



Newborn Screening
Saves Babies
One Foot
at a Time

Jordan

THIS IS A PERSONAL STORY SHARED BY A FAMILY WHOSE CHILD HAS
PROPIONIC ACIDEMIA (PA)

My name is Jordan Franks. I was born in Evanston, Illinois on January 28, 2000. I could see the pride and love in my parents' eyes as they beheld what they believed was their perfect little creation - all 8 lbs. 14 oz. of me. Unbeknownst to them, and the doctors and nurses, who attended to me, I was afflicted with a rare genetic disease. No one understood the signs - vomiting after birth, disinterest in nursing, rapid breathing, and sniffly nose. I thought my symptoms were loud and clear, but after two chest x-rays and the decision that it was birth trauma, I was sent home. My parents were told to bring me to the pediatrician in a couple of days to check my rapid breathing.



My mom tried feeding me and I just wasn't interested and acted like I didn't like the taste. How could she possibly know that the protein in the milk was poisoning me, causing elevated ammonia levels in my blood and acidosis? The next morning I was lethargic and wouldn't startle. My mom immediately took me to the pediatrician. My temperature was 93.6 degrees and I was dehydrated. I was down to 7 pounds 12 ounces and my doctor contacted our local hospital to inform them that we were on our way. After numerous labs and being hooked up to IV fluids, my parents were told I probably had a metabolic disorder. A what??? The doctors were able to get my acidosis under control; however, my ammonia levels went from 500 to 1050 and I was transported to Children's Memorial Hospital in Chicago for hemodialysis.

After two catheter surgeries, they were finally able to perform the hemodialysis and get my ammonia levels under 300. I survived the procedure, but I was in a coma and hooked up to everything including a respirator. My parents were sad and confused and didn't realize the seriousness of my condition until the doctor came back with a diagnosis. I had Propionic Acidemia, an inborn error of metabolism. My parents ran straight to the nearest computer to surf the net. Luckily, they found OAA (Organic Acidemia Association).

I was sent home at 3 ½ weeks and started vomiting the next day. By early morning, I was breathing heavy so my parents took me to our local ER to find out that my ammonia was back to 300. Transport arrived to take me back to Children's Memorial. For the next three weeks of trial and error and fluctuating ammonia levels between 40 and 200, my doctor made adjustments to my formula. Since then I have had 6 more hospital stays, g-tube surgery, viral gastroenteritis, and pancreatitis. My full strength formula currently consists of whole milk, water, PFDII, XMTVI Maxamaid, Polycose, canola oil, valine solution and isoleucine solution. I also receive Carnitor, iron, polyvisol and flagyl (10 days each month).

Although I am 14 months old, alert, active, and happy, I am developmentally delayed and at an 8 month level. I can move around the floor, but I haven't started crawling yet. I love babbling, but I don't say consonants yet. I

hate food and on a good day will take 2 teaspoons of baby food by mouth. The rest of my formula goes through my g-button. I receive speech therapy, occupational therapy, physical therapy and developmental therapy. My nurse, Pauline, is wonderful and comes 4 days a week to help with my care. My mom and dad have become experts at being my advocate. They can usually tell when my levels are off and make the necessary formula adjustments.

My prognosis for the future is uncertain. What is certain is that I will need constant medical care and therapy to maintain my condition. My story is a familiar one for children with metabolic conditions. There will always be questions of what if. I was diagnosed sooner. Would I be crawling and walking and saying "mama" and "dada".

One could say that my survival is a miracle. Perhaps the reason that I was destined to live is to be a voice for all the babies who have not yet been born. With timely comprehensive newborn screening, other babies will have an opportunity for early diagnosis and proper treatment and therefore a better quality of life.

On a lighter note, my big brother, Ryan, is 4 and constantly trying to stimulate me by giving me kisses and putting toys in my hands. After my diagnosis, Ryan was tested with CNBS and I am thankful the results were negative. My grandparents, great-grandparents, aunts, uncles and cousins have been incredibly helpful and supportive. My parents feel like they now have an extended family consisting of their new friends from all of the internet support groups and are so grateful for all of the support. Every day they see me do new things and know I am a miracle and that it's important to cherish every day.

Sincerely,

Jill Franks

Written March 2002 by Jordan with a little help

from his Mom Jill Franks

Highland Park, Illinois.