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UNIVERSITÄT
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Tübingen

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Elucidation of the genetic causes of hereditary retinal diseases

(HNE_2022-Vers. 09.02.2022)

Information Letter

Dear Sir or Madam,

You or members of your family have been diagnosed with a hereditary retinal disease. Hereditary retinal diseases are a group of rare eye diseases that lead to functional failures in the perception and transmission of light stimuli in the light-sensitive layer of the eye (= retina). The knowledge about the causes and the development of these diseases is still very limited. We aim to improve this knowledge, because there is no cure for these diseases yet.

In the following we inform you about the aims of the study, the collection, use and storage of your biospecimen in our research biobank, as well as the procedures and measures for the protection of your personal data.

Aim of the study

The Molecular Genetics Laboratory of the University Eye Hospital Tübingen is conducting a long-term study to investigate the genetic causes of hereditary retinal diseases. Our investigations aim to identify the genes involved in these diseases and to detect the disease-causing changes (mutations) in the genetic material DNA (deoxyribonucleic acid) or the messenger RNA (ribonucleic acid). The starting material for the isolation of DNA and RNA is one or more blood samples, which we request from you by this information letter. If you agree, your blood, DNA and/or RNA samples will be included in our biobank, which has been in existence since 1992. Here they will be linked to the corresponding personal and medical data related to your health and disease. This biobank is operated by the Molecular Genetics Laboratory, Institute for Ophthalmic Research, Centre for Ophthalmology, University of Tübingen (contact persons Dr. Susanne Kohl, Dr. Nicole Weisschuh, Prof. Bernd Wissinger).

Genetic tests are performed on the basis of your DNA or RNA samples. In a genetic examination, the base sequence is determined or the structure of certain genes (e.g. deletion/duplication analysis, copy number analysis) is examined. In the human body, our genes provide the information for the production of various proteins that are important for the structure of cells and tissues and for metabolism in the retina. Changes (mutations) in these genes lead to the fact that the corresponding proteins are not formed or not formed in sufficient quantities, or that their biological properties are changed. As a result, there are deficiencies or malfunctions in the retina, which lead to the clinical picture.

If the causative gene is unknown, indirect studies on your DNA can be used to identify the physical location of a causative gene within the genome by a technique called linkage analysis. This requires familial blood tests including affected and unaffected family members. The comparative reconstruction of the inheritance pattern of the genomic segments from generation to generation allows the determination of the segment that is inherited together with the disease predisposition. Such family examinations require a high degree of cooperation from your family. For legal reasons, we are not allowed to contact your family members without prior consent. We therefore ask you, if necessary, to arrange contact with your family members, to arrange for them to be contacted, or to obtain appropriate written consent for contact. For the discussion of your disease with other family members, we may ask you for a corresponding release from medical confidentiality.

As an alternative to linkage analysis, modern high-throughput DNA sequencing technologies are also used in this study. These allow the sequencing of the coding (= protein-forming) sections of all known genes (= exome) or the entire genetic material (= genome) of humans to be determined in a single experiment. These techniques are excellent for identifying as yet unknown "disease genes" for hereditary retinal diseases; however, they also carry the risk of inadvertently detecting further gene alterations that may indicate a predisposition to another disease that may only occur in the future (additional findings). We do not aim for a systematic search for such additional gene alterations. Additional findings will not be reported in the context of this study.

The results obtained in the course of this study serve primarily to gain scientific knowledge and to confirm the clinical diagnosis. A treatment or therapy of your disease cannot be derived from this at the present time in the vast majority of cases. In the medium term, however, the investigations are aimed at developing new treatment concepts on the basis of the scientific findings; many of these are dependent on the exact knowledge of the underlying gene mutation.

The detection of the disease-causing alteration in the genetic material, however, allows a more precise diagnosis to be made in addition to the clinical examination, and in some cases enables to draw conclusions about the future course of the disease. In addition, the hereditary risk for your children can usually be determined very precisely.

Regardless of your participation in this study, however, you have a right of not knowing, i.e. you can decide for yourself whether you want to be informed about the results of the examination. However, participation in this scientific study does not give you a legal right to have certain genetic tests performed or to be informed of the results of the tests.

Minors

For the examination of underage patients, the consent of both parents is required. Investigations for the purpose of pre-symptomatic (before the onset of the disease) detection of a predisposition and for the determination of a predisposition carrier in minors are not performed and are not subject of this study.

Unlimited study duration

Unfortunately, the detection of the disease-causing hereditary mutations has not been successful in all cases so far and requires in some cases very lengthy and complex research investigations. In addition, the biospecimens obtained serve us as valuable reference material for new sampling and testing methods. Therefore, the study is not limited to a specific time frame and the biomaterials and data will be kept and made available for medical research for an indefinite period of time.

Voluntariness and revocation

We hereby expressly state that participation in this study is voluntary. If you do not wish to participate or wish to withdraw your consent at a later date, you will not suffer any disadvantages as a result. By participating, you consent to the use of your blood/DNA and/or RNA samples for the aforementioned studies for an indefinite period of time, or until you may decide to withdraw your consent. You may withdraw your consent to participate in the study at any time without giving any reason, either orally or in writing. To withdraw your consent, please contact: Molecular Genetics Laboratory, Institute for Ophthalmic Research, University of Tübingen, Elfriede-Aulhorn-Str. 5-7, 72076 Tübingen. In this case, your blood/DNA/RNA sample will be destroyed, your personal data will be deleted and the results obtained to date will be anonymized. Data from analyses already performed cannot be removed.

Disclosure of samples to third parties

In the case of supplementary genetic testing at external institutions (universities, research institutes and research companies for non-commercial purposes), the blood/DNA/RNA sample is passed on in encrypted (pseudonymized) form. The samples and data hereby cannot be assigned to your person without the cooperation of our institution.

Sample material and risks during blood collection

Blood collection (for DNA 5 - 20 ml venous whole blood, EDTA tubes; for RNA 10 ml blood in PaxGene or comparable tubes) is performed by a physician or a trained specialist. In individual cases, local bruising (hematoma), thrombosis, inflammation (thrombophlebitis), bleeding, infection, accidental puncture of an artery or nerve, which may also be accompanied by prolonged discomfort, dizziness and syncope, may occur. However, these usually subside within a few days. A blood sample can also be taken by your family doctor / medical officer and the samples sent by mail. In this case, please enclose your signed consent form and make sure that your blood tubes are clearly labeled with your complete name and date of birth.

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Information on Data Protection

Within the scope of the study "Elucidation of the genetic causes of hereditary retinal diseases", personal data (name, date of birth, address, previous findings, study-related findings including imaging procedures, results of study-related genetic examinations, etc.) will be collected and processed. If necessary, your medical data will also be included in the processing. Your data will be documented and archived in paper form and/or in a protected electronic database at the Centre for Ophthalmology of the University Hospital of Tübingen, to which only authorized staff members including doctoral candidates bound to professional and data secrecy have access. All employees involved are bound to secrecy.

Any collection, storage and transmission of data from your biomaterials in the context of research projects involves confidentiality risks (e.g. the possibility of identifying you), especially with regard to information about your genetic material. These risks cannot be completely excluded and increase, the more data can be linked together, especially if you yourself publish genetic data on the Internet (e.g. for genealogical research). The data directly identifying you (name, date of birth, address, etc.) remain at the institution where the samples and data were obtained. Information concerning your health (medical data) is stored separately from the biomaterials. Therefore, this data cannot be assigned to your person without the cooperation of our institution.

Publication of the totality of your genetic information (total exome or total genome) does not take place.

Your personally identifying data will not be disclosed to researchers or other unauthorized third parties, such as insurance companies or employers.

A prerequisite for the use of the biospecimens and data for a specific medical research project is, in principle, that the research project has been evaluated by an ethics committee. This study was also positively advised by the local ethics committee (ethics vote 116/2015BO2 of 09.02.2022).

The biospecimens and data collected in the study can also be used and processed for future research projects of the clinic or the institute.

The biospecimens and data will be processed and used as part of the RetDis biobank and database for an indefinite period of time.
The information obtained in the course of this study may also be transferred for scientific purposes to cooperation partners within the scope of the European Data Protection Regulation and to cooperation partners outside the European Economic Area, i.e. to countries with a lower level of data protection (this also applies to the USA).
The research results from the study will be published in anonymous form in professional journals or in scientific databases. When such research results are published, your identity will not become known. However, we can use a patient list to trace the data back to you in the event of queries.
You can request information about your stored data at any time and have the right to have incorrect data corrected. You can also request at any time that your data be deleted or made anonymous so that a reference to your person can no longer be established.
The study director (Prof. Dr. Bernd Wissinger, deputy Dr. Susanne Kohl, Molecular Genetics Laboratory, Institute for Ophthalmic Research, Centre for Ophthalmology, University of Tübingen, Elfriede-Aulhorn-Str. 7, 72076 Tübingen, Germany) is responsible for data processing and compliance with legal data protection regulations.
In the event of complaints, you may contact the data protection officer of the University Hospital Tübingen or the data protection officer of the state of Baden-Württemberg:

Data Protection Officer
University Clinics Tübingen
Martin Schurer, Data protection team,
Calwerstraße 7/4, 72076 Tübingen,
Tel. 07071 29-87667,
E-Mail: dsb@med.uni-tuebingen.de

State Commissioner for Data Protection and Freedom of Information in Baden-Württemberg
Dr. Stefan Brink,
Postanschrift: Postfach 10 29 32, 70025 Stuttgart
Tel.: 0711/615541-0, FAX: 0711/615541-15,
E-Mail: poststelle@lfdi.bwl.de

For the collection, storage, use and disclosure of your data, your expressed consent is required by signing the declaration of consent to data protection.

The legal basis for the processing of your data are Art. 6, 7, 9, 89 of the General Data Protection Regulation in conjunction with §§ 4, 5, 6, 8, 9, 12, 13 of the State Data Protection Act of Baden-Württemberg in the version applicable as of May 25, 2018.