



MEDIZINISCHE
UNIVERSITÄT
INNSBRUCK

PhD Position in Lipoprotein Genetics

Division of Genetic Epidemiology
Medical University of Innsbruck

Duration: 3 Years, immediately available.

Description of the project: Lipoprotein(a) [Lp(a)] is the strongest genetic risk factor for cardiovascular disease. Lp(a) concentrations are controlled predominantly by one gene locus (*LPA*) which, however, presents a highly complex structure. Thus despite its clinical relevance, no native cell model for Lp(a) research exists and many aspects of Lp(a) genetics are still poorly understood.

Our group recently described a highly frequent genetic variant in the KIV-2 region (“4925G>A”), which reduces Lp(a) by up to 70%. In-vitro reporter data suggest a modification of splicing behavior but the exact mechanism is not yet understood. This project aims to investigate the effects of KIV-2 4925G>A in mRNA from liver biopsies with different Lp(a) phenotypes using targeted (qPCR, ddPCR, Northern blotting) and untargeted (RNA-Seq) approaches, characterize allelic expression patterns of *LPA* and investigate the use of organoid cultures as cellular model for Lp(a)-research.

The Division of Genetic Epidemiology Innsbruck: We are a highly interdisciplinary institute with long-standing experience in Lp(a) research, genetic epidemiology and NGS bioinformatics and a large network of scientific collaborations. We are equipped with state-of-the-art genetic analysis technologies and all currently available Lp(a) analysis techniques (ELISA and Western Blotting, PFGE genotyping, ultra-deep sequencing, KIV-2 qPCR and ddPCR, Nanopore sequencing) are available in-house. Our bioinformatics group has extensive experience in NGS data analysis and we carry on fruitful collaborations with cell biology and RNomics groups and with clinical partners.

Candidate profile: We are searching for a hard-working PhD student with a background of molecular biology, molecular medicine or related disciplines and a genuine interest in genetic research and computational biology. We offer a very productive and collaborative environment in an interdisciplinary institute with a broad expertise ranging from genetic wet-lab work to complex bioinformatics and statistics. The successful candidate should have previous experience in establishing novel methods as well as in standard molecular biology techniques (nucleic acid isolation, electrophoresis, PCR/qPCR/RT-PCR, mammalian cell culture). Previous experiences with next-generation sequencing data analysis, Linux operating systems and/or R programming are a strong asset but not strictly required. However, a genuine interest in learning NGS bioinformatics is essential. Successful applicants will be paid according to the rates of the Austrian Science Fund (FWF) and will be part of the PhD school “Genetics, Epigenetics and Genomics” of the Medical University of Innsbruck.

Contact: For questions or applications please contact Stefan Coassin, PhD at: Stefan.Coassin@i-med.ac.at or 0043 512 9003 70576

References

1. Coassin, S. *et al.* A novel but frequent variant in *LPA* KIV-2 is associated with a pronounced Lp(a) and cardiovascular risk reduction. *Eur Heart J* **38**, 1823–1831 (2017).
2. Kronenberg, F. & Utermann, G. Lipoprotein(a): resurrected by genetics. *J Intern Med* **273**, 6–30 (2013).