CHOPS Rare Diseases Foundation announces scientific collaboration with Rare Disease Translational Center at Jackson Lab

A new project is launched to develop targeted therapies for CHOPS syndrome and AFF4 gene mutations through Antisense Oligonucleotide (ASO) technology.

The Jackson Laboratory's Bar Harbor campus, home to the Rare Disease Translational Center (RDTC).

PALERMO AND BAR HARBOR, MAINE, ITALY - US, ITALY, December 4, 2025 /EINPresswire.com/ -- The Fondazione CHOPS Malattie Rare ETS (CHOPS Rare Diseases Foundation), the only organization in the world dedicated exclusively to CHOPS syndrome, is proud to announce the launch of a scientific collaboration with the Rare Disease Translational Center (RDTC) at The Jackson Laboratory (JAX), one of the world's leading institutions in genetics and translational medicine.

The partnership focuses on the design and in vitro testing of ASOs, an advanced technology based on synthetic molecules designed to modulate or correct AFF4 gene expression. Mutations in AFF4 are responsible for CHOPS syndrome, a severe neurodevelopmental and multi-organ disorder with fewer than 40 cases diagnosed worldwide.
CHOPS syndrome takes its name from the acronym describing its major clinical features: Cognitive impairment and Coarse facies, Heart defects, Obesity, Pulmonary involvement, Short stature and Skeletal dysplasia.

The AFF4 gene plays a vital regulatory role in gene expression, acting as a molecular "timer" that determines when key developmental genes are switched on. In CHOPS, this mechanism becomes dysregulated, triggering widespread effects on growth, development, and organ function.
The project will be carried out at the RDTC at JAX in Bar Harbor, Maine, USA with financial support from the CHOPS Foundation.
ASOs represent one of the most promising technologies in genetic medicine: short synthetic DNA sequences capable of "switching off," correcting, or modulating faulty genetic messages.

> This collaboration represents a decisive step toward targeted therapies for CHOPS syndrome and strengthens our global effort to accelerate research for this ultra-rare condition."
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> *Manuela Mallamaci, Founder and President, CHOPS Foundation*

The aim is to intervene directly at the biological root of the disease, paving the way for personalized and potentially transformative treatments.

"Our strategy is to activate multiple lines of high-level research in parallel, accelerating the path toward a targeted therapy for CHOPS syndrome," says Manuela Mallamaci, Founder and President of the CHOPS Rare Diseases Foundation ETS and researcher at the University of Palermo, Italy. "This collaboration with the RDTC represents the third line of research we are launching: after basic research, essential for understanding the molecular mechanisms of the disease, and after our drug repurposing program, which aims to identify existing medications with potential therapeutic benefit for CHOPS, this ASO project marks another decisive step toward highly innovative, targeted therapeutic approaches. Partnering with the RDTC allows us to combine the strength of our global network with the expertise of a scientific institution of outstanding international standing."

Jennifer SanMiguel, lead scientist at the RDTC, comments, "We are thrilled to join forces with the CHOPS Foundation on this groundbreaking initiative. Starting at the cellular level enables us to identify ASOs that demonstrate strong target engagement, a critical first step before advancing to studies that assess safety and efficacy. This work forms part of a broader, multi-pronged approach led by the CHOPS Rare Diseases Foundation to explore diverse therapeutic strategies for CHOPS syndrome. Our contribution focuses on building a solid foundation for future possibilities with the shared goal of moving science forward for this ultra-rare condition."

"Developing an ASO therapy for CHOPS exemplifies our commitment to accelerate precision medicine for rare genetic disorders. This collaboration reflects the RDTC's core goals: to transform cutting-edge research into tangible therapeutic solutions, foster strategic partnerships, and deliver hope to patients and families affected by conditions with no current treatment options," says Konstantinos Vanezis, scientific investigator at the RDTC. "Working closely with CHOPS strengthens our ability to translate scientific innovation into meaningful impact for the rare disease community."

This collaboration adds to the projects already underway with UT Southwestern Medical Center and Baylor College of Medicine (Texas), University of Essen (Germany), Children's Hospital of Philadelphia (Pennsylvania), Unravel Bioscience (Massachusetts).
To date, the CHOPS Foundation has funded six international research projects, investing over $400,000 and establishing the first patient-specific biobank dedicated to CHOPS syndrome.

The CHOPS Foundation was established in 2023 following the personal experience of Manuela

Mallamaci and Giovanni Zampella, parents of Mario, now 4 years old, one of the very few children in Italy diagnosed with CHOPS.
“Our battle is both scientific and deeply human, combining science, ethics, and long-term vision,” Manuela adds. “Research on ultra-rare diseases is not only of vital importance for my son’s life, it is a driver of innovation for global health: what we discover in CHOPS can illuminate therapeutic pathways relevant to many other conditions. Every new partnership brings us closer to a cure and strengthens a commitment that affects us all".

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