UiO : University of Oslo

Oslo University Hospital

GENETIC AND FUNCTIONAL STUDIES OF PROSPECTIVE MONOGENIC DISEASES

BRIEF SUMMARY OF THE PROJECT

You are invited to participate in this study because you 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 inherited diseases. Therefore, your doctor at the hospital has selected you to participate in this study.

WHAT IS THE STUDY ABOUT?

The project will collect and record personal information about you. Depending on your disease, we will collect one or more biological samples from you. The hospital personnel will take a blood sample and possibly stool and swab samples (swabs from skin and mucosa) during your 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 doctor at the hospital coordinates the sampling - please discuss with your doctor 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 samples will be transported and stored at “Rare disease biobank” at the Norwegian Centre for Molecular Medicine (NCMM) at the University of Oslo (UiO). Your biological material will be used in experiments to facilitate future research and for this reason, we will create induced pluripotent stem cells (iPS) and immortalized cell lines derived from your blood or skin cells. This way we will create an in vitro model to study monogenic diseases.
Yours’ and your parents signed consents for participation in this study are 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.

POSSIBLE BENEFITS AND DRAWBACKS OF TAKING PART

The study improves our understanding of genetic diseases, which will eventually help us to find new therapies. If your condition lacks a diagnosis, we might be able to find the underlying genetic cause of your condition. This enables better genetic counseling 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, in addition to your already scheduled tests.

WHAT WILL HAPPEN TO MY TEST RESULTS AND HEALTH INFORMATION?

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 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 specify 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 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. Doctors and researchers involved in this project: Hans Christian Erichsen, Emma Haapaniemi, Janna Saarela, Sebastian Waszak, Monika Szymanska will have access to this list.

Information about you will be anonymized. Your data and biological samples are very precious to us therefore we will keep the data and store your biological samples in a research biobank “Rare diseases”. After the project ends your biological samples will be anonymised and stored until 2035 due to requirements and conditions set by regional committees for medical and health research ethic. 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 will be destroyed after 2035.

PARTICIPATION IS VOLUNTARY

Participation in the project is voluntary. If you wish to take part, your parents will need to sign the declaration of consent. 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 analyzed 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 or researcher: Monika Szymanska, email: moniksz@ncmm.uio.no, tlf:46504016