

HEART TO HEART

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At the center of the human body is the heart. Not long after conception, we can see the heart through ultrasound as it pumps blood, carrying oxygen to the rest of the body. Sometimes, there is a defect in the heart that alters its ability to perform this job of circulating blood. A congenital heart defect may affect how blood flows through the heart or the blood vessels near the heart.

Children with CdLS are at a higher risk for having a congenital heart defect than children with typical development. For parents, hearing that their child has a heart defect can be devastating. Fortunately, there are treatments to correct many of these problems.

Ultrasound often plays a key role in first identifying a congenital heart defect. This helps both doctors and families prepare for and treat the presenting condition. Following, Karen describes her daughter Emma's heart condition diagnosed in utero:

“We knew while Emma was in utero that there was something wrong with her heart. We could see it on the ultrasound. We were told that there were four things wrong with her heart.

A Tetralogy of Fallot (TOF) is a cardiac anomaly that includes:

- 1) ventricular septal defect or VSD, 2) right ventricular hypertrophy,
- 3) moderate pulmonary stenosis, 4) overriding aorta.

The doctors preferred that any candidate for surgery be at least five pounds. We knew Emma was not going to be the required weight when she was born but the surgery for a TOF had to be done right away. We decided to have Emma at Jackson Memorial Hospital in Miami, FL. They were the best equipped to do the surgery.

The sun was shining that day and Emma was born with a VSD (a hole in the wall between the left and right ventricle of the heart), a right aortic arch and a pulmonary stenosis – no TOF. This was great news. The VSD was eight millimeters. She would need surgery for the VSD and the stenosis, but the surgery could wait until she was stronger. She had regular ultrasounds and echocardiograms. Emma left the hospital on July 20 at 4 lbs., 8 oz. She was on two medications for her heart; digoxin and lasix.

We feel Emma's feisty personality is part of the reason she is doing so well. We have to see her doctor on a regular basis and we monitor her closely. She has taught all of us a few lessons on how to prove the odds



wrong. Keep it up Emma!”

Many children are diagnosed with a heart condition at birth. Following, Molly (mother of Alex) discusses his early diagnosis:

“Alex was born at 38 weeks gestation, at 4 lbs., 5 oz. He was immediately transferred to a neonatal intensive care unit and examined by many specialists, including a cardiologist. He was diagnosed as having Tetralogy of Fallot. We were followed by the cardiologist for the first months of his life, with special instruction to watch for “blue” spells, which indicate a lack of oxygen.

We were very fortunate that Alex never had a blue spell. The doctors kept saying that the heart repair would be scheduled when he showed distress, and probably when he grew bigger.

As we know now, and as we were learning at the time, Alex was not going to get bigger for a very

long time. I remember he only weighed 12 pounds at one year of age.

Alex was scheduled for surgery when he was 22 months. Before surgery, Alex could not sit by himself and could only do a commando crawl. After surgery he quickly learned to sit and crawl by himself, as his energy level definitely increased.

Alex continues to be followed by a cardiologist every two years. It has been suggested that there may be more surgery in the future as he grows, but so far he has managed to do well. There remains a very loud murmur (leakage) that can even be heard with an ear to his chest. The doctors have assured us this can be typical with some heart repairs. Alex remains small – 63 pounds at 17 years old. He has no restrictions on his activities. There would be no way to keep this young man with CdLS down anyway.”

Although many heart conditions are diagnosed at birth, they can also be detected when a child is older and exhibiting symptoms. For Stephanie, her diagnosis didn't come until her teenage years and was quite a surprise to her family. Following, Shirley reflects on her daughter Stephanie's late diagnosis:

“Our daughter Stephanie, who is 17 years old, was recently diagnosed with an atrial septal defect (ASD), or hole in the heart. This came as quite a surprise to us since this had never been mentioned with all the other testing and surgeries she has had. Stephanie has many of the classic conditions of CdLS, even though she was not diagnosed until she was three years old. When I was pregnant with Stephanie, I had several sonograms to monitor her small size, but there was never any mention of





an ASD during that time. However, Stephanie's cardiologist said it is something that she was born with, but it started out small. Over time, Stephanie developed Type II diabetes and started taking medication to help her body be less resistant to the insulin. With the help of a dietitian and medication, we thought we were on our way to a healthier Stephanie. However, her triglycerides were not under control. The ASD was diagnosed when we went to a cardiologist at the Lipid Clinic at the Hershey Medical Center. As part of the exam, an EKG and echocardiogram were done. That is when the ASD was seen. On the echocardiogram, the cardiologist said it looked to be about 14 millimeters, which is approximately a half-inch sized hole. The right side of her heart was enlarged. Prior to knowing about the ASD, Stephanie was getting more lethargic and was not participating in the activities she had over the years. She used to do swimming, basketball and bowling with the Special Olympics. Even doing things and going places with family were getting more difficult for her.

When her metabolic disorder was diagnosed about three years ago we thought that was what was making her feel bad and not have energy. But it just seemed that she wasn't getting her energy back after working with the nutritionist and taking the medication. A heart condition just didn't enter my mind. I guess I thought if she had a heart condition it would have been caught while she was a baby and never thought she had a problem with her heart. I think we were so caught up in Stephanie's severe reflux and her seizures we just didn't think there was anything else.

Further testing to determine the exact size and shape of the hole were done a week later to reveal that it could be repaired by a device. The device, called an Amplatzer Occluder, was inserted through a catheter in the groin area up to the heart and then placed in the hole to repair it. The device is made of a soft mesh material that is inserted in the hole and then opens on each side of the hole to close it off. Over time the cells/tissue in her heart will grow over the device and it will permanently close the hole. This procedure sounded incredible to us and we were (and still are) amazed by modern technology.

She had the procedure done on May 15, 2008, stayed overnight in the hospital and was discharged the next day. The doctor said within four days she could return to her normal activities, with no limitations, and would need to take one aspirin every day for the next six months to help prevent any clotting. She had a month follow up and will have another one in two months, and then at six months. At the follow-up visit another echocardiogram was done and it showed the device in place and doing well. The other exciting news was that the enlargement on the right side



of her heart was back to normal. This was something they thought would take nine to twelve months to return back to normal.

This procedure has been just amazing and Stephanie is doing really well. She does have more energy and stamina than she did. It is noticeable! I just wish we would have known about this sooner. However, now I am a strong advocate for all babies or older children and adults that are diagnosed with CdLS to have an echocardiogram as soon as they are diagnosed so that it can be corrected as soon as possible. I am just so thankful that Stephanie's ASD was found and corrected so now she can start feeling better and get back to her activities."

The heart symbolizes so much more than the physical. If we think of descriptions we use – heart- felt, warm heart, kind heart, heart of gold – the heart signifies the depth of our being. We offer our heart-felt thanks to the families who shared their stories so other families and caregivers can provide better care to their loved-ones with CdLS.

