Website address:
https://www.gesundheitsindustrie-bw.de/en/article/press-release/patient-association-raises-10000-euros-work-pura-syndrome-donation-research-rare-neuronal-developmental-disorder-ulm-university

Patient association raises 10,000 euros for work on PURA syndrome - Donation for research into rare neuronal developmental disorder at Ulm University

PURA syndrome is a rare developmental disorder of the brain for which there is currently no cure. The patient association PURA Syndrome Germany was founded last year. Since then, the affected families have been collecting donations with great dedication – and have decided to donate the majority of this money to research into the syndrome at Ulm University. Recently, a family from the patient association travelled to Ulm to present the head of the Institute of Pharmaceutical Biotechnology, Professor Dierk Niessing, with the symbolic cheque for 10,000 euros.

It has been exactly ten years since PURA syndrome, a rare neuronal genetic defect that causes severe developmental disorders from birth, was first described in medical literature. The relatives of patients celebrate this "birthday" as a positive event: many families have since received a definitive diagnosis for the first time, where uncertainty previously prevailed. Children with PURA syndrome suffer from learning disabilities and mental developmental delays, as well as seizures, reduced muscle tone, eating difficulties and breathing problems. The PURA Syndrome Germany patient association, which was founded a year ago, marked the tenth anniversary of the description of the genetic defect by making a generous donation to Ulm University for research into PURA syndrome: A family affected by the syndrome recently handed over a cheque for 10,000 euros to Professor Dierk Niessing, head of the Institute of Pharmaceutical Biotechnology, who has been researching the mechanisms of the syndrome for many years. The money was collected by the families united in the patient association with a great deal of commitment in their environment.

The family that presented Niessing with the symbolic cheque has a young daughter with PURA syndrome and travelled from Munich for the occasion. "We hope that our donation has made a valuable contribution to research into PURA syndrome, and we would be delighted to support the researchers again next year," says Lisa Obst from PURA Syndrome Germany. "Us researchers are very enthusiastic about the personal commitment of those affected and their determination to support research at our university," says Professor Niessing. His laboratory is the leading scientific institution in Germany for PURA syndrome research and is mainly conducting stem cell research to understand which signalling pathways in the cells are disrupted by the genetic defect. This could lead to new approaches for therapies in the future, to better treat the symptoms.

In Germany, only a little more than 35 patients with PURA syndrome have been genetically identified so far, and about 700 affected individuals are known worldwide. Dierk Niessing has helped to bring together German families. The scientist and his research group have a very special and close relationship with those affected: They regularly give lay presentations, advise families and support those affected in organising joint events. "We are very grateful for this impressive support from the association and hope that our research will contribute to a better understanding of the disease mechanisms and ultimately to insights that can be used for therapeutic purposes," says Niessing.

About PURA Syndrome Germany

The non-profit organisation PURA Syndrome Deutschland e.V. represents the interests of PURA patients and their families or caregivers. It supports families by providing a network and facilitating the exchange of information, and shares background information and the latest research findings. In addition to regular get-togethers, the association organises an annual meeting for all families in German-speaking countries and contacts, as well as contributions to the annual international PURA Syndrome Conference.

Press release

15-Nov-2024
Source: Ulm University

Further information

- PURA Syndrome Deutschland e.V.
- Ulm University