Henley's Story

by Jess Johnson

Henley was born May 26, 2017, at 40 weeks and 1 day. About 24 hours after she was born, we noticed her respirations seemed really fast. She was moved to level 2 nursery care and a pediatrician ordered lab work and chest x-rays, which we were told were normal. Henley was given oxygen and doctors told us it was transient tachypnea of the newborn, also known as TTN. Although her rapid respirations hadn't resolved, she was discharged from the hospital four days later.

At home, we noticed Henley seemed very pale and was extremely fussy. She would not rest unless she was being held, which meant I spent nearly every night sleeping in the recliner, holding her.

June 2nd we had our well-baby visit and the doctor noted she had not gained weight. A couple weeks later at a recheck she still wasn't gaining appropriately.

On June 29th she was now in the 4th percentile for weight. Doctors told us to bottle feed her instead of breastfeeding, so we could see how many ounces she was consuming. Henley absolutely refused bottles. Instead, we used a baby scale to record pre and post feeding weights. Henley's pediatrician wanted her consuming at least 18 ounces in a 24-hour period and said that should allow her to gain weight appropriately. Henley's pre and post weights showed she was consuming between 19 and 24 ounces in a 24-hour period. I continued to express concern about her color, rapid respirations, and uncontrollable fussiness. Her doctor prescribed acid reflux medication.

Henley's pediatrician ordered bloodwork the first week of July. Although a number of things were outside the "normal" range, he said he didn't believe there was anything wrong. According to counts she was anemic, had elevated liver enzymes, and her ANC was 0. His only recommendation was to discontinue breast feeding and switch to a bottle. He said we didn't really know how much Henley was consuming and there wasn't anything with her bloodwork that was alarming. I explained that I did know how much she was consuming because I had been doing the pre and post feeding weights and that we had tried the bottle and it just didn't work. All we were doing was burning up valuable calories fighting with her. He suggested a nurse visit again the following week to check her weight.

I was in tears by the end of the conversation and called Kyle. I was certain something was wrong with Henley and that something was being missed. During the conversation Henley was sleeping and I witnessed her drastic color change to grey, although I could see she was breathing. I hung up the phone with Kyle and took a picture of her. This way I could *show* the doctors what I was seeing.

I truly didn't know what to do. I didn't want to see a pediatrician in the same office, because I wanted a fresh set of eyes and ears to see and to listen to everything I was saying and to review the bloodwork. The following week I drove to the next closet pediatrician's office, two hours away. I told her Henley's story and symptoms, start to finish. She listened and reviewed labs and looked at the photo I had taken of Henley's color change. This pediatrician said she was going to hospitalize Henley based on the following:

- Failure to thrive (her minimal weight gain, despite eating frequently and consuming the ounces needed to gain appropriately)
- Pale skin color, sometimes greyish in color
- Rapid respirations
- ANC of zero (Absolute Neutrophil Count)/ Severe Neutropenia
- Elevated liver enzymes
- Low hemoglobin (8.4)
- Extremely fussy at times

The next 72 hours were an emotional roller coaster. We went from thinking we would be told that our daughter maybe needed an iron supplement and was anemic to being hospitalized, undergoing a bone marrow biopsy, and needing a blood transfusion. A week later she was flown by air ambulance to University of MN. Fast forward another 4 weeks and we finally had the genetic diagnosis of Shwachman-Diamond Syndrome.

Since diagnosis, Henley began GCSF due to an ANC that remained at 100 at best. She also began receiving Creon, but continues to struggle to gain weight. Post diagnosis her marrow continued to fail and she required blood transfusions due to anemia. At 5 months old, in October of 2017, we met with transplant teams at the University of MN and also Boston Children's Hospital. Both teams recommended transplant and we began making plans to transplant around Henley's first birthday.

In April 2017, our family traveled over 500 miles one way to Minneapolis, MN to begin chemotherapy and prepare Henley's body for a bone marrow transplant. It was frightening and emotional.

Fast forward to January 2019, and Henley is a thriving, happy, busy toddler. Henley is +256 post-transplant and spends her days trying to keep up with her three-year-old twin sisters. Although the road to where we are today has been a bit bumpy at times, our family is incredibly thankful for the gift of transplant. We will continue to keep her isolated, until her marrow shows through blood work that it has fully recovered. We expect to see that happen in the next 100 days.

Henley, and our journey with her, has been the blessing we didn't know we needed. I realize not everyone who encounters a challenge like this in their life would view it as a blessing, but we have chosen to do everything we can to make good out of every day. This journey has changed our lives forever. We do our best to share Henley's story and encourage people to join the bone marrow registry.