



BIOMEDICAL CENTER MUNICH BIOMEDIZINISCHES CENTRUM MÜNCHEN



# Doctoral research position (m/f/x) Epigenetics of rare diseases and sex as biological variable Biomedical Center Munich (BMC) LMU Munich Prof. Dr. M. Felicia Basilicata (she/her)

## Background of the lab and project

Currently at University Medical Center of the Johannes Gutenberg University in Mainz, we are going to join the LMU faculty and the Department of Physiological Chemistry in September 2025. Collectively 58 million people worldwide are diagnosed with rare developmental disorders (DD) leading to intellectual disability and difficulty to thrive. While diagnoses of the underlying causative genetic mutations are massively improving thanks to routine next generation sequencing in the clinic, the question most frequently asked by affected families - what is the treatment? - remains largely unanswerable. In the lab, we study how biological sex influences embryonic development and how extrinsic and cellular environment contribute to the phenotypic presentation of selected neurodevelopmental disorders. The most enriched class of causative mutations in DD are genes belonging to the chromatin machinery, as for the Basilicata-Akhtar syndrome. Despite their diverse nature, mutations in these genes converge like light in a prism and refract into neuronal susceptibility and immune dysfunction generating a broad spectra of variable presentation at systemic levels. A striking feature of DD is their pronounced sex bias, with males typically affected at least twice as frequently as females. This pattern exists alongside females generally exhibiting heightened immune system sensitivity and greater susceptibility to autoimmune conditions. We examine how event occurring in the nucleus influence organelle communication networks (plasma membrane, mitochondria, ER, lysosomes) and ultimately contribute to cellular homeostasis. Our research investigates both how impaired organelle communication drives disease manifestation and how sex chromosomes and hormonal factors shape the action of these molecular determinants. By identifying common feedback mechanisms, we aim to develop sex-specific therapeutic approaches for these rare developmental disorders.

Webpage: https://www.physiolchemie.abi.med.uni-muenchen.de/research/fbasilicata/index.html

#### What can you learn with us:

Research focus:

- How do sex chromosomes and hormonal factors influence chromatin organization during embryonic development?
- How does organelle crosstalk impinge on cellular resilience and plasticity during DD disease onset?

#### Methodological expertise in

- Advanced CRISPR/Cas9 genome editing for knockout and Knock-in for molecular degrader based protein interference
- Stem cell culture of disease models in murine and human induced pluripotent stem cell culture
- Differentiation paradigms to multiple neuronal lineages using 2D and organoid based protocols
- High-resolution microscopy and functional assays (Flow cytometry, Confocal microscopy, Light sheet)
- Single molecule analysis
- Multiomics data generation and analysis, including CRISPR screening, RNA-seq, Cut&Tag, proteomics, metabolomics.

Who you are

For our Molecular Gene Regulation Research Group, we are looking for a team player PhD student who is experienced in the lab, enthusiastic about science and on tackling questions on sex chromosome biology, neurodevelopmental processes, cell metabolism and epigenomics and has a strong interest in exploring scientific questions in an interdisciplinary way. Computational skills are an asset but not compulsory.

## What do we offer

- Young, supportive, collaborative, interdisciplinary and international environment at BMC/ Martinsried Campus
- English as main working language
- Fully funded 3-year PhD position (with possibility of extension, TV-L pay scale with full coverage of social security, health insurance)
- Access to state-of-the-art core facilities (Bioimaging, Biophysics, Flow cytometry, Genomics, Proteomics and Bioinformatics)
- Mentoring on personal and professional development for both academic and non-academic careers

### How to apply

If you are interested in joining our group please submit as one PDF the following documents (Max 5MB - *Surname\_GeneRegulation*):

- CV
- Motivation letter
- Academic Transcripts and Degree certificates
- Contact email of two references

Via email to: mfbasilicata@uni-mainz.de

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