

IVA family story

Jacob Sachs

(as told by his physician, Dr. Helmut Niederhoff)

Dr. Niederhoff from Freiburg University Hospital in Germany, wrote to OAA wanting to share this story of a patient that was recently diagnosed with Isovaleric Acidemia using the Tandem Mass Spectrometer, the newborn screening equipment necessary for early detection.)

This male infant by the name of Jakob Sachs was born on August 21, 1998. The routine general newborn screening for hypothyroidism and treatable inborn errors of metabolism was done on August 25 (5th day of life), the Guthrie-card with dried blood spots being sent to the Heidelberg University Children's Hospital screening lab. This lab, by the way, was set up in the late sixties by Professor Horst Bickel, one of the pioneers of the dietary treatment of PKU. That's why the Heidelberg lab has become the screening center for the state of Baden-Wuerttemberg to which Freiburg belongs. As a sort of further innovation this screening lab in Heidelberg started in 1998 to feed their tandem mass spectrometer with dried blood spots from the general newborn screening, in order to detect not only PKU, but also organic acidemias, before the baby becomes sick.

Prior to that, gas chromatography and mass spectrometry (GC-MS) had been used in the Heidelberg University Children's Hospital for selective screening in infants showing signs of a metabolic disease. This, however, is being done by several labs in our country, including our Freiburg University Children's Hospital.

The Sachs baby happened to be the first case of Isovaleric Acidemia (IVA) detected by this new routine of the Heidelberg lab. Thus Jakob and his mother were transferred on his 11th day of life from their maternity ward to our Children's Hospital in Freiburg. We confirmed this inborn metabolic disease by GC-MS while the newborn was still in good shape: a mature boy who did not show any sign of IVA other than the lab findings including an already slightly elevated value of blood ammonia!

That means, our therapy came just in time. Reduced intake of protein, particularly of its essential component leucine, supplemented with carnitine and glycine in order to support effectively the excretion of increased amounts of isovaleric acid and related metabolites. For these are the products which cause the untreated child's serious metabolic trouble, since the genetic defect of IVA is located in the pathway of leucine break down.

Within a few days we got Jakob on his special, calculated diet. At time of discharge mother gave 300 ml of her breast milk that was mixed with another 300 ml of a leucine-free amino acid mixture supplemented with vitamins, essential minerals, and trace elements per day. The blood ammonia level returned promptly to normal. Urinary organic acids and blood amino acids were kept within tolerable limits.

The hospital stay lasted only 7 days and no further admission to any hospital has been necessary so far. Minor illnesses at home have been handled by dietary adjustments. Jakob is now 12 months old. His development is quite normal and his growth curve is following the lower percentiles of normal. Prerequisite for this favorable course, however, was teaching the parents all they have to know about IVA. They have been trained to cope with times of malaise and intercurrent disease with and without fever being supported by communication with our specialized pediatricians.

Cost Benefit Analysis

(according to hospital days in the Freiburg University Children's Hospital)

Early diagnosed organic acidemia versus late diagnosed organic acidemia due to Tandem-MS (Example:

Jakob Sachs with IVA):

Hospital Stay:

7 days

Subsequent admissions: None

Outcome:

Normal development with special diet

Total of hospital days: 7

Costs per day:

US \$546.00

Total Costs:

US \$3,824.00

Late diagnosed organic acidemia due to clinical signs (a former infant with IVA):

Hospital Stay:

38 days

Subsequent admissions: 5 times with a total of 32 days

Outcome:

Moderately handicapped child with retarded development inspite of special diet.

Total hospital days:

70

Costs per day:

US \$546.00

Total Costs:

US \$38,242.00

In addition to this one has to add the costs for special rehabilitation by physiotherapist, etc.

You may also take this letter or parts of it as a contribution to the OAA newsletter. I asked Jakob's parents for permission beforehand.

I wish you all the best for continuing your work!

Yours sincerely

Helmut Niederhoff, M.D.

From: October 1999 Organic Acidemia Association Newsletter

VLCAD family story

The Economic Cost of Not Screening Brett's Story

This is a personal story shared by a family whose child has [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

(over)

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 then 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

<http://www.savebabies.org/familystories/brettVLCAD.php>

LCHAD
family story

Adam's Story



2 years 10 months
by Valerie Fulton

After a stressful pregnancy Adam was delivered by emergency C-Section at 35 weeks weighing 4 lbs 4 oz. I had developed a rare disease of pregnancy called HELLP Syndrome** and we both would have died within hours from my rapidly progressing kidney and liver failure if the pregnancy were allowed to continue. We were rushed by ambulance to the nearest Kaiser facility with an intensive care nursery where the C-Section was performed. Adam recovered quickly and required only a few hours on the respirator. When Adam was 6 days old we were both out of intensive care and well enough to go home.

Adam progressed satisfactorily for the next 4 months although he showed some delay in motor development. I was concerned that at 3 months of age he could not lift his head when on his tummy. His two older brothers had done this with ease at this age. However, both were term babies weighing 9 pounds each, so the delay was attributed to his prematurity.

At 4 months Adam began to sleep through the night, which eliminated his middle of the night feeding. For the next month his health was stable, but there was little or no weight gain. Two or three days prior to his hospital admission, we noticed that he was not feeling well and seemed to have a slight cold. He had recently stopped taking solids and tended to frequently spit up in the morning. Also his sucking appeared to be weaker. Until this time Adam usually consumed from 20 to 26 oz of Enfamil with iron daily, but 2 days before admission he took 16 oz and only 9 oz on the day of admission. While waiting to be seen in the ER he appeared to want to spit up but was too weak to do so, and he was irritable and crying. His condition rapidly deteriorated until he appeared to be completely lethargic and non-responsive with a glazed look in his eyes. He was immediately tested for Meningitis, and his father and I were questioned about a possible drug overdose. His vital signs were stable and he weighed 12 lb 14 oz. Blood was drawn and other tests were administered. Initial symptoms appeared to be hypotonia, hypoglycemia and hepatomegaly. He had an enlarged liver and a blood sugar of 32. He was put on a glucose IV solution, which stabilized his condition. He remained in the hospital 5 days for further tests. Dr. John Mann of the Kaiser Department of Genetics was the principal consultant, and recommended several tests for genetic and metabolic defects. Urine and blood samples were taken and sent to several research labs around the country that specialize in this type of testing. During this hospital stay Adam's formula was changed to 2 to 3 oz Isomil every 3 hours. He was discharged after 6 days. Four days later the initial test results came back showing abnormal organic acids as well as fatty acids in the urine. This indicated a possible defect in the fatty acids metabolism.

After 8 days at home, he was re-admitted for a complete metabolic work up. His weight had gone down to 12 lbs. Adam was given many tests including head ultra sound, EEG, and MRI. Chest X-rays showed borderline cardiomegaly. A CT Scan of Adam's liver showed hepatomegaly with diffuse fatty infiltrate. A punch biopsy of his left thigh was taken and

sent to Dr. Hale at Philadelphia Children's Hospital for a cell culture analysis. A variety of urine and blood plasma samples were collected and sent to Dr. Neil Buist at Oregon Science University for study of organic acid excretion and serum guanidine organic acid levels.

During this hospital stay Adam was irritable fussy, and sucking poorly. When discharged 5 days later his weight had increased to 12 lbs 2 oz, which was still below the 10th percentile. Since there was evidence that Adam had a problem metabolizing long chain fat, his formula was changed to Portagen, a middle chain fat formula, as recommended by Kaiser's Regional Metabolic Nutritionist Elena Jurecki. Also L-carnitine was added to his formula. Carnitine is a protein present in everyone's body, which is thought to help move fat in and out of the mitochondria of the body's cells.

The results of the urine tests and cell culture confirmed the initial diagnosis. Adam had Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD), a deficiency in an enzyme that metabolizes long chain fat within the mitochondria of all body cells for energy production. This is an extremely rare condition which has only been diagnosed since 1987 with only a few cases reported world-wide. It is one of a family of disorders of fatty acid oxidation caused by a recessive genetic defect.

While Adam was at home this time his sucking stopped completely. We attempted to feed him for several days with an eye dropper which proved to be extremely stressful for both Adam and us. During this period Adam had an echocardiogram administered by pediatric cardiologist Dr Claude Roge at Kaiser San Francisco. It showed some myocardial thickening, but his heart was functioning normally.

At his next clinic visit 5 days later Adam's Pediatrician, Dr. Arthur Stein, was quite concerned by his poor color and weight (11 lb 12.5 oz). He immediately re-admitted Adam to the hospital and started him on a low fat formula of Tolarex supplemented with Isomil with a minimum fat requirement of 15% (a normal infant diet is approximately 55% fat). Since his sucking had dramatically decreased, he was started on NG tube feedings initially at 2 oz every 4 hours, which he tolerated well. During this hospital stay we were visited by Dr. Mark Lipson of the Kaiser Area Metabolic Consulting Center in Sacramento who provided one of the few encouraging statements we had heard. He said that Adam's heart seem to be less affected than most of the other 10 or 12 cases of this disease so far documented world-wide.

Frankly at this third hospital admission, I was certain Adam would not be coming home again. He was so weak, not eating, and even having difficulty swallowing. At one point his weight dropped to less than 10 pounds. His Pediatrician was also very pessimistic. We were asked if we wanted resuscitation if Adam's heart or breathing stopped, and we were given specific instructions as to how to proceed if Adam died in the hospital, or after we had gone home.

However, the strict regimen of frequent low fat feedings by NG tube gradually brought him back to a metabolically stable condition. Adam was able to go home after 17 days in the hospital. Except for a few set backs, his health continued to improve. His weight and strength increased rapidly. Within a month Adam could raise his head. Regular echocardiograms showed a decrease in the myocardial thickening. Within 6 months his heart appeared normal. However, his gross motor skills were somewhat delayed, and he didn't walk until he was 15 months old. His speech was also delayed due to his feeding problems and the NG tube.

Adam remained on the NG tube feedings continually from 6 months to 24 months. He was initially fed 6 times a day (including 2 am) His formula] consisted of a combination of Polycose, Isomil and Provimin with an L-carnitine supplement. This provided him with a high carbohydrate diet with the 15% minimum fat required for essential growth. At approximately 20 months Adam visited the hospital for an overnight fasting test. He went 12 hours without eating His blood sugar levels were checked at regular intervals. It was determined that he could fast as long as 12 hours without developing serious side effects. His daily diet was decreased to five 6 oz bottles supplemented with 2 tablespoons of cornstarch in each bottle. His middle of the night bottle was eliminated and his late night bottle supplemented with 3 tablespoons of cornstarch. Cornstarch is a very complex carbohydrate that takes a long time to metabolize. It acts as a time release carbohydrate for Adam. The elimination of the middle of the night feeding was not only a great relief to his father and I, but it also made it possible to consider removing the NG tube.

By approximately 20 months of age, when not ill, Adam was taking most of his daytime feedings orally. But occasionally, without warning, he would vomit after a feeding, possibly because the NG tube was affecting his gag reflex. At these times we became very frustrated, especially when it happened two or three times in a row.

From 10 to 24 months Adam experienced frequent ear infections (approximately one every 6 weeks). At 14 months he had a severe case of Chicken Pox, which the doctors were pleased he could weather without hospitalization. By his 2nd birthday we were able to remove the NG tube occasionally when Adam wasn't sick and was eating well enough. Whenever we had to reinsert the NG tube we noticed a regression in sucking and more frequent vomiting. He always ate better and vomited less frequently when the NG tube wasn't in. At 23 months his formula was changed so that it was completely fat free, consisting of non-fat milk, Polycose, and Provimin with a supplement of cornstarch and L-carnitine. He now has three to four 6 oz bottles of formula a day with the cornstarch supplement, and he has more solid food, which supplies essential fats.

Adam will be 3 on December the 21st. For a baby who was not expected to live at 6 months, he has developed into a normal rambunctious "terrible two". He is more active and gets into more trouble than either of his brothers at this age. His only hospitalization since his initial illness and testing, was because he had swallowed pennies which lodged in his throat. He has grown in height and weight. He is very intelligent and curious, and talks up a storm. Possibly because of his high carbohydrate-low fat diet Adam has more energy than the rest of his family. He is often still up and running around after the rest of us are ready to retire for the night. Adam's doctors are amazed at his progress. He is a real miracle child.

* LCHAD = Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency, a deficiency in an enzyme that metabolizes long chain fat within the mitochondria of the of all body cells. Almost all of the fat in a normal diet is long chain fat.

** "HELLP Syndrome is characterized by microangiopathic hemolytic anemia, elevation in SGOT/SGPT, and thrombocytopenia in pre-eclampsia. In addition to biochemical changes in liver function, hepatic rupture, hemorrhage, and death may occur." From "Complications in Pregnancy" (A manual for OBGYNs)

This story came from Adam's LCHAD website. More information is available at <http://www.adamslchad.com/>

GA-I family story

Hi Y'all! A little southern greeting to start our story!

My name is Lacy Posey. My husband, Shannon, and I have 2 children, Cody and Cheyenne. Cody was born on December 15, 2002. Although we are facing the “extremely picky eater” phase, he is a happy, healthy, rambunctious 3 year old boy unaffected by any metabolic disorders. Cheyenne was born on August 31, 2005. She has Glutaric Acidemia Type I (GA-1).

My pregnancy with Cheyenne was perfect. I gained 30 pounds, ate fairly healthy and stayed active. We opted out of the prenatal screenings because we didn't care if she had any type of disorders or syndromes; that wasn't going to change our minds about the pregnancy. No one in our families had ever exhibited any signs of genetic disorders before. Besides, no one ever thinks that it will happen to them. We weren't informed of exactly what tests are done on delivery or the option of having more done. I don't know that even with that information we would have done the screenings because of the lack of family history. We now realize how important it is to get the word out about the availability of these tests and how crucial they can be to your baby's health.

My delivery was induced because I was 2 days past my due date. When I arrived at the hospital at 6:30 am I had begun labor, so it was more of a help-along rather than an induction. The delivery lasted less than ½ an inning (the TV was on ESPN). Cheyenne Grace was born at 1:03 pm weighing in at a healthy 7 pounds 11 ounces and 20.5 inches. She was so perfect! Our well baby stay was uneventful. We were released after 48 hours.

I took Cheyenne by my & my husband's offices on the way home. Then we went to our new house (we had only moved in 5 days before she was born) to see her brother and grandparents. About an hour after arriving home, Cheyenne was hungry. I got my new rocking chair, Boppy and blanket all ready in her new room to nurse. She wouldn't latch on right away so I waited a couple of seconds and tried again. The lights were off, so I couldn't see very well. Shannon came in to check on us and when he opened the door and the lights from the hallway landed on us, I could see that Cheyenne was blue. I handed her to Shannon immediately. He tried talking to her and moving her around to get her to breathe. When that didn't work, he began rescue breathing. I was on the phone with the paramedics. Cheyenne began breathing again, but soon had another apnea episode. When the paramedics arrived she was back to normal. Their devices are not made for 2 day old infants, so they suggested that we take her back to the hospital for a complete evaluation. We left for the hospital—thankfully only 10 minutes away. I want everyone to know that during all of this, my son was amazing. He wanted to know what was wrong with his sister and why “those men” were there. He has gone through just as much emotional strain as we have and he is an incredible big brother!

At the hospital, I asked for the pediatrician on call and the nurses immediately took Cheyenne to the NICU and us to a room. I was crying by this point. I had no idea what was going to be done. I thought we had just gone in to talk to the doctor and go home. I didn't realize the seriousness of the situation. The NICU RN asked us lots of questions about the events at home, pregnancy, delivery and well baby stay. I was so rattled! A few minutes later she came back in and asked to sign a consent form for all kinds of test. We agreed.

I went home to be with Cody while Shannon stayed at the hospital. I came back a couple of hours later. The first time that I got to see her, they had started an IV in her head and she was in an NICU bed under the lamp without anything on except a diaper. The doctors didn't want to take any chances of there being an infection before the test results came back, so they started antibiotics for a brain infection as well as anti-seizure medications. Her small veins had a difficult time tolerating the IV lines for more than 24 hours. Shannon and I stayed at the hospital continuously. They were gracious enough to let us stay in an unoccupied room only 2 doors down from the NICU. It didn't have a shower, but we would take turns going home to clean up and spend as much time as possible with Cody. Cheyenne had 5 more episodes during the first 24 hours in the NICU. After beginning the anti-seizure medication, they stopped. As for feeding, I continued to breastfeed the entire time she was in NICU. She seemed happy and healthy...especially compared to the infants around her. She was by far the biggest one in there! On day four the doctor ordered CT, MRI and EEG. The EEG was normal, but the CT and MRI showed bleeding and excessive fluid in her head. Thank God that we had a doctor that was educated enough to order a full battery of tests, including metabolic. I have heard so many of your stories about doctors that didn't even know a metabolic condition was in the realm of possibilities. We were released on September 8, 2005 after 6 days. Still no answers, but seizures were the best diagnosis and the nurse said that we would be receiving test results over the next 2 to 30 days.

On September 13, 2005 around 6 pm, we got the call. I knew it was strange for our pediatrician to call us and especially that late, but he had always been great so I just shook it off and answered the phone. He said that one of the tests had come back abnormal. He told me (of course my husband and son were out getting dinner!) that Cheyenne had Glutaric Acidemia Type I. He told me all about what he had researched, but urged that I not look it up until we see a specialist. I agreed. The only metabolic doctor in Alabama happens to practice near us. She called that night and scheduled us to come in for a 2 day admission to have more tests run, teach us about the disorder and start her diet. I thought "This is nothing...no meat, no problem! Wow, was I wrong! I don't think it ever really hit me until we got home and I began to read through all of the pamphlets and books we had been given. We had to switch to Similac (my breastfeeding was out the window...I cried), Glutarex-1 began being shipped and the talk of G-Tube surgery began. Our metabolic doctor still does not believe the apnea seizures are related to Cheyenne's GA-1, so we are blessed to have found this disorder.

Cheyenne has only had 1 admission for metabolic issues. She was vomiting (not extensively), but our doctor wants to be overly cautious because she thinks that Cheyenne's condition was caught before any major damage was done. We had the G-Tube placed soon after this admission when she was only 6 weeks old. She eats so well by mouth. During one of our check ups, our doctor recommended physical therapy (PT) services to keep her on track. We are provided these through Early Intervention. Our PT has stated that Cheyenne's movements are erratic but not excessive. She also has been right on schedule with her cognitive, social and emotional skills, and her motor skills were only slightly delayed...but she has made up for that already!! She only comes out to our house once a month and it is mainly for monitoring purposes; she will come more often if the need arises. Since the initial PT visits, Cheyenne has not shown any more signs of a movement disorder...YEAH!!

Cheyenne began solid foods on schedule with infant cereal and now eats just about every vegetable, fruits and even spaghetti! She loves food! As for medication, she is on carnitine, riboflavin, zantac, pantothenic acid and co-enzyme Q10. She is allowed 6.5 grams of protein per day (soon to increase to about 10 when she is a year old). We only use her g-tube to feed her

once a day for medicine and to keep her & us used to using it. Our doctors are so impressed that she eats so well by mouth! She weighs almost 21 pounds and is 31+ inches tall with big blue eyes and the blondest hair—like a little angel.

Overall Cheyenne's development has been typical. She smiles, laughs, plays and loves so much! She adores Cody and he is very protective of her. I believe that they are best friends. The dogs, Chloe & Chyna, are even in on the action and she loves to play with them. She just got her first pair of shoes and will be walking any day now! I am thankful that we do not have any major medical, financial or emotional difficulties. My husband and I both work full time. I work for the Shelby County School System in the Special Education Department and he is in the building industry. My parents, sister and in-laws take turns keeping the children during the week so we have a wonderful support system. We all wear the NBS bracelets. Every time someone asks, we are eager to tell our story. We are blessed to have Cheyenne's condition caught even without NBS and so early in life. With the tools to manage her disorder, we feel that we are already ahead of the game. As she continues to meet her milestones, we continue to be cautiously optimistic and enjoy every day we have with each other and our wonderful, beautiful children!

Thank you for listening and all of your feedback from the list serve. This support has been enlightening, heartfelt and appreciated!

from: <http://www.oaanews.org/Cheyenne.htm>

Christopher Palladino, MMA, Cbl C, Age 4-1/2—Newborn Screened!

After 4-1/2 years, I'm so happy to be able to tell Christopher's story. He has been such a blessing in our lives and continues to lead us to God. The first year and half was difficult, but with anything, time heals and the sun eventually comes out from behind the clouds. Life has never been so bright.

Christopher was born on August 2, 1999 on the most beautiful summer day. The next day was my 30th birthday. Having him was the best present I could have received. Three days from his due date, he was ready to meet the world, but his heart rate wasn't tolerating labor. The next we knew, we were heading into the operating room for an emergency caesarean. We were relieved when he came out kicking and screaming. He was very alert and the doctor scored him a nine on his Apgar. We all breathed a sign of relief.

The next four days in the hospital were spent learning how to care for our new baby. We were both nervous first time parents. Luckily, the nursing went well aside from some latching problems and he woke often to eat.

The first few days were a bit scary and we were happy to have our follow-up appointment at the pediatricians. I wanted to see if we were doing things right. We left feeling confident things were going the way it should.

We arrived home to a message on our machine from our pediatrician. It sounded urgent and we called him back immediately. I couldn't understand what could be wrong, because we were just there and everything was fine. He said one of Christopher's newborn screenings came back positive. I wasn't even sure what he was talking about since I just opted for all the tests the hospital provided to be safe and really wasn't paying a great deal of attention to what they were all about. We wasted no time retesting him and now all we had to do was wait for the results. We were told that it was possible it could be a false positive and hoped they were right.

The next few days were pretty frustrating. We tried to take our minds off of it, but I was not successful. The stress was causing me trouble nursing him, so I started to pump to help the situation, but the only thing I would think about was that something was seriously wrong. The phone rang after three days and we were told he again tested positive. He couldn't tell us much since he didn't know what he was dealing with. He re-

assured us that the best doctors in the world were at Children's Hospital in Philadelphia and they would be there waiting for us to help.

The NICU was frightening. There were so many tiny, sick babies and now ours was among them. The doctors wasted no time and began testing and monitoring him. I remember watching in tears as they tried to get blood from a small vein in his head. This lasted for over fifteen minutes and I had about all I could take. Our parents had some with us and thankfully could take turns being with him.

We still weren't told much, but everyday we learned something new. First he had elevated



Christopher, MMA (right) with sister, Maria

homocystine and methionine and he had a disorder called MMA. He would be getting B12 shots along with amino acids until we knew what type of MMA he had. The results wouldn't come back until the fibroblast could grow and be tested. He turned out to be a B12 response so he was treated as though he had Cobalamin C until we knew for sure.

Over the next couple of days we learned how to mix his meds and formulas, and how to give an injection. The nursing was put on hold since his diet had to be monitored and quite honestly, I was too much of a basket case to continue. The last

day we were getting ready to go home and anxiety set in. How was I going to take care of him? We decided to take it day by day. Our parents were with us every step of the way, keeping us calm and helping in any way they could. We are so very lucky that we still have their support today.

Christopher was eating well. He was gaining weight and growing fast despite projectile vomiting. Those days were so frustrating. We didn't know if he had reflux or allergies, or why he was vomiting. We didn't want to give him any more medication so our doctor suggested adding cereal to his formula. That didn't help and seemed to make him colicky. We switched to soy and saw a little improvement. We were happy for even the smallest gifts.

For the next two months, Christopher's weight stayed the same and he began to refuse his bottle, taking an ounce or two at the most. I dreaded taking him to his weight checks. My mother-in-law came over often to help and we'd try different things to get him to eat. We changed feeding positions and tried other rooms in the house. There probably wasn't anything we didn't try.

Nothing helped and his eating continued to decline until he got sick at 6 months with RSV and stopped drinking altogether.

Because of the RSV and his refusal to eat, we ended up back at CHoP. We spent over a week there getting used to the NG tube and seeing several specialists. Someone different was in every day. We saw an ophthalmologist who brought our attention to his nystagmus and retinal scarring. They weren't sure if it was progressive and only time would tell. That was a pretty big bomb dropped on us. We were already feeling defeated having to use the nasal-gastric tube to feed him. We were in need of help and many prayers. I'm sure that's what got us through.

We were soon home again getting used to our new life with a NG tube, a feeding pump and a nebulizer. We were having a harder time dealing with the possibility of him losing his eyesight. We contacted Early Intervention and began services to prepare for whatever the future would bring.

It was wonderful to see Christopher progressing developmentally. He was sitting at 6 months and seemed to be using the vision he did have very well. With the help of therapists he seemed to be right on target with his cognitive and gross motor skills. His fine motor was mildly delayed and feeding skills ended up being the biggest delay to date. His vomiting continued and his oral intake was dwindling down every day. This became our toughest battle yet.

When Christopher was ten months old he had a severe allergic reaction one day when I kissed him after eating cheese. His face swelled and he had hives all over him. When we took him to the metabolism clinic they suggested seeing an allergist. Skin testing were done and he showed positive results to milk, egg, peanut, and soy. That was news that left us sick to our stomach since we had been feeding him this for almost 11 months. We hoped this new information would be the reason for all of the vomiting. He was switched to Neocate and his vomiting decreased tremendously to being occasional.

Now that we seemed to find the source of our problem, we started to make plans on getting him to eat. I joined feeding email support groups and the g-tube list to get as much info as I could. We decided against a behavioral approach because we felt that wasn't his problem. Kluge Rehab in Virginia was recommended by so many on the feeding group that we decided to try it out. When Christopher was around 15 months and just starting to walk, we flew to Virginia.

At Kluge we worked on getting him to drink his formula. After a week it looked as though we were making progress, unfortunately, this was short lived and only lasted a month or two. He had been put on medicine to

increase his appetite (I can't recall the name), but as soon as he went off it, he went back to normal

We arrived home right in time for Christmas and our big move to a new house on New Year's. We considered this a new beginning for us. We were moving away from our families, our biggest support and now had to rely on each other during the rough times. We adjusted well and we all loved our new home. Our move brought us closer to CHoP too, right in time of start another feeding clinic there. I didn't want to use a behavioral approach, but the opportunity presented itself, and we had to at least try it out.

We arrived at CHoP for their intensive day treatment expecting to stay two weeks and after that he would be eating. But when the two weeks were up, we weren't any better off than when we came. If anything, he seemed more determined not to eat. We decided to stay another two weeks, but that too proved unsuccessful. Christopher again was eating nothing by mouth. We tried everything we could, and I was finally ready to let it go.

Christopher's development was taking off. He was using two- and three-word sentences and running and playing like any other kid his age. He loves cars and pretend play and being the boss. He has so much energy and is such a happy child. We couldn't be more proud of how far he has come. Just as things were starting to get a little predictable, we found out I was pregnant. Worries surfaced over the possibility of our new baby having Cobalamin C. We decided to see a genetic counselor who suggested an amniocentesis to see if she had the disease. After much prayer, we decided not to have the amino since wouldn't change anything. Luckily, Maria was born healthy and now Christopher had a new sister to share mommy and daddy with.

Right before Maria's birth we moved again to a bigger house and have seen our prayers answered. Christopher is doing so well. He's an extremely active four year old full of questions (many of which I can't answer). He loves cars, books and playing with his grandparents. He gets to see them every week. He still doesn't eat, but our hopes are that, in time, he will. We have begun home schooling and I'm so impressed with his development.

We have so much to be hopeful for in the future. Our family has become stronger and our house is filled with love. I thank God for every day that he's in our lives.

Robin & Ken Palladino
1764 Evangate Drive
Allentown PA 18104
610-433-3599
rpalladino683@pngusa.net

PROP family story

Gabriel L.

Hi, our son Gabriel was born in London. He was diagnosed with propionic acidemia at 2 weeks of age after spending his first hours of life hyperventilating and with severe acidosis and high ammonia. Luckily, the medical team at Great Ormond Street Hospital for Children was very good at stabilizing him and at diagnosing him very quickly. Despite never showing actual fits, Gabriel was then diagnosed with infantile spasms (a type of childhood epilepsy) at 5 months of age after deteriorating progressively. We were again very lucky that he responded well to treatment with vigabatrin. He was weaned off medication at one year of age and has been seizure free since. At that same age,



he stopped eating by mouth completely and a g-tube was inserted in his stomach for feeding. His formula currently comprises pediasure, polycose, XMTV1, vitamins and flax oil. He is been very stable metabolically for the past two years and is been followed up by the metabolic team at Children's Hospital of Philadelphia every six months.

Gabriel's development was very slow until 18 months of age but he has made tremendous progress since he started therapies with the Early Intervention Program when we moved to the United States. He just transitioned to a special education pre-school program where he continues receiving OT, PT and ST. Since he started school, his gains have been really amazing. He just started talking and is able to comprehend perfectly and speak words in all three languages he hears. He has also gained cognitively and matured a lot. He has still a lot to catch up being his major challenges his low tone and his speech but we embrace his enthusiasm and effort and try to offer him as much support as we can.

We'll be happy to hear from other parents with children with PA and to share any useful information we may have.

Sincerely,

Marisa Cotrina and Juan Carlos López
Parents to Gabriel López, 3.9 PA

from: <http://www.pafoundation.com/Gabriel.htm>

CPT-1 family story

Michael and Crystal's Story ~ Carnitine Palmitoyl Transferase I (CPT 1)

November 17, 1995 was a day we will always remember. Our son, Michael Wurz, who was 2 years old on November 13, became ill during the night with diarrhea. We let him sleep in the morning until 9 o'clock and when we tried to wake him, he was unresponsive and rigid. We rushed him to the hospital 40 miles away [we live on a farm 40 miles from town] where they did a little blood work and right away called an air ambulance to transport him to a bigger hospital 100 miles away. The doctors did a lot of blood work and different tests to determine what was wrong ~ **the symptoms had pointed to Reye's Syndrome, but with some inconsistencies, which led to some doubts about what was really wrong.**

Our doctor, who was very kind and helpful and did his very best to treat Mike, did talk to us about metabolic disorders, but was unable to come up with anything other than Reye's Syndrome. Mike was in a coma for 10 days and was not expected to live, but **after 2 weeks he started to show some improvements.** However, he was very weak, his muscles had deteriorated, and he was not able to hold his head up, much less walk like he was able to before. Mike, who was in the hospital for 4 weeks, started showing seizures before he was discharged, which he still has occasionally even though he gets medication.

Since we were told Mike had Reye's Syndrome and had partially recovered, we felt all he needed was to recover his strength and he would be the same happy child he had been before. but we were wrong.

Three months later on March 13, after a single episode of diarrhea, **he was back in the hospital with severely low blood sugars.** After they were brought back to normal, he quickly recovered, but left the doctors scratching their heads. Even after still more blood tests, nothing new was discovered. After this we started monitoring Mike's blood sugar levels that were normal except when he was not feeling well which caused us a lot of anxiety.

The doctors changed their minds about Mike's illness, when a year later on February 15, 1997, our daughter Crystal became very ill with some of the same symptoms that Mike had his first episode such as low blood sugars and weakness in her muscles. Tests were still inconclusive as to what was wrong.

After doing a skin biopsy on Mike in April 1997, which was sent to the Institute of Metabolic Disease in Dallas, Texas, it was discovered that **Mike had a metabolic disorder called CPT I and because Crystal had had much of the same symptoms, it was concluded that she also had the disorder, which was later confirmed through DNA testing.** There were only a couple of known cases of CPT I in the USA at the time, which explains why the doctors couldn't make a correct diagnosis.

After her illness Crystal had a slight limp for about a year and her muscles seemed to be weaker in her right leg up to 2 years later, but now is healthy and strong and seems to be keeping up with her classmates. Mike's episode left him with seizures, poor muscle tone and developmentally delayed. He is going to school and seems to be learning, but very slowly.

In October 2000 **both kids participated in a dietary therapy study at Baylor University Medical Center, done by Dr. Charles Roe and Dr. Jay Cook, which involved a low fat diet supplemented with a special oil called Triheptanoin. Testing done on both proved it to be beneficial.** In school we are seeing very positive results with Mike. He is more attentive and is able to concentrate better but still has a lot of problems.

We have an **older son, Patrick, who does not have the disorder and our youngest, Benson, does not have the disorder but is a carrier.** In both Mike and Crystal the disorder didn't seem to affect them until after their second birthday ~ between the ages 2 and 4 Mike was in the hospital 10 times, once with pneumonia, then he had to have his appendix removed. Not enough food intake usually led to hospitalizations, but since 4½ years of age, thankfully, we haven't had any illnesses where he had to be hospitalized. The disorder didn't seem to affect Crystal as severely as Mike, as she never got as sick as he, nor as often.

After a few episodes of 'hypoglycemia' we learned to watch them carefully whenever they aren't feeling well, and to try and keep their intake level up, which means getting up during the night and giving them juices or Gatorade.

The monetary costs have been very high for both Mike and Crystal, but the real heartache was seeing them sick and not knowing what was wrong or what to do about it, but as doctors learn more about these disorders, they should become easier to manage. **The expanded screening that is now available at birth, for 30+ metabolic disorders, is something that every parent should be made aware of and requested for their child, for their own peace of mind and for their child's well being. *(Please note ~ Mike and Crystal had diagnostic FOD testing done and it is a different test than the supplemental NBS test).** Hopefully in the near future hospitals will be required to get this screening done on all newborns.

Ben and Debbie Wurz

from: http://www.fodsupport.org/michael_crystal.htm

Metabolism Meltdown: When Protein Proves Toxic

Dawn Cassada, HMG, Age 6 and Krystopher Arnold, MMA, Age 6

By Loretta Coureas

Reprinted from Kidstuff magazine with permission from Children's Hospital of The King's Daughters, Norfolk, Virginia

NORFOLK, VA... At 4 months of age, Dawn-Eagan Cassada lay in a coma in a Pennsylvania hospital.

Her mother was visiting relatives there when Dawn-Eagan became desperately ill and was rushed to the Pittsburgh hospital.

Doctors struggled to find out why she was so sick. The baby looked like she was suffering from a virus, one most children would shrug off in a few days.

But the doctors knew the true culprit must be something far more complicated.

It took a skin biopsy and other tests for them to learn that the protein in her baby formula was making her ill.

The root of her illness was a deadly inherited metabolic disorder. It is so rare, affecting one child in approximately every 100,000 births, that it isn't one of the disorders routinely screened for in newborns.

It's called *3-hydroxy-3-methylglutaryl-CoA lyase* deficiency.

"Don't bother trying to pronounce it," said Dawn-Eagan's dad, Chris Cassada of Virginia Beach, Virginia. "Just call it HMG."

Dawn-Eagan, who is now 6 years old and lives with her father and grandparents in Virginia Beach, was born with the particular genetic combination that -- like Russian roulette -- sometimes does and sometimes doesn't produce HMG. It means her body can't metabolize leucine, which is just one of the many amino acids found in all protein.

Her father describes how HMG affects Dawn-Eagan: "When she eats foods that contain protein, instead of providing nourishment, the protein deposits toxins in her tissues. Too much protein and she gets very sick. When a virus comes along, she could end up in a coma or worse."

Baby formulas contain protein. So, from the day she was born, Dawn-Eagan's feedings were actually building toxins in her body that put her on a collision course with a common virus.

The medical books predict childhood death in undiagnosed cases of HMG. So this bright, pretty girl is one of the "lucky ones," according to medical geneticist Virginia Proud at Children's Hospital of The King's Daughters in Norfolk, Va.



"If HMG hadn't been discovered when it was," Dr. Proud said, "she could have died or become severely handicapped. But we expect her to have a normal life -- as long as she watches what she eats for the rest of her life."

Diet is the key to her survival. Dr. Proud explained that to keep her healthy and growing, Dawn-Eagan's family must make sure she strictly limits the amount of protein she eats. Since proteins are essential for growth, she must eat some small helpings of protein and supplement her diet with specially formulated "medical food" prescribed by her nutritionist.

"She can do well on a basically low-protein vegetarian diet," says CHKD nutritionist Melody Persinger Yeargin. "But she needs other things to help her grow. The medical food she gets daily ensures that she will have a balanced diet, regardless of what else she eats."

Everybody in Dawn-Eagan's household -- including her grandmother, Kathy Cassada, and her Aunt Jennifer and Uncle Bill, know they must monitor, measure and log everything the child takes in daily. They make sure they know the exact amount of leucine contained in everything she eats, being careful not to exceed the maximum amount she can tolerate. An excess could result in damage to her heart, brain, lungs, kidneys or liver.

The nutritionists at CHKD arm the families of children with metabolic disorders

with enough knowledge to detect offending amino acids lurking in foods. "Keeping the child's body nourished with the proper balance of nutrients on a very low-protein diet can be a full-time job for the parents," said Yeargin, who specializes in working with children with metabolic disorders.

"Their basic 'food' is the prescribed medical food they take daily to make sure they get the carbs, fats, partial proteins, vitamins and minerals they need. Each patient's formulation is specific to his or her problem. But children need to eat other foods as well."

Dawn-Eagan is learning what she can and cannot eat.

"Tomato soup, croutons and cheese," Dawn-Eagan says when asked what she prefers. "No meats," she adds.

"She knows meat can make her sick," her dad said.

Tiny liver transplant patient goes home

Determined parents, dedicated hospital staff and luck made 3-month-old Sydni a survivor

BY TIM BONFIELD
The Cincinnati Enquirer



Judy Quinlan cuddles her daughter Sydni in her room at Children's Hospital Medical Center. (Glenn Hartong photo) | ZOOM |

More than 200 patients have received liver transplants at Children's Hospital Medical Center since 1986, but only three of those children were smaller than Sydni Quinlan.

After surviving at least two near-death crises and undergoing intensive care for a good chunk of her first three months of life, Sydni went home to Batavia this month cured of a rare genetic condition that doctors almost overlooked.

Sydni owes her life to determined parents, a small army of experts at Children's Hospital, and a large dose of luck.

"We almost lost her twice," said Sydni's mother, Judy Quinlan. "I'm here to tell you that when it comes to taking care of kids, all hospitals are not the same."

To Mrs. Quinlan and her husband, Rick, the ordeal offers a fresh reminder about the importance of an age-old medical precept: getting a second opinion.

Sydni was born Oct. 28, the apparently healthy third child of the Quinlans. Two days later, the couple took the baby home from the hospital.

On the third day, Sydni couldn't be awakened.

"She was cold. She wasn't breathing right. Her arms, her face, her legs were ashen white," Mrs. Quinlan said. "We called our pediatrician, who told us to take her to the emergency room, right away."

Mrs. Quinlan declined to say which hospital they went to first. "It's probably a very good hospital. I don't want to hurt its reputation," she said.

But from the start, Mrs. Quinlan said, the emergency staff struggled with Sydni's case. She said it was a Saturday night and no one in the emergency room felt comfortable enough to draw blood from an infant, so a labor-and-delivery staff member was called to assist.

"Then, the local ER didn't find anything wrong enough with Sydni to keep her, so they sent us home," Mrs. Quinlan said. "They told us repeatedly that whatever she had wasn't life-threatening ... that we should call the pediatrician again on Monday, and if we were still concerned we should go to an urgent care center."

"They thought we were new parents who were overly concerned," Mrs. Quinlan said. "So we left."

That was about 3 a.m. By 6 a.m. Sydni was getting worse. "She was breathing like a little goat, just bleating," Mrs. Quinlan said.

The couple tried calling the urgent care center, only to find out the office doesn't open until 8:30 a.m. So they called their pediatrician again. This time, the doctor told them to go straight to Children's Hospital.

"When we got there, the doctors practically ran her to the trauma room," Mrs. Quinlan recalled. "She stopped breathing a few minutes after that."

Sydni had lapsed into what appeared to be septic shock, possibly from a bacterial infection that had run out-of-control. The emergency staff at Children's revived Sydni, stabilizing her enough to move her to neonatal intensive care, where she was placed on a respirator.

That was just the beginning. Soon, her lungs began to hemorrhage. Later, her liver began to fail. She would go through six dialysis treatments to cleanse her blood.

Fighting the shock symptoms made it hard for doctors to spot Sydni's disease. The picture came into focus on the third day in intensive care, when a blood test revealed a sharp spike in Sydni's ammonia levels, which can cause brain damage.

The test helped doctors determine that Sydni had citrullinemia, a genetic condition that occurs once in every 50,000 to 100,000 births. Patients with this condition have a liver that cannot process ammonia, a waste product created as the body breaks down proteins from food.

Sydni's condition is so rare that doctors at Children's Hospital wouldn't expect it to be detected at any community hospital emergency department.

It can take two or three days after birth for the ammonia to build up to health-damaging levels, said Dr. Nancy Leslie, a geneticist at Children's Hospital who worked on Sydni's case.

"For some of these kids, it's like falling off a cliff," Dr. Leslie said. "They can look sort of OK for a while, then just hours later they look awful."

Once diagnosed, Sydni's parents faced two choices.

They could skip surgery and put Sydni on a diet that sharply limits protein intake to keep ammonia levels low. Then doctors would use various medications and treatments to cleanse the blood during flare-ups. The risk to this approach is that every serious or long-lasting spike in ammonia levels would further damage her brain, and possibly kill her.

Or they could try for a liver transplant. A successful transplant does cure the genetic condition. But the risks include waiting for a donor, surviving the operation, then spending a lifetime on immune-suppressing, anti-rejection medications that carry risks of triggering infections, cancer and organ damage.

The prospect of a real cure led the Quinlans to choose the transplant. For the next several weeks, Children's Hospital nutrition experts custom designed a diet to make Sydni gain weight as fast as possible while controlling ammonia levels in her blood. The bigger she was, the better her chances of surviving transplant surgery.

Then, luck kicked in.

Some patients can spend years on the waiting list for a liver transplant. The disparities in waiting times are so sharp from region to region that transplant centers, patients groups and regulators spent much of last year fighting over ways to reform the system.

Sydni waited just four days. She went on the list Jan. 15 and was in the operating room on Jan. 19.

Her case had a high priority because of the risk of further brain damage, said Dr. John Bucuvalis, the liver disease expert who put her on the waiting list.

Even so, it was sheer coincidence that a 22-pound child died in Galveston, Texas and was close enough in size to Sydni to make the transplant possible.

Transplant surgeons Maria Alonso and Frederick Ryckman used the left lobe of the donated liver to fit inside Sydni's 8-pound body. The smallest baby to get a liver transplant at Children's Hospital weighed 6.5 pounds.

Even though the organ was trimmed down, it will grow as Sydni grows.

"Every time I look at Sydni, I think about that other child," Mrs. Quinlan said.

Despite the rare circumstances in Sydni's case, doctors and Mrs. Quinlan said they drew two lessons from the ordeal.

First, parents should trust their gut feelings about their children. Parents know — often better than doctors — when something is wrong with their child.

"If you are not comfortable with a doctor's opinion, get a second opinion. Don't wait," Mrs. Quinlan said. "Our waiting may have caused even more damage."

Second, that Greater Cincinnati is lucky to have Children's Hospital. Mrs. Quinlan marveled at the sheer number of health professionals involved in Sydni's care: trauma specialists, liver experts, transplant surgeons, geneticists, nutritionists, entire teams of nurses and technical staff.

Dr. Deborah Borchers, Sydni's pediatrician, said she often recommends that parents with sick children under 2 years old go to Children's Hospital, even if other hospitals may be closer.

"Without slamming any other hospital, my opinion is that Children's Hospital is one of the top five or 10 pediatric hospitals in the country," Dr. Borchers said.

DONATIONS

Medical bills for Sydni Quinlan reached \$86,000 before she received her transplant, which is expected to cost more than \$300,000. Medications could cost \$20,000 a year.

Her parents' medical insurance will cover most of Sydni's medical bills. Even so, a family friend has established a fund to defray uncovered costs. Send donations to:

"Cents for Baby Sydni Fund"

P.O. Box 453

Batavia, Ohio 45103.

**ASA
family story**

Dylan Clark



Dylan was born on October 22, 2003. He weighed 8 pounds and 3 ounces and was 21 inches long. He was our first child, and the pregnancy and birth were normal. Dylan was eating without any issues and showed no signs of ASA. We received the call when he was 6 days old that there were abnormalities in his newborn screening and we needed to see a specialist that weekend. This was the beginning of an education on ASA as well as how to get around insurance. Further tests confirmed that Dylan did have ASA.

Dylan is now 2 1/2 years old. If it was not for the monthly blood test and strict diet, you would never know he had ASA. We have been lucky that his case is not as serious as others. His levels have never been elevated to a point of concern. He now sees the specialist every three months. His development has been a little behind the curve. He

did not walk until he was about 13 months old (not that late, but his little brother walked at 10 months). His speech is the area he is most behind. He still talks in 2 or 3 word phrases (note I did not call them sentences).

He loves Thomas the train and baseball. I am not sure how those go together, but he is always asking to watch one or the other.

As I mentioned above, Dylan has a little brother, Brady, who is now 1. Brady does not have ASA or any other known conditions. Dylan is extremely protective of his brother in public and beats him up at home (typical brothers).

Dylan's physical growth has been normal, in fact he has always been tall for his age. He is as happy and healthy as any other little boy. We are very lucky to have caught his condition so soon and to have such great medical help!

from: <http://www.geocities.com/oliphint4/Dylan.html>

TFP **family story**

Stephen's Story by Diane and Val Nielsen

Stephen Nielsen was diagnosed with Trifunctional Protein Deficiency (TFP) at the age of 8 months. Prior to a nearly fatal crisis brought on by the common flu, abnormalities in Stephen's behavior and development were not so unusual to warrant great concern. Mild developmental delays had been noticed and discussed with Stephen's pediatrician. The rapid decompensation due to metabolic crisis came as a complete surprise. Despite a diagnosis after only two days, Stephen did not immediately respond to treatment. He quickly decompensated, suffering from extreme weakness, liver and brain problems, severe lung failure and partial heart failure. Stephen spent 3 ½ weeks on life support, part of that time on ECMO, a type of heart/lung bypass. He came within hours of death. After nine weeks at Children's Hospital, Stephen returned home to our grateful family. His strength and development were back to about a three-month old.

Recent testing revealed that Stephen is an extremely bright child, but has some difficulty holding working memory and some challenges in small motor skills. He is still a little weak in gross motor skill compared to his peers, and has some permanent damage to one of his eyes. But to look at Stephen now, you would never know he has had such difficulties. His current good health is the result of years of therapy to "catch up" and vigilance by family members to follow the diet protocol. Today Stephen is a bright, energetic kindergardener full of ideas about the world. He delights in teasing his three older sisters and keeping things lively in our home.

TFP is a mitochondrial disorder in which the body is deficient in enzymes required to break down fat for energy. It is commonly initially diagnosed as LCHAD, or Long Chain 3 Hydroxyacyl CoA Dehydrogenase Deficiency. Fat builds up, becomes toxic, and fasting for even three or four hours may cause hypoglycemia. In infants, awareness is critical, as the newborn's body has fewer back-up resources for energy. Supplementation of antioxidant vitamins, a very low fat diet, a special oil containing fat Stephen can burn for energy, and avoidance of fasting keeps him healthy. It's not difficult, but is essential.



3MCC family story



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Seth

This is a personal story shared by a family whose child has [3-Methylcrotonyl-CoA Carboxylase Deficiency \(3MCC\)](#).



Our son Seth was born on November 26, 2002, healthy, beautiful and responsive. We would never suspect anything to be wrong with him until we got the call from the Metabolic Genetic Clinic when he was five weeks old. At the time of the call Seth had a bad cough, which eventually developed into Respiratory Syncytial Virus (RSV).

At the time of Seth's birth the California Expanded NBS research study was taking place. I was aware of this study, so I had signed the consent to have the extra testing done. The clinic contacted us with a sense of urgency to bring him in within 24 hours. I found this information disturbing, and my husband and I were skeptical. The following day after the call we brought Seth in and the same test was repeated. The results took five days to process, and in the meantime Seth was admitted to the hospital with RSV. Seth's doctor, Susan Winters, called us at the hospital (she was out of state at the time) with the lab results. It was confirmed that this was not a false positive, and the doctor ordered Carnitine and Biotin immediately. He was able to go home two days later, recovering well. The results of the two series of labs did not make total sense to the medical team, so they took a biopsy for further enzyme testing. Based on the biopsy results, they identified Seth's diagnosis as a partial deficiency of 3-Methylcrotonyl-CoA Carboxylase Deficiency (3MCC). I have read so many unfortunate, sad stories of affected children. This is a happy one in that Seth is normal, healthy and has no disabilities due to his disorder. It was discovered only by the expanded testing which is now state mandated. My son has a delightful personality, and he is bright and beautiful. His life was saved, and his quality of life will be normal because of the expanded newborn screening. Thank you for your efforts to ensure that all children have the appropriate NBS testing.

Sincerely,

Brenda, Seth's Mother

<http://www.savebabies.org/familystories/Seth3MCC.php>

TYR-I family story

http://www.joshuascore.org/

Go Bookmarks 1 blocked Check Look for Map AutoFill Send to



Joshua Holdner 2007

Video
[Joshua's story - view the video](#)
by Duke Children's Hospital



Read about Josh in Duke Children's Hospital newsletter



Pictures Courtesy of Duke Children's

Welcome!

News

cutting / Silent au

→ [Learn more about "Joshua's house" - 23 August ribbon cutting](#)

Duke Children's Champion 2006 ~ Josh Holdner

Josh was chosen as the 2006



Champion for Duke Children's Hospital. [Click here](#) to read the story.

About Tyrosinemia Type I

Tyrosinemia Type I is characterized by a lack of the enzyme FAH (fumarylacetoacetate hydrolase), which is needed by our bodies to break down the amino acid Tyrosine. Failure to properly break down Tyrosine, resulting in an accumulation of this amino acid in the body, has adverse effects on the liver and may eventually cause severe liver disease. High levels of Tyrosine may also accumulate in the kidneys and central nervous system.

Symptoms associated with Tyrosinemia Type I include failure to gain weight, unusually slow physical development, jaundice, fever, diarrhea, vomiting, and an enlarged liver. A child diagnosed with Tyrosinemia does not usually live to his 10th birthday. A liver transplant could be the only known treatment for this disorder, and because of its complex

This site is about a boy, a devastating disorder, and hope. The boy is Joshua Holdner, now 9 years old. The devastating disorder is a rare genetic metabolic disorder known as Tyrosinemia Type I. Hope comes from those of us who know Josh and his family, and all the volunteer efforts to raise money for research of this disorder. And now, maybe, from you too.

Joshua Holdner is one of approximately 100 children in the United States—and the ONLY one in the state of North Carolina—to have this rare genetic disorder.

Get a more personal look at Josh and his family by viewing [this video](#) done by Duke Children's Hospital, as well as the [photo gallery](#). And be sure to check out the [events](#) page.

What you can do

Frankly, we need your money along with your prayers. To date, there is little research available. This is where you can make a difference.

- Attend our fundraisers—visit this site often to learn what activities are planned. See [events](#).
- Go to [donations](#) to donate online, or send your tax deductible donation to:
Joshua's Cure
P.O. Box 1106
Cary, NC 27511-1106
- Learn more—navigate this site using the links across the top and down the left side, in addition to any links within the pages. The skateboard photos above link to a PDF₁ file that provides more information.

Add your hope to ours, and your dollars to ours, to help us learn how to save these young lives. Our efforts are voluntary, so all your donation dollars go to research. Keep Joshua, his family and the other children suffering from this disorder in your thoughts and prayers. Together we can all make a difference!

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