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Name:

DOB:

Patient Idenfication

Clinic Identification (stamp)

International Registry for Osteopetrosis

INFORMATION SHEET FOR LEGAL GUARDIANS

Dear parents / legal guardians,

The attending physicians have diagnosed osteopetrosis in your child. This disease may cause many problems in different organs, with a very variable presentation for each patient. Some patients show have such severe disease that treatment by bone marrow or stem cell transplantation is necessary. Other patients present a milder form of the disease and do not need to be transplanted.

Osteopetrosis is a rare inherited disorder. Several different genetic varieties of osteopetrosis have been described during the past decade, but so far we only have limited knowledge about the disease course in each different genetic type of the disease . In order to gain more information on this rare disease, the treatment options and long-term consequences in adult life, a European study has been initiated with the aim to collect medical data of the patients and to store them in a **computerized database (registry)**.

The information to be collected would include blood counts, bone marrow analysis, results of X-rays , ultrasound examinations, MRI and CT scans, as well as information on all the clinical symptoms of your child, their treatment including details on any haematopoietic stem cell transplantation procedure and information on how your child copes physically and psychologically with the disease. The collected data will allow comparison of different disease manifestations, treatment options and disease course, enabling the doctors and researchers to better understand how these rare diseases develop, how they influence patients' lives and how we can better help the patients.

The data will be collected by the attending physician during diagnosis and treatment, they will be filled in special designed questionnaires and sent to the supervisor of the registry at the University Children's Hospital in Ulm, Germany: forms to be completed include an initial clinical information form, a form on the transplantation procedure (if applicable), and annual follow up forms concerning clinical status and disease course. Patients' data will then be transmitted in a pseudoanonymized manner to the electronic Osteopetrosis documentation system, where they will be stored. „Pseudoanonymizing" means to delete and/or change the data which allow identification of a patient, but keeping a patient's study identification number related to the plain text data. This means that the patient's name and address will be deleted and that they will be identified within the registry only by their unique study identification number. The original plain text data will be stored separately from the patient's study identification number. Only the study staff (that is the supervisor of the central registry, PD Dr. med. Ansgar Schulz, and the study data coordinator , Mrs Sandra Steinmann) are able to relate the unique registry identification number back to the original patient.

Data cannot be accessed by any external person who is not participating in the study. The computer software used encrypts data so that only the above mentioned study staff have access to the data according to the guidelines. The right to allow access to the system and to obtain the control of this access has been delegated to the study staff of the University Ulm, who are responsible for overall control of all data.

If a stem cell transplant has been performed and if you agree, the data may be transferred to transplant databases (as the MedA/MedB data of the European Group of Blood and Marrow Transplantation EBMT). For this purpose a separate written consent is necessary.

Independently from the registry and only if you agree, a **genetic analysis** will be performed in order to determine the specific genetic type of osteopetrosis that your child is suffering from. In this case genetic counseling can be arranged if you wish. If you consent to the genetic diagnosis by a separate signature, the results will be collected together with the other data of you child in the pseudoanonymized database.

These data will be generally be archived for 10 years in the database system. In the following case you and/or your child will be asked again for consent: data storage for more then 10 years after the end of the study, at the moment when your child reaches adulthood, as well as in the case of planned transmission to another database or archive system.

We would like to include your child in our registry. If you agree that a questionnaire concerning your child's disease course can be completed and sent to the central registry in Ulm, Germany, please sign the declaration of consent. A separate consent form has to be sign for the genetic analysis.

Before you decide to participate in the registry, you should know the following facts:

Voluntariness

The participation in the registry is voluntary and you can withdraw at any time and without mentioning the reasons. In this case, data concerning your child will be destroyed. An eventual withdrawal from the study will have no impact on the medical care of your child.

Accessibility of the study supervisor:

In case of any further questions, you can reach PD Dr. Ansgar Schulz (or his substitute PD Dr. Manfred Hoenig) by telephone +49-731-500-57271 (beyond office hours +49-731-500-57192).

Insurance

During the participation in this study you benefit from insurance cover according to the general liability.

Professional secrecy / data privacy protection

All persons, who are taking care of you during this study, are subject to the medical professional secrecy and are committed to the data secrecy.

The study-related results may be used in anonymized form for scientific publications.

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Physician City, date, hour, stamp of the clinic