

FOD Communication Network
(Fatty Oxidation Disorders)

'All In This Together'

June 1995

Volume 5 Issue 2

The **FOD COMMUNICATION NETWORK Newsletter** was created and is currently edited by Deb and Dan Gould ~ 805 Montrose Drive, Greensboro, NC 27410 (336) 547-8682
Any questions or comments should be directed to them.

From The Editor

Yes! **We've made a CHANGE!** And believe it or not, as far as change goes this was EASY. Wouldn't it be nice if all change was like that? I guess it can't hurt to hope! Anyway in order to **provide needed support to more families** dealing with genetic metabolic disorders we have broadened our scope. Lately I have been receiving calls from some LCHAD families seeking more research information, as well as the names of others families they could connect with. Along with families, professionals have also suggested this change. So as of this issue, **the FOD (Fatty Oxidation Disorders) Family Support Group will be focusing on ALL of the fatty oxidation disorders (FODs).**

If you are working with a family with any of the following disorders please have them write us so we can send them our newsletters. **Our Support Group** will now include the following disorders: **MCAD, SCAD, LCAD, LCHAD, CPT I & II, Carnitine Acylcarnitine Translocase, VLCAD, Trifunctional Protein Deficiency, ETF Dehydrogenase Deficiency (GA II or MADD), Carnitine Transport Defect, and HMG.**

Since our last issue, we have received a few more family and personal experience stories, as well as questions to be answered. As always, these are greatly appreciated. However we always invite more of you to contribute your own ideas and stories.

I NEED YOUR HELP!! If we are going to make this OUR newsletter, then we need to work together. PLEASE...

So if you'd like to contribute anything to our newsletter, Families AND Professionals, please send things by December 1, 1995 so they can be included in our Jan '96 issue. Tell us how you are coping as a family, about your hospital experiences, what some of the difficult issues are for you, or what has been the greatest gift your child has brought to your lives. Professionals ~ it would be great if you could tell us about your current research and how the findings might benefit our Families. All letters/articles will be appreciated!

In this issue, Dawn Greene shares with us her opinion about the benefits of this newsletter. Alex Holloway, from England, shares her story about her son, Daniel, and how they have dealt with MCAD since 1991. Thank you also to Randall Burns for

sharing their hospital experiences and for offering some suggestions to make that experience less stressful.

As in other issues, questions are answered, nutrition information is offered, and Medical and Pharmaceutical Updates are discussed. **NEW Families:** Please remember to **send in the Family Questionnaire** (included in the Family Packet I sent you) so you can be included on the networking Family List, as well as continue to receive the newsletter.

Again, we hope that you find this issue informative and helpful in networking with other families. If you would like to contribute to our Jan 1996 issue, we would greatly appreciate your suggestions, questions and/or articles.

Deb and Dan Gould, Co-Editors
fodgroup@aol.com
336-547-8682

Letters to the Editor

Dear Deb: I wanted to write you a short note to say Thank You for all the time and hard work you put into the newsletter. Every time I look at my last newsletter, I get upset that any health care professional would discourage a family with an MCAD child from getting your newsletter. Every time I read about a child that has died from MCAD, I do cry, but it also makes me realize how lucky we are that Austin is alive. If it weren't for you and this newsletter, I would still be depressed and upset not knowing anything about MCAD. Austin turned 3 on Jan 1 and is doing great ~ only 2 hospital stays! Thanks again!

Ron, Dawn, Amber, and Austin Greene
South Dakota

Dear Deb: I received your address from NORD. Our daughter, Cari, has MCAD. She is 14-yrs-old and is seeking a pen pal with another 14-yr-old with MCAD. Cari is an A, B, & C student. She "shot puts," plays basketball, and cheerleads. She plays the trombone in the school band. She loves animals, bike riding, and walking. Can you help us find her one? Thank you.

Al, Sherry, and Cari Thorn
Monroe, MI

Dear Deb: We have an 18-month-old with LCHAD and we lost our first child, Sarah. To those with the same disease, Jane has been doing very well with her MCT oil diet, three-hour eating schedule (she has a gastrostomy tube), and evening drip schedule. I am interested in compiling more information on diet, medication, feeding schedules, health complications, and types of lab tests done. Please contact me if you would like to network and exchange ideas and information.

Jenny Carroll
Prairie Du Sac, WI

From England With Love

Thank you so much for all the information that we received this morning. It was very helpful as there is not a support group specifically for MCAD families in Britain. There is, however, The Research Trust for Metabolic Diseases in Children, which is where we got your address.

From reading all of the touching stories in your newsletters, we have realized how lucky we are not to have lost Daniel. Daniel was born April 24, 1991 and we were very happy as he seemed normal and healthy. I had difficulty trying to breastfeed though, as he found it hard to latch on. I was told that he was getting enough, as babies didn't need much for the first couple of days. He cried a lot the first night, but was much quieter the next day. I was looking forward to taking him home where I thought I would be able to relax more and feed him more easily.

However, when the midwives came to show me how to bathe him, they started looking worried. He seemed to be breathing strangely and was floppy. A pediatrician looked him over and tested his blood sugar. It was so low they rushed him down to special care and put him on a glucose drip. This made it difficult to breastfeed, so I switched to the bottle.

He came out of the hospital a few days later. We just assumed he got sick from not getting enough milk from me. So we put the experience behind us. No other explanation was given for his low blood sugar.

Everything went okay until he was between 15-20-months-old. Then Daniel's behavior changed. He no longer spoke the 30 or so words he had learned and he became withdrawn, hyperactive, and very difficult to manage. I frequently spoke with my health visitor and doctor about my worries, but no one could give me an explanation.

On April 15, 1993, Matthew was born. He fed well from birth and was a very happy baby. Then when Daniel started playgroup, they decided he needed a one-on-one helper to be able to cope with him.

In September 1994, Daniel ate his breakfast and then was violently sick. I called the doctor and was told it was a tummy bug and that I should restrict his eating, but make sure he had plenty to drink.

The next day Daniel didn't get up, so I made him up a low sugar black currant drink. He still hadn't gotten up by 11:00 so I brought him downstairs and laid him on the sofa. He was very sleepy. I got worried and called the doctor again. He didn't know what was wrong and said he would come back later to check on him.

We woke him up to give him drinks, but he kept going back to sleep. Then he started moaning in a strange way. When he opened his eyes he couldn't seem to see us or respond to us so we called the doctor and he told us to go to the hospital.

When we got there, they discovered his blood sugar was dangerously low. It was very scary because all his veins had shut down and they couldn't get the glucose drip in. Eventually they got it in place and Daniel came to.

The doctors tried to talk to him and we explained that he only used a few words. They seemed to think there was a link between this and his illness. They gave him a brain scan and some steroids as they suspected he had either a hormonal or metabolic condition. He stayed in hospital for a week. Then in October he went in to be fasted so that they could take blood, urine, and a skin biopsy in a fasted state. We finally got the results ~ MCAD.

Our son, Matthew, was also tested and he is a carrier. Daniel's behavior is improving. He is not so withdrawn and has even said a few new words. He enjoys playgroup now and is getting easier to manage. Being in hospital set him back a bit, but with lots of patience he has come through it well. Slowly we feel we are turning the corner.

We are still coming to terms with MCAD and at the moment it's hard to think about anything else. We are hoping that this is the answer to the learning and behavior problems as we have been blaming ourselves for not being good enough parents and that's even harder to bear.

Our prayers and good wishes go out to all the families that have lost a child to MCAD. **Let's hope in the future all babies can be tested for it so these tragic deaths can be prevented.**

We would love to carry on receiving the newsletter. Thanks for all your help.

Alex and Mark Holloway
Oxford, England

Be Prepared!

I'm not one to write letters or articles, but what has prompted me to this time was your piece Deb, 'Will Somebody Listen To Me!' It was a very good piece and I'm sure it will benefit all who read it. It very well could have been me who wrote it, and maybe I should have some time ago.

Twice we were faced with the same dilemma. We knew the warning signs. When Kathryn's fever increased and her blood glucose level took a dive, we immediately went to the Children's Hospital emergency room with protocol in hand. As usual, we were met by that brick wall ~ waiting in line, then finally pre-interview.

The receptionist, intake nurse, and the physician on duty would all be given a copy of the protocol. They would glance over it, we would stress the immediacy, and we would be told "We need to take a few samples for tests."

After the wait, usually 3-5 hours, the physician would finally follow the protocol's recommendation of IV glucose and L-Carnitine. Kathryn would be admitted for a 4- and a 5-day stay in the hospital. We were left with the feeling of helplessness as though we did not know what we were talking about, as though no one was listening. A nurse once even politely asked us to not interfere while the physician was conducting his exam.

At that time we were still trying to locate a pediatrician we felt comfortable with, one who we felt took an honest interest in Kathryn's condition. After an exhaustive 3-year search, we did locate one. She studied the literature we had given her, established a contact with Drs. Iafolla and Roe, and even requested that we supply her a copy of this newsletter.

We expressed to her our **concern over the length of time it takes for the ER personnel to accept and follow the protocol**. We were relieved to learn that all it would take was for her to **place us on an "Emergency List"**. Upon our arrival we are led directly to a room where Kathryn is prepared for the IV glucose and L-Carnitine and our pediatrician is called. **You could say that we are pre-registered for an emergency visit**. No hassle, no worry.

We consider ourselves very lucky indeed. I would strongly suggest that all MCAD parents check with their physicians to see if their hospitals offer a similar process. It never hurts to ask and the less you have to worry about the better.

Again we look forward to and appreciate the time you put into this wonderful newsletter. We hope to visit Duke again soon. It would be great to see you and the boys again as well. We wish you and yours the best and as always each of you remain in our prayers.

Randall, Dianna, Crystal and Kathryn Burns
Eules, TX

Adam's Story, 4½ years old ~ LCHAD

After a stressful pregnancy Adam was delivered by emergency C-Section at 35 weeks weighing 4 lbs 5 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 admission to the hospital, 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 that 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 metabolism of fatty acids.

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 of serum guanidine organic acid levels.

During this hospital stay Adam was irritable and 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 Elaina Jurecki. Also L-carnitine was added to his formula. Carnitine is a protein present in everyone's body that 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 that has only been diagnosed since 1987 with only a few cases reported worldwide. 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 eyedropper that 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 bad 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 for 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 that 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 seemed to be less affected than most of the other 10 or 12 cases of this disease so far documented worldwide.

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) a formula that 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 again visited the hospital for an overnight fasting test. He went 12 hours without eating, and 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 presence of 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 every 6 weeks}. At 14 months he had a severe case of Chicken Pox that 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 that supplies essential fats.

Adam will be 3 on December 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 due to swallowing pennies that lodged in his throat. He has grown rapidly and is approximately 90th percentile 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.

***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.

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

Val Fulton
San Jose, CA

Joseph Marsella Update, 2 years old ~ VLCAD

Joseph is our son with Very Long-Chain Acyl Coenzyme A Dehydrogenase Deficiency (VLCAD). I am so thankful for having him in my life, although he is definitely in his Terrible Twos! I have to say that the last two years were hard, but it has gotten better. As I write this letter, I have to think back on our daughter, Toni Marie. Her life was so short! She would have had a 4th birthday today, October 8th. Fighting each day to keep Joseph healthy is hard, but a lot easier than to bury a baby.

Joseph is such a happy child and with each smile and laugh, gives me strength to keep going. As the children grow, you realize that it was worth it because life is so very precious!

When Joseph became ill last May and was hospitalized in our small facility with doctors that are unfamiliar to Joseph's disorder, the proxy letter from our specialist was very helpful. Proxy letters should be written by your doctor. It describes your child's metabolic disorder and the appropriate treatment measures that need to take place during illness. It should also provide your doctor's telephone number or beeper number so that they can be consulted during these episodes (and any other health care provider familiar with your child's condition).

Under stress you can forget something of importance, or become so upset that you can't speak at all. With a proxy letter, it is easy to hand it to someone who is there to help your child. It also makes it easier for you as a parent to remain calm in the situation. I think that everyone should obtain a proxy letter from their doctors even if it is just for a back-up safeguard!

After Joseph's illness, being his first episode, you do realize what is missing from your letter and what you need to say, point blank! I would also suggest getting your proxy letters updated yearly, and make copies to keep in your wallet, diaper bag, glove compartment, doctor's office, and your local ambulance service. This would help to familiarize people with your NAME, DISORDER, and what they have to do, without wasting time!

Heather Marsella
Port St. Lucie, FL

Questions and Answers

[Please Note: This question and answer column is designed to answer questions, both medical and practical, on FODs and their treatment. Answers to questions are solicited from those who have had firsthand experience dealing with FODs. These include physicians, parents of FOD children and children/adult FODers 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 1995, 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: Now that my 9-year-old MCAD son is getting older, what is the possibility of either decreasing his Carnitor® dosage or withdrawing the medication completely? Do you have any current research on children that have stopped the medication?

Answer: Dr. Roe stated that he and another researcher are exploring the possibility of a research project involving MCAD children on Carnitor®. If the project becomes feasible, it may take form sometime in early 1996. The researchers would like to determine if there is an optimal carnitine dosage for MCAD children. They are also

interested in looking into what conditions and at what age children might be able to cease taking the medication. We will keep you posted if and when this study is implemented. It is also important to note that there are some researchers/physicians that do not suggest or prescribe carnitine for their patients for various reasons. If any of you are in that position, we would greatly appreciate it if you could tell us how things are going for your child. For example, have you had any difficulty with 'episodes' and was any form of carnitine used in an emergency situation? Let us know.

Question: As an MCAD parent, I have an interest in how you determine who is a carrier. Many of my siblings are now having their own children and are interested in getting tested for being a carrier. Can you explain what they need to know and where they can call to have the necessary diagnostic procedures done?

Answer: At our new Lab at Baylor, we will be able to test for MCAD and for MCAD carrier state (only if homozygous mutation), as well as selected mutations of LCHAD. A whole blood spot is needed for the testing. DNA testing for carrier state has not been as successful with some of the other fatty oxidation disorders, however. Sometimes it may be necessary to send samples to an overseas Lab that is equipped for that type of testing. Of course, if you have any questions, you can call Dr. Roe's Lab at 214-820-4533. There are also other labs throughout the U.S. that do similar testing. I will try to get a listing of those labs for our next newsletter (*see our current website under Medical Information/Diagnostic Labs).

Question: Why is carnitine suggested for some of the fatty oxidation disorders and what is its purpose?

Answer: The production of energy takes place in the mitochondria, the 'power plants' of the cell. Fatty acids are the preferred energy source for the mitochondria, especially for muscle and heart cells. Entrance of fats into the mitochondria requires a carrier molecule called carnitine. Once inside the mitochondria, the fats are metabolized in a process called fatty acid oxidation to produce ATP (a chemical form of energy).

Carnitine also serves a second important role in the mitochondria where it functions to remove toxic organic acid (OA) compounds. This scavenging function is important since high concentrations of these OA compounds impair the mitochondria's ability to produce ATP. After removing these compounds from the mitochondria, the carnitine-organic acid compound is eliminated in the urine. This loss frequently leads to carnitine deficiency if supplemental carnitine is not provided.

If carnitine concentrations are inadequate, entry of fatty acids into the mitochondria are blocked. In addition, scavenging of the toxic OA compounds is impaired leading to a buildup inside the mitochondria. This results in a depressed production of ATP and causes the person to exhibit signs and symptoms of low energy production (i.e. floppy appearance, poor growth, weakness, lethargy) and dysfunction of multiple organs including heart, muscle, liver, and brain.

We receive a significant amount of carnitine from foods we eat. The average diet contains approximately 100-300mg of carnitine derived mostly from red meats and dairy products; strict vegetarians have low carnitine intakes. Our bodies can also synthesize carnitine from proteins we eat. Newborns have less synthetic capacity to make carnitine than children and adults. Carnitine has been added to most infant formulas and is also found in mothers' breast milk. Carnitine is supplemented in many of the special formulas designed to treat organic acidemias, but usually additional carnitine administered separately is recommended in the treatment of some of the fatty acid disorders.

Especially during times of illness and metabolic stress, the defective enzyme results in a buildup of the toxic OA compounds. These compounds bind to carnitine in the mitochondria and are then excreted in the urine. This excessive loss may lead to a carnitine deficiency if not adequately replaced. (*see our current website under Pharmaceuticals/Sigma-Tau for more info on Carnitor®)

(This information was taken from an article written by Elaina Jurecki, M.S., R.D. and John Baker, M.D. of the Regional Metabolic Center, Kaiser Permanente. Oakland, CA. It was printed in the Organic Acidemia Newsletter August 1994.)

Pharmaceutical Update

Some of you have requested information about Sigma-Tau Pharmaceuticals, Inc., the producer of Carnitor® and the financial backer of our newsletter. If you would like to correspond with them, call 1-800-44 7-0169.

I would like to especially thank Ken Mehrling and Sigma-Tau for all of their support over the last several years. It is through their generosity that we are able to provide the current newsletter, as well as Family Packets of past newsletters for our new families. **Thank you from all of us! We are truly 'all in this together!'**

Medical Update

We aren't the only ones making changes around here. Dr. Charles Roe, our Group's Medical Advisor, has recently made a big move to Texas. His new address is:

Dr. Charles Roe
Baylor University Medical Center
Institute of Metabolic Disease
3812 Elm Street
Dallas, TX 75246
214-820-4533
214-820-4853 (fax)

Now that we are including several other disorders in our newsletter, we thought it might be helpful if we learned about some of them and how they might be similar to or different from MCAD. Rachel Slaugh, a genetic counselor from Washington University School of Medicine in St. Louis, Missouri, has provided us with some information about LCHAD. She also tells us about Dr. Strauss (*in 2000, now at Vanderbilt) and his work in that area. Thank you, Rachel, for taking the time to write and send in your contribution!

Note: If any of you, parents or professionals, have information about the other disorders, please feel free to write and let us all know what you know! We're always interested to know what other researchers are working on, as well as how you are coping as families with the different treatment protocols.

LCHAD Deficiency: A Genetic Syndrome with Similarities to MCAD Deficiency

LCHAD (Long-chain 3-hydroxyacyl-CoA Dehydrogenase) deficiency is a genetic syndrome with many similarities to MCAD deficiency. Like MCAD, it is caused by an enzyme defect in the beta-oxidation cycle. This results in an inability of the body to break down fatty acids into a usable energy source.

LCHAD deficiency can present in many ways, such as hypoglycemia, lethargy, SIDS or Reye-like syndrome. LCHAD deficiency can also cause a child to have poor muscle tone (hypotonia) and problems with the functioning of the heart (cardiomyopathy). As with any inborn errors of metabolism, LCHAD deficiency is inherited as a recessive genetic condition, so there is a 25% chance of recurrence with each pregnancy.

Unlike MCAD deficiency, LCHAD deficiency can cause medical complications during pregnancy for the mother of a child who will have LCHAD deficiency. People who are carriers for MCAD deficiency have no medical complications from being a carrier (*although in 2000, some MCAD carriers have reported such symptoms as hypoglycemia) since they have one copy of the gene that produces an enzyme that works correctly. Usually a carrier of LCHAD deficiency will not have medical problems either. However, women who are carriers of LCHAD deficiency may have problems during a pregnancy if the unborn child has LCHAD. These problems may include: anorexia, vomiting, abdominal pain, and jaundice during the third trimester of pregnancy. If untreated, it can cause liver failure in the mother. This may result in the need for liver transplant or death. These complications are called maternal acute fatty liver of pregnancy or AFLP syndrome (*see our current website under Medical Information). It is not known why women have these complications during pregnancy or why the problems occur only when the fetus has LCHAD deficiency.

The treatment for LCHAD deficiency is similar to the treatment for MCAD. Like MCAD, an infant or child should avoid fasting. Illness in the child, such as the flu, may necessitate medical treatment or hospitalization for the child. Carnitine, often prescribed for MCAD, is not always given to a child with LCHAD deficiency (*although many of

our LCHAD Families today use carnitine). LCHAD children are usually given MCT Oil (medium chain triglycerides) prescribed as a supplement in the diet.

LCHAD deficiency was discovered in 1989. The gene for LCHAD has recently been found and the different alterations (mutations) in the gene are being characterized. One laboratory that is looking for mutations in the gene is Dr. Arnold Strauss' lab at Washington University School of Medicine in St. Louis. Investigators in the lab have also found mutations in the gene that causes MCAD deficiency. Recently, the genes of 3 children with LCHAD deficiency, all of whose mothers had AFLP, were analyzed. In the 6 copies of the gene (2 copies in each of the 3 children), 5 copies were found to have the same change (mutation) of the DNA. This is called the G1528C mutation; named for the location of the mutation. The last gene had a different mutation called C1132T. The G1528C has been found in other children with LCHAD, so it appears to be a common mutation. The researchers in the Strauss lab have also found other mutations. It is not yet known how many different mutations may occur in the gene or how common any of the already identified mutations will be.

Identification of the mutation allows for accurate diagnosis of the LCHAD deficiency, either as a newborn or prenatally. This may be important so the treatment of the infant can begin in the newborn period before the child has a potentially life-threatening episode. It may also help to recognize which pregnancies are at risk for maternal complications of AFLP. Early treatment during the pregnancy may reduce the severity of the maternal complications.

At this time, the work in Dr. Strauss' lab is being done at a research level. He hopes that in the near future, some of this testing will be available as a clinical test. Further studies will continue to identify mutations so that information will become available about the frequency of the different mutations. This will allow for increased accuracy in diagnostic and carrier testing. Dr. Strauss is committed to helping families where there is a question about the diagnosis of MCAD or LCHAD deficiency and would be glad to evaluate DNA samples if there is a reasonable suspicion of one of these conditions.

Summary: LCHAD deficiency is a newly identified genetic syndrome caused by a deficiency of an enzyme in the beta-oxidation cycle. Since both MCAD and LCHAD are caused by enzyme deficiencies in the same metabolic cycle, they have many of the same medical symptoms such as hypoglycemia, lethargy, and in severe cases, may first present as SIDS. Treatment for each is both similar and different. Fasting should be avoided and illnesses may precipitate complications. Individual treatment plans and Emergency Protocols should be coordinated by a physician with expertise in these disorders. The gene for LCHAD deficiency was recently identified and research is currently looking for mutations in the gene. This will allow for earlier diagnosis and therefore better and earlier treatment.

Rachel Slaugh, M.S.
Genetic Counselor
Division of Medical Genetics
Children's Hospital
One Children's Place
St. Louis, MO 63110
314-454-6031

Love Messages

(Please see our most current online issue)

'The soul would have no rainbow, had the eyes no tears.'

John Vance Cheney

Recipes

'Cooling Off the Low Fat Way'

Summer means being hot and getting cool may mean eating lots of ice cream. Perhaps you don't even need this excuse for indulging! But let's take a look at some tasty alternatives to ice cream that contain fewer calories and fat. The following listings are for a 3/4 cup or a 6 oz serving, not a jumbo or double scoop.

ICE CREAM: ingredients are sugar, cream, whole milk, milk fat, and flavoring. By law there must be at least 10% milk fat. A 3/4 Cup serving will have 220-440 calories with 11-28 grams of fat. Haagen Daz, Frusen Gladje, and Ben & Jerry's are the highest on the scale. A single serving provides 40% of the fat and 75% of the saturated fat a person should ingest daily.

ICE MILK: ingredients are sugar, milk, 3-7% milk fat, and flavoring. By law there must be at least 2% milk fat, but not as much fat as ice cream. A 3/4 Cup serving will have 145-190 calories and 3-7 grams of fat.

FROZEN YOGURT: ingredients are yogurt, sugar, gums (makes it creamy), flavoring. Yogurt uses cultured milk, which may help people with trouble digesting the lactose (milk sugar) in ice cream. A 3/4 Cup serving will have 190-330 calories which are low in fat. You really must read these labels to identify which are low in fat. TCBY's product has no milk fat while Tofutti's have 19 grams of fat.

DAIRY DESSERT: ingredients vary and you must read labels. This category means the item has varying amounts of fat and often nutrasweet is used.

SHERBET: ingredients are sugar, water, some milk and flavorings. One serving will yield about 200 calories and 3grams of fat.

SORBET: ingredients are sugar, fruit, water, and flavors. A serving is 170 calories with little or no fat.

NON-DAIRY means that no milk products are used. Often these are very high calorie items due to addition of soy or corn oil. There is no calcium so there is little nutritional advantage in these, unless you are lactose-intolerant.

These listings don't consider the wide variety of toppings so popular these days. Keep in mind that these toppings will add calories, fat and cholesterol depending on your choice. Fruit or sprinkles are your best choice.

You can make many cool creations at home with the blender or food processor using ice milk, low fat yogurt, fruit, and juices. Freeze your own popsicles or ice cubes using fruit juice. Children like to help make these and they will consume a better snack than sugar-laden pops or fat full ice cream. (*when your child is ill and not eating well, regular sugar popsicles will give him/her some calories and may avoid the low blood sugar that can trigger a metabolic crisis.)

So indulge in a frozen treat, but watch the serving size and the ingredients. Also, keep your refrigerator stocked with melon, berries, peaches, plums, and other fresh fruit. And head to the local pool for some exercise. Stay cool!

Peach Cream

1 cup frozen, canned or fresh peach slices (no juice added)
1/2 Cup vanilla yogurt
1/4 Cup milk
1/8 Tsp nutmeg or cinnamon (optional)

In a blender, combine all ingredients and blend till smooth. Makes two 4 oz servings.

Cow in the Apple Orchard

1 3/4 Cup of milk
1/3 Cup (one-half of 6 oz can) frozen apple juice concentrate (do not dilute)
1 Cup vanilla yogurt
1/4 tsp cinnamon

In a blender, combine milk and apple juice concentrate. Blend till smooth. Add yogurt and cinnamon. Makes five 6 oz servings.

Juice Float

Vanilla ice milk
Orange Juice
Lemon-lime carbonated beverage

Drop a scoop of ice milk into an 8 oz glass. Fill the glass 3/4 full with orange juice. Pour in enough carbonated drink to fill the glass. Give a stir with a spoon. Add a straw and enjoy.

Strawberries Through a Straw

3 cup frozen or fresh strawberries
1 1/2 Cup unsweetened pineapple juice
2 Tbs honey (optional)

In a blender, combine ingredients till smooth. Makes four 6 oz servings.

Fruit Pops

1 - 16 oz can pear or peach slices, drained or 2 cups sliced fresh fruit
1 - 8 oz carton vanilla yogurt
1/2 tsp lemon juice
3 Tbs sugar or honey

In a blender, combine all ingredients until smooth. Pour mix into 5 oz paper cups and cover each with foil. Insert a wooden stick in middle of foil and freeze till firm. Can also freeze in regular popsicle cups.

You can use a food processor or blender in these recipes. Experiment with other fruits and make your own creations. Yogurt was substituted for ice cream in the original recipes. You could use milk for an occasional change.

*'Have patience with everything unresolved in your heart
and try to love the questions themselves...
Don't search for the answers, which could not be
given to you now.
because you would not be able to live them.
And the point is to live everything.
Live the questions now.
Perhaps then, someday far in the future,
you will gradually, without even noticing it,
live your way into the answer.'*

*Rainer Maria Rilke
Letters to a Young Poet*

June 1995
Volume 5 Issue 2

[Please Note: Our Group began in 1991 as the MCAD Family Support Group ~ in 1995 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, Newborn screening, treatment recommendations, research, and names of FOD researchers/Labs.]