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When a spontaneous mutation turns off autophagy in the brain

Tübingen molecular biologist and hope tree e. V. join forces in research into the neurodegenerative disease BPAN

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Prof. Dr. Tassula Proikas-Cezanne (4th from left) with her research group in the new auditorium at the University of Tübingen.

Deciphering the causes of rare, neurodegenerative diseases is a lengthy process that is associated with a lot of hope and suffering for those affected. In a new research project, Professor Tassula Proikas-Cezanne from the University of Tübingen will in future investigate the molecular causes of the rare disease "BPAN (beta-propeller-associated neurodegeneration)". In doing so, she works with doctors in Tübingen, London and Copenhagen as well as the patient organizations "Hoffnungsbaum eV" in Germany and "BPAN Warriors" in the USA to investigate the cellular causes and potential therapeutic options. The project is funded by the German Research Foundation (DFG) as part of the SFB 1177 special research area in Frankfurt.

BPAN (beta propeller-associated neurodegeneration) is a very rare, congenital neurodegenerative disease caused by a mutation in the WDR45 / WIPI4 gene on the X chromosome. From birth, patients are characterized by a severe developmental disorder, which is associated with epilepsy, mental and physical disabilities and pronounced impairments in language acquisition. Painful muscle cramps and symptoms of Parkinson's disease develop later. The life expectancy of patients has been significantly reduced, so far there is no therapy. BPAN belongs to a group of more than ten diseases called NBIA (neurodegeneration with iron deposition in the brain). Around 20 patients with BPAN are known in Germany.

The autophagy researcher Professor Tassula Proikas-Cezanne, molecular biologist at the University of Tübingen and discoverer of the WDR45 / WIPI4 gene, will be involved in the Collaborative Research Center SFB 1177 ("Molecular and Functional Characterization of Selective Autophagy") over the next four years. Spokesman: Prof Dr. Ivan Dikic, Goethe University, Frankfurt aM, http://www.sfb1177.de) study the process of autophagy in brain cells of BPAN patients. Proikas-Cezanne recognized a few years ago that the WDR45 / WIPI4 gene has an important functional importance in the process of autophagy. Autophagy is a cellular degradation and renewal system that is disrupted in the nerve cells of patients with many neurodegenerative diseases, such as M. Alzheimer and M. Parkinson. Due to the mutation in the WDR45 / WIPI4 gene, autophagy in BPAN is severely impaired, however the causative molecular mechanisms are unknown. The scientist wants to elucidate these mechanisms, and she will work closely with the neurologist Professor Matthis Synofzik from the University Hospital Tübingen and other colleagues in London and Copenhagen.

The researchers are supported by "Hoffnungsbaum eV", the German patient organization for the promotion of research and treatment of NBIA. The association has initiated, co-financed or supported research projects several times over the past 15 years and is also very well networked internationally. “In this strategic partnership, we can act in particular as a contact and information interface between the researchers and our patient network. In addition, there is of course the knowledge of the families concerned about the BPAN disease of their children, ”explains Angelika Klucken, chairwoman and co-founder of“ Hoffnungsbaum eV ”.

"I am happy and excited that we have found such a renowned and committed colleague in the fight against BPAN in Professor Proikas-Cezanne," says Markus Nielbock, father of seven-year-old Emilia, who has BPAN, and second chairman of "Hoffnungsbaum eV" , "We will do everything we can to support you and Professor Synofzik."

For Tassula Proikas-Cezanne, this research project also stands out from other projects in her research group. "The fact that a spontaneous mutation of a gene that I discovered years ago can trigger such a serious disease obliges me to concentrate our research interests here," she says. "The personal contact with children suffering from BPAN also motivates me to do basic research to help alleviate their suffering."

Tassula Proikas-Cezanne / Antje Karbe

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