



My name is Molly Tingley, and I lead patient outreach at [SLC6A1 Connect](#). I live in Iowa, and my daughter, Malia, was diagnosed with SLC6A1 at the age of 3. You have joined a club that nobody ever wants to join, but we are warm, welcoming, and here to help. If there is one thing we want you to know today, it is that you are not alone.

Many of our families receive zero information or support when diagnosed. Unfortunately, your experience is not isolated. Diseases like SLC6A1 are often categorized as *too rare to care* because we have so few patients that doctors, biotechs, and researchers are uninterested. SLC6A1 Connect is dedicated to changing that paradigm and ensuring that future patients do not walk the same paths as families who came before them. **We are solely focused on developing treatments and ultimately a cure for SLC6A1. We represent the patient.**

If you are located outside of the U.S., please let us know ASAP so we can connect you with one of our dear sister organizations.

History

SLC6A1 Connect was founded by [Amber Freed](#) in 2018 when her son Maxwell was diagnosed. Amber was told nothing could be done and that she should go home and give Maxwell the best life possible. She left her career in equity research and now focuses 100% of her time on SLC6A1. Like-minded parents have joined our cause from all over the world, including [Kim Fry](#), our Vice President/Fundraising Chair; [Laura Bermingham](#), our Program and Conference Organizer; and [Sandy McEntee](#), who handles all special projects.

What is SLC6A1?

To learn more about SLC6A1, you may find the following resources helpful:

- Watch this [video explaining SLC6A1](#).
- Watch this [video to understand genetic disease](#).
- Read the [SLC6A1 Neurodevelopmental Disorder article](#) by Goodspeed et al.
- Review this [SLC6A1 information brochure](#).

To-Do List

We understand your head is spinning, so we will prioritize next steps for you.

1. Are you seeing the correct doctors? Depending on your child's age and needs, you need a neurologist, developmental pediatrician, psychiatrist, occupational therapist, physical therapist, speech therapist, and a strong educational team. Our [Centers of Excellence](#) include medical teams who are familiar with SLC6A1 and have experience caring for our patients.
2. You must sign up for our patient registries in order to be notified of current and future breakthroughs.
 - [Patient Database](#): This information helps us reach out to keep you informed with news and information about future clinical trials and treatments. We need to know who you are so we can serve you best.
 - [The MATRIX](#): Matrix is running a Natural History Study for SLC6A1 Connect. Data is collected through surveys completed by patients, parents, and caregivers. You may also upload important medical records. We are asking all families worldwide to participate.
 - [Simons Searchlight](#): Simons collects data through surveys completed by parents and caregivers. This information is shared with researchers studying SLC6A1. Gift cards are provided for completing surveys. Learn more about Simons Searchlight and how to participate [here](#).
 - [Citizen Health \(US Families Only\)](#): Citizen Health partners with SLC6A1 Connect to use health data to improve care and drive research toward treatments and a cure for SLC6A1. It is also a free platform to retrieve, store, and share your loved one's medical records. Signing up takes about 10 minutes.

Treatments

Traditional treatments for SLC6A1 have primarily included anti-epileptic medications, along with medications and therapies used to manage comorbidities such as ADHD, anxiety, and other behavioral challenges. It is never a good time to be diagnosed with a rare disease like SLC6A1, but we have made incredible strides with [Centers of Excellence](#), a [repurposed drug](#), a [patient who has received gene therapy](#), and more treatments on the horizon.

More recently, researchers have identified an FDA-approved medication called glycerol phenylbutyrate (Ravicti), originally approved for urea cycle disorders, as potentially addressing the underlying cellular mechanism of SLC6A1. A [pilot study](#) involving nine SLC6A1 patients demonstrated safety and efficacy. The medication is expensive, and insurance approval can take time, so we recommend beginning conversations with your doctor as soon as possible. Our best advice is to engage with other parents and review our [playbook for obtaining coverage](#).

Additional Information

We hope the information above answers many of your basic questions, but we also encourage you to explore [our website](#) and [Resource Guide](#). You can stay connected by joining our [Facebook Support Group](#), following us on [Facebook](#) and [Instagram](#), and connecting with [SLC6A1 Connect](#) and [Amber Freed](#) on LinkedIn. Lastly, we host an annual International Symposium and Family Conference that brings together families, researchers, and clinicians. Videos and slides from the [2025 Symposium](#) are available on our website. Registration is open for the [2026 Conference](#), scheduled for December 3-4 in Denver, Colorado.

While this diagnosis may feel overwhelming today, please know that you are now part of a community that will walk alongside you every step of the way.