HCU Patient Stories

Colbie

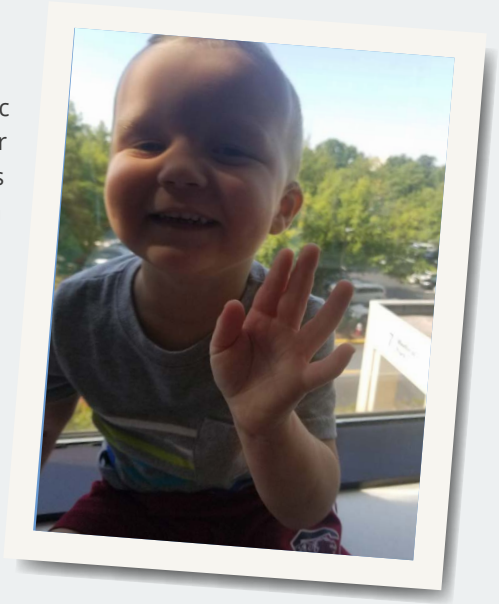
Our daughter, Colbie, suffered a massive blood clot in her brain shortly after her 3rd birthday in June of 2012. She was flown for treatment from Montana to Seattle Children's Hospital, where doctors were confounded as to what may have caused the blood clot and small strokes. After a week's stay and many tests, they still had no idea and chalked up the cause as chance or a freak incident. They allowed us to return home with follow-up appointments scheduled six weeks out. It wasn't until a month later, in July 2012, when we were on the follow-up visit, that the doctors tested her blood for homocysteine. Her results alerted the doctors to the possible cause of the blood clot - her homocysteine levels were so abnormally high that Colbie's blood had become toxic and prone to clot. **We blamed ourselves for not having a second heal prick test done on our kids as part of their newborn screening - the state of Montana does not require a second test as some states do.**

— Sarah and Cole Sullivan, parents to HCU patient Colbie, 14, Montana

Elliott

At two years old, Elliott was diagnosed with Homocystinuria, a rare genetic metabolic disorder. Being missed in his newborn screening test led to a delayed diagnosis for Elliot and resulted in nearly losing him to this treatable disease. He spent 29 days in the ICU with blood clots in the brain, and he had a stroke. Had we known at birth that he had this treatable disorder, we could have avoided a lot of suffering. **I've since learned that his methionine level at birth was 44, and the cutoff in our state is 65.** I urge South Carolina and other states to lower their MET cutoffs to identify all babies with classical HCU at birth.

— Liz Carter, parent to HCU patient Elliott, 7, South Carolina



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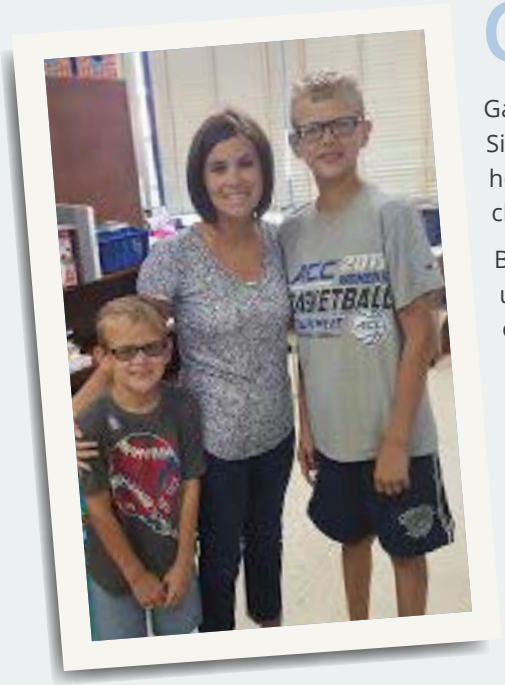

Gavin & Zakary

Gavin and Zakary were not diagnosed with HCU until Gavin was ten and Zakary was 5. Since the boys were diagnosed later in life, we battled constantly with controlling their homocysteine levels due to the difficulty they had with adjusting to the required diet changes and medicines.

Both Gavin and Zak had several side effects from not finding out that they had HCU until they were older. They both had a severe learning disability, and both had to have eye surgery. Zakary had a mild case of scoliosis, and they both had several behavior problems. Sadly, things worsened for Zak on Thursday, October 19th, 2018. Through complications from his late diagnosis, Zak left us to play baseball with the angels in the outfield. **If we had found this on their newborn screening, I believe it would have been much better for both boys in many ways. I pray that they will be able to find a way to catch HCU better on newborn screenings so that it gives children a better chance of life.**

— Amanda, parent to HCU patients Gavin and Zak, North Carolina

Landon

Landon was diagnosed with HCU at age 4. When he was born in 2007, our state had not yet begun testing for Homocystinuria as part of newborn screening. Our journey to diagnosis was very long and hard. Landon was misdiagnosed several times, and from 9 months until age 4, we went through five pediatricians and traveled all over our state to see specialists. Upon examining Landon for the first time, our fifth pediatrician seemed upset. He looked me in the eyes and said, "Your baby will die soon; he is very sick and needs to get out of the state." He sent us to Cincinnati Children's Hospital where, after an initial misdiagnosis, he was correctly diagnosed with Homocystinuria, and we began treatment. Six months after we began diet modifications and medications, Landon's body began to respond. With a lot of OT, PT, and speech therapies, Landon has finally been able to catch up with his peers in most ways.

We almost lost Landon. **The damage to his body and brain was severe and caused a lot of health issues, muscle damage, and developmental delays that could have all been avoided with a simple blood test at birth.** Through our journey, we have learned how to be better advocates for our children, encourage others to advocate for their children, and follow their gut instincts regarding their health and safety.

— Rachel and Justin, parents to HCU patient Landon, 17, West Virginia

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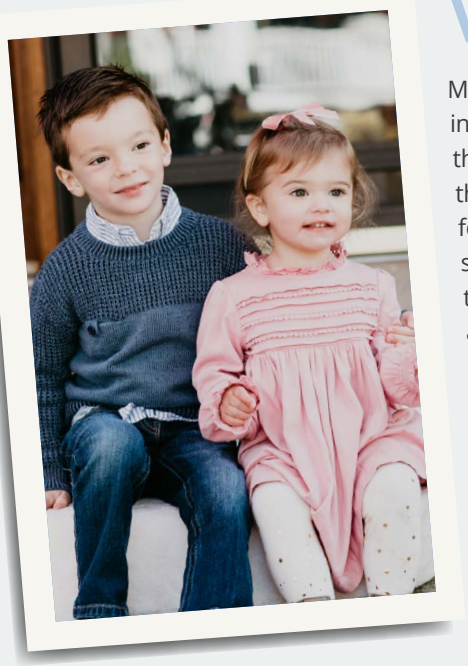

Virgil & Annie

My HCU heroes are, of course, my kids, Virgil and Annie, who were diagnosed with HCU in 2016 and 2019, respectively. When Virgil was born in 2016, the newborn screening at the hospital (or "PKU test") returned one abnormally high result. After about two weeks, the diagnosis was official: Virgil had Homocystinuria, a rare metabolic disease. Fast forward to 2019, when Annie was born. After going through vitals and blood pressure, she checked the computer for blood test results: her homocysteine level was 60, and the typical range was 0-10. EXPLETIVE! Annie was also diagnosed with Homocystinuria. Like any new parents, we freaked out, and when we visited Google MD, we freaked out some more. Most sites describing the effects of Homocystinuria mention a high risk of eye lens dislocation, delayed brain development, and an increased risk of stroke and blood clots. None of the sites contain the caveat, "these risks are close to zero if treated properly through diet." **Our doctors reassured us that, with a special metabolic formula and following a rigid "low protein" diet, he would develop normally and lead a happy, healthy life. Four years later, that has come to pass.**

— Tom and Kristen Hawkins, parents to HCU patients Virgil, 8, and Annie, 5, Illinois

Sarah

I was diagnosed with Classical Homocystinuria through newborn screening. While I am sure that the news must have been scary for my parents, we now know what a huge blessing it was to get a diagnosis at birth. My parents had two children before me - my brother, Jackson (now 21), and my sister, Emily (now 19), so the news that I had a rare disorder came as a total surprise. My mom once told me this story, which she now calls her 'testimony.' She said that she was on a trip with my dad, and she kept having this sudden gut feeling that something was wrong with her pregnancy, so she went and checked in with her doctor when they were back in town. The doctors kept telling her that there was nothing wrong and that, as far as they could tell, I was a super healthy baby. But my mom knew deep down that something just wasn't right. My parents moved to North Carolina from Abingdon, Virginia, during the pregnancy. **To make a long story short, moving to North Carolina was what my mom thinks of as God's intervention because if we had not made that move, my HCU would not have been detected through newborn screening due to the difference in newborn screening between NC and VA.**

— Sarah May, HCU patient, 16, North Carolina

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