Finding Meaning in Medicine: The Power of Hope and Resilience

"The question is not what you look at, but what you see."
-Henry Mark Thoreau, Journal August 5, 1851

After our Neurology module in medical school, my classmates and I were well versed in cataloguing symptoms, memorizing medications, and reading CTs and MRIs rapidly. Even during our clinical encounters where we had honed our physical exam and history-taking skills, diagnostic accuracy and clinical acumen were paramount. I always assumed that social workers would assist patients and their families with finding resources like support groups and educational programs on their journey to wellness, never realizing the full impact of what living with a rare, long-term, and incurable genetic disease like spinocerebellar ataxia type 15/29 would mean on a day-to-day basis. Until I met Luke Fischer and his family, I wasn't sure what role, if any, a physician could realistically have in helping families cope with a chronic disease.[1]

One hot Sunday in July, Luke's family pulled up at his father's office building in a large silver van. As I shook hands with Luke's sister Madeline and father Mark, his mother Anne helped him from his car seat to the ground, instructing him to meet me and shake my hand. A seemingly frail and slender eight-year old, Luke grinned and wobbled towards me on the concrete pavement, with his mom holding his hand for support because of his cerebellar atrophy. I'd read up on the symptoms and presentation of spinocerebellar ataxia type 15/29 the night before: nystagmus, muscle hypotonia, dysmetria, cognitive delay, seizures, tremors, and speech problems with a congenital nonprogressive presentation. His IPTR1 gene on chromosome 3 was mutated, and Anne spoke of negative experiences with his past neurologists in her email correspondence. This was all I knew at the start of our encounter. I smiled, crouched down, and shook Luke's hand with both of mine to say hello.

Our discussion began inside an office building with a large floor fan aimed at the five of us. Luke played with toys while we introduced ourselves. As I learned, neither Anne nor Mark had a family history of any hereditary genetic diseases. Yet, from the moment Luke was born, it was clear something was wrong. He was slow to open his eyes, and all of the normal milestones of infancy were delayed. In fact, at 18 months, he couldn't even support his head well enough to sustain eye contact. At four years old, Luke was diagnosed with spinocerebellar ataxia type 15 due to a de novo missense mutation of the IPTR1 gene. Ironically, this was the very same gene my lab had been studying when I was an undergraduate, a fact that Anne noted when she emailed me for the first time. Sadly, I could proffer very little information about the laboratory's work, which was more relevant to apoptosis, cell biology, and various types of cancer rather than spinocerebellar ataxia types 15 and 29. The discomfort I felt made me wonder why a simple literature search could turn up so many articles on IPTR1 and cancer biology and such few articles on spinocerebellar ataxia related to IPTR1. While on one hand it must be so frustrating to know so little about a child's condition, practically, how much funding could the NIH devote to research on rare genetic diseases like spinocerebellar ataxia 15/29 that affected so few?

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[1] All names have been changed in order to comply with HIPAA. This includes the names of the patient, his family members, and his special-education advocate.

As we became more comfortable with one another, Mark and Anne let me perform a abbreviated neuro exam on Luke and play with Luke and Madeline, an 11-year old young woman who I learned was a stellar honor roll student and Luke's biggest idol. Luke chased us as we walked backward. He fell down every four steps, but refused to take my hand when standing back up, picking himself back up with relentless persistence. He wanted to do everything his sister could do, and his cerebellum was not going to stop him.

"The human capacity for burden is like bamboo- far more flexible than you'd ever believe at first glance."
— Jodi Picoult, **My Sister's Keeper**

Resilience and hope- the unquestionable foundation of the Fischer family's very existence. It was clear from their story that the four of them refused to accept any setback as permanent. Passive TV screen time was out of the question. Mark playfully rough-housed and practiced karate with his son, treating him like he would any child. Madeline would entertain Luke with Scooby-Doo and Spiderman toys, pushing him to walk longer distances and cheering him on during gait-training on the driveway in the morning. Every week, Anne took Luke for speech language therapy, physical therapy, and applied behavioral analysis therapy. Mark and Anne collaborated with Luke's teacher so playground activities would involve him as an active participant. All the kids in his class were required to play with Luke in order for them to obtain toys. This helped Luke make immense strides in his social development and verbal language skills, while also building up his self-esteem. Spinocerebellar ataxia was not a burden; it was a catalyst for close-knit family unity.

Unable to comprehend how the whole family could be so tenacious given the incurability of spinocerebellar ataxia, I asked Mark how he and his family could have such a positive outlook. He replied, "Neuroplasticity. If Jonathan Keleher from NPR could thrive without any cerebellum at all, Luke should do quite well with one." Luke is doing just that- thriving. Although it took Luke two years to sit up by himself and four to take his first steps, he's made incredible gains, now keeping up with his grade level in school. His family believes in him and refuses to let him stop believing in himself.

Challenge is the pathway to engagement and progress in our lives. But not all challenges are created equal. Some challenges make us feel alive, engaged, connected, and fulfilled. Others simply overwhelm us.

- **Brendan Burchard, The Charge: Activating the 10 Human Drives That Make You Feel Alive**

Life within the rare disease community is no easy feat. Mark lamented that budgeting $22,000 a year plus deductibles for Luke is difficult. He's not sure how he and Anne will pay for Madeline's college tuition, and regularly wonders how one-income families with chronic diseases survive. Existing entitlement programs don't benefit Luke nearly enough considering the nature of his condition. Speech therapy, which amounts to $60/session, is not covered by either parent's health insurance. Still, for his family, watching Luke's immense motor, speech, and cognitive gains is worth every sacrifice.

Sadly, not all families have fared as well as Luke's. Kara, the special education advocate who represented the Fischer family, told me over the phone during our one-hour chat that there is an 85% chance or higher than parents of children with disabilities will divorce. Therapy and treatment for acute conditions are expensive and for chronic diseases unthinkable. Parents like Mark worry their children will outlive them and their child's siblings will become caregivers burdened with huge medical bills. Isolation is another somber reality Mark and Anne face. Parents of other children, even close relatives, express worry when their children play with Luke because they fear he is fragile. Finding a babysitter can also be a real challenge. The stress of caregiving means that leisure time is scarce, and a respite is much needed.

Education is another arena fraught with difficulties. In schools with limited special-needs education budgets, especially those in rural or inner-city settings, children may get placed into low-quality programs that do not adequately foster individualized learning and the acquisition of the skills of daily living. Teachers may also be unfamiliar with various types of learning disabilities. For instance, a child with dyslexia or dysgraphia may be misdiagnosed with ADHD because of their teachers' assessments. In fact, much of the controversy surrounding Luke's educational setting had to do with his teacher's unfamiliarity with spinocerebellar ataxia. There was also a focus more on his limitations than his abilities, and school administrators felt uncomfortable having a child on campus with a rare genetic disease they knew nothing about. Cerebellar dysfunction didn't automatically equate to major cognitive deficits, a common misconception among the administrators at his first school. Fortunately, while advocating for Luke's inclusion at his first school rather than a transfer to a school with a special education program, Kara and school administrators came to an agreement consistent with IDEA, a law guaranteeing that Luke receive an education in the "least restrictive environment" possible. This federal law guarantees a free and appropriate public education (FAPE) where a student who has a disability (due to a genetic disease like spinocerebellar ataxia) has the opportunity to be educated with non-disabled peers to the greatest extent possible. Yet, not every parent has the financial means to hire an advocate or attorney equipped to challenge a school district in due process court if there is contention regarding a child's least restrictive environment. Schools serving many children with disabilities may also be reluctant to make too many allowances for any one student because other students and their families could demand equal benefits.

In most cases, special needs education only lasts until age 21 or 22, at which point children receiving services must have a transition plan in their Individualized Education Program. Options generally include work-study programs, vocational school, and specific training programs through the Department of Assistive and Rehabilitative Services. For patients with rare genetic diseases, the diagnostic odyssey can be lengthy, impacting the receipt of disability resources and development of a reasonable, long-term education and professional plan for the child.

What is even worse is that physicians themselves are not always well-equipped to treat patients with rare genetic diseases. Luke's parents saw four neurologists at a well-regarded children's hospital who failed to reach a diagnosis, did not make a referral to a geneticist, and were evasive about their lack of knowledge. Because they couldn't diagnose Luke, his first physicians didn't provide the family with adequate social service resources. This pattern is all

too common. Lamarque Polvado, founder of the non-profit technology company CareStarter, developed an application with a virtual social worker who delivers ten appropriate healthcare resources in the patient's community/vicinity to address this need. The CareStarter database includes information on therapy, medical information, respite care, nutrition, and family support services, and has already gained traction in some parts of Texas.[2] Physicians who learn that a child has been having problems with his/her special needs education can also refer families to special needs advocates like Kara who may represent them in due process court.

The statewide disability organization for the state the Fischer family and I live in published on their website that our state, though one of the most populous in the U.S., has the second worst financial outcomes for individuals with intellectual and developmental disabilities in the country.[3] Unemployment rates for those with disabilities, some of whom suffer from rare genetic diseases, are roughly twice the statewide unemployment rate of 5.9%.[4] However, the disability community is not as cohesive as other minority communities because there are so many types of disabilities- adult, geriatric, pediatric, motor, language, and speech, to name a few. Moreover, if caregivers are looking after their loved ones with disabilities, it becomes so much harder to lobby politicians for increased funding of special needs educational programs. It was hard enough for the Fischer family to meet with me for a full two hours without Luke getting antsy and impatient; political lobbying would be out of the question. To make matters worse, the state we live in also claims sovereign immunity in cases involving the American with Disabilities Act (ADA), a federal law that guarantees civil rights to people with disabilities. This means that the state is immune from disability lawsuits brought under the ADA. There are many gains to be made in civil rights for those who are disabled and funding for special education.

In the meantime, as a medical student, I can help advocate for children with disabilities at a statewide level through the state's pediatric society or on behalf of the statewide disability organization. Another idea could be to contact CareStarter, and find out (with faculty mentorship) how to implement and popularize the app at the medical institution I'm training at. However, asking social worker colleagues what resources already exist in the metropolitan area for the rare disease community might be a more appropriate place to start. I also plan to share what what I've learned with fellow medical students in the Genetics Track, a 'minor' for students in medical school interested in Medical Genetics, so that they know how important it is to connect patients and families with resources.

What I took away from meeting Luke and his family was a better sense for how a single mutation could irrevocably alter so many lives, including my own. The medical school preclinical curriculum can reduce a person's disease condition to a series of symptoms treated with medications and/or therapy under adverse time constraints. Reevaluating a disease in terms of its impact on a family- financially, socially, personally, and even politically, caused me to realize that physicians and medical students need to truly listen to patients and family members

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[2] Polvado, Lamarque. "Caring for Kids." Filmed Dec 2013. YouTube video, 16:27. Posted Dec 2013. < https://www.youtube.com/watch?v=o_gNwQztWG8>

[3] Source Withheld. Would provide identifying information on the patient's and author's location.

[4] Source Withheld. Would provide identifying information on the patient's and author's location.

even if that may mean a longer clinical visit and running behind schedule. Only by listening will we able to address our patients' needs and help them obtain the resources they so badly need. Handing out contact information for support groups rather than generic disease information sheets could go a long way, so that family members would be better able to cope with the emotional burden of caregiving. Helping special education advocates connect with families can have a lasting positive effect on a child's academic and professional trajectory. Placing patients and their families in touch with local, state, and/or national organizations may empower those with rare genetic diseases to have a voice in politics, and perhaps even seek elected office. If we as physicians can help our patients have a fighting chance in the world by focusing on their abilities and enabling their caregivers to foster their child's talents rather than magnifying their limitations, we can empower children with rare genetic diseases to unlock their full potential.