Master thesis – Molecular mechanisms behind the neurodevelopmental disorder PURA syndrome

Location:
Helmholtz Zentrum München, Neuherberg.

University supervisor:
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A heterozygous mutation in the *PURA* gene can cause the neurodevelopmental disorder PURA Syndrome. First described in 2014, today there are over 620 diagnosed cases worldwide, with numbers constantly increasing. The PURA protein is expressed in every human tissue. Apart from already published detailed structural data, very little is known about the underlying mechanisms leading to the severe symptoms including neurodevelopmental delay, intellectual disability, hypotonia, and epilepsy.

The Niessing group at the Institute of Structural Biology at Helmholtz Munich studies the structure and function of the PURA protein. Our overall goal is to understand PURA's cellular functions and the molecular consequences of a PURA deficiency in PURA syndrome patients. Therefore, we use genetically modified immortalized cell lines and patient-specific induced pluripotent stem cells (iPSCs). High-throughput techniques in these cells already gave us first ideas about PURA's interactome and in which pathways PURA may play a role. These data need further validation to identify specific molecular mechanisms that cause the severe disorder. Ultimately, we want to provide information that can lead to the development of treatment strategies for PURA syndrome patients.

Interested? Join us on this crucial and challenging task for your master thesis or contact us if you want to be part of this as an intern.

What to expect?

- Cell culture (human neural stem cells, iPSCs, fibroblasts)
- Differentiating into neural progenitor cells
- Molecular cloning
- Real-time qPCR and western blotting
- Immunofluorescence experiments
- Working with different CRISPR/Cas9 systems
- Proteome interaction studies
- Interpreting and evaluating results
- Independent lab work

Contact: carolin.ketteler(at)helmholtz-munich.de (direct supervisor); For further information on the PURA Syndrome visit: https://www.purasyndrome.org/; And to check out our lab: https://www.helmholtz-munich.de/en/stb/research-groups/niessing-lab

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