

Quill & Scope

Volume 9 *Volume IX*

Article 13

2017

Sidekick

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Recommended Citation

Yum, B. (2017). Sidekick. *Quill & Scope*, 9 (1). Retrieved from

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SideKick

Brian (Kyu Won) Yum

They say doctors save lives. Doctors are heroes. One day, I picture myself seeing patients, diagnosing life-threatening problems and miraculously making them all better. I picture myself being the hero. It had not occurred to me that my duty as a doctor would be the sidekick, the supporting character for a different hero, until I met Max and his family.

Max is a patient diagnosed with a rare disease called Vanishing White Matter disease (VWM). He cannot run, walk, or jump like other kids. Instead, he must crawl and is bound to a wheelchair. However, rather than sulk about his condition, Max uses his seemingly never-ending smile and boundless enthusiasm to bring joy to those around him. By meeting Max and his family, I realized my role as a future physician may sometimes be constrained. I can be limited by the lack of available knowledge surrounding a disease. In such a case, a doctor's role is no longer simply to treat, but rather to support and ensure that those similarly affected by poorly understood diseases do not get marginalized in our society.

When Max's mother, Sarah, first told me that Max had VWM, my first instinct was to look up the condition online and read articles about it. VWM, I found out, is an inherited childhood leucoencephalopathy due to a mutation in one of the five subunits of eukaryotic translation initiation factor eIF2B. Though it is a housekeeping gene that is present in every cell, the mutation predominantly affects oligodendrocytes and astrocytes, which leads to neurologic symptoms such as ataxia. The disease is progressive, ultimately resulting in death, and the progression is accelerated by febrile infections or trauma. Currently, there are only about 170 living cases of VWM worldwide. I had an academic understanding of VWM. However I made a critical mistake in assuming that I this knowledge would allow me to grasp how this disease could affect not only Max, but his family as well.

But luckily for me, Max was a little kid more than ready to teach an overconfident medical student a thing or two. Upon meeting Max and his family, I quickly realized that the question I needed to answer was not "what is VWM," but rather "who is Max?" Who is this 4-year-old boy and how is VWM affecting his own life and that of his family? The traumatic experience Sarah and her husband experienced when they first discovered that Max could not walk and the adjustments Max and his family had had to make every day – those are what I needed to grasp to begin to understand what VWM was.

Sarah and her husband first saw symptoms of VWM when Max suddenly fell one day and lost his ability to walk. What must have been a terrifying experience as parents only got worse as they did not discover Max's proper diagnosis until months after the initial signs appeared.

There may be several reasons why a diagnosis took so long, and Sarah's experience offers valuable lessons about the pitfalls of healthcare. First, doctors and paramedics did not suspect VWM because it is so rare. Instead, they convinced Sarah that it was likely a hip injury that would improve with time. After all, it was the more likely explanation at the time. Second, since this event took place while Max's family was in England, the National Health Service prevented the family from seeing a specialist. Rather, they had to go through their designated health maintenance organization to obtain a referral, which prolonged the testing required for proper diagnosis. Third, in order to reduce healthcare costs, there has been an ongoing effort worldwide to curb the number of unnecessary tests administered to patients. One unintended consequence of this effort was that the moment the doctors believed that Max's injury was minor and not the manifestation of a bigger problem, Max was suddenly denied further testing, resulting in additional doctor visits that could have been avoided. Max's situation reminds me that the criteria for what is deemed unnecessary must be carefully defined – caution must be used to avoid excessively relying on cost reduction to the point of possibly harming patients. Once I start treating patients myself, I must be open-minded about the possibility that a patient may have a disease I had never come across. We are constantly taught in medical school to listen to the patient's stories and not jump to conclusions. And yet, it will always be a challenge to remain open to the possibility that our patient may require a diagnosis that we may never have seen before.

Sarah and her family moved to the U.S. in order to care for Max. The family was frustrated with the English system and its policies. They were losing time. Sarah and her husband believed that while healthcare in the United States was more expensive, they would have more freedom to see doctors of their choosing.

Unfortunately, the move to the States was not the end of their healthcare woes. While there was freedom to select their own doctors, Sarah had to deal with 5-6 doctors in order to get all the necessary medical attention for Max. Moreover, Max was waitlisted for Medicaid and is not expected to be able to obtain Medicaid benefits for another 3 years. Tragically, Max might not have 3 years to live.

In addition to having 5-6 different doctors for Max, Sarah must deal with is the fact that the doctors themselves do not know enough about VWM to treat Max adequately. During every initial visit, Sarah finds herself educating and informing the doctors about VWM. This reversal of roles is the sad truth that accompanies many rare diseases: patients or their parents are put in a situation where they have to be their own advocate and take it upon themselves to gather as much information as possible. Part

of being a responsible physician is having the humility to admit that there are still many diseases that we do not understand. In these circumstances, the doctor becomes incapable of treating; instead, the doctor must contribute in other ways, by providing comfort, support, and resources to the family.

When asked about how much support was available for someone like Max, Sarah confessed that while her family and friends have been more than supportive, there are very few organizations that offered her resources, as VWM is so rare. The lack of resources shine light on the bitter truth that unlike breast cancer, which can affect almost every family, rare diseases like VWM do not get the attention they need due to a lack of awareness Sarah has been a vocal advocate, posting online blogs detailing her story and trying to spread word about the realities of VWM and other rare diseases. I hope that their message gets across to as many people as possible but I believe more can be done. I plan to be a part of that movement through rare disease advocacy here at New York Medical College.

Despite the lack of information out there about VWM, I was pleasantly surprised to find that out of the 170 families dealing with VWM, Sarah knew more than 20 families throughout the world. What made that possible? Facebook. Financial resources are limited and there isn't an official website detailing VWM, but Sarah is part of a Facebook group with families across the world who are dealing with the same obstacles that she is. Sarah explained that the Facebook group offers invaluable information as families share their experiences and detailed progression of their children's symptoms. Sarah admitted that it helped to hear how others were doing, and it helped to know what symptoms to look out for as Max's disease progresses. It is astonishing to see the rise of social media increase access and communication for the victims of rare diseases. Fur-

thermore, through Facebook, I will be able to follow how Max is doing and help share his story.

The true heroes in Max's case are his family.

Oftentimes when people get sick, they look to the doctors to heal them, to be their heroes. However, when a disease is so rare and poorly understood, doctors can only do so much. Max's family stepped forward to take care of him when nobody could. Max's parents are not only his biggest advocates, but also his greatest admirers. They help him to maintain his positivity and live his life to the fullest. They take care of him every single day and provide everything he needs in his life, whether it is going to multiple doctors' appointments every day or just simply being by his side. Whatever Max requires, Max's parents are there for him, armed with endless love and support. Hearing Max's story helped me realize that a doctor's role is not always to be

the hero. Sometimes, our role is to be the sidekick – to read up on research being done, connect the family to the best experts, and provide as much support as possible to ease the burden. And that is a role I am more than willing to embrace.

If Superman couldn't fly, would he give up? My bet is that he would start running as fast as he could. For Max, VWM may have taken his ability to run, but it did not take away his ability to move forward. Max never gave up, and his family was there for him every step of the way. In meeting with Max and his family, not only did I learn to be open-minded about a patient's diagnosis, but I was also humbled by the heroics of the family in the face of adversity. As a future physician, I hope to be a trusty sidekick who cheers, supports, and spreads awareness about VWM and other rare disease communities, so that we may expedite the progress to understand the disease and find a cure.

