Through the prism: 17q12 Stories

From the moment you meet him, Alex shines with joy and energy. "He's crazy about anything with wheels, especially dump trucks as big as he is! He loves dancing, bowling, swimming, and (dangerously for the wallet) sushi. We're so proud and thankful to watch him grow and discover all the things he loves."

"For us, there was a lot of empowerment in receiving a diagnosis."

Alex is three years old, the younger son of Christina and Craig, who run the **Brentwood Chiropractic Clinic**, a wellness centre in Sherwood Park, Alberta. Along with his big brother Teddy (5), Alex fills their days with excitement, play, and curiosity. Christina and Craig kindly shared some of what they've experienced and learned as a family.

Looking back, Christina and Craig recall how their journey began. As they explain, "We first realized something was going on with Alex in utero. He had echogenic kidneys, so we did follow-up testing and ultimately received a diagnosis of ARPKD (*Autosomal Recessive Polycystic Kidney Disease*). We wanted to know if we were carriers, so we advocated for a genetic test and discovered that Alex had a 17q12 deletion, which was not something we had ever considered. It doesn't change who he is—it's just information about how we can best support him."

However, one of their biggest challenges was the absence of follow-up care, something common for many families facing rare diagnoses. As they recall, "We were like, what do we do now? We went to Facebook groups, and through the 17q12 community, we connected with others. It was a great way to learn the spectrum of the diagnosis. There's a lot of worry, but also a lot of hope. The community provided support, information, and helped us become better advocates for our son. We think there are real opportunities to collaborate across advocacy groups working on 17q12 symptoms."

A turning point came when they connected with *the Robin Hood Association.* "They provided early monitoring and encouraged us to apply for **PALS (Playing and Learning at School).** We knew Alex would likely face developmental challenges. When he started, he was nearly three and non-verbal. Now, he can speak in short sentences, ask for what he wants, advocate for himself, and follow instructions."

Thanks to PALS, they saw an absolute explosion in Alex's vocabulary, his receptiveness to speech, and his ability to express his needs. "The play between himself and his brother has changed exponentially for the better because he's able to communicate. Alex wakes up every day calling for "Teddy Bear"which is his name for his brother, and they play beautifully together."

Building on their clinic work and inspired by the *Nephrotic Syndrome Foundation* welcome packet, Cristina and Craig want to help create a more formal, proactive welcome for Canadian families receiving a 17q12 diagnosis.

"There's still so much stigma against kids who are different, not just for the children, but for parents and siblings, too. It's important to know you're not alone and to have a proactive circle of care. When we talk to Teddy, we explain Alex's kidneys are sick, using words he understands. We'd love to see more support on how to explain diagnoses to siblings and family, and more formal resources for people diagnosed later in life, so they know, *you're part of this community now, and we're here to help you.*"

Their advocacy expanded further when Christina and Craig participated as panellists and attendees at this year's 17q12 Virtual Forum. "It gave us incredible insights into the disease, symptoms, and research. We connected with **Dr. Alexander,** who is studying magnesium exchange and its role in kidney function. Knowing more has helped us advocate, especially when daycare policies don't account for medical potty-training challenges. This knowledge opens doors to inclusion through awareness."

As they reflect on their journey, Christina and Craig remain eager to connect with others. "We always say that if anyone in the community wants to reach out, we're here. We're passionate about accessible health care, good treatment, diagnosis, and supporting families on this journey."

We thank the Breckenridge family for opening their hearts and sharing their journey with us.