UiO : University of Oslo

Oslo University Hospital

Genetic and functional studies of prospective monogenic diseases

WHAT IS THE STUDY 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 samples 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.

POSSIBLE BENEFITS AND DRAWBACKS 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.

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. 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 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 project will collect and record personal information about you. 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. 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 long-term storage, in a research biobank "Rare diseases biobank". 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/guardians will need to sign the declaration of consent. 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