



Behind the Screens

Missouri Department of Health and Senior Services
Newborn Screening Program

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Featured Disorder

Mucopolysaccharidosis II (MPS II), also known as Hunter Syndrome, is an inherited disorder. MPS II encompasses a wide spectrum of symptoms at varying levels of severity, but is often characterized by distinctive facial features, a large head, hydrocephalus, enlargement of the liver and spleen (hepatosplenomegaly), umbilical or inguinal hernia, and hearing loss. Additional symptoms include joint deformities and heart valve abnormalities. The most severe types of MPS II can also cause a decline in intellectual function and a more rapid disease progression. The disorder can be managed with enzyme replacement therapy but there is no cure (babysfirsttest.org). Early diagnosis and intervention give babies with rare but serious conditions like MPS II the best chance at a healthy life. Missouri added MPS II to the newborn screening panel in 2018.

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Causes:

MPS II is considered a type of lysosomal storage disorder. The body's connective tissue is made up of molecule chains called mucopolysaccharides. Normally our body is able to break down these chains, dispose of them and replace with new ones. People with MPS II lack the enzyme that helps to break down these chains. Damage can be caused by the progressive 'storage' or buildup of these used materials in the cells (mpssociety.org).

Prevalence:

MPS II is considered a rare condition found in 1 in 100,000 to 1 in 170,000 males (babysfirsttest.org). Females are generally unaffected carriers of this condition.

Treatment:

At this time, there is no cure for MPS II. However, with early detection and proper treatments such as enzyme replacement and physical therapy, symptoms can be managed. Depending on the age of onset, some symptoms can be delayed or prevented. The life expectancy for people with the severe type is 10 to 20 years. Individuals with the less severe type typically live into adulthood and their intelligence may or may not be affected.

To learn more about MPS II visit mpssociety.org or babysfirsttest.org.

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Did You Know?



Patient

Spotlight

Hudson

“We were so very excited to welcome our fourth child to the family in August of [2019], and our three oldest kids were ecstatic to meet their new baby brother. They came to see him the morning after he was born, and by that evening, we were begging to be discharged so we could go home and start our life as a family of six. Adding one more to the group was a seamless transition, and Hudson just happily joined right in! We requested discharge so quickly that we barely had time for visitors, and we had to ensure we actually stayed long enough for him to have his heel prick test completed. Being that he was our fourth child, we never really thought another thing of the newborn screening. I remember anxiously counting down the days until I received my oldest child’s clear screening, and this time, I had forgotten it even existed. We were busy, we were experienced parents, and life was as smooth as it could be with three young children and a newborn baby.

On Saturday morning, August 17, at 11:17 am, I was dozing off for a nap with nine-day-old baby Hudson, and my cell phone rang. I saw that it was our pediatrician’s office and almost let it go to voicemail since I knew his follow up newborn appointment was on Monday. I was certain they were just giving me the general courtesy call to remind us of the upcoming appointment, but for some reason, I drowsily answered the call. Strangely enough, it wasn’t an automated call, the receptionist, or even a nurse on the other end of the line. It was our pediatrician, but that didn’t seem weird to me at the time because I was a sleep-deprived mom of a newborn who was waking up from a rare nap, and I was so excited to touch base about our sweet, new baby boy.

Unfortunately, that excitement was short-lived and quickly replaced with the absolute most confusing and desperate

moments of my life. After our quick greetings, the doctor said she was calling about the newborn screening. Her familiar, friendly and casual tone wasn’t the same as usual, and she sounded much more serious, so she quickly had my attention. As I was realizing this probably wasn’t going to be a general call to let me know everything was clear, the doctor shared that there were some concerning results on Hudson’s screening. We have a friend whose child was diagnosed with PKU via the newborn screen several years ago, and a pit quickly formed in my stomach as my mind flashed to questions and concerns that would encompass learning to care for a child with the very specific and limited diet that accompanies a PKU diagnosis.

Before my imagination could carry me much further into the worry of the PKU world, the pediatrician asked if my husband or I had a family history of Cystic Fibrosis (CF). I quickly mentioned that we didn’t and thought, “Goodness! Of all the things I was terrified about with general obstetric checkups and ‘what about if’ scenarios when I’ve been pregnant with each of our four children, there’s absolutely NO WAY that this is the case in this scenario. I’m so glad to know this is one of those things I don’t even have to worry about, and I’m certain there’s a mix up, and I’m not even worried about this because I know it’s not possible.” I was so certain this wasn’t a possibility that in fact, looking back, I even vividly remember a moment when I saw a CF screening poster in my OB’s office during my pregnancy, and I recall feeling a sense of relief that since CF didn’t run in our families, that would be at least ONE thing I didn’t have to worry about.

I’m not 100% sure what all our doctor said during those first few moments. In fact, I probably don’t even remember a significant majority of it, but I do recall her telling me that

she didn't want to have to tell me over the phone but that she knew I'd rather have the information sooner than later. She didn't know if my husband would have been at Monday's general newborn appointment with us, or if he would have been back at work... or if I would be there with all of our other children as well... so she made the difficult choice to call me. For that, I am forever grateful. She gave us the opportunity to process our darkest moments in the privacy of our home with each other. It gave us the opportunity to truly wrap our minds around the information and come to the realization that this wasn't just a nightmare before we truly jumped into the medical realm that is CF.

I remember telling her there was NO way this was accurate or possible. Those moments are literally frozen in time in my head. I remember exactly what my bedroom looked like, how the blankets were crumpled at the foot of the bed, where Hudson was sleeping, the hand-drawn picture my daughter taped to the wall by my nightstand, and the look on my husband's face as he was worriedly trying to figure out what the conversation was about. I remember mouthing to him that they thought Hudson had CF, and his expression was one of total shock and disbelief. I kept asking the pediatrician if there was any way the screener was wrong. Was it another child's results? How was this possible if it doesn't run in our family? Could CF occur as part of a genetic anomaly and not be hereditary? Could anything else possibly have triggered such a high IRT level to indicate that he might have CF? I kept explaining it away in any way imaginable because it truly did not compute in my mind. He was our FOURTH child, and NONE of the others had CF. We didn't know our children, ourselves or anyone in either of our family to even be CF carriers! The doctor slowly, patiently, carefully and methodically explained over and over again as my brain processed the pieces one at a time. I'm certain she probably repeated herself many times before each of the parts of the story could truly sink in. I remember her explaining that not only were his IRT levels high on the initial test, but they also conducted genetic testing which resulted in two specified gene mutations. She told me about all the reading and research she had done that morning to see if there was any other possible diagnosis and was able to explain

that while it could possibly be CRMS, with his extremely high IRT levels and two genetic markers it would be exceptionally doubtful. Her ability to slowly and repeatedly explain all of those pieces made me

come to the realization that it wasn't an error... and that his sweat test in the coming days would in fact very likely diagnose him with CF.

Knowing this information provided by the newborn genetic testing was a huge initial blow, but it was so beneficial in the long run. Once I heard about his probable diagnosis, I was able to connect some dots of symptoms I had seen and dismissed as typical newborn nuances. Early identification provided us with the gift of early intervention. We were given the opportunity to process what was going on, conduct research and get our son into his pediatrician and then a high quality Cystic Fibrosis Clinic within days. We quickly gained a strong understanding of his disease, and Hudson received his first medications within just a few days of the initial call. Even though I still worried during the weeks between our newborn's diagnosis and the completion of official CF testing for each of our older children, it was also reassuring to know all three had passed the newborn screener for CF that we found had been put into place in the state of Missouri just months before our oldest son was born.

Empowering parents and families with knowledge and resources allows for children to have an advantage when battling genetic disorders that are identified by newborn screenings. We are forever grateful that our son didn't have to battle through the first years of his life with never-ending health issues that we didn't have the answers to and weren't able to address specifically. Additionally, when Hudson was critically ill with respiratory viruses twice during his first winter, we knew exactly what type of obstacles to watch for, and we were more prepared to fight the battle of weeks in the PICU. Had we not known that Cystic Fibrosis was a contributing factor to significantly worsening and complicating any viral respiratory illness, his medical team would not have been on the lookout for some of the issues he had, and his treatments would not have been as tailored to his needs. Knowing he had CF during those grueling weeks very well could have saved his life during that time.

Overall, I can't say enough about the importance of newborn screening and the gratitude we have toward the ability to receive an early diagnosis. While some may feel that hearing that news while holding your nine-day-old baby is crushing, and while we agree that we felt that way at the time, we now feel differently being on the other side of that experience. We have learned that the pain in those early days provided us with the opportunity to address Hudson's diagnosis head on, immediately begin medications and treatments, and begin to wage the battle of adding years to his life due to knowledge and early intervention."

Hudson's mom



COVID-19 Resources

As we all strive to put this pandemic behind us and find our new normal, we also want to stay informed. Visit the resources below to learn more about COVID-19 information and vaccine guidance.

Missouri Department of Health and Senior Services
health.mo.gov

Centers for Disease Control and Prevention
coronavirus.gov

World Health Organization
who.int/health-topics/coronavirus

New and Expecting Parents
expectinghealth.org/index.php/discover/coronavirus-covid-19

Families with Children who are Deaf or Hard of Hearing
handsandvoices.org

Did You KNOW?

Did you know that meconium ileus (MI) is NOT the same as meconium present at birth or during labor?

MI is a bowel obstruction that occurs when the meconium in a baby's intestine is even thicker and stickier than normal meconium, creating a blockage in the ileum. The earliest signs of MI are abdominal distention (a swollen belly), bilious (green) vomit and no passage of meconium stools.

Most infants with MI have a disease called cystic fibrosis, which is one of the disorders detected through newborn screening. Newborns with MI are at risk for false negative newborn screens for cystic fibrosis; therefore, if a baby has MI, it is important to mark the box indicating so in the altered health status portion of the blood spot form. This will flag the specimen and allow the Newborn Screening Laboratory to initiate second tier DNA testing to provide the most accurate screening results possible.

Tech Tips

Delayed identification of children who are deaf or hard-of-hearing, but who passed the newborn hearing screening is a concern. Repeatedly performing the newborn hearing screening increases the likelihood of obtaining a pass outcome by chance alone. Therefore, to avoid false-negative screening results, the Joint Committee on Infant Hearing (JCIH) renewed the following recommendations:

1. No more than two high-quality hospital-based screenings should be performed prior to hospital discharge.
2. Only one high-quality outpatient rescreening should be performed prior to referral to a pediatric audiologist for the child who needs follow-up testing after the outpatient rescreening.

To read all of the JCIH recommendations for a hospital newborn hearing screening program, see the "JCIH Year 2019 Position Statement: Principles and Guidelines for Early Hearing Detection and Intervention Programs" at www.jcih.org.



thank you

for your contribution in ensuring the best possible start for Missouri newborns!



MISSOURI DEPARTMENT OF HEALTH AND SENIOR SERVICES

Bureau of Genetics and Healthy Childhood
Newborn Blood Spot, Hearing, and Critical Congenital Heart Disease Programs
573.751.6266 or 800.877.6246

Missouri State Newborn Screening Laboratory
573.751.2662

www.health.mo.gov/newbornscreening



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