

CNS Family Newsletter

First Edition

March 01, 2001

*A Note From Timothy & Kathleen Hiscock

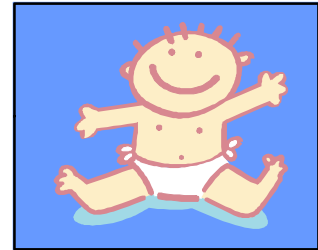
Hello! We hope you will find this newsletter as a helpful way to stay connected to other CNS families and to learn of current and upcoming news. Our son, Thomas, who is 18 months old, has CNS Type I. We felt fortunate that we were able to locate other CNS families through the internet and by telephone. We felt that we could organize a newsletter to improve communication with the CNS community. We will send a newsletter out twice a year, and more if we have a sufficient amount of information provided by you.

It's About Time!

I am very excited about starting our first Crigler-Najjar Newsletter! This is something I wished would have happened since Derick was six-months old. That was eleven years ago! Thanks to Kathleen and Timothy Hiscock for making it happen! I hope it can be an informative newsletter; one that is sharing and caring.

Katie Martin

Recent Births



Eugene and Vera Martin of East Earl, PA had a healthy baby boy named Ryan on 10/12/00. Ryan's sister, Regina, has CNS.

Michael and Eva Hoover of Milton, PA had a healthy baby girl in November. Ester May's brother, Eugene, has CNS.



Recent Publicity

Weekly Reader's Current Science recently did an article on CNS, which featured Derick and Amy Martin. The article focused on the future of gene repair as a possible cure for this disease. To order a copy of Current Science, call 1-800-446-3355. The issue number is 10; volume 86. The date it came out was 1/19/01.

The Jacksonville Community newspaper recently printed an article on CNS, which featured Ric DeVere and Melanie Bryant, both of Florida. You can see the full article by visiting the CNS website at www.crigler-najjar.com.

Going To Press

Do not forget to send in a story about your experiences with CNS so that Brookside Book Store & Printing can print and sell our book. Send your submission by **April 30**, to:

Floyd and Katie Martin
R.R. # 1 Box 177 A
Mifflinburg, PA 17844

Double space your story, using only one side of the paper. Pictures may be sent in, but will not be returned.



CNS Family Newsletter

Past and Present Events

On July 22, 2000, John and Miriam Martin hosted a CNS barbeque. 14 children with CNS attended. Dr. Morton and his wife, Caroline, also attended.

On September 16, 2000 the Clinic for Special Children held it annual auction. Several CNS families attended and had dinner at Yoder's with Dr. Michael Blaese of Valigen.

The weekend of October 06, 2001 there will be a CNS conference in Hamburg, Germany.

In Their Own Words

Marlin's Story

Our son Marlin was born on June 13, 1984. A day later, we took our healthy baby boy home from the hospital, or so we thought. As new parents, we did not think anything of it when he started to get yellow.

At five days old, my mother saw him again and said we better take him to the hospital right away as he was really yellow. The doctor said he needed to be admitted to the hospital to be put under lights. His bilirubin level was 18. He was in the hospital for three days and his points came down to 8. We thought that they would stay down, but of course, they did not, so we kept him in the sun a lot.

When Marlin was four weeks old, our doctor said that he needed to go for tests to see why his bilirubin was not staying down. We went to Hershey Medical Center in Hershey, Pennsylvania. When we got there, they took a blood test and Marlin's points were up to 30. They did a blood transfusion right away. Years later, Dr. Morton told us that a blood transfusion would not have been necessary.

At the hospital, they made him fast for 12 hours so they could do a stomach test to see if he had CNS. A tube was put through his nose down to his stomach to get a bile sample to send to a lab in California. It took a couple of days until the results confirmed that Marlin had CNS Type I. We knew a little about the disorder because my cousin had a son who died of CNS at two years old.

Marlin stayed in the hospital for four days. We got him a light to use at home, and he was put on a medication called Agar. It came in a powder form which I had to mix with his formula. It did not dissolve in his formula; it was like sand. He took it in his bottle until he was five years old, and then I thought he could drink it out of a cup with a straw, but he would not drink it. We didn't feel that Agar ever helped his levels at all. When he was on Agar, his bilirubin level was 18. After he was off Agar for two weeks, his bilirubin level was 17.

At five years old, Marlin's doctor, Dr. Jeffrey Masels of Hershey Medical Center, moved to Michigan. At the same time, our family doctor quit too. The new doctor did not know anything about CNS and did not take much of an interest in it either. So we were without a doctor who understood CNS. Marlin did not get seriously ill during that time, but we took him to the doctor for flu shots and for blood tests. At the time, we were very uneducated about the disorder.

At nine years old, we started going to Doctor Morton, and we were very pleased with the care we got from him. It was great to use a doctor who understood the disorder. Between the ages of nine and thirteen, Marlin's levels ran between 22 and 28. After he started taking Ursodiol, his levels ran between 16 and 21.

He sleeps under 16 four-foot bulbs between 8-8 1/2 hours each night. His nights are not as long as they used to be when he was younger. He currently takes 16,000 mg. of creatine a day as he get tired, mostly in his legs.

CNS Family Newsletter

Marlin's Story continued

Over the years we used to wonder if we should have a liver transplant done, but we are never quite ready for that step. After we heard about gene repair, we are glad that we have not done a liver transplant. We are still hoping and praying that gene repair will be available in due time.

Marlin has an air conditioner in his room during the summer, as we can't open the windows because the blue lights attract the bugs. Even if the windows are closed, some bugs and flies manage to get in and pest him so that he cannot sleep.

Leon and Lydia A. Newswanger

Gabriëlla- a patient's story

Before I begin my story let me first introduce myself. My name is Gabriëlla. I am 25, I live in The Netherlands and I have Crigler-Najjar syndrome type II. I don't have to sleep under lights, because the medication, formally used for epilepsy, called Phenobarbital works well for me.

Living with Crigler-Najjar is a special experience. When I was little the children at school used to call me names like "Chinese", "Indian", "Yellow" and "Vanilla". Those kind of words hurt for a child that can't help having an illness that produces more yellow color than any other person. I never quite understood those children because they all knew what it was that was causing that strange yellow color. Still they kept on calling me names. People can act strangely to something they don't know or somehow can't understand.

I later learned that when I started to visit the hospitals for other reasons than my routine Crigler-check-up. People always assumed that I had the temporary contagious jaundice disease. Assuming that I was ill with that disease would I be walking around, instead of being in my bed getting rest?? I don't think so.

These kind of things are everyday - well ordinary - things for a patient with Crigler-Najjar to encounter. It teaches you that some people are just scared. It teaches you to stand up for yourself and make clear that what you have isn't contagious. It is not something temporary. And I will get ill if I don't take my medication! It has happened twice now that I didn't take enough medication. Once by mistake; the attention was going out to my ankle-operation and the doctors forgot about Crigler-Najjar for a moment and once as an experiment. I had trouble with headaches. My doctor suggested a reduction of my medication. I was used to taking 150 mg per day. I dropped it only to 120 mg and my bilirubin went up within two weeks. I became slow in thinking and acting. I was also very moody. Then I realized that it was not going as it should, so I stopped the experiment.

Everyone who doesn't have Crigler-Najjar has to realize that this is a very delicate disease. A patient can live very good with it as long as the patient listens to his or her body. A patient needs others help, not their judgments. Like for instance I don't have to use lights. I have met people who have the opinion that I am therefore not ill enough. Don't ever judge someone about something you know nothing about. We need to do this together; by talking, writing, asking, by meeting each other. Supporting each other. That is the only way that we can learn to make the lives of the patients as good as possible.

To the patients I would like to say listen to your body. It tells you when something is wrong. I can feel it when I'll be getting ill because of Crigler-Najjar. It happens when I am tired and still have a lot to do. My family and friends and people at work help me, remind me of Crigler-Najjar. That's the way that I have found to work

CNS Family Newsletter

Gabriëlla- a patient's story continued

best. Also no doctor can tell you how you are feeling. Only you can. A bilirubin level doesn't say everything. It is a guideline, but you will learn to know when you need rest. I know my limits. And you will too. You will learn by falling down and getting up again. But hey, that's how it goes for everyone, Crigler-Najjar or not.

Pediatric Liver Transplantation

Therapeutic decision making in the treatment of Crigler-Najjar Syndrome Type I is clearly complex. Conservative medical management is efficacious but has persistent risks of unanticipated decompensation with the development of irreversible brain injury from kernicterus. In addition, conservative medical management imposes a difficult life-style, which clearly affects quality of life. Advances in gene therapy present great promise for the treatment of this disorder, although the time frame for these advances is unpredictable. Liver transplantation remains an important option in the treatment of Crigler-Najjar Syndrome Type I and the following is an overview of the process of liver transplantation.

Standard orthotopic liver transplantation involves the removal of the native liver with subsequent replacement of a graft in the same position. Typically the graft can come from two sources, cadaveric or living-related. Cadaveric organs come from individuals who have died in circumstances where liver function is intact. Cadaveric livers can be utilized whole or can be either split or reduced so that a liver from an adult can be made to fit into a child. Cadaveric grafts are allocated according to a waiting list. Children with Crigler-Najjar Syndrome Type I are afforded special consideration in waiting. Waiting times for organs varies by region and can range from many months to several years. Living donor grafts typically come from close relatives of the affected individual, although good samaritan donation has occurred in special circumstances. Parents of children with Crigler-Najjar type I are by typically carriers for the disease with diminished but adequate bilirubin conjugation capacity. As such they are potential donors, but should be carefully assessed. Being a living-related liver donor carries the risks of major abdominal surgery, including most significantly a risk of death that ranges from 1:1000 to 1:5000. A typical donor may be hospitalized for four to seven days and can resume normal activities four to twelve weeks after donation.

The typical orthotopic liver transplant takes approximately 4 to 8 hours. Recipients initially recover from surgery in an intensive care setting for a period of one to seven days. Most children are ready for discharge from the hospital two to four weeks after surgery. The initial life after liver transplantation is complicated and requires a number of medications, office visits and blood work. This period of intense medical evaluation typically lasts until three months after transplantation. During the first year after liver transplantation, readmission to the hospital for complications is common. These complications can include infection, rejection and surgical problems. Medical care subsequent to the first year after liver transplantation is much less cumbersome. Immunosuppression regimens typically require the administration of one or two medications. Blood testing is done every eight to twelve weeks and physician visits are required quarterly. Longterm risks after transplantation are decreased after the first year but still include infection, rejection and the potential for the development of malignancy. In addition, adherence to medical regimens is critical as in the current era immunosuppression must be maintained for life.

The overall efficacy and quality of life after liver transplantation is excellent. One year survival rates at major centers involved in pediatric liver transplantation range from 85 to 95%. Longterm survival rates are between 70 and 85%. Technical improvements in liver transplantation continue to increase the success of this procedure. Innovative procedures have been performed which include transplantation of liver

CNS Family Newsletter

Pediatric Liver/Liver Transplant Program Continued

cells through a catheter and replacement of only part of the native liver. The advantage of the first procedure is the avoidance of surgical risk. Both new procedures do not necessarily commit the recipient to this therapy life-long as the native liver remains in place. In the context of unacceptable complications of immunosuppression the transplanted liver (cells) can be sacrificed. These innovations remain investigational. In addition advances in post-transplant medical regimens have decreased the risk of significant complications like malignancy and have improved the quality of life after transplantation. Of longterm survivors after liver transplantation 60 to 75% have an outstanding outcome with normal liver function and a normal way of life. Growth and school performance is typically normal. Many of these children who have undergone liver transplantation have gone on to have families of their own.

Sukru Emre MD
Surgical Director

Benjamin Shneider MD
Medical Director

Pediatric Liver/Liver Transplant Program
Mount Sinai School of Medicine
New York, NY
212 241 6227

Helpful Hints

Light Bulbs

Dauphin Electric carries the Philips bulbs for your phototherapy unit. They have the 2-foot F20T12-BB and the 4-foot F40BB bulbs. Find them on the web at www.Dauphinelectric.com. For prices, email herb@dauphinelectric.com. Their phone number is 570-523-3232. Their fax number is 570-524-4280.

Other

For reasonable prices on Creatine powder or L-Carnitine check out
VITAMIN WORLD
1-800-381-9000 or
vitaminworld.com

CNS Family Newsletter

From the Children

In Derick's Words

God could have made all leaves alike and all snowflakes alike, but he did not. He made them all different. He made all of us different too. No two people are exactly alike.

You are a special, one-of-a-kind individual.

"Though there are billions of people on earth, God knows and loves me. I am a special someone. There's only one me!"

Derick Martin

A Note From Us

Next Issue

We will be gathering information to put in the next issue of the CNS Newsletter. We hope to have it done prior to the annual benefit auction in support of the Clinic for Special Children. Please send in anything you would like to see included.

Addresses

We will soon be sending you a list of names and addresses of the CNS families who are on our mailing list, so you can contact other families if you wish. If you do not wish to have your name and address included in this list, please let us know.

Important Notice

****This newsletter does not provide medical advice. Contact your health care provider before making any treatment changes.**

CNS Family Newsletter

Photos of the Famous



Thomas and Amy



Dr. Morton with ten of the fourteen CNS patients that attended the gathering at the home of John & Miriam Martin



benefit auction for the Clinic for Special Children in Leola, PA



several CNS families dined together after the benefit auction in September of 2000

*** Please send us articles and pictures for the next issue.**

