Institut für Humangenetik
des Klinikums rechts der Isar
der Technischen Universität München
Director: Prof. Dr. Th. Meitinger
Trogerstr. 32, 81675 München
Tel. 089/4140-6381; Fax -6382

Klinikum rechts der Isar Technische Universität München

Information about the study on "Identification of causes of genetic syndromes by genome-wide sequencing"

Dear patient,

We would like to inform you about the study indicated above. For many genetic syndromes the cause has not been identified yet, or may not be identifiable by previously used technology. This also seems to apply to your disorder. With the present study we try to detect the genetic basis of your disorder by using novel technologies that allow for examination of nearly all genes of an individual at once. This study is supported by the Klinikum rechts der Isar and by the Helmholtz Zentrum München.

What data will be generated by this study?

In the present study, genes (DNA) and possibly also gene products (RNA, proteins, metabolites) will be extracted from your biosamples (blood, cells, tissue samples) in order to identify the genetic causes and risk factors of your disorder by using methods of genome-wide gene analysis. "Genome-wide" means that the genetic information of nearly all genes will be analyzed as completely as possible. For that purpose, DNA sequencing techniques will be applied that address the entire genome or relevant parts of it (i.e., whole genome sequencing or exome sequencing, respectively). Besides genetic data, clinical (medical) data will also be used in this study. The information derived from the genetic and clinical data will not serve only for the research on your disease but will also be applied as a control in the research on other diseases.

Which results of the study will be disclosed and how will they be disclosed?

In this study we will not only analyze genes that are already known to relate to your disease. We will also search for novel genetic causes that are not known as yet. Results that are likely to be the cause of your disease will be disclosed to the medical doctor who admitted you to this study. If additional results will be generated in future studies, we will also try to disclose them to your doctor but we cannot guarantee that all future results will be disclosed.

What will be done with secondary results?

Genome-wide analyses may produce results which do not relate to the disease that was the reason for your participation in this study. Such secondary results may include risk factors for certain cancer dispositions, for instance, or information on the carrier state for an inherited disease. Currently, we do not list such secondary results systematically, but we try to identify those that have specific relevance for therapy or timely detection of disease. Results of that kind of relevance may be disclosed, but we cannot guarantee regular, complete, or updated disclosure. We will not disclose carrier states for autosomal-recessive diseases or genetic factors that influence drug sensitivities (i.e., pharmocogenetic sequence variants).

How long will the study take?

This study is open-ended. The research on genetic causes of disease by genome-wide sequencing has been initiated only recently. We expect that sequencing techniques and human genetic knowledge will further improve over the next years. By participating in this study you agree that your samples may be investigated again at a later time, that the data may be reanalyzed, and that the samples may be stored without limit of time.

Where will the data be archived?

The data will be archived at the Institute for Human Genetics, Technical University Munich. Data and biological samples may possibly be exchanged with cooperating institutions in the context of research projects. Before any data exchange is done, the data will be stripped of personal identifiers such as name or address. We will take every precaution to secure your data. **However, we cannot completely exclude the possibility that the data may be stolen in the event of a security breach.**

Where will the data be published?

Data may be published in scientific journals and databases. The European Genome-phenome Archive (EGA; http://www.ebi.ac.uk/ega/) and Decipher (http://decipher.sanger.ac.uk/)) are examples of databases that store genome-wide data. Genetic and clinical data (including images if you agree) will be published only after personal identifiers such as name or address have been removed. Access to this information will only be available to *bona fide* researchers who will use the information for good scientific reasons, and will safeguard the information. However, there is a remote possibility that a third party assigns the data to you. This is possible because genetic data are specific for an individual person. Someone who has obtained partial genetic data from you may compare this data to the sequence data generated in this study and thereby assign the latter to your person.

How to withdraw from this study?

At any time, it is possible to withdraw from this study by sending a written request to the physician who admitted you to this study or to the director of the study. In case of withdrawal your data and samples will not be used for any further investigation. However, data and results that have already been generated or published in scientific journals or databases prior to the date of your request cannot be reversed, undone, or withdrawn. The standard of your medical care will not be affected if you withdraw from the study.

Is it possible to obtain sequencing data?

On request, we will provide you with the "raw" sequence data. Raw sequence data do not contain any medical interpretation. The request can be sent to the physician who admitted you to this study.

If you have any further questions, please do not hesitate to contact us (see phone number above).

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Patient data	
Name	Prename
Date of birth	Phone.:
Street	
ZIP	City

Specimen:
Diagnosis:
Physician:

Consent form for the study on "Identification of causes of genetic syndromes by genome-wide sequencing"

Participation in the study

I confirm that I have read and understand the information sheet for the above study and that the physician undersigned below, orally explained to me the aim, the procedure, and the meaning of this study. I have been given copies of the information sheet and the consent form. I agree with the participation in the above study, with the donation of biosamples (blood or tissue samples) and the unlimited storage of these biosamples and the corresponding results.

Yes ☐ No ☐

Use of the biosamples for other scientific studies

I agree that the biosamples and the generated data may be used in other studies that pursue a different scientific objective. I understand that the biosamples and data may be used in these other studies as controls.

Yes ☐ No ☐

The participation in the study is voluntary

I understand that the participation in the above study is voluntary and that I may withdraw my consent at any time without causing any disadvantage for myself.

Yes ☐ No ☐

Disclosure of results

I understand that those findings that are likely to be the cause of my disease will be disclosed to me by the medical doctor who admitted me to this study.

Yes ☐ No ☐

Secondary results

In this study, secondary results may be generated that do not relate to the disease which is the objective of the study. Secondary results may include risk factors for certain cancer dispositions, for instance, or information on the carrier state for an inherited disease. I understand that such secondary results may be disclosed to me if they have relevance for therapy or timely detection of disease. I understand that I do not have a contractual claim on disclosure, completeness or updating of these results.

Yes ☐ No ☐

Storage and publication of data

I agree that my biosamples and data will be exchanged with cooperating research institutes. I understand that this exchange will be done only after personal identifiers such as name or address have been removed.

I agree that my data may be published in scientific journals and stored in scientific databases if all personal identifiers have been removed. The European Genome-phenome Archive (EGA) and Decipher are examples of databases that store genome-wide data and serve for research purposes only.

I understand that in principle the data published in databases may be assigned to me by a third party who has obtained partial genetic data from me and compares this partial data to the databases.

Yes ☐ No ☐

Participation in the study does not provide any financial profit

I understand that I will not benefit financially in any form when I participate in this study by providing blood or tissue samples.

Yes ☐ No ☐

Participant

Name:

Date: Signature:

Physician

Name:

Date: Signature: Address: