Taylor's Story

Taylor was born on May 16th, 2020 after a perfectly healthy pregnancy! She was our second child so I kind of knew what to expect when we got to labor and delivery. The doctor that delivered Taylor immediately said the words "enlarged spleen" after the APGAR. This caught me off guard, but no one seemed concerned, so I quickly forgot and embraced our baby girl. The following day they wheeled Tay away for an Ultrasound of the liver and spleen, and all were within normal limits so they released us for discharge. Her bilirubin was high, so they adamantly noted we needed to follow up with the pediatrician the next day. We made the appointment asap, and her pediatrician ordered the bilirubin check by the heel stick. We got the call that it was still high, so we had to come in the next day for another heel stick. She gave us orders for a bilirubin blanket. The heel sticks went on for a few more days, until the doctor referred us to VCU Children's Hospital. I got a call from a GI doctor via virtual appointment asking lots of questions, and she sent us for lab tests. We quickly got a call back saying her liver levels were high, platelets were dangerously low, and bilirubin was too high, and they were admitting us to the hospital. Taylor was 6 days old.

Taylor and I were admitted to VCU Children's Hospital for a total of 7 days. Scott wasn't able to visit. At this time they were only allowing one parent and no visitors. This broke my heart for him, and I was trying to be strong for our family. I really didn't realize how serious this was, as no one really explained anything. We had no answers. It was honestly a revolving door to our room. Genetics, GI doctor, Ultrasound techs for abdomen and brain, nurses, Eye Doctor, Infectious Disease doctors, etc – asking me all kinds of questions. "Were you sick during pregnancy?" "Did you travel any?" I didn't know which doctor was from which department. I was exhausted and scared to death that we could get Covid. They did so many blood draws from her little veins, her arms were black and blue from getting bad sticks. Two separate times, the staff had to get an anesthesiologist doctor and his team to use their equipment to get a good stick. I had a lactation consultant helping me nurse, but I was so stressed I had to supplement. They tested her for everything from HIV to certain infections to Covid. During our stay, she had a 3-hour HIDA scan to see if her ducting from the liver was working correctly. We didn't get many answers from that. We were discharged with a surgery date 2 weeks later to see if she had Biliary Atresia and more genetics lab work on the schedule. During surgery, the surgeon found no Biliary Atresia but also performed a liver biopsy. We would have some results in about a week. I was so happy to be home with my family, but it was also such a dark time for us.

Taylor's Genetics team requested additional labs and 2 weeks later we got the call – Taylor has Gaucher Disease, a rare genetic disease (lysosomal storage disorder)*. They gave us all the details about how there's no cure and mentioned DUKE has a great genetics team. This was all surreal. I immediately got on social media and started researching Facebook groups. I found one and the mom immediately told me to contact one in Fairfax – world renown Genetics Dr right in our backyard. She wanted to see us immediately. I was in shock. It was an all-day meeting, and she received an emergency dose of an Enzyme Replacement Therapy infusion intravenously. We didn't know the prognosis, but with her onset symptoms she told us "she may die before her 2nd birthday". She told us Germany makes a medication that could help, similar to a clinical trial, that will slow the brain damage. We would need to find ways to get this medication as she can't prescribe it. Taylor will need a port for weekly infusions for the rest of her life.

Life has been full of ups and downs since then. Currently, Taylor is walking and doing great! Our little 3 year old receives weekly ERT infusions through her port from a home health nurse. She has a cochlear implant on her right ear, due to disease progression. She is speech delayed, but she gets weekly speech therapy and is making progress! She has an eye disorder, which affects her balance. Taylor just had her IEP approved and will be getting Special Education services! Her disease progression has slowed with her oral medication taken every 8 hours and ERT weekly infusions. We see specialists what seems like ALL the time, but we would do anything for this sweet girl. Taylor is very opinionated, loves to cuddle, and is always trying to make people laugh. She is such a happy toddler!!! Her favorite thing is her inclusion ballet class. She isn't promised a long life, but we've already seen miracles happen for the last 3 years. We hope to one day see a cure. I've been so lucky to connect with all of these great moms locally through New Path and the community for support. I want to give this same support back to any family that needs it!

Taylor is diagnosed with Neuronopathic Gaucher Disease, type 3. This disease means she lacks an enzyme to break down fat and affects her organs and bones. Her type affects the brain, which the medication from Germany will help slow down the brain damage. The weekly infusion replaces the enzyme. This disease can cause brain damage, seizures, cognitive problems, and other issues. She can survive into adulthood.