Information for participants

Genotype-phenotype correlation on GRIN-related disorders

Universitätsklinikum Leipzig
Medizin ist unsere Berufung.

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Dear parents, dear legal guardian,

Your child or person in care has been diagnosed with a GRIN-related disorder. With this letter, we invite you to have your child or person in care in the above-mentioned study. Please read the following information carefully. Then you can decide whether you would like your child or person in care to participate in the study. Please take enough time and ask the study group any questions that are important for you.

Procedure of the study

In this study, we want to investigate clinical and genetic findings of GRIN-related disorders and collect data of affected individuals in a local registry at the Institute of Human Genetics at the University of Leipzig, Germany.
In the study we will collect retrospective data on genetic causes, symptoms, course of the disease, treatment and findings of your child or person in care. The data will be evaluated and stored pseudonymized.
Registration to and participation in the study will require the completion of an online questionnaire. A case-specific login and a case-specific ID number will be generated. Depending on your consent, you can then enter personal and anamnestic data as required. Due to the case-specific login, you can edit / correct / delete / complete the entered data at any time.
The study group at the Institute of Human Genetics in Leipzig will have access to all data in order to perform genotype-phenotype analyzes.

Aim of the study

- Create a registry for GRIN-related disorders
- Better understanding of genotype-phenotype correlations
- Publication of the study results

Risks

The study is based on analyses of pseudonymized clinical and genetic data. There are no significant risks or inconveniences to participants or relatives.

Benefits

There is no immediate personal benefit from participating in this study. However, the study is intended to provide a better understanding of the disease, the course of the disease and the correlation between the different genetic variants and the respective phenotypes. These findings may also be relevant for an improved therapy in the future and thus may possibly benefit your child or person in care as well as other patients with GRIN-related dirsorders in the future. A further benefit of the registry is (depending on the individual consent) the possibility to re-contact study participants with potentially relevant novel information, such as information on future independent therapy studies.

Legal framework

The legal basis for the processing of the personal data is your voluntary consent in accordance with the GDPR and the Declaration of Helsinki (Declaration of the World Medical Association on Ethical Principles for Medical Research on Human Beings).

Data security

During the study, medical reports and personal information from your child or person in care will be stored in the database in pseudonymized form without limitations and in accordance with medical confidentiality laws and data protection regulations. All data will only be used for the purposes of this study.

Consent to the processing of personal data and right to retract

Processing and storage of your personal data is only legitimate with your voluntary consent (Article 6 GDPR). You have the right to retract your consent at any time without justification. However, retraction of consent does not affect data that has already been processed/published prior to the retraction. Neither non-participation nor retraction will have any disadvantages for your child or person in care with regard to further treatment, etc. In case of retraction, all personal data will be deleted (Article 7, paragraph 3 GDPR).

If you have any inquiries, concerns, requests, etc, please contact the study group at the given address.

We would be very thankful for your participation in our research project!