

HLH Diagnostic Investigations

Patient label including full name, date of birth, and sex

Referring physician

Name: _____
Address: _____

Email: _____
Phone: _____ Fax: _____

Basic clinical data:

Underlying disease: _____

Immunosuppressants at the time of venipuncture: _____

Start of immunosuppressive therapy: _____

Haemoglobin _____ g/dL
Neutrophil count _____ /nL
Leucocytes count _____ / μ l
Platelet count _____ /nL
Ferritin _____ μ g/L not done
Fibrinogen _____ mg/dL not done
Triglycerides _____ mg/dL not done
CRP _____ mg/L
Infectious trigger _____ (e.g. EBV, CMV)
Fever yes no
Hepatosplenomegaly yes no
Haemophagocytosis yes no
Partial albinism yes no
Consanguinity yes no unknown

Prior to shipping it is **mandatory** to contact the physicians of the German HLH study center:

Paediatric Haematology and Oncology, University Hospital Medical Center, Hamburg-Eppendorf
PD Dr. Kai Lehmborg (Mobile: +49 152 22816726, Office: +49 40 7410 54209, k.lehmborg@uke.de)
Prof. Dr. Gritta Janka (Mobile: +49 172 5447780, Office: +49 40 7410 54369, janka@uke.de)
Fax: +49 40 7410 58250

- Morphology** (Lab Hamburg)
Bone marrow aspirate smears (2x stained, 4x unstained), CSF cytospin (mandatory: CSF cell count ____/ μ L, CSF protein ____ mg/dL)
- Gene sequencing** (Lab Hamburg)
5 mL EDTA blood, ideally send samples of parents as well. A signed informed consent (patient or legal guardian) for each is **mandatory**.

Tick requested genes:

- Perforin (FHL 2) SH2D1A (XLP)
 UNC13D (FHL 3) BIRC4 (XIAP)
 Syntaxin 11 (FHL 4) RAB27A (Griscelli)
 UNC18B (FHL 5) ITK
 LYST (CHS) AP3B1 (HPSII)

No express shipping required. Include this sheet and an address for billing purposes.

Contact person lab Hamburg:

Dr. Udo Zur Stadt
Tel.: +49 40 7410 52743
zurstadt@uke.de

Shipping address Hamburg:

Laboratory for Paediatric Haematology and Oncology
N21
University Hospital Medical Center Hamburg-Eppendorf
Martinistrasse 52
20246 Hamburg
Germany

- Immunology** (Lab Freiburg)
- perforin, degranulation
 - for male patients additionally SAP, XIAP
 - in case of EBV infection additionally CD27
- 10-15 mL EDTA blood (infants at least 5 mL)
- 2-3 mL serum
- 10 hairs of the patient (if albinism is suspected)
- 10-15 mL EDTA blood of a healthy unrelated person is **mandatory**.
- A signed consent form (patient or legal guardian) is **mandatory**.

It is **mandatory** to arrange the day of shipment with the lab.

Shipping overnight (within 24 hours of blood sampling). No cooling required. Sample must be delivered until 9 AM in the lab. Include this sheet and an address for billing purposes. Results are available within 2-3 days.

Date and time of venipuncture: _____

Contact person lab Freiburg:

Ilka Fuchs
Phone: +49 761 270 71010 or -71070
Fax: +49 761 270 9671070
ilka.fuchs@uniklinik-freiburg.de

Clinical contact person Freiburg:

Prof. Dr. Stephan Ehl (+49 761 270 77300, stephan.ehl@uniklinik-freiburg.de)

Shipping address Freiburg:

Medical Center – University of Freiburg
CCI Advanced Diagnostic Unit at ZTZ
Breisacher Str. 115, EG
79106 Freiburg