Elsewhere in the genome

15q13.3 deletion

Interview By Julia Katz

> ***“Being a mom of a child with disability is the most special thing in the world. I was meant to be his mom, we have a special bond. He’s my whole world and I feel so blessed. S is my best friend and I love him more than I can tell you.”***

We all have unique stories to tell. For the first edition of our newsletter, we wanted to share the story of one of the families that has partnered with us to understand better the impact of genetics on mental health. It's a story about kindness, challenges, and growth, and it underscores the importance of advocacy across research and clinical care, but most importantly, within communities that can work together towards a shared goal.

We spoke with Ms. G, who told us a bit about her experience of being the mother and daughter of someone with a 15q13.3 deletion, and finding that she had this rare CNV herself in the quest for answers. She has done a fantastic job advocating for the 15q13.3 deletion community, and she created a group called “**Unique and Special 15q13.3 Microdeletion”.**

She learned that her son had the 15q13.3 microdeletion back in 2008, when he was 38, after his psychiatrist ordered a genetic test called chromosomal microarray. “I thought: he doesnt have anything like that. Nobody ever said he could have something like that or that we needed to do genetic testing. Nobody. Two weeks later, I got a phone call, and they said your son has a 15q13.3 microdeletion. I cried for two or three days because I knew that doctors had been missing something. Im a carrier and I felt bad at first. I blamed myself, but my mom and sister have it too. It would’ve been helpful for the family to know about that earlier. Before genetics, he got a lot of different diagnoses.”

“He has a heart of gold and a really good sense of humor”, says Ms. G. “He is 52 years old now and he likes to watch movies and sports with me. He loves John Wayne and Chuck Norris movies, as well as old western movies. A few years ago he was in a group home where they wrote to Chuck Norris and Chuck Norris wrote back with a picture and a signature! He loves moms homemade burritos and football and Tom Brady. He used to work at a grocery store, and a food court at a mall; all vendors loved him. He was also in special Olympics and ran track, bowling and golf.”

Unique and special 15q13.3 microdeletion

Ms. G had to figure out a lot of things on her own while her son was growing up. They moved around a lot and found mental health services as a helpful first step that connected them to other resources like case managers, social security, and information about guardianship. “When I lived in Georgia I went to NAMI (National Alliance on Mental Illness) It helped being able to talk to other parents.”

“I joined a group when I found out about the diagnosis. After that, I started reading about how to start a group and started my own group, which I named Unique and Special 15q13.3 Microdeletion. At least once a week a new parent joins the group; we have around 500 parents, including people from England and other countries. Somebody will say - I’m new to the group, help , I’m not sure what to do – we try to help them and give them advice, and support each other throughout. It is a good way to connect with others about research, resources, and emotional support. We have also had a positive experience with docs and wanted to have them involved in the group.

What advice would you offer to parents navigating the system new to the 15q13.3 community?

“Mental health is the place to start, get a good neurologist and good psychiatrist. It is hard to find good psychiatrists” says Ms.G, stating that they have been lucky with their current psychiatrist, of which they speak very highly. “Advocate for education, speak with genetic counselors, do your own research”

What has it been like to advocate for your son?

“I always advocated for S.” says Ms. G, who then refers to experiences from other families, “Because some can’t advocate for themselves, we have to do it for them. I’ve always gone to mental health services, which helped connect with other services. The genetic diagnosis allowed him to get more services”.