

**Declaration of consent to participate in the study on  
Identification and characterisation of genetic alterations  
in rare diseases**

**at the Institute of Human Genetics / University Centre of Rare Diseases Leipzig**

Family number:

Subject number:



**Institute of Human Genetics**

Philipp-Rosenthal-Str. 55, 04103 Leipzig  
Prof. Dr. med. Johannes Lemke, +49 341 97 23800, [johannes.lemke@medizin.uni-leipzig.de](mailto:johannes.lemke@medizin.uni-leipzig.de)  
Prof. Dr. med. Rami Jamra, +49 341 97 23803, [rami.aboujamra@medizin.uni-leipzig.de](mailto:rami.aboujamra@medizin.uni-leipzig.de)

**Board of the Centre for Rare Diseases**

Human Genetics, Prof. Dr. med. Johannes Lemke, Dr. med. Constanze Heine  
Internal Medicine, Prof. Dr. med. Christoph Baerwald  
Pediatrics, PD Dr. med. Skadi Beblo, Dr. med. Maria Arélin  
Laboratory Medicine, Prof. Dr. rer. nat. Uta Ceglarek  
Neurology, Prof. Dr. med. Florian Then Bergh

**A General remarks**

We have been informed by the consulting doctor / physician mentioned below about the purpose, course and significance of the study and the benefits and risks that may be associated with it. We have read the written information. All our questions have been answered to our satisfaction. We have received a copy of the information and consent form. We had plenty of time to think about and finalize our decision to participate in this study. We are aware that participating in the study is free of charge for us and that we are not paid for the use of our blood-, oral mucosal- or tissue samples, and that we have no entitlement to any compensation, bonus, or other financial benefits that may result on the basis of the research with our samples.

**B1 Consent to the collection and use of blood or tissue samples**

We agree to the collection, use and unlimited storage of a venous blood sample and / or oral mucosa sample from me / us and our minor child or patronized relatives and hereby entrust the blood or tissue samples taken from us to the above mentioned responsible persons. We agree that the samples under the responsibility of the above mentioned persons are stored in encrypted form (ie so that assignment to us as persons is only possible via further aids - such as a reference list) for studies like the above mentioned and detailed in the Information sheet, which includes the installation of cell lines for genetic and molecular biological analysis. These methods also include the analysis of genetic sequence by genome-wide- or partial sequencing, genome-wide analysis of gain and loss of genetic material, and studies on established cell lines.

We agree that the samples taken or clinical data may also be used in scientific projects other than those mentioned above.

Yes  / No

We agree that the samples taken or clinical data may be sent to other clinics or institutes in Germany or abroad within the framework of the above-mentioned and other scientific cooperation projects.

Yes  / No

**B2 Revocation of approval for sample use**

We know that we can revoke our consent to the use of our samples at any time without giving reasons to the above-mentioned institution / person and that this does not affect our further medical care. We are aware that the destruction of the samples and the associated clinical information at our request is not possible, if they have been so encrypted ("anonymized") that a connection between the samples and us as persons is not or is only possible with a disproportionately large amount of time, cost and labor.

**C1 Data protection declaration in accordance with the EU General Data Protection Regulation (GDPR)**

We agree that the persons mentioned in the introduction or one of the employees of the institutions named in the introduction may take a look at our original medical records. Personal data is stored pseudonymised on data servers, partly on 'cloud' servers within Europe. We agree that data concerning us (including, in particular, medical data from our medical records) is stored in the study database in encrypted form under the responsibility of the above-mentioned institution, exchanged with cooperation partners and project managers involved in the study and in a pseudonymised form for publication the study results are used.

We agree that the data collected in the study may also be used for other **scientific cooperation projects** on rare diseases at other hospitals or institutes in Germany or abroad.

Yes  / No

We agree that the data collected during the study may be entered in pseudonymised form in **variant and phenotype databases**.

Yes  / No

We agree to the **storage of photos** of me / us / the patronized person in the encrypted project database.

Yes  / No

We agree to the **use of photos** of me / us / the patronized person in the context of medical or scientific lectures (without naming name, address or similar personal information).

Facial features completely recognizable: Yes  / No  / We ask for prior consultation

Only individual parts of the face may be shown or use of eye bars: Yes  / No  / We ask for prior consultation

Only certain parts of the body may be shown (specify if necessary .....): Yes  / No  / We ask for prior consultation

We agree with the **publication of photos** of me / us / the patronized person in a medical or scientific journal, including their online edition (without naming name, address or similar personal information).

Facial features completely recognizable: Yes  / No  / We ask for prior consultation

Only individual parts of the face may be shown or use of eye bars: Yes  / No  / We ask for prior consultation

Only certain parts of the body may be shown (specify if necessary .....): Yes  / No  / We ask for prior consultation

**C2 Revocation of consent to data usage**

We know that we may revoke our consent to the use of our collected and generated data at any time without giving any reason to the institution or person named above and that this does not affect our further medical care. We are aware that in the case of anonymous storage of our data, their deletion at our request is not possible.

**D Information about study results**

If, within the study described above, results are obtained that are of direct relevance to the initial question, I would like to be informed about this.

Yes  / No  (index person)      Yes  / No  (mother)      Yes  / No  (father)      Yes  / No  (relative:.....)

If, within the study described above, incidental / secondary findings are collected that are not related to the initial question, but nevertheless have medical relevance for me (eg. regarding the possibilities of therapy, prevention, early diagnosis, etc.), I would like to be informed about this and take into account that there is no claim to completeness.

Yes  / No  (index person)      Yes  / No  (mother)      Yes  / No  (father)      Yes  / No  (relative:.....)

**E1 Participants of the investigation**

\_\_\_\_\_  
*Name*                      *Date of birth*              *reference number*

\_\_\_\_\_  
*Name*                      *Date of birth*              *reference number*

\_\_\_\_\_  
*Name*                      *Date of birth*              *reference number*

\_\_\_\_\_  
*Name*                      *Date of birth*              *reference number*

\_\_\_\_\_  
*Name*                      *Date of birth*              *reference number*

\_\_\_\_\_  
*Name*                      *Date of birth*              *reference number*

**E2 Signatures**

**Index person** (if necessary legal guardian)

\_\_\_\_\_  
*Name*                      *Date of birth*

**further relatives** (if necessary legal guardian)

\_\_\_\_\_  
*Name*                      *Date of birth*

**mother** (if necessary legal guardian)

\_\_\_\_\_  
*Name*                      *Date of birth*

**father** (if necessary legal guardian)

\_\_\_\_\_  
*Name*                      *Date of birth*

Place, date, name and address or stamp of the consulting doctor / physician