What Mason Taught Me: The Sad Truth About Rare Diseases

I was first taught about the disease called hepatocellular carcinoma (HCC) during my first year of medical school in a pathology course. I was taught that it was a liver cancer induced by exposure to Hepatitis B virus, Hepatitis C virus, or alcoholic cirrhosis. I was taught the pathophysiological changes in the liver in each of these settings and how they set the stage for neoplastic activity. I was taught that HCC was, thus, highly preventable. But medical school did not teach me that HCC also affects children who have never been exposed to alcohol or hepatitis, that about one child per million develops unexplained HCC, or that the overall 5-year survival rate for children with this disease is just over 20%. Medical school did not teach me about the implications that come with developing an exceedingly rare disease; Mason[1] did.

Mason was 12 years old when I met him in a hospital waiting room. A cancer survivor myself, I was there for check-ups, so my anxiety was particularly high that day. As soon as I spotted Mason, I immediately forgot about my own concerns. A teenage boy playing on his laptop and wearing a hat to cover his chemo-stricken hairless head, he held a bucket in his lap for catching vomit. Though I knew I could not take away his illness, I figured distracting him was the next best option.

Although he was initially reluctant to respond to my efforts to make small talk, Mason gradually let his guard down. When he asked why I was there, I was honest; I told him of my previous diagnosis and treatments. Perhaps this earned me his trust, for he went on to tell me about his cancer. He said it was liver cancer, "the kind that alcoholics get," but that he "had never taken a single sip" of alcohol. By the time his cancer had been discovered, he explained, it had filled his liver and metastasized to his lungs. Liver transplantation was the only cure for a disease this advanced, but Mason was not eligible for a transplant unless the cancer in his lungs could be eradicated. Unfortunately, the cancer in his lungs was resistant to both chemotherapeutics and radiation therapy. Though he was still receiving various treatments, the treatments aimed to slow the growth of his tumors and alleviate his side effects. In short, Mason the 12-year-old knew he was dying.

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[1] Patient's name has been changed.

That was the day I became friends with Mason. From that point on, every time I went to the hospital, I looked for him. I sat with him as he received his chemotherapy treatments, listening to him as he told me stories about his friends back home and the dreams he once had for his future. We played board games and cards and developed our own inside jokes. I convinced him to stop wearing his hat everywhere—to stop being ashamed of his baldness. He taught me how to play video games and introduced me to the many different flavors of potato chips that exist. He was like the little brother I never had.

Throughout the year, I also grew to know Mason's mother. When Mason was asleep or too sick to visit me, I would talk to her instead. She told me about his diagnosis, describing nearly a year of appointments with various primary care physicians and specialists who attempted to explain his nausea and abdominal pain. Finally, one physician had decided to order Mason's liver function tests, but it was too late; by the time HCC was discovered in Mason's liver, it had already filled his lungs. Mason's mother was told that Stage IV HCC was highly evolved and could only be cured by removal. She was angry with the many doctors who had never considered cancer to be a possible diagnosis, but she was also guilty she hadn't been able to advocate for her son throughout his months of symptoms. She knew that the delay in Mason's diagnosis had been his death sentence.

Mason was, nonetheless, determined to fight for his life. Because of the rarity of his disease, his only option was to enroll in a variety of clinical trials, many of which came with debilitating side effects. Mason was constantly nauseous, achy, and fatigued, and the pain in his liver was difficult to control, no matter what painkillers his physicians prescribed. Once, Mason admitted to me that he often felt jealous of the children around him who had leukemia. I did not have to ask what he meant; I understood. With an incidence of 1 in 500 children, acute lymphoblastic leukemia (ALL) has a cure rate of over 90%. Yet this disease was once considered fatal. Because of its relatively common occurrence compared to other childhood cancers, the amount of funding towards research and clinical trials for the treatment of ALL has led to its cure. I now understand that Mason's disease is no more scientifically complicated than ALL; it just occurs far less

frequently. Because of this, funding for HCC research and treatment, like funding for all rare diseases, is difficult to come by. And because of this, Mason never got his cure.

I will never forget my last conversation with Mason. He was lying in bed, his eyes closed, holding onto a morphine pump and squeezing it every few seconds. He was thin, too thin, and he no longer looked like the budding teenager I had seen just months before. He was a little boy again. I walked into the room alone and sat down in the chair beside his bed. I was scared to speak, worried that I would wake him, and I didn't want to disturb him if he was feeling anything close to peace.

"Mason," I said, "I don't want to bother you, so if you don't want to talk then don't worry about it. I just want to be close to you for a little while." I knew it would be our last time together.

He slowly opened his eyes and forced a painful smile onto his face. "That's okay," he said, saying my name with fondness, "I can talk for a little while."

"How are you feeling?" I felt stupid for asking such an obvious question.

"Not so good," he said, "Pretty bad, actually."

"I'm so sorry you have to do this," I said to him as I began to cry. "I'm so sorry."

"It's not your fault," he answered, wise beyond his years.

I looked at him for a little while, feeling comfortable in the silence.

"I'm going to be a doctor for kids like you one day. I'm going to try to change things," I said.

"I know, Maggie," he looked at me for a moment, eyes filled with certainty.

"I'm going to let you rest now," I said, noticing the way he had begun to wince again. I stood up, leaned over him, and kissed him on the forehead. Then I walked out of the dark room.

Mason died of metastatic HCC within a year of our first meeting. Today, when I go to the hospital for my appointments, I catch myself looking for him. Since his death, I have researched childhood HCC as well as many other rare childhood diseases. To say that these diseases are unfair does little good. However, my friendship with Mason and his mother taught me that the rarity of a disease directly impacts the patients who have this disease. I now believe that raising awareness of rare genetic disorders, like those that lead to HCC, is one way that physicians can contribute to finding cures and treatments for

children like Mason. The scientific community (including pharmaceutical companies that are key for financial support) will not make an effort to understand and treat rare diseases until the general public considers this a priority.

Second, Mason's story taught me the importance of early detection of many diseases, especially cancers. Unfortunately, many primary care physicians are inexperienced with the detection of rare diseases. Medical schools should address these diseases thoroughly in spite of their infrequency. It is most of the primary care physician who discovers or diagnoses a patient with a rare disease—not the specialist. Primary care physicians should, thus, remain open-minded and keep their differentials broad. It is far better to be overly cautious in medicine than to jump to incorrect conclusions.

My relationship with Mason also exposed me to a stigma that often surrounds illness. Because many diseases are, at least in part, caused by lifestyle choices such as smoking, alcohol, UV-ray exposure, and poor diet, people who become ill are often judged. Mason, himself, admitted that he was embarrassed to have a disease that was associated with alcoholism. Many of the disorders and diseases caused by genetic abnormalities, however, are not linked to lifestyle. Whether genetic mutations are inherited or spontaneous, most of them cannot be prevented by healthy habits. I now believe it is my responsibility as a physician to help dispel common misconceptions and preconceived notions regarding diseases and their causes. It is also a physician's responsibility to advocate for patients and help them to better understand their conditions so they do not place blame on themselves. Guilt and self-blame can only reduce quality of life for patients with rare diseases—and lessen the likelihood that others will stand up to help raise awareness about these diseases.

Finally, Mason taught me about the experience of chronic and terminal illness. Mason knew that his disease would eventually end his life, but he chose to continue treatment anyway; he chose to be hopeful. It is important for physicians to honor the decisions of patients with poor prognoses, whether this means continuing treatments or stopping them altogether. What Mason needed most from his healthcare providers, family, and even me, was support and acceptance. During our last conversation, I promised Mason that I would be a doctor for children like him. He knew that I wanted to treat children with rare diseases, children who are often overlooked in the medical and

scientific community. While I hope to contribute to scientific advances in the treatment of rare diseases, my greatest goal is to serve as an advocate for patients who are often overlooked—patients like Mason.