2015 Annual Report for New Mexico's Newborn Screening (NBS) Program

Sawyer-A Family's Story

Inside this Edition

There are many things people warn you about when you announce that you are expecting. You will get more unsolicited advice about sleepless nights and changing diapers than you ever expected to. You will often say, "I don't care what it is, as long as it's healthy!" to anyone asking if you have a preference on the gender. But what is never talked about is the possibility that the baby isn't 'healthy' or how to handle a roller coaster of a new medical diagnosis with your 6-day old baby. That's where our story starts.

My husband and I were a young, healthy, newly married couple. When we found out we were expecting again, we were overjoyed because we had lost our first pregnancy to a miscarriage. As things progressed, we became more and more excited about the thought of bringing a new baby into our lives. Although I suffered from hyperemesis, the pregnancy was normal. Our daughter looked perfect on every ultrasound, was growing well, and developing as she should. At 32 weeks I went into preterm labor, not uncommon for a woman in her early 20's that is pregnant. We were unable to stop contractions but I was put on medications to stop me from progressing, was put on modified bed rest and continued to carry our sweet baby until 38 weeks when I was induced for Pregnancy Induced Hypertension, also not uncommon for a pregnant woman my age.

Sawyer was delivered by C-section on December 12th. She was absolutely perfect. I ended up having severe complications so Sawyer was taken to the nursery to be cared for while I recovered. In the nursery, they gave her formula, which becomes a very important part of our story. The rest of the hospital stay was uneventful and we returned home after 72

hours with our beautiful baby girl. One day when Sawyer was 6 days old, we were resting on the couch around 7 pm. All of a sudden the hospital phone number came up on my phone. I answered assuming it was a reminder call for our follow up appointments that week and was shocked to hear our Pediatrician on the line. She informed us that Sawyer's Newborn Screening had come back abnormal and that we needed to come in immediately the next morning to have blood work redrawn. We were told to feed her every 2 hours around the clock, no breast milk, and to come in to the ER if she was hard to wake up. We were shocked. We had been given no disease name except for that it was a Fatty Acid Oxidation Disorder. We were told the chances of Sawyer actually having the disorder was very rare and that these are flagged quite frequently as the parameters on the state test are set very low to reduce the chances of a child being missed. We returned the next day for blood work, were told to follow the precautions and to return if we had any other concerns.

At 3 weeks old, Sawyer became ill with a respiratory virus so we were admitted to the hospital for observation. On January 9th, a group of physicians entered our hospital room to inform us that they had received the blood work back that had been done 2 weeks prior. It was confirmed that Sawyer had Very Long Chain Acyl-CoA Dehydrogenase Deficiency (VLCADD). We started testing the next morning to look for heart and liver damage associated with the disorder and thankfully Sawyer had none. We met with Genetic Specialists, Metabolic Physicians and a Dietician to get Sawyer switched over to a specialty formula and to start our life with this diagnosis. We then learned that babies with her disorder that are breastfed within the first few days of life often have severe complications, including death. The formula Sawyer received while I was recovering probably saved her life.

Life after diagnosis was a blur; Sawyer started losing weight and was refusing to eat well. At 4 months of age, Sawyer was admitted for refusal to eat and received her first Nasogastric (NG) tube as an attempt to give her a break from eating and to help her to gain weight. She was officially diagnosed Failure to Thrive at 4 months old and we were devastated. We fought with the NG tube for 2 months and after multiple admissions for eating concerns, we decided to have a Gastrostomy tube (G-Tube) and a Port-a-Cath placed. The G-tube was our attempt to slow admissions down while giving Sawyer the nutrition she needed to keep her VLCADD stable. The port was for intravenous (IV) access during admissions and for frequent blood work as most of her veins had been destroyed by the thick IV fluids.

The admissions continued every 2-6 weeks for frequent vomiting and diarrhea. We had every test done to try and determine the cause. It was found that Sawyer had Bradygastria, or slow movement in the stomach causing the vomiting. We were able to start her on medications and a partially blended diet to stop the vomiting. We then found a clinical trial that was accepting new patients with VLCADD to test a new treatment. Due to Sawyer's history, we were accepted into the trial when Sawyer was 9 months old and we have never looked back.

The admissions finally started to slow down as Sawyer's vomiting stopped until March 2014 when Sawyer had a Gastrointestinal bleed of unknown origin. The hospital ran every test under the sun and were unable to find the cause. We made a decision to move to Houston, Texas to get a second opinion at Texas Children's Hospital. We relocated in April 2014 where they found that Sawyer had a second rare diagnosis called Mastocytosis that had been causing the frequent fevers, diarrhea and ultimately the Gastrointestinal bleed. Sawyer was placed on medications to regulate both disorders and the admissions have almost completely stopped. Sawyer was admitted 11 times her first year of life. In 2015 we only had 2!

Sawyer is now 2 years old and doing amazingly well. Developmentally she is right where she should be with only a little bit of muscle weakness. She has mild heart enlargement and a slightly enlarged liver, but everything is functioning as it should and there has been no worsening damage in 6 months! She is still receiving complete nutrition from her g-tube and we plan to have her port removed by the end of this year!

We weren't expecting this life, but we wouldn't change it for the world. Sawyer is a blessing to our life and we are so incredibly thankful for her. I owe her life to the Newborn Screening. Before these disorders were placed on the Newborn Screening, most babies were dying before diagnosis. Because of this screening and early intervention, Sawyer is living and thriving.

If you are a parent of a child with a rare disorder, please know you are not alone. There are many resources available for you to get plugged in to the support you need. If you would like to follow Sawyer's daily life, you can find us at www.facebook.com/uniquelysawyer where we update frequently on Sawyer's progress with her disorders.

We would like to thank all of our amazing care teams who have taken care of Sawyer from the Nurse Assistants, Nurses, Physicians, Dieticians, Physical Therapists, Speech Pathologists, Laboratory Technicians and Pharmacists. Although you may not see that you have influenced our lives, we would not be here without you.

-Michael, Brittany and Sawyer

Why Screen Babies?

All babies are screened, even if they look healthy, because some medical conditions cannot be seen by just looking at the baby. It is only with time that some conditions discovered by newborn screening may affect your baby's brain, physical development, social skills, school performance, or cause other medical problems. By then the damage may be permanent. Finding these conditions soon after birth and getting treatment early can help prevent some serious problems (such as brain damage, organ damage, and even death) and promote normal growth and development of your child. Since treating your newborn soon after birth is so vital to their health, it is important that your doctor have a current phone number or address so they can reach you if your child needs follow-up care. If they do contact you, make sure to follow their instructions promptly in order to increase your child's chances of living a long, healthy life.

When and How is Newborn Screening Done?

Newborn Screening Blood Spot Test (For Endocrine and Inheritable Conditions)

A health professional will take a few drops of blood from the baby's heel. New Mexico requires two blood spots samples. The first blood sample is collected within 24-48 hours of birth and a second blood sample is collected at 10-14 days after your child's birth. The blood sample is sent to a newborn screening lab for testing.

Otoacoustic Emissions or Auditory Brainstem Response Test (For Hearing Loss)

Hearing screening is easy and not painful. The screen required by New Mexico plays soft sounds and your baby's responses to those sounds are measured. All babies should be screened for hearing loss no later than 1 month of age. It is best if they are screened before leaving the hospital after birth.

Pulse Oximetry Test (For Heart Defects)

Pulse oximetry is fast and easy test that does not hurt your baby. A small sensor is placed on a baby's right hand and one foot to measure the oxygen level in their blood. Pulse Oximetry should be done prior to leaving the hospital.

What is Newborn Screening Program?

The Newborn Screening program ensures that all babies with an abnormal screening results receives appropriate diagnostic testing and that confirmed cases receive early and timely care. The newborn screening program at the state level does this by coordinating information and medical care amongst doctors, nurses, laboratory technicians, social workers, and medical specialists. The program also works to keep costs affordable, reduces health disparities, and provides equal access to care and assistance.

One of the main purposes of the Newborn Screening Program is to educate parents and healthcare providers about newborn screening. New Mexico state law requires that all infants born either at birthing facility or with a midwife receive a newborn screen, NMSA §24-1-6 (2014) and NMSA §24-1-6.1 (2001). Parents can refuse screening by sending a completed refusal form to the state's newborn screening program.

New Mexico does not do research with blood spot samples. All samples are destroyed within a year of the child's birth.

Screened Conditions

Amino Acid Disorders

Argininemia (ARG); Argininosuccinic aciduria (ASA); Citrullinemia, type I (CIT); Citrullinemia, type II (CIT II)Phenylketonuria (PKU);Homocystinuria (HCY); Hypermethioninemia (MET);Tyrosinemia, type I (TYR I);Tyrosinemia, type II (TYR II); Maple syrup urine disease (MSUD);

Endocrine Disorders

Congenital adrenal hyperplasia (CAH); Primary congenital hypothyroidism (CH)

Hemoglobin Disorders

Hemoglobinopathies (Var Hb); S, Beta-thalassemia (Hb S/ßTh); S, C disease (Hb S/C); Sickle cell anemia (Hb SS)

Fatty Acid Oxidation Disorders

Carnitine acylcarnitine translocase deficiency (CACT);
Carnitine palmitoyltransferase I deficiency (CPT-IA),
Carnitine palmitoyltransferase type II deficiency (CPT-II);
Carnitine uptake defect (CUD); Glutaric acidemia, type II (GA-2);
Long-chain L-3 hydroxyacyl-CoA dehydrogenase deficiency (LCHAD);
Medium-chain acyl-CoA dehydrogenase deficiency (MCAD);
Trifunctional protein deficiency (TFP);
Short-chain acyl-CoA dehydrogenase deficiency (SCAD);
Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD)

Organic Acid Conditions

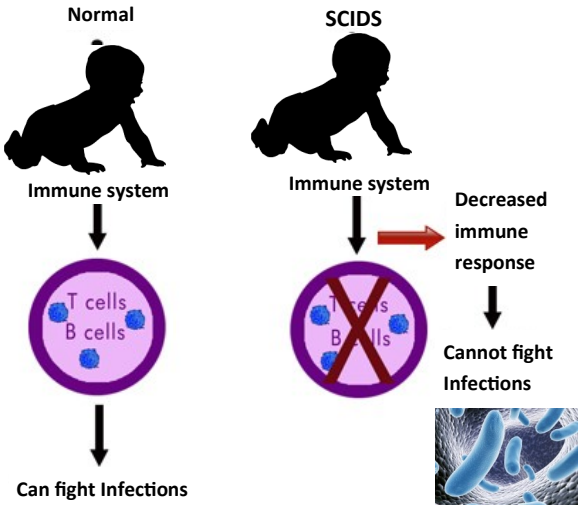
2-Methyl-3-hydroxybutyric academia (2M3HBA); Glutaric acidemia type I (GA1);
2-Methylbutyrylglycinuria (2MBG); 3-Hydroxy-3-methylglutaric aciduria (HMG);
3-Methylcrotonyl-CoA carboxylase deficiency (3-MCC); Isovaleric acidemia (IVA);
3-Methylglutaconic aciduria (3MGA); Beta-ketothiolase deficiency (BKT);
Holocarboxylase synthetase deficiency (MCD); Isobutyrylglycinuria (IBG);
Malonic acidemia (MAL); Methylmalonic acidemia (cobalamin disorders) (Cbl A,B);
Methylmalonic academia (methymalonyl-CoA mutase deficiency) (MUT);
Methylmalonic acidemia with homocystinuria (Cbl C, D, F);
Propionic acidemia (PROP)

Other Disorders

Biotinidase deficiency (BIOT); Galactosemia (GALT); Cystic fibrosis (CF);
Severe combined immunodeficiency (SCID); Hearing Deficiency;
Critical Congenital Heart Disease (CCHD)—Hypoplastic left heart syndrome,
Tetralogy of Fallot, Pulmonary atresia (with intact septum), Total anomalous
pulmonary venous return, Tricuspid atresia, Truncus arteriosus,
Transposition of the great arteries

Conditions added to NBS Panel-SCIDS

T cell receptor excision circle (TREC) screening was added to New Mexico's Newborn Screening Genetic Panel in the fall of 2015. Two additional blood spots are collected along with blood spots already submitted for New Mexico's Newborn Screening Panel. Blood spots are sent to the Oregon Public Health Laboratory for evaluation. Abnormal results (low TREC) are first reported to New Mexico's Newborn Screening program which then alerts your baby's doctor and the Pediatric Immunology Division at the University of New Mexico's (UNM) Health Sciences Center. TREC screening identifies babies at risk for developing Severe Combined Immune Deficiency Syndrome (SCIDS). SCIDS is an inherited deficiency in both the cellular and humoral components of the immune system which if not treated early with enzyme replacement and/or stem cell transplantation will result in the death of your baby. Although SCIDS is a rare condition in the general American population, SCIDS has a higher occurrence in New Mexico. TREC may also identify low T cells counts not caused by SCIDS but caused by other conditions.



Since January of 2015, seven newborns with abnormal TREC results have been identified by the Newborn Screening Program and reported to the Pediatric Immunology Division at UNM. The majority of infants detected as having abnormal TREC levels but who did not have SCIDS occurred early in the year prior to the re-adjustment of the low end of the SCIDS Screening protocol. Babies diagnosed with SCIDS have successfully been treated with stem cell transplantation.

-Rebecca Vaughan, CNP at UNM Pediatric Specialty Clinic

Conditions added to NBS Panel-CCHD

Congenital heart defects are a commonly occurring birth defect that affect 8 in 1,000 live births. Some of these heart defects have minimal symptoms in childhood and are detected during routine well child care when a heart murmur is heard. However, 2-3 in 1,000 newborns have a 'critical' congenital heart defects (CCHD) and cardiac surgery is required within the first year of life. These defects are life threatening without advance cardiology care.

A newborn baby with a critical congenital heart defect can appear normal in the first days of life, and may be discharged from the hospital before the heart defect is recognized. Newborn pulse oximetry screening detects a problem at 24 hours of age so confirmatory cardiac ultrasound can be done.

Pulse oximetry is a simple method of detecting oxygen in the blood with a small light placed on the hand and foot. Oxygenated blood transmits the light at a wavelength that is detected by the sensor. Newborns with critical congenital heart defects can have lower levels of oxygen in the blood but not enough to appear 'blue' on their physical examination.

In 2014, New Mexico and other states mandated that all birthing facilities perform pulse oximetry screening before babies are discharged home. Early, lifesaving, cardiology care is given to babies that have a critical congenital heart defects. With early cardiac surgery, the majority of the children have a good quality of life, similar to their peers.

-M. Beth Goens, MD at UNM's Pediatric Specialty Cardiology

Is There Support for Families?

One of the concerns many families have when they first learn that their baby has one of the disorders detected by the Newborn Screening Program is the possibility of increased health care costs. Fortunately, the state of New Mexico has various policies and programs in place that can assist families in obtaining the best care for their child.

Under the Affordable Care Act, both public and private health insurers (which does not include grandfathered plans) are required to fully cover the costs of diagnosing your baby with a metabolic or genetic disorder that is listed on Secretary's Advisory Committee on Heritable Diseases in Newborns and Children. Most health insurers will cover treatment (including clinical services, medical prescription drugs, nutritional management and medical foods), although durational limits, caps, deductibles, coinsurance and copayments may apply. If your baby is identified as having one of the disorders detected by newborn screening, the Affordable Care Act makes it illegal for your health insurer to deny health coverage to your baby. For families that have difficulty paying for newborn screening services (including treatment) or who do not have health insurance, please contact the Newborn Screening Program for additional resources.

The New Mexico Newborn Screening Program is housed within the Children's Medical Services (CMS), another program in the Department of Health. Families who have a baby identified by newborn screening can receive care coordination from a CMS Social Worker to help them obtain the care they need for their baby and identify other community resources. CMS sponsors community-based pediatric specialty clinics around the state for metabolic, genetic, endocrine, and cardiology conditions. CMS also partners with family support groups such as Parents Reaching Out (PRO), Education for Parents of Indian Children with Special Needs (EPICS), Hands and Voices, Community Outreach Program for the Deaf (COPD), and the Sickle Cell Council of New Mexico in addition to other national parent support groups such as the National PKU Alliance. For further information, see their website at http://nmhealth.org/about/phd/fhb/cms/cyshcn/