PhD Position in Hereditary Cancer Genetics

We search for a motivated PhD student to join a 3-year FWF-funded project that is part of the EUROpean NETwork of the NeuroFibromatoses-schwannomatoses and related disorders (EURONET-NF), a European Joint Program-Rare Diseases (EJP-RD) project, which aims at improving the molecular diagnostics of these disorders. The starting date is planned for summer/early fall 2023.

The applicant will work in the research laboratory of the Hereditary Cancer Genetics section of the Institute of Human Genetics of the MUI (https://www.i-med.ac.at/tumorgenetik-erbliche-tumoren/research.html). This lab is closely connected to the diagnostics laboratory of this section. In close collaboration with the other partners of this EJP-RD project the applicant will develop and implement new diagnostic tools that are mainly based on different DNA and RNA next generation sequencing (Illumina, PacBio) techniques with the aim to uncover genetic and genomic alterations that escape the currently applied sequencing techniques. Furthermore, it is planned to implement the novel tools into our routine NF diagnostics repertoire. A specific part of the project focusses on the role of a rare childhood cancer predisposition syndrome characterized by a constitutional mismatch repair deficiency (CMMRD) as an alternative diagnosis to neurofibromatosis type 1 (NF1). The applicant will collect with the clinical partners of this project suspected sporadic NF1 children without identifiable mutation in the relevant NF1 gene that fulfill clinical pre-selection criteria for CMMRD testing. Using a novel NGS-based CMMRD screening assay (to be implemented by the applicant our laboratory) and subsequently genetic testing these patients will be analyzed for CMMRD and by this the sensitivity, specificity, positive and negative predictive value of these criteria will be evaluated.

To successfully perform this project the candidate PhD student should have:

- a masters degree in a natural science discipline or medicine and a keen interest in hereditary cancer genetics and genetic diagnostics
- a solid molecular genetics background and at least basic skills in next generation sequencing and bioinformatics
- interest and capability of (self-)organization and diagnostic accuracy

We offer:

- an interesting project with immediate clinical implications
- integration into a network of six European (Spain, France, Netherlands, Belgium, Poland and Austria) partner laboratories working together in a EJP-RD project, with the possibility to attend common meetings and short-term research exchanges
- payment according to FWF salary for PhD candidates for 3 years

Applications should be sent along with a letter of motivation, information regarding the desired requirements and a CV to Ao.Univ.-Prof. Dr. Katharina Wimmer (katharina.wimmer@i-med.ac.at).