

Affiliation: Masaryk University, Faculty of Medicine
Study program: PhD program Biomedical Sciences
Specialization: Biochemistry and Molecular Biology
Workplace: Department of Clinical Immunology and Allergology
Institution shared with St. Anne's Faculty Hospital, Faculty of Medicine, MU
Form of study: Full-time

Supervisor: Prof. MUDr. Tomáš Freiberger, Ph.D. (tomas.freiberger@cktch.cz)

Title: Screening of genetically determined hypercholesterolemia in newborns

Brief annotation:

Cardiovascular diseases belong to the deadliest diseases affecting people worldwide. Familial hypercholesterolemia (FH) is a genetic disorder that affects 1 out of 250 – 300 individuals and significantly increases the amount of LDL-cholesterol in blood, therefore increasing the risk of coronary heart disease development. Early diagnosis and treatment can significantly reduce mortality especially in the group of young adults, but also in elderly population. Sequence variants in LDLR, APOB, PCSK9 (and others) are associated with the disease. The phenotype of this disease can have an important polygenic component determined by the co-occurrence of the polymorphisms linked to the increased levels of LDL-cholesterol and/or increased risk of cardiovascular diseases.

The work focuses on the evaluation of biochemical and molecular biological parameters of FH in newborns of the Czech Republic population with regard to the possible introduction of population screening of this disease. Attention will be primarily focused on the role of molecular genetic diagnostics in screening and on the analysis of monogenic and polygenic risk of atherosclerosis development. Modern methods of massive parallel sequencing and bioinformatic data analysis will be used.

Funding: The proposed project has a grant support. Student will be employed on 0.5 contract (on top of the scholarship) that will be funded from the grant resources or institutional support.

Requirements: Great level of English (both written and spoken). We offer friendly environment and prefer team players.

Requirements to successfully obtain a PhD degree in the Biomedical Sciences PhD program:

- Have at least one first author publication in an international research journal with an impact factor above the median IF in the field or have two first author publications with the IF in Q3. Importantly, the affiliation to the Faculty of Medicine, Masaryk University must be listed.
- Gain a set of minimum credits (240 ECTS in 4-year study period), pass 4 faculty-wide courses + an English language course and 4 field-specific courses.
- Pass the doctoral state exam.
- Take an active part in teaching.
- Participate in the annual PhD conference.
- Present research outcomes at least once at an international conference (poster, presentation).

Masaryk University, Faculty of Medicine

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Please quote the Reference Number in your reply.

- Spend at least one month abroad on an internship.

Information about the supervisor:

Prof. Freiburger is the author or co-author of more than 100 publications in journals with impact factor, with the citation index >2900 (WoS) and h-index 23. He is the author of 1 chapter in scripts, 4 chapters in textbooks and 4 chapters in monographs. He was the principal investigator of 6 grants and co-investigator of 16 grants of the Ministry of Health or the Ministry of Education. He is involved in a number of international projects. He led 5 students to the successful defense, and more students are heading towards the defense soon.