



NATIONAL NIEMANN-PICK DISEASE FOUNDATION, INC.



Volume 1, Issue 1

Spring 2002

FAMILY SUPPORT SUPPLEMENT

Families "In The News" and Raising Awareness

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On March 28, 2002 Congressman Mark Foley (Fl) held a Press conference at the Good Samaritan Medical Center in West Palm Beach Florida. The purpose of the press conference was to lobby for support for two landmark pieces of legislation *The Rare Diseases Act of 2002*, (H.R. 4013) and the *Rare Diseases Orphan Product Development Act of 2002*. (H.R.4014) This legislation would provide \$25 million dollars annually to the office of Rare diseases (ORD) and the Orphan Product Research Grant. Currently their annual funding is \$14 million dollars annually. If this legislation is approved by congress and then President Bush, the funding could begin as early as this year.

The National Office of Rare Disorders (NORD), of which NNPDF is a supporting member, sent out a request for families with rare disorders to attend this press conference. We are pleased to announce the NNPDF was very well represented by seven NPD family members and three HFTP members from that area. Amy Lee of West Palm Beach was asked to be one of the speakers at the press conference. Amy's emotionally charged message and Sherry Richardson who also attended with her son Ryan were featured in the majority of the media coverage on the event.

Amy Lee stated the following after the

event,

"The NNPDF is a supporting member of NORD. Because I am in West Palm Beach, and because my family has experienced first hand, the tribulations of having a child with an

"orphan" disease, I jumped on the opportunity to be one of three speakers at the press conference. Although public speaking is not my forte', I did my college best to represent all of our families.

Sherry Richardson, recognizing the importance of the legislation, was on the front row with Ryan (age 1 1/2, NP-C) in her lap. All of us were proudly wearing our Fox NFL NPD lapel pins!" (Please refer to page 5 in this packet for a copy of Amy Lee's presentation.)

Sherry Richardson contacted the foundation after the press conference with a note of heartfelt thanks.

Continued on page 4....



This pin was worn by the FOX NFL Sportscast team during their Sunday, January 6th, 2002 broadcast.

NNPDF—A Special Family Connection

Dear National Niemann Pick Disease Foundation Family,

Just when I thought I could be alone in my life preserver with no caring about what my child is going thru, my "family" comes along and wraps me in its' arms and rocks me to sleep with a security that I know will only

last a bit but is welcomed just because it comes from this family.

Thank you and much love,
A NP Mom



Foundation Office Updates

◆ Annual Membership Renewals Now Due

The annual memberships with the foundation expire on December 31st of each year. The \$20 membership fees assist the foundation with the printing and postage expenses related to the newsletters, family directory and family support documents. If you need to renew your membership, please complete the membership application form at the back of this packet and forward to the foundation office.

◆ 2002 - 2003 Family Directories

The new family directories were mailed out to NPD immediate family members in February. If you are a foundation member with an afflicted NPD family member and did not receive a directory, please contact the office. **NOTE:** During the first printing of the directory a page of family names was inadvertently left out of the directory. Updated pages have been forwarded for insertion.

◆ Family Conference Packets

The 10th Annual NNPFD Family Support and Medical Conference will be held in New Orleans, LA on July 18th—21st, 2002. If you have not received the registration packet please contact the office.

Deadlines: May 1st, 2002

Conference Registration, Hotel Reservations, Helping Hand Stipend Applications, First Time Fund Raiser Contest Results.

Foundation Office Contact Information

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Important Contacts:

Niemann-Pick Regional Representatives

Please contact your regional representative for information on the many services and family support needs offered by the Foundation.

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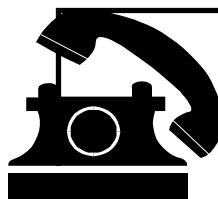
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(Note: All of the above members of the board & consultants to the foundation volunteer their time and expertise to NNPDF. Thank you.)

Calling All Contacts:



There are many NNPDF projects that can be helped by business contacts you, your family, friends or co-workers might have. We are always trying to minimize operating expenses and costs related to our programs and services. In-kind donations that absorb either all or part of a project can make a big difference to our bottom line. The more costs that are offset by in-kind donations—the more funds that we can channel to research. Publications, including the newsletter, the family directory, “They have only their childhood..”, and educational brochures can benefit from in-kind donations of paper or printing. Professional services, equipment and supplies used by NNPDF are similar to those at any small business, including general office supplies, grant writing, telecommunications services, photocopy machine, etc. Please keep the foundation in mind.

The Ara Parseghian Medical Research Foundation is pleased to host and sponsor an International Conference for Niemann-Pick C Disease in Tucson, Arizona on May 29-31, 2003. The 3-day conference will be held at the Westin La Paloma Resort and Spa.

For more information, please contact Glen Shepherd, Executive Director, APMRF 1760 E. River Road, Suite 115; Tucson, AZ Telephone: 1-520-577-5106; E-mail: victory@parseghian.org. “A Goal For Life”

We're Looking For:

- ◆ *Seasonal Photos of your children for use in a NPD Annual Calendar.*
- ◆ *Holiday drawings by your NP child for foundation note cards.*
- ◆ *Articles of interest for use in the Family Support Newsletter.*
- ◆ *Thank you to all those who have sent in photos & articles for use the Family Support Inserts.*

Fundraising News: “Meeting The Challenge”

The year 2002 brings with it a new promise and new hope as the NNPDF turns 10 years old. It is amazing when one thinks about the courage of those initial handful of families who took the initiative to establish an organization to offer family support and to foster scientific research for a cure for their affected children.

It has taken a team effort to propel the foundation over the past 10 years and it will take a team effort to move the organization ahead today and into the future. The top priority for all of us – volunteer leaders, foundation members and board members, the Scientific Advisory Committee, professional staff and donors alike – are the children and family members with NPD. For all those who have lost their lives to Niemann-Pick Disease, for those who are just beginning or those who are in the midst of their battles against this relentless illness, and for all the children of the future who deserve a life free from disease, we must solidify our partnership.

This year the foundation will focus on a nationwide awareness campaign and fund raising events to benefit the NNPDF and its goals. I encourage each family to “Meet the Challenge” and look within their family struc-

ture, friends and community support to create their own “team” and sponsor a fund raising event during the year 2002.

Now is the time to support the organization in anyway you can. Time and talent as well as financial support are all required to make this next year a true milestone in the foundations history. Each of you will have an opportunity to support the foundations fund raising goals this year and I hope you and your “team” will all join in the challenge.

I want to thank all of you who have so warmly welcomed me to the NNPDF community this past year. I look forward to working with you throughout the year ahead and to meeting many of you at the family conference in July.

Nadine Hill; NNPDF Associate Director



“Rare Diseases Press Conference - Family Feedback”

Continued from page 1...

“E-mail from Sherry Richardson regarding the Rare Diseases Press Conference”

Hello to all:

Today (3/28/02) Amy Lee (aunt of Jimmy, 14 w/NPC) did a remarkable job in representing the National Niemann Pick Foundation, at a press conference for the Rare Disease Orphan Product Development Act, H.R. 4014. Amy talked about the foundation on how it came about and told of her nephews fight with NPD. I can say that tears were flowing in the entire room. People who will watch our story tonight will know how serious Niemann Pick Disease is and that it is taking the lives of so many innocent children and adults.

I was very happy to hear from Congressman Foley that within the next two weeks, President Bush will be signing this bill that will give researches of rare diseases 100 million dollars broken into a four year period. This is a wonderful day for the foundation and our kids. I took Ryan and my oldest daughter so the public can see first hand what Niemann Pick really is. I want to thank Amy Lee for a job well done and I know it was tough getting through that, but I thank you from the bottom of my heart.

One thing we can all do to help is to contact your local Congressman and tell them to sign H.R. 4014, The Rare Disease Orphan Product Development Act of 2002.

Thinking of you, Sherry Richardson - Ryan NPC 22 months (Please refer to Amy’s speech on facing page.)

Editors Note: Amy Lee and her team of co-workers at HFTP have been instrumental in raising awareness into the plight of NPD individuals and the work of the NPD foundation. Thanks Amy.

NNPDF Family Member, Amy Lee, Represents Foundation at Press Conference

Speaking Notes – Amy Lee (Thursday 3/28/02)

Good morning Congressman Foley, and members of the media. My name is Amy Lee, like the others who are speaking this morning, I'm here to talk to you about a disease most people have never heard of. It is called Niemann-Pick Disease and is usually referred to as NPD. My nephew Jimmy Dees has NPD.

I work to raise funds for the National Niemann-Pick Disease Foundation, a sponsor member of the National Organization of Rare Disorders (NORD). For the second year in a row my friends, family and associates are eagerly preparing for our upcoming charity golf tournament in June. The proceeds from which will benefit the NNPDF's family services and research programs. The NNPDF was established in 1992 and is a voluntary, non-profit organization comprised of parents, relatives and friends committed to finding a cure for their children.

All of the diseases that fit the criteria of an "Orphan Disease" have many things in common – when your child or your loved one is afflicted, you deal not only with the heartache of their suffering, but desperation in your search to learn just what you are facing. What is it? What causes it? How do you treat it? And - Is there a cure? The common experience of those afflicted with a rare disease is unbearably stressful and painful, being shuffled from specialist to specialist, being subjected to test after test. This desperately needed and genuinely welcomed legislation will fund research grants that could result in a dramatic increase in scientific knowledge that would lead to a greater understanding of rare "orphan" diseases.

Not knowing fills you with overwhelming fear and the feeling of isolation. It consumes your life, and changes your whole family.

NPD has changed the Richardson's lives. Sherry is here today with her son Ryan who is 22 months old - and a victim.

Niemann-Pick Disease affects mostly young children. It is an inherited metabolic disorder that is dangerously unpredictable and always fatal. In some cases the onset of symptoms are subtle, like difficulty with upward or downward eye movements, clumsiness or unsteady gait, learning difficulties or the inability to perfect fine motor skills, making the probability of a diagnosis at this stage - minimal. Until the onset of the more severe symptoms like seizures, difficulty swallowing, enlarged liver or spleen, and sudden loss of muscle tone, it is difficult to even draw medical attention.

My nephew, James Alfred Dees the 4th, is a remarkable young man. We all call him Jimmy. When he could, he called me his favorite Aunt. As a young child he was like every other healthy, happy child who was a little awkward. When he was seven to eight years old he began showing subtle symptoms of NPD. As time progressed the

symptoms got worse.

The doctors horrified his father, my brother, with speculation, using terms such as Fetal Alcohol Poisoning and Shaken Baby Syndrome. On December 31st, 1996 - Jimmy - at nine years old had his first violent seizure. There were more seizures in the months that followed. Finally - in July of 1998 Nemours Clinic in Jacksonville Florida delivered an accurate diagnosis - Niemann-Pick Disease. Jimmy was ten years old.

Today Jimmy is 14. He is either in his wheelchair or in his bed. His condition is very fragile and requires round the clock licensed nursing care. He breathes through a tracheotomy tube in his throat. He is fed through a gastrostomy tube in his stomach. It has been exactly two years this month since we last heard his precious voice. On his good days we can get a couple of quality "Three Musketeer" sword fights out of him. On his not so good days we may get a thumbs up.

Rare disease means most of the time you don't get the right diagnosis at first. Often it takes years. The National Commission on Orphan Diseases estimates that one third of the people suffering from a rare disease will not be accurately diagnosed for three years or more following the onset of symptoms.

What all of the families represented in this legislation share - is the heartache of not knowing, the heartache of a misdiagnosis at one time or another, and the overwhelming feelings of fear, isolation and helplessness.

Congressman Foley on behalf of the NNPDF, I thank you for your compassion and insight. For realizing that if the government does not provide these funds, no one else will.

And I thank you on behalf of my daughter Nicola, who is 19, healthy, and here today. She has a renewed hope that the increase in funding will provide the peace of mind she so desperately seeks with genetic testing. The genetic testing that has not yet been perfected to the point of accurately detecting whether or not she is a carrier who could potentially pass Niemann-Pick Disease on to her children.

Thank you. Amy Lee



Web Site Updates At "www.nnpdf.org"



NNPDF Store

The web site continued to have an increase in traffic during 2001, averaging over 2,500 visits per month to the NNPDF mail page. Since the site was created in August 1997, there has been a consistent increase in monthly traffic. To date, there have been over 83,000 visits to the site. The rate of increase slowed in 2001, probably because of delays in the OGT-918 trial. A significant boost in visits occurred after the Fox Sports broadcast (Jan. 2002) when over 1,110 visits were recorded in one week. With renewed progress in the OGT-918 trial, we anticipate that the rate of visits will increase again in the coming year.

New Pages or Major Revisions

Keeping the website "fresh" encourages repeat visit, helps members feel part of the Foundation and promotes the Foundation as a professional organization. However, it is the researchers and families (particularly Board members) who create the news that we report - **please let us know of any news items that come to your attention.**

Although there were no major additions to the NNPDF web site in the past year, many of the 125 pages on the site were updated. In addition, a "NNPDF Website on CD" is now available for people with limited Internet access.

The "Store" pages highlight items and companies that may be of interest to members, although the Foundation does not actually sell any products. The "Store" pages were reviewed and the NNPDF Store was expanded. The NNPDF Store features public awareness items, such as the poster, bumper stickers and lapel pin, with a donation suggested for each item.

Mailing Lists

Mailing list activities grew significantly in 2001. There are now four mailing lists. All mailing lists are hosted by Yahoo.

- General list
- Type B list
- Sibling list
- Board member list

Membership in the general mailing list has grown again, with approximately 150 people subscribed at the end of the year. The general list has the largest membership and the greatest amount of activity.

FundRaising

NNPDF is registered with Helping.Org which allows us to accept contributions over the Internet. Donations via Helping.Org have been increasing, although it is still a small percentage of total revenue. Placement of a donation button on the main pages has made this option more visible.

Research and Grants

The new online process to handle research grants is functioning quite well. Eleven grant applications were received and forwarded to the SAB. The process is allowing the SAB and Research Subcommittee more time to concentrate on the merits of grants.

Future Direction

The Internet is becoming a wider method of communication and our website is increasingly visible - not only to members but also to potential donors, state regulators, medical review groups, and other third parties. Because the website contains a wide variety of information from so many sources, there is a potential for questions to be raised, especially with fund raising, medical information, and truth in charities oversight.

Goals for 2002

We will continue to expand the science features on the website to make the concepts understandable to the lay person. We also hope to improve the navigation on the site to make it easier to find specific pages. We appreciate your feedback and want to make this your website.

Respectively submitted by your Web masters, Doug and Janet Pease

NNPDF has available a number of items to help with your fund raising event or public awareness campaign. These items were originally created by members and supporters and donated to the Foundation.

Items provided by the Foundation are free of charge. However, a small donation is requested to offset the cost of this service.

NNPDF T-shirt

T-shirts with the Foundation logo and website address. Check with the NNPDF office concerning size availability. *Contributed by Evelyn Hughes, in memory of her son Jason Hughes.*

Donation suggestion: \$10 each

NNPDF Bumper Stickers

"Help Us Cure Niemann-Pick Disease" with the Foundation logo, website address and toll free phone number.

Contributed by Janet and Doug Pease, in memory of their nephew Adam Ward and in honor of their niece Amanda Ward.

Donation suggestion: \$1 each

NNPDF Lapel Pin

Lapel pin as featured on Fox NFL Sports for NPD Sunday. (Refer to pin photo on front page.)

Contributed by Hunt and Annette Ozmer, in honor of their daughter Hunter and Bob Eadie, in honor of his son Kevin.

With special assistance from Rod Carter of Carter Communications.

Donation suggestion: \$5 each

NNPDF Poster

A striking poster with dozens of pictures of the NP children.

Contributed by Tim and Julie Kersteins in honor of their son Chandler.

Donation suggestion: \$1 each

NNPDF Website on CD

A useful CD-Rom of the foundation website for those with no Internet (or limited) access.

Contributed by Doug and Janet Pease. Suggested donation: \$5 each



Fund Raising News

Raising Awareness in 2002 - - - October is National NPD Awareness Month

The following fundraising events which have occurred or will occur in various localities contribute to the goal of reaching our needed monies for sponsoring research. We want to thank Melissa King, director of fundraising for her help in encouraging these families to participate in these fundraising efforts. Additionally we, thank all of the families and friends who have made these events so successful. I have included in this newsletter many examples of their efforts to illustrate their fundraising ideas. Special thanks go to all of the following.

First Time Fund Raiser Contest "kicks" off the NPD foundation's 2002 focus on raising awareness and funds into Niemann-Pick Disease for much needed research.

Warm up for the Family Conference in New Orleans by joining our **First Time Fund Raiser Contest**. The rules are easy

- (1) You must be a first time fund raiser
- (2) Receipts must be turned into the Foundation by July 7th
- (3) Officers, Board members, and their relatives are not eligible

The person or family who raises the most money wins the grand prize: complimentary Executive Level room during the Family Conference, with continental breakfast, evening hor'd oeuvres, and turn down service. The winner will also be recognized at the Conference and in the Newsletter.

If you need help or would like a copy of the Fund Raising Do's and Don'ts, contact Melissa King, Director of Fund Raising. Here are a few ideas for starters:

- Send out a Friends and Family letter
- Have a Church Supper
- Request a donation from your employer
- Have a Garage Sale
- Ask your friends for ideas!

Everyone participating will be First Timers - so everyone has a good chance at being the top fund raiser. The Fox NPD Sunday was a great way to start the year. Let's build on that experience. NNPDF depends on member fund raising for the money spent on research and support programs like the Family Conference. When more people raise money, more gets done to fight Niemann-Pick Disease.

For the Children ...

****Established Fund Raising Events Nationwide****

2nd Annual Lee King Benefit Buddy Tournament - - - June 8th, 2002



Plans are already underway for the 2nd Annual "Lee King Buddy Benefit Fishing Tournament" to be held on June 8th, 2002 on Lake Eufaula in Eufaula, Alabama. Hosted by Melissa and Jimmy King, along with Sam Williams, a long-time sup-

porter and friend of NPD foundation and the King family, this event is sure to please everyone involved. Last year's tournament raised over \$11,000 to benefit the foundation. As this was a first time event, the organizers were elated with these results. They have set their sites high and hope to double the benefit proceeds this year. Dedicated and enthusiastic volunteers were a key to helping this event prove so successful. Community involvement and donations towards raffle prizes and the silent auction made by local businesses were also instrumental in building excitement over the event. Of course, beautiful weather and anglers willing to take part in a day filled with fishing excitement and share their "fish stories" wrapped up a great event.



Pease/Ward NBA Hornets Raffle

The foundation office recently received an update from Doug and Janet Pease regarding their annual fundraiser project. This year they will be raffling a pair of season tickets to the NBA Hornets who are set to move from Charlotte to New Orleans next season. If they sell all of the 1,500 raffle tickets the proceeds would be over \$12,000! Ticket sales have been swift thus far and they have a committed group of contributors and volunteers waiting in the wings to help move this project along. Interested parties may contact Janet and Doug at webmasters@nnpdf.org.

"Golf Across American in 2002"

Annually there are a number of Golfing events held to benefit NNPDF. If you are interested in putting together an event - please contact the office.



GOLF "FORE" STACEY - GOLF CLASSIC

**June 29th, 2002 – Spring Creek Golf Center
Cold Spring, Wisconsin**

Barb and Gary Vorpahl, along with friends & family, and sponsored by AAL – Aid Association for Lutherans, will play host to this annual event. The 4-person scram-

(Continued on page 8)

Fund Raising Highlights - - continued

(Continued from page 7)

ble golf outing cost of \$30 per person includes golf, food, door prizes and flag prizes. Many local businesses continue to take part in the day by sponsoring holes, donating items for the silent auction and joining other participants for a sun & fun-filled day with the sticks.

5th Annual Hunter's Hope Golf Tournament
September 13th, 2002 - Roanoke, Virginia

Not to be forgotten is the Hunter's Hope Golf Tournament sponsored by Annette and Hunt Ozmer, of Roanoke, Virginia. Held annually in September, this Captain's Choice – Shot Gun Start event draws loyal participants year after year. Corporate, business and individual sponsors assist the event with a wide variety of donated goods and financial sponsorships. Numerous raffles, hole prizes and mulligans add excitement to the days events. The skilled (lucky) player who shoots a hold-in-one can drive away with the grand prize of a new car!!!



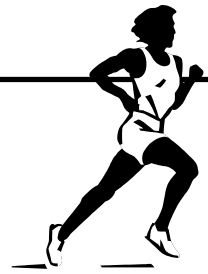
Fort Atkinson, WI Fundraising Highlights

The tradition of community support for the NNPDF and the Barb & Gary Vorpahl family continues to be strong in Fort Atkinson, WI. Local theatre veterans, Estelle Wiesmann and Bob Horton, presented three one-act plays on Thursday and Friday, March 22 and 23 at 7:30pm and on Sunday, March 24 at 2pm. The charity benefit "Dessert Theatre" offered delectable delicacies and fragrant coffees for the theatre goer during the intermission. Funds raised by this event will assist the foundation in procuring a National Niemann-Pick Disease Type C Center at Columbia University Medial Center in New York City. The new Natl. NP-C Center would act as the main coordinator on clinical issues and drug trials.



"Eighth Annual Bridge Extravaganza"

Local supporters of the NPD foundation and the Vorpahl's, whose daughter Stacey, a courageous and beautiful young 17 year old with NP-C, have also planned their Eighth Annual "Bridge Extravaganza" fundraiser to benefit NNPDF. Bridge card players of all levels are encouraged to join the fun on April 13, 2002 from 1pm-4:30pm at the First Congregational United Church of Christ in Fort Atkinson. The entire registration fee will be donated to the foundation for research. Friends and local business will pay all other expenses. Many door prizes and refreshments for all participants will be distributed along with prizes for first and second place teams.



"A Goal for Life"

Fun Run and Walk
October 26, 2002
Newport News Park
Newport News, VA

Brenda Eadie, mom to Kevin age 8 (NP-C), has notified the NPD foundation that she and many of her family and friends will again host "A Goal For Life" fun run & walk in October of this year. This is a great time to show your support for Niemann-Pick families by taking part in this fun-filled event. Registration for the fun run and walk starts at 9am with the run/walk beginning at 10 am.



How Can YOU Help To Support NNPDF?

Fund raising is an integral part of the NPD foundation. The support of families who raise monies annually during their various fund raising events supports many aspects of the foundation. They raised funds from friends and relatives, neighbors, local businesses and corporations and other community groups to provide the important family support services offered by NNPDF. Donations led to the hosting of unique family support and medical and scientific conferences to share the new knowledge, counseling services for parents trying to cope with the eventual death of their child, the formation of an incredible support network for affected families and numerous informational packets.

Melissa King, Director of Fund Raising, is currently working on updating the foundations "Fund Raising Packet" which will help families to get their events "off the ground". She has many ideas and suggestions that will fit any set of circumstances - so be sure to contact her for help. If you have had a successful fund raiser - we want to know about it. Please forward your information to the foundation office for inclusion in upcoming newsletters. Thanks.

Fund Raising can be fun & easy. Here are some ideas of events that families have held in the past.

- * Raffles
- * Dessert Theatre
- * Back-yard barbeque
- * Charity Nights at a Minor League Baseball Game
- * Bingo/Card/Trivia Game Night
- * Ice Cream Social
- * Sell products through home based business consultants fund raising (ie: Avon, Tupperware, MaryKay).
- * Wine-Tasting Parties
- * Lemonade stand
- * Rummage Sale
- * Flower sale

Of course, you can be creative and use your imagination!
Let us know of any great fund raising ideas that you have!



“Pennies From Heaven”



Terry Long, mother of James Long (NP-C), sent in the following note detailing a fund raising project that her Dad did on behalf of her son James and the NPD foundation.

Dear NNPDF,

Over the years I've read about various fund raising efforts for NPDF but I think my small story is worth telling.

My 87 year old Dad has been saving pennies for about 17 years. Each time we would visit him in North Carolina my son, Jim, would marvel at the pennies in the basket in his grandfather's bedroom.

Recently on a visit to Dad's we discussed cashing in the pennies. We knew there were too many to roll and no one could lift the basket! So I scooped them up into three different buckets. My Aunt and I then loaded the buckets into her car and off we went to the nearest coin counting machine. About 20 minutes later, we received a receipt for 12,173 pennies or \$121.73.

My Dad would like to donate this money to the NNPDF in memory of his only grandchild, our only child, Jim, who died on February 17, 2001 of Niemann-Pick Disease Type C at the age of 16.

A heroic fundraising effort by a devoted grandfather for a courageous grandson.

This is YOUR Newsletter!

**We Need Pictures....
Articles.....Your stories.....Your
Inspirational Moments.....**

I would like to thank all of you who contributed to this edition of the NNPDF Newsletter. I ask for your help and support in continuing this much-needed communication. We really do need your input!! We would like to include more pictures. Please send seasonal photo of your NPD child/adult for the newsletter.

Please submit information, articles, ideas, stories, parent tips, fundraising events, dietary tips, anything you come across that can be used in our newsletter to the foundation office.

Thanks.

**I found a penny today
Just laying on the ground.
But it's not just a penny,
This little coin I've found.
Found Pennies come from heaven,
That's what my Grandpa told me.
He said Angels toss them down.
Oh, how I loved that story.**

**He said when an Angel misses you,
They toss a penny down.
Sometimes just to cheer you up.
To make a smile out of your frown.
So, don't pass by that penny,
when you're feeling blue.
It may be a penny from heaven,
That an Angel's tossed to you.**

“Contact your local shopping centers with this creative fund raising idea.” Thanks to Holly Roberts, mom of Erin (Age 20 years, NP-C) for sending in this update.”

Annual Charity Night

The Annual Charity Night will be held at The Hanover Mall on Sunday, Nov 25. We are proud to announce that 100% of the ticket proceeds will benefit your charity! Charity Night raises funds for your organization with no overhead costs to you. The \$5 ticket sale proceeds are yours to keep.

The Hanover Mall will pay for the printing of the tickets and programs, arrange the door prizes, entertainment and music while allowing you to keep ALL the ticket proceeds. It's that easy! Just about everyone goes shopping in Nov & Dec for Christmas, Hanukkah or New Year's gifts for family or business associates. Charity Night is your opportunity to raise funds by asking people to do something they already do while making a small contribution to a cause they already support.

The opportunity for a special evening of shopping at major regional malls is an enticing one. The decorations, music, door prizes and special events simply add to the ambiance of their experience.

Your members will have a chance to select holiday merchandise while selection is at its' best. Many stores will have special sales and promotions offered only during Charity Night. In addition, the organization with the most ticket sales will receive an extra \$750 donation directly to their Charity!!
(The NPD foundation was the lucky recipient of the \$750 prize!)

*Watch your thoughts, for they become your words.
Watch your words, for they become your actions.
Watch your actions, for they become your habits.
Watch your habits, for they become your character.
Watch your character, for it becomes your destiny.*



A word from NPD Board Member - Kyle Hoffman Type B Family Representative

Hello to all NPD-B families. The National Niemann-Pick Disease Foundation (NNPDF) completed a very successful board meeting last week in St. Louis. I was fortunate enough to attend this meeting and have a short recap of some of the important issues at hand. The meeting minutes will come out some time in the future and I will be sure to fill you in on anything that I may have missed with this correspondence. I want to discuss three things - fundraising, family conference, and update on clinical trials. This e-mail is lengthy but I feel it has some very important information. Please be sure to read it in its entirety.

Fundraising

The NNPDF is a not for profit foundation totally funded by fundraising, donations, and memberships. I want to emphasize the importance of fundraising. The foundation does not receive large government grants. The money that the foundation uses to fund research, education and family support, family conference and other activities comes from us and our family and friends. There is a myth about fundraising - it is difficult. This is not true, it can be as simple as giving a family member, friend, or co-worker a NNPDF donation card. Golf fundraisers are nice and they do bring in numerous donations but we don't have to do one. A garage sale bringing in fifty or a hundred dollars will do. What matters is that we all are doing something. Last year approximately \$135,000 was raised through fundraising. Only a handful of type-c families raised this money. The foundation is looking for more representation by the type-b families. We are getting close to trials and a possible therapy, but it is not a cure. We need to work together to raise funds to find a cure for this disease. You will be receiving packets on fundraising and contest in the near future.

Family Conference

Last years conference was a huge success. We are looking forward to this year being the same. Doug and Janet Pease along with Missy and Jim Ward will be hosting the conference in New Orleans on July 18-20. I hope you will be able to attend. Genzyme gave the foundation several thousand dollars last year to help type-b families attend. We are hoping they will do the same this year. Please don't let money issues be a reason not to attend. The foundation does provide a stipend on a needs basis. This is a great opportunity to meet other families in the same boat as you. Also, doctors from Mount Sinai will be there with updates on the trials. The foundation is looking for ideas on what you would like to hear at the conference. Please send in any ideas you may have. I see some people have already done so, thank you.

Clinical Trials Update

The following is an update from Dr. Schuchman at Mount Sinai. The process of completing the international survey is still underway. This consists of collecting clinical and laboratory data on approximately eighty type b patients. The following is a list of sites involved in the survey - USA(Mnt. Sinai), France(Lyon), Italy(Trieste), Germany(Mainz), England(Manchester), and Brazil(Porto Allegra). Dr. Schuchman says the study should be completed by early spring. Everything is on track to begin phase 1 and 2 trials in the summer. This will likely be undertaken at Mnt. Sinai on a group of 6 - 10 patients. If the results look good a broad base phase 3 trial will begin at multiple sites. FDA must approve the phase 1 and 2 trial results before the trials can move on. Dr. Schuchman assures us that Genzyme has the ability to provide more than enough enzyme for the trials.

One concern that Dr. Schuchman has is the ability to complete the trials. He is worried there may not be enough participants that have enrolled in their database. He stated that the FDA would place some limitations on who can participate in the trials (i.e. Age, degree of clinical severity). He asks that if you have not contacted Mnt. Sinai to please do so (Dr. Schuchman - EDWARD.SCHUCHMAN@MSSM.EDU or Dr. Wasserstein - melissa.wasserstein@mssm.edu). By talking with them and being placed in their database does not mean you or your child will have to participate in the trials.

I want to be frank with everybody for a moment, please read this with an open mind. I know placing yourself or your child in a clinical trial is a scary proposition. Tiffney and I took Luke to see the doctors at Mnt. Sinai last spring after his diagnoses. We heard about the trials before we went up there to see them. Weighing the pros and cons we decided that we would participate if we could. This was not an easy thing to do. Unfortunately because of Luke's age, he is four; he is too young to participate. Even if you or your child is doing OK today we must think about down the road 5-10-20 years. This is a progressive disease; there is no escaping this reality. Luke seems to be a normal four year old except for his swollen liver and spleen. But we realize it is just a matter of time before other organs will be affected. Some people are already experiencing this affect. We must do something now while there is momentum. Because of the rarity of this disease, if something happens like not having enough patients to complete the trials everything could come to a grinding halt. Who knows when it would be started back up, if ever? The cold truth is that Genzyme is a publicly held company ran by a board of directors and its stockholders. If they decide to stop the project produces the enzyme for Gaucher disease, which is very similar to Nimann-pick type b. This treatment has been a great success with very little side effects.

Tracy,

Please insert the two-page document titled "Living with Niemann-Pick" from the magazine "Lifeline" pages 8 & 9 and place on these 2 pages of the Family Support (pages 11 & 12)

Thanks. Nadine

If you have to shrink the story via the photocopier. Thank is fine.



Tracy,
Please place page 9 of the LifeLine story here. Thanks.

Kyle Hoffman-Type B Family Update-continued from page 10

Lets show the foundation our support by getting out there and raising funds for our children, family, and friends. This year's family conference is going to be a blowout. Come and meet old friends and new ones alike. I have never been to New Orleans but I hear it is like no other place in the world. I hope to see you there. The clinical trials look like they are on track. Lets keep are fingers crossed and keep on praying for success. If anybody has any questions I can be reached with one of the following methods, typeb-nnpdf yahoo list serve, home e-mail khoffman@wwdb.org, home phone 816-847-0266.

Your friend

Kyle Hoffman
NNPDF Board Member
Type B Family Representative
1-816-847-0266
E-mail: khoffman@wwdb.org

National Niemann-Pick Disease Foundation, Inc.

Presents the

10th Annual Family Conference

***Astor Crowne Plaza
New Orleans, Louisiana***

July 18 - 21, 2002

Please contact the foundation office for a registration packet and join us for this information and helpful networking family session. This year's conference is being hosted by Missy and Jim Ward along with Janet and Doug Pease. One change to this year's conference is an invitation to families' physicians to attend the conference. Call the foundation office for additional information. Join us!



Men Do Cry

I heard quite often "men don't cry"
Though no one ever told me why
So when I fell and skinned my knee
No one came to comfort me.

As I grew to reasoned years
I learned to stifle any tears
Though "be a big boy" it began
Quite soon I learned to be a man.

The one long night I stood nearby
And helplessly watched my child die,
And quickly found to my surprise
All tearless talk was lies.

And still I cry and have no shame
I cannot play that "big boy" game
And openly without remorse
I let my sorrow take its course.

So those of you who cannot abide
A man you've seen whose often cried
Reach out to him with all your heart
As one whose life's been torn apart.

For mean do cry when they can see
Their loss of immortality
And tears will come in endless streams
When mindless fate destroys their dreams.

Foundation Membership Renewals Due

Please Note: All annual family memberships expire on December 31st of each year. If you have not yet had a chance to renew your membership—please complete the Membership Application form at the back of this packet and forward along with the \$20 annual membership fee.

Membership fees assist the foundation is offsetting the printing and mailing costs for newsletters, family

"In The News"

Newspaper articles which appeared in local print around the country are located at the back of this packet. We welcome all families to forward their local interest news stories for placement in the Family Support Packet. We enjoy hearing from you all.

Communicating With Health Care Professionals

Communication

Your description of your NPD child condition will aid the doctor and others in planning treatment in an atmosphere of harmony and rapport. You will need to be frank and clear about the information you bring to your doctor and do your best to talk to him/her about things that can't easily be explained.

Gather as much information as possible. This enables you to prepare for doctor visits with a list of intelligent questions in treating symptoms associated with this disease.

Have One Person to Turn to First

It is important to have one health care professional who knows your child well. This professional, your primary care provider, may be a local pediatrician, family doctor, nurse practitioner or other staff member at a clinic. The primary care provider has two functions. *One*, he or she is the person you turn to first; *two*, he or she is the person who helps you coordinate the services your child receives.

How Do You Choose a Primary Care Provider?

Begin your search for a primary care provider by thinking about what you need most from a health care professional. You may, for example, want to find someone who is easy to talk to or who has a particular interest in your child condition. On your first visits to doctors or other professionals, observe their reaction to you and your child.

- Do they make eye contact with your child? Talk directly to your child?
- Treat your child with the same concern as your other children?
- Do they really hear what you have to say?
- Do they accept your feelings?
- Is the professional interested in your questions?
- Do they respect your experience and skill as your child's primary caretaker.

Your Relationship with your child's doctor

When you have chosen a primary care provider, be sure that the doctor or professional knows you want them to take on this role. Establish an understanding on several key issues early in the relationship. For example, it helps to agree on what constitutes an emergency and what you should do if one occurs.

Think of your primary care provider as someone you can talk with:

- Discuss your child's growth and development as well as strictly medical issues.
- Talk about your child's progress as well as his or her problems.
- Tell the primary care provider about your fears and anxieties and about any changes in your family that might affect your child.
- Ask if you can make appointments just to talk at times when you have serious concerns about your child, and ask about fees for such consultations.

Making the Most of your time with the Doctor

Prepare for routine visits by thinking through the things you want to ask and/or tell the doctor **and write them down**. Be sure your child's medical history is up to date so you can answer the doctor's questions. Keep a Health History Form

The following suggestions will help you be more prepared to ask questions:

- Identify your concerns. Are you worried about specific things?
- Keep a running list of questions that occur to you between doctor's appointments.
- Make a list of any questions that your child might have. This is a good way to reinforce your child's role in his/her health care.
- After you and your child have identified and written down your concerns, consider them in order of importance. You will usually have a limited amount of time for questions.
- Make sure you understand the doctor's answers. Feel free to write down what the doctor says and ask him to repeat the answer if necessary.
- If you have to leave with some of your questions unanswered, ask if it would be convenient for you to call and ask the remaining questions.

Concerns

Parents often have concerns in the areas listed below. You may have concerns that do not appear on this list, and all these concerns may not be of equal importance to you.

Diagnosis

- Is a firm diagnosis possible at this time? If not, when?
- Is there anything to be gained by a second opinion?
- What are the immediate concerns? The long-term concerns?
- Will this condition get better, get worse, or stay the same

Medications

- What is the drug meant to do?
- Are there possible side effects?
- How much should the child take? At what times? With or between meals?
- Should the child take the medications until the symptoms disappear? Or until the medication is gone?
- What are the effects of combining this drug with other medication the child may be taking? (Include over-the counter drugs such as cough syrup, aspirin and aspirin substitutes.)

Tests

- What does the doctor expect to learn from the test?
- What stress or side effect can the test cause?
- How long will the test take?
- Referral to a specialist or Clinic
- Why is the child going to the specialist or clinic?
- What will the specialist do?
- Will you go to the specialist on an ongoing basis or for a limited period of time/
- Who will get the reports of the evaluation?
- How will the findings be coordinated with the child's overall health care?

Hospitalization

- Estimate the length of stay.
- What are the names of attending and consulting doctors?
- Will you be able to stay with the child?
- Does the hospital have a program to prepare children before they are admitted?
- What will it cost? What costs are covered by insurance and what costs are not covered?

Surgery

- Explain the surgery in everyday terms.
- What will happen if it is not done?
- How long will it take to recuperate?
- What are the possible complications?
- Is there anything to be gained by a second opinion?
- Will therapy or follow-up surgery be necessary?
- What will be the long-term effect of the surgery?

Insurance

- What procedures require pre-approval?
- What is your annual deductible?
- What is your lifetime cap?
- What protections do HIPPA and the new HHS Privacy Regulations Offer?

Stay sensitive to privacy issues. Who needs to know the information? If in doubt, don't disclose. Never give blanket disclosure authorization.

Remember

You know your child better than anyone. You are the expert on your child's care.

Take a proactive role in any medical decisions. Arm yourself with as much knowledge as possible to make informed decisions with any medical recommendations or procedures.

Communicating With Health Care Professionals

Tracy,
Please place a copy of the “Home File
Checklist” page here.

Thanks—Nadine

- - - For Your Information - - -

Health Care Web Sites

Clinical Trials

Get clinical trial and medical research info from the huge National Library of Medicine database. Type a word, phrase or disease name relating to a clinical trial into its search engine or search by clinical trial site or sponsor. www.clinicaltrials.gov

InfoNet

A service of the Johns Hopkins Medical Institutions, this site acts as a gateway to many health-related websites. Patient-advocacy services offer aid for everything from birth defects to Gulf Way Syndrome; they also cater to elderly, low-income and special-needs patients. www.Infonet.welch.jhu.edu/advocacy.html

National Institutes of Health

This government-run portal is a good starting point for finding on-target and reliable health information. Find an A-to-Z listing of health reports and studies. For more specialized questions, click on HEALTH HOTLINES for a list of toll-free telephone numbers www.nih.gov/health

National Library of Medicine

The world's largest medical library, online or off. Be sure to check out Medlineplus, a huge database of research literature. Use it to find good health sites run by a variety of institutions. www.nlm.nih.gov

Physician's Reference Guide

The Physician's Reference has long been the top guide to FDA-approved drugs. There's also information on herbal medicine and a section called the Getting Well Network that offers guidance to detecting, preventing and treating a wide range of ailments. www.pdr.net

Quackwatch

Dr. Stephen Barrett is superman to Lex Luthors of the medical industry. His site aims to help consumers fight health-related quackery. www.quackwatch.com

National Organization of Rare Diseases NORD

The NNPDF is a member of NORD and follows much of their legislative actions. To view current legislative matters refer to: <http://www.rarediseases.org/cgi-bin/nord/new>

Family Directory

Please extend a warm welcome to the Burd family, our newest Niemann-Pick Disease Foundation family member.

- Amanda Burd
2024 E. Oak
Enid, OK 73701
580-977-4608
E-mail: blondie_73701@yahoo.com

Children: Alexis Michele (NP-C) DOB: 11-17-01 Ashlee Ray DOB: 08/09/00
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Directory Changes

Since the printing and mailing of the January 2002-2003 Family Directory, we have been advised of the following changes. Please make a note of this in your family directory. Thank you.

- ◆ Wendy & Roy Futterman
75 Central Park West, Apt #7D
New York, NY 10023
(212)787-9043
New Addition:
Katherine Sara DOB: 2/25/02
(We are also saddened to advise families that the Futterman's precious son, Billy (NPA) died on 3/27/02. Please accept our condolences.)
- ◆ Anthony & Lisa Leoni
Please correct their e-mail address to:
aleoni.a@pacbell.net
- ◆ Woodrow & Theresa Long
Please note a correction to their zip code.
42 Pond Hill Road
Wallingford, CT 06492
203-265-7890
- ◆ Murray & Donna McNab
Please add their new e-mail address:
mcnab_md@shaw.ca
- ◆ Hunt & Annette Ozmer
Please note their new home address:
5971 Saddleridge Road
Roanoke, VA 24018
540-774-0944
- ◆ Roy & Brenda Teller
Please note a small correction to their telephone #:
619-934-8053

Families as advocates

BEING A HEALTH ADVOCATE FOR YOUR CHILD WITH SPECIAL HEALTH CARE NEEDS

Parenting a child with special health care needs is not easy. The 30,000 members of Family Voices, most of us parents of kids with disabilities or chronic conditions, want you to know you're not alone on this amazing journey. We're there, too. And we know that the sooner we parents become knowledgeable and strong advocates for our children, the smoother life becomes for our entire family. Our advocacy tips focus on health issues, because that is the mission of Family Voices. But you can use these ideas at your child's preschool, child care center, classroom, in the community, and with your extended family and neighbors. In a later Family Voices, we'll describe how to advocate for other children by improving public and private health systems serving youngsters with special needs. We begin, however, with one child.

- Believe with all your heart that your child, like all children, is wonderful --- even when she or he gobbles up so much of your time and energy. Tell all the world about this precious gift! Remember, though, that your child with special health care needs is not the **heart** of your family, but a **part** of your family, sage advice from veteran mom, Florene Poyadue.
- As soon as possible, make contact with another family who also has a child with special needs. Hearing from another parent who never sleeps, feels inadequate, and is also frightened about the future will change your life. We know it's not easy to talk with a stranger about your child, but it's worth it. Most communities have a parent organization, usually listed in the phone book, that matches experienced fathers and mothers with new parents. Some groups bring parents together around a certain diagnosis, Cerebral Palsy, for example. Parent groups provide free peer support and are excellent resources for learning all about this new world.
- Find out everything you can about your child's diagnosis from your pediatrician, early intervention program, parent group, local library, state program for children with special health care needs. Surf the Internet, where every imaginable diagnosis has a website full of information and resources. If you lack a computer or don't know how to surf, ask someone to help. Every community now has a location with free internet access. Family Voices has a list of Internet addresses.
- Learn about the hospitals, pediatric specialists, therapy centers, equipment banks, early childhood programs, and state and private agencies that your child will use. Ask other parents for advice about where to get the best care.
- Keep records. Of all phone calls, doctor visits, insurance bills, Medicaid notices, and forms related to your child. Take notes. Request copies of everything. Put this paperwork in one place --- a box in the kitchen, a notebook, a bedroom drawer.
- Become an expert on your child's health insurance plan, whether it's through your employer or Medicaid, especially the benefits paid for by the plan. If you've moved into a managed care system, make sure you ask lots of questions about how it works.
- Insist that all your child's providers, from pediatricians to social workers, practice **family-centered care**, which acknowledges and supports families as they raise their children in their homes and communities. Ask us for family-centered care materials.
- Develop strong partnerships with health and other professionals in your child's life, so that you can work together and use the expertise and skills each of you brings to the care of your youngster. It's especially helpful to have one professional who knows you, your family, and your child very well, and who will advocate with you as a partner.
- Know that YOU are your child's best advocate. No one else will do the job as well. Use all the information, contacts, friends and skills you have gathered to practice your advocacy firmly, but with kindness and humor. As your child grows up, teach her or him to be an advocate, or prepare a sibling or friend to

FAMILIES AS ADVOCATES (continued from previous page)

do so. Because you won't be around forever. In the meantime, it's wise to take care of yourself, even when you think you can't.

- Ask Family Voices for help. Call us tollfree (1-888-835-5669) to find brochures, books, and websites about family-centered care, family-professional partnerships, fathers networks, information in languages other than English, support groups, and training conferences.

NOW, GO FORTH AND ADVOCATE!

The preceding article and the following resource information were taken from the "Family Voices" web site at www.familyvoices.org. NNPDF gratefully acknowledges the resources that "Family Voices" makes available to families.

"Family Voices" is a national grassroots network of families and friends speaking on behalf of children with special health care needs. This eight year old organization listens to families, provides information, and acts as a clearinghouse for information about children. Volunteer Coordinators run projects meant to improve the health and well being of member families. Following is a list of contact information, by state, which may be helpful.

STATE RESOURCES & COORDINATOR CONTACT INFORMATION

ALABAMA:

The Alabama system is fragmented and resources are not always available. Susan Colburn, 1-800-846-3696; 334-613-2284; scolburn@rehab.state.al.us

ALASKA: A pressing problem facing these families is finding respite and individual assistance workers.

Kathy Allely; 907-273-0448; kathya@stonesoupgroup.org

ARIZONA: Arizona families find that referrals to specialists are difficult to get from the managed care systems.

Judie Walker; 602-242-4366; walkjaws@aol.com

ARKANSAS: The limited amount of services and lack of providers in rural areas creates a challenge.

Rodney Farley; 501-682-1461; Refarley@juno.com

CALIFORNIA: The state with the largest Voices membership finds care coordination and access is a pressing problem Juno Duenas; 415-282-7494; sfdmiss@aol.com

COLORADO: A failure to follow health conditions as children grow, results in costly surgeries, lost of function, and dissolution of families. Lorri Park; 303-447-8447; Lparkpris@aol.com

CONNECTICUT: Problems in Connecticut include a lack of care coordination and inconsistent application of the "medical necessity" definition. Molly Cole; 860-679-1504; mcole@nsol.uhc.edu

DISTRICT OF COLUMBIA: This new organization hopes to facilitate family involvement in program and policy development. Gail Johnson; 301-470-0256

FLORIDA: Families in remote areas of Florida encounter inadequate coverage for therapies and home care. Conni Wells; 850-926-3514; cjwells@sprynet.com

GEORGIA: Georgia Voices group has taken advantage of its opportunities to collaborate with other organizations. Cindy Arceneaux; 770-338-2390; carceneaux@aol.com

HAWAII: Since the only tertiary care center is on Oahu, families who do not live there must fly to the island to receive care. Leolinda Parlin; 808-524-8313; leolinda@resqconsultants.com

IDAHO: Families must travel long distances or wait for extended periods to receive care for their children. Ellen Hunt-Landry; 202-344-9187; ellenfcf@micron.net

ILLINOIS: A lack of coordination, collaboration, or cooperative agreements among multiple programs causes children to "fall through the cracks". Faye Manaster Eldar; 773-274-8683; familyvoicesillinois@yahoo.com

INDIANA: Waiting lists for in-home support are very long. Donna Gore Olsen; 317-257-8683; FamVoicesIN@aol.com

IOWA: Parents with private insurance have conflicts with Iowa's system of care. Paula Connolly; 515-223-6714; LDNAIOWA@hotmail.com

KANSAS: Finding adequate coverage for specialty care and obtaining health services at school, are problems in Kansas. Joe Porting; 785-296-8625; jporting@kdhe.state.KS.US

(Continued from page 19)

- Family Voices - State Coordinator Info.

KENTUCKY: Long waiting lists for services and inability to afford insurance are problems in Kentucky. Veronica Brown; 502-479-7465; VWB511@aol.com; PiNK@KYP2P.org

LOUISIANA: Health services in schools, medical necessity and care coordination create problems for Voices families. Phyllis Landry; 504-299-9176; familyla@Bellsouth.net

MAINE: Maine families are challenged to coordinate services, fund home services and to retain home care providers. Beverly Baker; 207-582-2504; bbaker@mpf.org

MARYLAND: Poor access to health providers and services as well as inadequate coverage for therapies exist for Maryland families. Gail Johnson; 410-859-5304; mdfv@ppmd.org

MASSACHUSETTS: A critical problem for families is finding qualified providers and awareness of supports and services. Polly Sherman; 508-947-1231; polly.Sherman@state.ma.us

MICHIGAN: Communications break down between health care providers, which results in lack of care coordination. Ramona Adams; 800-292-7851, ext. 110; radams@voyager.net

MINNESOTA: Mental health service coverage is poor, and accessing public programs is difficult. Carolyn Allshouse; 952-838-9000; callshouse@pacer.org

MISSISSIPPI: Mississippi families have problems with payment and access to health care; also, communication between education and health systems is poor. Aretha Lee; 601-714-5707; Aretha@parentpartners.org

MISSOURI: A parent facilitator provides information and support to parent groups. Sarah Zerr Munday; 816-455-2977; shzkemo@aol.com

MONTANA: Inconsistency in approval for services from the Medicaid system and obtaining services at school, are problems. Tera Randall; 406-232-6321; randallracen@mcn.net

NEBRASKA: Rural areas have trouble accessing health services and non-English speaking families have problems obtaining information in their language. Rene Ferdinand; 402-475-4407

NEW HAMPSHIRE: Lack of home nursing causes many parents to leave their jobs. Martha Jean Madison; Terry Ohlson-Martin; 603-271-4525; 800-852-3345 x 4525; MJM10KIDS@hotmail.com; WWTOM@tds.net

NEW JERSEY: Problems being addressed include adequate insurance coverage and health related services at school. Lauren Agoratus; 609-584-5779; Agoratus@cs.com; Louise McIntosh; 908-277-2883; lkmcintosh@aol.com

NEW MEXICO: State's Medicaid program for children

with special health needs is not working for mental health problems. Patricia Holmes; 505-865-3700; nmfamilyvoices@aol.com

NEW YORK: Providers are not familiar with requirements of special needs children causes problems between medical and educational system. Ruth Walden; 518-474-2001; rxw03@health.state.ny.us

NEVADA: Obtaining information regarding services and obtaining insurance are two problems faced by families. Marcia O'Malley; 800-216-7988 X 3003; momalley@unr.edu

NORTH CAROLINA: There is no "one" place for families to get information for insurance, rights, waivers, or advocacy. Grace Sisco; 704-892-1321; ECACNCFV@aol.com

NORTH DAKOTA: Home and community-based services are not available in rural areas. Donene Feist; 701-493-2333; feist@daktel.com

OHIO: Family Voices Chapter is working to reduce the shortage of home care staff for families. Kathy Bachmann; 614-752-2930; bachmannhq@aol.com

OKLAHOMA: Family advocates serve on many boards and task forces to help provide services for families.

Lyn Thoreson; 405-373-3300; ThoresonL@aol.com

PUERTO RICO: Adequate coverage for pediatric specialty care is lacking. Pedro Cruz; 787-263-7969; pedrocruz@hotmail.com

RHODE ISLAND: Few pediatric providers accept the state Medicaid, and there are few other insurance options. Dawn Wardyga; 800-464-339 X58; 401-727-4144 X58; familyvoices@ripin.org

SOUTH CAROLINA: Rural families have trouble getting to services and communication between systems is poor. Sarah O'Brien; 803-898-0348; obriensa@columb60.dhec.state.sc.us

SOUTH DAKOTA: Transportation to health providers is poor in this rural state. Barb Beck; 605-361-3171

TENNESSEE: Inadequate coverage for therapies and denied initial requests, create a burdensome appeals process. Dara Howe; 615-383-9442; familyvoices@tndisability.org

TEXAS: Inadequate specialty care is a problem. Contact New Mexico office.

UTAH: Long waiting list for support services and fragmentation between medical and educational systems create problems. Gina Pola-Money; 801-272-1051; utahfamilyvoices@juno.com

VERMONT: Families must be the "middle man" due to lack of coordination within the system.

Nancy DiVenere; 802-655-5290; Nancy.Divenere@partopartvt.org

VIRGINIA: Restricted home health care and lim-

(Continued on page 21)

(Continued from page 20)

- Family Voices - State Coordinator Info.

ited access to specialty care create problems for Virginia families. Heather McCabe; 703-960-2361; drmom9@early.com

VIRGIN ISLANDS: Families lack stateside federal programs, funding, and limited culturally sensitive information. Cheryl Miller; 240-776-6179; mangomomma@yahoo.com

WASHINGTON: Families struggle with inconsistency in medical necessity decisions and lack of coordination among services. Judie Ebbert-Rich; 360-866-8254; 360-866-4154; judierich@olywa.net

WEST VIRGINIA: Rural families cannot reach specialists and battles exist between health and education services. Gail Foley; 304-873-1834; gfoley@iolinc.net; Colleen Anderson; 304-293-4692; canderson@hsc.wvu.edu

WISCONSIN: Families are challenged by low reimbursement rates for home care and waiting lists for services. Liz Hecht; 608-263-7148; hecht@waisman.wisc.edu; www.wfv.org

WYOMING: Obtaining affordable private insurance and adequate coverage are problems faced by Wyoming families. Becky Lancaster; 307-857-3962;

RESOURCES FOR FAMILIES

- **The Medicaid Clearinghouse**, a service from Families, USA is now on the Web at: <http://www.handsnet.org/medicaid>. Find out the latest on state and federal Medicaid law, regulations, changes, and Medicaid-related sources.
- **"State in Health"** is a monthly newsletter from the Center for Community Health Action. Good, believable information. Write: Center for Community Health Action, c/o Families USA Foundation, 30 Winter St. Boston MA 02108.
- **Heartfelt Press** has published a baby book for babies with special needs called "Under the Plum Tree" and a memory book for ages one to five called "A Rainbow in My Pocket." Both colorful books come from families and therapists, inspired by babies and children like ours. Phone (913) 272-1155.
- **"Health Issues for Children and Youth with Disabilities" newspaper** provides health information with a focus on our kids and families. The latest issues include some positive articles on managed care and children and youth with special needs. Write IHD, Univ. of Minnesota, Box 721, Minneapolis, MN 55455-0392.
- **"Family Village"** is the electronic place to go to get your questions answered, to search for other families who share your situation to find resources. Aply and creatively run by Linda Rowley, this a friendly place. [Http://www.familyvillage.wisc.edu](http://www.familyvillage.wisc.edu). E-mail: familyvillage@waisman.wisc.edu.

Homecare Resources

- "How to Choose a Home Care Agency; A consumer's Guide" is available for free by sending a self-addressed stamped letter to the National Association for Home Care, 519 C. Street NE, Washington, DC 20002-5809.
- "Managing Your Personal Assistance Worker," written by Consumers in Action for Personal Assistance in 1998, includes information on worker management, communication skills, hiring skills and many other topics. The handbook is free to In-Home Support Services (IHSS) consumers in San Francisco. Other are charged \$6 to offset some of the cost of printing and mailing. Call 415/243-4477 or email info@sfihsppa.org.
- "Caregivers and Personal Assistants; How to Find, Hire and Manage the People Who Help You (or Your Loved One!)" by Alfred H. DeGraff, MA, SEA, from Saratoga Access Publication is \$24.95 and can be ordered online at www.saratoga-publications.com or by calling 970/484-5595.

Paying the Medical Bills

A Briefing Paper from the National Information Center for Children and Youth with Disabilities (NICHCY). The purpose of this briefing paper is to enable the family and/or care providers to make a full search of possible ways to meet medical expenses. Financial needs solutions vary from family to family. Specific plans for resolving difficulties will depend on the family's personal financial situations, public or private family health insurance policy, the presence or absence of individual health insurance policy, the child's medical needs, the state in which the family lives, and family members' understanding of their rights; responsibilities and options. NICHCY "Briefing Papers" are produced in response to requests from parents, professionals and other concerned individuals. Individual copies of these and other NICHCY inquiries. For further information or assistance, or to receive a NICHCY "Publications List", contact: NICHCY, P.O. Box 1492, Washington, D.C. 20013-1492 or call 1 800 695-0285.

A Better Day

- Take one cup of spirit and one cup of hope
- Add a pound of strength, mix well, take a deep breath
And smile.

RESOURCES, REVIEWS & WHATNOT

The Compassionate Friends Has a Home Page On The Internet

• Thanks to Glen Heavilin, who writes, "The TCF page is listed in many search tools now: Yahoo, Web Brower, Infoseek, etc. When you are at the search screen type Compassionate Friends, then click on the search button. One or more listing for TCF will come up in blue text. When you click on the blue words you will link to the page. After you are there add it to your hot-list and you can return directly by choosing TCF from the list. Our WWW address is: <http://pages.prodigy.com/CA/ycq97a/lycq97tcf.html>. To locate a local chapter in your area, just e-mail TCF at wntc86a@prodigy.com. Chapters, which would like to create and maintain a home page, can be linked to the TCF home page. There is no charge for Prodigy home pages at the time. American On Line (AOL) has a forum for bereaved parents, called "You're Not Alone," which meets every Tuesday night, beginning at 8PM, EST. To join in: Go to Keyword, type PIN. That brings you into Parents Exchange. Choose that-and from there choose Parenting Conference Center and Home-front Hall. There is also an active folder in AOL under PIN/Parent/Exchange/Family Life/Death of a Child.

Resource Guide from Exceptional Parent Available through STSAD Office

For the second year in a row, "Exceptional Parent" magazine has provided NTSAD with copies of their annual Resource Guide. You can receive a free copy of this Guide, which lists directories of national organizations, associations, products and services by calling NTSAD.

Do Self-Help books really help you help yourself? Like the story of the three bears, some are too long, some are too short and some are just right. Here are three "just right" booklets that are well organized, easy-to-read and contain helpful hints for family caregivers, professionals, and those suffering from chronic illness.

- "Self-Care Now! 30 Tips to Help You Take Care of Yourself and Minimize Caregiver Burnout"
- "Self-Care Now! 30 Ways to Overcome Obstacles that Prevent You from Taking Care of Yourself"
- "Self-Care Now! 30 Tips to Help You Take Care of Yourself When Chronic Illness Turns Your Life Upside Down"

The author, Pauline Salvucci, MA, is a personal and professional coach, and principal of Self-Care Connection in Cape Elizabeth, Maine. For the last ten years, Pauline worked within a medical family practice, where

she counseled chronically ill individuals and family caregivers. For more information on how to obtain these booklets, contact: SelfCare Connection, 66 Starboard Drive, Cape Elizabeth, ME 04107, phone 207/799-9363, E-mail: Booklets@SelfCareConnection.com, or visit the website at www.SelfCareConnection.com.

Are you kidding?

Doctors will make housecalls?

It's true! The American Academy of Home Care Physicians (AAHCP), as a public service, has included on its website a list of member physicians who make house calls. Patients and family members who are searching for a physician to come to their home can go to the website at www.aahcp.org and click on "House Call Network" to access the database which is arranged by zip code served. Once the appropriate zip code is located, the site visitor has access to the provider's name and contact information. There are over 3,000 zip codes listed on the site. However, yours may not be one of them. Ask your physicians or healthcare provider if they are aware of this service and whether they would become a member.

The AAHCP, established in 1988, is a 501 (C) 3 organization of physicians, other care professionals, and agencies dedicated to promoting and improving the art, science, and practice of medicine in the home. For more information, contact Constance Row, AAHCP, P.O. Box 1037, Edgewood, MD 21040-1037, phone: 41/676-7966, fax 410/676-7980, or e-mail: aahcp@mindspring.com.

Question: Can a workbook take the crisis out of caregiving? Answer: It can help.

Hard questions, Simple Answers: A workbook to Take the Crisis Out of Caregiving, Caregiving Solutions, 118pp., \$24.95, is one of the best I have seen so far. It truly does pose the hard questions and we all know that simple answers are hard to come by. Nobody ever says this caregiving business is easy or simple. However, this workbook systematically outlines the issues and information you will need to take care of any older adult. Its practical, detailed, and easy-to-use approach enables you to record personal information and pertinent dates in a step-by-step manner. The workbook focuses on those life issues specific to older adults. The author, Elaine Petes, MA is a social services professional and private geriatric care manager who has assisted seniors, the frail elderly and their families for over twenty-five years. The workbook was inspired by those caring-but-frustrated family members who needed advice and counsel. For more information or to order the workbook, go to www.caregiving solutions.com or contact by phone: 949/253-4140, fax 949/723-1967, or email: caregiving@solutions@earthlink.net.

What's New - The Genome Action Coalition - TGAC HOTLINE

Legislative Update:

GENETIC NONDISCRIMINATION: IS A DEAL IN SIGHT?

Ever since an amendment to the Health Insurance Portability and Accountability Act of 1996 added the words “genetic information” to the list of reasons that could not be used to discriminate in health insurance, there has been discussion about expanding the scope of the provision. HIPAA applied only to group health plans, but not individual plans. It had relatively weak enforcement provisions. It was not comprehensive genetic nondiscrimination legislation – nor was it ever intended to be.

Now, six years later, there is increasing evidence that Congress may finally be ready to take this issue on. As regular readers of the TGAC Hotline know, Senate Majority Leader Tom Daschle (D-SD) made this a high priority issue when the Democrats took over the Senate last year. However, some questions about scope and practicality of the bill in hearings, followed by the tragic events of September 11th, derailed the legislation for a time.

Now, with the Senate Health, Education, Labor and Pensions Committee poised to act, eight members of the Senate (seven Republicans and Independent Senator Jim Jeffords), led by Senator Olympia Snowe (R-ME) have introduced legislation that, while not identical to the Daschle bill, contains sufficient common ground to suggest that the opportunity for compromise may exist.

In her floor statement introducing the legislation, Senator Snowe said, “...given the advances in the science, there are two separate issues at hand. The first is to restrict discrimination by health insurers and the second is to prevent employment discrimination, based upon genetic information.”

With regard to health insurance, she said, “...we must ensure that we protect genetic information, genetic tests, as well as information regarding a request for genetic testing, from being used by an insurer against the patient.” Then, moving to discrimination in the workplace, Senator Snowe says, “...the issues surrounding protecting genetic information from workplace discrimination are new. And to that end, the legislation I introduce today creates these protections in the workplace. As demonstrated by the Burlington Northern case, the threat of employment discrimination is real and therefore it is essential that we take this information off the table...”

To be sure, there are differences between the Daschle and Snowe bills. However, for the first time in six years, we now have senior members of both parties saying, as Senator Snowe did when she introduced her legislation, “...the need for protections against genetic discrimination by both health insurers and employers is becoming more urgent everyday. If, because of concerns about the way information could be used, people are unwilling to use the potential unlocked by the Genome project to take proactive steps to protect their health and that of their loved ones, then we will never reap the true benefits of the discovery.”



Thinking of Moving?

You are important to us. Please keep in touch....

We don't like losing members who forget to tell us that they are moving, so please remember to let us know your new address. Apart from anything else it prevents us wasting precious resources on printing and postage, so please help us keep the database “Up-to-Date”! Keep us informed of new addresses, telephone numbers, e-mail address and any interesting news about your child. You, our members, are very important to us.



We have received word that the APMRF will be moving to new offices on May 1st. Please make a note of the following:

3530 East Campo Aberito
Suite 105

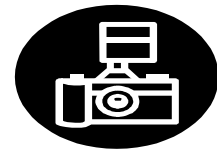
Tucson, AZ 85718

Phone: (520)577-5106

Fax: (520) 577-5212

E-mail: victory@parseghain.org

Web Site: www.parseghian.org



Photographers Needed!

If you are a “shutterbug” or have an eye for taking candid photographs, we welcome your talents to capture those special moments at the family conference in July in New Orleans.

These photographs would then be used in an upcoming edition of the NNPFD Family Support Packet. Please contact the foundation office if you are interested.

Thank you.

Research Update – Marie Zahner, NNPDF Director of Research

- It has been five years since the NPC1 gene was cloned at NIH in early 1997 and a little over a year since the fortuitous identification of NPC2. No one expected that determining the function of NPC1 would prove as difficult as it has. However progress is being made and there are at least two theories about its function. One is that it is a pumping mechanism and the other is that it is a shuttle mechanism. In either case, finding a treatment will be a challenge – a challenge that we must meet. To this end we continue to fund the best of the research grant applications that the NNPDF receives twice a year. Of the six applications that we received for the November deadline, we have funded two and asked several others to resubmit. We expect more applications in May, 2002.

*****NEW RESEARCH*****

- **Anita H. Corbett, Ph.D.** of the Emory University School of Medicine will be funded as of August, 2002 to study the “Functional Analysis of Niemann-Pick C1 Protein”. Dr. Corbett believes that her experiments will open a new field for studying the function of both the NPC1 and NPC2/He1 genes. By understanding what these genes do in normal cells researchers can understand what goes wrong in NP-C patient cells. Her proposal will test whether a very simple yeast based system can be a useful tool to answer some of the most important questions about the cause of the disease, which could lead to accurate diagnosis and effective treatment.
- **Melanie Jean Dobson, Ph.D.** of Dalhousie University has been funded to study “A Yeast System for Analysis of Sterol Trafficking Defects in NPC.” This is another yeast scientist and she will be looking at pathways of sterol synthesis and regulation that have been conserved from yeast to man. Yeast contains a single gene with remarkable similarity to the human NPC1 gene. Dr. Dobson’s approach to determining the function of human NPC1 is to analyze the yeast version of this gene.
- **Dr. Lee-Way Jin** “Abnormal Intracellular Accumulation of amyloid B (AB) protein in the NPC Disease Brain” Dr. Jin is part of a strong team of scientists who have been actively involved in studying Neurodegeneration in Alzheimer’s Disease. Dr. Jin suspects that there may be a common mechanism of neuronal death shared by NPC and Alzheimer’s Disease. By drawing on the wealth of research and drug discoveries of Alzheimer’s Disease we can greatly accelerate the drug discovery to alleviate neuronal death in NPC. I will keep you posted the status of this grant application.

*****CONTINUING RESEARCH*****

- **Dr. Daniel Ory** was funded in August, 2001 to study “The Role of HE1 in Cholesterol Trafficking.” He reports that from his studies a new model is emerging for participation of both NPC1 and NPC2 in the processing of cellular cholesterol. He believes his research is helping us understand how defects in cholesterol metabolism lead to disease in individuals with NPC. Dr. Ory also believes from his research that NPC disease may be a candidate for application of stem cell technology.
- **Dr. Robert Maue** was funded in August, 2000 to investigate the effects of growth factors on a population of neurons particularly devastated in NP-C disease. He has also been analyzing the expression of membrane proteins, including growth factor receptors, in cholesterol-enriched membrane domains. Dr. Maue’s work related to gene therapy is cautiously optimistic. Mice injected with adenovirus to carry the good copy of the NPC1 gene still appear to be flourishing after seven to eight weeks. He will continue to monitor the motor skills and coordination of the injected mice, as well as their growth and longevity.
- **Dr. Futerman** was funded in February, 2001 to study “Neuronal Development and Death in Type A Niemann-Pick Disease.” Over the past year, Dr. Futerman has been trying to determine the mechanism by which nerve cells die in a mouse model of Niemann Pick A disease. Nerve cells are taken from the brain of these mice, and then cultured in a petri dish, and challenged with various agents that may or may not kill the cells. He has found that different agents have different effects. One surprising result is that a key molecule involved in nerve cell growth, the nerve growth factor, kills these neurons equally well in both control and NPD-A neurons, whereas another agent, glutamate, does not kill NPD-A neurons. Moreover, he has seen changes in the growth and development patterns of these neurons. Together, they are beginning to understand what regulates neuronal growth, development and death in NPD-A nerve cells, which will hopefully allow them to identify new therapeutic targets.

As you can see our foundations is funding an ever greater number of scientists and we are helping to make strides in the various diseases that have the common name Niemann-Pick. It may be a slow arduous process but we work diligently and with great resolve to do the best for our children. Our success depends on the dedication of all the families and on the generosity of our contributors. As always, a special thank you to all.

Respectfully Submitted: Marie Zahner - Director of Research

Parent - to - Parent

Coenzyme Q 10 (CoQ10)

There has been much discussion recently about **Coenzyme Q10 (CoQ10)** on the NNPDF Website's mailing list. Several NPC families have reported positive results using this coenzyme.

What is CoQ10?

CoQ10 or Ubiquinone is an antioxidant best known for its protective effect on the heart. CoQ10 is produced naturally in the body (its alternate name, ubiquinone, reflects the ubiquity with which it is found throughout the body), but its levels decline significantly with age, and many medications (including cholesterol lowering drugs) further deplete it. CoQ10 plays a crucial role in the production of energy in the mitochondria, tiny "energy furnaces" found in almost every cell in the body that create energy from the combustion of nutrients and oxygen. CoQ10 is beginning to be noticed as a nutrient that combats age-related neurological decline, both by maintaining optimal energy production in the brain and by protecting it from oxidative damage generated by free radicals – something to which the fat and oxygen-rich environment of the brain is especially vulnerable. Because of these properties, it may well play a role in preventing such neurodegenerative diseases as Alzheimer's and Parkinson's disease and has sparked the interest of NPC Researchers.

Dr. Patterson has reviewed literature and recent studies on CoQ10. He feels, before we can go any further with clinical and related laboratory studies on this dietary supple-

ment, we need to demonstrate that a secondary deficiency of CoQ10 in NP-C exists. Dr. Patterson is recommending a preliminary analysis be done to check for a CoQ10 deficiency in NPC individuals. In order to do this **Dr. Patterson is requesting blood samples from NPC individuals be sent for analysis.**

Your doctor can take the blood sample and send it to the laboratory of Dr Salvatore DiMauro. The details for your doctor are:

- Not currently on CoQ10 (if you are thinking of using CoQ10, please obtain a blood sample first)
- Not currently taking a statin, such as Lovastatin or Mevacor
- 10 mls of blood (5 mls in a child) should be drawn into a plain tube and allowed to clot; the serum so obtained should be labeled with:

Patient's name

Date of birth

"For Coenzyme Q assay; Niemann-Pick Type C patient."

- The labeled specimen should be sent to arrive on a weekday.

The mailing address is:

Salvatore DiMauro, MD

4-420 College of Physicians & Surgeons

630 West 168th Street

New York, NY 10032

Dr. DiMauro is providing the preliminary analysis at no charge. Since this is a research study (not patient diagnosis), results cannot be processed immediately. We will report the results on the NNPDF Website on Newline.

We ask you to please consult with your doctor before beginning or ending any treatment, including dietary supplements

Information on OGS Clinical Trial in Niemann-Pick Type-C Disease

Information on OGS Clinical Trial in Niemann-Pick Type C Disease

On 7 Mar 02, Oxford GlycoSciences (OGS) announced that they will be conducting a clinical trial of their glucosylceramide synthase inhibitor OGT 918 in Niemann-Pick Type C disease (NPC). The study will be conducted at 2 clinics specialising in the care of patients with NPC at Royal Manchester Children's Hospital in the UK and Columbia University in the US. Fifteen patients will be enrolled at each centre. The study will assess the safety and effectiveness of OGT 918 for the treatment of NPC. In order to do this, the study employs a no treatment control arm, whereby one-third of patients enrolled i.e. 10 patients (or 5 from each study centre) will NOT receive the trial therapy for one year. The study has been initiated and patient screening has commenced at Manches-

2002 R&D UPDATE *PR Newswire - March 7, 2002*
OGS Building a Sustainable Pipeline: New Programmes and New Clinical Trials OXFORD, UK

Today Oxford GlycoSciences Plc (LSE: OGS, Nasdaq: OGS1) will present progress on its drug discovery and development pipeline and describe its novel R&D strategy. The Company will announce a number of advances in its pipeline at an R&D update, including: the expansion of OGT 918 trials into potential new indications; commencement of a development programme based on a novel molecule, OGT 923, for lysosomal storage diseases; pre-clinical efficacy data on OGT 2492, the lead candidate in a wholly-owned small molecule cancer programme; and progress in the OGS-Medarex collaboration.

Dr Raj Parekh, OGS' Chief Scientific Officer said: "Over the last 18 months, OGS has implemented an innovative process for the rapid selection of druggable protein targets and the discovery of drug leads. This process, built on the quality and number of targets

2002 Research & Development Update - Oxford, UK - Continued from page 25

emerging from our proteomics platform and close access to leading drug discovery technologies from Medarex and NeoGenesis, is generating a large number of drug programmes. These will fuel our pipeline growth this year and beyond in our current focus areas of glycolipid storage diseases, cancer and fungal infection using small molecules and therapeutic antibody approaches."

Drug Candidates:

Cancer:

- Data will be presented on OGT 2492, OGS' first small molecule inhibitor of heparanase I, an enzyme involved in the growth and spread of many cancers, including breast cancer. The compound, which is orally active, shows good selectivity, pharmacokinetic and efficacy profiles in pre-clinical studies.
- The first candidate emerging from the OGS-Medarex collaboration is a fully human antibody that binds to and neutralises the heparanase I enzyme. This antibody, which is in the Medarex T12 programme, is part of the comprehensive OGS-Medarex-Genmab cancer campaign announced earlier this year.

Lysosomal Storage Diseases:

- The Company's lead compound Vevesca (OGT 918) is under regulatory review in Europe and the United States for Type I Gaucher disease. Publication of the enzyme replacement switch/combination data is progressing and is currently under peer review. The Company will today announce that it is initiating new clinical studies to investigate the safety and efficacy of OGT 918 in Niemann-Pick Type C disease, Neuronopathic Gaucher disease and Late Onset GM2 Gangliosidosis.
- The Company will also announce a new programme based on OGT 923, a new chemical entity which has shown efficacy in pre-clinical models of Sandhoff and Niemann Pick Type C disease. OGT 923 is now in development at OGS with a target date of first dose to man (Phase I) in Q4 2002.

Dr Chris Moyses, OGS' Chief Medical Officer said: "Evaluating potential new indications for OGT 918 and commencing a second development programme underscores our commitment to the therapeutic area. We continue to lead the evaluation of novel oral agents for substrate reduction therapy for patients with Gaucher disease and related glycolipid storage disorders."

Lead Discovery:

To expand the drug candidate pipeline in 2003 and beyond, OGS will describe how its drug discovery strategy is being used successfully to build its pipeline. To date the Company has:

- Selected 27 cancer targets involved in various disease mechanisms that have entered the NeoGenesis collaboration;
- Identified breast and renal cancer antigens which are

being investigated as monoclonal antibody targets as part of the Medarex/Genmab joint venture;

- Identified multiple targets involved in the growth of several pathogenic fungi, which have entered the NeoGenesis collaboration;
- Received high affinity ligands against targets from NeoGenesis, which are the basis of ongoing lead optimisation programmes.

Michael Kranda, OGS' Chief Executive Officer, said "We have aggressive goals for building our pipeline, namely multiple first dose to man programmes a year. These objectives are realistic considering our innovative and productive discovery and development strategy and I believe will create substantial shareholder value."

The R&D update will take place in London on the 7th of March. Please contact Mo Noonan at Financial Dynamics on +44 (0) 20 7242 8695 for details. A corresponding US session will be organised in New York on the 8th of March. Please contact Hemal Parikh at Feinstein Kean Healthcare on +1-617 761 6787 for details.

Notes to Editors

OGS has developed a patented technology platform in the emerging field of proteomics, the comprehensive study of proteins, integrating proteomics with genomics to create an innovative drug discovery platform. OGS' proteomics collaborations with major pharmaceutical and biotechnology companies include Bayer, Pioneer Hi-Bred/DuPont, Medarex/Genmab, GlaxoSmithKline, NeoGenesis and Pfizer. OGS has technology development collaborations with Applera, Cambridge Antibody Technology, Packard BioScience and the Institute for Systems Biology. OGS has also entered into a joint venture, Confirmant Limited, to develop the Protein Atlas of the Human Genome™.

OGS has drug research discovery programmes in central nervous system, cancer, infectious disease and glycosphingolipid (GSL) storage disorders. OGS has had submissions to regulatory authorities accepted for review in both Europe and the US for its development compound, Vevesca (OGT 918), for the treatment of type 1 Gaucher disease. Vevesca (OGT 918) is an investigational drug and has not received approval for marketing in any country.

This release contains forward-looking statements, such as the commercial potential and success of OGS' collaborations and drug candidates. Factors that could cause actual results to vary significantly from those expressed or implied by these and other forward-looking statements include the success of OGS' research and development strategies, the validity of its technologies and intellectual property position and strategies, the medical conclusions on which Vevesca (OGT 918) is based and uncertainties related to the regulatory process. For further information please contact:

Oxford GlycoSciences Plc
 Michael Kranda, Chief Exec. Officer
 Dr Chris Moyses, Chief Medical Officer
 Dr Raj Parekh, Chief Scientific Officer
 Dr Stephen Parker, Chief Fincl Officer
 Tel: +44 (0) 1235 208000

NPDG (UK) News Updates

From Susan Green, NPDG (UK) received on February 21, 2002

A Clinical Trial Nurse has been appointed for the OGT-918 trial and the first patient is scheduled for initial tests on Monday, February 25th. The trial is underway in the United Kingdom!

- The initial phase will last 12 months with a second 12 month extension possible. The study aims are to assess safety, tolerability, and efficacy (effectiveness).
- A maximum of 30 patients will participate in the U.K. and the U.S., with approximately 15 being tested in each country.
- Patients must be over 12 years of age.
- The patient must be able to swallow and participate in clinical assessments.
- During the initial phase, approximately one third of the participants will not receive OGT-918. This control group will be chosen at random and will undertake clinical assessments only. During any extension, all participants will receive OGT-918.
- Up to seven clinic visits will be required during the initial phase with up to five additional visits during the extension. Some or all visits may require two days to complete the required tests. All visits will be done on an outpatient basis.
- Assessment tests include blood samples, full body MRI, nerve conduction velocity, eye movement, hand movement, nutritional review, and a general physical exam.
- Oxford GlycoSciences will pay reasonable costs related to the clinical trial, including travel, accommodations, and meals.

From Susan Green (NPDG (UK) received on January 21, 2002

The United Kingdom has approved the OGT-918 clinical trial for children 12 and older. Patient Information Letters have been mailed to all known NPC families in the U.K. The letter describes the inclusion criteria and the procedure for applying for the trial. A research nurse has been appointed and the trial is scheduled to begin February 4th. If you do not receive the OGS letter in the next week (by January 24th), please notify the Niemann-Pick Disease Group (UK). The United States is still reviewing the application for the trial. We will update this page as soon as possible with news about the U.S. trial.

NP-A & NP-B Research *By Robert Desnick PhD MD*

Progress towards the Treatment of Type B Niemann-Pick Disease Since the cloning of the acid sphingomyelinase gene in 1992, research on Types A and B Niemann-Pick disease (NPD) has progressed at a very rapid pace. DNA alterations (mutations) in the gene have been identified, a mouse model of the human disease has been constructed and characterized, and efforts to develop effective therapy are rapidly advancing. Highlights of these accomplishments were presented at the recent Millennium meeting by Dr. Robert J. Desnick, Professor and Chairman of the Department of Human Genetics, Mount Sinai School of Medicine, New York. They will be briefly reviewed below.

Background:

Types A and B NPD are both caused by abnormalities in the gene which makes the lysosomal enzyme, acid sphingomyelinase. This enzyme is ordinarily found in special compartments within cells called lysosomes and is required to metabolize a lipid, called sphingomyelin. If the enzyme is absent or not functioning properly, this lipid cannot be metabolized and accumulates in the cell, eventually causing the malfunction of major organ systems. Although Types A and B NPD are both caused by the same enzyme deficiency, the clinical manifestations are

these patients are very different. Type A NPD is a severe neurologic disorder which generally leads to death before 5 years of age. In contrast, patients with Type B NPD generally have little or no neurological involvement and may survive into late childhood or adulthood. The underlying reason for this dramatic difference in the two forms of the disease is not fully understood, and at present it is not possible to accurately predict the severity of the disease by enzyme testing.

Both Types A and B NPD can be diagnosed by measuring the enzyme's activity in white blood cells or cultured cells from a small skin biopsy, although this enzyme test is not very reliable for detecting carriers of the disease. In contrast, gene analysis provides accurate carrier detection as well as prenatal diagnosis for both Types A & B NPD. This is particularly useful in the Ashkenazi Jewish community where three abnormalities in the gene account for greater than 95% of the Type A NPD patients. In Type B NPD there is one common mutation that is present in about 30% of patients; the other mutations are family specific.

(Continued on page 33)

Tracy,
Please place the two page article from the Winter
2001/2002 NTSAD magazine.

Page 6 & 7—Science & Medicine (Focus on NPD)

Thanks—Nadine

Tracy,
Please place the second page of the Focus on N-P article here.

:)

Memory loss in NP-C

Loss of memory can have consequences on daily life in many ways, leading to communication problems, safety hazards and behavioural problems. In order to understand how memory is affected by NP-C, it is useful to consider the different kinds of memory.

Episodic Memory

This is the memory people have of events in their life ranging from the most mundane to the most personally significant. Within episodic memory, there are memories classed as short term (having happened in the last hour) and those classed as long term (having occurred more than an hour ago). Young people with Niemann-Pick Disease often do not seem to have any difficulty remembering distant events but may, for example, forget having done something five minutes ago. Memories of distant events although not greatly affected tend to interfere with present activities. This can sometimes result in the person acting out routines from the past, which are no longer relevant.

Semantic Memory

This category covers the memory of what words mean e.g. a flower or a dog. Unlike episodic memory, it is not personal, but rather common to all those who speak the same language. It is the shared understanding of what a word means, which enables people to having meaningful conversations.

As episodic and semantic memory are not located in the same place in the brain, one may be affected and the other not.

Procedural Memory

This is the memory of how to carry out actions both physically and mentally, for example, how to use a knife and fork or tie shoelaces. The loss of procedural memory can result in difficulties carrying out routine activities such as dressing and washing. This includes things which have become automatic. For this reason, some children who have difficulty finding their words can still sing fairly well. Their procedural memory is still intact whereas their semantic memory (the meaning of words) has been damaged.

Apraxia/Aphasia/Agnosia

Apraxia is the term used to describe the inability to carry out voluntary and purposeful movements despite the fact that muscular power; sensibility and co-ordination are intact. In everyday terms this might include the inability to tie shoelaces, turn a tap on, fasten buttons or switch on the TV.

Aphasia is the term used to describe a difficulty or loss of the ability to speak or understand spoken, written or sign language as a result of damage to the corresponding nervous centre. This can become apparent in a number of ways. It might involve substituting a word which is linked by meaning (e.g. time instead of clock), using the wrong word but one which sounds similar (e.g. boat instead of coat) or use a completely different word with no apparent link. When accompanied by *echolalia* (the involuntary repetition of words or phrases spoken by another person) and the constant repetition of a word or phrase, the result can be a form of speech which is difficult for others to understand or a kind of jargon.

Agnosia is the term used to describe the loss of the ability to recognise what objects are and what they are used for. For example, a person with agnosia might attempt to use a fork instead of a spoon, a shoe instead of a cup or a knife instead of a pencil etc. With regard to people, this might involve failing to recognise who people are, not due to memory loss but rather as a result of the brain not working out the identity of a person on the basis of the information supplied by the eyes.

Communication

Children and Young people with NP-C may have difficulties both in the production and comprehension of language, which in turn lead to other problems. Most children also lose the ability to read and the ability to interpret signs.

Behaviour

A common symptom of NP-C is wandering, both during the day and at night. There are a number of possible reasons for this wandering but due to communication problems, it is often impossible to find out what they are. Other symp-

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toms affecting behaviour include incontinence, aggressive behaviour and disorientation in time and space

Physical changes

Weight loss can occur as a result of the person 'forgetting' to how to chew or how to swallow. Because of damage to the brain the muscles involved in chewing, swallowing and speech do not always receive the correct message to enable them to function effectively.

Progressive difficulties - dementia

There may be *moderate memory problems*, such as forgetting names, school books etc. but, due to the mild nature of these problems, they might not be immediately noticeable and it might be assumed that the child is simply not paying attention. A young adult may be concerned and may try to prevent others noticing the problem due to embarrassment or worry about what friends may think.

Similarly members of the family or friends might try to play down the importance of the problem and make excuses for them. However, the memory problems experienced by young adults with NP-C are less likely to go unnoticed if they are in employment.

The problem may be further aggravated by accompanying *difficulties with attention*. A combination of these two problems may lead to difficulties accomplishing tasks, in school or at home, involving several sub-stages and even in following the thread of a conversation. Another consequence is that the young person will find him/herself searching for words. Although this does not generally affect the person's interest in communicating, s/he might tend to use simpler words and shorter sentences.

Orientation in time can be affected and children can tend to show signs of *disorientation in space*. Wandering and even getting lost in a familiar environment such as the home e.g. not knowing where to find the toilet, or in school e.g. turning up at the wrong class.

Some children develop *strange habits* (for example not wanting to change their clothes or insisting on wearing a certain item of clothing) and some have preference for particular colours. They may not want to tidy away their clothes or toys (if they can't see them then they might not remember where they are). Others may show a *lack of spontaneity* and activity and develop a tendency to be complacent.

There may be *problems with abstract representation*. Money, for example, loses its symbolic form and it might be difficult to manage spending money. Associating geometric forms with real objects might be difficult; i.e. they would not be able to reproduce a cube as it is too abstract. Puzzles and games may prove too difficult. This stage can often be extremely stressful for the child or young person as s/he often has an awareness of what is happening.

As memory and concentration become more difficult they will become more dependent on others. It becomes more *difficult to interpret stimuli* such as taste, sight and hearing and they may be *hypersensitive to touch*. This has repercussions on daily life in the form of loss of appetite, the inability to read and sometimes, *visual hallucinations* may occur. *Insomnia* may become a problem as the distinction between day and night loses its significance. They might tend to sleep more during the day, but less at night. The notion of time and space is affected. Activities such as washing and dressing can be impossible to accomplish alone due to memory loss, confusion and difficulty in manipulating objects.

Movements become uncoordinated. Children become less stable on their feet and may have falls. *Incontinence* may occur either as a result of failing memory, communication problems and practical difficulties or because the signals for needing to go to the toilet are no longer registered or recognised.

Language problems become more marked, including the inability to fully understand the spoken and written word, as well as difficulty speaking and writing. It is not unusual for children to constantly repeat the same words or sentence (*echolalia*).

It is important to remember those children & young people with NP-C do not all have the same symptoms in the same order and with the same degree of severity. However knowing something of the general pattern can help parents to know what to expect and to recognise symptoms as they arise.

Tracy,
Please place the article titled “Aquatic Therapy” from the SNAP
Report—Summer 2001.

Thanks.

In Loving Memory Of
Kathryn Grace Taft

**Who would have been two years old on March 27th,
2002**

Rainbows appear only on dreary, rainy days. They beautify the world for a few brief moments. These moments, however, can be spectacular. You were our brief rainbow. You entered our lives and stayed for but a short while. Nonetheless, the memories of those moments when you blessed us with laughter and delight, joy and smiles, charm and beauty, mischief and silliness, giggles and love ... made the deluge of helplessness bearable. Rainbows, however brief, make the world a brighter, lovelier place. How grateful we are that we had you. Our brief rainbow. Miss you Katiebug.

Love Daddy and Mommy and All who love you. (Baby Kate passed away on Feb. 17, 2002.)

Tracy,
Please copy and place the photo of Baby Kates memorial card here. Thanks—Nadine

Love Lives

Love is this
That you lived amongst us these few years
And taught us love

Love is this
That you died amongst us and helped us
To the source of life

With all our love
We wish you bon voyage
Love Lives

Lindy Hemmy

It is with intense sadness that I must announce the loss of Billy Futterman on Wed March 27, 2002. He was 2 yrs old and had NP type A. He was our beloved middle child and we all miss him very much. We are beyond sad. Thank you to this group for being there. It helps to know others out there know how we feel.

Wendy and Roy Futterman

*Note: A story about Billy and his struggle with NPD appears on page 12 of this publication.
Our hearts go out to Billy's family.*

NP-A & NP-B Research Updates

(Continued from page 27)

Preclinical Studies of Enzyme Replacement Therapy for Type B NPD:

The construction of a mouse model of NPD in 1995 dramatically enhanced our efforts to develop therapy for these diseases. These mice have many similar clinical features of Types A and B patients, and have been used to evaluate treatment for both forms of this disorder. Enzyme replacement therapy refers to the infusion of normal enzyme into patients with Type B NPD. The first prerequisite for such therapy is that we much produce large amounts of the normal enzyme. This was accomplished in our laboratory several years ago using DNA technology. When we there infused this enzyme into the bloodstream of the NPD mice, the enzyme reduced the abnormally elevated sphingomyelin levels in the liver, spleen and lungs to near normal levels. These results indicated that this therapeutic approach is likely to be effective in patients with Type B NPD.

Based on these exciting findings, we undertook collaboration with Genzyme Corporation to develop enzyme replacement therapy for Type B NPD patients. We hope to begin Phase ½ clinical trials in the United States in the next one to two years.

In summary, research on Types A and B NPD is progressing at a very rapid pace. Current efforts are focused on therapy for Type B NPD. We are very interested in studying Types A and B NPD patients from around the world, and encourage individuals to contact us for mutations analysis. All that is usually required is that a small amount of blood be shipped to us. We also have clinic staffed with physicians specially trained in the diagnosis and management of Types A and B NPD, and offer the opportunity for patients to be evaluated by our expert staff.

The preceding article was reprinted with permission from the Niemann-Pick News Sheet published by the NPDG (United Kingdom). Thank you.

Tracy,
Please place a copy of the foundations membership application here.

Thanks—Nadine
Follow this up with the copies of the newspaper articles.

Thanks :)