EUROPEAN
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Kosla, Malgorzata
PACS2 Research Foundation

Malgorzata (Gosia) Kosla, President of the PACS2 Research Foundation, a patient-led global research collaboration focused on the PACS2-related neurological disorder, also known as DEE66.

Her journey into rare disease advocacy began in February 2022, when one of her twin daughters, Lena, was diagnosed with this ultra-rare developmental and epileptic encephalopathy. Gosia and her husband, Piotr - then directors at international corporations - decided to apply their project management expertise and leadership experience to benefit not only their daughter but also other children affected by the disease.

Starting from virtually no scientific research on PACS2 syndrome, they developed a comprehensive drug development roadmap and, within just two years, established collaborations with more than 10 research institutions worldwide. Together, they launched a coordinated research program spanning basic science, translational research (including drug screening and RNA-based therapies), and clinical studies.

In 2024, research on PACS2 syndrome reached a major milestone by securing the first two grants ever awarded by the U.S. National Institutes of Health (NIH) for this condition. This was followed by a grant from the Polish National Science Centre (NCN) in 2025. The world's first personalized antisense oligonucleotide (ASO) therapy for a PACS2-related disorder, being developed by the n-Lorem Foundation, is currently in the final preclinical stage. Gosia also serves as a patient representative in the ePAG of ERN EpiCARE and on the Polish Rare Disease Council to the Minister of Health. In 2025, she co-founded the E+ Alliance, an initiative supporting patients living with rare and complex epilepsies across Europe.