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INSIGHTS

Family reflections: our journey with Down syndrome coupled with a congenital heart defect

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I was 46 years old and pregnant with our sixth child and going in for the first ultrasound of the baby. This was my fifth child after the age of 35 and I was not particularly concerned as long as there was a heartbeat. There was a heartbeat, and despite the ultrasound tech's somewhat cold demeanor and no doctor available to speak to me after the appointment, I thought nothing of it. Later that evening, I received a call from the OB/GYN who had been unable to meet with me earlier that day. He informed me that the ultrasound tech had concerns because of the nuchal fold measurements that were obtained during the ultrasound and that they were referring me to the perinatologist. The whirlwind had begun.

Subsequent ultrasounds indicated that the baby, a little girl, appeared to be at risk for Trisomy-21 (Down syndrome) along with a host of other concerns including fluid around the heart and other details that are now blurred in my memory. I do remember the doctor commenting that things did not look good and that we would "see if the baby was still alive" with the 16-week ultrasound. That hurt to hear. But the baby was indeed alive and thriving at 16 weeks and many of the concerning items seen in the previous ultrasounds had simply disappeared. Despite the high risk that the baby had Down syndrome (maternal blood tests came back with a 9 out of 10 chance that the baby had Down syndrome), we thought that we were in the clear. We could handle Down syndrome. But at 22 weeks, the heart defect was revealed (a large atrial septal defect (ASD) and multiple smaller ventricular septal defects (VSDs) as well as a valve defect that I never completely conceptually understood) and a whole new world of worry opened up for us.

Fast forward to the 38th week of my pregnancy, Bennett Grace Alexander was born weighing 8 lb, 8 oz. My prayers were answered in that she nursed like a champ and did not need to spend any time in the NICU. We were home in 4 days.

Echocardiograms again confirmed the heart defects seen in utero that would be closely monitored. We were not on any oxygen or medications. Compared to so many other babies who are born with Down syndrome and congenital heart defects (CHD), her first few months of life were relatively simple, peaceful, and easy.

Bennett battled some jaundice as a newborn, like five out of six of my children, but overall, continued to do well and meet milestones: rolling over at 8 weeks, sitting by 7.5 months, etc. At the same time, there were signs of heart failure that I am not sure I was willing to admit/accept until now, looking back on things. Bennett was born in January, so it was cold and her hands were always like ice. The doctor even dismissed it saying, "some kids have cold hands and feet." But now, after her surgery, I know that this was a sign of her heart failure. Head sweats were common, as was vomiting. These could be attributed to the temperature of the room and overfeeding, but in reality, they were signs of heart failure. And she was ALWAYS tired and ready to sleep.

At 8 months, her echocardiogram revealed signs of increasing pulmonary hypertension. In a 24-h period, Bennett was put on oxygen at night, and sidenafil to treat the pulmonary hypertension. Her surgery was scheduled and it was only a few weeks away. We had to physically and mentally prepare for her surgery and we had to do it quickly. I must confess, I remained largely uneducated about Bennett's condition and the details of what exactly was going to be done in the operating room during her surgery. I could not handle the details, I just wanted her to survive and her heart to be fixed. I could not wait to have this all behind us.

Her surgery was successful in closing the ASD and all but two very small VSDs (they eventually closed on their own). Her valve defect was repaired, leaving only mild regurgitation in her newly formed mitral valve. When Bennett left the hospital, she required oxygen at night and was still on sildenafil and lasix. She quickly got off of the lasix and her pulmonary hypertension soon resolved and she now no longer requires the oxygen or sildenafil.

We were out of the hospital in only 5 days, but hands down, that was the hardest thing that I have ever been through in my life. (I can't imagine what it was like for our little heart warrior.) Bennett's mitral valve regurgitation is now categorized as "mild to moderate"

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and is being closely monitored. She is on enalapril in order to lower her blood pressure and reduce the load on the leaky mitral valve. We visit our pediatric cardiologist every few months. But Bennett is growing and thriving and learning how to walk and talk, doing all the toddler things that she should be doing.

The biggest issue that Bennett faces continues to be the leakage of her mitral valve, which seems to be a common lingering issue for repairs of this sort. There is a delicate balance between suturing the cleft in order to close the leak and suturing it too much resulting in stenosis or an undesired stiffness in the valve leaflets. Bennett's cardiologist has told me that there is a very good chance that she will need to have this valve repaired again at some point in her life. The thought of her going through open heart surgery again takes my breath away. But we have time. There is time for the medical science involved to evolve and the procedures to improve. That being said, I would love to see research being directed to find other ways to make these repairs, utilizing less invasive procedures that are safer and avoid the need to open up the chest (ex. catheterization procedures).

For those of you who are doing research in this area, I simply want to thank you. Your contributions have saved my daughter's life, and many others along with it. What can be done in the realm of pediatric heart surgery is amazing, and I encourage researchers to keep foraging forward. Keep improving the outcome of these procedures, keep reducing the invasiveness, keep lessening the risk, keep learning how to save these little lives.

COMPETING INTERESTS

The author declares no competing interests.

ADDITIONAL INFORMATION

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