PhD Position in Translational Neurogenetics

Location: Department of Neurology, TUM School of Medicine and Health with TUM Graduate Centers & Medical Genetics Center (MGZ)

Supervision: PD Dr. I. Cordts, Neurology, TUM & Dr. A. Benet-Pagès, Bioinformatics, MGZ

Start: As soon as possible

We invite applications for a **PhD position** in an interdisciplinary and translational research project at the intersection of **neurogenetics, bioinformatics, and clinical neuroscience**. The position is jointly embedded at the Department of Neurology (TUM) and the bioinformatics team at MGZ, combining clinical research on rare neurological disorders with expertise in long-read sequencing technologies. You will work in a motivated, skilled, and supportive team within a highly interdisciplinary environment, benefiting from close supervision, access to cutting-edge sequencing tools, and integration into a translational neuroscience network. We offer professional development opportunities including conference attendance and targeted training, an innovative workplace equipped with state-of-the-art technology, and a centrally located office in beautiful Munich.

About the Project

The PhD project consists of two complementary research approaches:

1. **Targeted long-read sequencing of the GAA repeat expansion in Friedreich's Ataxia**
   Using state-of-the-art PacBio sequencing (PureTarget), this subproject aims to characterize repeat structures (length, interruptions, motif patterns, and methylation) in a multicenter cohort of patients with Friedreich's Ataxia. These genetic features will be correlated with clinical phenotype, biomarker data, and response to omaveloxolone treatment.
2. **Long-read genome sequencing in rare neurological disorders**
   This subproject focuses on a unique cohort of patients with suspected genetic motor neuron diseases in whom no genetic cause has been identified through conventional diagnostics. The aim is to use genome-wide long-read sequencing (PacBio HiFi) to uncover complex genetic variants, e.g., structural variants and novel repeat expansions, that could underlie disease pathogenesis.

Both projects involve applying, refining and adapting innovative long-read analysis pipelines, with strong emphasis on integrating and analyzing genetic data with clinical and biomarker-based phenotyping.

Your Profile

- You hold a Master's degree in **Bioinformatics, Neuroengineering, Medical Informatics**, Computational Biology, or a related life science field.
- You have a **strong background in data science**, ideally with experience working on **genetic datasets and bioinformatics data analysis pipelines**. You are motivated to acquire and refine advanced methodological skills in genetic data analysis, with strong interest in clinical translation.
- You have good skills in **programming languages** (e.g., Java, Python, Bash, R) and have experience with workflow languages (e.g., Nextflow, CWL), version control systems (e.g., Git), containerization tools like Docker, and project management tools (e.g., Jira).
- You have proficiency in **statistical methods**, preferably including work with longitudinal modeling in clinical or human cohort studies (e.g., linear mixed models).
- You are confident **working independently** and enjoy solving challenging methodological problems.
- You communicate effectively, work with precision, and are open to **interdisciplinary collaboration** – particularly between neurology and genetics in physically separate research environments.
- Excellent proficiency in **written and spoken English** is required.
- Own funding/scholarship not required but appreciated.

To apply, please send your CV, a brief motivation letter, and academic transcripts to isabell.cordts@tum.de and anna.benet-pages@mgz-muenchen.de.

Additional information: The position is initially for one year, extendable until Ph.D. completion. Salary follows the German public service scale (TV-L E13). Candidates with severe disabilities are given priority if equally qualified. By applying, you consent to the processing of your personal data (see TUM privacy policy, Art. 13 GDPR: http://go.tum.de/554159). Submission confirms your acceptance of this notice. For details on TUM doctoral programs, see: https://www.gs.tum.de/gs/weg-zur-promotion/.