	Serum Parameters (je 3 mL serum)
\square SAP in Lymphocytes (YLP1 in boys) ¹	\square XIAP in Lymphocytes (XLP1)	Anti-Pneumococcal Polysaccharide
\square XIAP in Lymphocytes (XLP2 in boys) ¹	\square NKG2D on NK cells ¹ (MAGT1. EBV)	Antibodies (IgG, 9 Serotypes)
\square CD27 on T cells (in EBV) ¹	CD27 in Lymphocytes ¹ (CD27 deficiency)	vaccination; not under IgG substitution;
Serum Parameters (3 ml serum)	BTK in Monocytes ¹ (XLA)	normal postal shipping possible)
Soluble IL2 Receptor (sCD25)	WASP in Lymphocytes ¹ (WASP)	

Weeks after vaccination

- Sample Storage (as per page 2)
- Material is to be stored
 - (consent enclosed)

TCRαβ DNT cells (5 mL EDTA blood) 4. Comments and signature

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For feedback: Email:

ALPS Panel

3ml, every 4 weeks)

Date

T Cell Function¹

(CID/SCID)

T cell activation markers CD25, CD69¹

ALPS Biomarker: sFasL, Vit B12 (from Li-Heparin, Γ T cell proliferation¹ (PHA, αCD3, αCD3/CD28)

Name (legible) and signature of the doctor

Phone:

X

CCI Immunodeficiency Diagnostics

Requirements for sample material

For each submission, a telephone registration with an appointment is required before shipping. Unannounced samples cannot be processed. Please ask for the amount of blood or serum required for your request when making an appointment. Please fill out the request form completely. The following information is required for processing:

- Complete patient data (name, first name, date of birth) on the request form and blood tube
- Date and time of sample collection
- Contact details of the sender for sending the results
- ▶ Insurance status (laboratory referral form form 10BF or 10)
- ► Billing address:

¹ For functional blood tests, an additional blood sample from a healthy adult, unrelated control person is required. This is important in order to exclude artifacts from storage and transport as much as possible. Please check the request form to see if this applies to the tests you want.

Shipping conditions

The samples must be sent at room temperature and, unless otherwise stated, must arrive in the laboratory within 24 hours of blood collection and no later than 9 a.m. on the day after blood collection. Only use shipping services that offer a corresponding service (e.g. DHL, TNT, GO Express). The blood tubes must be packed in a leak-proof outer tube or bag with absorbent material and the packages must be marked for shipping with "medically exempt sample" or "UN 3373". In extreme temperatures (<5°C and >30°C), please ensure that the samples are well insulated to avoid cell damage.

Send the sample to the following address: University Hospital Freiburg

CCI - Diagnostics in the ZTZ Breisacher Str. 115, EG

79106 Freiburg

Prof. Dr. Stephan Ehl (Focus: All immune deficiencies in childhood; in particular: SCID, combined immune deficiencies, immune deficiencies with autoimmunity, HLH) and **Prof. Dr. Klaus Warnatz** (Focus: antibody deficiency syndromes, combined immune deficiencies, complement deficiencies in **adults**) are available to you as clinical contacts.

Ped. secretariat.: 0761 270-77550 (Prof. Dr. Ehl), Adult secretariat: 0761 270-77640 (Prof. Dr. Warnatz)

The results of individual analyses can be influenced by the patient's clinical condition and medication, so the correct interpretation of the test results is only possible in the context of clinical findings. Please provide these to us (e.g. in the form of doctor's letters). Further tests for immunodeficiency diagnostics are possible after consultation.

The costs for the requested tests are available on request.

Sample storage

Your patient's material may also be of interest for scientific questions in the future. We would therefore like to ask you to inform the patient about our study: "CCI Biobank" and to ask them to give their consent by signing the consent form "Examination and storage of biomaterial" (available at <u>https://www.uniklinik-freiburg.de/cci/immunologische-diagnostik.html</u>). It is not possible to store samples without a consent form. Thank you for your support.

We are happy to answer any questions you may have. The laboratory team

Legend:

- 60114291 ForManFR / 06.05.2025
- ADA2 = Adenosin deaminase 2 ALPS = Autoimmune Lymphoproliferative Syndrome
- BTK = Bruton's Tyrosine Kinase
- CGD = Chronic Granulomatous Disease
- CMC = Chronic Mucocutaneous Candidiasis
- HIES = Hyper-IgE Syndrome
- HIGM = Hyper-IgM Syndrome HLH = Hemophagocytic Lym
 - LH = Hemophagocytic Lymphohistiocytosis
- IBD = Inflammatory Bowel Disease

- IPEX = Immune Dysregulation, Polyendocrinopathy, Enteropathy, X-linked Syndrome LAD = Leukocyte adhesion defect
- (S)CID = (Severe) Combined Immunodeficiency
- TLR = Toll-Like-Receptor
- XLA = X-linked Agammaglobulinemia
- XLP = X-linked Lymphoproliferative Syndrome
- XMEN = X-linked immunodeficiency with magnesium defect, EBV
- infection and neoplasia (MAGT1 Mutation)
- WASP = Wiskott-Aldrich-Syndrome Protein

¹ Blood from a healthy control person is absolutely necessary. Please be sure to include this with the patient sample.
 ² A current daily differential blood count is necessary to calculate the cell count. Please include this or send it later in electronic form.