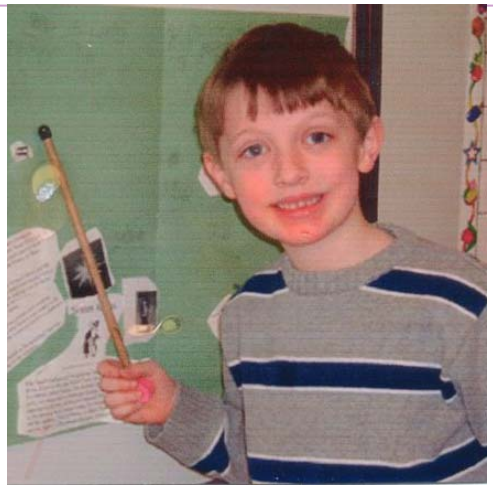
Save Babies Through Screening Foundation, Inc.

Alex

This is a personal story shared by a family whose child has Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCADD).

Alex's Happy Story



When I tell people in the metabolic disorder community Alex's happy story, I get a reaction synonymous to that of, "I just had a baby a week ago. I have 5 nannies and a personal trainer. I sleep 9 hours a night, uninterrupted, and I work out 2 hours a day. I'm already losing weight." This is a luxury afforded to a rare few. So is the inexpensive comprehensive newborn screening with tandem mass spectrometry, which Alex had at birth.

Our first child, Alex, was born October 1, 1998. He had his first well-baby visit when he was 5 days old. During the appointment, the pediatrician continually raved about him. Whatever a 1-week-old was supposed to be doing, he was doing all that and more. Alex did so well we were told we could skip his 2-week well-baby visit. I'm not bragging, I'm making a point about how silent MCADD is. One hour later, the pediatrician phoned and told us his newborn screening indicated he had MCADD. We were told to immediately call the endocrine nurse at Children's Hospital of Philadelphia. So much for being such a perfect baby.

The endocrine nurse explained MCADD and its treatment well - Alex can't convert fat reserves to sugar and ketones, and therefore he can't afford to fast. Avoiding fasting is normally not a problem for him, but can be a problem when he gets sick and can't eat. Since he may fast at times such as illness, including flu, vomiting, ear infections, and general illness, he may require occasional hospitalizations in which he'll need IV dextrose. When he does fast, he needs IV dextrose to avoid serious consequences including coma, cardiac arrest, and sudden death. We were given a strongly worded letter of instruction, to take with us to the hospital when Alex needs IV dextrose. This letter is necessary because the medical community is ignorant of MCADD and its crucial treatment. But Alex's prognosis is excellent!

Alex has never yet been hospitalized (as of March, 2000). Our goal was to keep him illness-free for one year. After that, we figured, when it happens, we'll just have to deal with it.

Most MCADD parents are not this lucky. Since MCADD is silent, and since MCADD is usually not part of the newborn screening, many children who have MCADD don't know it until after brain damage or death has occurred. I cannot tell you how fuming we would be if this had happened to Alex, and if we had later learned there is an inexpensive test to detect this silent disease presymptomatically!

We joined the FOD Family Support Group and I was absolutely appalled to learn that over 90% of the nation's babies do not receive the comprehensive newborn screening with tandem mass spectrometry that saved Alex's life. We're talking 4 drops of blood and $20, what's the big secret? I couldn't even believe it! My son was screened only because Bryn Mawr Hospital, where he was born, does it routinely on all the babies just because it's a good idea. It's not the law here. If Alex had been born in some of the other hospitals in my area, he would never have been screened. I thought surely someone's working on this problem to change it. I failed to find any formal movements to fight this problem, so I knew I had to do something to change it. I've been working on it for over a year and it has been quite maddening knowing that we FOD people hold the key to many medical mysteries, yet so many people, including family, doctors and pathologists, ignore us.

Last year we joined the Tyler for Life Foundation (now *Save Babies Through Screening Foundation*) so that we could help educate parents and medical professionals on expanded newborn screening available. Every child with MCADD and other fat oxidation disorders deserves the same bright chance our son has, and no one should ever have to lose a child to a disease that can be treated so easily.

Written April 2000 by Alex's Mother
Alex was born on October 1, 1998

UPDATE - Alex's First Hospitalization Went Well
Alex finally had his first hospitalization, and I thought people might be curious to hear how it went. Alex was screened at birth for 30+ serious metabolic disorders and that's how we found out he has MCADD. It is NOT law here in Pennsylvania to screen newborns for 30+ serious metabolic disorders, contrary to what some pediatricians and some metabolic specialists think. My hospital just does the screening routinely on all their newborns, and I sure am glad they did. (Some hospitals here do, some don't.) Alex is now 2 years, 4 months (as of February, 2001). He has been sick before with mild vomiting but he has never needed hospitalization, until January 31, 2001.

First let me make a point that while we do live in Pennsylvania, there are no Amish in our area. Suburban Philadelphia has about as many horse and buggies as the Catholic Church has female Popes - there aren't any.

Having said that, you're not going to believe this.

We appeared at the pediatrics unit in Bryn Mawr Hospital (the pediatrician calls ahead and has us admitted so we can bypass all that ER / fill out the paperwork / your kids looks fine / it's just a little vomiting / what is MCADD / have a seat and wait 3 hours stuff). I calmly and simply told the person at the desk in the pediatrics unit, and the nurse standing there, "My pediatrician called ahead. I'm supposed to ask for the pediatric doctor. My son has MCADD. It's life-threatening. I have a protocol letter."

OK, are you ready??

The nurse said, get this.. "Oh yes, we've had several little kids in here with these kinds of things."

SEVERAL??!

So I told her, "That's because this hospital screens them at birth, so they have a chance to prevent metabolic crisis."

Several kids, as in, screen them at birth so they can treat them early as per the protocol letter, in an ordinary localhospital like this one. Several kids, as in, metabolic disorders are not so rare when someone's looking for

them. Several, she said. As in, oh, I heard about this and I've even seen some of these things. This is not a metabolic clinic, mind you - this is just an ordinary local hospital with a pediatrics unit.

I believe her too because not one professional asked me anything about MCADD - just the normal, have you been here before for this, what's happening with Alex - normal questions. You'd think he was being treated for an ordinary boo-boo, honest to God. Everyone acted like they've done this before - and you know what, I think they have.

Treatment was an idealistic dream. I swear, we could have made the "This is how you treat these kids in ideal circumstances" training video. They admitted us immediately. They read the protocol letter, prepped him, hooked him up to the IV, and the pediatric doctor telephoned the pediatric endocrinologist on call at Children's Hospital of Philadelphia (where we go for consultation) to discuss whatever issues of concern to them. No, they are not metabolic specialists at our local hospital. It did not matter. Everyone knew how to follow directions and knew how to call the specialist.

The pediatric doctor at Bryn Mawr Hospital, who's been there since summer, told me they now keep a file on all these special kids they're seeing there. She told me they'd add Alex to the file, and next time I can just call the pediatric unit directly, let them know we're coming, and that we're one of the special cases in the file.

Alex was on the IV for about 24 hours. They unhooked him and let him go home after he ate his normal lunch. During a period of about 23 hours, he took only 1 ½ ounces of juice but that was OK because he was on the IV during most of this time. The unanswered question is why was he sick. Chris and I felt like we were coming down with colds but we did not actually get sick. Alex got a runny nose and congestion Tuesday evening, and he ate only half his normal food amount, but he did drink. On Wednesday he ate absolutely no food. He stopped drinking late Wednesday morning and a few hours later he threw up. That's when we left for the hospital. By the time we got to the hospital, Alex had a fever of 101.5 under arm. His sugar was 69, the low end of normal (implying we got there in time - you can do that when someone gave you newborn screening and two years' notice. The time elapsed between his first vomit and receiving IV dextrose was 1 hour and 45 minutes - that includes time driving to the hospital, parking, finding the pediatrics unit, prepping him, etc. They did admit us immediately. When we left the hospital the next day, his labs were still not back - they had analyzed a poopy diaper to see if he might have had a virus and if so, which one.

When these kids come off their long sugar trips, (i.e., are unhooked from IV), do they always bounce around like a nut, or is my little guy just really happy to be home? Actually, he did get sick the next day at home, but he did eat and drink enough to keep from having to go back in the hospital. He was sick for a total of 5 days, with hospitalization happening days 1 and 2.

I cannot even believe I might possibly be speaking with my pastor right now, making funeral arrangements, if my hospital had not spent frickin' $20 two years ago for a routine supplemental newborn screening test. Oh, and speaking of church, at least one family in my church has children with CAH (congenital adrenal hyperplasia, a serious, life-threatening metabolic disorder detectable through the $25 comprehensive newborn screening program at NeoGen). Treatment is different from MCADD (different stuff in the IV), but the hassles for the families are the same. They did not have the supplemental newborn screening, and they found out when their son had a heart attack as a baby.

My point is, metabolic disorders all together are not so rare - we are everywhere. You just wouldn't always know it by looking at the patients. Metabolic disorders are not an Amish thing, they're an all-populations thing. People just don't know it because they're not looking for metabolic disorders, much less keeping files on these kids at local hospital pediatrics units.

Well, isn't it important to know, and to push for, what is possible and doable. There's no reason every single hospital everywhere can't be doing what my hospital did.

UPDATE - July, 2006

Alex has now completed the first grade. He has been hospitalized four times, all between the ages of 2 ½ and 5. All hospital stays were less than 24 hours. Alex plays soccer, loves trains, drawing, camping, and playing tag, and is generally as active as any kid his age. Alex likes himself, and we are very proud of him. At his age, he understands that if he vomits, it’s very important that his mom or dad be called immediately because he can get very sick very fast. He also knows that eating sugar before bedtime “helps him not throw up”.