

PhD POSITION

Hereditary Cancer Genetics group

<https://www.i-med.ac.at/tumorgenetik-erbliche-tumoren/research.html>.

THE PROJECT

Improving the diagnostics of Neurofibromatoses-schwannomatoses and related disorders

The 3-year Austrian Science fund (FWF)-funded project is embedded in 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 genetic tumor predisposition syndromes.

In close collaboration with the other partners of this EJP-RD project we 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 in the relevant genes that escape the currently applied techniques.

A specific work package 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). A novel NGS-based CMMRD screening assay (to be implemented by the applicant in our laboratory) and subsequently genetic testing will be used to test for CMMRD in a large number of suspected sporadic NF1 children without identifiable mutation in the relevant NF1 gene that fulfill clinical pre-selection criteria for CMMRD testing.

YOUR PROFILE

- 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

To 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).