**#missionCURE: we drive treatment development for rare genetic disorder**

Few sentences of your family story and/or on your child (hobbies, character, what you love about them), eg. For three beautiful months we were just normal parents – dying from no sleep but happy with our amazing twins, Lena and Zuzia. We laughed that Lena would be actress in future with her touching smile and very social personality. Then suddenly, in first day of Christmas, Lena started having series of epileptic seizures. We spent end of the year in the hospital and our world literally fell apart. All examination looked normal. But then the results of whole exome sequencing came…

Mid-February 2022 we already knew: a pathogenic variant in gene PACS2, in heterozygous state, the autosomal dominant mode. The description of disease leave no hope: „Key features of patients are moderate [8-10 years old] to severe [0-2 years old] intellectual disability, variable developmental delay [including limited or no speech], cerebellar dysgenesis and other central nervous system malformations, muscle hypotonia, facial dysmorphism, and behavioral disturbances [autism spectrum disorders].

Searching on Google did not help much. Just a few dozen cases globally. No cure.

But today there is hope. There is PACS2 Research Foundation working for patients impacted by PACS2 syndrome around the world. **The main goal of the Foundation is to drive treatment development by financing and coordinating scientific research**. They have already managed to engage incredible scientists and physicians to work on disease understanding and drug development, i.a. Oxford University, Northwestern University (Chicago) or Charles River Laboratories. In less than one year, from no interest in the disorder, there are 5 teams exploring different treatment options. At the moment the biggest challenge is financing. Without it the hope will stay just the perfect plan written on a piece of paper.

**Would you help?**

DONATE: <https://www.pacs2research.org/donate>

Read more about the foundation roadmap: <https://www.pacs2research.org/roadmap>