



Newborn Screening
Saves Babies
One Foot
at a Time

Brett

THIS IS A PERSONAL STORY SHARED BY A FAMILY WHO HAS A CHILD WITH
VERY-LONG-CHAIN ACYL-COA DEHYDROGENASE DEFICIENCY (VLCAD)

Our long-awaited third child arrived February 9, 1998. Watching the video of his birth you can hear my almost hysterical cries, "Is he healthy, is he healthy?" Only my nearest and dearest friends knew of my fear to try for another baby due to haunting premonitions that my third child would not be healthy like my first two were. Much to my happiness, my doctors and midwife assured me that he was perfect.

Then how can it be that almost 3 months later we would be back in the same hospital, me hysterical, but this time the doctors unable to assure me that he would pull through? I recall the events of Friday, May 1, 1998 as if it were yesterday...how I made those 3 a.m. phone calls to my husband, friend, Debi, parents and in-laws. How one-by-one I sent each of his grandparents into his room to kiss his almost lifeless, cold, little body good bye. How can this be? He was healthy, reaching all of his milestones.

I knew something was wrong with him; maybe just a little physical therapy was needed because his muscles seemed to be weak, so I brought him to the neurologist (even though people said I was being neurotic and should just relax!). And she tells us to go straight to the hospital because his liver is taking up most of his stomach and he was having a difficult time breathing? Twelve hours later he suffers a heart attack because his heart was as thick as an adult male's due to all the fat that he was unable to break down since birth? His sugars and temperature are so very low and his liver so very large and fatty? He can't breathe so a tube is stuck down his throat? This cannot be happening!

He was taken to a larger hospital with my husband beside him, my dear friend, Debi, and I following behind the ambulance. Will he be alive when we get there?

The doctors scratch their heads, conference, bring in specialists who scratch their heads and conference some more. The cardiologists tell us that the needle that has been inserted into the sac around his heart to drain off the excess amounts of fluid that have been collecting there needs to be taken out the next morning and then nothing further can be done for his damaged heart. The nurse gently asks us if we would like her to call the priest. Last rites??? This cannot be happening. This is the stuff you only read about or see on TV! The priest offers to baptize him and we refuse. After all, the caterer has been booked and the church date set. We were not allowing ourselves to admit "out loud" that we could lose him. Brett was then blessed. Not normally one to pray, I prayed so very long and hard along with thousands of others. Prayer chains for Brett crisscrossed the country. The next day the needle was removed, and to everyone's shock, his heart started healing.

Thirty days in the ICU, a tracheoscopy, and gastrointestinal tube operation later, still no definite diagnoses. I knew I would crack if I didn't get an answer soon so through the Internet we found a support group for Fatty acid Oxidation Disorders. This was what the doctors finally narrowed their list of possible disorders down to. Which one of the FODs they did not know. We would later come to learn that Brett had an FOD. Simply put,

Brett was born missing the enzyme that breaks down fats into energy. After his supply of sugars is depleted, his body cannot get energy from stored fat to function. This causes his blood sugars to plunge leaving him vulnerable to seizures, coma or congestive heart failure. He must never go more than four hours without adequate calories. In the event he cannot eat due to a stomach virus, he must go to the hospital for an IV of glucose until he can hold down food again.

I was unable to leave Brett's side that month in fear he would die without me being able to say good-bye. However, during the Memorial Day holiday weekend, I finally went home to visit my other children while my husband stayed with Brett. The packet from Deb Gould, founder of the support group had just arrived. I sat for hours on end reading every back issue of their newsletter. The stories could have been our story in that the details were almost exactly the same! I cried myself to sleep that night, woke up and decided that I better pull myself together if I was going to be of any use to Brett. I started calling each and every family on the support group's list, thirsty for any information. One call would change the course of events. I called Heather Marsella who lost her beautiful 5-month-old daughter, Toni Marie, to an FOD called VLCAD, and also had a VLCAD son, Joseph. She told me about her doctor, Dr. Rinaldo, at Yale University (presently at Mayo Clinic), offered to place a call to him at his home, and within the hour I was speaking to him making arrangements to have Brett transferred to Yale the next morning by his hospital's special ICU ambulance team.

Within three hours of arriving at Yale, we had our diagnosis-one of the most rare FODs called VLCAD. It was truly one of the happiest days in my life. To finally have a name, and this time it was not a fatal name like the previous "we think he has" disorders. We began to, and continue to educate ourselves about VLCAD and feel confident in caring for our son's medical needs. Through e-mail, we compare notes daily with other members of the FOD Family Support Group. Although there is no cure for this serious metabolic condition we feel lucky to have found the very best doctors and specialists in the world to work with in forming a care plan for our son. We are meticulous in keeping his fat consumption to under eight grams per day, feed him every four hours around the clock, and follow a detailed protocol when he starts coming down with ANY type of illness.

Three weeks later, June 10, 1998, we brought our son home. How my mom and I wept to see him back in his crib. So what if there was a room full of medical equipment, tubing hooked up to his trachea, oxygen tanks, a feeding pump, monitors beeping and nurses. He was home. How good my bed felt compared to the hospital's floor, and how well I slept knowing my dear friend Susie, herself a newborn ICU nurse, was watching over Brett that night.

It has taken \$800,000 dollars worth of care to get Brett to the point he is now. And how is he? He is perfect. Yes, he still has a g-tube, and he racks up frequent visitor points at our hospital. Dismissed from speech and occupational therapy, he still receives physical therapy to build his upper body. But along with his medical alert bracelet he wears a big smile and brings joy to all who are lucky enough to meet him as strolls up and down the aisles at Shop Rite with his, "Hi lady, Hi man" greetings. He is a constant source of pure joy and pride. He has taught us more than we can ever teach him.

It is most painful to me to think about all the babies who died and continue to die DAILY. It pains me to think that a simple \$20 newborn screening blood drop test could have spared their lives. \$20 could have spared Brett the trauma he endured. \$20 could have saved the taxpayers and insurance company \$800,000. But unfortunately, lawmakers don't agree. Their research tells them that it is more cost effective to care for those babies who were NOT lucky enough to HAPPEN to be in the hospital when their hearts started giving out, and because of it suffered brain damage, have seizures, and are in wheel chairs. In other words, they would rather pay for the care of a child who was injured due to delayed diagnosis than to pay \$20 to screen each baby at birth. Only North Carolina screens every baby at birth for ALL of the treatable disorders that modern science can test for. North Carolina believes that babies' lives are worth \$20. North Carolina understands that in reality, it is more cost-effective to diagnose a baby PREsymptomatically. And I am not speaking as a hysterical mom.

This is fact.

Our lives are forever changed from the events we went through almost two years ago today. I left the successful Parent Toddler program I owned in town so I can spend time being an advocate for newborn screening. Some day I believe all babies will be tested at birth for treatable disorders like the one Brett has, but so much has to be done. Through the Internet, we have formed a special advocacy group to divide and conquer the endless amount of work. I don't believe I will personally begin to heal, and be able to put this trauma behind me until ALL babies are tested at birth. Only then will I believe that all of the babies who have suffered or died will not have done so in vain. Those babies who have been lost to these disorders are heaven's angels now. If not for them, it would not be possible for us to have our earth angel, Brett Parker Revinski.

Sincerely,

Gina Revinski

Written November 2001 by Gina Revinski

New York