Website address:
https://www.gesundheitsindustrie-bw.de/en/article/press-release/novel-therapy-phelan-mcdermid-syndrome-drug-development-be-funded-17-million-euros

Novel therapy for Phelan-McDermid syndrome Drug development to be funded with up to 1.7 million euros

An international team led by Professor Tobias M. Böckers from Ulm University Medical Centre has been awarded up to 1.7 million euros to develop a novel therapy for a rare, syndromic form of autism. The research project focuses on a gene whose loss of function can manifest itself in severe impairments in language, behaviour and everyday functions of those affected. The project is funded by the non-profit organisation SPARK NS, which aims to promote efficient drug development in the field of autism and Parkinson's disease.

A translational research project to develop a novel therapy for Phelan-McDermid syndrome (PMS) is being launched at Ulm University. This is a rare, genetic neuronal developmental disorder that leads to a genetically diagnosable form of autism. The research centres on a gene called SHANK3. "This gene's loss of function can lead to disorders of nerve cell synapses and severely impair language, behaviour and everyday functions, among other things," explains Professor Tobias M. Böckers. The Director of the Institute of Anatomy and Cell Biology and Dean of Studies for Medicine is leading the "A Therapeutic for Shank3-related Autism (PMS)" project based at Ulm University, which is one of nine projects to receive new funding from the non-profit organisation for translational research SPARK NS in 2026.

As part of an international team, researchers from Milan and Ulm are developing and testing antisense oligonucleotides - short, synthetic nucleic acids that are used to treat genetic diseases - that are intended to increase the formation of the functional SHANK3 protein. The active substances had already been identified in a broad-based screening process and are now being further validated in patient-derived cell models and preclinical models. "Our aim is to create the basis for an initial clinical trial," says Professor Böckers. In parallel, biomarker measurements are being established, for example in blood and cerebrospinal fluid, in order to assess effects at an early stage and precisely control treatment. "The Phelan McDermid outpatient clinic of our colleagues in neurology offers the best conditions for this." SPARK NS not only provides funding, but also a high level of additional support. The aim is to increase the chances of therapeutics actually reaching the market and directly benefiting patients.

"The funding from SPARK NS gives us the opportunity to take a promising approach towards therapy - with the clear aim of opening up real prospects for families who currently have little more than supportive measures," says Tobias M. Böckers. "We want to be able to objectively measure as early as possible whether treatment reaches the brain and has an effect, and thus prepare the way for an initial clinical trial in a responsible and speedy manner."

About SPARK NS

SPARK NS is an independent, non-profit organisation for translational research based in the USA. Founded in 2023, the organisation aims to more effectively translate promising academic discoveries in the neurosciences from the laboratory to the clinic. In addition to financial support, the non-profit organisation offers researchers advice, training in drug development and translational research, a collaborative community and support after completion of the financial support programme. The SPARK NS Translational Research Programme currently comprises 22 projects in the USA, UK and Europe, which together can receive up to 44 million US dollars in funding and support for drug development.

Press release

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Further information

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