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UNIVERSITÄT LEIPZIG
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Patient Information

for participating in the project

Angelman Syndrome Online Registry

at the Institute of Human Genetics, University of Leipzig Medical Center

Projectmanager:
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Dear patients,
dear parents,
dear legal guardians,

your child or person in care has been diagnosed with Angelman syndrome. 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 the Angelman syndrome and collect data of affected patients 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 (in accordance with the European General Data Protection Regulation (EU-GDPR): replacing the name and other identifying characteristics to exclude or make it significantly more difficult to identify the data subject).

Registration and participation in the study will require the completion of an online questionnaire with an online consent form. A case-specific link and a case-specific return-code will be generated. Depending on your consent, you can then enter personal and anamnestic data as required. Due to the case-specific link (URL) and case-specific return-code, you can edit, correct, delete or complete the entered data at any time.

For some of the questions it is possible to upload documents (e.g. molecular genetic reports, findings on electroencephalography and magnetic resonance imaging, doctor's letters, etc.) which may then be viewed by an employee of the study and transferred pseudonymously to the study database. 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 the Angelman syndrome
- Better understanding of genotype-phenotype correlations
- Publication of the study results
- Improving care for affected patients based on the results of the registry evaluation

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 Angelman syndrome 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

Medical confidentiality and data protection regulations will be observed. During the study, medical findings and personal information from your child or relatives will be stored in pseudonymized form in the database and, if necessary, with your consent, electronically stored in a personal file in our patient management system. The study management will take all reasonable steps to ensure the protection of your data in accordance with German data protection standards and those of the European Union. The data are secured against unauthorized access. Data collected during the study will be kept indefinitely after study completion or until a request for deletion is made. The data will be used exclusively 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). The consent is voluntary. 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 would like to exercise one of these rights, please contact the project managers.

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!