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Preclinical research on rare, early-onset diseases in children

From molecules to realistic therapeutic approaches: Fraunhofer ITMP receives funding for three translational projects developing new treatment methods for rare, hard-to-treat diseases in infancy or early childhood.

Göttingen and Hamburg. In the first Joint Transnational Call of the European Rare Disease Research Alliance (ERDERA) on “Preclinical therapy studies for rare diseases using small molecules and biologics” Fraunhofer Institute for Translational Medicine and Pharmacology ITMP has received funding for the CHAMPION, VALUEKCNQ, and SynLeigh projects. All three projects address rare diseases in the early pediatric stage, which have so far been very difficult or impossible to treat. The aim of the funding is to develop potential treatment options for a rare lysosomal storage disorder, a severe mitochondrial disease, and rare genetic epilepsies. “Approximately five out of 10,000 people suffer from a rare disease. Those affected face major challenges, from often late diagnosis to the lack of therapy. With our interdisciplinary research, we address exactly these issues and want to advance potential treatment methods for affected patients in collaboration with our international project partners,” explains Prof. Dr. Gerd Geisslinger, managing director of the Fraunhofer ITMP in Frankfurt. At the Göttingen and Hamburg locations, ITMP will conduct three-year projects in collaboration with international consortia to validate drugs that could lead to improved therapies for affected infants and young children. Patient organizations will also be consulted, serving as voices for the needs and experiences of affected children and their families. The three projects are funded with a total budget of approximately six million euros.

Research on treating severe lysosomal diseases in childhood

Within the CHAMPION project, new therapeutic approaches for Multiple Sulfatase Deficiency (MSD) will be developed. MSD is an extremely rare, degenerative, and currently incurable metabolic disorder, affecting about 1 in 500,000 people. It is caused by an inherited deficiency of the enzyme FGE. Symptoms include developmental delays, liver and bone problems, vision loss, epilepsy, and loss of motor and cognitive abilities. A potential therapeutic approach could involve drugs and their precursors originally developed for other diseases. Laboratory tests have already identified 56 substances as potential MSD therapies. In CHAMPION, these substances will be further studied for their therapeutic potential in MSD. Understanding the mechanisms by which these medications may act in MSD is also a key focus of the project. “In addition to additional research on the 56 substances, we will develop the best formulation for patients. We will also investigate whether drugs or substances that act in MSD might be suitable as therapies for similar diseases, such as other forms of dementia,” explains Prof. Dr.

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Lars Schlotawa, CHAMPION project coordinator at Fraunhofer ITMP Göttingen and pediatric neurologist at the University Medical Center Göttingen. The project is supported by an international consortium with expertise in drug development and MSD, as well as a patient organization.

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Development of new therapeutic approaches for rare genetically determined epilepsies

The VALUEKCNQ project addresses severe epileptic seizures beginning in early childhood. These are often caused by changes in the KCNQ family of genes, which encode neuronal potassium channels. These play a crucial role in modulating neuronal excitability. Mutations in these genes are a major cause of epileptic seizures and are linked to a broad spectrum of epilepsy phenotypes, including severe developmental disorders and epileptic encephalopathies (KCNQ-DEE). KCNQ-DEE manifests in the first months of life and is characterized by treatment-resistant seizures and developmental delays. The incidence is about 1-5 per 100,000 births. Therapies for these neurodevelopmental disorders are currently unavailable. "We target the activation of neuronal Kv7 channels, which could be a therapeutic option for treating epilepsy. In the predecessor project TreatKCNQ funded by EJP RD, we already searched for new substances that can increase the opening probability of Kv7 channels and identified a potent and selective candidate, JNJ-37822681, which was previously tested as an antipsychotic in late-stage clinical development. Now we want to explore whether JNJ-37822681 could also be effective in related forms of DEE with evidence of Kv7 channel involvement, such as Dravet syndrome," explains Dr. Ole Pless, head of the research group and VALUEKCNQ project coordinator at Fraunhofer ITMP Hamburg. During the project, preclinical evidence for the efficacy and safety of JNJ-37822681 is expected to emerge, which could improve therapies for KCNQ-associated diseases. An international consortium of research institutions with expertise in KCNQ biology and pathobiology, drug development, clinical treatment and care, and patient advocacy organizations has been established for this purpose.

Two therapeutics for treating Leigh syndrome discovered

The SynLeigh project focuses on Leigh syndrome spectrum disorders (LSS), which cause neurological developmental delays and movement disorders and typically present in childhood. The most common forms of LSS are incurable; about one in 40,000 people is affected. So far, there is no effective approved therapy. The international consortium aims to identify therapeutic interventions and potential synergistic effects to create a roadmap for the development of clinical trials for LSS. Rather than studying single drugs in isolation, SynLeigh specifically investigates synergistic combinations of promising agents to enhance their combined effect. Two of these therapeutic approaches have already received orphan drug designation from the European Medicines Agency (EMA). These insights will inform the ongoing project to evaluate the efficacy and toxicity of these molecules and their possible synergy. The goal is to develop a mechanistic understanding and a practical strategy to establish treatments for affected patients.

The project is coordinated by Prof. Dr. Alessandro Prigione, Department of General Pediatrics, Neonatology, and Pediatric Cardiology of the Heinrich-Heine-University Düsseldorf. Dr. Ole Pless of Fraunhofer ITMP serves as a partner.

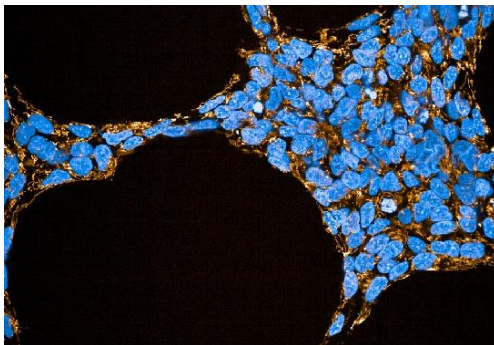
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High-throughput microscopy analysis of human cells.

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Microscopic image of living neural progenitor cells derived from induced pluripotent stem cells (iPS) from a patient with Leigh syndrome. The nucleus is blue, and the mitochondria are stained orange.

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