Graduate Programme no.: FP10 | Graduate Programme name: Translational Medicine
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Theme: Genetic Medicine (link to home page) | Project title: Molecular Genetic and Cell Pathological Studies of Inborn Errors of Mitochondrial Metabolism
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Project description (approx. 300 words)

The research program at Research Unit for Molecular Medicine is centered on Molecular Genetic and Cell Pathological Studies of Inborn Errors of Mitochondrial Metabolism. We have for many years performed diagnostics and research on fatty acid oxidation defects as well as a few other mitochondrial metabolic disorders. The aim has always been to improve the knowledge about these diseases as precondition for more rational management of patients.

Today we know a lot, but we also know that the genotype in itself does not tell us very much about pathology and clinical expression of the various diseases. To improve this situation, we have, during the last few years, embarked on a number of large scale mitochondria proteomic studies, which aim at answering some of the questions related to the cellular pathology of these disorders.

In addition to an advanced protein mass spectrometer (Orbitrap; Thermo) and facilities for both prokaryotic and eukaryotic cell cultures the laboratory is equipped with instruments for DNA sequencing and qPCR as well as knowledge and equipments for traditional genetic and molecular biological analyses.

The PhD projects we want to ‘offer’ and which it is possible to apply for through an application within the framework of ‘Genetic Medicine, will all in some way be centered on the elucidation of the patophysiology of mitochondrial metabolic disorders. The projects can primary use in vitro studies and/or investigation of model cells and animals as well as patients cells, of which we have collected a large number and permission to study them deidentified. The projects can also be technology driven, as f.ex. assessment or development and use of methods for detection and determination of modified mitochondrial proteins.