David R. Cox Scholarship for Rare Compassion Essay

"The Unsung Heroes: Parents of Children with a Rare Disease"

In today's world of fast-paced medicine, scientific advancement is always evolving. As a medical student, it seems like before I can memorize the pathophysiology, diagnosis, and management of a disease, a new treatment's been discovered. Understandably, for practical purposes, medical education tends to focus largely on "high yield" diseases. In turn, there are a myriad of medical conditions that I will simply not be taught about. Essentially, medical advancement is propelled forward based on supply and demand. Unfortunately, this means that rare diseases tend to take a back seat. The Global Genes: Allied in Rare Disease program has opened my eyes to the barriers that exist in our healthcare system, the challenges families dealing with rare diseases face, and to how I might contribute towards this cause.

As someone who deeply cares for the wellbeing of others and who felt compelled to dedicate my life's work to that purpose, I started my journey to becoming a medical doctor in 2016, at the University of Saskatchewan. Every day, I'm thankful for this education, as it has harnessed my compassion and bolstered my knowledge, both on the importance of patient-centered care, and around sociocultural awareness. I've learned meaningful lessons about Indigenous culture and health provision in Canada, around stigma and marginalization of vulnerable populations, and about advocacy for patients in need. Beyond my school and community work, this path has driven me to seek out additional opportunities where I could get involved as an emerging medical student; to continue building a sense of compassion and understanding for what patients go through, and how to be of service to them, as a future physician.

The opportunity to match with a patient with a rare disease was particularly appealing to me for several reasons: pragmatically, for self-education and to fill in gaps in my own medical

training which textbooks don't provide; spiritually, to identify with someone dealing with a rare disease and build a sincere appreciation for the challenges they face; and emotionally, out of an authentic disapproval of anyone feeling helpless or alone, and to be a support and advocate for them if I could. As a child, born to a family member with a disability, my blind father relied on me from an early age to guide him. This meant that I learned early on how to lead by example and look out for others. It made me realize what it might be like for someone to be in the dark, dependent on others, much like someone with a rare disease might be, when little to nothing is known about it.

In February 2018, I received notice that I was matched to a family with a 10-year-old girl named "Tara". I immediately recognized her diagnosis of "Trisomy 9" as a chromosomal disorder, but I'd never even heard of it before. My genetics lectures had always focused on the well-knowns: Trisomy 13, 18, 21. Tara's mother, "Jenna", was very open and forthcoming about Tara's medical condition, and she shared that she had signed up for the matching program, "to help young doctors". I remember reading those words and being amazed, that someone whose daughter was diagnosed with this disorder and who had undoubtedly overcome a lot, would enlist themselves for the benefit of someone else they didn't even know. I immediately realized how much of an advocate patients with rare diseases must be. That concept, grasped, provoked within me a bittersweet feeling of thankfulness for her participation, and simultaneous frustration for its requirement in the first place; both juxtaposed by the all-important dilemma: how best to tackle the issue of advancing knowledge and cures for rare disease, when triaging resources must be priority-stratified?

Tara is a healthy little girl with a normal life expectancy, but it wasn't always that way. When Tara was born, her parents knew, "something wasn't right". Jenna graciously sent me a

detailed notebook she'd compiled over the years, literally outlining every physician visit dating back to 2007. In just a short period of time, Tara and her family had endured over 95 visits to various specialists. Family doctors, pediatric orthopedic surgeons, cardiologists, ophthalmologists, and eventually geneticists – all were priceless in Tara's health experience. Reading through Tara's historical timeline gave me a sobering understanding of how little I understood about what patients with rare diseases (and their families) go through. Reflecting, I compared how easy my health experience in life has been to hers, and how much of her family's life has been dominated by illness and medical visits. More than anything, reading through the notebook from back to front made me realize how miraculous Tara was, and how incredibly strong her family has had to be.

Tara was born with a hole in her heart and a left sided vena cava, and a dislocated hip. Although these were manageable, she developed numerous other health problems in her first few years, including full body muscle spasms, chronic constipation, a wandering eye, and abnormal gross motor skill development. It wasn't until Tara started to interact with other kids that her parents knew she wasn't the same as the others. One day, when a friend's child who was younger than Tara tried to play with her, she turned to Jenna and asked, "What's wrong with her?" I tried to imagine myself as a father and in her position, receiving this question from an innocent child. It immediately broke my heart. Tara's parents promptly booked an appointment with their family doctor regarding her development, for which they received a referral to an audiologist and speech pathologist. Just in case, their doctor also ordered genetic testing for possible chromosomal abnormalities. When the genetic testing results came back, that was when the real medical "adventures" began for Tara's family. Tara had a very rare and not well understood disease.

Although there are several variants of Trisomy 9, each ranging in their symptom severity, having three copies of chromosome 9 in every cell tends to have some characteristic consequences.

Suddenly, Tara's cardiac defect, joint dislocation, seizure-like spasms, and developmental delays started to make a bit more sense; a syndrome of symptoms associated with Trisomy 9p (which looks like 47, XX, +del (9) (q13)). As soon as Tara was given this formal diagnosis, Jenna knew the life of her family would never be the same again. Little did she know, yet, how she would become Tara's champion, her personal researcher, her greatest historian, her biggest fan and advocate, and how devote she would be toward seeing her daughter live her best life.

Out of the 7,000 or more rare diseases recognized, there are less than 150 documented children or adult cases of Trisomy 9, worldwide. Feeling alone and scared, Jenna's family promptly set out to learn as much as they could about Trisomy 9 and what it meant for them, and sought out others like Tara. They eventually found and joined a small community of families with similar diagnoses. Connecting with others who understood their challenges allowed them to exchange information and support each other, and it provided some needed esprit de corps. I came to understand, through my exchanges with Tara's family, how vital these communities were for them. These families were immensely helpful not only with regards to remaining hopeful, but especially with helping set expectations, both for Jenna's family's personal education and awareness, and for that of their doctors.

Fast-forward eight years, longstanding joint problems finally resolving, and care provision gradually lessening, and Tara was finally "discharged" in 2016. Her parents could breathe at last and move forward, with care "as needed". When I inquired about how Jenna and her husband dealt with their daughter's diagnosis and her constant requirements over the years, her honest answer surprised me. Tara's diagnosis had taken her 8 years to accept, and often caused despair, fatigue to the point of exhaustion, and even resentment towards Tara. Inside, Jenna knew that Tara would never grow up the way she had imagined her daughter to before she was born. She described

dealing with diapers and total dependence for 10 years as a "total energy sapper". She quickly countered this point, however, by telling me that she finds peace in reminding herself that, "Tara will always be my little girl". Her answers gave me the impression that the first 8-10 years of Tara's life were a whirlwind for her poor loving parents, whom may never have gotten the chance to step back, or recover. I also gathered how enthralled parents can be for their children with special needs, and how easy it might be to let their own needs slip for the sake of their kin's.

I taught myself as much as I could around Trisomy 9, but I honestly learned more about rare disease, sacrifice, and the importance of social connection from Jenna and her family, than any website, textbook, or course could ever teach me. They gave me a more holistic, human perspective on what getting a diagnosis like Tara's meant. It later dawned on me how little is known about diseases with such a low prevalence, and how important advocacy and public awareness is. Through all my conversations with Tara's mother, I still had a few burning questions. I needed to know what Jenna's most positive and negative experiences were with parenting a child with a rare disease, what her most meaningful learning lessons were, what she felt was lacking on behalf of the medical community, and what might be done better if she could advise me as an aspiring doctor. Her authenticity and sincere explanations empowered me to write this paper.

One of the biggest positives for Jenna and her husband was to see their daughter become more independent than expected. When they first got Tara's diagnosis, they described it as incredibly disheartening. They met with very well educated doctors and geneticists, and nobody had any real idea what to expect, in terms of Tara's development and prognosis. Her doctors didn't think Tara would ever be able to even dress herself. When Tara started to do this one day, it therefore came as an awe-inspiring moment for her parents, that Tara might be able to live, at least, some kind of independent life in the future. Jenna shared that, "the celebrations of the little things

are so much more special with Tara". It came as no surprise to me that one of her biggest lessons, parenting Tara, has been patience. She has had to be extraordinarily patient with her daughter, with herself and her husband, with other parents, teachers, with doctors, and just with the world.

One of the things that upset me the most to learn was Jenna's biggest fear: the future. With the many unknowns of Tara's condition, it has made future planning next to impossible. Jenna and her husband want to know that Tara will be alright during school and in life, like any parents would, but most things in school have been extremely difficult to organize. Seemingly forever in wait, at the mercy of aptitude testing and school recommendations, Tara's family has been unable to look ahead with much certainty, or without paralysis. Questions like, "will Tara live with us, or in a group home?" remain unanswered and naturally add to the daily stress of the family. Jenna has coped with this so far, by trying not to look ahead too much, focussing on one day at a time, and working on what she can control. Both her and her husband have, together, been saving money for Tara, so that she is provided for. "This way", Jenna imparted, "she will not have to depend on her sister when her parents are gone". If that wasn't enough, the other major fear they have around Tara's future, is her safety. The notion of Tara, with special needs, maturing physically into a woman one day, but not mentally, is terrifying for Jenna and her husband.

When I asked Jenna what she felt could be improved by the medical community (e.g. raising awareness, research, advocacy) and what, in her eyes, were some potential barriers, she first recounted how very fortunate they have been with Tara's doctors. Jenna revealed, however, that no one in the medical community really understands Tara's diagnosis, and it has been largely up to her and her husband to advocate for Tara, always. I noticed, in Jenna's answers, that she was somewhat unable to put a finger on specifically what she would change. Perhaps that epitomizes

the dilemma around what can be done to improve patient care for rare disease, since it's a complex, case-by-case issue.

Although Jenna might not have outlined specific problems for me to tackle, she did impart some wise advice for me. I had asked her how I could be of maximal service to a patient with a rare disease, based on her history. Most importantly, she exclaimed, "always listen to the parents". Jenna mentioned that there were numerous occasions where doctors dismissed things she and her husband were concerned about, which was a terrible feeling. Having their concerns validated was a huge blessing for them, even if every problem Tara was experiencing couldn't be fixed, or addressed right away. She also advised me to "Be available". Although Jenna recognized the inherent challenges in carving out extra time when balancing care for hundreds of patients, she loved that if she needed to see her doctor, he would make time for Tara. When Jenna called the doctor's office, the administrative staff all knew who she was, and that they were special. They always knew who she was when she said, "I am Tara's mum."

Reflecting on potential areas for improvement in helping patients with rare disease, perhaps working on setting up more efficient childhood trajectory planning would be beneficial. How do we as a medical community reconcile a problem of tackling the unknowns around rare disease and special needs patient care, while balancing scientific advancement? In a system where public sponsorship and government funding is required to drive medical innovation, research around rare diseases are scarce and it doesn't get the attention it deserves. Jenna and her family should not have to wait and constantly have their future planning bottlenecked, by having a chromosomal defect that is not in their control. Obviously, each person's disabilities require individual assessment which can be difficult, but Tara deserves equal access to education and anything else

life can offer others. I believe it is our responsibility, as a society, to give them a voice and help see that happen.

Perhaps Gandhi had it right when he proclaimed, “Be the change you wish to see in the world”. Maybe taking individual steps towards improving my own awareness, and that of those around me, is key. If I can at least recognize syndromes like Tara’s, and if I can contribute to making an early diagnosis, involve the right care people, and facilitate putting supports in place for families, that to me could be a win. If I can take leadership and advocate for needed progress and community support, and collaborate to protect parents of children with rare disease, I think that, too, would be meaningful and impactful. Perhaps the little things matter most (like Jenna said about Tara), and that is how I might be able to help make a difference; building strong patient-doctor relationships, making extra time for those in need, and ensuring special needs patients are indeed treated specially.

Sometimes it’s easier to look ahead than it is to look behind, but it’s also said that you can’t know where you’re going unless you know where you’ve been. That is why I chose to listen and learn from Tara’s story. I am deeply appreciative of them sharing their lives with me, and I aspire to use the lessons I’ve learned to positively impact my future practice in medicine. Tara, and others like her, don’t get the same opportunities others do, so let’s not forget about rare disease or take for granted how spectacular our lives are. Life is fragile, and to think that the simple swap or deletion of a gene, like the flick of a light switch, can completely alter your life, it’s very solemn. In short, there may not be anything ground breaking in the lessons I have described or put forward, but I believe kindling the power of the human spirit and working together is paramount. Acting, and trying to make an individual difference may also cause a butterfly effect, which collectively, can help bring much needed awareness and light to rare disease around the world.