Vaun Sanford Way and Infantile Onset Pompe's Disease

My boyfriend, Brian Way, and I are learners. As volleyball coaches, we emphasize the importance of being a GREAT learner and how that will lead you to being the BEST volleyball player you can be. We ourselves, spend much time learning more about teaching, coaching, volleyball, and anything that we can. When we found out we were expecting, we did what we do best- research and LEARN! I bought *What to Expect when Expecting*, *What to Eat when Expecting*, and had several pregnancy apps. Brian and I read articles on parenting and brain development in the first year. We were determined to be informed.

Our research was halted, as we needed to decide where to start our lives as a family. Brian was coaching volleyball at UMBC in Baltimore, Maryland. I was coaching at Kansas City Kansas Community College. Both of us were ready for new coaching opportunities, but I was reluctant to make a move since we were going to be first time parents. I had built a close relationship with my OB at St. Luke's on the Plaza in KC, MO and didn't want to transfer mid pregnancy. Brian, the most patient person in the world, wanted to keep me comfortable (I was a really emotional pregnant lady!), and he moved here to Kansas City. We rented an apartment in Overland Park, KS, and considered moving delivery to St. Luke's South down the street from us, but my doctor didn't deliver there. We went back to learning all about birth and delivery and settled into our new home.

Under advisement of my OB, Brian and I decided to take a new parents class at our local hospital before we delivered. We laughed when I couldn't swaddle the baby doll and cringed at the idea of using a rectal thermometer on our baby. The teacher made mention of a newborn screening and that our baby would be pricked on the heel, his DNA tested, and we'd most likely never hear anything about it ever again. A bit skeptical, I made a joke about how this really was a government conspiracy, and didn't talk about it again.

After 27 hours of labor, and a delay in active labor, our son Vaun Sanford Way was born on May 28, 2016 via emergency C-section at St. Luke's hospital in Kansas City, MO. He was 9 lbs. 7 ounces with a full head a thick, black hair. We were instantly in love with him. Brian's parents flew in from New Hampshire and spent a week with their new grandson and the new parents.

Brian's parents left on Friday morning. We took pictures and had a tearful good bye. Exhausted, Brian, Vaun and I began to sit and relax as a new family of 3. Our rest was interrupted by a phone call. I was only half listening (or maybe only half awake) as the woman on the other end stated her name and that she had some results from his stay at St. Luke's. Uncertain of what she was talking about, I cut the person on the phone off and hung up. A minute later they called back and slowly explained that Vaun had a flag on his newborn screening for a disease called Pompe's. They told us not to Google Pompe's, enjoy our weekend, and that they would see us Monday in the metabolic genetics clinic at Children's Mercy hospital.

I had put the lady on speakerphone the second phone call. Us learners had Googled Pompe's no sooner than when she said the name of the disease. Pompe's Disease. It didn't even sound real. I assumed it had something to do

with exposure to a volcanic eruption. Before we ended the call, we learned Pompe's Disease is a genetic disorder in which a complex sugar, glycogen, builds up in the muscles and impairs its function. The words FATAL and FIRST YEAR stood out of what we read next. We were confused. Vaun had no issues latching or feeding. He was a healthy sized baby. Brian and I didn't have any babies with health issues in our families. I spent the weekend thinking this must be a false positive, while Brian continued to read and research. We'd have to wait till Monday to sort things out.

Monday morning we headed to Children's Mercy in Kansas City, MO. Brian was prepared with questions and I came prepared for an apology that they worried us with this false positive. Dr. Heese spoke to us about the disease and test. He told us they would do some blood work on Vaun that day and assess him, along with a urine test. We would then be sent to cardiology for an echocardiogram and an EKG. Some of the blood work would be sent out for DNA testing along with the urine test, but the rest of the results we'd get that day. Dr. Heese's assessment was Vaun looked great! We headed to have Vaun's blood drawn then to cardiology. Still convinced this was all a mistake, we waited in the dark room where we had the echo. The cardiologist finally came into the room with a big smile. With a calm, bright voice, she explained that Vaun had hypertrophic cardiomyopathy, or as she explained thickening of the heart wall; symptomatic of Infantile Onset Pompe's disease. She said she was really excited to be working with us and that Dr. Heese would join us soon to explain what needed to happen next. Our whole world was shattered in those words. With tears, I wanted to punch the cardiologist for her lack of sensitivity. Excited was certainly not the word I'd use to describe that moment. Dr. Heese confirmed that Vaun's CK levels were extremely elevated and coupled with the hypertrophic cardiomyopathy; it was enough to formally diagnose Vaun with IOPD.

We drove home in silence with tears. The question of "how much time will he have" ran through my mind. Our beautiful new baby boy had an extremely rare, fatal disease. How did this happen? How do we save him? Can we save him? Unable to accept this news as reality, we relied on our strengths. We began LEARNING what we could about IOPD. We worked till the early morning, but we learned about enzyme replacement therapy and Dr. Priya Kishnani at Duke University. Dr. Kishnani is the foremost expert on Pompe's disease in the United States. We knew she would be able to help us. We sent an email and hoped to hear from her. Ten hours later, we did! We still remember the hope her words restored to us. She reminded us to focus beginning ERT as soon as possible, and that she would continue to communicate with Dr. Heese about Vaun's case and treatment. We also learned that the target for starting ERT with cases like Vaun was no later than 30 days of life.

Strengthened by our new knowledge, we headed to our next appointment with Dr. Heese. It would be the first of many times that Brian and I would have to push and advocate for the best care for Vaun. The staff asked if we needed to plan around any vacations or time to settle into the new family routine before starting treatment. Vaun would need to not only begin ERT but he would have a 6-week immune suppression treatment to assure his body would accept the

replacement enzyme. While some may appreciate the consideration of personal time from the staff, we felt like the team didn't understand the necessity to get treatment started before Vaun was 30 days old. We stayed hard on our timeline, and within a couple days we had plans to begin Vaun's treatment by 23 days old. On June 21, 2016 Vaun Way began enzyme replacement therapy. After much debate with Vaun's cardiologist and requesting a new doctor to replace that cardiologist, Vaun had outpatient surgery to place a port catheter to make infusions smoother and less stressful for him. We met our deadline and now began life as parents of Vaun.

The next few months muddled together. Vaun finished the immune suppression treatment, we began a routine of seeing doctors and having infusions every other week. Brian and I continued to research, read and learn more about Vaun's diagnosis. We set up therapy services with Infants and Toddlers of Johnson County. Vaun moved to bottle feedings in hopes of less fatigue when eating for him. Honestly, we spent much of the time waiting to see Vaun's health decline. Instead, he grew! Vaun began rolling over, holding his own bottle, laughing at dad, and all the things people celebrate in those first few months with a new baby. Our first piece of news that treatment was working was at Vaun's cardiology appointment when he was 6 months old. Vaun's echo showed a healthy heart that functions properly. He no longer suffered from hypertrophic cardiomyopathy.

Before we knew it, we were celebrating Vaun's first birthday. He amazed Brian and I. He was a growing, thriving little toddler. Vaun had learned so much that first year. Brian and I worked hard to believe that Vaun's story could be a different one, even when his geneticists told us this would be the best his life would ever be. We were discouraged from believing that Vaun could live a more healthy, typical life than that of what we read about when Vaun was diagnosed. We decided it was time to visit Dr. Kishnani at Duke and learn what more we could or should be doing.

Once at Duke, we quickly learned how special Vaun was. Cardiology, Pulmonology, Physical therapy, Speech therapy and Dr. Kishnani herself- they all had the same look of amazement on their faces when they met Vaun. Dr. Kishnani showed us studies and taught us that early diagnosis, immune suppression, and beginning enzyme replacement therapy so early had greatly impacted Vaun's life. Vaun walks independently. He breathes without the assistance of a ventilator. His heart functions healthy and normal. All things we never believed he would do. Dr. Kishnani recommended continued physical and speech therapy, along with and increases in Vaun's dosage of ERT. We took our recommendations home, only to be met with resistance from Vaun's geneticists. We began looking for a new doctor, and found our way to Children's of Omaha. Vaun began receiving weekly physical, speech, and occupational/feeding therapy at Britain Center at Advent Health, a therapy community for children under the age of kindergarten. Again, Vaun grew and learned.

Vaun is now 3 and half years old. He LOVES trains! Vaun can put together 100 piece puzzles without assistance. His favorite color is read and his favorite food is pepperoni pizza. Vaun has weekly, double dosage ERT infusions.

He continues to have therapy at Britain center, along with 2 days of preschool classes in a peer model class there. We visit Dr. Kishnani and her team yearly, and bring recommendations back to his therapists and Dr. Timothy Smith at KU Medical. Dr. Smith specializes in managing pediatric patients with multiple specialties involved in their care. He's a true learner. He works to collaborate and communicate with Vaun's specialists and because of that, Vaun truly gets the best care.

Like I said, Brian and I are learners. Our journey has taught us a lot! While many things have impacted Vaun's story, the first thing that did was his diagnosis. The Missouri Newborn Screening for Pompe's disease saved Vaun's life, and saved mine and Brian's. Nine days old. He was nine days old. The difference between 9 days and even 30 days are huge in terms of treatment for Pompe's disease. Vaun didn't have to lose function of muscles to be diagnosed. He didn't almost die for someone to decide he may have a rare genetic disorder. Not only did early diagnosis keep Vaun alive, it impacted his quality of life. He can walk and run and jump, things I took for granted before I knew my son might never be able to do those things. But while adding Pompe's disease to Kansas newborn screening would be life changing for these children, what happens after is just as important. I've watched as other states have tested for Pompe's, but children are still waiting 1, 2, even 3 or 4 months for a diagnosis. Vaun's result from the screening came on a Friday, and on Monday we saw a geneticist. That is crucial, especially for IOPD children who need to begin ERT as soon as possible. I urge you to have a plan or system of how these screenings lead to a quick diagnosis and early treatment.

Thank you for considering adding this test to the Kansas newborn screening and thank you for listening to our story. Our only hope is to be able to share what we've learned and hope that it can help another child and family.

Paloma Juarez
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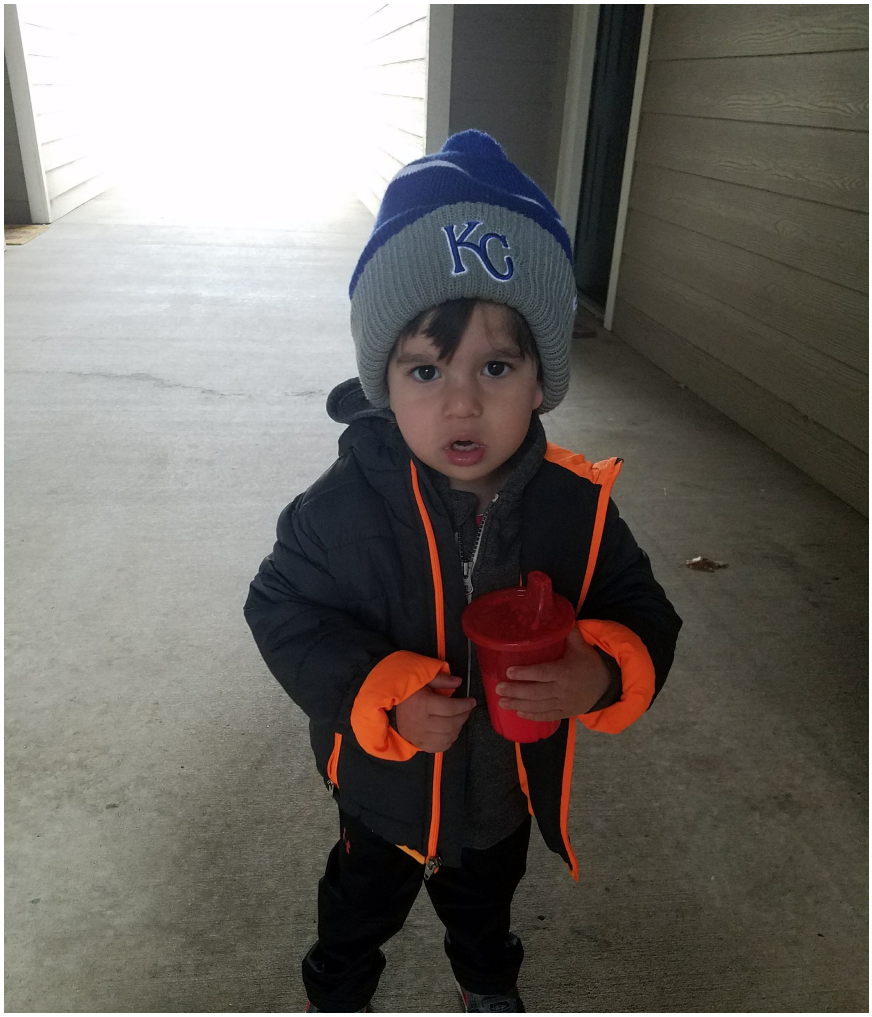
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