Formular drucken | Dieses Formular ist am Bildschirm ausfüllbar | Formular zurücksetzen

Patient information

Result-/Return address

alternativ clinic stamp

Name of clinic

Adress

Fon Fax

UNIVERSITÄTS KLINIKUM FREIBURG
CCI - Advanced Diagnostics Unit
Breisacher Str. 115 • 79106 Freiburg
Phone: +49 (0)761 270-71010
Fax: +49 (0)761 270-9671070
cci.diagnostik@uniklinik-freiburg.de
www.cci.uniklinik-freiburg.de

Centre of Chronic Immunodeficiency
SPONSORED BY THE Federal Ministry of Education and Research

Diagnostics of Immunodeficiencies CCI

Tests are available after prior arrangement only (Please refer to shipping conditions on page 2)

Sample announcement: Ilka Fuchs +49 (0)761 270-71010 or lab -71070 - clinical questions: +49 (0)761 270-77550

1. Preliminary diagnosis

Date of collection:

Time of collection:

2. Please indicate invoice recipient (please tick)

- ☐ "GKV" patient (Please add "Überweisungsschein" (only within Germany))
- ☐ Patient, please add address
- ☐ Sender / hospital
- ☐ other:

3. Biobanking (please refer to page 2)

Material can be stored: ☐ no ☐ yes (please add consent form)

☐ Serum without IgG substitution ☐ Serum during IgG substitution

4. Single Tests (5 mL EDTA blood each)

Standard Lymphocyte phenotyping

- ☐ Lymphocyte subpopulations with naive and activated T cells (CD3+, CD4+, CD8+ T cells, B cells, NK cells, CD45RA, HLA-DR; please add blood counts for absolute values)
- ☐ advanced T cell activation- & differentiation markers (TCRαβ/γδ, HLA-DR, CD3, CD4, CD8, CD45RA/RO, CD27, CD28, CD57)
- ☐ CD31+ CD45RA+ T cells "recent thymic emigrants"
- ☐ CD11b/CD18 (**LAD Type 1**)
- ☐ HLA-A, B, C (**MHC I-Defect**)
- ☐ B cell phenotyping memory, switched, IgG+, IgA+, plasmablasts, transitionals, CD21low
- ☐ CD40 on B cells* (**HIGM**)
- ☐ CD40L on T cells* (PMA/Iono stimulation; CD40Ligand, CD69; **HIGM**)
- ☐ Dendritic cells* (pDC, mDC; **GATA2 deficiency**)
- ☐ MAIT cells* (Vα 7.2/CD161)
- ☐ NKT cells* (Vα24/Vβ11)
- ☐ TCRαβ DNT cells (**ALPS**)
- ☐ regulatory T cells
  - ☐ phenotypic (CD45RO, CD25high, CD127low)
  - ☐ FoxP3 intercellular* (e.g. for **IPEX**)

Repertoire

- ☐ extended T cell repertoire (24 Vβ chains on T cells)

Intracellular proteins

- ☐ BTK in monocytes* (**XLA**)
- ☐ SAP in lymphocytes* (**XLP1**)
- ☐ XIAP in lymphocytes* (**XLP2, IBD**)
- ☐ WASP in lymphocytes* (**WASP**)
- ☐ further proteins**: _____ (to verify genetic mutations, only after consultation)

Functional tests on lymphocytes

- ☐ ADA2 enzyme diagnostic* (second Ü-Schein Nr10 with note "ADA2" necessary; contact: Mrs Tucci +49 761 270-43740)
- ☐ Ca-Flux* (TCR and Thapsigargin stimulation)
- ☐ IL-2 / IFNγ production* (PMA/Iono stimulation)
- ☐ IL-4 / IFNγ production* (PMA/Iono stimulation)
- ☐ IL-17 / IFNγ production* (PMA/Iono stimulation; **CMC/HIES**)
- ☐ NFκB p65 phosphorylation and IκBα degradation* (PMA/Iono stimulation; e.g. for **CARD11, MALT1, BCL10**)
- ☐ STAT1 phosphorylation in T cells* (IFNα/γ stimulation; **CMC**)
- ☐ STAT5 phosphorylation in T cells* (IL-2 stimulation; **X-SCID**)
- ☐ STAT6 phosphorylation in B cells* (IL-4 stimulation; **X-SCID**)
- ☐ T cell activation markers* (TCR stimulation; CD25, CD69)
- ☐ T cell proliferation* (PHA and TCR stimulation)

NK cell function

- ☐ NK cell degranulation* (**HLH**)
- ☐ Perforin in NK cells* (**HLH**)
- ☐ NKG2D on NK cells (**MAGT1/XMEN**)

Functionality of macrophages/ monocytes

- ☐ Functional test of TLR* (stimulation of monocytes with LPS; **IRAK-4, MyD88**)
- ☐ Functional test of XIAP/NOD2* (stimulation of monocytes with L18MDP; **XLP2, IBD**, verification test, only if XIAP stain was abnormal)
- ☐ IL-10R function* (**IBD**)
- ☐ Interferon-Funktionstest* (**IFNAR, TYK2, STAT1, STAT2**)

Granulocyte function

- ☐ Oxidative burst* (**CGD, IBD**)

Serum parameters (3 mL serum each)

- ☐ **ALPS** biomarkers (sFasL, Vitamin B12)
- ☐ sCD25 (soluble IL-2 receptor)
- ☐ anti-Pneumococcal-Polysaccharide-antibodies (IgG) (ideal before and 4 weeks after vaccination; not during IgG-substitution)
  - ☐ before vaccination
  - ☐ _____ weeks after vaccination

5. Comments and signatures

Comments:

Date

✕ Signature and stamp of physician

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UNIVERSITÄTS KLINIKUM FREIBURG

Diagnostics of Immunodeficiencies CCI

Directions for shipping of blood samples

Tests, apart from serum parameters, are by prior arrangement only. Please notify us of samples by phone (Ilka Fuchs: +49 (0)761 270-71010 or lab: -71070) or by e-mail (cci.diagnostik@uniklinik-freiburg.de) as early as possible. Unannounced samples cannot be processed. Please let us know if there are any changes in shipping time.

Please complete the request form with all necessary information, including:

- ► Patient data (Surname, name, date of birth), also on the sample tubes
- ► Date and time of sample collection
- ► Contact information of sender for communication of results
- ► Contact information of invoice recipient

Samples, apart from samples for anti-Pneumococcal-Polysaccharide-antibodies, have to reach the lab **within 24 hours** after blood sampling and **before 9 AM** the next day. Please use only couriers that offer this service.
Samples have to be shipped at room temperature. If necessary protect samples from extreme outside temperatures.
Please use following address:

Medical Center - University of Freiburg
CCI - Diagnostic at ZTZ
Breisacher Str. 115, EG
79106 Freiburg - Germany

For basic diagnostics usually 15 ml EDTA blood and 3-5 ml serum are sufficient. In special situations (e.g. babies) please ask for minimum amount.
For functional assays, an additional blood sample from a healthy, non-related, adult is necessary to control for shipment-dependent artefacts. Such assays are marked with '*' in the request form.
For clinical questions please contact **Prof. Dr. Stephan Ehl** (**Paediatrics:** all immunodeficiencies; mainyl SCID, combined immunodeficiencies, immunodeficiencies with autoimmunity, HLH) or **Prof. Dr. Klaus Warnatz** (**Adults:** Antibody deficiencies, combined Immunodeficiencies, complement defects).
Office: +49 (0)761 270-77550.
Clinical information about the patient is necessary for the correct interpretation of our results. Please add available medical reports to the samples.

Please ask for the exact prices for your tests of interest.

Biobanking

Biomaterial of your patient might be of interest for further research. Please inform your patient about the possibility to store biomaterial in the "CCI Biobank" and ask for consent. In case the patient agrees, please add a signed consent form (to be downloaded from: https://www.uniklinik-freiburg.de/center-forchronicimmunodeficiency-cci/diagnostics/immunologie.html?L=1) to the patient sample and mark on the first page of this form that samples can be stored. Biobanking without the patient's consent is not possible. Thanks for your help.

If you have further questions do not hesitate to contact us
CCI Advanced Diagnostic Team

Key:

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 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

* Blood of a healthy control necessary. Please send it together with patient sample.
** Proteindetection for validation of genetic defects, only after consultation: verified: BAFF-R, Carmil-2, CD16, CD20, CD21, CD25, CD27, Coronin 1A, DOCK-8, ICOS, IL-7R, Munc13, Munc18, Rab27A, TACI
on research base: CTLA-4, IκBα, LRBA

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