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INSIGHTS

Family reflections: the journey of a CDH family

Cassandra Whitten[1] ✉

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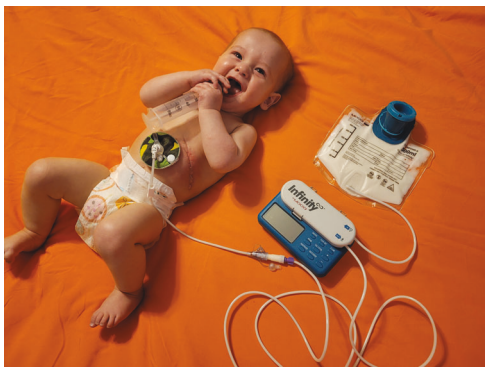
During week 18 of my pregnancy, I had my first anatomy scan. The scan was normal but due to a blood clotting disorder that I have, I was referred to a Maternal-fetal Medicine (MFM) doctor. I had my first appointment with them at 20 weeks and they conducted their own scan, after which we were told that our son's diaphragm was not fully developed and that his stomach and intestines were in the left side of his chest cavity. This condition is known as congenital diaphragmatic hernia (CDH) and is a life-threatening birth defect. Due to this condition and my blood clotting disorder, my pregnancy was considered high risk and required that I be seen more often by my MFM and receive ultrasounds on a regular basis to determine my baby's growth and development. This would provide vital information about what we could expect once he was born. Because his defect was life threatening, it was recommended that I utilize a hospital that had experience with CDH cases and had a level 4 NICU with extra-corporeal membrane oxygenation (ECMO) availability. There was only one hospital in the state that met all the criteria and thankfully, it was only an hour away from our home.

Given the complications of my pregnancy, it was decided that my delivery would take place in the operating room to ensure that the respiratory team would be available as soon as my baby was born. On May 13, 2021, Jeremiah Douglas "JD" Whitten was born via C-section, weighing 8 pounds 15 ounces.



Without us even being able to see him, JD was rushed out and immediately intubated because he was unable to breathe on his own. With the excess organs in his chest cavity, his left lung had not properly developed, his heart was pushed to the right side of his chest, and his right lung was experiencing tremendous pressure, all of which compromised his ability to breathe.

[1]CDH Stars and Angels Leader, Vicksburg, MS, USA. ✉email: info@pedres.org

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Since birth, JD has undergone many surgeries and treatments. While in the NICU, he was resuscitated multiple times, the first being only 36 h after birth. At 4 days old, even with the assistance of the ventilator, he was unable to sustain viable oxygenation levels and underwent his first surgery: he was placed on an ECMO device. In short, this device takes the place of the heart and lung functions. It draws the blood from the body, oxygenates it, and returns it. Without this incredible device, JD would not be with us today. Countless physicians saw him every day, adjusting multiple settings in order to sustain his life and allow his body to rest, heal, and strengthen. His battle for life had only just begun.

Day 10 brought yet another surgery. A team of surgeons opened his chest cavity, removed the organs from his chest, and repaired the hole in his diaphragm that had allowed them to get there in the first place. Due to the high-quality sonograms I had while he was in utero, we knew that his stomach and intestines were in his chest cavity, but it was not until now that the doctors discovered that his spleen, colon, and part of his liver were also there. The day following his repair surgery, JD's kidneys began to fail. Fortunately, this did not require another surgery as the doctors were able to add a dialysis machine into the ECMO circuit. At 2 weeks old, the physicians believed that his lungs and heart were strong enough to take over their functions without the need of ECMO and dialysis; JD proved that they were right.

Over the course of his 67-day NICU stay, JD was intubated for 42 days. His tube required suctioning multiple times a day. He had multiple ultrasounds, countless x-rays, a bronchoscope, and was on a variety of medications, including paralytics, sedatives, and pain management drugs. He also required medication for reflux, pulmonary hypertension and several other issues; he is still on some of these medications today.

He has undergone seven hospitalizations, nine different operations, and has been deemed disabled by the Social Security Administration. The treatments he has received over the last 17 months have proven successful; however, they do not come without the occasional failure or side effect. One such example is that his diaphragm can reopen, allowing his organs to again enter his chest cavity; this happened when he was 7 months old and could happen again. Due to his extensive medical history and disability diagnosis, he now attends a pediatric nursing facility daily instead of a typical daycare and is still followed closely by multiple doctors.

As a parent of a CDH baby, one of the most common questions that I get is: "why does this happen"? This question has never really been scientifically answered but there are generally two theories of thought: a genetic anomaly; or a simple mishap during development. My husband, I and JD have all had genetic testing done to see if there is a common factor that may help determine if his defect was genetic. We were told that there was no genetic explanation for his defect. While our case was determined not to be caused by a genetic issue, several studies have concluded that genetics may be a likely factor; the DHREAMS study being one of them.

Determining the cause is very important to us as JD is my first live birth of three pregnancies. Because of this and JD's condition, we are extremely nervous about trying to have another child. I believe more research is needed in order to provide better insight to those parents who have already faced this diagnosis but still want to grow their family. This insight would enable them to make a better-informed decision and could give them the confidence to realize their dreams.

I would also encourage research into the side effects of the treatments for CDH and how to mitigate them. Because he was on ECMO, JD must now be monitored for the next few years for possible hearing loss, esophageal muscle paralysis, impaired speech and more. Due to his long intubation period, he did not learn the "suck-swallow-breathe" process that is so important for infants and is now 100% dependent on a feeding tube. His 2-month stay in the NICU meant that he had developmental delays in all areas. This has required him to receive speech, feeding, occupational, and physical therapy. While the hospital advised us that side effects were possible, no one could predict the extent or severity of them. More research could help minimize or eliminate them.

Lastly, I believe we could do more to help diminish the impediments to the bonding process between parents and a child while in the NICU. Many times, being able to hold your child is not possible until weeks, or even months after they are born. Occasionally these babies are under the influence of medication, so even eye contact (and the release of serotonin) is not possible. These factors, among others, make some parents feel as if they are not able to connect with their child or fear that their bond will not be as strong.

As a scientist myself, I understand that we may never solve all of the problems associated with child birth and development, but I do believe that with dedicated research and appropriate funding, we can certainly do more. Personally, I cannot thank the research community enough for all of the work that has already been accomplished. Without this research and the discoveries that have already been made, I know that my son would likely not have made it. I genuinely appreciate their work more than words can express. Thank you!

COMPETING INTERESTS

The author declares no competing interests.

ADDITIONAL INFORMATION

Correspondence and requests for materials should be addressed to Cassandra Whitten.