

## PKU: Owen - Learning the Weight



Our son Owen was born on a Monday afternoon in September. He had a rocky arrival and ended up in the level 2 NICU. He had to stay until Thursday morning, when they determined that his rough entrance had not caused any harm and he was safe to go home. At some point during his hospital stay, I remember signing permission for the newborn screen, not thinking much of it, not realizing it would save my son from severe developmental delays. The Thursday after he was released, we

got a call stating that his newborn screen had shown signs that he had Phenylketonuria (PKU). I remember them saying that it was not likely a false positive because the number was so high. They told us to drop what we were doing and go get the second screen done. I remember questioning them, because he was still about four days from being two weeks old. They assured me I needed to get it done immediately. They said he would need to be on a very strict diet for the rest of his life, and to not give him any formula until we had spoken to the clinic - just breast milk. We made a clinic appointment for the following Tuesday where we would be getting special formula for him and talking with the doctor and dietician. I asked if we needed to get the special formula sooner and they assured me it was safe to wait until the appointment. The wait seemed like forever.

My husband and I dropped what we were doing and took our precious newborn to the lab. We watched with heartbreak as the phlebotomist poked his heel and squeezed and squeezed to fill all five circles on the filter paper. Then we wrapped him up and went to Grammy's as planned for our weekly Thursday Night Dinner. I remember telling my parents and siblings about the call and how we had to rush to the lab. And of course there was plenty of web searching to see what we could find out about PKU. We mostly found that, if he started followed a life-long strict diet low in protein, he could avoid developmental delays and behavioral/psychological problems, but was it already too late? We prayed for a false positive. After all, no one in either of our families had PKU. But we would just have to wait until Tuesday.

That Tuesday we arrived at the clinic and met some people who changed our lives forever. We were told that Owen for sure had PKU, and that it looked like he had the classic (or most severe) type, because his blood phenylalanine levels were so high. We were told that it would take some time and monitoring to get his levels back down to where they need to be for him to grow and develop normally, but that he would, in fact, grow and develop normally. The special formula was going to bring his blood phenylalanine levels down. He was to only drink special formula for two days, and then we would slowly add in measured amounts of breast milk, depending on how much phenylalanine was showing in his blood work. He would need to come back to the hospital three or four times a week for measurements and blood levels, to ensure he was growing at a healthy rate, and to ensure he was

getting the proper nutrition that would bring his levels back down to a safe point. We met with several people at that clinic appointment, a geneticist who explained how he came to have PKU, a social worker, a psychologist, and a nurse. We met a dietician who has turned out to be one of the most amazing people on this planet. We are so incredibly grateful for her and what she has done for our family. Finally, we met the metabolic doctor - an incredible man who has his patients' best interests at heart at all times. We were given a huge book with the phenylalanine levels of every food imaginable for later use, a large packet of information, and a case of special formula which had all the other amino acids of protein in it, except for the one his precious tiny body couldn't break down on its own.

It felt like it took forever before they finally told us his blood levels were in the safe range. In reality it was just a couple of months. I remember thinking and worrying about brain damage, knowing that the longer his blood was "toxic" the closer he was to having brain damage. But he kept meeting or exceeding all of his milestones. He smiled on time, rolled over on time, sat up on time, crawled on time, and was even walking at 10 months. He was growing at a rate the doctors weren't concerned about. With the help of his doctor and dietician, we figured out how much formula he needed and how much phenylalanine was safe for him each day, and he was developing on track. Slowly we worked down to doing blood levels only twice a week, and then down to once a week. We started him on solid foods, weighing out every portion. Our dietician gave us a phenylalanine number to aim for and we made sure we hit it, without going over, every day. Of course we did. And every day it became easier.

So we built a new routine. Every day we wake up, measure out and mix the medical formula, weigh each item of breakfast, each item of lunch, and figure out ahead of time how much of each food he could have for dinner. If he only has 75 phenylalanine points left for the day, he can have one regular taco shell for 40 points, 100 grams of tomato for about 25 points, and 5 grams of vegan cheese for about 10 points. Oh no, did we forget to write down that orange he ate earlier? We need to subtract points for that and recalculate dinner. And we didn't leave room for dessert, so he'll have to have something phenylalanine-free for dessert. Believe it or not, we often have the opposite problem. He has 200 points left for dinner, what are we going to give him to fill that number? After all, his body needs just the right amount of phenylalanine to function properly. He's going to need something higher in phenylalanine, but something that's still appropriate for his diet. But we figure it out, and we get e-mails and calls from the dietician and doctor stating what a wonderful job we are doing keeping our precious son in such good metabolic control. It's rewarding, and so are his smile, along with the reports from his preschool teacher saying he is doing fabulous, and that first hit off the tee. He's going to be okay!

Sometime around age three and a half a switch flipped in his little brain and he suddenly didn't like his medical formula. This same formula that he drank every day, three times a day suddenly was gross. He stopped drinking it. He wasn't getting his needed protein for the day. We couldn't figure out why, but suddenly what used to be simple became a huge struggle. We bribed him to drink it, we begged him to drink it, we added strawberry syrup to it, we added chocolate syrup to it, and we tried five to ten other formulas. Nothing worked. We had to remind him every minute to take a drink. It took three hours to finish one cup of formula, just in time to start the next cup. We struggled for about a year, working with him every day to get him to drink all of the formula that was so important for his growth and development. He gradually, thankfully, learned to love it again.

By that time, we were going to clinic once every couple of months, still doing monthly blood draws, though by now we were doing them from home to save the time it took driving to the hospital. Every time we went to clinic, Owen would cry and cry about getting on the scale and getting his head measured. It was like torture. He was terrified to be there, and there was nothing I could do to soothe him. Each blood draw was just as heart wrenching as the first one - tears and screaming every time. And the insurance? Talk about tears and screaming. I had to call and fight with them every time I needed a new case of formula or tried to get coverage for low protein foods, and every time I ended up in tears.

Owen is now seven years old, and is in first grade. He reads above grade level and excels in math and spelling. He plays baseball and loves camping and fishing. He wakes up early every morning and weighs out his own breakfast. He knows how many grams of cereal he can have, how many grams of formula he needs each day. He knows he cannot have a birthday cupcake brought in from the kids at school, but knows that Mom has lovingly left one of his low protein cupcakes in the nurse's office at school, just for that occasion. Does he occasionally get bummed out because he can't have a donut or ice cream? Sure. Does he let it stop him from asking for a low protein muffin or sorbet? Not for a second. He never uses his food as an excuse for bad behavior or fits. He accepts his diet and is very understanding of the fact that he eats differently because it is good for his brain and development. He now only goes to metabolic clinic every six months with no tears and screaming. He does his own blood levels from home with Mom and Dad once a month, to make sure his blood phenylalanine levels are where they should be and that phenylalanine goal we strive to hit every day is just right. We have somehow managed (I think I cried enough) to get insurance coverage for all of his foods and formula (it's a miracle!). Thanks to the amazing metabolic team, he has had several opportunities to meet other kids and adults with PKU. Last summer he got to attend Camp Knot a Phe, a family camp for individuals with metabolic disorders put on by the PCH Metabolic Team and Arizona Network for PKU and Allied Disorders (ANPAD), and he is so excited to return this summer.

Owen has a younger brother who does not have PKU. He has 7 cousins who do not have PKU. No one at his school has PKU. He understands that PKU is unique. I am so grateful for the Office of Newborn Screening and that Owen's PKU was detected by it. Our lives would be so different if he hadn't had that test. He would not be our amazing, bright, energetic, playful son without the newborn screening.

