UiO : University of Oslo

Oslo University Hospital

INVITATION TO PARTICIPATE IN A RESEARCH PROJECT

Genetic and functional studies of prospective monogenic diseases

You are invited to participate in this study because you/ your child/ the one you consent for/ have agreed to donate blood and other biological samples for a research purpose as a healthy control.

WHAT IS THE PROJECT ABOUT?

This project has two main aims: i) to help undiagnosed patients with hereditary diseases get a confirmed diagnosis, and ii) to better understand why some gene defects cause serious conditions and to develop therapies for the underlying defect. Our study needs healthy control samples because by comparing healthy and patient samples we will better understand diseases we study. The sample will be used in laboratory experiments. These include different studies of the cells and their DNA. We will also create induced pluripotent stem cells (iPS) and immortalized cell lines derived from blood or skin cells. This way we will create a patient's and healthy control models to study genetic diseases.

FORESEEABLE BENEFITS AND PREDICTABLE RISKS AND BURDENS OF TAKING PART

The study improves our understanding of genetic diseases, which will eventually help us to find new therapies. The sample donation will help researchers and doctors to understand the mechanisms of disease, which enables better genetic counselling and therapy. The study involves taking blood and other samples, which we take during your routine hospital visits and procedures.

VOLUNTARY PARTICIPATION AND THE POSSIBILITY TO WITHDRAW CONSENT

Participation in the project is voluntary. If you/ your child/ the one you consent for/ wish to take part, you will need to sign the declaration of consent on the last page. You can, at any given time and without reason, withdraw your consent. This will not have any consequences for any future treatment. If you decide to withdraw participation in the project, you can demand that your tests and personal data concerning health to be deleted, unless however, the personal data concerning health and tests have already been analysed or used in scientific publications. If you at a later point wish to withdraw consent or have questions regarding the project, you can contact the researcher and medical doctor: Janna Saarela, email: janna.saarela@ncmm.uio.no tlf: +47 22840553 or Emma Haapaniemi, email: emma.haapaniemi@ncmm.uio.no tel. 41384367

WHAT WILL HAPPEN TO YOUR PERSONAL DATA CONCERNING HEALTH?

The duration of this study is 10 years, the project will end on 31.12.2030. Any personal data concerning health that has been recorded about you/ your child/ the one you consent for/ will only be used as described in the purpose of the project. You have the right to access information that has been recorded about you and the right to stipulate that any error(s) in the information that is recorded is/are corrected. You also have the right to know which security measures have been/will be taken when your personal data concerning health is processed. Information about you will be anonymised, all information will be processed and used without your/ your child/ the one you consent for/ name or personal identification number, or any other information that is directly identifiable to you. A code links you and your personal data concerning health via an identifier list. Only doctors and researchers involved in this project will have access to this list. After the project ends the information about you will be stored until 2035 due to requirements and conditions set by regional committees for medical and health research ethic. Your data is very precious to us therefore we might renew or apply for new permits for further research after the study period ends and contact you again to ask for a new consent. If you do not wish to renew your consent, all your data will be destroyed after 2035.

SHARING OF PERSONAL DATA AND TRANSFER OF PERSONAL DATA ABROAD

By agreeing to participate in the study, you are also consenting to that your child/ the one you consent for/ research data as well as biological material can be transferred to another country as a part of research collaboration and publication. This can be a country where law differs from the European Data Protection Law. Norwegian REK does not have the authority to consider later use of information stored in a database abroad. The collaborating institutions have their own ethical permits and they follow the common EU and Nordic countries guidelines. The project manager will ensure that your personal data concerning health is kept safe. Depending on the hypothesis and data analysis method, the collaborating scientists can access the data via TSD web portal service (https://data.tsd.usit.no/). In this case, a federated login will be provided to external researchers that will grant them access to the relevant TSD project. In addition, we might upload the data to European data storage infrastructures such as EBI/ELIXIR's EGA archive (https://www.ebi.ac.uk/ega/about) to give collaborators controlled access. This is mainly in cases where TSD does not support relevant programs or when data needs to merge with multi-national cohorts. The code that connects you and your personal data concerning health will not be released. Your signed consent for participation in this study is retained in NCMM. The samples and data will be coded, and all information will be processed at the laboratory using this code and not your real name. You can't be identified from the resulting scientific publications either.

WHAT WILL HAPPEN TO THE TESTS YOU HAVE TAKEN?

The project will collect and record personal information about you/ your child/ the one you consent for. The hospital personnel will take a blood sample and possibly stool and swab samples (swabs from skin and mucosa) during routine hospital visit. Skin biopsies will be collected during routine procedures performed under anaesthesia, unless you agree differently. Your local hospital coordinates the sampling, please discuss with your treatment team for further instructions. You choose which samples to give and can consent for only specific samples (i.e. blood) while declining others (i.e. skin biopsy).

The tests taken from you/ your child/ the one you consent for/ including blood samples, skin biopsies will be stored at research biobank located at the Norwegian Centre for Molecular Medicine (NCMM) at the University of Oslo (UiO). The persons responsible for the biobank are Emma Haapaniemi and Janna Saarela. We will keep the biological samples in a research biobank until the end of the project. After the project ends your biological samples and data will be anonymised and stored until 2035 due to requirements and conditions set by regional committees for medical and health research ethic.

Since the samples collected from you/your child/ the one you consent for, are very precious to us, we will renew or apply for new permits after the study period ends and contact you to renew and sign new consents. If you do now wish to renew your consent your samples and data will be destroyed after 2035. If you don't want your/your child/ the one you consent for biological samples to be stored in research biobank, you can decline it in the consent below.

INSURANCE

All patients are insured by the national Pasientskadeordningen. The scientists are insured by their employer (University of Oslo, Oslo University Hospital). There is no monetary compensation to the participants.

FINANCE

The study is financed by the combined 50M NOK startup funding from Haapaniemi & Saarela groups (2019-2023) at Norwegian Centre for Molecular Medicine. In addition, we have dedicated grants from Academy of Finland (2.7M NOK 2016-2020), Barncancerfonden, and Instrumentarium Foundation (1.5M NOK total).

APPROVAL

The Regional Committee for Medical and Health Research Ethics has reviewed and approved the Research Project *REC* (ID:77492).

In accordance with the General Data Protection Regulation the NCMM director, Janna Saarela and the project manager Emma Haapaniemi are independently responsible to ensure that the processing of your personal data concerning health has a legal basis. This project has legal basis in accordance with the EUs General Data Protection Regulation, article 6 no. 1a, article 9 no. 2a and your consent. You have the right to submit a complaint on the processing of your personal health data concerning health to the Norwegian Data Inspectorate (Datatilsynet).

CONTACT INFORMATION

If you have any questions regarding the research project, you can get in touch with: Janna Saarela, email: janna.saarela@ncmm.uio.no tlf: +47 22840553 or Emma Haapaniemi, email: emma.haapaniemi@ncmm.uio.no tlf. 41384367 or Monika Szymanska, email: moniksz@ncmm.uio.no tlf. 46504016

Visiting address:
Centre for Molecular Medicine Norway (NCMM)
Forskningsparken
Gaustadalléen 21,
0349 Oslo
Mail address:
P.O. Box 1137 Blindern
0318 Oslo, Norway

You can also get in touch with the Institution's Data Protection Officer (personvernombud) if you have any questions related to the use of your personal health data concerning health in the research project Roger Markgraf, email: personvernombud@uio.no

I CONSENT TO PARTICIPATING IN THE RESEARCH PROJECT AND THAT MY PERSONAL DATA CONCERNING HEALTH AND BIOLOGICAL MATERIAL CAN BE USED AS DESCRIBED ABOVE

	Yes	No
I agree to collect the following samples from me/my child:	☐	☐

☐ blood

☐ skin biopsy

☐ swabs (skin, mucosa)

☐ stool

I agree to store mine/my child's biological material in research biobank	☐	☐

City/Town and date

Participant's Signature

Participant's Name (in BLOCK LETTERS)

As parents/guardians of_____ *(Full name)*
we consent for him/her to participate in the research project

City/Town and date

Parent's/Guardian's Signature

Parent's/Guardian's Name (in BLOCK LETTERS)

City/Town and date

Parent's/Guardian's Signature

Parent's/Guardian's Name (in BLOCK LETTERS)

Consent on behalf of a representative

As next of kin for _____ *(Full name)*
I hereby consent to that, he/she can participate in the research project.

Place and date	Next of kin signature
	Next of kin name (IN BLOCK LETTERS)

I confirm that I have given information about the research project

Place and date	Signature
	Role in the research project