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Questions and answers (FAQ)
Dup15q patient registry

What is the Dup15q registry?

The Dup15q registry aims to collect data on the natural history of Dup15q syndrome, a rare disease, in a database. Digital questionnaires are used to collect information on disease presentation (e.g., epilepsy, child development, other symptoms, comorbidities), clinical findings (e.g.: genetic tests, EEGs, MRIs), treatments used (e.g.: medications, assistive devices) and families' quality of life. It is also possible to send results (e.g.: molecular genetic results, diagnostic test results) to the registry team by e-mail. The project is initially scheduled to run for 10 years, and aims to enable long-term observations through annual data collection. The software used for this is called REDCap (Research Electronic Data Capture). All questionnaires are digitized and can be completed via any Internet browser.

Why do we need a multilingual European Dup15q register?

There is currently no Dup15q registry in German or French. From now on, affected German- and French-speaking families will be able to participate in a patient registry for Dup15q. The Dup15q registry enables data on Dup15q patients to be collected and processed centrally at the University of Heidelberg in Germany. In addition, the Dup15q registry will be expanded to include multiple languages to reach affected families across Europe. The database will also facilitate contact with affected individuals for future German and international studies.

Who can participate in the Dup15q registry?

The Dup15q register includes people affected by Dup15q, regardless of age, severity or nationality. Questionnaires are completed by parents, relatives or caregivers of people with Dup15q. People with microduplications in the 15q11 to 15q13 region, outside established Dup15q syndrome, can also participate. Questionnaires are currently available in German, French and English. Other languages will be added over time.

How often is data collected?

The first data collection will take place via a questionnaire immediately after your registration.
You will also automatically receive a link to the follow-up questionnaire at annual intervals.

What happens to the data I enter?

The personal data you enter on the consent form (registration) and the data collected via questionnaires on disease progression, therapies and quality of life will be stored separately. This means that only the person in charge of the study can establish a link between the person and the data collected, using a pseudonymization key.

If anything has changed (for example, if you have a new e-mail address), please inform us as soon as possible. To do so, please contact the study director or a member of the Dup15q registry team (see below).

Will my data be published?
Pseudonymized (= de-identified) data is stored in a database on the REDCap (Research Electronic Data Capture) server. The data collected is analyzed at regular intervals by the Dup15q registry team with the aim of describing the natural history of the disease.

What happens to the documents I share?
You can send the results of the study participant's reports/tests (EEG, MRI, molecular genetic diagnostics, etc.) directly to the patient registry e-mail address. They are stored separately from the rest of the data. The results are then transferred to the database by a member of the Dup15q registry team.

Do I have to remove personal data (e.g.: name, address) from the documents I supply?
All documents provided will be reviewed by a member of the Dup15q registry team. Relevant results will be de-identified (= pseudonymized) and transferred from the documents to the study database. This ensures that the database contains only de-identified (= pseudonymized) data. As a result, you don't need to remove any personal data from the documents you send.

Can I change my data later?
You can change, delete or add to your entries at any time using your personal link and a return code. For instructions, click on “Save and continue later”.

How can I let you know if something has changed (e.g.: my e-mail address)?
If something has changed (e.g.: you have a new email address), please let us know as soon as possible. To do so, please contact the study director or a member of the Dup15q registry team (see below).

Will my data be published?
The data collected will be published in pseudonymized or anonymized form. This is mainly to provide information about Dup15q for further research. The resulting public attention is intended to increase general knowledge about Dup15q and stimulate interest in new studies with Dup15q patients.

How can I withdraw my consent?
You may withdraw your consent at any time. This can be done both in writing and verbally. If you revoke your consent, you can decide whether the data collected from you for the purposes of the study should be deleted or whether it can continue to be used for the purposes of the study. Even if you initially agree to further use, you may later change your mind and request that the data be deleted. There will be no inconvenience to you if you withdraw your consent. If you wish to withdraw your consent, please contact the study director or a member of the Dup15q registry team (see below).

Why is genetic testing required?
In order to establish a possible link between genetic variants and disease severity, a genetic diagnosis must be established. Such a connection between genetic diagnosis and disease onset is called “genotype-phenotype correlation”. The aim is to determine whether a specific genetic variant in the Dup15q region is associated with a certain degree of severity, or even with certain symptoms.

Why are symptoms and development investigated?
The specific symptoms of Dup15q, the additional symptoms that accompany it and any secondary diseases are investigated to create the most accurate picture possible of the disease. This allows us to describe the natural history of Dup15q.

Why do we ask about behavior?
In many cases, Dup15q manifests itself through typical behavioral patterns, which are also recorded in the Dup15q register. Like the symptoms and their evolution, these behaviors are part of the Dup15q clinical picture. Describing them should provide a better understanding of the disease.

Why do we ask about therapy?
Information on anti-epileptic treatments, technical aids and other forms of therapy is collected in order to study existing therapeutic options for Dup15q and their ability to relieve symptoms. This enables us to examine the impact of different therapies on the quality of life of people with Dup15q and their families.

Why do we ask about socio-economic status?
The “Family situation” section asks for information about the affected person's family and their living conditions. It's important to consider patients holistically in their environment. This helps us to better understand the symptoms, the consequences of the disease and the resources available.

Why is quality of life assessed?
Assessing quality of life is one of the most important aspects of the Dup15q registry. Improving the quality of life of affected individuals and their families is one of the main objectives of Dup15q research.

Do I have to complete the questionnaire in one go?
You can stop your questionnaire at any time and resume it later. There's a button at the bottom of the page for this purpose: “Save and continue later”. Don't forget to write down your return code. You can receive your personal link to your questionnaire by e-mail.

I've lost my return code: how can I access my questionnaire again?
If you have lost your personal return code, please contact a member of the Dup15q registry team (see below) and follow their instructions to access your questionnaire again. You can also restart the questionnaire from the beginning. However, all data entered up to that point will be deleted.

Why are certain questions mandatory?
Questions considered particularly important need to be answered. This applies in particular to the questionnaires used to calculate the defined Dup15q scores. Even if not all questions are compulsory, we ask you to answer the questionnaire as completely as possible.

Why can't I answer “Don't know/No answer” to certain questions?
As the scores used are already established internationally, the response options are predefined. To enable internationally comparable Dup15q scores to be established, the response options are predefined. It is therefore impossible to add a “Don't know/No answer” option. This ensures accurate score calculation. If you have any doubts about answering these questions, please check “Never” or “No”.

Who can I contact if I have problems or questions?
If you have any difficulties, questions or comments, you can contact the study director or a member of the Dup15q registry team at any time:

Study director:
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Other members of the Dup15q registry team:
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