

MCAD Communication Network
'All In This Together'

January 1993

Volume 3 Issue 1

From The Editor

Happy Holidays & New Year! We hope that all of you had a safe and healthy holiday season. Even though 1992 flew by, it seems like ages ago since our last newsletter in June.

In this issue, two more MCAD families share their experiences and feelings about how MCAD has touched their lives ~ Many thanks to Cheryl and Brian Rosenberger (expecting twins in March!) and Andrea and Phillip Franklin. These stories really make our newsletter personalized, so please seriously think about sending your own Family Story.

Also in this issue questions are answered (please send those in too ~ we only have 1 for this issue), pharmaceutical and medical updates are reported, Love Messages are remembered, and a revised Family List is included. Additionally, we have included an article about MCAD for parents and families written in understandable language, as well as a drug assistance program pamphlet for families needing help acquiring their child's medication.

In our June issue, we will tell you about a special group of Virginia middle school children whom have 'adopted' MCAD as their yearlong project. They are very motivated to get the word out about MCAD. Their teacher, Carolyn Stamm, is the force behind their excitement. We'll fill in the details in June.

Again, we hope that you find this issue informative and helpful in networking with other families. If you would like to contribute to our June 1993 issue, we would greatly appreciate your suggestions, questions and/or articles.

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Kylie, Our First Born

It was a sunny day in June when I came home to tell my husband our news ~ I was expecting our first child! Being the one that had always wanted the children, I was excited!! Not being around children much in his life, Brian was not so sure.

As time wore on, Brian got more curious and excited. Then the day came, it was February 7, 1990. After an average labor the Dr. announced that it was a girl and we had

decided that her name would be Kylie Ann. Brian was elated. The tears came quickly and in less than 30 seconds, Kylie had wrapped her daddy around her little finger.

We went home on Friday and the weekend was busy. Come Sunday night we were finally settling down, just the three of us. The phone rang and I was talking to my boss. Just as our conversation had ended I looked at Kylie in her cradle and she was turning blue. After what seemed like hours, though it was minutes, we were at the hospital and the Doctor came in to tell us that they were unable to revive her. We had lost our firstborn child.

The next nights were extremely long and Brian now realized that children were to be a part of his life and we had lost our baby girl. After much testing, four months later our medical examiner called on a Friday to tell us that if they could find nothing by Monday they would just call the cause of death a "liver disorder" and close the case. That Monday morning Dr. Cohl called to tell us there might be some answers. Over the weekend he had been reading an article written in a magazine by Dr. Roe at Duke University (*in 2000, now at Baylor in Dallas) and what he was reading about was a genetic disorder called MCAD. He asked permission to send the necessary samples to this Dr. Roe for testing. We approved, got the needed information and it was only days later that we got the call that our daughter had died of MCAD.

The relief was that we knew what the cause was. The worry was that we knew nothing of MCAD. We have since learned much, as have most of you.

By the grace of God, we have since had another daughter, Kaleigh Rae, born June 8, 1991 and she is not a carrier. We have recently found out that we are expecting twins in March. Brian and I are extremely excited at the thought of having more babies, but we are also apprehensive because of the worry that comes with this until the day we get the news, MCAD or not. No matter what the news, we know that it rests in God's hands and that He will take care of us and our new baby. Thanks to all the research by Dr. Roe and others, we will make it.

Cherryl Rosenberger
Cedar Springs, MI

Brandi Lei

Our story begins on December 2, 1986, when my husband and I were very gifted with a beautiful and very healthy daughter, our firstborn, named Brandi Lei Franklin. She was a very smart child and very loveable, too. She was never sick a day of her life. She was so healthy.

One night, when my husband was working third shift, he said goodbye not knowing it would be the last time he would see our little girl alive. She had been running a little bit of a temperature and I called the Doctor. We were told she was probably cutting a tooth,

since she was thirteen months old. So I gave her some Tempra and put her to bed with me that night.

Well, on in the night, she got worse and I called for an ambulance to come to the house. She had stopped breathing and I was trying to give her CPR. My husband was on his way home, but when he and the rescue team got there, she was gone. It was so sudden. It still hurts so badly. The worst memory I have was before she stopped breathing, she looked at me and her eyes went back into her head. I just knew she hadn't died from SIDS.

Her father and I are both carriers and since then we have had one son. He is a carrier also. He's 2-years-old and doing fine. We are very lucky to have known about what our daughter died from so that our son could be tested and to know that he will be fine. He is our only living child and he is quite a miracle. He didn't take her place of course, no one could ever, but he is filling that void we have had in our hearts for so long. He was born July 10, 1990. Thank you and God bless all of you.

Andrea and Phillip Franklin
Hendersonville, NC

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 1993, 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: We are having trouble getting insurance reimbursement for my child's carnitine. What can we do about this?

Answer: Ken Mehring, Director of Marketing and Sales at Sigma-Tau Pharmaceuticals (*in 2000, Ken is a VP at Sigma-Tau), the makers of Carnitor®, suggests that you make sure that your physician specifically prescribes Carnitor® instead of just carnitine. Carnitine can be sold over-the-counter in health food stores, but it is **NOT** the same as the drug Carnitor®, which is an FDA-approved drug. The health food supplements are not covered by insurance and it is **NOT** what Drs. recommend (*see article on the differences between Carnitor® and carnitine on our current website). You may also want to have your Dr. write a letter to your insurance company explaining those differences and that it is an important medication for the treatment of your child. One additional suggestion might be to contact Sigma-Tau and ask about their Drug Assistance Program.

Pharmaceutical Update

Since Kendall-McGaw Pharmaceutical recently announced they would no longer produce their VitaCarn product, Sigma-Tau Pharmaceuticals, Inc., has now become the only company producing a levocarnitine drug. Their product is CARNITOR®. As mentioned in the Q & A section, having your physician specifically order Carnitor® (not just carnitine) may help you avoid problems with insurance reimbursement. If your pharmacy is having difficulty obtaining tablets and/or liquid, contact Sigma-Tau at 1-800-447-0169 and Ken Mehrling's staff will be glad to help you.

Ken also informed me that the Carnitor® intravenous solution is expected to be FDA-approved sometime this month. It will no longer be an experimental drug. Presently, if an IV is needed in an emergency situation, your physician can call Sigma-Tau directly to have it sent immediately. We'll keep you posted as to whether that procedure changes once the solution is FDA-approved (i.e., your pharmacy may be able to have it on hand). Also enclosed in this issue, is a pamphlet about a Carnitor® Drug Assistance Program sponsored by Sigma-Tau and administered by the National Organization of Rare Disorders. This program assists eligible financially needy patients to obtain the drug Carnitor®. If you would like more information or an application call NORD at 1-800-999-NORD.

Medical Update

This issue's Update contains several excerpts from Dr. Roe's revised chapter in **Metabolic Basis of Inherited Diseases**. The revision, however, will not be in print until possibly 1994.

NOTE: If any of you are involved with other MCAD researchers, please ask them if they would like to contribute an update on their research efforts and findings. We would like to know what's going on with MCAD across the country.

Since 1982-1983, approximately 200 patients have been identified with MCAD; some after the death of a sibling. Many of those deaths were initially diagnosed as SIDS or Reye's Syndrome and later determined to be MCAD by testing post-mortem blood or tissue.

By researching many of these children/families, it has been found that there are varying degrees of MCAD presentation even within the same family. In 1 family with 4 children, 2 of the brothers' deaths were attributed to SIDS and Reye's. After the 2nd child's death, several different tests confirmed MCAD in all 4 children. The oldest brother (now 10) and youngest sister (now 7) have had different experiences. Both eat frequently, avoid fasting, and are treated with oral carnitine.

The brother has been without symptoms and has never had an illness episode related to his deficiency. From birth to age 2, the sister has had several illnesses (viral infections, vomiting, lethargy, decreased oral intake) requiring hospitalization and treatment with IV

glucose and carnitine. For the last 4 years, she has been, episode-free, even with the chicken pox.

The variability of symptoms within this family emphasizes the need to evaluate siblings of previously identified patients. It is also suggested that parents be tested ~ it's possible that an apparently unaffected parent is MCAD deficient.

A clinical review of 94 MCAD families (104 affected children, 25 deaths) revealed the following: most patients presented symptoms between 3 and 15 months of age; few reports of 1st symptoms after age 4 and less recurrent illness after age 4; symptoms requiring repeated hospitalizations after age 20 are rare; 1st episode death occurred for 12 of 63 children in 1st year of life, 9 of 41 children after 12 months of age, and 4 died with a later episode; and that MCAD deficient children are at serious risk for death with an initial or later illness. An MCAD diagnosis was not established at the time of death for the 25 children. It was made post-mortem. In contrast, after the deaths an MCAD diagnosis was established for the 79 survivors. **Following diagnosis, there were no metabolic deaths, suggesting that early diagnosis (i.e., neonatal screening) before onset of symptoms may be associated with the decrease in mortality.**

NOTE: North Carolina is in the midst of a newborn screening pilot study that could possibly result in the screening going national. Blood spotted on the newborn screening card is sent for analysis by tandem mass spectrometry. (*in 2000, NC is the only state that mandates testing of newborns for ALL 30+ disorders that can be detected through tandem mass spectrometry ~ several other states may test for MCAD, but most of the other disorders are on a supplemental or pilot study basis at this time ~ see our current website for more NBS information).

Additionally, early diagnosis and initiation of treatment for this disease may also help lower the risk of long-term disability in survivors. Some of the possible dangers associated with the delay in early diagnosis found in a survey of 78 MCAD deficient survivors (all older than 2 years) revealed several unexpected problems with 29 patients. These included: global developmental disability, speech and language delays, behavior problems, attention deficit disorders (8 of 9 children were females), chronic muscle weakness, seizure disorder, cerebral palsy, and failure to thrive. The development of muscle weakness was strongly correlated with the length of time between symptom presentation and the initiation of measures to prevent further illness episodes. These minor delays seem to place these patients at a higher risk of long-term disability

Another interesting finding of 172 MCAD deficient patients is that MCAD is almost exclusively found among Caucasians of Northwestern European origin. There is, however, 1 Pakistan and 1 American Black and some patients of Southern European and North African origin. There have been none identified in the Far East, nor any carriers found by newborn screening in Japan. It has been suggested that 1 of the several MCAD mutations may have arisen in the UK and Germany and then spread to nearby countries.

Along with specific dietary changes, supplementation of L- Carnitine is advocated for

MCAD patients. From a research study of an asymptomatic MCAD deficient patient, it was shown that L-Carnitine IV therapy during an illness assisted in the removal of potential toxins that might accumulate under fasting stress and infection conditions. It was suggested that carnitine supplementation was useful during illness, but may not be necessary during healthy times. For the MCAD patient, however, the dilemma is not knowing when illness will occur. Apparently, there are no toxic effects from supplementation, except for occasional loose stools and a fish-like body odor when receiving high doses.

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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.]