

# 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 \_\_\_\_\_ / $\mu$ L

Platelet count \_\_\_\_\_ /nL

Ferritin \_\_\_\_\_  $\mu$ 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](mailto:k.lehmborg@uke.de))  
Prof. Dr. Gritta Janka (Mobile: +49 172 5447780, Office: +49 40 7410 54369, [janka@uke.de](mailto:janka@uke.de))  
Fax: +49 40 7410 58250, Email: [hlh@uke.de](mailto:hlh@uke.de)

### Morphology (Lab Hamburg)

Bone marrow aspirate smears (2x stained, 4x unstained), CSF cytospin (mandatory: CSF cell count \_\_\_\_/ $\mu$ 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:

- |  |   |
|--|---|
| <input type="checkbox"/> Perforin (FHL 2)    | <input type="checkbox"/> SH2D1A (XLP)       |
| <input type="checkbox"/> UNC13D (FHL 3)      | <input type="checkbox"/> BIRC4 (XIAP)       |
| <input type="checkbox"/> Syntaxin 11 (FHL 4) | <input type="checkbox"/> RAB27A (Griscelli) |
| <input type="checkbox"/> UNC18B (FHL 5)      | <input type="checkbox"/> ITK                |
| <input type="checkbox"/> LYST (CHS)          | <input type="checkbox"/> MAGT1              |
| <input type="checkbox"/> CDC42               | <input type="checkbox"/> HAVCR2/TIM3        |

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

### Contact person lab Hamburg:

Dr. rer. nat. A. Kruchen, F. Oyen (BTA)  
Tel.: +49 40 7410 52721 or – 54742  
Fax: +49 40 7410 58931  
[a.kruchen@uke.de](mailto:a.kruchen@uke.de) [f.oyen@uke.de](mailto:f.oyen@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
- 10-15 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](mailto:ilka.fuchs@uniklinik-freiburg.de)

### Clinical contact person Freiburg:

Prof. Dr. Stephan Ehl (+49 761 270 77300,  
[stephan.ehl@uniklinik-freiburg.de](mailto: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