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Partnering with the Patient

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By Catherine Loftus

Many children look forward to jumping into the pool on a hot summer day, building a snowman to celebrate the first snowfall, or running to the door in excitement to greet a friend coming over to play. These are activities that inspire joy in the life of a child. But for children like Maddy*, living with Alternating Hemiplegia of Childhood (AHC), these activities are also triggers. When Maddy gets too hot or too cold—or too excited, upset, or exhausted—she may experience a debilitating episode of painful spasms and partial or total body paralysis. Since these episodes often accompany particularly salient moments of life, treating Maddy is a juggling act. The goal is to reduce episodes while allowing Maddy to live fully.

Since AHC is a rare neurological disease, finding a diagnosis can be a harrowing experience. First, a healthy-appearing infant or toddler begins having episodes of weakness or paralysis that resolve. The child often presents to the ER with "hemiparesis", a medical term referring to paralysis of one side of the body. The initial presentation can look suspiciously like the effects of a stroke, which is often what clinicians first suspect. Neurologists are often puzzled when the symptoms suddenly switch to the other side of the body or disappear entirely. At this point, the family is usually sent home. Then, a few weeks later, the same thing happens again. After trips to many specialists, across a spectrum of fields, the correct diagnosis is finally reached. Yet, for families of AHC kids, this is just the beginning. There is no cure for AHC, and symptoms can vary significantly from child-to-child.[1] For example, dystonic posturing and seizures are two characteristics that vary from patient-to-patient. Maddy experiences dystonia, but does not have seizures. Despite the diversity in symptoms, management is limited to a few drugs, shown to provide only a modest benefit, and vigilance—caregivers learn over time how to avoid triggers and recognize and manage symptoms.

The name Alternating Hemiplegia of Childhood is really a misnomer. Although attacks lessen in frequency and severity as children enter adolescence and adulthood, symptoms persist throughout life and are usually accompanied by pervasive developmental delay. Flunarizine is a calcium channel blocker that has been observed to reduce the severity of AHC attacks and the number of episodes in some children. Considered the best treatment option by leading specialists, Flunarizine only works in certain cases and is not yet approved by the Food and Drug Administration. Although further research is needed to establish the long-term effects of flunarizine, AHC children do not have the luxury of time. Attacks, aside from being disruptive, also contribute to permanent cognitive and motor impairment. By reducing the frequency of these attacks, the goal is to improve the child's long-term prognosis. Armed with this knowledge, families of AHC children often use flunarizine despite the risks.[2]

Drugs that induce sleep, such as benzodiazepines, can also be used to control symptoms. Sleep, for unknown reasons, is an effective weapon against the debilitating effects of AHC. Patients experience no symptoms while asleep, and although they may recur upon waking during particularly severe episodes, there is usually a period of improvement before this happens. The mechanism behind sleep's effectiveness, like much else in AHC, has yet to be characterized.[1] When Maddy shows signs that an episode may be imminent, such as yawning frequently and appearing exhausted, she is encouraged to sleep. In some cases, this measure can prevent an episode from occurring altogether.

The poem "Welcome to Holland" characterizes the experience of raising a child who is different, comparing the discovery of a child's disability to stepping off a plane and realizing the

plane has landed in Holland instead of Italy. As the initial shock wears off, the narrator discovers that Holland is different from Italy, but has its own joys. "Holland has tulips. Holland even has Rembrandts."[3] For AHC families, they step off a plane and they aren't in Italy or Holland or any other recognizable place. There is no blueprint on raising a child with AHC; it is entirely uncharted territory. Families often travel hundreds of miles to reach specialists that are few-and-far between. And apart from dedicated specialists, most medical professionals have no formal training in treating a child with AHC. They simply draw upon diseases they have seen—or worse, make erroneous assumptions.

When Maddy needs to go to the emergency room, it can feel like gearing up for battle. Even when her family calls ahead and makes sure the hospital is aware of her rare condition, they are often unprepared for such an unusual case. One day, Maddy was admitted overnight to the hospital after a long day waiting for tests to be done. Waiting is hard for Maddy, and finally her mother sought out a physician to find out what the hold-up was. She discovered that they were waiting for a neurological consult. One of the tests required a tube to be put up Maddy's nose, and the physician was hesitant due to his lack of familiarity with her condition. Maddy's mother was surprised. She knew that Maddy would not tolerate the tube well, both from her experience with her daughter and her extensive knowledge of her condition. "I could have given you that information. I wish you had asked me," she said simply. The physician looked a bit taken aback. Admittedly, he had not spoken to Maddy's family since their initial intake session earlier that day.

As a medical student, that interaction resonated with me. It makes sense that Maddy's mother, who has lived with her condition for years, would have a profound understanding of what works for her daughter and what doesn't. Not only can she draw upon own intuition as a parent and caretaker, she can also draw on knowledge of what has worked successfully in the past. She possesses a different type of knowledge — an expertise no neurologist would have. This type of knowledge can be easy to overlook in a clinical setting, and yet, when carefully considered, can greatly enhance a patient's quality of care. As a future medical professional, experiences like this are formative for me. Sometimes the expert that is needed to crack the case is the only person in the room not wearing a white coat.

In rare disease cases, medical professionals should not underestimate the family's wealth of knowledge about the condition and should not be afraid to tap into it. Rare disease families can be an asset. These families pore over resources for years, gaining a sophisticated knowledge of their child's disease that is often second only to leading experts in the field. Meanwhile, the vast knowledge a physician needs to be successful is compacted into four years of medical school. Medical education necessarily emphasizes conditions that are commonly seen on the wards, with everyday perpetrators like diabetes and head trauma demanding a larger slice of the pie. Physicians are taught that patients ask the questions and doctors provide the answers. But, in reality, the physician-patient relationship is much more nuanced. In rare disease cases, often physicians are the ones with the questions and patients are the ones with the answers.

I am very fortunate to have met Maddy and become a part of her journey. And, in turn, she has become a part of mine. As a future physician, I now recognize my responsibility to advocate for children like Maddy, so that when they enter a medical setting, they are encountering medical professionals who are adept at treating rare diseases. Rather than educating physicians about the intimate details of every rare disorder, however, it is more important to promote flexible thinking through team-based exercises during the first two years of medical school. Doctors are well-versed in integrating new information with what they already

know; they are trained to do this from the outset. Yet, there lies an inherent danger in forcing new information into an existing schema. Physicians should not be afraid to share what they know, but must also admit what they don't. They must be willing to accept help from their patients, even though that concept may seem antithetical to their training. Medical education must be adapted to place a greater emphasis on effective physician-patient communication during the formative preclinical years. Well-rounded physicians are better equipped to provide quality care to patients with rare diseases.

AHC is one example of a rare disease, but there are many others. Some share similarities, and some do not. Yet these diseases share a community, and it is our job as future medical professionals to collaborate with the rare disease community in pursuit of a common goal. We must spread awareness for diseases that many people have never heard of. We must educate the public and medical professionals alike. We must help children like Maddy. All the while, we must recognize that our efforts may impact other children—children who have not yet been born to families who have not yet heard the name of the disease that their child may someday have. These are the stakes. Thanks to collaboration with patients, like Maddy and her family, my experience has taught me that the rare disease community is eager to meet this challenge. It is our job, as future physicians, to partner with them.

*Patient name has been changed to protect privacy.

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