Informed Consent for Participation in a Clinical Molecular Genetic Research Study

Information Sheet for Adults or Parents of minor Children

Study Title: Genetic und functional Analysis of Immune Disorders

Centre for Chronic Immunodeficiency
University Hospital Freiburg
Breisacherstr. 115, 79106 Freiburg
Germany
Tel.: +49 761/270-77732
Prof. Dr. B. Grimbacher

We invite you and/or your child to participate in an immunological genetic research study.

1. The participation in this study is entirely voluntary.
2. It may be that your participation in this study will not benefit you personally, but the knowledge gained can benefit others.
3. You may withdraw from participation in the study at any time.

The job of the immune system is to defend us against germs. However, clinical experience shows that the immune system of humans does not fulfil this task with equal efficiency. Some people are more susceptible to infections than others. The reasons for this are complex, however, the genetic makeup of each individual plays an important role. Thus, we plan to investigate the genetic architecture of the immune system within the frame of this study.

Purpose of the Study

The purpose of the study is:

1. To characterize disorders of the immune system that lead to an increased susceptibility to infection.
2. To find the changes in the genetic material that underlie these disorders.
3. To obtain a better understanding of the function of the immune system through identified immune disorders.
4. To be able to make more precise and earlier predictions of who is at risk to develop or pass on to his/her offspring a genetic disorder of the immune system (improved genetic diagnostics).
5. To gain insight into previously unknown disorders of the immune system.
6. To create basic knowledge for improved treatment of immune disorders.

Study Procedure

At least 5 ml of EDTA blood will be taken for genetic and phenotypic workup. From the blood, DNA and if necessary cells for a phenotypic workup will be isolated. In the course of this study we will analyse genes, which play an important role in the immune system, as well as their gene products. In case of given consent, study participants with abnormalities may be contacted again for further blood samples in order to perform further analyses. In addition, we may occasionally also ask for stool samples, a saliva sample or other body fluids for further workup. If you are interested in the final results of the study, we will explain them to you and be available for questions.
Upon arrival in the laboratory all samples will be pseudonymized. This means that the researcher does not work with patient data, but with codes. Only the responsible medical staff can trace the codes back to any personalized data. The latter is necessary in order to inform patients of their results if so desired.

Benefits of this study

This study will expand our knowledge of the functioning of the immune system. This means you may not necessarily draw a direct benefit for yourself, your child, or other family members. The following, however, could be of interest:

1. The information that you have a healthy immune system.
2. The information that a specific change in the genetic material is the reason that you or your child have frequent infections.
3. That new and better treatments could be used if a specific cause for your problems is found.
4. That you can use information about risks for your family planning.

Risks and possible consequences of this study

The phenotypic analysis of your immune system does not pose any immediate risks. If you have consented to genetic testing, the genetic code (DNA) of your and your family will be examined. You should be aware that the knowledge of the genetic origins of a disease could have an impact on your future life.
Information regarding parentage may be discovered in the course of this research. Aspects of adoption or fatherhood may be touched. It is our practice not to share this information with you if you do not request it or there are no direct medical or reproductive implications for you or your family.

Storage of genetic material

The DNA will be extracted from the samples. The samples will be stored for 25 years, unless you withdraw from the study, or you want the samples to be destroyed at the end of the study. You have the right to withdraw from the study at any time without any negative consequences. Moreover, during the course of the study, possibly serum or cells might be collected from you and stored. The above applies to these as well.
In addition, we intend to make use of the collected samples within the frame of joint research projects in Germany as well as internationally with collaborators from e.g. public and private institutes, research centers or the pharmaceutical industry. To this end, the samples as well as pseudonymized or coded information on the samples may be made available to these collaborators.

Confidentiality

If the study results are presented in medical journals or at conferences or passed on to third parties (eg academic colleagues, pharmaceutical industry), the study participants remain anonymous, unless you have consented to the publication of your case in writing. Medical records of patients are held in accordance with applicable legal regulations.
We will **not** disclose any information from this study about you or your family to insurance companies or employers.

Problems or Questions

Should you have any problems or questions relating to this study, your rights as a participant, or a study-related injury, please contact the principal investigator Bodo Grimbacher at the Centre of Chronic Immunodeficiency, University Hospital Freiburg, Breisacherstr. 115, 79106 Freiburg, Germany.

Consent for:

Patient label

It is recommended that you keep a copy of this document for future reference and your personal records.

Please specify whether you would like to be notified about conclusive information regarding your heath or the health of your child.

_____ I agree to the phenotypic analysis of my immune system or the immune system of my child.

_____ I agree to the genetic analysis (possible regarding the whole genome).

_____ I do **not** want to be notified of information collected in this study about me or my child.

_____ I would like to be notified of information collected in this study about me or my child.

_____ I would like to allow the investigator to contact me by phone if necessary.

_____ I would like to have my blood and information completely anonymous. Anonymization cannot be undone at a later time, meaning the data collected will never be traced back to my person.

Please complete the appropriate section, A or B:

A. Adult patient consent	**B. Parent's permission for a minor patient**
I have read the explanation about this study and have been given the opportunity to discuss it and ask questions. I hereby consent to take part in this study.	I have read the explanation about this study and have been given the opportunity to discuss it and ask questions. I hereby give permission for my child to take part in this study.
_____ (Date and signature of adult patient)	_____ (Date and signature of parent/guardian) _____ (For relatives, please indicate relationship to the patient)

_____
(Date and signature of explaining person)

_____
(Date and signature of principal investigator)