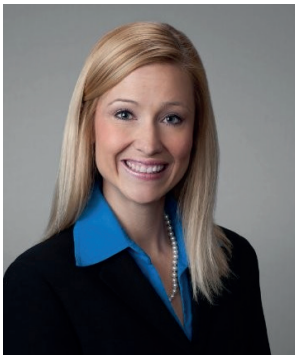
CELL & GENE THERAPY INSIGHTS

INTERVIEW

The power of patient advocacy in connecting industry, academia and patients: the story of SLC6A1



AMBER FREED and her husband Mark are the parents of adorable twins, Miss Riley James and Mr Maxwell Norman. Maxwell was 18 months old when the Freed family received his devastating diagnosis of SLC6A1. Ms Freed left her career in equity analysis the day Maxwell was diagnosed and dedicated her life to finding a cure. In 18 months, Amber has single-handedly driven multiple translational treatments forward and become a leader within the rare disease community. Ms Freed serves as the Founder and CEO of SLC6A1 Connect. SLC6A1 Connect's work has elevated awareness and created an ecosystem that can systematically help fund and consolidate research and treatment efforts. Her efforts have been highlighted in the Huffington Post, Buzzfeed, Bloomberg, CNBC and many more. Ms Freed was featured in the best-selling book, Shortcut to Prosperity, as an example of grit well before her skills were put to the ultimate test. Prior to Founding SLC6A1 Connect, Ms Freed served in a variety of equity and financial analysis roles, most recently in consumer equity research with Janus Henderson Investors. Prior to Janus, Ms Freed was a Vice President with Stout, Risius & Ross in Houston, Texas, focusing on private company and personal valuations. Ms Freed has also served in roles with RK Capital Management, Dividend Capital Trust, and KPMG LLP. Ms Freed attended the University of Denver for both undergraduate and graduate school, receiving degrees in Accounting on an academic scholarship. She was nominated for the Global Genes Rare Champion of Hope Award and sits on the Board of CombinedBrain. Amber can be reached at any hour of the day to advance science.

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Q How did you first learn about SLC6A1 and become an advocate?

AF: I spent my career in equity research and gave birth to twins in March of 2017 named Maxwell and Riley. They are the light of my life.

At 4 months, I noticed Maxwell wasn't progressing like his twin sister and he had bizarre symptoms; like the inability to use his hands. Every doctor dismissed my concerns but mother's intuition said differently as Maxwell missed every developmental milestone.

I remember the day Maxwell was diagnosed. My husband and I were called back to a cold, sterile room at the children's hospital full of doctors with sad faces. Genetic testing revealed that Maxwell had a rare neurological disease called SLC6A1. We were handed a five-page research article from Denmark and the doctors acknowledged our understanding of the disease would quickly surpass their own. We had no idea of what Maxwell's future would hold. All of the dreams we had for our baby slipped through our fingers as we tried to digest every parent's worst nightmare. It was the lowest moment of our lives and a sadness for which there is no words.

In that moment I decided to fight like a mother. If anyone was going to cure SLC6A1, it was going to be me. A determined mother does better work than any doctor or detective. I left my career the same day as Maxwell's diagnosis and have devoted 80 hours a week to curing this disease myself.

Q What is known about SLC6A1 so far, and what research still needs to be done?

AF: SLC6A1 encodes a GABA transporter, GAT-1, and mutations in the gene cause a progressive neurodevelopmental disease. It begins with a movement disorder, speech apraxia, intellectual disability, and develops into a debilitating form of epilepsy. There are currently no drugs that effectively treat SLC6A1. The patient organization is pursuing novel translational approaches and there is a large unmet need.

Q A lot of your advocacy work has focused on the development of a gene therapy approach to treat SLC6A1 – why is it considered a good candidate?

AF: It's a perfect candidate for a gene therapy approach. SLC6A1 a monogenic, haplo-insufficient, loss of function, and the required genetic material fits well into an adeno-associated viral vector that is already being used for spinal muscular atrophy and retinal diseases. SLC6A1 is also a candidate for an RNA approach such as an antisense oligonucleotide, or some micro-RNA approaches.

Q What stage is the potential gene therapy currently at?

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AF: We began preclinical work on a gene therapy approach in the fall of 2018 in conjunction with the University of Texas Southwestern. We are now finishing preclinical studies and progressing towards a clinical trial. The next steps will be toxicology, completing a natural history study, manufacturing and actually holding the clinical trial itself.

Q How do you connect with the right people in industry and academia?

AF: I believe that the value proposition and the authenticity of our patient organization has advanced our advocacy efforts by decades. Many scientists and companies may want to engage with patient organizations, and in many ways I think the onus is on the patient organizations to reach out. We know our disease better than anybody and we must educate academics and companies about us.

To this end, I've designed a one-page overview of SLC6A1 to help interested parties 'speed date' with SLC6A1. I pride myself on our rapid response times, accessible registry/natural history study and collaborative culture. We are the most enthusiastic patient population you will ever meet – we are putting excitement back into inhibitory neurotransmitters. SLC6A1 Connect hosts an annual symposium and over 100 scientists and biotechnology companies attended last year.

Q As a volunteer organization, how do you reconcile your own goals with those of researchers or for-profit businesses you work with?

AF: Coming from a background in capital markets, I have a sound understanding of how collaboration leads to greater success. One of my first observations about patient advocacy, industry, and academics, is that everyone is fragmented. I aim to bridge that gap. We provide the patients, advocacy, and research access to unite key stakeholders.

We host a monthly virtual lab meeting where every interested party is welcome and shares thoughts. The virtual lab meeting has been instrumental in building strong relationships and

advancing our mission to cure children. Some examples of shared efforts include our registry, natural history study, creating centers of excellence and academic/biotechnology partnerships.

We now have partnerships spanning the USA, Europe and Asia. We originally thought this disease was incredibly rare. But once we got everyone in the same room to discuss this, we found out something amazing: it's not.

SLC6A1 was added to genetic testing panels in 2017. Prior to 2017, SLC6A1 essentially didn't exist. We now know that our prevalence is actually 1 in 38,000. We quickly realized that SLC6A1 is a newly discovered disease and is actually not so rare. We are the tenth cause of autism, sixth cause of epilepsy, and play a major role in many psychiatric conditions. Our quest to save Maxwell quickly transcended our little family, and we held the ability to impact a multitude.

Q What advice do you have for anyone in academia or industry looking to engage with patient advocates?

AF: I think that the patient advocate role within organizations themselves is very important. And there are many patient organizations that get lost – they may not have the right professional background to know how to reach out and get in touch. The more information you can provide on your website the better – and providing a contact is incredibly helpful.

Another area to consider is companies that have developed drugs and then shelved them, for whatever reason. There is a potential market out there for rare diseases. I would advise companies to keep an open line of communication with academics that are keyed into non-profit organizations, sit on the board, or maybe make new connections and test drugs in animal models. There's so much more that can be done and a large opportunity for pharmaceutical companies.

Q What are your key priorities and goals for your advocacy work for the next 2 or 3 years?

AF: We will advance a gene therapy. It is not a question, it is a fact. My second goal is to advance an antisense oligonucleotide therapy. My third goal is to raise awareness for rare disease overall. Rare diseases are an extremely lonely place to be, and they are often forgotten. We are only able to rely on a couple of scientists, and there's no infrastructure for us.

However, rare diseases are not as rare as they might seem, and what can be a breakthrough for one can be a breakthrough for

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many. I really want to shine a spotlight on the opportunities available and the amazing things happening right now in rare diseases. If anybody had said that children with spinal muscular atrophy would be walking at the age of four 10 years ago, no doctor would have believed it. On my darkest days, I watch videos of children who have received Zolgensma®, to keep my spirits up and to remind myself that this will happen for my son too.

Ultimately, we're an organization of mums trying to cure our children. I have put my entire retirement accounts into this and I spend day and night fundraising. If anybody is reading this and has an interest in SLC6A1 or GAT-1, please reach out to Amber Freed. I return text messages at 3am. I am relentless, and I won't stop until there is a cure for every child.

AFFILIATION

Amber Freed
CEO, Founder & Mom; SLC6A1 Connect

AUTHORSHIP & CONFLICT OF INTEREST

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