



Postdoc in Computational Genomics

A postdoctoral researcher position in Genomics, Bioinformatics, and Statistical Genetics is available in the Computational Genomics group at the Institute of Medical Genetics and Applied Genomics. We are looking for an early experienced, highly motivated young researcher with a strong enthusiasm to conduct cutting-edge research at the interface of computational biology and clinical genomics. The position is part of the EU-funded research project "Solve-RD – Solving the Unsolved Rare Diseases" (www.solve-rd.eu), in which we apply the latest sequencing technologies such as whole-genome sequencing, long-read RNA-seq and single cell sequencing to investigate 19,000 unsolved cases of rare genetic diseases. The successful candidate will be part of a large European network of researchers working on computational genomics, rare genetic diseases and personalized medicine. The working language in the laboratory is English.

Requirements: Applicants must hold a PhD or equivalent in computational biology, bioinformatics or a mathematical discipline, have a strong interest in the genetics of human disease, and a substantial publication record in high-quality international journals focused on the analysis of genomic or transcriptomic data. Proficiency in programming in R, Python, Perl, C/C++, or Java is highly desired.

The Project: Solve-RD aims to use novel omics technologies, improved computational methods and standardized analysis of large cohorts of unsolved rare disease patients in order to improve the diagnostic rates, to identify novel disease genes and variants and to investigate the contribution of splicing, regulatory, mosaic or structural variants. As part of the Solve-RD study we will obtain a combination of exome-seq, whole-genome-seq, short- and long-read RNA-seq data for up to 19,000 unsolved cases of rare genetic diseases. The postdoc will develop innovative computational and statistical methods for interpretation of large-scale omics data to identify novel disease genes and variants for rare genetic diseases. Specifically the postdoc will work on:

i) Development and application of methods for the analysis and combined interpretation of whole-genome and long-read transcriptome sequencing data for rare disease diagnostics.

ii) Automating interpretation of genetic variant pathogenicity using novel artificial intelligence concepts.

iii) Development of methods for interpretation of intronic/splicing and regulatory genetic variants (including point mutations and structural variants).

iv) Development of statistical tests for the identification of novel disease genes in large cohorts of unsolved cases.

The Institute: The University Hospital Tübingen is a highly specialized clinical center with more than 420,000 patients treated per year. The Institute of Medical Genetics and Applied Genomics is an integral part of patient care and research at the University Hospital and the Medical Faculty of the University of Tübingen, and currently employs more than 150 staff members. The Institute coordinates the Center for Rare Diseases (ZSE Tübingen) and, as member of the Center of Personalized Medicine (ZPM) and the Molecular Tumor Board we are strongly interacting with all clinical disciplines. The Institute is also part of several European Reference Networks (ERNs) for rare diseases and, since 2017, is one of the four German competence centers for Next Generation Sequencing (NGS Competence Center Tübingen, NCCT) funded by the DFG. Our facilities are equipped with state of the art technologies for NGS and single cell sequencing as well as large-scale compute clusters for NGS analysis. With a highly motivated group of bioinformaticians, biologists and clinicians the institute has established the broad application of whole genome sequencing in clinical diagnostics. For further information about our groups, please visit our websites: https://www.medgen-tuebingen.de/de/forschung.html.

What we offer: The initial appointment will be for two years, with possible extension. We offer remuneration in accordance with TV-L (collective wage agreement for the Public Service of the German Federal States) in addition to all the customary benefits granted to employees working in Public Services. Severely handicapped persons with equal qualifications are given preferential consideration. The University of Tübingen is anxious to increase its quota of female scientific staff, and therefore emphatically requests women to apply for this position. The Administration of the University Hospital is responsible for all employment matters. Personnel appointments will be made pursuant to the fundamental stipulations of the legal statutes for universities in Germany. Interview expenses are not covered.

To be considered, please send Cover Letter, Curriculum Vitae, Unofficial Transcript, and List of References (including contact information of three professional references) to Prof. Stephan Ossowski (<u>medgen.bewerbungen@med.uni-tuebingen.de</u>). Applications will be considered until June 1st, 2019, or until the position has been filled. Candidates finishing their PhD before 1st of September 2019 will also be considered. Please send your application and details on your qualification by mail or email to:

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