Monday, July 25, 2016

I was driving back to the office after running errands over lunch. I'd just hung up the phone with a nurse from the local children's hospital, who had called to discuss details of Maddie's upcoming heart surgery. I laughed to myself as I looked down at my phone, and saw the familiar phone number pop up once again. "She must've forgotten something," I thought.

"Hello, again," I answered.

But it wasn't the same nurse who I'd just spoken with moments earlier. Instead, it was the genetic counselor who had been assigned to our family's case. Maddie was diagnosed with epilepsy in March, and since that time we'd been awaiting complete results of genetic testing to determine the cause of the seizures. We'd gone through two rounds of testing thus far, and no genetic "matches" had been found (a match would indicate a cause). Based on what they knew about Maddie, they'd explored the most likely causes first. The last time we'd spoken with the counselor she was doubtful that we'd find a cause. Sometimes seizures just happen. In some ways, I was hopeful for an answer. But in other ways, no cause was a relief- we could hope that these seizures were an isolated incident that she would outgrow.

The counselor didn't spend time mincing words. She eagerly told me that they'd come up with a match. My heartbeat quickened, but I wasn't overly concerned. Based on what they'd told us, the match might tell us something as minor as a dietary restriction. I asked her to hold so that I could conference Nate in on the call.

No answer on his end. No matter. I was eager to hear the news, so as I walked back into my office, I asked her to proceed with the information.

Thinking back to this day, things seemed much simpler. She told me that the match indicated that Maddie had a chromosome deletion called 1p36 Deletion Syndrome. That it was a rare genetic condition, and that it indicated that Maddie may have severe developmental and

physical disabilities. As I approached my desk, her words began to sink in. I felt a lump form in my throat and tears come to my eyes. I quickly ducked into a private conference room.

“What does that mean?”

She told me about how children with 1p36 often have hearing and eyesight loss. How they might experience delays in walking and talking. How her heart condition and the seizures were all related to this chromosome deletion.

“Will she live a normal life?” I asked.

The counselor paused… “I think she’s going to need some help.”

I became hysterical. She suggested that I try calling Nate again as a tactic to calm me down.

This time he answered, and I tried to recap what the counselor had said. He urged me to try to breathe. I could hear myself starting to hyperventilate, but couldn’t stop. I remember the counselor saying to me that Maddie was still our daughter. That she was the same baby I knew and loved.

She asked if we’d noticed any milestone delays to this point. I replied yes, but that we’d thought it was because of her heart condition. She always seemed so bright eyed and knowing- I’d never considered that there might be cognitive or other issues to contend with.

I remember Nate telling me that I needed to come home, and asking if he could pick me up. I responded that I would be okay.

The counselor scheduled an appointment for us to speak with a geneticist in two weeks and promised to email us information on the diagnosis. She warned not to dig too deeply on the internet, and to consider the information she sent as the only 100% reliable resource.

Meanwhile, a trusted coworker had texted me to see if I was okay. I was sure that the thin walls of the conference room did nothing to muffle the sounds of my agony, but I didn't care. I asked her to come in, and I quickly explained the situation.

I remember crying "I can't do this!" We'd already gone through so much since Maddie's birth: The NICU, diagnosis of a heart condition, seizure activity resulting in a hospital stay, pneumonia and time spent in the Cardiac ICU, going home on oxygen, and a surgery to remove a growth.

This was asking too much. How could we survive so many struggles in less than a year? And this news was unlike any of the other medical diagnosis that we'd received. There was no 'cure," no end, no foreseeable time that things would become 'normal.'

It left us with many questions. Had we moved from parents to caregivers? Would Maddie ever be 'typical?' Would she wear princess dresses, and do ballet; would she graduate from high school and one day walk down the aisle? It seemed everything that I'd dreamed for my little girl was now in question because of one phone call.

And the odd thing was, the genetic counselor acted so proud to share the information. As though they'd discovered a hidden gemstone in an unlikely place. They weren't expecting to find a match, but lucky us, they had.

Later the counselor told me that the reason they'd not checked into this diagnosis in the first two rounds of testing was that it was so improbable. Maddie did not display any of the physical characteristics of a child with 1p36. She passed her newborn hearing screening. Her vision seemed to be normal.

That evening Nate and I cried together. We took turns Googling the diagnosis, and quickly shutting the browser window after we read a few sentences, unable to process the info staring back at us.

I held Maddie and looked deep into her eyes, fishing for predictions on what her life would hold. I tried not to let the things I'd learned affect my love for her. And they didn't. I was relieved to see that the counselor was right about one thing: Maddie was still my daughter. The same baby I'd grown to know and love over the last eight months. And the same daughter I knew I would love to the depths of my soul for the rest of my life.

I'm not going to say that I somehow figured out how to handle her diagnosis with grace and dignity- I didn't. I'm not going to say that I don't still struggle with the information shared that day- I do. And I'm not going to say that I think any of this is fair- life isn't designed to be fair.

But I am going to say that God has a purpose for this. That we serve a God who doesn't make mistakes. That Maddie's life is every bit as valuable as anyone else's, and that she will be a blessing to many. That worrying about the future is a complete waste of time. And that Nate and I are privileged to be chosen as special parents for this very special little girl.

Galatians 1:5

To God be the glory forever and ever! Amen

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Cristin Riebel is a mother to Madison Riebel, a very special girl with a rare chromosome deletion. Follow our family’s daily adventures of joy, confusion, and coping as we not only learn to navigate parenthood, but also learn the ropes of raising a child with special needs at MadsEverydayMiracles.com