April 2016

Dear KCNQ2 parents,

I want to start out by welcoming you to this community. You will soon find that in addition to the fear and worry that comes with a diagnosis of KCNQ2 for your child, you also gain entry to one of the most amazing and supportive group of families imaginable, filled with the most precious resource possible: people from all over the world who will be able to say to you "I know what you are going through," and really mean it. I say this at the outset because in some of my most difficult moments, this is what kept me going: I am plugged into this group. They have all been there.

Our cherubic daughter Lucy was born in August 2014 (6 pounds and 9.5 ounces, 20.5 inches long, ten fingers, ten toes, one nose). Despite over two days of exhausting labor, my husband and I could hardly contain our joy. After a smooth pregnancy and a long but uncomplicated birth, our perfect little girl was here! What more could we ask for?

It sounds so naive now but it honestly never occurred to me that anything like this could happen. My family was known for having fat, healthy, happy babies that quickly slept through the night and breezed past development milestones. I spent my pregnancy decorating Lucy's nursery and daydreaming about naps and snuggles through my maternity leave. I can't remember even once thinking that our daughter would be anything but healthy.

Things did not work out this way. A few days after her birth, Lucy started making little movements. At first we giggled every time because it seemed like she was just trying to pass gas, but we soon suspected that these movements were something more ominous. We explained our concerns to the pediatrician who said that babies make all kinds of movements and it didn't worry him. Even then I felt I could see him viewing our concerns through a 'first time parents worried about everything' filter. I tried to assuage myself that everything was OK, but the movements started to become more pronounced over the next few days and as I confessed my growing fear to my mom she insisted that we head back to the doctor's office for answers. I will be forever grateful for that push.

We went to the doctor again, this time armed with a video of what we had come to call one of Lucy's "episodes." After speaking with several nurses, who all agreed that it was probably gas related, the senior pediatrician came in and it was as if our little girl knew that this was the time: just minutes after he arrived her arms went out, she looked to the left and I knew that it was starting again. I jumped up and interrupted the doctor to make sure he saw. Within minutes Lucy and I were in the back of an ambulance racing toward Children's Hospital in Washington D.C. In between the blaring sirens, the EMT asked me basic questions, Lucy's full name, our address. I looked at this tiny baby, in her car seat, strapped to the gurney sleeping through all of this chaos. The EMT asked another question and I didn't hear it against the scream of the siren. "What is her social security number?," he shouted. I convulsed into tears and could barely speak, blurting out "she doesn't have one yet." I just wished I could scoop Lucy up and we could run away. She was ten days old.

The next two weeks in the NICU were a blur. Each day we stumbled toward the scrum of doctors to join morning rounds, eager to hear if the previous day's tests had yielded any answers. As the obvious answers were ruled out—no infection, no meningitis, no brain injury—we felt a quick surge of joy followed by the crushing let down. The neurology team let us know that each negative result pointed us closer to a more intractable, more dangerous form of

epilepsy—one that couldn't be cured and that had lasting impacts. I remember just wishing she had meningitis, or something else that could explain the seizures. The doctors told us not to turn to Dr. Google, at least not yet. I went against their advice during one long and scary night in the NICU and saw only terrifying words—profound delays, poor prognosis, and early death. I was still physically healing from Lucy's birth and couldn't even wrap my head around this reality.

When we were finally able to bring Lucy home, we were terrified. What if we messed up her medicines, what if something happened while we were asleep, what if the seizures started again? Well, the seizures did start again. We changed medicines. We learned to always have our phones on hand to tape any suspicious movements. We adapted. It was not easy.

In addition to the seizures and the sometimes crippling fear of what the future would bring, we also struggled to deal with Lucy's severe colic and reflux. She screamed constantly for her first four months. It was heartbreaking, often starting before she even opened her eyes. As we hunkered down trying to survive each day, I found myself feeling incredibly isolated from many other people in our lives, especially other new moms. It didn't help that no one in our family was getting much sleep.

But slowly it got better. One day as Lucy napped in my arms I saw her smile in her sleep. My heart melted. Then eventually she started smiling when awake and later even laughing—the most beautiful noise I had ever heard. While our day-to-day lives are more complicated than most peoples—medicines, medical appointments, therapies, exercises—we have also found a delicate balance, always searching for the next thing that will elicit giggles from our little girl.

And while everyday I continue to learn, there are three things I'd like you, as a parent new to the KCNQ2 world, to know.

First, while this diagnosis, and indeed this whole world, may seem completely foreign and overwhelming now, you will become the world-renowned expert on your child. Doctors will learn to trust your judgment and ask your assessment of how a treatment course should proceed. They will ask you to relay how other kids with KCNQ2 have responded to a similar protocol. Sometimes you will need to assert this authority and explain to a doctor why you want to try a different medicine or why you think your child needs a specific type of therapy. Sometimes it will be frustrating to make your voice heard. And sometimes this will feel scary, really scary, to be the one calling the shots. But the reality is that this condition is so rare that doctors will rely on you to tell them what they need to know.

Second, this can be an incredibly isolating experience. If you have a newborn, and he or she is anything like Lucy, then you are just trying to survive these tough first few months. But the diagnosis can also be very isolating, as you look around at other babies or children the same age. There is so much comparing that goes on that it can be hard not to see the differences. But as I mentioned up top, you also become part of the KCNQ2 family. If you join the Facebook group (which I highly recommend), you will likely find yourself sharing parts of your life that no one else sees and asking questions that you wouldn't ask anyone else. You will start to feel, as many of us already do, that you have a new expanded family around the world that really knows what you are going through.

Third, this amazing community that will provide you with so much support will also rely on you. Every new family that joins the KCNQ2 group is another source of hope, strength, and expertise. Every new diagnosis can teach families, doctors, and the research community more about KCNQ2, especially since many cases of KCNQ2 are caused by previously unknown

mutations. If your child was diagnosed later in childhood, you will be able to share so much of what you have already learned. In a group this small, every new case can provide critical information about the world of KCNQ2 and every parent is a wealth of knowledge.

Lucy is a year and a half old now and our family is still figuring out our journey day by day. I am incredibly grateful for the KCNQ2 community and know that everyone in the group looks forward to welcoming you.