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HelmholtzZentrum münchen
Deutsches Forschungszentrum für Gesundheit und Umwelt

PURA Syndrome: Mutations in neuronal nucleic acid binding protein Pur-alpha lead to neurodevelopmental disorders

PURA Syndrome is a rare disease caused by a mutation of the gene PURA encoding the protein Pur-alpha (Pura) expressed in every human tissue. This protein is involved in major cellular processes including transcription, mRNA transport and translation as well as neuronal development. Defects in the gene cause neurodevelopmental delays, leaving the patient with a lack of speech, difficulties with motion and even epileptic seizures in some more severe cases. First diagnosed in 2014, today over 200 patients worldwide are known. Although knowledge on Pur-alpha function is increasing over the last 20 years since the proteins discovery, there is still a lot to understand about the disease and its role in the human body.

At the Ulm University, the Niessing group is currently building up. However, our second lab at the Institute of Structural Biology of the Helmholtz Zentrum München is fully functional (head: D. Niessing). There we study the function of Pur-alpha in different human cell types. This includes studies in genetically modified HeLa cells and patient specific induced pluripotent stem cells (iPSCs). Using these cells we want to identify protein and nucleic acid interaction partners of Pur-alpha.

Our goal is to generate a more adequate disease model of the PURA syndrome and by this potentially identify mechanisms that cause the severe disorders previously described. 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 bachelor or master thesis or contact us if you want to be part of this as an intern.

Main tasks in our lab are:

- Cell culture (HeLa-cells, iPSCs, patient fibroblasts)
- Molecular cloning
- Real-time qPCR and western blotting
- Overexpression Experiments using PiggyBac system
- Working with different CRISPR/Cas9 systems
- Whole transcriptome & proteome interaction studies
- Structural biology, protein expression and purification
- Interpreting and evaluating results
- Independent lab work

Contact: lena.molitor@helmholtz-muenchen.de (direct supervisor) or dierk.niessing@ulm-university.de

For further information on the PURA syndrome visit: https://www.purasyndrome.org/

And to check out our lab: https://www.helmholtz-muenchen.de/stb/research/groups/research-group-niessing/research/index.html