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Creation and evaluation of a patient registry for Dup15q syndrome

Information leaflet for legal guardians/representatives

Dear parents and guardians,

We cordially invite your child or ward to participate in the above-mentioned study. Please read the following information carefully. You also have the possibility of receiving an oral explanation. You will find contact details in the information brochure below. You can then decide whether your child or ward should participate. Please allow plenty of time and ask the study team any questions you may have.

What is Dup15q syndrome?
Dup15q syndrome is a rare chromosomal abnormality caused by at least one extra copy of the 15q11.2-q13.1 region of chromosome 15. Symptoms include decreased muscle tone (muscular hypotonia), motor, language and cognitive development disorders, behavioral problems and sometimes seizures that are difficult to treat.

What are the aims of the study?
- Creation and evaluation of a multilingual registry of Dup15q patients
- Long-term documentation of disease progression in Dup15q patients to better understand this rare disease
- Study of the link between genetic diagnosis (DNA defect) and clinical picture (disease characteristics)
- Facilitating contact with Dup15q patients for future studies (e.g.: clinical trials)

How does the study work?
Our study aims to collect and evaluate clinical and genetic information from patients with Dup15q in a registry. As part of this study, we aim to collect data on the following aspects:
- Disease manifestation (e.g. epilepsy, infantile development, other symptoms, comorbidities)
- Clinical findings (e.g. genetic tests, EEG, MRI, diagnosis of autism)
- Treatment (e.g. anti-epileptic treatment, technical aids)
- Your family's quality of life

For this purpose, we use an online questionnaire that you can easily fill in at home. Once you've registered, we'll ask you once about your developmental progress and other important medical and day-to-day aspects (time required: 1 h to 1 h 30).

We'll then ask you to report any changes or new aspects every year. You will receive a reminder with a link to the questionnaire (time required: approx. 30 to 45 minutes). This questionnaire will enable us to follow the evolution of your child's or ward's illness over a long period.

Is there a personal benefit for us or our child/ward?
Analysis of data from the Dup15q patient registry is intended to improve general understanding of this rare disease. It should make it possible to compare current treatment options and prepare for the development of new therapeutic options. Every participant in the study stands to benefit directly from these long-term results. Placing your child or ward in the full spectrum of people with Dup15q syndrome can help you better understand the severity of the disease and the prospects for the future. Finally, the registry aims to facilitate contact with Dup15q patients for future studies. Participation in the study does not give rise to any financial compensation in the form of an expense allowance.

What are the risks associated with participation?
The study does not present any risks for participants. No additional examinations, treatments or therapeutic modifications will be carried out as part of the study.

Data protection information
Practical information about the patient registry:
Once we have received your consent and e-mail address, a file with a pseudonymized ID number will be automatically created in our Dup15q database using REDCap software. You can then enter all relevant information about your child or ward in the register. With your personal access code, you can save, modify, add to or delete your data at any time. You can send your child's or ward's test results (EEG, MRI, molecular genetic diagnostics, etc.) directly to the Dup15q registry team, and these data will be stored separately. These results will be transferred to the database by a member of the study staff. This ensures that the database contains only de-identified (pseudonymized) data.

General:
Medical confidentiality and data protection rules will be respected. During the study, medical results and personal information about you and your child or ward will be collected and stored electronically on a separate study server. Study-relevant data will be stored, evaluated and, if necessary, shared with participating universities/clinics exclusively in pseudonymized form. As part of this study, pseudonymized data will also be shared with third countries outside the EU and the European Economic Area for analysis purposes. These are countries for which the European Commission has determined an adequate legal level of data protection.
Pseudonymization“ refers to the processing of personal data in such a way that it can no longer be attributed to a specific data subject without the use of additional information (”keys"). This additional information will be stored separately and will be subject to technical and organizational measures designed to ensure that the personal data is not attributed to an identified or identifiable natural person.

The study management will take all reasonable steps to ensure that your child's or ward's data is protected in accordance with the European Union's data protection standards. Data is protected against unauthorized access. Decryption is only authorized by the study director. As soon as the purpose of the research permits, personal data will be anonymized. Anonymization means modifying personal data in such a way that the person concerned can no longer be identified, or only at disproportionate cost or delay. As the evolution of the disease must be documented over the long term, the data will be kept until you revoke your consent or until scientific results are available that no longer require further investigation.

The data you provide or collect as part of the study will be used primarily for the research questions described in this information notice. However, further studies using this data may be required in the future, and will be addressed in other research projects. The exact research questions cannot be specified at this time. However, the research focus would be limited to the following research areas: research projects on Dup15q-associated diseases. These future research projects will be examined separately by the relevant ethics committee. You will not be asked to provide any further information or to give your consent.

You will only be contacted again at the e-mail address you have provided regarding other study projects, such as therapeutic studies, with your express consent.

The person responsible for collecting personal data relating to the study is:

Study director
Prof. Dr. med. Maja Hempel
Heidelberg University Hospital
Genetics outpatient clinic
Im Neuenheimer Feld 440
69120 Heidelberg
Tel: +49 6221 56-5081
E-mail: maja.hempel@med.uni-heidelberg.de

Other members of the Dup15q registry team:
Verena Romero
Sebastian Burkart
Camila Gabriel
E-mail: Dup15q.register@med.uni-heidelberg.de

If you have any questions about data processing and compliance with data protection requirements, please contact the facility's Data Protection Officer:

Dr. iur. Regina Mathes
Im Neuenheimer Feld 672
69120 Heidelberg
Tel: +49 6221 56-7036
E-mail: datenschutz@med.uni-heidelberg.de
Internet: https://www.klinikum.uni-heidelberg.de/organisation/zentrale-einrichtungen/datenschutzbeauftragter

In the event of unlawful data processing, you have the right to lodge a complaint with the following supervisory authority:

State Commissioner for Data Protection and Freedom of Information Baden-Württemberg
P.O. Box 10 29 32, 70025 Stuttgart
Königstraße 10a, 70173 Stuttgart
Phone: +49 711/61 55 41 - 0
Fax: +49 711/61 55 41 - 15
E-mail: poststelle@lfdi.bwl.de
Internet: http://www.baden-wuerttemberg.datenschutz.de

Confidentiality risks (e.g. the possibility of patient identification) exist whenever data derived from a patient's genetic material is collected, stored and transmitted as part of a research project. These risks cannot be completely eliminated and increase with the number of data items that may be linked, especially if you publish genetic data online yourself (e.g. for genealogical research). The study management assures you that it will make every effort, within the limits of the state of the art, to protect patient privacy and will only share data with projects that have an appropriate data protection policy.

Voluntary participation / Withdrawal
Participation in the study is voluntary. If you wish to participate, we ask you to sign the enclosed consent form. You may withdraw this consent at any time, in writing or verbally, without giving any reason and without any inconvenience. To withdraw your consent, please contact the study management or the staff in charge of your care. If you withdraw your consent, you can decide whether the data collected about you for the study should be deleted or whether it can continue to be used for the purposes of the study. Even if you have initially agreed to further use, you may later change your mind and request

deletion of the data; please also contact the study director or staff in charge of you for this purpose. If data has already been included in scientific evaluations or anonymized, deletion is no longer possible.

Will participation incur any costs for us? Will we receive any payment or reimbursement of expenses?
Participation in the study is free of charge. You will not receive any compensation for the use of your child's or ward's data.

Additional information/contact information for any questions:
For further information and to obtain information on the general results and conclusions of the study, please contact Professor Maja Hempel (tel.: +49 6221 565081, email: maja.hempel@med.uni-heidelberg.de), the study director.

We would love for you to participate in this research project!