The Community Health Clinic

A NEWSLETTER FOR THE CHC

SEPTEMBER 2023

Special Newborn Screening Edition

IN THIS ISSUE:

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60 YEARS OF NEWBORN SCREENING

In celebration of Newborn Screening Awareness Month, The Community Health Clinic is sharing family experiences, the history of newborn screening, as well as information from Indiana Department of Health, Genomics & Newborn Screening Program and Expecting Health Navigate Newborn Screening Program. We are delighted at the opportunity to reach our community with the many important reasons why newborn screening is such an important part of every newborn's life.

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History of Newborn Screening

In 1960, Dr. Robert Guthrie developed a test that would screen newborn babies for a single disorder, phenylketonuria (PKU). This disorder, if not detected early and treated, left children mentally handicap and disabled. Dr. Guthrie knew all too well what the outcome could be because his niece had PKU; she was not detected as a newborn, but at age 13 months when she was already delayed. Dr. Guthrie wanted all newborns to have this test to determine if they too could be affected with PKU. It was a simple prick of the heel to collect five blood spots on a special filter paper. That blood was then sent to the lab where detection of elevated phenylalanine could be discovered, and early diet intervention could begin for those newborns, changing the outcomes for their life. Over the years, more screening methods have been developed using this same process of five blood spots and thus the development of and expansion of newborn screening. Newborn screening detects "hidden" conditions that may not be detected until serious and sometimes fatal symptoms occur.

The promise of newborn screening has expanded to most states, screening for 35 of the 36 conditions recommended by the federal government. The recommended uniform screening panel (RUSP) consists of the following conditions: **Indiana screens for 35 of the 36 disorders on the RUSP.**

Inborn Errors of Metabolism

- Organic Acid:
  - 3-Methylcrotonyl-CoA carboxylase deficiency (3MCC)
  - Methylmalonic acidemia (methylmalonyl-CoA mutase)
  - Isovaleric acidemia (IVA)
  - Holocarboxylase synthase deficiency
  - B-Ketothiolase deficiency
  - Propionic acidemia
  - Methylmalonic acidemia (Cobalamin disorders)
  - Glutaric Acidemia Type 1
  - 3-hydroxy-3-methyglutaric aciduria (HMG)
- Fatty Acid Oxidation Disorders:
  - Carnitine uptake defect/carnitine transport defect
  - Medium chain Acyl-CoA Dehydrogenase deficiency (MCADD)
  - Very Long chain Acyl-CoA Dehydrogenase deficiency (VLCADD)
  - Long chain L-3 hydroxyacyl-CoA dehydrogenase deficiency
  - Trifunctional Protein deficiency
- Amino Acid Disorders:
  - Argininosuccinic Aciduria
  - Citrullinemia, Type 1
  - Maple Syrup Urine Disease (MSUD)
  - Homocystineuria
  - Classic Phenylketonuria (PKU)
  - Tyrosinemia, Type 1
  - Guanidinoacetate Methyltransferase deficiency

Endocrine Disorders

- Primary congenital hypothyroidism
- Congenital adrenal hyperplasia

Hemoglobin Disorders

- S, S disease (Sickle Cell Anemia)
- S, Beta-Thalassemia
- S, C Disease

Other Disorders

- Biotinidase Deficiency
- Critical Congenital Heart Disease
- Cystic Fibrosis
- Classic Galactosemia
- Glycogen Storage Disease Type II (Pompe)
- Hearing Loss
- Severe Combined Immunodeficiency (SCID)
- Mucopolysaccharidosis Type 1 (MPS1)
- X-linked Adrenoleukodystrophy (X ALD)
- Spinal Muscular Atrophy due to homozygous deletion of exon 7 SMN1 (SMA)
- Mucopolysaccharidosis Type II (MPS2)

Since July 2013, The Community Health Clinic has collaborated with the Indiana State Department of Health (ISDH) to provide follow-up care for infants with positive findings on the Indiana Newborn Screen (NBS). The CHC currently provides follow-up care when an NBS is positive for inborn errors of metabolism (IEMs), Severe Combined Immunodeficiency (SCID), Spinal Muscular Atrophy (SMA), or X-linked adrenoleukodystrophy (X-ALD). The Community Health Clinic is located in northern Indiana, nestled within the third largest Amish settlement, in Topeka, Indiana. While the clinic serves a large population from the Plain Community, The CHC provides excellent, affordable metabolic and genetic care to all patients referred to the clinic. This includes newborns detected by the Indiana Newborn Screening Program. The CHC is one of two metabolic clinics within the state of Indiana, providing access to care for patients locally. The CHC provides Newborn Screening Follow-Up for the above conditions in the following 19 Indiana counties: Adams, Allen, Daviess, Dekalb, Dubois, Elkhart, Fulton, Jay, Kosciusko, LaGrange, Marshall, Noble, Orange, Parke, St. Joseph, Steuben, Washington, Wayne and Whitley.

We provide NBS follow-up through our Das KIND program. Das KIND in German means 'the child.' The CHC's Das KIND program provides the following services:

- Rapid and direct contact from the CHC to the family upon receiving a positive screen.
- Education for the family and access to a 24/7 call service throughout the confirmatory testing process and after a diagnosis is made.

We build collaborative relationships with local primary care providers to impart relevant education and serve as a resource to allow for effective and efficient care of mutual patients.

BRUCE'S "EXCITING" STORY

As we recently celebrated our son Bruce's first birthday, it is still a whirlwind to remember the events leading up to and following his birth. With a year of review, here is the "exciting" story that got us here today.

Bruce is our fourth child and like many families with multiple children we, as parents, try to simplify our lives as much as possible. Bruce had other ideas. From the day he was born he has been much more than simple. My wife went into labor around 2am on a day I was at work overnight. She called me to come home and had the midwife already on the way to our house. My wife was going to have a home birth like she had with the other children. I came home around 2:30am to my wife standing at the front door in a pool of water telling me, "I wasn't kidding; I'm in labor!" The midwife arrived a short time later around 3am and started to get things set up. My wife moved to the living room and so began the "excitement."

Moments after getting somewhat comfortable sitting on the floor in the living room, the midwife checked to see how far along my wife was. She was ready to push! We thought, "Great! The other kids are sleeping and will wake up to a new baby." No hassle, no long, drawn-out labor like the other children had been. No baby sitter needed, etc. Easy, right?...Wrong. My wife gave 3 or 4 good pushes and out popped a……leg. And I mean a leg. Knee down to foot. The midwife and I looked at each other like "uh oh." The midwife said we needed to go to the hospital. Now we are typically not hospital people if at all possible. Bad experiences, self-sufficient, whatever the reason, we've always avoided hospitals. Well that was the only option for us, so off we went. Van loaded up with the midwife and Momma on all fours and a leg hanging out. While pulling out of the driveway we remembered that there are three young children asleep at the house, easy to forget amidst the hustle and bustle of labor. We called a friend a few minutes away and she came over to babysit.

We arrive at the hospital and the doctor tells my wife we will have to push the baby's leg back in and perform a C-section. My calm, cool, collected wife says, "No problem, it's our fourth kid, do what you got to do." I was shocked by her demeanor. Carefree, not getting too caught up in the moment. She's amazing!

So into the OR they go and healthy baby Bruce is born at 5:14am via C-section. Not exactly our plan, but when does everything go to plan, right? Following the birth was the usual checklist of tasks to accomplish before going home; hearing test, blood work, etc. And of course the **newborn screening**. So in our urgency to get out of the hospital, we pushed the nurse to check these boxes off as quickly as possible. Everything went smooth and we were almost out the door until…. "There is an issue with Bruce's newborn screening."

The doctor came into the room and discussed the potential issue without going into detail relaying that there is an abnormality and we will need to follow up with his PCP as soon as possible. We were out the door with our healthy baby and headed home, FINALLY.

The following day we visited the pediatrician and he told us Bruce would need a secondary newborn screening to again check into this abnormality. We assumed nothing was wrong, maybe just a bad sample. By the looks of things and the nurses' and doctors' remarks, we had a healthy normal baby. So we went to a nearby clinic and got a newborn screening. A day or two later we get the results from the pediatrician that the abnormality is still there.

The newborn screening showed a condition called Profound Biotinidase Deficiency (BIOT). This is a very rare condition, so at the guidance of the pediatrician and the people behind the newborn screening, we were advised to get a full blood test done to confirm this condition.

Now at this point we are a few days past leaving the hospital and the last thing we wanted to do was go to more appointments and get more tests. Being at the hospital for two days had put our lives out of routine enough. We wanted to relax with our

new baby. That is when we got a call from Community Health Clinic in Topeka, IN. The nurse there explained exactly what was going on and expressed the urgency to address the issue. Bruce had BIOT and despite his "normal" appearance on the outside, there was a condition going on that had great potential harm. Over the next week or so we were in contact with Community Health Clinic regularly and had an appointment in person to make sure we knew everything we needed to know. This was great given the rabbit-hole one can dive into on the Internet.

Bruce's condition was confirmed and the steps to address it were spelled out. With a simple pill once a day, Bruce will appear and develop in the same way our other three kids will. What started out as just another box to check before leaving the hospital, Bruce's life was saved from a condition that we would not have seen on the outside until it was too late. This was all due to getting a newborn screening done. GET A NEWBORN SCREENING DONE. Thanks to this simple step and the amazing staff at Community Health Clinic in Topeka, IN, Bruce now has the opportunity for a full and healthy life. We just hope it's with a little less "excitement" than it started.

– John Davis and Family

WAYLON'S JOURNEY

Our son Waylon was born on December 31, 2020. Just 24 hours after being home from the hospital we received a phone call from the geneticist, Dr. Ammous, whose clinic was an hour away. She informed us that our son was tested for over 50 things on his newborn screening and one of them came back positive. We were unaware of what newborn screening was until his diagnosis. His Dad and I were both in shock because I had genetic testing when I was pregnant and we were beyond confused as to why we weren't aware of this prior. Dr. Ammous let us know that for us to be aware of this condition, prior to birth, we would have had carrier screening done. We never knew that this test existed. When the results came back low risk from my prenatal testing, we thought everything was fine. Dr. Ammous let us know that we both had to be carriers of SMA in order to pass it on to our child. We had a 1 in 4 chance of either having a child with nothing or having a child that is affected. Being first-time parents and only being home 24 hours with our precious baby boy before receiving such devastating news was a feeling I'll never have words to describe. Dr. Ammous brought us hope by informing us on three FDA-approved treatment options.

SMA stands for Spinal Muscular Atrophy and it is a genetic disease that causes the muscles to become weak and waste away. People with SMA lose a type of nerve cell in the spinal cord which controls the muscles. We took him to the clinic the same day we got the call, to confirm what type he had and how many copies he had of his backup gene. There are four types of SMA and he was so symptomatic when we took him in, that Dr. Ammous knew on the spot, even before getting bloodwork back, that he was most likely type 1, which is the most severe form of SMA, with only two copies of the backup gene. Type 1 children typically show symptoms by six months of age, where Waylon showed symptoms at day five of life with belly breathing; bell-shaped chest, and no head control. His neurologist used the word "floppy" often. While Waylon was lying down on the table she demonstrated by picking up both arms and letting go. His arms went straight back and hit the table. He had absolutely no control over stopping them and when you held him, his arms would lay behind him due to super low muscle tone. One day before receiving treatment, at 26 days of life, Waylon showed the first movement we had ever seen in his arms.

After he received gene therapy at 27 days old, it helped ease my mind that the treatment would halt the progression of SMA. A month later his crying got louder and we started seeing movement in his arms. Waylon also had slow weight gain and severe belly breathing which led him to being on bipap at night. This has helped him with breathing and widening his chest. Waylon has therapy three days a week and continues to make progress weekly. He amazes us with his dedication to continue to work hard and do things that we never thought we would see. He sits unassisted, rolls both ways, plays with toys, but his biggest struggle is lifting his head. It stops him from being able to crawl or sit himself into a seating position. For our family, it is not IF he will crawl, walk, or meet other milestones, it is when. He just had his yearly evaluation and they said his fine motor skills were on track for a child his age. The fact that he can sit unsupported is unheard of in the natural history of SMA and before treatment.

At the time of Waylon's diagnosis, all we could think of was "why us"? Then we came to the realization that there are people who are denied treatment or have to wait months to receive it. The sooner the treatment is received, the better the outcome. We were fortunate enough to be able to get therapy so we would say we are doing pretty well. There are times when we wish he was progressing more, but we remind ourselves that we are blessed to have him with us today and he is thriving. Looking back at the newly diagnosed days we never saw ourselves where we are now. By no means are we saying this is easy, but when you have a little one that depends on you daily, it gives you every reason to keep on giving them the best life possible. We will continue to advocate for Waylon and be active in the SMA community through fundraising for Cure SMA and other organizations. Waylon's Mom, Jessica, states, "No matter how weak you may feel after receiving your child's diagnosis, just know in the end you'll come out stronger than you ever knew you were. You will fight for your kid until your very last breath."

My name is Hanna Quintana,

I have a son, whose name is Ozias Reed.

I gave birth to him August 18th, 2022. He was perfect. Me and his father were thrilled. As we got ready to head home, after two days of testing, making sure his feeding was okay, we got his newborn screen back. His test came back with a positive screen for Adrenoleukodystrophy. My father had passed away from this disease, and me and my son's father were heartbroken. However, the newborn screen allowed us to catch it in enough time that we were able to set up a team of doctors in neurology and endocrinology. The newborn screen gave me peace of mind. Knowing that I was walking into motherhood with a new challenge. The newborn screen helped because I was able to get the support I needed right away.

I believe there should be more tests on the newborn screen and would highly recommend it. Even if you have a family history of no illnesses. This will help you better prepare for the best future for your child.

UPCOMING EVENTS

CHC Open House & 10th Anniversary Celebration
September 9th

Walk for Rare, Run for Hope
October 7th at 9 a.m.
The CHC
Register Online

Daviess County Community Education
October 11th
The Daviess County Produce Building

SNIP 1 Family Day
October 21st
The CHC

VLCADD Berne Education
November 17th @
the South Adams Senior Center

Isaac's Story

On April 14, 2019, our son Isaac was born. He appeared to be a healthy, normal infant at birth, weighing 8 pounds and 15 ounces. Little did we know that something was wrong.

At four days old, The Community Health Clinic in Topeka, Indiana received notification from the newborn screening lab that Isaac's newborn screening indicated that he may have SCID (Severe Combined Immunodeficiency). The Community Health Clinic immediately made contact with us to let us know what the findings were on the newborn screening.

We had to do more bloodwork to confirm the diagnosis. We still had a little bit of hope that everything would turn out to be just fine. But two days later his diagnosis was confirmed. He did indeed have SCID. This meant he had little to no immunity, so if he happened to get an infection of any sort, he would not be able to fight it off on his own. We were told to keep his body checked daily for any rashes. If he happened to get an infection, our first sign would probably be with him breaking out in a rash. That would be his body's way of showing us that he can't fight it off.

We kept Isaac checked daily. At eight days old he started with a rash on the upper part of his leg. We called The Community Health Clinic, and it wasn't long until they were at our house examining his rash. They told us we needed to pack our bags and go to Riley Children's Hospital. So that's what we did. We had a four-day hospital stay. During that time, they did a lot of bloodwork and testing. We had doctors telling us what to expect in the next three to four months. They explained that the only cure for Isaac is to have a bone marrow transplant, or a cord blood transfusion done at the age of three months. He needed to be kept healthy up until that time. Isaac also had a skin biopsy done to find out what the rash actually was. It turned out to be an infection, but it was quickly and effectively treated at Riley. After four days, we were sent home. Again, we were placed in strict isolation.

Three weeks later we had a checkup at Riley to have more testing done. Now the doctors were searching for an unrelated bone marrow donor for Isaac. We were waiting and praying for a match. A couple weeks passed, then one

day the doctor notified us to tell us that they had found a perfect match for our son! So, when Isaac was 2 ½ months old we went to Indianapolis to start the bone marrow transplant journey.

The first thing they did was have surgery to place two central lines in his chest. Those were placed for blood draws, blood transfusions, immune infusions, and bone marrow transplant. Isaac had seven doses of chemo. The day for his transplant came and was followed by four more doses of chemo. Isaac had his good days and his bad days, but overall, everything seemed to be going well. Finally, they released us to go over to the Ronald McDonald house.

Again, we had to keep our eyes open for any rashes or any other signs of infections. Although the transplant was done, the doctors told us it'll be months or even a year before Isaac's immune system would work properly like a healthy person. It wasn't long until he started having symptoms of an infection, so we were sent to the emergency room and had another short hospital stay. After a little over nine weeks in Indianapolis, we got to go home. We still had weekly checkups at Riley and weekly checkups at The Community Health Clinic to keep his central lines clean and sterile and to check to see if his counts were still gradually coming in.

One week before Isaac turned one year old, we got the exciting news. His immune system was working like it should. Now he's a healthy and energetic little guy.

Although at times we thought it was a little difficult, we are very blessed with how healthy he is now. He has some issues with his kidneys, which probably comes from his chemo treatments. Otherwise, he remains healthy. He is currently off all his medications. It was well worth it to finally have a healthy son.

May God bless you all,

Leon, Karen, & Isaac Hostetler

ENGAGING FAMILY LEADERS IN THE NEWBORN SCREENING SYSTEM

Navigate Newborn Screening
An Expecting Health Program

Newborn screening is more than a screen at birth, but a complex system with many stakeholders. We believe that families are integral to driving system change and supporting positive family experiences within the newborn screening system. The Newborn Screening Family Education Program (Navigate Newborn Screening) is dedicated to developing opportunities for all families to learn about newborn screening and to develop training and educational resources that build confidence for families to become leaders in the newborn screening system.

In the fall of 2021, The Newborn Screening Family Education Program established the **Navigate Newborn Screening Ambassador Program**, in which we have recruited over 20 family leaders across 20 different states and 16 unique condition groups to participate in a 10-month program to learn more and develop opportunities to serve as leaders within the newborn screening system.

Ambassadors are trained to connect with others, build awareness and knowledge of newborn screening, and serve as leaders throughout the system. Ambassadors are available to share their experiences with diverse audiences, engage in programs and project development as well as collaborate with other families or organizations to implement systems change programs.

Hear more from one of our Navigate NBS Ambassadors from Indiana…

When our son Grant was born in February of 2021 I didn't think much about newborn screening. I didn't really know what it did, I just knew it was something that happened before you brought your baby home from the hospital. I never expected to get a phone call that something had come back abnormal on our son's newborn screen. Grant was diagnosed with severe Pompe disease from newborn screening in Indiana. When we went to his first appointment, his doctor was so thankful for his newborn screening diagnosis. At the time I didn't realize what a big deal that was. Thanks to Grant being diagnosed on newborn screening, he was able to begin treatment for Pompe disease at just three and a half weeks old. Early diagnosis and treatment have been life saving for Grant. Today, he is a thriving and wild two year old because of the access he had to newborn screening. After his diagnosis I learned that not all states screen for the same things and that newborn screening is different depending on the state you live in. For Grant, he got lucky. He was born in the right state at the right time. Indiana only started screening for Pompe disease a few months before he was born. I have become passionate about sharing his story and advocating for newborn screening because I believe that every baby deserves newborn screening. Newborn screening is life saving and allows for babies that are born with rare but treatable diseases the access to early diagnosis and treatment.

I will be forever thankful for the opportunity to be a part of the Navigate Newborn Screening Ambassador Program. I have been able to connect with other families that are similar to mine and truly understand what it is like to have a child with a rare disease. I have been able to form a relationship with the Indiana newborn screening lab and I am working with them on ways to provide more education on newborn screening to expecting mothers. It has also allowed me to become connected with the Texas newborn screening lab and I will be traveling to Texas in a few months to share our story with newborn screening and the life-saving difference that it has made for our son. Through being a part of the Navigate Newborn Screening Ambassador Program I have gained more confidence and knowledge to be a better family leader in newborn screening. I am excited to see the difference I can help make with newborn screening and family education.

I think that it is so important for families to understand what newborn screening is and what it does. My sister-in-law had just delivered her first child and didn't know anything about newborn screening. She was told that they needed to do a heel prick on her daughter so that they could do the newborn screen. Since she didn't know anything about it, she questioned them and considered not having it done. After our son was diagnosed, she has learned the importance of newborn screening and wishes that it would have been explained better to her before the birth of her first child. Since then she has gone on to have two more

children and has made sure a newborn screen was done each time. I think there are so many people who have found themselves in this place of just not being educated on what newborn screening is for. I think there are several ways that there could be more education about newborn screening. It should be part of the prenatal education process when expecting parents take prenatal classes. It should also be something that an OB-GYN goes over with their patients prior to delivering a baby. There could also be videos about newborn screening that play in the waiting room of OB-GYN offices. The more opportunities there are for newborn screening education, the more likely families are going to understand the importance of it.

TO LEARN MORE ABOUT OR CONNECT WITH THE NAVIGATE NEWBORN SCREENING AMBASSADORS, YOU CAN

1. **Watch these videos:**
   https://www.youtube.com/playlist?list=PL6M2gEwQoV25pYKKTwN7GnK1cf1kiDL40

2. **Read more here:**
   https://www.expectinghealth.org/sites/default/files/inline-files/Meet%20Our%20Ambassadors%20Booklet_0.pdf

3. **Connect with us** by emailing
   mraia@expectinghealth.org

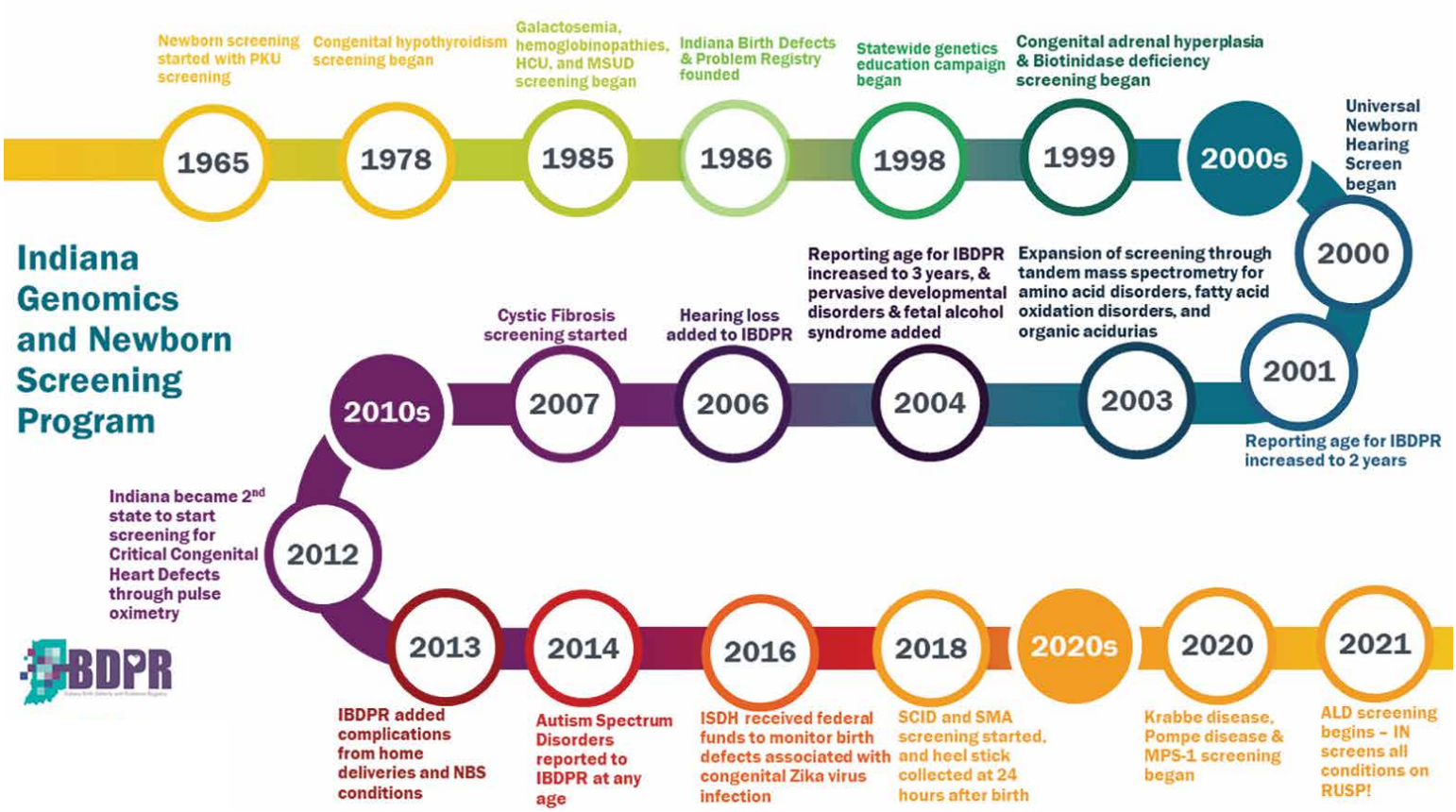
Indiana Department of Health, Genomics & Newborn Screening Program

Newborn screening is a simple and quick way of detecting serious health conditions within days after a baby is born. Every baby has this set of three screens done to look for signs of over 50 rare genetic conditions. The health conditions that screening looks for often do not show any signs until the condition starts causing serious health problems. Many of the conditions that screening looks for are passed down in families through their genes. Some communities have a higher risk of developing conditions because their families carry genes that cause the conditions. The Indiana NBS Program partners with the Community Health Clinic to make sure that all families and communities with higher risks of conditions have access to screening and other health services that can greatly improve their babies' health outcomes. It is especially important for families with higher risk of conditions to have their baby screened within 24-48 hours after they are born and mail the screen sample within 24 hours. Quick detection and intervention can be life-saving.

The program values being family-first, equitable, engaging, innovative, and advocates for families of babies with rare health conditions. Please reach out to their program with questions or concerns about screening by calling 888-815-0006 or emailing NewbornScreening@health.in.gov.

Metabolic Formula Program:

This July, the Indiana NBS Program has launched a more approachable statewide metabolic formula program to help provide more convenient and equitable access to metabolic formula for patients with IEMs in Indiana. The Metabolic Formula Program's aim is to prevent the high financial and emotional burden of inconsistent access to metabolic formula by offering assistance. Abby Hall, the program coordinator, will work closely with the metabolic clinics and dietitians to identify patients and families with no or poor medical formula coverage. Once identified, the clinic will



NEWBORN SCREENING
INDIANA

reach out to Abby who will then work one on one with the family to get them metabolic formula as needed. The program is available to any patients or families currently prescribed metabolic formula or supplements.

This new program has been both a passion project along with a needed update on the old program, according to program coordinator. Abby previously worked as a metabolic Dietitian at the Riley Children's Hospital metabolic clinic and through this has over 10 years of experience working with metabolic patients. She knows first-hand how expensive and difficult it is navigating metabolic formula is and is thankful to be able to update this program for the families in Indiana. If you feel that this program would benefit you, please reach out to your dietitian or Abby directly at 317-775-0320 or emailing MetabolicFormula@health.in.gov.

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The Mission of the Community Health Clinic (CHC) is to provide excellent and affordable medical care consistent with the needs of the Amish, Mennonite and other rural northern Indiana communities with a focus on individuals and families with special health care needs. The CHC embraces, incorporates and promotes participation in research to advance medical knowledge and improve care.

Jody Werker, RN
Newborn Screening Coordinator

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