Jill Hawkins & The FAM177A1 Research Fund

With only a handful of known cases worldwide, FAM177A1 Associated Neurodevelopmental Disorder is considered one of the rarest of rare diseases, presenting immense challenges for affected families and the research community working to understand the disorder and develop effective, life-saving treatments.

Jill Hawkins is on a mission. As the founder of the FAM177A1 Research Fund, she is laser-focused on improving the lives of those affected by this ultra-rare genetic disorder and ultimately finding a cure. After a 13 year diagnostic odyssey, Jill and her husband Doug's two children, Charlotte and Cooper, were among the first in the world to be diagnosed with FAM177A1 disorder in 2019. At the time, the gene responsible for the condition was severely understudied, with only a handful of known cases globally. Jill quickly realized that if she wanted answers and a cure for her children, she would have to take matters into her own hands.

"When our kids were diagnosed, there were five papers ever published that even referenced the gene, and none of them explained what the gene was," Jill recalls. Undeterred, she created a beacon of information online, building a Facebook group and website to connect with other affected families and researchers. Her efforts paid off, as Jill was soon contacted by scientists intrigued by this mysterious gene.

A Rare and Complex Disorder

FAM177A1 Disorder is an extremely rare, autosomal recessive genetic condition caused by the loss of function in the FAM177A1, a gene having a critical role in neurodevelopment. Affected individuals have a complex set of symptoms typically including intellectual disability, significant delays

well as a unique gait that appears to regress over time. Many also suffer from uncontrolled seizures, which put them at risk of Sudden Unexpected Death in Epilepsy (SUDEP), a leading cause of death among those with difficult-to-treat epilepsy. In addition to the seizure risk, those with FAM177A1 disorder often exhibit behavioral disturbances including autism. Jill's children, now 19 and 14 years old, are among the oldest known patients with the disorder and require assistance with every aspect of daily life. "They're medically complex, vulnerable kids," Jill says. "We're treating symptoms, not the root cause. The seizures are awful, and the medications have brutal side effects."

"We are the among the rarest of the rares. I personally know six patients in the US, including my two, and about 20 other patients around the world. But we will find more now that it's been identified, even though we'll always be ultra-rare."

Determined to Change the Trajectory

Jill created the FAM177A1 Research Fund just a couple of years ago. The organization's mission is to not only bring hope to families, but to foster a patient-centered, collaborative research community with the shared goal of curing FAM177A1 disorder.

Jill has wasted no time making progress. Through the foundation's efforts, the gene has moved from the "variant of uncertain significance" category to "likely pathogenic," improving diagnostic accuracy for new patients. Jill has also united researchers across disciplines, breaking down silos and creating a network focused on understanding the disorder and developing treatments.

"Being a patient-centered, collaborative research organization is pulling in the researchers and clinicians and industry people that don't necessarily work together," Jill explains. "They're learning from the families, meeting the kids, and it's very motivating for them."

The foundation has also established a robust biobank of patient-derived cell lines, plasma, and blood samples, as well as various animal models to support its research initiatives. One exciting project is the development of a gene therapy program, which Jill believes holds great promise for curative treatment. The foundation is currently designing the viral vector and testing it in disease models,

with plans to initiate the program this year. In addition, the FAM177A1 Research Fund has funded a proteomics study to identify potential biomarkersand a drug repurposing project using a fruit fly model that uncovered a list of FDA-approved drugs that could potentially alleviate symptoms.

"Our disease is well-suited for gene therapy, and that would be the curative thing we would go after."

FAM177A1 RESEARCH FUND

To learn more about FAM177A1, please visit www.fam177a1.org

A Generous & Supportive Community

Jill's tireless efforts have not gone unnoticed. The foundation's Inaugural "FAM JAM" gala in Seattle raised an impressive $250,000 in a single night, and Jill has been buoyed by the generosity and support of the rare disease community.

"I've been so struck by the generosity of the rare disease community to share resources and share learnings," she says. "I'm a little bit further along the road, and I'm like, 'Come on, let me pull you forward. Let me pull you forward. I don't want it to be as hard for you as it was for me.'"

As Jill continues to navigate the challenges of rare disease research, her unwavering determination and the foundation's strategic roadmap offer hope for the future. With gene therapy on the horizon and a growing collaborative network, the FAM177A1 Research Fund is poised to not only help hose affected with this disorder, but possibly smooth the journey to treatment for those affected by other ultra-rare diseases.

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