

**MCAD Communication Network**  
**'All In This Together'**

**December 1991**

**Volume 1 Issue 2**

**From The Editor**

First of all, Dan and I would like to apologize for taking so long in getting this issue of the newsletter out to you. We experienced an unexpected delay in the editing for the Medical Update section. We had hoped to have it sent out by September, but I guess it's better late than never! We will have two newsletters a year ~ one around May/June and the other in December/January. Thanks for being so patient.

Dan and I would also like to thank all of the families (15 out of 30) that returned the questionnaire enclosed in the first issue of this newsletter. Your family information, ideas, and suggestions were greatly appreciated.

We were pleased with the return rate, but we are still hoping that the remainder of the families will return their questionnaires. Enclosed you will find the MCAD Family List. We hope this will stimulate some networking with other families.

Several families suggested on the questionnaire to have a section of the newsletter devoted to MCAD families 'telling their story' about how MCAD came into their lives and how it changed their lives forever. In this issue, Dan and I will tell our story which began 6 years ago with the death of our first child, Kristen.

Also included in this issue is a Question & Answer section on MCAD issues, Pharmaceutical News, and a Medical Update on current MCAD research. Again, we hope that you find this newsletter interesting and useful in your efforts to cope with MCAD. Remember, 'We Are All in This Together!'

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**Out of Death Came Life**

The afternoon of June 12, 1986, in Champaign, Illinois, was a joyous yet sorrowful time in our lives. As we held our newborn son, Kevin, tears streamed down our faces. They were tears of joy, celebrating the miracle of life and also tears of sorrow mourning the tragedy of a child's death. Eleven months earlier, Kevin's sister, Kristen, a sister he would never experience growing up with, died suddenly for no apparent reason.

On Sunday, July 21, 1985, Dan and I thought the world had come to an end. Our beautiful, happy, and healthy 21-month-old daughter was declared dead at 6:05pm. We

could not believe this was actually happening to us. The night before, Kristen was laughing and running through the sprinkler, and now she was motionless on the emergency room table.

That morning she had awakened with flu-like symptoms; forcefully vomiting several times. She had never been sick before, so I knew something was definitely wrong. I sat by her all day and kept in contact with the advisory nurse and on-call physician. They said the flu was going around and to make sure that she took fluids to avoid dehydration and to let her sleep.

Early that afternoon, Dan returned from a weeklong business trip. He knew something was wrong when a close friend picked him up at the airport instead of Kristen and me. He was disappointed that Kristen wasn't awake yet so he could give her a beautiful stuffed animal to add to her collection.

Two hours after Dan's return, I was getting very concerned that Kristen was sleeping too much and not taking enough fluids, so I called the nurse for advice. While I was on the phone, Kristen stopped breathing. The nurse called for an ambulance, and I yelled for Dan. We immediately began CPR. We both thought to ourselves, "This is NOT really happening!" What was even more amazing was when a police officer arrived, he looked at us on the floor doing CPR and accusingly (my perception) asked "What's going on here?" as if he was implying that we had done something to Kristen. Despite feeling frightened about what was happening with our daughter, I can still clearly recall my internal reaction and thoughts about this arrogant and uncompassionate officer ~ and it's not anything I can repeat here! The ambulance seemed to have taken forever to get to us. Unlike the officer, the paramedics told us that we did a good job restoring Kristen's breathing and pulse and that they would take over.

On the way to the hospital, I knew we had lost her, but Dan kept asking God to save her. The ER team did their very best, but 2 hours after being admitted, Kristen's ventilator was turned off and she died. To see our baby lying there motionless was heart shattering. We will forever remember those horrible feelings of shock, numbness, and indescribable gut-wrenching pain.

With the help of family and friends, Dan and I made it through the wake and funeral. The hardest time was when everyone went back to their homes and we were alone in our once cheerful home. Not being able to cuddle and kiss Kristen and hear her say mama and daddy was so very difficult. Even Samson, our yorkie, knew something was wrong. That HUGE 'blackhole' feeling was felt by all of us.

One week later, the autopsy results came in. The outstanding finding was fatty infiltration of the liver. After consulting several Drs., Kristen's death was determined to be from Reye's Syndrome (RS). Dan and I had a difficult time accepting that conclusion, because she had not taken aspirin nor had previous bouts of chicken pox or flu, which are typical characteristics of Reye's. Even after reading about RS, we still couldn't believe the

results. It's shattering enough to have a child die, but even more frustrating when you don't know why. And for us, RS was NOT the REAL why!

While struggling through the grief process, Dan and I made the difficult decision to continue our family. Seven months after Kristen's death and while awaiting our second child's birth, we received an article from my sister's pediatrician in Chicago. From conversations with my sister, the pediatrician was aware of Kristen's questionable autopsy results. When she read about a metabolic deficiency that mimicked RS in a Pediatric Journal, she immediately thought of us and sent the article.

Dan and I were certain that MCAD was the cause of Kristen's death and when our next child was born he or she was going to be tested. Our pediatrician contacted some RS researchers as well as metabolic experts, and they felt that Kristen's death could not be this newly discovered deficiency. We did not accept their opinions because they could not rule-out MCAD!

On June 12, 1986, Kevin was born and we were adamant about having him tested. After his birth, a blood and urine sample were sent to Duke University Medical Center and within 24-48 hours we received a call from Dr. Charles Roe ~ Kevin had MCAD. Our initial reaction was devastation. We thought we were going to lose Kevin, too.

When Kevin was 4-weeks-old, we flew to Duke Medical Center for 1 week to participate in Dr. Roe's research study. At the time, Kevin was the youngest child to be diagnosed with MCAD. Those of you who have had the pleasure of meeting Dr. Roe, know that he has a very calming way about him ~ even if you can't understand all the biochemical terminology! After talking with Dr. Roe, we felt somewhat assured that MCAD was treatable, that we had a better idea of what was going on inside Kevin, and that MCAD was the cause of Kristen's death. We wanted to know for sure though.

Fortunately, some of Kristen's liver tissue had been saved by the medical examiner, so Dr. Roe was able to test that and positively determine that she had MCAD. We now had a definite cause of death and not the nebulous NONdiagnosis of Reye's Syndrome.

To say the least, Kevin's first year or two were stressful. Once we got into a routine and had gone through some colds and fevers, we felt more comfortable about the illness protocol. Fortunately, we haven't had to use the intravenous carnitine we keep at the hospital, but if we have to, we feel confident that it will stabilize Kevin during an illness along with the glucose IV.

Seventeen months after Kevin's birth, our third child, Brian was born. All during the pregnancy I dreaded that he too would have MCAD ~ fortunately, he is only a carrier.

Kevin is now 5-years-old and doing well. He is a very big, strong, and active boy. His high carbohydrate and low fat diet probably play a part in his healthy look. Brian, almost 4, also adheres to our 'heart healthy' diet. However, we modify his diet by using 2% milk instead of skim, and regular yogurt instead of the nonfat kind, as young children need

some fat for normal growth. Smaller frequent meals and snacks are stressed (2-4 hours) and prevent Kevin from becoming hypoglycemic (low blood sugar). We are very well aware of how serious that could be for an MCAD child.

After 5 years of dealing with MCAD, Dan and I still have a concern that something could happen to Kevin, but we have it more in perspective now than when we first heard his diagnosis. We are all too aware that neither of our children is totally protected from injury or death ~ Kristen's sudden death SHOWED us that! Even though it gets stressful sometimes when he's sick, we feel much more confident of his positive prognosis for a healthy lifetime.

Dan and I often think about how we found out about MCAD ~ if we hadn't heard about it when we did, Kevin wouldn't be with us today. Of course, we would have liked to have learned about it in time for Kristen, but it was so new that most Drs. had never heard of MCAD before. We are hoping that will change through more funded research, more exposure at conferences, more physicians reading their journals, and ALL of you spreading the word to your families, friends, and any medical professionals that you come in contact with.

Kristen's death not only affected us personally, but also professionally. Dan is a Sport Psychologist at the University of North Carolina at Greensboro and has reaffirmed his commitment to studying children and sport as well as elite athletes. I have moved from being a K-12 Physical Education teacher to working privately with bereaved parents and their families. I am presently working on my master's in Counseling at UNCG.

Now that you have an idea of the 'Gould's story,' it would be terrific if we could print your MCAD story in this newsletter. This would be a great way for all of us to get to know each other. Who knows, maybe someday we'll have an MCAD family conference where we could all meet in person!

Writing about your MCAD child(ren) can also be very helpful in coping with the stress (among other emotions!) of being a bereaved parent and/or an MCAD parent. At least, it has been for us. So, if you wish to share your story, please send it to us for our next issue. You don't have to be a best-selling author to send your story ~ write from your heart and your story will be just fine.

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## Questions and Answers

[Please Note: This question and answer column is designed to answer questions, both medical and practical, on MCAD and its treatment. Answers to questions are solicited from those who have had firsthand experience dealing with MCAD. These include physicians, parents of MCAD children and children themselves. It is our hope to provide

general guidelines in responding to questions posed as opposed to specific foolproof solutions. Additionally, it is especially important to note that our Medical Advisor, Dr. Charles Roe, (at printing of this newsletter in 1991, he was at Duke University Medical School and now, in 2000, at Baylor in Dallas) has read and approved responses to all medical questions. However, because of the individual nature of each case, it is always important to discuss these guidelines with your physician before making any changes.]

**Question:** Our child was recently diagnosed with MCAD. How can I explain the seriousness of MCAD to relatives, friends, teachers, etc., without alarming them?

**Answer:** If you have suffered the death of a child(ren) and/or a near-death episode, conveying that information to relatives, friends, teacher, etc., will quickly enlighten them as to the seriousness of MCAD, as well as to the ramifications of not diligently treating your present MCAD child. Not having the individuals panic after hearing that information is a real balancing act, however.

One possible way of approaching this situation is by having a talk with the friend etc., explaining some of your concerns and fears of coping with an MCAD child(ren). After discussing how MCAD has affected your family (death or near-death of a child), shifting your discussion from death to focusing on your present child's MCAD concerns and needs, as well as on the positive prognosis for a healthy lifetime, may be a helpful way of keeping the friends etc., from panicking. They are aware of what can happen, but their immediate concern is to meet your child's needs while at school, daycare, etc.

In your discussion, you can explain a few guidelines that need to be followed (i.e. eat frequently, low fat meals and snacks, take carnitine at certain times). Your guidelines will vary depending on what situation your child is in (i.e. all-day daycare, school, short-term babysitter). Some parents find it helpful to have a complete schedule written out giving times for carnitine, meals, etc. This way the caregiver does not need to guess or make major decisions about when to do these things. They may feel more comfortable if everything is written down. Be sure to highlight the most important details, but try not to make it so detailed so as to overwhelm the individual. Also include situations where you NEED to be called (i.e. when the child vomits, hasn't eaten for hours etc) so the caregiver will KNOW when an illness protocol will need to go in effect.

In summary, stressing the seriousness of MCAD is important, but having the individual meet your child's needs is the immediate concern. This balancing act may become easier over time and as you feel more confident in discussing your child's well- and sick-routines.

**Question:** My 3-year-old daughter is a finicky eater. Knowing that hypoglycemia (low blood sugar) can cause a problem for an MCAD child, how can I get her to realize that she needs to eat?

**Answer:** One approach is to offer smaller and more frequent (2-3 hrs.) snacks and meals and not to rely on your 3 main meals for her caloric intake. If you spread out her food

over the course of the day, it may not be as stressful on you always trying to get her to eat so much at those meals.

Other suggestions include: offering high carbohydrate snacks (bagels, pretzels, fruit); keeping a snack container in your car for those times you're running late, keep offering new foods (even though you've tried everything!), and keeping high calorie drinks available especially when she is sick. Be very open with your daughter about why food is important for her body and that it is used to help it work. An age-appropriate analogy may be helpful. For example, explain that toys that use batteries only work well when the batteries are charged up. By playing with the toy a lot, energy is used up and the toy begins to slowdown and doesn't work as well. To keep the toy in working order, the batteries need to be kept fully charged ~ just as your body needs food to be fully charged in order to run its best.

**Question:** How can I explain MCAD and its treatment to friends, etc., in layperson's terms?

**Answer:** MCAD is an inherited metabolic disorder in which both parents carry the gene and EACH child has a 25% chance of inheriting it. Fatty acids cannot be broken down thoroughly enough to be used as energy so the body needs carbohydrates/sugar in order to function.

Carnitine helps to keep blood sugars steady (as does eating often) and is vital for breaking down some fatty acids in the cells, as well as for helping the body get rid of the toxins it produces in that process. If the child experiences fasting and/or an illness, a buildup of toxins can occur in the body, which could be fatal if not treated immediately with IV Glucose and sometimes IV carnitine. Long-term daily treatment usually includes avoidance of fasting, taking carnitine, and maintaining a relatively low fat/ high carbohydrate diet with snacks between meals. If there are other medical concerns due to previous severe episodes, then those medications/treatments would be included in the daily protocol (\*also see Dr Roe's article 'MCAD Information for Families' on our current website, [www.fodsupport.org](http://www.fodsupport.org) under Medical Information).

**Question:** Besides taking carnitine and maintaining a low fat diet, what other considerations should parents of MCAD children be aware of?

**Answer:** As previously mentioned, informing others of the seriousness of not diligently meeting your child's needs is imperative. Some people may think that a child skipping a meal is no big deal, but for an MCAD child, it can possibly cause some problems. On the outside, MCAD children look so normal that others may forget that on the inside their bodies are different. Wearing a Medic Alert tag (1-800-ID-ALERT) may help others remember that certain precautions need to be taken with this child.

Illness can be a stressful time for MCAD families. Possibly preventing some illness may take some pressure off. Having yearly family flu shots (discuss this first with your

physician, for children over 1) cannot guarantee 100% protection from the flu, but if the child does get sick the symptoms may not be as serious.

Many times, parents cannot control avoiding other children with illness (daycare, school), but having your neighbors and friends tell you when their children are sick can help you somewhat monitor after-school playing. Another concern to be aware of is to let your child's teacher, babysitter, etc., know when your child has been on double doses of carnitine. Diarrhea is sometimes a side effect and for some children it can be unsettling or embarrassing for them if an accident occurs. Thinking ahead and preparation can decrease some of your stress as well as your child's.

Another area to be aware of is your child's speech and motor development. Twelve percent of MCAD children have had some speech and motor delays, but have later 'outgrown' them or worked through them with speech or occupational/physical therapy. Don't hesitate to discuss your possible concerns with your physician. He or she may recommend testing.

## **Pharmaceutical News**

Ken Mehrling of Sigma-Tau Pharmaceuticals, Inc., has been a tremendous informational source. He furnished us with the helpful handout that explains the role of carnitine in our metabolism by comparing it to how a furnace works. The scientific pathways are also shown. For those of us who don't remember or never had high school biochemistry, the furnace analogy is easier to understand!

Dan and I would like to express our gratitude to Ken Mehrling and Sigma-Tau for offering to cover all the expenses for this newsletter. Their financial support, as well as educational and moral support is greatly appreciated.

## **Medical Update**

Dr. Roe and his colleagues, Drs. Kahler and Iafolla (\*in 2000, all are no longer at Duke) are extremely busy working on several research studies. All of the studies are funded by state and/or federal agencies.

So far, researchers have identified several mutations in MCAD children and their families. In one study, the Duke medical team will be studying the DNA of MCAD families to see which mutation is present. It is important to be able to prove that the mutation is causing the disorder. From this information, treatment strategies can be formulated.

Our son, Kevin, who has one of the mutations, recently spent 3 days at Duke undergoing tests to determine if taking the vitamin, riboflavin, would convert him to a carrier state. The test results were intriguing, but further investigation is warranted before any conclusions can be drawn.

In another study, in conjunction with the North Carolina Medical Examiner's office, all North Carolina deaths under the age of 2 will be screened for biochemical markers for several biochemical disorders, including MCAD. Seventeen diseases can now be screened for by using post-mortem blood and fresh liver tissue samples. From the 249 (1989) cases already screened, 1 was determined to be MCAD and 4 were carriers. It was important that the families be notified and made aware of the findings and their ramifications. Other metabolic causes were discovered in a few cases too.

On a related issue, during the summer, Dr. Roe met with Tom Moran, the Director of the SIDS Alliance, in Washington, D.C. Mr. Moran was very interested in what Dr. Roe was saying ~ that 1-3% of the SIDS cases may be MCAD. Dr. Roe discussed his ideas with him about having a national SIDS research center and about much of the research going on in North Carolina. Mr. Moran, according to Dr. Roe, was intrigued by some of the ideas. Each was encouraged that the lines of communication were opened between the SIDS group and researchers interested in finding the causes of SIDS.

Also in NC, researchers will be screening for 17 disorders in every child born in the state (@100,000 live births per year). If this study goes well, national implementation of newborn screening for inborn errors of metabolism is a very strong future possibility.

In another study, 6000 neonates will be screened over the course of 6 months. The purpose of this study is to determine the frequency of the MCAD gene in the population, as well as looking for the biochemical markers for inborn errors and the mutations that go along with MCAD. MCAD is estimated to occur in 1 in 10,000 live births and 1 in 50 live births as a carrier state. If an MCAD child or carrier is determined, families will be notified and further testing will be implemented.

Dr. Kim Iafolla, a Neonatologist/Geneticist, has a special interest in researching premature apnea (stopped breathing). In a preliminary study of 20 premature babies, 7 of the babies had apnea related to an obstruction and 13 exhibited central apnea of prematurity. All the babies with central apnea showed metabolic abnormalities as seen in MCAD with the level of octanoylcarnitine decreasing as the baby got older. In the future, Dr. Iafolla plans to look at administering supplemental carnitine to premature babies to determine if it prevents the premature apnea from occurring, or if the biochemical abnormality she found is reversible with carnitine.

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**[Please Note: Our Group began in 1991 as the MCAD Family Support Group ~ in 1996 we expanded to include all of the Fatty Oxidation Disorders (FODs). Please be sure to read the most current newsletters to get the most updated information on FOD diagnosis, treatment recommendations, research, and names of FOD researchers/Labs.]**