



The **Boztug Lab at LBI-RUD**, the Ludwig Boltzmann Institute for Rare and Undiagnosed Diseases in Vienna, seeks to fill the open full-time position of a

## MEDICAL GENETICIST

The focus of our group is the identification of rare monogenic disorders of the immune system and hematopoiesis to explain pathophysiological manifestations in humans (Ozen A, Comrie WA, Ardy RC et al *NEJM* 2017; Salzer E et al *Nat Immunol* 2016; Dobbs K, Dominguez Conde C, Zhang SY, Parolini S et al *NEJM* 2015). In the context of this work, we apply a combination of state-of-the-art genetic and genomic technologies including next generation sequencing, and a wide range of cellular and molecular techniques to elucidate novel pathways important for immune system homeostasis.

We work in close alliance with the CeRUD Vienna Center for Rare and Undiagnosed Diseases ([www.meduniwien.ac.at/cerud](http://www.meduniwien.ac.at/cerud)) and the Undiagnosed Diseases Program and the Undiagnosed Diseases Network International of the National Institutes of Health (<http://www.udninternational.org>).

To complement our team, we are looking for a highly motivated and well-organized **Medical Geneticist** with a strong interest in research of rare and undiagnosed Mendelian diseases.

As our new team member you will be fully integrated into a team of experienced technicians and genetics/NGS experts, and will work in close collaboration with bioinformaticians, as well as clinicians and researchers from local and international hospitals and research centers. We have established a streamlined workflow for NGS-based genetic analyses that requires continuous maintenance and optimization as well as development and implementation of novel assays, tools and infrastructure. This includes e.g. the migration of our current patient/genetic database to a new IT platform, the systematic and safe exchange of data with international research institutions, and the generation and implementation of standardized procedures and workflows. For the latter, you have ideally already gained experience and/or thorough theoretical knowledge of the practical and legal requirements of a laboratory accreditation process.

### Your tasks will include:

- setup of Standard Operating Procedures for genetic analysis workflows
- oversight of operation and maintenance of current lab material and equipment, incl. evaluation and ordering of additional equipment as needed
- independent research, testing and implementation of novel NGS technologies to improve and advance our current pipeline (e.g. whole genome sequencing, Novaseq technology, epigenome sequencing)
- management of our patient/genetic databases, including migration to a new IT platform
- development and implementation of a standardized way of sharing clinical information using Human Phenotype Ontology (HPO) terminology
- management of data sharing activities using the Matchmaker Exchange platforms
- support for the analysis and interpretation of genetic data

### Your profile:

- you hold an academic degree in medicine or life sciences – ideally you are an MD with research experience and/or experience in diagnostics/laboratory medicine (board certification as human geneticist would be an asset)



- alternatively, you hold an advanced (MSc or PhD) life science degree with research experience in an international environment and/or pharmaceutical industry
- you have substantial knowledge and experience in a regulatory environment (GCP, GCLP, related to EMA standards) - required
- you have very good written and oral communication skills in English and German - required
- you have experience in immunology and/or genetics - advantageous
- you know the Austrian Genetic Engineering Act (Gentechnikgesetz) and have experience in biobanking – advantageous
- you communicate and collaborate easily with different kinds of people from diverse cultural background
- you work with highest accuracy and reliability due to your high level of self-motivation and self-organization, as well as your perfect time management skills
- you are a proactive person and eager to actively initiate and direct projects

**Your benefits from working with us:**

- a challenging position in a meaningful, inspiring, and international setting
- the possibility to take over responsibility for your own projects
- an excellent work climate and the possibility to join our social (cultural or sports) activities
- a competitive salary according to FWF personnel costs (<http://fwf.ac.at/en/research-funding/personnel-costs>), starting from 2500€ (monthly gross), taking into account your specific background and qualification

**The Institute:**

The Ludwig Boltzmann Institute for Rare and Undiagnosed Diseases (LBI-RUD) is directed by Kaan Boztug, a world-leading expert in primary immunodeficiency research, and is situated in a beautiful, bright, and perfectly designed building with outstanding design and state-of-the-art laboratories and technology platforms. The institute is strategically located next to one of the world's largest university hospitals in the heart of the city and is deeply integrated with its renowned partner institutions, namely the CeMM Research Center for Molecular Medicine of the Austrian Academy of Sciences, the Medical University of Vienna, and the Children's Cancer Research Institute of the St. Anna Children's Hospital. It is therefore perfectly situated to benefit from strong translational research long established by leading-edge research groups.

For more information visit: [www.rare-diseases.at](http://www.rare-diseases.at)

**Application Details:**

Please apply with a cover letter describing your career goals and explaining why you are the ideal candidate for this position, your detailed CV and contact details of 2-3 referees. Send your application in one single PDF file to: [application@rud.lbg.ac.at](mailto:application@rud.lbg.ac.at) (using the reference code **#MedGen17**).

Initial application deadline is 19<sup>th</sup> of November 2017, but applications will continuously be reviewed and the call will remain open until the position is filled.

***Join Austria's new research institute for rare and undiagnosed diseases research!***