Rachel Heilmann, The Rory Belle Foundation, & NARS1-Associated Disorders

"The Rory Belle foundation will exist for as long as it needs to exist. The enduring matter is that rare isn't rare, right? We are a massive community of families that are directly impacted by something that nobody wanted... You don't choose this path, you don't choose trauma and grief, but you do choose what to do with it."

Life changed for Rachel and her family when her daughter Rory was born, 3 weeks early, very small, and with microcephaly. From her earliest days, Rory faced significant health challenges, including persistent vomiting and failure to thrive. These issues led to multiple hospitalizations within her first few months, notably a sixteen-day stay during which she experienced her initial seizure. In an effort to understand the cause of the problem, Rachel's family provided samples for whole exome sequencing, a time-consuming process further delayed by the COVID pandemic.

At eight months, while Rory was in the hospital due to her failing liver, her geneticist finally received the sequencing results. Rory was diagnosed with an ultra-rare NARS1 genetic disorder. At the time, research on NARS1 was nearly non-existent, Rachel recalled, saying "There was nothing – you Googled NARS1 and you got a French make-up company – there were no publications." Rory's geneticist tapped into their network and was able to find one researcher who was working on this gene, who indicated that, at that time, only 30 individuals worldwide were known to be affected.

Unfortunately, Rory's condition worsened. To improve quality of life, doctors prescribed numerous drugs to lessen the seizures, decrease spasticity and pain, and reduce fluid buildup from Rory's failing liver. Despite all her medical struggles, Rory found moments of joy. They weren't enough though. At seventeen months, after a brief life marked by severe medical struggles, Rory passed away.

The Rory Belle Foundation

Early after Rory's diagnosis, two publications on NARS1 were released which enabled Rachel to reach out to researchers and eventually connect with Dr. Stephanie Efthymiou of the University College London Institute of Neurology. Together, Stephanie and Rachel formed a Facebook group to connect with other families affected by NARS1. Through the group, Rachel met Terry DeClercq, a mother of a child with NARS1, and together they founded the Rory Belle Foundation. For Rachel, the Foundation was an inspiration from Rory. "That girl wanted to fight. She wanted to live, and she fought so hard to do that." The Foundation "extends the life of Rory through awareness and research of the NARS1 disease."

"Regardless of whether your child passes away or struggles every day to live, it's grief, right? We all share a common thread of grief and for a life that our children will never have... And I think when you are in the middle of your grief, you have a choice to be swallowed up by the waves, or you find the surfboard and the gratitude and you hit the wave and you move forward with something that's bigger than you and your daughter, with the hope that other people don't have to do it."

Rachel considers herself lucky to have had the healthcare knowledge and clinical training to have been able to understand the processes through which her daughter endured. But she also realizes that many parents do not have that same expertise. "What do people do who don't know how to advocate, how to ask the questions, or to understand what the physician is saying?" It is here that the Rory Belle Foundation functions as support for parents, as well as serving to advance research into treatments and cure for this devastating condition.

Fostering A Research Community

Thanks to a research fellowship at COMBINEDBrain, while the Rory Belle Foundation was still a fledgling organization, Rachel was able to supplement her clinical background with a "crash course in neuroscience" which in turn led to her role as Director of Research and Community Engagement at the MED13L foundation. The goal for both MED13L and Rory Belle Foundations is cure, but to get there, per Rachel, "requires putting things in place." The knowledge of these "things" – meaning the resources and the tools necessary to be ready for pre-clinical and in-human trials – is what Rachel brings to each foundation.

But of course, research is a team effort. An initial focus for the Rory Belle Foundation was expanding the limited research being conducted on NARS1. When Rory was first diagnosed, there was only one researcher in the world actively studying the condition. In just two years, the foundation has grown that network to ten researchers and driven the development of cell, yeast, and mouse models for research and drug testing. Beyond this, the Rory Belle Foundation has been proactive in engaging the families and scientific community, submitting abstracts, delivering presentations, and creating educational materials to raise awareness about NARS1. As importantly, the foundation has connected with the greater rare community to learn from each other.

This communal approach extends beyond research methodologies to a support system for parents. The Rory Belle Foundation is, as Rachel puts it, "building the community of NARS1 families that are affected, but also exposing them to other [rare disease] groups, to recognize... you're really not alone."

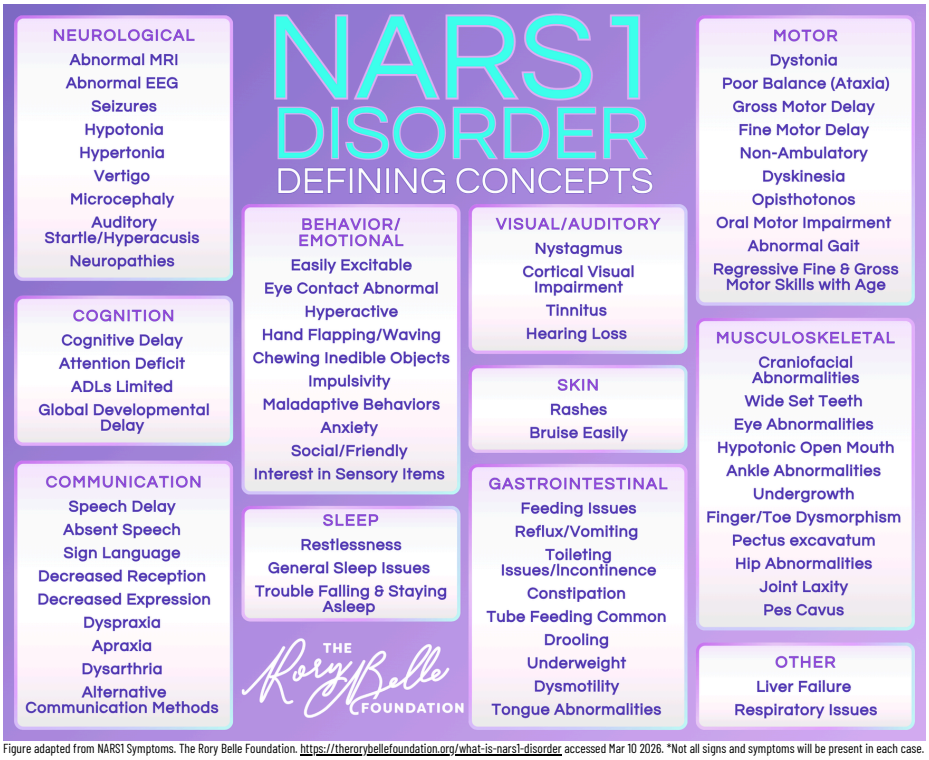

Figure adapted from NARS1 Symptoms. The Rory Belle Foundation. https://therorybellefoundation.org/what-is-nars1-disorder accessed Mar 10 2026. *Not all signs and symptoms will be present in each case.

With less than 100 known cases worldwide, NARS1-associated disorders are exceptionally rare. Mutations in the NARS1 gene may be inherited or arise spontaneously, with pathogenic variants leading to impaired activity of Asparaginyl tRNA synthetase 1, a crucial enzyme involved in protein synthesis. Those affected have highly varied and often severe symptoms, including failure to thrive, seizures, and global developmental delay.

The Path Ahead

The Foundation's efforts have already yielded significant results and is better positioned for sustainability and capacity due to the recent award of a Rare as One grant from the Chan Zuckerberg Initiative. These funds are critical for the mission, which Rachel shared is "ultimately to find a cure" while acknowledging the path ahead, "We like to say at the Rory Bell Foundation that we're bringing joy today and hope for tomorrow for all individuals affected by NARS1 disorders."

Rachel Heilmann is a clinical pharmacist and the Director of Research and Community Engagement for the MED13L Foundation. She co-founded the Rory Belle Foundation shortly after the passing of her daughter from NARS1-associated disorders. In a recent episode of Rare Awareness Radio, she shared her personal journey and the achievements to date and to come for the Rory Belle Foundation.

To Learn More

The https://therorybellefoundation.org/ website provides a host of resources for families, researchers, clinicians, advocates, and other healthcare stakeholders.

Other resources for those looking to learn more about collaborating foundations mentioned in this podcast or the rare community in general include:

- COMBINEDBrain - https://combinedbrain.org/
- MED13L Foundation - https://med13l.org/
- SLC6A1 Connect - https://slc6a1connect.org/
- FAM177A1 Research Fund - https://www.fam177a1.org/
- Rare Awareness Radio - https://rareawarenessradio.org/
- Principled Research Resources L3C - https://www.principledresources.com/rare-awareness-radio

Rare Awareness Radio is made possible by support from Meeting You Podcast, OMSLife Foundation, and Principled Research Resources L3C.