Alberta Newborn Screening Program
Family Experiences

Alberta Health Services
Alberta Newborn Screening

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Baby Gabriela's story

September 2023

Newborn screening made a profound impact on the family of Arthur and Camila Iwaniszyn.

"We understood what newborn screening was at a high level, but didn't know how important it was to some people," says Arthur.

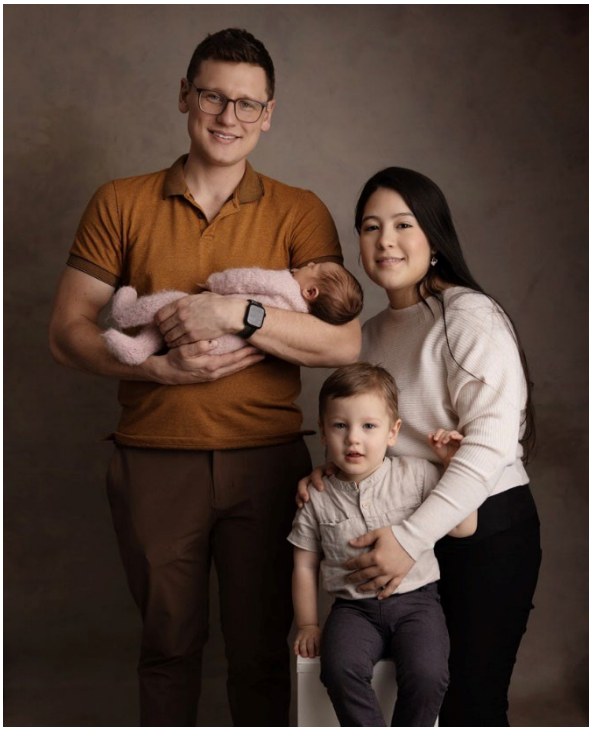
When their daughter Gabriela was born, their midwife took a routine bloodspot sample, as part of the Alberta Newborn Screening Program (ANSP) – a provincewide screening program offered to all babies by Alberta Health Services (AHS).
Gabriela was diagnosed with spinal muscular atrophy (SMA). "I didn't think SMA would happen to us or Gabriela, but getting her screened ensured we could give her a head start in life," says her mom Camila Iwaniszyn. The program aims to prevent health problems, improve newborn health and save lives.

The program works to prevent health problems, improve health and save the lives of Alberta newborns through early diagnosis and treatment of specific conditions such as spinal muscular atrophy, cystic fibrosis and more. Using a quick heel poke, drops of blood are collected on a special card and tested for 22 treatable conditions.

Two weeks after Gabriela's sample was taken, she was diagnosed with spinal muscular atrophy (SMA), a condition that causes progressive muscle weakness over time.

It affects the ability to crawl and walk and, sometimes, even the ability to breathe and swallow.

"When we received the phone call, it felt like a blur and the panic set in. We had no idea what SMA was," says Camila. "But we were determined to find out more information and help our daughter."

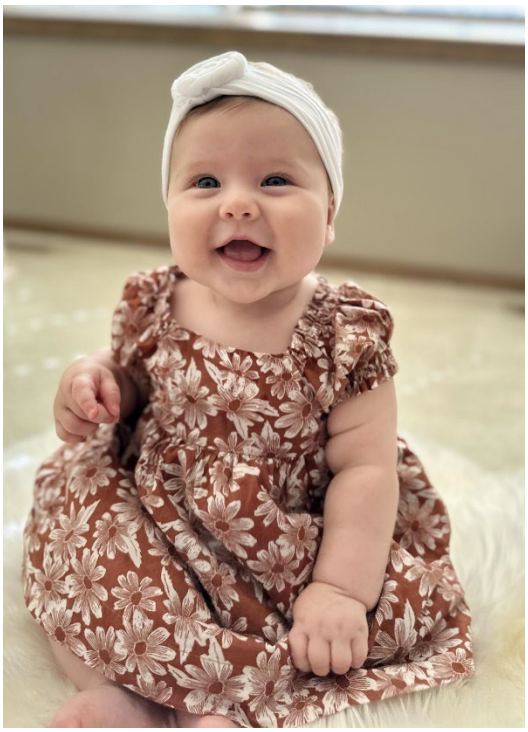


Here, Gabriela is shown being held by her father Arthur, with her mom and brother Jacob.

The Calgary family then began their journey to find treatment for their daughter, with a team of healthcare providers on board to help them along the way.

"It was overwhelming dealing with so many people involved, but they were working really fast," Camila adds. "I found it comforting to know so many people cared about her. Her doctor already had a treatment in mind — and we just had to wait and see if she was eligible."

After a week, the family found out Gabriela was eligible for treatment. Today, after receiving treatment, Gabriela is a happy six-month old, and meeting all her developmental milestones.



Here Gabriela is at 5.5 months old sitting independently.

“Now that we’ve seen how screening works, the impact it had on us and the steps we took to help Gabriela, we’re more grateful than ever to have had screening, or we never would have known,” says Arthur.
Dr. Jean Mah, Pediatric Neurologist at the Alberta Children's Hospital, adds: “Newborn screening saves lives. It gives children the best chance to live healthy and normal lives.”

The Iwaniszyn family hopes more parents-to-be will realize how important newborn screening is for their babies.
“I didn’t think SMA would happen to us or Gabriela, but getting her screened ensured we could give her a head start in life,” says Camila. “Screening is a good safety net. It can save both their life and quality of life.

Mighty Hudson's story

May 2023

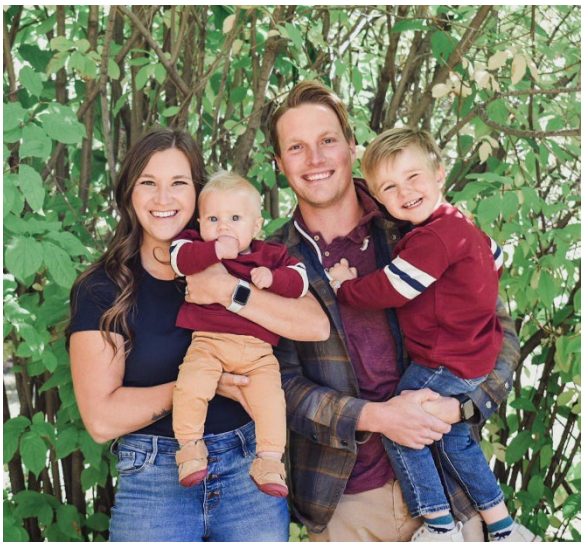
Hayley Cowie and her family never expected to receive an abnormal genetic-test result just days after their bundle of joy, Hudson, was born.

The Spruce Grove couple's first child arrived in June 2019 and underwent newborn blood-spot screening while in the hospital as part of the Alberta Newborn Screening Program (ANSP) — a provincewide program offered to all babies born in Alberta by Alberta Health Services (AHS). Screening is offered before discharge from hospital and involves a quick heel poke to collect a few drops of blood for testing.

"The program works to improve newborn health and save lives of Alberta newborns through early diagnosis and treatment of certain conditions such as sickle cell disease, cystic fibrosis, metabolic conditions, endocrine conditions and more," says Dr. Huiming Yang, medical director Provincial Screening Programs. "Some of these conditions can be fatal if not caught and treated early."

Hayley and her husband Ian were notified in July by their primary doctor that their son's screening results had come back positive for Severe Combined Immunodeficiency (SCID) — a genetic disorder that can cause abnormalities in the immune system, making infants susceptible to developing re-occurrent and life-threatening infections.

Hudson was the first child in Alberta to be diagnosed with SCID under the ANSP. "We panicked when we first got the news of the positive screen results — and we were shocked because there was no history in our families," says Hayley. "We thought it could have been a false positive. Waiting to hear the test results to confirm was the hardest part."



The Cowie family strikes a sunny pose: mom Hayley, left, Max, dad Ian and Hudson.

Armed with the knowledge of this new diagnosis, however, the Cowie family and their healthcare team sprang into action with a treatment plan.

"Because of newborn screening, we were given the chance to fight early," says Hayley. "The immunology team at the Stollery Children's Hospital was amazing and walked us through so many uncertain times. They had a plan right away and all we had to do was stick to it."

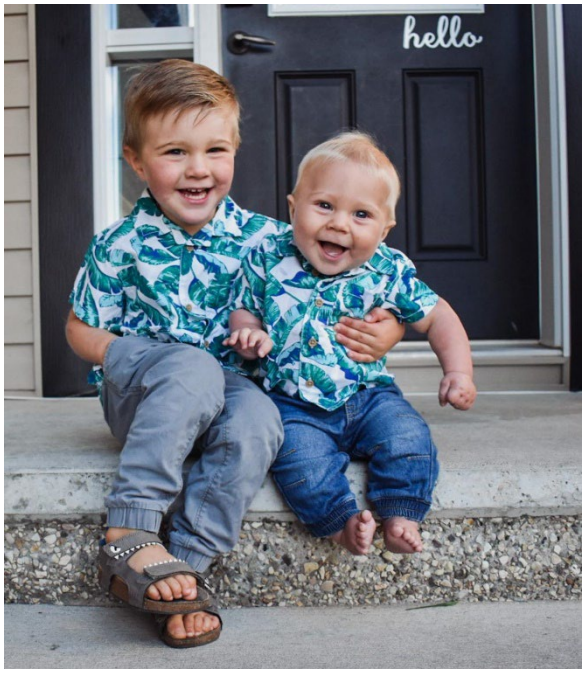
The family explored options for treatment and geared up to do a bone-marrow transplant, with family and friends open to being donors. In August, Hudson was accepted into a gene-therapy trial program at St. Jude Children's Research Hospital in Tennessee.

“To be on the trial, you had to have no previous infections. We were lucky Hudson’s condition was caught early, or we’d be in and out of the hospital system, and may not have qualified for the trial,” adds mom Hayley. “He was able to complete treatment quickly and was completely healthy within six months.”

“Screening is quick and safe,” says Dr. Yang. “Performing newborn screening within a baby’s first 10 days of life ensures they get timely access to the care and treatment they need for healthy development.”

Now almost four, Hudson is a fun-loving big brother to his baby brother Max.
“He’s doing great and there are no concerns at all,” says Hayley. “He has lots of energy and loves anything outdoors.

“For any parents who may be worried, this is a minimally invasive procedure — and I’m a strong advocate for newborn screening.”



Here is Hudson with his little brother Max.

Baby Jacob's story

September 2021

Ten days after he was born, Andrea Fernandez de Soto's son Jakob was diagnosed with severe combined immunodeficiency (SCID). The genetic condition affects his ability to fight infections, and his body can't keep him from getting sick.

The family was told he likely only had a few months to live. "It was pretty tough, pretty awful," Andrea recalls. "We were so upset. We never imagined this would happen to us."

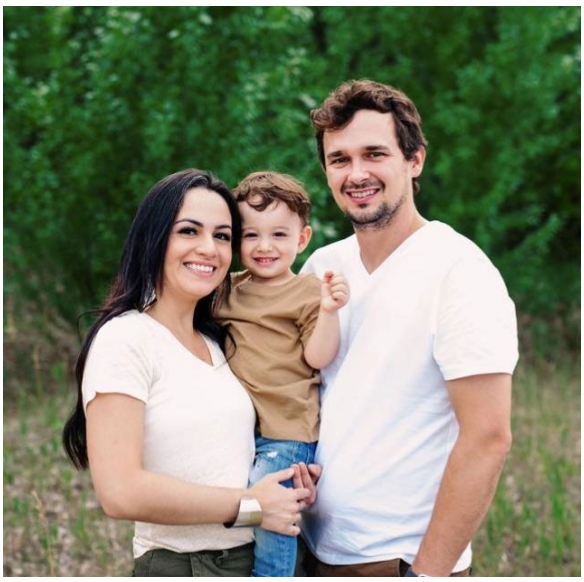
But because the family had agreed to newborn screening, the condition was discovered early, and little Jakob quickly received treatments – treatments that will likely continue the rest of his life.

He receives antibiotics every day, and enzymes twice a week that help his lungs heal and stay healthy.

It's been a long couple of years for the family. Andrea was diagnosed with cervical cancer not long after Jakob's diagnosis. She has since completed radiation and chemotherapy, and she has written a book about Jakob, to be released this coming December.

Now, however, Jakob is a happy, bright little guy who loves to watch cartoons and is learning three languages: Spanish, his mom's language; Polish, his dad's language; and English. And last month, the family celebrated Jakob's second birthday.

"It was such an emotional celebration, an amazing time," Andrea says. "We are happy that he's doing well and beating the odds."



Here baby Jacob is with his Mom and Dad.

Screening is the best way to find out if your baby has a condition that you, your doctor or your midwife might not know about.

"My friends always ask me, 'How did you find out about Jakob?' I say he was diagnosed because of the newborn screening," Andrea says.

"Without that newborn screening, we would not have Jakob with us right now. I tell my friends who are expecting now: Your baby is going to be tested."

Early screening ensures babies get treatment when it can help the most and may even save lives. It's quick, effective and can be done, free of charge, shortly after birth.

Baby Monet's story

September 2019

Josh Gaudet and Matt Urquhart have big advice for other parents: get your newborn screened as soon as possible.

"I can't imagine not doing it," Josh says. "Trust the (healthcare) professionals. Just trust them."

Newborn screening is the best way to find out if your baby has a condition that you, your doctor or your midwife may not know about. Screening is quick, offered free of charge, and can be done shortly after birth. "It's easy to do," Matt says. "And the supports are there."

Through newborn blood spot screening, Josh and Matt's adopted daughter, Monet, was diagnosed with classical phenylketonuria.

Best known as PKU, it is a hereditary metabolic disorder that keeps the body from digesting protein properly. Without early treatment, a baby's brain will not develop correctly. Every day counts.

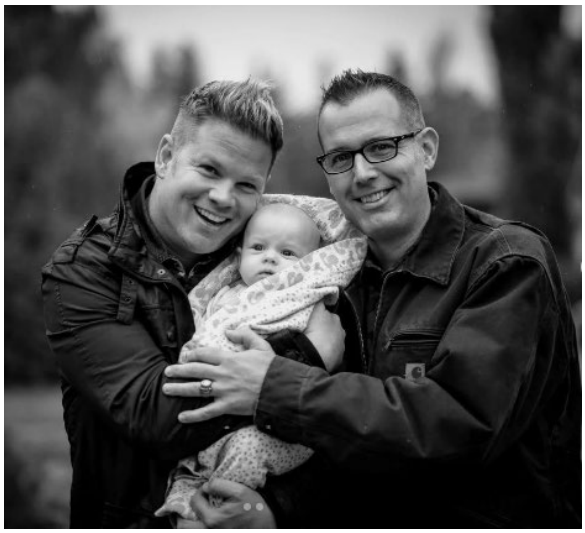
"The PKU diagnosis is scary when you read about it at first," says Matt. "If it is untreated and you feed normal food or breast milk, it will cause irreversible brain damage."

But if it is diagnosed at birth, PKU can be completely controlled by diet. "And the child can grow and develop normally," Matt says.

Within a day of the diagnosis, the family had met with a doctor and a team of AHS nutritionists. Monet was soon getting the special medical formula she needed. "She can't have certain proteins. No meat, dairy, eggs, fish," Matt says. "You just give enough

protein for growth."

These days, Monet is a lively four-month-old who laughs when her nose is touched and smiles and laughs when she hears the sound of her parents talking. "She's exactly where she needs to be for optimal growth," Josh says. "She's hitting all milestones or exceeding them. She's growing."
And both Josh and Matt are grateful for the help they've received from AHS.



Baby Monet with her parents Josh and Matt.