NARRATIVE MEDICINE

The Sword of Damocles: Living with Neurofibromatosis Type 1

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ABSTRACT

Both as a physician and as a patient, having neurofibromatosis type 1 has been an important part of my life. In my practice as a physician and as a professor, I have tried to reflect on the reality of living with a genetic condition and how it affects the way I live and practice medicine.

INTRODUCTION

Some time ago, while examining a child with café-au-lait spots, a pediatric neurologist who had been one of my professors in medical school said to me: "Neurofibromatosis is like a Sword of Damocles … You never know when the bad things are going to happen." I never forgot those words.

Damocles was a member of the court of Dionysius II, king of Syracuse in Sicily. As told by Cicero,[1] Damocles was conversing once with the king about how fortunate the king was, as a man endowed with great power and authority. In response, the monarch graciously offered for Damocles to trade places with him. The courtier eagerly accepted. Damocles sat on the king's throne, surrounded by servants and luxury, only to discover that a huge sword hung above his head, held only by a single hair of a horse's tail. The sword might fall on him any second and, then again, it might not.[1]

I am a physician, and I have neurofibromatosis (NF) type 1 (NF1), a complex autosomal dominant neurocutaneous condition that mainly causes tumors and pigmentary lesions.[2] These tumors usually affect the skin and the nervous system. For as long as I can remember, my condition has felt, indeed, like a Sword of Damocles: A silent, constant, perhaps even dangerous companion. Many years before I got my medical degree, I remember my parents trying to explain to me why my skin resembled that of a giraffe's, a trait I used to hate and be very self-conscious about, but now have come to terms with.

THE UNCERTAINTY OF A DIAGNOSIS

I am my mother's youngest son; her pregnancy was normal, and I was born only a couple of weeks before term. Shortly after I was born, my parents noticed freckles under my right armpit and spots on my chest and arms. They had some freckles themselves and figured that it was probably a family trait, so they took me home. When the spots started to multiply and grow, my parents became concerned.

Because no one knew why I had these spots, I spent a great part of my childhood visiting hospitals and physicians' offices. I was very scared at first, of needles mostly. I felt hospitals were cold. After some time, I grew to feel comfortable in hospitals. Everyone was always so nice, the nurses and the physicians made the children laugh, and they gave them sweets and smiles. Hospital time became less scary and more fun because I got to skip some boring classes in elementary school.

It was less fun when I had brain computed tomography scans and magnetic resonance images (MRIs), which were long and gave me a headache, or when my father had to take me to a cancer center, and I was examined, poked, and prodded, by about 15 physicians. Those physicians shared a distinct look of bewilderment that screamed "I have no idea what is wrong with this boy."

Despite this daunting situation, my parents were great; they were very involved and protective. They even had "Fake Christmas Eve" the night before my first brain MRI. I stayed up late with my brother doing a treasure hunt for presents so that I would be exhausted and asleep by the time they took me to the hospital, thus ensuring I would be able to stay still during the test.

I vividly remember visiting the office of a pediatrician and looking at her libraries filled with leather-bound medical texts, neatly organized stethoscopes, blood pressure cuffs, and fountain pens. The room was filled with the sound of classical music. It was an incredible place, and I wished I could someday work in an office like that. Moreover, the physician was so knowledgeable and kind, that I was always in awe of her presence. I admired the fact that someone could be that smart, well-spoken, and kind.

Physicians found that my neurodevelopment was normal, other than the fact that I was a clumsy child, often dropping things or falling down. Gathering from what I have been told and what I can remember from early childhood, most of my struggles came from emotional development. Although apparently I had good academic performance, teachers often worried that I spent a lot of time by myself and that I often looked sad and somber.

I did not have a diagnosis for many years, and for some time, I was misdiagnosed and thought to have Noonan syndrome with multiple lentigines (then known as leopard syndrome). This lack of a diagnosis was anguishing for me and my family. Every fever or minor ailment was a major concern for my parents. When I turned 9 years old, we were referred to a pediatric neurologist and a geneticist. It was these specialists who first said the word *neurofibromatosis* to me. I could hardly pronounce it then, yet now, not a day goes by when I don't say it to someone.

I remember when I first did an online search for the term *neurofibromatosis.* A myriad of Web sites and shocking images

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stared back at me. In disbelief and with my scant medical knowledge then, I remember drawing one simple conclusion from that exercise: "I am definitely going to die before anyone else I know."

MEDICAL SCHOOL

The feeling of safety that I experienced when I was in physicians' offices stuck with me. I felt at home in hospitals. I set aside the idea of becoming a journalist, like my father, and I applied to medical school.

More than that, I almost felt I had to study medicine. I wanted to help people. As cliché as it sounds, it was true. I was not naïve; I did not become a physician because I thought I would find a cure for NF1. It was not about that; it was about listening to these people, these families. People like me, like us, who were left in the dark with a diagnosis hanging above their heads and not having a clue of where to go from there, how to keep on living knowing there was something wrong with their child.

I eagerly waited for the day I would receive a class on NF. The first time it happened, I was in my second year. I remember it vividly—partly because I was happy to see my own geneticist teaching the class and partly because I was terrified. Every single picture, every statistic, everything suddenly became very real.

I realize, of course, that I could have it much worse and that I am clearly not the first physician to have a particular condition or disease. (As much as we try to drive our humanity out of ourselves to become of use to our patients, sobering realities such as physical illness become a harsh reminder.) Nonetheless, the prospect of not knowing when something will hit you is daunting. I have grown to like my spots and my freckles, but what happens when it becomes more than that?

WORKING AS A DOCTOR WITH NEUROFIBROMATOSIS TYPE 1

I have heard the story 100 times. "Our baby was born, someone [a physician, a nurse] saw the spots and told us our child would not live long, or they scared us beyond comprehension. We have forever resented that [physician/nurse] and the harm he did to our family." The first time I heard it, it was my father telling the story; the latest, a 30-something-year-old mother in whose son I had just diagnosed NF1.

I remember one patient I came across while attending a surgical oncology rotation: A 15-year-old girl with severe chronic pain in her upper extremities. Physical examination findings and an MRI revealed a malignant peripheral nerve sheath tumor that developed from a preexistent plexiform neurofibroma in her right arm. The tumor was very large and invasive, and she needed to have her arm amputated because of it. I had to scrub in for the procedure. I was ready, and then I felt myself gasping for breath. I scrubbed out and broke down into tears. Hiding outside the operating room, I called my father, and I was scared and embarrassed. I felt inadequate and unfit to be that girl's physician. I was. I seemed unable to concentrate on her and see her as my patient. Rather than going over the steps for the surgical procedure, I saw myself on that operating room table. Although I wanted to help that girl, I could not shake from my mind the thought "What if this happens to me?" How should a physician act in such a scenario? Should I have refrained from treating her and sought out a colleague? Or should I have referred the patient to a different hospital because her case affected me in a very personal way? How can physicians take care of patients while taking care of themselves?

Fortunately, not all of my stories with NF1 through medical practice are that sad. I will never forget the first time I saw a 10-year-old boy with café-au-lait spots and freckles. He was crying because he was scared and did not understand; nor did his parents. I sat down with him, rolled up my sleeve, and showed him my arm that is full of macules and freckles. "You see?" I said, "We're just the same. Everything is going to be okay."

After some time practicing, and as many physicians do, I started to adapt. Medicine has brought me an inexplicable amount of joy, as well as a large share of grief. Some days are bad, and some are good. Nevertheless, encountering patients with NF1 was something I was unexpectedly unprepared for. I decided to delve headfirst into my fears and work in NF1 research for the betterment of people's lives, to aid in their understanding of this somewhat elusive condition.

My family, my partner, and my mentors (I am fortunate to have many of them) are incredibly supportive of me. However, at the end of the day, it is hard to get all my thoughts and feelings together and remind myself that I have to forget about being a patient, forget to be afraid, and provide comfort instead of expecting to be comforted.

The main NF1-related symptoms I have experienced in my adult life are paresthesias and pain in the areas where I have neurofibromas. I must have some of these neurofibromas removed because of the discomfort as well as the risk of their transformation to malignant peripheral nerve sheath tumors (MPNST).

As a university professor, I am involved in research regarding NF1, and I see patients with wildly different stories, on a daily basis in clinical genetics consultations. I have often told patients that whenever their physician tells them they have a certain amount of risk of cancer developing (1%, 50%, 90%), the number itself does not matter, whether high or low; when you hear that it is you, or your children, who may have a severe complication, every number becomes 100%. What happens when that becomes a reality?

Although having NF1 and having to think about it every day at work is very hard at times, I believe that getting involved in NF1 research has been a rewarding experience. It is not easy telling people that there is no cure for what their child has. It is hard to see patients become sick or die, not to mention that it is hard to wonder if that could be me someday. A number of factors went into making that career decision, including how it would affect me personally, my mental health, or my judgment. However, what I have found is that the pros very much outweigh the cons. Nothing feels quite as satisfying as knowing that I am part of a team of people from all over the world who have dedicated their careers to making life for families with NF1 better, to understanding an elusive condition such as this so we may anticipate severe complications such as malignancy or to

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do early intervention in children with learning disabilities.

My work with patients and medical associations has brought about another unforeseeably difficult thing to manage: Praise and admiration. People who meet me, especially mothers and fathers, are brought to tears, embrace me, and compliment my courage and my strength. I do not deserve all that praise.

I value their words and actions and believe that most of the credit goes to my parents, my brother, my partner, my friends, and my other family members; they're the ones who have been brave. I understand that families see me as living proof that their children can have a normal life and that they appreciate that someone with NF1 is taking care of them, someone who knows what they are going through. Caring for patients with NF1 has become my greatest honor and responsibility. I do believe these people have helped me to become a better physician, helped me understand what they are going through, and convinced me of the importance of continuing to work in NF1, both in research and in the clinic.

Working with these families and meeting parents and children with NF has been an extraordinary experience. Sitting down and listening to people's stories is both humbling and enlightening. Even when most genetic conditions do not have a specific treatment, people often find solace and peace with only a diagnosis. You can see it in their eyes; just knowing becomes key in coming to terms with a condition.

PARENTHOOD?

As a young person with NF1, concerns are mainly centered on you and what is going to happen to you as an individual. As you become older and get into a romantic relationship, concerns shift toward what is this new person in my life going to think? What will she say when she finds out about the risks? What is going to happen to our children? Are we even going to have children?

This is an actual discussion I have had with my partner, a neuropsychologist, on a number of occasions. It has never been easy. Her love and constant support of my condition have brought us this far, and I have no idea where I would be without her or where I would go. We both want to have children, a family of our own. At first, of course, children were as far from our minds as possible, and just knowing what could happen, makes you think, makes you worry. It is true, at least for us, that there is no such option as "leaving it up to chance." I have often discussed with patients that there is a 50:50 chance of NF1 inheritance in every pregnancy. Many couples take those odds, but others do not; as a patient, physician, and prospective father, I know I never could. For those wanting their own children, this leaves the expensive option (both financially and emotionally) of preimplantation diagnosis and in vitro fertilization—another long road ahead for prospective parents.

CLOSING REMARKS

I am grateful for all the people and circumstances in my life that have helped me to better understand my diagnosis. This gratitude comes from a rather complicated mixture of both knowledge and ignorance. There are some things I choose to believe. I hope, for the most part, people will find it easier to live with a chronic disease once they understand that balance is the most important thing. Knowledge can be liberating at times, and it can also be a burden.

My diagnosis of NF1 brings me closer to my patients in a way nothing else could. Having journeyed through the uncertain path of having a genetic condition provides some insight. I know the weight that my words have. Sometimes a single word a physician says, even a little thing, can haunt a patient and a family forever.

Empathy is an innate social skill, almost as much as it is a developed skill. Mostly, people relate and empathize with experiences they have lived themselves. I am very glad to be able to find a silver lining to having NF1. Humanity makes us better physicians, despite what some may believe.

There is also a deeper meaning to be understood from the story of Damocles. With NF1 it is very hard not to worry, not to be anxious and lose your mind over the day when the sword may finally drop, the day when things start going wrong.

It is, I believe, the responsibility of the physician to remind families to try to look past the complexity and severity of NF1, to live their lives to the fullest one day at a time without wondering when the disease is going to turn bad. That is the thing with chance: The day may never come.

Living with that "what if?" question and repeating it to myself daily is one of the hardest things I have ever had to do. Still, I have to try to stop wondering and focus on living a life of meaning, of purpose, perhaps even of being able to help someone. After all, the looming threat of death and disease hangs above all of us, patients and physicians. It is up to us to find the strength to stay together with those you love for good times and bad. Even in the face of adversity and fear of what may come, we must all strive to find happiness. ❖

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