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 been diagnosed with a genetic condition. The study has two main aims: i) to help undiagnosed patients get a confirmed diagnosis, and ii) to better understand why some gene defects cause serious conditions, and to develop therapies for the underlying defect. For certain patients, we study how microbes and other environmental factors modify the disease presentation. The study will improve our general understanding of different genetic conditions, as well as strives to find common underlying links between different hereditary diseases. Therefore, your doctor at the hospital has selected you to participate in this study.

WHAT IS THE PROJECT ABOUT?

The project will collect and record personal information about (you/ your child/ the one you consent for). For some participants we will collect new information and material, and for others we will collect already collected information and material.

Depending on your disease, we will collect one or more biological samples from 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. Bone marrow, cerebrospinal fluid, tumor and 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).

If you / your child/ the one you consent for have previously participated in other research projects and donated biological samples to research biobanks or clinical storages we will access these samples to use them in our experiments. Your doctor who has access to your medical history will inform us in which research biobank or clinical storages your materials is stored. The collaborating clinicians will coordinate the access to your samples with managers of research biobanks/clinical storages. If these research biobanks/clinical storages require additional consents, we might contact you again to sign a new consent.

The samples will be transported and stored at “Rare diseases biobank” at the Norwegian Centre for Molecular Medicine (NCMM) at the University of Oslo (UiO). Your signed consent for participation in this study is retained in NCMM. Your biological material will be used in experiments in which we will create induced pluripotent stem cells (iPS) and immortalized cell lines derived from blood or skin cells from you/your child/the one you consent for. This way we will create an in vitro model to study monogenic diseases. 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.

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. If your /your child/ the one you consent for/ condition lacks a diagnosis, we might be able to find the underlying genetic cause of your condition. This enables better genetic counselling and, sometimes, helps to target the treatment.

Disadvantages are that the study results may not be clear and, in most cases, won't bring you any immediate benefit. If your disease is currently undiagnosed, we are often unable to rapidly find the cause of the condition, as it often takes years to verify a novel gene defect. 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 have received this information now and we encourage you to spend some time to revise this request and consult it with others. You can deliver your consent to project manager by mail or to a nurse during your scheduled hospital visit. 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 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. 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: Hans Christian Erichsen, Emma Haapaniemi, Janna Saarela, Sebastian Waszak, Monika Szymanska, 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 information genetic and 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. If this is to happen, you will receive new information from us. 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.
The code that connects you and your personal data concerning health will not be released.

WHAT WILL HAPPEN TO THE TESTS YOU HAVE TAKEN?

The tests taken from you/ your child/ the one you consent for will be stored in a Research Biobank connected to Research Project. Your biological samples, including blood samples, bone marrow, skin biopsies, stool samples and mucosal swabs will be stored at research biobank, called "Rare immune disease" 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 will be 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 might renew or apply for new permits for further research after the study period ends and contact you to ask for a new consent. If you do now wish to renew your consent your samples 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.
We will mainly collaborate with scientists from the Center for Hematology and Regenerative Medicine (HERM) at Karolinska Institute (Sweden), Hospital for Children and Adolescents (Finland) and the Institute for Molecular Medicine Finland (FIMM). 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.

GENETIC TESTING

- Genetic Counselling

  Genetic counselling will be given verbally and/or in written form by your attending clinician at the hospital. The attending clinician will inform you about the genetic origin of your disease and if you have a treatment-relevant mutation, he will discuss with you and help to find the best treatment options. In a case where we won't be able to provide the genetic diagnosis, your attending clinician will further discuss it with you and may refer you to clinical geneticist for further consultation.
- Incidental findings

  If you already have genetic data available, we will reanalyse the data to search for mutations in novel genes, and to identify additional genetic changes that affect disease progression. Your doctor who has access to your medical history will inform us in where is your genetic data stored. The collaborating clinicians will coordinate the access to your data with managers of sequencing facilities. If these facilities require additional consents, we might contact you again to sign new consent.

  In addition, we might study the genomes of single cells of your sample to better understand the disease pathology. For selected patients, we might study the genomes of microbiomes that reside in the skin, gut or mucosa. We also might try to correct your gene defect in the sample cells by using new gene editing technologies, to better understand the disease process and develop future therapies. Generally, we don't report these results back unless we believe they have direct relevance to your care.
- Possible re-identification

  Even though your name and personal identification number is removed, the genome sequencing is so unique that it can in theory never be regarded as completely anonymous.

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.

FOLLOW-UP PROJECT

As one of the disadvantages is that we won't be able to find causative mutation related to your illness in the present project, we might contact you in the future when new technologies arise, to follow-up on your case and find a diagnosis for you.

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

Based on an agreement with University of Oslo, Sikt Data Protection Services has assessed that the processing of personal data in this project is in accordance with data protection legislation. For more information you can contact Data Protection Services by email: (personverntjenester@sikt.no) or by telephone: +47 53 21 15 00.

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 consent to collecting the following samples from me/my child/ my family member:	☐	☐

☐ blood

☐ bone marrow

☐ cerebral spinal fluid (CSF)

☐ tumor biopsy

☐ skin biopsy

☐ swabs (skin, mucosa)

☐ stool

I consent to my/my child's/my family member's genetic data reanalysis	☐	☐
I consent to accessing my/my child's/my family member's biological material stored in existing biobank/clinical storage	☐	☐
I consent to accessing my/my child's/my family member's genetic data stored in existing sequencing centres	☐	☐
I consent to store mine/my child's/my family member's biological material in research biobank	☐	☐
I would like to receive feedback on the outcome of the study and the gene sequence abnormality and on the carrier status associated with the main condition	☐	☐

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