



Universitätsklinikum  
Hamburg-Eppendorf

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## Consent Form for Gene Sequencing and Immunological Characterisation in Patients with Haemophagocytic Lymphohistiocytosis

Patient.....

Date of Birth.....

Dear parents, dear patients,

Several different genetic defects are known to be associated with Haemophagocytic Lymphohistiocytosis (HLH). Currently, defects in the following genes are known: Perforin, Unc13D, Syntaxin11, and Unc18B. In other immunodeficiencies a propensity to develop HLH is described as well (Chediak-Higashi-Syndrome (Lyst), Griscelli syndrome type II (Rab27a), XIAP deficiency (BIRC4), X-linked Lymphoproliferative Syndrome (XLP, SH2D1A), Hermansky-Pudlak-Syndrome type II (AP3B1), ITK deficiency. In less than 10 % of patients with assumed genetic HLH, no defects can be identified.

In most cases of HLH with a proven genetic defect a stem cell transplantation is necessary to cure the disease and prevent relapses. A known genetic defect may as well be used for prenatal testing. It is the aim of the research groups in Freiburg and Hamburg and cooperating scientists worldwide to identify further genes associated with HLH to better understand and treat the disease and more easily take the decision for or against a stem cell transplantation. For the genetic analysis, we usually require 5 mL of blood. This investigation will be performed at the Paediatric Haematology and Oncology Department of the University Hospital Medical Center in Hamburg, Germany.

In addition, further investigations will be done to characterize certain properties of cells of the human immune system. If these investigation show abnormal results conclusions can be drawn which genetic defect may be present. The investigation requires 10-15 mL of blood, in infants 5mL. The analyses will be performed at the Center of Chronic Immunodeficiencies at Freiburg University Hospital.

We ask for your permission to keep remaining material of the sample that is sent to us to potentially identify genetic defects in the future by the research groups in Freiburg or Hamburg. The samples and clinical data of you / your child will be stored in a pseudonomised form in the study center in Hamburg or the respective labs. Only the study center with the two collaborating laboratories in Freiburg and Hamburg can assign a name to each sample. No third parties may be informed of any aspects of your samples without your consent. You may withdraw your consent and ask for the elimination of the respective samples and data at any time.



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## Consent form

I agree that investigations will be performed that can identify known genetic defects associated with HLH. They include genetic sequencing and immunological characterisations of NK- and T-cells. **YES**  **NO**

I agree that the result will be transmitted to the following physician(s)/person(s) **YES**  **NO**

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Name (e.g. treating physician)

I want to be informed about the results. **YES**  **NO**

I agree that the samples will be kept for 30 years, to be able to identify not yet discovered genetic defects in the future. **YES**  **NO**

I agree that the samples and the obtained data will be made available to other physicians involved in HLH research in a pseudonomised form and may be published in scientific journals in an anonymised form. **YES**  **NO**

By law data must be deleted after 10 years. However, they may be of relevance for you or your family later. If you agree we may keep them for 30. I agree that the data obtained may be kept for 30 years in Hamburg and Freiburg. **YES**  **NO**

I am aware that I can withdraw my consent at any time without explanations, which will not have any negative effects for me. All my questions have been answered.

.....  
**Date**

.....  
**Father**

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**Date**

.....  
**Mother**

.....  
**Date**

.....  
**Patient** (required if  $\geq 16$  years of age)

.....  
**Date**

.....  
**Translator** (if required)

.....  
**Date**

.....  
**Physician**