Westfälische Wilhelms-Universität Münster

UKM Universitätsklinikum Münster

Klinik für Kinder- und Jugendmedizin
– Allgemeine Pädiatrie –

Univ.-Prof. Dr. med. Heymut Omran
Direktor

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_____
Surname, Firstname

_____
Date of birth

Patient Information and Informed Consent Form:

"Characterization of diseases of the ciliated epithelium including Primary Ciliary Dyskinesia (PCD)"

Dear parents,
Your child`s diagnosis or suspected diagnosis of primary ciliary dyskinesia (PCD) has been made. We would like to ask if you and your child are willing to participate in a scientific research project to study the molecular basis of PCD and provide us with relevant material (blood, respiratory epithelial cells, ciliated tissues, sperms) for research purposes. Participation is voluntary. If you or your child do not wish to participate or discontinue participation at a later time, you will be at no disadvantage.

Background

The surface of the airways (respiratory epithelium) is equipped with movable, hair-like structures (cilia) that transport particles (e.g., dust, pollen, bacteria) by a percussive movement. When the cilia do not beat properly, the cleaning of the respiratory tract is disturbed, so that there may be recurrent infections. This occurs in PCD, which is a genetic congenital disease that can be caused by changes in different genes. As of now, numerous (more than 30) genes are known to be altered in PCD. A gene defect can be detected in approximately 60% of patients, however many genes responsible for PCD have not been identified. The genetic defect in a patient is transferred in most cases from healthy parents, who each have a healthy and a defective copy of the gene in question, to the affected child. A person suffers from this disease when both copies of a gene are defective.
Moving cilia are found not only in the respiratory tract, but also in many other organs, e.g., the (female) fallopian tubes, the (male) sperm, the cerebrospinal fluid spaces, the ears and the so-called node of the embryo. These organs may also be affected in PCD patients.

Aim of the study

The aim of this study is to find the exact cause (the responsible gene) for the occurrence of PCD and learn how this defect affects the function of the cilia. The findings are intended to better understand the progression of the disease and to develop new treatment options in the long term.

The course of the study

We would like to ask you for permission to use your blood or isolated DNA for genetic studies and the sample obtained from the nasal brush biopsy from your child (respiratory epithelial cells) and tissue samples of cells (possibly also sperms) from your child to examine cilia. We only use material (blood, cells from the nose, tissue, sperms) that was already taken for diagnostic or therapeutic purposes and would otherwise be discarded.

A single blood sample (5-10 ml) supplemented with heparin or EDTA for preservation is required from each person. This means that you and your child and perhaps also the siblings would donate a blood sample.

The genetic material (DNA) is isolated from the blood and studied. It is used solely for the molecular investigation of hereditary lung diseases. The DNA is stored until the end of the study and its identity is encrypted.

The samples obtained from the nasal brush biopsy (respiratory epithelial cells) and other tissue samples carrying cilia are to be used for research purposes only. We can visualize some components of the cilia (antibody staining) with this method. For example, we can confirm a frequent error in patients (outer Dynein arm defect, > 50% of all PCD cases PCD) with this method. Using these samples, we can also develop other diagnostic procedures.

Benefits and Risks

You and your child will probably have no direct benefits from the study, however, you and we may gain a better understanding of the disease.

There are no health risks from participating in the study because all measures were already performed for diagnostic or therapeutic reasons.

Data management and data protection

The legal basis for processing the aforementioned personal data is the consent in accordance with Article 6 (1), subsection a EU-DSGVO. The study enrollment takes place only if you and your child consent. However, we request permission to save data that is important to improve the diagnosis and understanding of PCD. This includes information about the medical history (presence of typical symptoms) and results of the performed diagnostics (nasal NO measurement, high frequency video microscopy, electron microscopy, immunofluorescence microscopy) as an encrypted number code. Study-related patient material is stored as a pseudonym (i.e., a number code, not a name). All staff involved in the study strictly adhere to medical confidentiality. Only a few authorized employees have access to the patient data list encrypted as number codes.. All disclosure of data is done only by the number code. Relevant intermediate results are published. Individual subjects cannot be identified in the publication. The stored data may be forwarded in pseudonymised form to a co-operation partner in the context of

these publications. All search attempts are logged in lab books. The data is securely stored in an EXCEL table.
The data storage is subject to the provisions of the Data Protection Act and medical confidentiality. International guidelines for the "Good Clinical Practice" (GCP) are fully taken into account.
All electronically recorded data is stored on a server on which software updates are regularly recorded. A monitoring service checks the availability of the register and notifies in case of failure. It is comprehensible which change was made to the data by which user. Modified / overwritten records are retained. The security of the data is ensured by appropriate measures. Of all electronically recorded data, daily backups are made on tapes. The creation of the backup copies is checked regularly. In the event of a physical or technical incident, the information may be recovered by information technology personnel.

The data and tissue are removed or disposed after completion and analysis of the study.

You as parents have the possibility to obtain information about your stored data at any time.

The following person is responsible for the processing of personal data:

Klinik für Kinder- und Jugendmedizin – Allgemeine Pädiatrie – University Hospital Münster
Univ.-Prof. Dr. med. Heymut Omran
Albert-Schweitzer-Campus 1, Gebäude A1
48149 Münster
T +49 (0)2 51 - 83 - 47732

Contact data of the data protection official
Thomas Claes
Date protection officer of the UKM
University Hospital Münster
Albert-Schweitzer-Campus 1
48149 Münster
T 0049 251 83-49694
datenschutz@ukmuenster.de

Notification of rights of the party concerned
According to Article 13 II b *Datenschutzgrundverordnung* (DSGVO – Basic Regulation on Data Protection), you are entitled to the rights listed below.

Information (Article 15 DSGVO and section 34 BDSG)
You are entitled to information regarding the personal data referring to you that are collected, processed or made available to a third party.

Right of protection(Article 21 DSGVO and section 36 BDSG)
The data subject shall have the right to object.

Data transferability (Article 20 DSGVO)
You are entitled to transfer of personal data relating to you that you have made available to us. This allows you request that we transfer these data either to you or, if technically possible, to some other party.

Deletion (Article 17 DSGVO and section 35 BDSG)
On expiry of the periods of custody stipulated by law, you shall be entitled to have your data deleted.

Restriction on processing (Article 18 DSGVO)
In special cases, you shall be entitled to restrict the possible processing of your data. This shall be the case when the data processing is unlawful or you dispute the accuracy of the collected data. You can also demand that processing be restricted when the data are subject to a deletion obligation after the purpose has been achieved, but are, however, required by you for asserting some legal claims. An application has to be made for restriction on processing.

Correction (Article 16 DSGVO)
You are entitled to ensure that any incorrect personal data are corrected. This expressly does not include correction of data in external media/with third parties.

If you wish to exercise one of these rights, please contact the data protection officer at UKM.

You are entitled to file appeal with the supervisory authority:

Landesbeauftragte für Datenschutz und Informationsfreiheit Nordrhein-Westfalen
Postfach 20 04 44
40102 Düsseldorf
Tel.: 0211/38424-0
Fax: 0211/38424-10
poststelle@ldi.nrw.de

However, we request permission to save data that is important to improve the diagnosis and understanding of PCD. This includes information about the medical history (presence of typical symptoms) and results of the performed diagnostics (nasal NO measurement, high frequency video microscopy, electron microscopy, immunofluorescence microscopy) as an encrypted number code.

Designation of the Ethics Committee

As required by law, the scientific research projects have been approved by the ethics committee. This does not mean that the ethics committee has endorsed your participation. It is your responsibility to carefully consider the information in this document and then decide whether you want to participate in the study.

Questions

We will inform you and your child in person about the planned study. For further information, please contact Prof. Dr. H. Omran.
We will inform you during the course of the study as new information becomes available. You are free, of course, to elect not to receive this information.
A discussion of these results should be made with your supervising physician and genetic counselor.

Consent to participate

With regard to the content and the implementation of the study, I agree/ we agree that my/ our child participate in the study to investigate the molecular basis of primary ciliary dyskinesia (PCD). Health risks from the study, except the blood collection, are not expected.

With this signature I /we consent that the results obtained in this study are made available to the treating physician. I/ we can withdraw my/ our consent at any time with my/ our doctor, without notice and for any reason, and without incurring any disadvantages for the treatment of my/ our child.

I/ We understand this information and have no further questions.

Consent to Privacy

1. I/ We agree that in the framework of the research project, the data from my/ our child including health status and medical history, gender, age, weight, height, and ethnic origin (if required by the study) be recorded and encrypted as a pseudonym (name replaced as number code) and made available to:

a) the sponsor of the study for their scientific evaluation

b) the competent Authority to verify the correct execution of the study.

2. Also, I/ we agree that an independent and authorized person who evaluates the study or competent Authority within the research study can inspect the existing personal data from my/ our child, if this is necessary for the review of the study.
I/ We also acknowledge my/ our consent to the method of collection and storage of personal, encrypted data as described under "Data management and data protection". In the case I/ we cancel my/ our participation in the study, the data and tissue from my/ our child will be removed or disposed.

I/ We have have been informed, that my consent for recording, storage and utilization of the data can be withdrawn at any time without any disadvantages at the above named responsible. Such cancellation will not affect the lawfulness of the data processing carried out on the basis of the consent applicable until it was revoked.

I/ We have received a copy of the patient information and consent and agree to the implementation of the study.

I/ We wish that my attending physician is informed
_____ (name) of findings collected in this study.
Please tick: Yes ◯ No ◯

I/ We wish to be informed of findings that are collected as part of the study. Please check: Yes ◯ No ◯

Contact address and phone number to receive findings of the study:

patient: Surname, first name **filled in by patient/ parent**	place/ date, signature (if applicable)
Name of parent (1): Surname, first name **filled in by parent (1)**	place/ date, signature
Name of parent (2): Surname, first name **filled in by parent (2)**	place/ date, signature
Physician: Surname, first name	place/ date, signature