

FOD Communication Network
(Fatty Oxidation Disorders)

'All In This Together'

January 1996

Volume 6 Issue 1

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

Happy belated New Years! Dan and I hope that all of you had a safe, healthy, and peaceful holiday season. Holidays can be an extremely stressful time and we hope that you were able to get through them in your own way. We were especially thinking of all the families that experienced their first holiday without their child. Our thoughts and prayers were and are with you.

I apologize for being so late with this issue. My family experienced its own kind of holiday stress with me being out-of-town on a unique and prolonged 'experiential on-the-job training' exercise. It was definitely a different and a once in a lifetime experience ~ NEVER to be repeated! If you recall my editorial article from a past issue called 'Will somebody listen to me!' ~ well, my holiday experiences related very well to not being heard! To say the least, I learned a lot by what I had to go through, but at the same time, there were many not so positive experiences. In the near future I hope to voice my experiences verbally and in writing, but for now, I will not bore you! Let's get on with this issue.

Two FOD Families will share their touching Family Story with us and a family asks for your help in finding 'pen pals' for their FOD children. We will also discuss suggestions for making our newsletter better. So please don't hesitate to drop us a note or give us a call. **WE NEED YOUR HELP!** Ideas and/or articles would be **GREATLY APPRECIATED!** Our next issue will be around July 1996 so please send things by May 15. Additionally, if for some reason you do not wish to receive this newsletter, we would appreciate it if you could let us know so we could keep our postage costs in check.

As listed in our last issue, the **Communication Network will address** the following disorders: **MCAD, SCAD, VLCAD, LCHAD, CPT I and II (Carnitine Palmitoyl Transferase), Carnitine acylcarnitine translocase, Trifunctional Protein Deficiency, ETF Dehydrogenase Deficiency (GA2 or MADD), and HMG**

As in other Issues, questions are answered, nutrition information is offered, and Medical Updates are discussed. We have also included an updated Family and Professional List. Please send us corrections if needed. Also if you send questions or articles, it would be nice to see a picture of your child and/or family. It's always nice to connect faces with

names ~ it will make our newsletter more personal. If you send one, send one you don't mind not getting back!

NEW Families: Please remember to **send in the Family Questionnaire** 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 FOD Families. Please Send Ideas and/or Articles By May 15, 1996.

Deb and Dan Gould, Co-Editors
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336-547-8682

Letters to the Editor

Dear Deb and Dan: We received a letter from the office of our MCAD specialist at the University Hospital explaining the Family Support Group. We read the enclosed articles with quite a bit of amazement because many of the stories could duplicate our experiences almost word for word.

We have 9 children in our family. Our youngest two, both boys, have been diagnosed with MCAD. One time at the hospital the Dr. told us to be calm and enjoy our son, then he sent us home. We were back at the hospital 2 hours later with a son who was unconscious, having seizures, and as we later found out, a blood sugar level of 11! We are very grateful for a family Dr. who continued to try and find help even though there seemed to be none anywhere.

The MCAD specialist in Denver wanted to know how closely related my wife and I were, so with a lot of information from relatives, we found that we are 6th cousins. No problem the Drs. say.

The boys have stayed out of the hospital by using Carnitor® twice a day. We also notice a behavior pattern of crying and easily being upset with Jerry and anger and temper control with Ben. The Drs. encouraged us to have them on multiple vitamins high in B vitamins (3 times daily dose, no sugar, no food colors). This has a very definite effect: Jerry's much happier and Ben handles things in a calm manner rather than being angry.

[Note from Deb: Families, please be sure to discuss this type of suggested treatment with your Dr. or specialist if you are considering it, especially about the safety of tripling the daily dose of vitamins and whether a cause-effect pattern can be established. Also be aware that other factors may have contributed to the changes in behavior. Fluctuating low blood sugar episodes or ADD/ADHD are just 2 examples of concerns that can impact behavior, such as the anger/crying episodes. There are established and effective treatments available for each of those issues that don't involve high doses of vitamins.]

Yes, we would like to receive the newsletter and my sons also expressed a desire to find someone to be pen pals with. Ben is now 8 years old and Jerry is 10. Both are active in

outdoor activities and in Cub Scouting. Jerry is also the catcher on the local Little League team. Thanks for your willingness to take the time and commitment to help other people in the same situation.

Sincerely,
Thomas and Julie Grant
Centralia, WA

*** Thanks for your letter and input. I hope your sons are able to connect with some other MCAD children. Let us know if things work out. And with 9 children, try to get some rest! DLG

Dear Deb and Dan: Thanks for giving me the opportunity to help with the layout of the newsletter. I have some ideas that you may have already thought about but I thought I'd throw them out there for others to respond to also. Add pictures to articles or inquiries ~ black & white photos work best (*all of these early issues have been scanned, condensed and edited, so the pictures and graphics have been removed for the online archive.) Make a photo address book or a 'Meet the Families' issue to offer in the future ~ this could possibly be a fundraising idea. Have families send in their favorite low fat recipes and mail everyone a copy. Try to get more FOD children to 'voice' their concerns or questions. Have a 'Kid's Corner' as a special part of newsletter. Many of the children are old enough to interact with others in a similar situation.

Judy Farrell
Menasha, WI

***Thanks, Judy. Those are all great ideas. What do the rest of you think? Let us know and if you have your own suggestions pass them along to us. It would be great if everyone sent in a favorite 'FOD' recipe and we were able to compile a 'Metabolic Disorder Cookbook.' We could even have it published! Think about it and let us know what you think and/or go ahead and send me your recipes. DLG

Dear Deb and Dan: It might be helpful to get on the Internet in the future.

Julie Grant
Centralia, WA

***That's an idea we have been interested in for a while. Jeff and Lisa Schmidt of Ohio have been able to secure a Web page for us on the Internet. To access the FOD Web page type in: <http://www.cinternet.net/FOD> (*in 2000, please note that our website is now www.fodsupport.org)

Dear Deb: I had to write a personal note to you while returning my questionnaire. I'm sure editing and coordinating the newsletter is a time-consuming task, and I have to thank you for making the effort.

When I received the Packet, I sat down and read straight through each newsletter. I cried and cried as I read of other families' losses and I prayed a prayer of thanks for some unknown reason my child was spared the fate of so many of the children.

The newsletter helped answer in layperson's terms many of the questions that we had. I really appreciate you sending the past issues. It allowed us to see how far the research has come in a relatively short amount of time.

Our daughter was diagnosed after her 2nd episode which, thank God and the Drs., was less severe than her first. During her 1st episode she stopped breathing during a seizure and had to be airlifted to the Children's Hospital in Greenville, NC. The newsletter helped us to see that in spite of the scary events of the past, with proper care, the future can be bright and less frightening. I want you to know if there is anything I can do to help with the newsletter or with anything else to help spread the news about MCAD, please contact me.

I was also startled to read about the health professional's response that your newsletter was 'too depressing.' The reality of MCAD not being treated is depressing! The sadness of these stories serves to make those of us who have children who survived even more thankful.

I have 3 sisters. One of my sisters has a 20-month-old son and is trying to have another child. She had not planned to be tested due to personal and insurance reasons, but after reading the newsletter, she has realized the importance of testing and is going to find out if she is a carrier (*see our current website under Medical Info/Diagnostic Labs for carrier and diagnostic testing info).

In closing, I would like to express my sympathy on the loss of your daughter. I'm sure words can't ease the pain, but I want to thank you for turning your sorrow and grief into hope and support for others. Thank you!

Sincerely,
Gail and Harold White
New Bern, NC

***Thank you for your thoughtful and touching letter. Your comments were greatly appreciated! DLG

Dear fellow FOD parents: I would like to make a request to parents who receive this newsletter. If you have not been in touch with the Goulds, please take the time to write or call and let them know if this newsletter is of help to you. If you have been receiving the newsletter for some time and feel it is no longer of value to you, please inform the Goulds of this. They will continue to keep you a part of the Network if you wish. It is difficult for Deb and Dan to know how parents feel about the newsletter and Network if they do not hear from you.

Sincerely,
Jenny Carroll
Prairie du Sac, WI

***Thanks for your comments and all your help, Jenny!

Birth Announcement

Welcome to the world Gabriella Rae Marsella, born June 3, 1995, at 4lbs 14oz. She is the sister of Joseph who has VLCAD. Congratulations to Heather and Phillip Marsella of Poughkeepsie, NY.

Our Beautiful Victoria

First of all I want to thank you for all you do for families like us dealing with MCAD. Finally we can talk about MCAD with families who understand. Our daughter, Victoria Wade Gerstung, was born on April 22, 1993 after only 2 hours of labor and seemed to be the picture of health. We now had our son, Kristopher, who is 5, and our little girl with blonde hair and blue eyes. What more could anyone ask for?

When she turned 6 months, we took the family on vacation to my parents in Florida. Victoria caught a cold while we were there and didn't want to wake up one morning (sound familiar?). I thought it was the cold medicine and let her sleep a little longer. When she didn't wake up and was extremely lethargic, we decided to take her to the emergency room before we drove back to Atlanta. They immediately noticed the low blood sugar, started an IV, and transported her to a children's hospital. We stayed for a week running a lot of tests and then they discharged her stating they found nothing: that we may have given her too much medication. We followed up with our pediatrician when we returned. At 8 months she was standing, taking her first steps, and had a vocabulary of about 7 words, so we thought she was doing fine.

At her December checkup, the Dr. scheduled Victoria for a Sweat Test (Cystic Fibrosis) on January 3, 1994, because she started to have lots of colds and her sweat did not taste salty. A few days before the test, she caught a stomach virus and the Dr. placed her on Pedialyte and off formula. On the night of January 2, she was extremely upset and would not go to sleep. My husband woke me up at 2 a.m. and asked if I could take her because he needed some sleep. When I picked her up, she began saying "mama" in a very strange voice and I knew something was wrong. I placed her on my bed and when I looked down she began having seizures. We rushed her to the Emergency Room where she continued to have seizures and the nurses could not get to a vein for an IV. As I was holding her she stopped breathing and her lips turned blue. We screamed for the Dr. to come help her and they made us leave the room.

It seemed like someone had thrown me into a black hole and I didn't know what to think, say, or do. My husband ran up and down the halls trying to get answers from somebody. Then a nurse came to us and asked what religion we were so she could get someone there

for us. When she told me this, I thought my baby was gone but she said no and just wanted to get a minister for us. I lost myself and told her that I knew that my baby was going to be OK and that we would not need a minister because if God couldn't hear my prayers, He was not going to hear his.

Our neighbors took care of Kristopher while this was going on. And I stayed on my knees praying until the Dr. opened that door and told us she was OK for now and that they would transport her to a children's hospital. When we saw her, she was so still with tubes sticking out everywhere. She remained in ICU in a coma state for a week when we found out she had MCAD, RSV pneumonia and that they needed to put in a central line because all the veins had collapsed (which meant Surgery). During the procedure they punctured her lung so they had to take X-rays 3-4 times a day. At the time, I was pregnant with our third child and could not be with her during the test. Victoria and I lived in the hospital for 30 days with my husband trying to work and carry on a somewhat normal life for our son.

Victoria is now 2 years old and extremely beautiful. She has lost everything she knew before and suffers from Cerebral Palsy. We are currently waiting for her wheelchair to arrive and are working to get some type of communication box to assist her with speech. She is on Polycose and ProViMin formula, cornstarch in her bottles at night, and Carnitor® 4 times a day. Kristopher is a carrier and due to the trauma, we lost our third child.

We have moved to Texas and have been fortunate to find a nanny who we love and who cares for Victoria (She keeps me sane). One concern that I have, that someone may be able to address is I worry about placing Victoria in school to be with other children. Is the risk of her catching a cold or virus worth it?

Deb and Dan, there are no words to tell you how much it means to have people like you. To save one life or to keep one child from having to live with Cerebral Palsy needlessly, your family has made their place in heaven.

Sincerely,
Jackie L. Gerstung

***Your thoughtful comments are greatly appreciated. Thank you! DLG& DG

To Medicate or Not

We are a family that uses carnitine, mostly during sickness and stress, and do not use it regularly during our children's well period.

After one day of stomach flu, our first boy, James, died in December 1986, at 19 months. The real cause of his death was unknown to us until nearly 3 years later. At the time, our pediatricians suggested Reye's Syndrome as the cause. So we learned what we could about that, but were never convinced that that was it. Then one day the Reye's Syndrome

Foundation sent a paper about MCAD. This sounded so very much like James' sickness that we tested Larry, not yet 3 years, and Andrew, 1 year. Andrew tested positive and Larry negative. This was September 1989. We established a contact point with Dr. Kahler at Duke (*in 2000, in Australia) by phone and found that to be very helpful.

Andrew was on carnitine as recommended by Dr. Kahler for his first several years. I don't remember when we started to decrease the amount, possibly when he was about 4 years. The knowledge that there are doctors that do not prescribe it during well periods combined with the fact that he seemed to handle his disorder well ~ he has never shown MCAD symptoms ~ helped us in choosing to use carnitine more as a standby for sick times. Also, the consciousness that he was nearly through the most critical years, and knowing that we had the oral carnitine and the IV in our refrigerator for whenever we wanted it. He is age 7 now. For the last 2-3 years we have given it only during a cold or fever or asthma attack. He has since outgrown the asthma.

Philip was born in July 1990, and did not have MCAD. Laura came August 1992 and tested positive. If I recall correctly, knowing that breast milk is high in carnitine and because my babies didn't seem to have trouble with stomach flu while I was nursing, I did not start with the Carnitor® regularly. I had it on hand, though, and would give it if I felt worried about her ~ if she had a cold or if there was extra stress like more going away or irregular schedules. When she was 5 months old, she did have a fever and throwing up which in turn brought lethargy and hospitalization. I kept up the oral carnitine for a while until she seemed quite back to normal. Then at 7 months she was throwing up again from an ear infection. We almost admitted her to the hospital, but she responded to a shot in the emergency room and did not need to stay. At 17 months she had a stomach virus that required an IV.

She differed from Andrew in that she was more prone to stomach upset and we, along with our pediatrician, thought it wise to keep her on the carnitine regularly. The lethargy at 17 months was the last we had trouble. She is 3 years old now and this summer I have not been giving it regularly. I always like to have a good supply of carnitine on hand though, and enough IV for a day or two. Then we are prepared for an emergency.

I realize we are 'on our own' in a sense, and I would not recommend this to anyone not comfortable with it. We have had a Dr. that is supportive and very workable with us, which has meant a lot. Also, the availability of Dr. Kahler by phone has always been a comfort. We did take Andrew and Laura once to Duke for a visit.

We are interested in any information research could provide on IF and WHEN the carnitine is no longer necessary. (*many FODers have a secondary carnitine deficiency, so if their carnitine levels are low they most likely need the daily carnitine supplementation. Note from Deb ~ as part of our research with Dr. Roe, we took Kevin off of his maintenance dose (1 tablet 4 times/day) of carnitine for 30 days when he was well and at the end of that trial period his carnitine levels were extremely low so he was put back on the Carnitor®. EACH FODer presents/responds differently to his/her

metabolic disorder so speaking with your Dr./FOD expert is vital as to what treatment is suggested).

At what point would an MCAD person be able to fast without symptoms? Would they dare do on an extended fast as an adult? (*PLEASE see our current website under Medical Information and the article about the 45-year-old MCAD woman that died when she was fasted for surgery ~ since the MCAD individual cannot breakdown fats to be used as energy, when the sugar/carbos are no longer available, there could be MAJOR ramifications at ANY age.) We would greatly appreciate any feedback from the rest of the FOD Families and Professionals. Thank you for the effort put into the newsletter.

Sincerely,
Karen Carpenter
Metter, GA

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 an FOD. 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 1996, 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: Do you have any more information on resources or on other National Support Groups that stress parent-to-parent support for a variety of disorders?

Answer: As noted in the last MUMS newsletter, there are two excellent books containing lists of groups for rare disorders. Write to:

The Self-Help Sourcebook. American Self-Help Clearinghouse. St. Clares-Riverside Medical Center, 25 Pocono Rd., Denville, NJ 07834, 201-625-7101(\$11, overseas \$15)

Directory of National Genetic Voluntary & Related Resources ~ Alliance of Genetic Support Groups. 35 Wisconsin Circle, Suite 440. Chevy Chase, MD 20815-7015
800-336-GENE (\$22)

Local Information Network and Communication System (LINCS) is an on-line service that provides a National Resource Directory to individuals with specialized resource needs. 3041 Olcott Street, Santa Clara, CA 95054-3222, 408-727-5775

National Patient Air Transport Hotline. This service will help you find free or reduced rates for air travel for medical purposes. Information specialists refer families to the service that best fits their needs and situation. 800-296-1217.

Exceptional Parent Magazine for Families & Professionals. Provides information and lists 1400 support resources for children and young adults with disabilities and special health care needs. They encourage families to submit articles about their specific disorder. 209 Harvard Street, Suite 303, Brookline, MA 02146, 617-730-5800

Nutrition

Guidelines for Eating Right for a Healthier Heart (*see our current web page www.fodsupport.org and past newsletters for nutritional information).

Pharmaceutical Update

Some of you have requested information about Sigma-Tau Pharmaceuticals, Inc., the producers of the liquid, tablet, and IV Carnitor® and the financial backer of our newsletter. If you would like to correspond with them, call 1-800-447-0169 or write to them at 800 South Frederick Avenue, Suite 300, Gaithersburg, MD 20877.

Medical Update

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 across the country and world, as well as how you are coping as families with the different treatment protocols.

LCHAD UPDATE:

Jenny Carroll, who has been very helpful as far as locating LCHAD Families to refer to the FOD Family Support Group, has been in contact with several professionals. Some are researching the connection between LCHAD, DHA deficiency, and retinitis pigmentosa, which can cause blindness. Questionnaires were mailed out in December 1995, to 16 physicians who agreed to participate in a Survey on diet and long-term care of their LCHAD patients. As of January 22, only 4 surveys have been returned.

The University of Wisconsin Biochemical Genetics Program needs your help in reminding your physician to complete this survey. The information obtained from this survey will be available to the Drs. that participated. The questionnaire will benefit your child and other LCHAD patients. Your help is greatly appreciated.

If your Dr. did not receive a questionnaire, they can receive one by contacting Sandy van Calcar, MS, RD at: U. W. Waisman Center, Biochemical Genetics Program
1500 Highland Avenue, Madison, WI 608-263-5981

The responses from 2 physicians/researchers are below and are somewhat technical so you may need to have someone translate them for you!

“It seems that the deficiency of DHA could be a reason for retinitis pigmentosa. Nevertheless, it also seems that other additional factors might contribute. For instance: the concentration of hydroxy-acids. From our experience, hydroxy-acids are normal in urine during clinical remission, but are always slightly increased in plasma. I think it will be interesting to evaluate the concentration of antioxidants in these children, because the hydroxyacids can easily act as free radicals. I have passed your information along to Dr. Manuela Martinez, who is experimenting with DHA supplementation on patients with peroxisomal disorders. We will evaluate polyunsaturated fatty acids, as well as antioxidant concentrations in our patient and will inform you of any interesting results.”
Dr. Ortigosa's address is:

Hospital Nuestra Sra. de la Candelaria
Servicio de Pediatria
Carretera del Rosario, s/n
38010 Santa Druz de
Tenerife SPAIN

Sincerely,
Antonia Ribes, PhD
Corporacio Sanitaria
SPAIN

"Thank you very much for your letter. We are currently studying the fatty acid patterns in patients with Long Chain 3 hydroxy Acyl CoA Dehydrogenase (LCHAD) with a view to understanding the lipid changes better. Once we have done this then we can plan logical treatment that might include supplementation with DHA but also the use of antioxidants. I am sure you realize, too early to decide how we will proceed and obviously once we have some results, I will contact you again."

Sincerely.
J. V. Leonard
Professor of Paediatric Metabolic Diseases
Great Ormond Street Hospital for Children NHS Trust and the Institute of Child Health
Great Ormond Street, London WC1N 3JH 0171-405 9200

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Dr. Charles Roe and his colleagues at Baylor University Medical Center submitted 2 article abstracts for this issue. The articles were recently submitted for possible publication in **Biochemical and Molecular Medicine**. Our July issue may cover these articles/topics more in-depth.

‘Very Long Chain Acyl-CoA Dehydrogenase Deficiency: Successful Treatment of Acute Cardiomyopathy’

Very Long Chain Acyl-CoA Dehydrogenase (VLCAD) deficiency is a severe defect of mitochondrial fatty acid oxidation characterized by hypertrophic cardiomyopathy, pericardial effusion, steatosis, and hypoglycemia often resulting in death by 4-5 months of age. The onset of cardiomyopathy and pericardial effusion is insidious and sudden, necessitating early diagnosis and intervention to prevent death. A family affected with this defect is described in which dietary therapy with medium-chain triglycerides (MCT) was associated with rapid reversal of these severe clinical symptoms.

Diagnosis by acylcarnitine analysis in the neonatal period can provide the opportunity for early clinical intervention. Prenatal diagnosis from amniocytes by enzymology or in vitro probe of the fat oxidation pathway with deuterated fatty acid precursors has also been successful and permits intervention at birth. Of 9 affected children, 7 untreated cases died within the first several months, while the remaining 2 cases have survived when treated with medium chain triglycerides as the major source of dietary fat.

‘Improved Detection of the G 1528C Mutation In LCHAD Deficiency’

Long-chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency, an autosomal recessive disorder of fatty acid oxidation, is clinically characterized by skeletal myopathy, Reye-like syndrome, or sudden unexplained infant death. A common mutation G 1528C has recently been reported.

To avoid nonspecific amplification from a ‘pseudogene’ and potential complications, we have developed a nested PCR/Pst I digestion method. Here, we reported mutation studies in 11 additional unrelated patients with LCHAD deficiency. Genomic DNA fragments (117 bp) were amplified by the nested PCR digested with Pst I and subjected to electrophoresis on 12° polyacrylamide gel. Four patients were found to be homozygous for the G1528C mutation, 7 patients were compound heterozygous, indicating significant genetic heterogeneity. The G1528C mutation has been found on at least one allele in all patients with isolated LCHAD deficiency, suggesting that it is an excellent marker for this disease. This DNA test combined with tandem mass spectrometric in vitro probe analysis easily identifies affected individuals and carriers in families that are compound heterozygous for G1528C.

Newborn Screening

NEWS RELEASE: ABCD Family Support Group (*in 2000, merged with the United Mitochondrial Disease Foundation) Advocates for Nationwide Neonatal Screening
Contact: Meridith Murray 703-827-8771 or 800-554-ABCD

Oak Brook, IL (October 10, 1995). A recent survey of pediatricians conducted by the Gallop organization showed metabolic disorders to be the second most difficult medical problem to diagnose. This survey has prompted a parent support group called Assistance

for Babies and Children with Carnitine Deficiency (ABCD) to call for nationwide neonatal screening.

“While this Survey provides dramatic evidence of the problems pediatricians have diagnosing metabolic disorders, the good news is that an easy and affordable screening test now exists.” said ABCD president, Eva Tameling. Using the PKU blood spot infants can be screened for 38 different metabolic disorders at a cost of less than \$20 per test.”

Tameling cites the similarity of symptoms of carnitine deficiency to other common ailments as the reason why diagnosis is very difficult for physicians. These symptoms include severe lethargy, failure to thrive, diarrhea, poor muscle tone, vomiting and/ or constant and recurring infections.

In the Gallup survey, pediatricians reported an average of 14 patients, with one or more of these symptoms, who had not responded to normal treatment. “This means as many as 450,000 infants and children could be suffering needlessly from undiagnosed metabolic problems.” said Tameling.

Undiagnosed carnitine deficiencies have been linked to cardiomyopathy, severe developmental disabilities, and according to some experts, as many as 25% or more cases of Sudden Infant Death Syndrome (SIDS).

ABCD has been active in promoting awareness of carnitine deficiency, working to educate both doctors and parents. Now that an inexpensive test is available, it is focusing its efforts on convincing health policy makers to support widespread neonatal screening to identify newborns with metabolic disorders.

In Pennsylvania, more than 80 hospitals are participating in such a screening program. The program was developed and is being implemented under the direction of Edwin W. Naylor, PhD, the president and lab director of Neo-Gen Screening in Pittsburgh, PA.

Noting that the majority of metabolic disorders have an effective treatment that works best when started as soon as possible after delivery. Naylor said “The concept is to catch them early, before they're sick. Neonatal screening can begin immediately to benefit tens of thousands of newborns each year.”

According to the study conducted by the Gallup organization, only 52% of pediatricians were familiar with carnitine deficiency and associated metabolic disorders, and of those, only 43% knew blood tests were necessary to confirm a diagnosis.

Carnitine, itself, is a natural substance produced by the body that is vital to the metabolic process. Carnitine deficiencies are classified as either primary, when the body fails to produce adequate carnitine, or secondary, when another illness or metabolic problem depletes the body's supply of carnitine.

“Knowing that there may be almost a half million children suffering now from these difficult to diagnose conditions,” said Tameling, “we will continue educating physicians about the symptoms of carnitine deficiency because symptomatic children can be diagnosed using the same simple and affordable test as newborns.”

“Screening is available now. Treatment is available now. It’s time to work together to reduce SIDS deaths and improve the quality of life of our infants and children.”

Survey Methodology

The survey results are based on telephone interviews with a national sample of 300 pediatricians currently in practice. Interviewing was conducted from March 9 through April 3, 1995. For results based on samples of this size, one can say with 95% confidence that the error attributable to sampling and other random effects could be plus or minus six percentage points. In addition to sampling error, question wording and practice difficulties in conducting surveys can introduce error or bias into the finding of opinion polls.

Question wordings:

In your opinion, what type of medical problem in children is hardest to diagnose?

(Do not read) (Open-ended and record all responses)

(If physician treats patients with listed symptoms ask: I see you have seen (number of patients) for (symptom). How many responded well or as expected to treatment and how many did not respond as expected to treatment and required additional work up?

(Open-ended and record actual #)

- A. Genetic Metabolic Disorders
- B. Cardiomyopathy
- C. Muscle weakness (hypotonia)
- D. Liver Disease
- E. Chronic TPN treatment
- F. Tx with valproic acid
- G. Premature infants receiving TPN treatment
- H. Hypoglycemia
- I. Kidney disease
- J. Failure to thrive
- K. Recurrent infections
- L. Diabetes

How familiar are you with carnitine deficiency and its symptoms?

Are you very familiar? fairly familiar? or not familiar?

What type of lab measurements, if any, are used to confirm the diagnosis of carnitine deficiency? (Open-ended and record all responses)

Love Messages

(Please see our most current online issue)

We would like to express our sincere condolences to the Westman Family of El Cajon, CA, at the death of their second son, Beau, on February 19, 1995. Our thoughts and prayers are with you.

We were also very sorry to hear that Jenny Carroll had recently lost twins when she was 10 weeks pregnant. It is not known at this time if this was related to LCHAD which Sarah died from in September 1992 and which Jane is living with. We're thinking of all of you.

We may not ever understand why we suffer or be able to control the forces that cause our suffering, but we can have a lot to say about what the suffering does to us and what sort of people we become because of it.

Harold Kushner

When Bad Things Happen To Good People

Donations Received

Even though our newsletter is provided through the generosity of Sigma-Tau Pharmaceuticals, Inc., we greatly appreciate any family or professional donations (however, because we are not a non-profit, they are not tax-deductible). Thank you to Kathleen and Patrick McCreesh of Monte Sereno, CA, and Mary and James D'Ambra of Hanover, MA for their generous donations in 1995.

Reminders

I have listed all the NEW families that returned the Family Questionnaire. If you do not see your name listed, it means you did not return the questionnaire. So please take the time to fill it out and return it to me so I can list you in our next Issue. Thank you.

NEWSLETTER HELP NEEDED!!

Does anyone know how to make Mailing Labels on their computer? I would like to be able to run off labels from the Family and Professional Lists instead of handwriting over 250 envelopes for each Issue! If you can help out, write or call Deb by May 15.

January 1996

Volume 6 Issue 1

[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, Newborn screening, treatment recommendations, research, and names of FOD researchers/Labs.]