Institute of Human Genetic
Klinikum rechts der Isar
Technical University Munich

Klinikum rechts der Isar
Technische Universität München

Declaration of Consent*

on the scientific study

Identifying the Causes of Rare Diseases using Genome-Wide Sequencing

* This declaration of consent also applies to persons who take part in the study but do not have the disease themselves (e.g. as parents, adult siblings or as participants in a control group).

Name: _____ First name: _____

DOB: _____

I was informed about the study in a personal conversation by the responsible physician

_____

I declare that I have read the study information for persons who are able to give consent and that all my questions about it have been adequately answered.

If applicable: I have been diagnosed with the following rare disease:

I consent that during my care or treatment the team of the above-mentioned scientific study may obtain information about me and my state of health and may use my already existing medical data as well as, if applicable, data on the course and treatment of my disease (so-called "patient data" as described in the study information). I also consent that this data is recorded, processed and stored in pseudonymised (i.e. encrypted) form.

I consent to the collection and use of my pseudonymised (i.e. encrypted) biosamples, in particular for genetic analyses, which may include the analysis of my entire genetic material - i.e. the genome - as described in the study information. I also consent to the collection and processing of blood samples up to a maximum total volume of 25 ml per year for further laboratory analyses.

Furthermore, I agree to the scientific use of other biosamples from me, such as tissue samples and/or bodily fluids, which were taken for diagnostic purposes and/or in the context of my treatment and are no longer needed (so-called residual materials), as part of this study.

Furthermore, I consent to the collection and analysis of the following additional biosamples from myself (please mark with a cross in the list below):

- □ Fingernail sample
- □ Urine sample (spontaneous urine)

- □ 24-hour urine sample
- □ Saliva
- □ Skin biopsy (tissue sample from the skin only for patients of whom a skin sample is taken anyway as part of the medical diagnosis or treatment, no additional sample is taken)
- □ Muscle tissue (only if a muscle sample is taken anyway as part of the medical diagnosis or treatment, no additional sample is taken)
- □ Cerebrospinal fluid (in patients only as an additional sample up to a maximum of 5 ml during a routine puncture that is being performed anyway)

I transfer ownership and all rights of use of my biosamples to the Klinikum rechts der Isar of the Technical University of Munich.

Communication of medically relevant results of data analysis from the study "Identifying the causes of rare diseases using genome-wide sequencing".

I wish:

☐ to be informed of any clues/hints/indications about the possible cause of my rare disease that may emerge from the study*

and/or

☐ to be informed of additional findings, that put **me or my offspring** at very high risk for any disease for which an effective therapy, useful preventive measures or examinations for early detection of disease manifestation are available.

The information is to be sent to me and/or the following physician:

name: …… first name: ……

address: ……

☐ **not to be informed** of analysis results from this study that are relevant for me.

* does not apply to participants who are not affected by the respective rare disease themselves

Recontacting

I agree to be contacted again by the physicians of this study. Such contact/contacting may be initiated, for example, to discuss additional aspects of my medical history and/or to request further information or biosamples from me.

yes ☐ **no** ☐

I agree that my **pseudonymised (i.e. encrypted)** patient data and biosamples may also be passed on to departments and research groups of the Klinikum rechts der Isar cooperating in this study and to the genetic research laboratory at Helmholtz Zentrum München for scientific investigation. The results of these analyses and the above-mentioned data will be stored, processed, and analysed on **servers at the Klinikum rechts der Isar of the Technical University of Munich**.

In addition, I can decide whether my double-pseudonymised (i.e. encrypted) data and biosamples may be passed on **beyond this study** for scientific purposes to other researchers/research institutions within and possibly also outside the European Union (EU). It should be noted that transfer to recipients in countries outside the EU is only permitted if one of the following conditions is fulfilled:

- The European Commission has ascertained/determined an adequate level of data protection in the respective country, **or** (if this has not been done)
- The Klinikum rechts der Isar signs contractual data protection agreements with its research partners that have been decided or approved by the European Commission or the competent supervisory authority. Research partners/institutions outside the Klinikum rechts der Isar of the Technical University of Munich will, of course, not be given access to my medical records.

I consent that my pseudonymised data and biosamples may be shared beyond the scope of this study, both within and outside the EU, under the conditions stated.

yes ☐ **no** ☐

Any publication of the results of the study "Identifying the causes of rare diseases using genome-wide sequencing" will only be made in a way that does not allow any direct reference to my person.

I agree that my patient data and biosamples will be stored for 30 years from the date of my consent unless I withdraw my consent before that time. In special cases, my data and biosamples may be of great importance for scientific purposes even beyond this time. In this instance, the study administration in consultation with the responsible data protection supervisory authorities and the local independent ethics committees would clarify whether further use of your data and biosamples is permitted. After expiry of the period of use, my biosamples will be destroyed and my personal data will be deleted. If deletion is not possible or not possible with reasonable technical effort, my data will be anonymised by deleting the identification code assigned to them.

My consent to the acquisition of patient data and the collection of biosamples during presentations/visits is initially valid for a period of five years from the signature of my declaration of consent. Should I present again at the Klinikum rechts der Isar or to the physician in charge after five years, I can give my consent again.

Right of withdrawal

My consent is voluntary!

I can withdraw my consent at any time without giving reasons and without any disadvantages arising for me. The legality of the processing based on my consent until the withdrawal is not affected by this. In the case of a withdrawal, the biosamples I provided for research will be destroyed and my patient data stored according to this consent will be deleted. If deletion is not possible or not possible with acceptable technical effort, my patient data will be anonymised by deleting the assigned identification code.

I have been informed about the utilisation of my patient data and biosamples as well as the associated risks and give my consent within the abovementioned context. I have had sufficient time to reflect, and all my questions have been answered satisfactorily.

I received a copy of the study information for participants and the signed consent form after the informed consent consultation.

city, date

first name and surname patient/participant (block letters)

signature patient/participant

I performed the informed consent consultation.

city, date

first name and surname physician (block letters)

signature physician