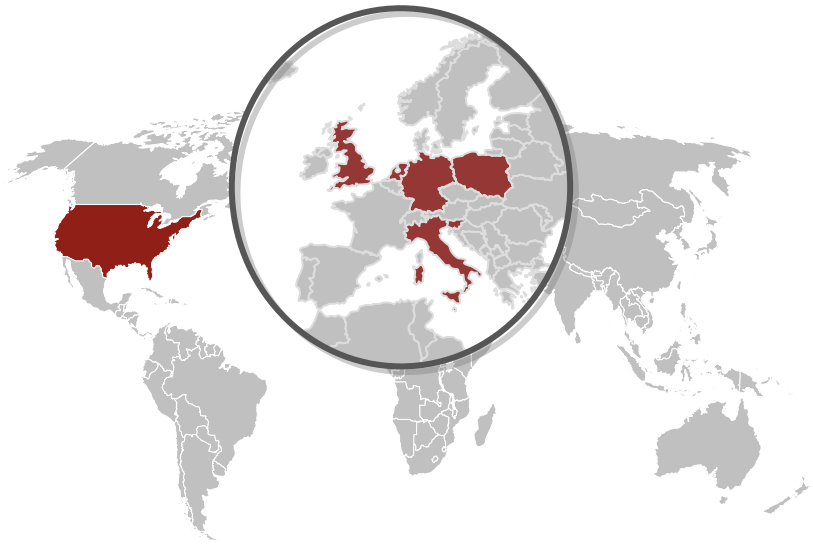
TIRCON

Treat **I**ron-**R**elated **C**hildhood-**O**nset **N**eurodegeneration



TIRCON – Information leaflet

Standardized NBIA patient registry and natural history study

Biomaterial bank und biomarker study

www.tircon.eu

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General information

This leaflet is intended to inform you about the patient registry and the biomaterial bank within the international project **TIRCON** (Treat Iron-Related Childhood-Onset Neurodegeneration). Both of these subprojects in TIRCON serve the goal of improving the research on various forms of the disease group **NBIA** or **Neurodegeneration with Brain Iron Accumulation**. NBIA is a group of very rare, genetic diseases which result in serious impairment of physical health. There is currently no cure for NBIA. Although the genetic cause has already been identified in many patients, still, little is known about how the disease evolves and proceeds or its biological basis. The international registry (incl. natural history study) and the biomaterial bank (incl. biomarker study), incorporated within TIRCON are expected to foster a better understanding of NBIA and the development of new therapies.

What is a patient registry and what are its aims?

A patient registry is a scientific database that records disease-related information from patients with a specific disease. The registry presented here collects the data of patients with **NBIA**. The registry was developed with participation of all TIRCON partners and was technically implemented by the Institute for Medical Statistics and Epidemiology at the Technical University Munich (TUM).
Under TIRCON, information from about 400 patients in Germany, Great Britain, Italy, Poland and the USA, with possible participation from other countries, will be collected. By collecting data from as many patients as possible, a better understanding of the disease's cause and course, which could eventually lead to development of new treatment strategies, is expected to be gained.

What are the biomaterial bank and the biomarker study?

As part of the biobank, blood, urine and possibly skin or muscle cells from patients or healthy control persons (e.g. relatives) will be stored. The NBIA biobank is conducted by the Institute for Human Genetics at the Technical University Munich. The biosamples will also be stored and processed there.
A biomarker study will be carried out with these biomaterial samples. Biomarkers are biological characteristics such as proteins which may give some indication of normal or abnormal biological processes in the body. Within this research project, DNA (desoxyribonucleic acid), RNA (ribonucleic acid) and metabolites (metabolic products such as carbohydrates and fats) will be analyzed. The biomarker study pursues the goal of identifying changes in genetic material or endogenous proteins and metabolic products which may cause or promote the disease or may correlate with the disease's course.

Who can participate?

- Patients with NBIA confirmed by imaging and/or genetic testing or with high-grade suspicion of such a disease
- For the biobank/biomarker study additionally: control persons – this could be: relatives who do not have (and will not have) signs of the disease or patients' friends and acquaintances

What do the participants need to do …

… if they agree to participate in the registry and the natural history study?

If there is written consent to participate in the study, data will be collected by the treating physician during each visit. For the longitudinal study, the follow-up visits typically occur annually or more frequently when needed.

The information collected by the physician will be saved electronically and become part of the NBIA database. These data will include among others:

- family history
- disease history
- clinical exams
- imaging
- results of any further technical tests
- medication history
- genetic test results
- various questionnaires (e.g. on quality of life, sleep)
- laboratory examinations

… if they agree to participate in the biomaterial bank/biomarker study?

If there is written consent to participate in this study, the treating physician will arrange a blood draw and collect urine, process the samples and send them to the biomaterial bank. The participants in the biomaterial bank are preferably asked to fast for the blood collection.
Under TIRCON, it is not envisaged to take samples of skin or muscle tissues. If, however, skin or muscle tissues have been sampled or will be sampled in the future during a routine examination, we would like to use these tissues and make them available to the biomaterial bank for research purposes.

Does simultaneous participation in both studies need to occur?

For the scientific processing, it certainly makes the most sense if a patient is included in the registry while also providing samples for the biobank. In this case, the results of both studies may be linked together resulting in what we would expect to be the largest gain of knowledge. Patients can be enrolled in both studies as part of an outpatient consultation at a clinical TIRCON centre.

If, for any reason, an enrollment in both studies is not possible or desired, a patient can participate in the registry/natural history study or in the biobank/biomarker study only. Participation in just the biobank/biomarker study may also take place outside the clinical TIRCON centre, for example at the general practitioner or at a local institution. In any case, strict compliance with the specifications for a blood draw is necessary. In this case we ask you to contact the biobank team (Dr. Prokisch, see below for contact details).

What are the benefits of participating?

The participation in the patient registry and the biobank will not provide any direct benefit to the participants. However, the collected data may result in new knowledge, for example about the causes and course of NBIA or the factors that influence the course of the disease. We ultimately hope to make better therapy concepts available for patients in future by the gain of knowledge.

Who can I contact with further questions?

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More detailed information about the registry and the biobank will be available at the study center next to you:

http://www.tircon.eu/nbia-clinical-network/tircon-clinical-centres