

neuro·fibroma·tosis®

THE NATIONAL NEUROFIBROMATOSIS FOUNDATION, INC.

SUMMER 2000

Volume 21, No.3

NNFF Day at the Lab® Program Goes National

Neurofibromatosis is not a regular Saturday morning topic for most teenagers. However, for teens with NF, discoveries and breakthroughs on this topic are important for them to learn about so they can better understand the disorder. That is why the NNFF, in an effort to educate young people about the condition, hosted three Day at the Lab®

Teens with NF Become Scientists for a Day

Finding a Gene on a Chromosome Map—Milwaukee

On March 25, 2000 teens from all over the state met at the Medical College of Wisconsin in Milwaukee to take part in the program. Dr. Mark Lubinsky answered questions about NF and took the teens on a tour of a laboratory where genetics research is being conducted.



Making friends and learning at the same time in Dr. MacCollin's lab in Boston.

programs this past spring in Milwaukee, Boston and Los Angeles.

The Day at the Lab Program began as a part of the NNFF International Summer Camp for Teens. Campers spent a day at the University of Utah Eccles Genetics Center, learning about genetics, taking tours of working laboratories and performing hands-on experiments that help them to learn more about NF.

The purpose of the Day at the Lab program is to help make research more real and accessible to the children. We want to make them feel more empowered and in control of their condition through education. This program is so popular among the teenagers at the camp that the Foundation decided to expand into other cities so children all over the country will have a chance to experience it.

For many of the teens attending these events, it was the first time they had met other children with NF. The children were able to learn about NF in a safe, fun environment and make new friends in the process.

After a brief genetics lesson, the children were ready to start doing genetics experiments of their own. One of the most popular experiments was finding the "mutated" gene on a chromosome map. Using pipe cleaners of different shapes, sizes and colors, they plotted the chromosome pairs out on a "map" and used the knowledge they had gained that

day to see which chromosome carried a mutation. This gave them the opportunity to learn about different types of genetic mutations and how these mutations manifest themselves in the body.

Shy at first, the teens gradually felt more comfortable asking questions as the day went on and by the end of the program they were eagerly jumping in with questions and sharing stories of their own experiences in living with NF.

Making Ice Cream from Liquid Nitrogen—Boston

Dr. Mia MacCollin and her staff at the Massachusetts General Hospital Cancer Center opened their laboratory to teens from all over New England on April 1, 2000. The children attending the program not only learned about NF but also got to perform experiments in an actual working laboratory where research on NF2 is taking place.

The teens truly felt like "scientists for a day" when they were given lab coats and official Mass General photo IDs to wear in

(continued on page 5)



NNFF 2000 Research Awards

2000 Research Awards Announced

The National Neurofibromatosis Foundation has announced the recipients of its research awards for the academic year 2000/01/02. The awards are as follows:

Adam Goodkind Family Fund

Dr. Sushmita Maitra, Duke University
Two-Year Young Investigator Award

"Role of Merlin in Proliferation and Cellular Differentiation in Drosophila"

With her experiments in the fruit fly/drosophila model, Dr. Maitra will work on the question: why do specific (Schwann) cells over proliferate and cause tumor formation when the NF2 gene mutates? Her experiments are also designed to further elaborate on the normal function of the NF2 protein Merlin in cells. She will study especially the interaction of the Merlin protein with another tumor suppressor gene to determine how this interaction causes defects in cell growth cell differentiation. Finally, Dr. Maitra will investigate whether NF2 tumors occur because Merlin interacts with still other genes.

The Marcy and Richard Horvitz Family Fund

Dr. Dominique Lallemond, Harvard Medical School/MGH Cancer Center
Two-Year Young Investigator Award

"Investigation of the role of NF2 Gene Product, Merlin, in Cell Adhesion, in Vitro and in Vivo"

Dr. Lallemond proposes to study two things, namely what role the NF2 protein, Merlin, normally plays in cell movements and cell adhesions, and secondly why the NF2 gene in its mutated form causes abnormalities in cell movement and cell mutations. These studies are of considerable importance to all NF2 research, but they also hold great potential for studies in cancer that seek to determine how cancer cells spread in the body and give rise to metastasis. Dr. Lallemond will carry out her experiments both in vitro (the petrie

(continued on page 7)

Former NINDS Director Co-Chairs Foundation's Research Board

Dr. Zach Hall has been named the Co-Chairman of the NNFF Research Advisory Board (RAB). He will serve in this capacity with Dr. Tyler Jacks, of the MIT Cancer Research Center.



Dr. Zach Hall, Vice Chancellor of the University of California/ San Francisco for Research

Dr. Hall is the immediate past Director of the NIH's neurological institute, NINDS. He has a Ph.D. in biochemistry from the Harvard Medical School, and now serves as the Vice Chancellor of the University of California/San Francisco for Research.

"Dr. Hall's record as a top flight scientist and as a leader of the nation's pre-eminent neurological institution, adds new and substantial strength to the Foundation's

research program and especially to our Research Advisory Board peer review panel," said NNFF Medical Director Dr. Allan Rubenstein. "With Dr. Hall's leadership we will move into exciting new directions which we expect to bring us closer to rational treatments and hopefully cures for both NF1 and NF2."

The RAB acts as the peer review panel for the rigorous annual review of grant applications. The board also provides overall direction for the Foundation's other research activities. (For a complete list of grants awarded at this year's meeting see page 1.)

NF

Research Provides New Insights Into the Link Between NF and Cancer

(Editor's note: This article is based on an interview with Dr. Luis Parada, Director of the University of Texas Southwestern Medical Center's Center for Developmental Biology. The research described in this article was made possible in part by a grant from the NNFF. Dr. Parada serves on the Foundation's Research Advisory Board.)

The Use of Mouse Tumor Models in NF Research

A recent issue of *Science* reported on groundbreaking research that is providing new insight into how and why patients with NF1 sometimes develop malignant tumors. Although a rare complication of NF1, malignancy is an important focus of current NF research.

A research team at the University of Texas Southwestern Medical Center, led by Dr. Luis F. Parada, has developed a mouse tumor model that is helping scientists learn how various genes interact to activate malignant tumor development. This mouse model provides scientists with a powerful way to develop and test strategies to inhibit tumor growth in patients with neurofibromatosis.

NNFF: How Did You Get Started in NF Research?

Dr. Parada: I was peripherally interested in NF because I was working on biology of the neural crest and on the sensory nervous system. When the NF gene was cloned, we realized that this gene was a key regulator for the development of neural crest derived tissues. This was exciting because this tumor suppressor gene (NF) provided promise not only for a better

understanding and treatment of the disease but also for learning more about how the nervous system is put together.

For the five years leading up to the discovery of the NF1 gene, my lab was working on generating mouse models to study gene function and human disease. Once the news about the NF1 gene was out, together with colleagues at NCI, we cloned the mouse gene for NF1 and undertook to make a mouse model of NF1.

"Mouse model research has far overreached our expectations..."

This research approach has far overreached our expectations. It has demonstrated that the NF gene when functioning normally is essential to the formation of the nervous system. I became more interested in NF the disease and began developing mouse models of NF1 in order to directly study the consequences of NF1 in patients.

NNFF: What is a mouse tumor model?

Dr. Parada: Since mice are 90-95% genetically similar to humans, they are useful for studying genetic disorders to which we can ascribe a defect in a specific gene, as is the case for NF1 and NF2. Mice get diabetes, they get hypertension and they get cancer. All of the things that can go wrong in a human being can go wrong in a mouse. In mice, we can try different approaches and gene manipulations that are not possible with human patients for ethical and safety reasons. Since mice are small and their lifespan is shorter than humans, everything happens more quickly in terms of disease manifestation so we can gather greater amounts of data. The mice used in our

(continued on page 3)

RESEARCH CALLS

Study of Mutations in Dermal Neurofibromas

The Medical Genetics Department at the University of British Columbia is studying the origins of mutations in dermal neurofibromas, to understand why people with NF1 often develop multiple neurofibromas. If you are a woman planning to have cutaneous neurofibromas removed and would like to help, please contact Patricia Birch at U.B.C. (604-822-2749 or birch@interchg.ubc.ca). By contacting her in advance of your surgery, she can help arrange for a tissue sample to be sent.

NF Graduate Student is Seeking NF Parents

Ann Dylis is a Registered Nurse who is studying for her doctoral degree at Boston College. She was diagnosed with NF as an adult and has a strong desire to give back to the NF community. Her dissertation will be on the parents of NF children. So far, the parents of NF children have only been studied in relation to validating their child's behaviors. This study will be about YOU. You can be a parent with or without NF, and your children can be any age.

If you are an NF parent and are interested in being involved, please contact Ann Dylis directly. Thanks for thinking about participating in this original and significant project! Contact Ann Dylis, Boston College, Cushing Hall, 140 Commonwealth Avenue, Chestnut Hill, MA 02467.

Tel: 617-552-1406;

Fax: 617-479-1282; email dylis@bc.edu.

(Reprinted with permission from the Texas NF Foundation January 2000 newsletter Volume VII)

NF2 Patients Needed for Natural History Studies

The natural history study will measure the growth rates of vestibular schwannomas in 100 people with NF2, in order to determine the normal growth rates for vestibular schwannomas. The study will focus on patients who have been diagnosed with NF2 on or after October 1, 1993. The goal of the natural history studies in NF2 is twofold:

- Develop predictive factors to determine growth rates for vestibular schwannomas in NF2
- Define all parameters necessary to conduct clinical trials for new therapies for NF2

The three-year study will be conducted at Massachusetts General Hospital in Boston, The House Ear Institute in Los Angeles, and Mt. Sinai Hospital in New York, as well as centers in Hamburg, Germany and Manchester, England. No new treatments will be part of the NF2 natural history studies and patients may not have any treatments during the study. Patients interested in participating should contact Gloria Yoon, a research assistant at The House Ear Institute.

Tel: 213-273-8008; email gyoony@mailhouse.hei.org.

NF Awareness Month—A Great Success!

May was International NF Awareness Month and as usual we were busy trying to get the word out about NF to as many people as possible. Chapters and individual volunteers from across the country helped to make NF Awareness a success. Following are some highlights from May.

NF Story on ABC's Nightline



Porter Colley talks to medical students about living with NF.

Ms. Porter Colley, a valued and active member of the NNFF, appeared on the May 5, 2000 edition of ABC's Nightline. For the past 15 years Porter, who has NF1, has spoken to first year medical students at Harvard and M.I.T. about what it is like to live with a genetic disorder like NF. This year, Nightline sent a camera crew to film Porter's presentation as part of a story on innovative teaching methods at medical schools.

Porter tells us, "there hasn't been a doctor to graduate from Harvard or M.I.T. in the past 15 years who doesn't know a little something about NF!"

Nebraska High School Students Honored by NNFF for Marketing Success



DECA Competition; from left to right: Amy Hayes, Alex Eggers (student), Christine Baker (student), Janet Butler (teacher and parent of child with NF), Jamie Pittenger (student).

Jamie Pittenger, Kristine Baker, and Alex Eggers were honored by the NNFF for

excellence in public service. The three teenagers selected the NNFF as their "charity of choice" in the Marketing Association for Students (DECA) competition.

The three young women have successfully helped to raise public awareness of Neurofibromatosis. As a part of the project, they distributed information on NF and sold green NF ribbons to DECA associations in the U.S. and abroad as well as to the Millard West High School students and faculty. Amy Hayes, the president of the Northern Plains Chapter, had the honor of presenting the awards on May 11th at the Millard West High School in Omaha.



Buzz Westfall, County Executive of St. Louis, declares May NF Awareness Month. Left to right: Sue Slocomb, Buzz Westfall, Deborah Medbury, and Craig Kuhl.

Hear Ye, Hear Ye...

The states of South Dakota, Oklahoma, Missouri and New Jersey have issued official proclamations declaring May NF Awareness Month! Thank you to all those chapters and volunteers who helped to make this possible.

Welcome To A New Chapter in North Carolina

North Carolina was officially voted a new chapter of the NNFF at the National Meeting on May 20, 2000. The president of the North Carolina chapter is Tara Skirzenski. The chapter is off to a strong start having organized support groups in six different locations throughout the state, organized a medical symposium on May 7 and planning a golf outing and charity auction to take place this fall. Congratulations Tar Heels!



Tara Skirzenski with Paula Dorinson at the NNFF Chapter Council Meeting.

e-Bay Auction Going Strong

This May the Foundation kicked off our first ever on-line auction on eBay. Due to the growing success and increasing interest in this auction, it was decided to extend the event through the entire summer. For more information log on to www.nf.org/auction/ or log on to www.ebay.com and search under "4NF" to view all items listed on the NF auction block.

Taking the "Dis" Out of Disability

Foundation President Peter Bellermann and volunteer Kerry Getzinger appeared together on a Washington, D.C. cable show called "Taking the 'Dis' Out of Disability" on May 26, 2000. The show focused on what it is like to live with NF and how persons and families living with NF can receive help and support from the Foundation. This cable network can be viewed in more than 700,000 homes in the DC-area.

Modern Day Stone Soup

In the folk tale "Stone Soup," a soldier enters a town of closed doors with nothing in hand. The soldier showed strangers that their small contributions, when consolidated, equal greater benefits for all. Wisconsin Chapter President Elaine Pankow, is a contemporary version of this soldier. She spent countless hours going door to door, asking local businesses to make contributions to a Prize Drawing for NF. In addition, Elaine recruited her place of work to support fund raising efforts and awareness for NF. Fleet Mortgage Company agreed to have four 'Pay to Dress Down' days, during which employees donated money for the chance to come into work in casual clothing. The Wisconsin NF chapter simultaneously held a fund raising barbecue on May 24th. Everyone contributed and no one went hungry, just like the villagers in the story of "Stone Soup."



Research Provides New Insights

(continued from page 2) research are genetically identical to one another so they react in the same way to each experiment. By using mice that have been inbred to be genetically identical, it gives a more reliable result and helps to better determine what does and doesn't work in therapies.

We created our mouse model of NF1 by manipulating the genes that caused the mice to develop manifestations of NF1 (tumors). It is interesting to note that in the genetically altered mice with NF1, these same mice also showed signs of learning disabilities. Learning disabilities affect about 50% of the NF1 patient population.

NNFF: How do the mouse models work to help you understand the link between NF and cancer?

Dr. Parada: In the mouse models used in the study, and similarly by Dr. Jacks and his colleagues, we manipulated the genes in the mice along with the genes that are known to be involved in the development

(continued on page 8)

www.nf.org

Back to School Shop While Raising Money for Research

It's that time of year again—back to school time!

Our Shop2Give on-line mall has a huge selection of clothing, sports equipment, electronics and books to



help get your child or yourself ready to head back to class. You can shop for computers and educational software from Dell or OfficeMax. Buy your kids the latest back to school fashions at JC Penny or Lands End. You can even buy sports equipment and books at The Sports Stores and Borders.com! Best of all, each purchase helps further NF research with no extra cost to you.

Purchase a favorite book, movie or CD at Amazon.com



and a percentage will go towards NF research. Search any book or music category at Amazon from the NNFF Bookstore at www.nf.org/pat8-1.htm and find the perfect book for even the most discerning person.

Is your child nervous about starting school or going away to college for the first time? Send them a hug—an E-Bear hug! E-Bears can be shipped anywhere in the U.S. and makes the perfect accompaniment to any

purchase from the NNFF Online Store. Log onto our secure site at <https://secure.nf.org/store/> and start shopping today!



Join Our On-Line Community

Hundreds of people from around the world have joined the NF Chat room and the number continues to grow! The chat room allows you to have "live" discussions about issues pertaining to NF right from your own computer.

The NF Chat Room does not require any special software or any specific browsers (such as AOL, Prodigy, etc.). Anyone, anywhere that has an internet connection can use it anytime—all you need is access to the World Wide Web.

The NF Chat Room is open 24 hours a day, 7 days a week, for the convenience of people in international time zones. Although the NNFF has scheduled 10 pm on weekdays and 3 pm on weekend afternoons (Eastern Standard Time) for discussions, we urge you to establish chat sessions of your own at times that are practical for you. Just schedule a meeting with your buddies online whenever you want and then convene in the Chat Room at the designated time.

To use the NF Chat Room, all you have to do is register once. To register for the NF Chat Room, follow these simple instructions:

- Go to <http://www.nf.org/chat/chat.htm>.
- Read the "Rules and Policies" statement and click "Agree".
- On the registration page, click the "register new user" link.
- Enter your nickname, a password, password confirmation and email address. Click on the "register" button.
- Click the "Login" link on the confirmation page.
- Enter the username and password you just chose.

Congratulations! You have now joined over 500 users around the globe already in the NF Chat Room. Just type your message and click on the "post" button to let others view what you have written.

Thank You

A sincere thank you goes out to all of you who participated in the on-line Ebay auction this summer. Whether you donated an item to the auction or bid on something on the NF auction block, you helped to make a difference. Every dollar made from the auction will go to fund NF research in the US and abroad. Your generous contributions are helping to move us closer to our goal of finding effective treatments and a cure for NF.



New Member Brings Marketing Expertise To Foundation Board

The newest member of the Foundation's Board of Directors, Al Kahn, brings an impressive record of marketing achievements to the organization that will strengthen the NNFF's growing public education programs.

Mr. Kahn is the Chairman, Chief Executive Officer and Director of 4Kids Entertainment, Inc. a Nasdaq traded company. While previously at Coleco Industries, Mr. Kahn



Newest board member Al Kahn.

devised and executed the marketing strategy for one of the most successful toys in history—the Cabbage Patch Doll.

As the head of 4Kids Entertainment, Inc., Mr. Kahn is responsible for creating yet another toy sensation worldwide, the popular Pokemon in its many applications.

"We are delighted to have Al join the Foundation's leadership," said Peter Bellermann, NNFF President. "His experience and background lend considerable strength to our

Public Education Programs. We are determined to increase public awareness for NF1 and NF2, and Al will help us to do so."



PEN PALS

19-year-old woman with NF1 seeks pen pal. Write to: Angie Muncy, 33610 Guilbert Road, Eastlake, OH 44095.

If you live on the East End of Long Island I want to hear from you! I am a 37-year-old man with NF looking for pen pals. Please write to: Jonathan Hubbard, P.O. Box 678, Water Mill, NY 11976 or e-mail FRUITLOOPS11968@aol.com.

All letters will be answered! 41-year-old woman looking for pen pals to write or email. Email R_Werth@msn.com or write to Regina Werth 1426 Summit Avenue, Racine, WI 53402. Hope to hear from you soon!

Recently diagnosed with NF1 and looking for pen pals. 40-year-old man with NF1 who enjoys music, the outdoors and poetry would like to correspond with others. Write to: Billy Dykes #75014, P.O. Box 1010 Canon City, Colorado 81212.

NF PROFILE

Representative Al Lindley *The Power of One*

Representative Al Lindley has served in the Oklahoma state legislature for the last 4 years. He knows first hand the impact that one person can have in educating people and raising awareness for a cause you feel strongly about.

Before Rep. Lindley became a public servant he was a concerned father writing

“People think that one person can’t make a difference. They are wrong.”

to his representatives to help his daughter. Rep. Lindley knows all too well the impact that a disorder like NF can have on a family. Between 1965 and 1990, he lost three brothers and one sister to NF. Rep. Lindley has NF himself but has been lucky to experience only minor manifestations of the condition.

In 1972 his daughter Wendy was born already showing signs of NF. By 1978 she had already been to several doctors and endured several surgeries. These complicated surgeries left Wendy paralyzed and unable to speak due to the severing of the 7th nerve of the sympathetic nerve chain and damage to the brain stem. In 1985 Wendy passed away due to complications of her condition.

Rep. Lindley's experiences with NF in his own family are what got him interested in politics. In 1984 he began by writing letters to his state representative and his

Congressman urging them to support the U.S. Army funding for NF research. Shortly after he began his letter writing campaign he learned that his congressman, Glen English, had become co-sponsor of the bill to support U.S. Army funding for NF research. When he got the chance to meet Congressman English he asked him how many letters he had received on this issue that made him support the bill. English's answer surprised him—only one letter.

“People think that one person can't make a difference,” Rep. Lindley said. “They are wrong. Politicians are regular people and should be approached on a personal level. I urge people with NF to get to know their state representative and work with them to create awareness of this condition.”

Rep. Lindley firmly believes that putting a “face” to this disorder is what helps create awareness that translates into benefits for the NF community. Before his daughter passed away, he would bring her to State Senate committee meetings so that elected officials could see for themselves the problems that families with NF face.

Rep. Lindley is committed to using his position as a state representative to make a meaningful contribution to people with disabilities. The state legislature often votes on legislation regarding health insurance and disability benefits. He has brought persons with NF to the floor of the state house to talk about

their experiences and work with their representative to affect change and educate the public. Dr. Mulvihill of the Children's Hospital in Oklahoma City has addressed the legislature on issues related to children and NF. Due to Rep. Lindley's efforts, this year will be the fourth consecutive year that Oklahoma has declared May NF Awareness Month.



Oklahoma State Representative Al Lindley.

Most recently, Rep. Lindley was the driving force behind a bill to create a task force that will address issues of assistance for persons with disabilities. In May, Governor Keating signed the bill (HB2356) enacting the creation of the Task Force to Review the Complaint System for Persons with Disabilities. Twenty-eight of the 101 House members were co-authors of the bill.

“It takes time to raise awareness and affect change in our health care system,” Rep. Lindley said. “People need to get involved, get to know the people who represent them and make important legislative decisions. You need to stand up and be counted so that legislators know that the NF community has important health concerns that need to be taken seriously.”

NF

NFFF Day at the Lab[®] Program Goes National

(continued from front page) the lab.

The children made gels where they inserted samples of different genetic materials and used a process called “electrophoresis” to determine what type of materials were being used in the experiment. Microscopes with different cell samples were set up for the children to examine and genetically



Children pose with Dennis Haskins of NBC's *Saved by the Bell* at the Los Angeles Day at the Lab program.

altered mice were on display as well. Each experiment brought a new round of questions about NF and genetics.

One of the most practical applications of science shown to them that day—in the children's opinion—was the use of liquid nitrogen to quickly freeze a concoction of strawberry ice cream! Children left the program with a greater understanding and excitement for genetics and learning about NF.

Making DNA From Scratch— Los Angeles

The House Ear Institute in Los Angeles was the site of the April 15, 2000 Day at the Lab program.

Drs. William Slattery and Gene Hung answered questions from teens about NF, took them on a tour of a genetics laboratory and worked with them to perform hands-on genetics experiments.

A special guest was also on hand to help the children with their experiments. Mr. Dennis Haskins of NBC's popular teen show *Saved By The Bell*, worked with the children to extract DNA from peas and help them twist string into a DNA double helix that simulated E. coli bacteria in the nucleus of a cell.

All three events were a great success and the children learned a lot about NF while making news friends and having fun. The Foundation plans to continue the program in the coming years in other cities throughout the United States.

NF

NF Marathon Team Update

Todd Mozzer is a confident man. He sets high goals and standards for himself, and always proves quite capable of reaching what he sets out for. The success of the NNFF Marathon Team Program lies in the dedicated efforts of people like Todd who are using their energy and enthusiasm to help move research forward towards a cure for NF.

Since November Todd is more than one quarter of the way (\$27,000) towards his goal of raising of \$100,000 for NF research. His drive to raise monies for the cause is a very personal one. As a father of two children, he hopes to set an example. His son Trevor, diagnosed with NF1 at 8 years old, will hopefully benefit most from Todd's heroic efforts.

In March Todd ran the Maui Marathon with Nanette and Michael Julian, who ran in honor of their daughter Elise, who has NF. Out of the 2,700 participants, Todd finished 197th, registering a time of 3 hours, 40 minutes.

"A lot of people ask me why I do this, and how hard was it? The last few miles are excruciating but Trevor, Andrew and Amy got me through it, the image of those faces are enough to finish the race," Todd said.



Todd Mozzer (far right) poses with Nanette and Michael Julian at the finish line of the Maui Marathon.

In addition to the marathon team, Todd has either organized or participated in (and often both) several different events. In April, he coordinated a 24-hour swim-a-thon called "Jump in the Pool for NF". He recruited 147 swimmers to raise money for NF. In May Todd recruited a Cajun band to perform at a bake sale. Friends and family were recruited to work the concession stands as the crowd, including local NF families, danced the night away. Todd has already signed on for another marathon and continues to plan events to raise money and awareness for NF.

"The [marathon] team is an amazing win-win deal," Todd said. "People talk and write about marathons all the time, but only the ones who make it to the finish line, and

neuro-fibroma-tosis®
NF Marathon Team
racing for research

UPCOMING MARATHONS:

| | |
|--------------------------|--|
| December 10, 2000 | Honolulu Marathon |
| January 14, 2001 | Disney World Marathon |
| February 24, 2001 | Myrtle Beach Marathon |
| March 18, 2001 | Maui Marathon |
| April 2001 | Country Music Marathon, Nashville TN |
| May 2001 | Vancouver Marathon |
| June 2001 | Rock N Roll Marathon, San Diego |
| June 2001 | Mayor's Midnight Sun Marathon, Anchorage |

those who gave it their all, only they will know what it really feels like."

While Todd's commitment and drive are extraordinary, we do not expect every individual to duplicate his efforts. One thing to take away from Todd's story is that participating in the team not only helps raise money and awareness for NF, but also makes you a proactive part of the process of working towards a cure. To get involved please call Anita Carter at our Marathon Team Headquarters (toll-free) 877-NF-RACES (637-2237) or email RaceForNF@aol.com.

Neurofibromatosis: Trusting God in the Midst of NF

Reviewed by M. Priscilla Short, MD, Dept of Pediatric Neurosurgery, University of Chicago and the Science, Research and Technology Division, American Medical Association. Dr. Short is also a member of the NNFF's Clinical Care Advisory Board.



Dr. M. Priscilla Short.

The book, written by Renee B. Wilson, is one mother's heartfelt account of her son Kevin's neurofibromatosis 1. Her account begins with the diagnosis of NF1 being given to them after a number of encounters with the medical profession. She eloquently relates her sense of anxiety in seeking additional medical expertise, her sense of guilt, and her needs to understand where or from whom the NF might have been inherited. Ms. Wilson receives great support from her faith. Other readers should not be offended by her reliance on faith as she balances it with recognition to seek medical attention.

One of the tragic themes of her account is the numerous occasions when the medical community appears to have let her down, in terms of not pursuing relatively significant clinical signs of developmental delay separate from the issue of NF. Given Kevin's

cutaneous finding of NF, this should have led to an earlier diagnosis and introduction of early intervention. Sadly the author blames herself for not having recognized signs earlier.

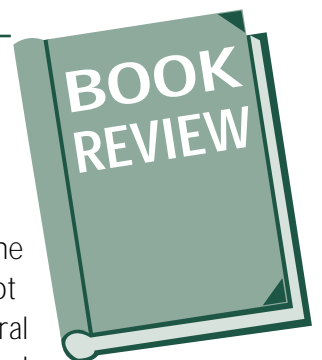
One of the best chapters is the account of Kevin's evaluation by the public school special education department. This chapter should be required reading for all parents whose children may need additional services to succeed regardless of a diagnosis of NF. Of importance to note, Ms. Wilson demanded no less of the school than she did of herself and came prepared to advocate for her child.

It is unfortunate that there are several errors in the preface that detract from the book. Ms. Wilson mixes up NF1 and NF2 in her description of the symptoms associated with NF1 (hearing loss is predominantly associated with NF2). Her characterization of Kevin's MRI findings of thickened optic nerve versus optic glioma will confuse some parents. Making a diagnosis of optic glioma requires a concomitant visual exam to discern visual defects as Kevin had upon his follow-up ophthalmologic examination. In addition, she refers to Kevin as having a "fibroma that affects his cerebellum" which is confus-

ing to the reader. Some children with NF may have some fine motor delay that is not related to any structural brain abnormality seen by MRI. Her description may be confusing to other parents who may assume that fine motor difficulty means there is a causative structural abnormality. It would have been helpful if one of the physicians with whom she has seen could have assisted her in the preface.

The tone of the forward is somewhat judgmental and speaks to concerns about how healthcare professionals communicate diagnosis to patients. Particularly for a genetic condition of variable severity as NF1, how the diagnosis is given may significantly impact on the probability of negative future events (e.g., future negative discriminatory events). For the majority of children with NF1, the diagnosis does not equate with certainty of a "lifelong change for [that] child."

For many Ms. Wilson's book will be inspirational. For the healthcare professional reading it, it should be viewed as a cautionary tale as to how we communicate to support patients.



CHAPTER NEWS: Waging the Fight Against NF at the Grassroots

Community Effort

A successful fundraising golf event was held in Staten Island, New York, on April 27th organized by Lydia LoPrimo. Organizers kept their costs low by soliciting friends, family, coworkers, and fellow community members (more than 100 people) to participate and contribute to the fundraising effort. Public service announcements for this NF Golf Tournament were made for an extended period of time on the sports radio station WFAN.

Grabbing Headlines Crossing Finish Lines

On April 29th, The Indiana Chapter of NNFF held their 10k run. Many of the 138 participants came due to pre-race coverage on a local news station, WISH TV-Channel 8. Local newsman and race participant, Scott Swan hosted two telecasts on April 26th and 27th. This publicity tapped into the potential to connect members of the NF community, while conducting public outreach and education. As Indiana Chapter President Dottie Whitehurst relayed, "a good time was had by all".

The Art of Fundraising



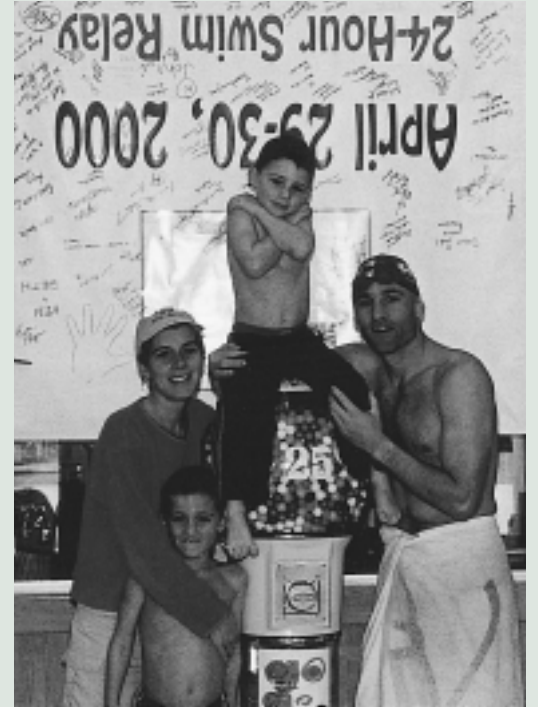
Painting auctioned to benefit NF research at the Robert Canaga Gallery on Portland, OR.

Throughout the month of May, six artists and a gallery in Oregon supported NF Awareness Month through fundraising. At the two sites of the Robert Canaga Gallery, charity art auctions were held to benefit The Washington Chapter of NNFF. Postcards with images of the artist's works were sold, with a percentage of the profits going towards this benevolent effort. The contributing artists included A.J. Boots, Robert Vetter, Patricia Millar, Nancy Warren, Mike Van, and Sarkis Antikajian.

Take the Plunge!

Todd Mozzier of the Connecticut Chapter organized 147-person swim-a-thon in April. A month before the event, Todd

educated volunteer swimmers on NF and distributed educational materials. Marcy MacDonald, who swam the English Channel, swam for a total of 22 hours covering more than 8,400 yards! Plans for next year's event are already in the works.



Left to right: Amy Mozzier, Trevor Mozzier, Andrew Mozzier, Todd Mozzier, at the 24-hour Swim Relay, April 2000.

NF

Research Awards

(continued from front page) dish) and in vivo, i.e. in the mammalian NF2 mouse model.

The Joseph S. Byrka Medical Research Fund

Dr. Janet Hock, Indiana University
One-Year Research Grant

"Skeleton Phenotyping and Bone Cell Function in Heterozygous NF1 Knockout Mice"

Dr. Hock seeks to provide an overview of how the NF1 gene product, neurofibromin, regulates skeletal development and bone cell function. She will test whether bone abnormalities are the result of defective neurofibromin or a secondary consequence of neurofibroma or abnormal hormone functions. Using the mouse model, Dr. Hock will examine whether there are abnormalities in how "bone building" cells (osteoblasts) respond when the animals are treated with a hormone that activates osteoblasts. She will also evaluate whether the NF1 mutant mice exhibit decreased numbers of precursor cells for either osteoblasts or "bone eating" cells (osteoclasts). A better understanding of these processes leading to skeletal abnormalities in NF1 should lead to the development of new drugs for treatment of common skeletal diseases to change skele-

tal growth and healing and thus improve the quality of life for people with NF1 and bone lesions.

The Barrie and Faith Sommerfield Family Fund

Dr. Michael Stern, Rice University
One-Year Research Grant

"Cell-Nonautonomous Regulation of Perineural Glial Growth by Drosophila in NF1"

Dr. Stern will address one of the basic questions in NF1, namely why does the loss of NF1 in Schwann cells cause the overabundance of neighboring, genetically normal cells and thus lead to the formation of neurofibromas?

He is especially interested in investigating whether the loss of a potassium channel is responsible for the over proliferation of perineural cells in drosophila, whether such a potassium loss leads to an excess of calcium that could trigger the release of signals that affect neighboring cells.

The Feinberg and Kany Families Research Fund

Dr. Wanda Salzer, USAF Keesler Medical Center, MS
Two-Year Young Investigator Award

"Molecular Basis of Plexiform Neurofibromas

and its Correlation With Response to the Farnesyltransferase Inhibitor R 11577"

Dr. Salzer is part of a group at the National Cancer Institute that has begun clinical trials, testing a farnesyltransferase inhibitor in plexiform neurofibromas in patients with NF1. Her particular study will examine what the effects of a new version of the drug, i.e. R11577, are on plexiform neurofibromas and on several ras pathways that are thought to be involved in the formation of plexiform neurofibromas.

Steven and Lottie Walker Family Foundation

Dr. Anna Tchoudakova, Harvard/MGH
Two-Year Young Investigator Award

"Genetic Analysis of NF1 Function in Drosophila"

Dr. Tchoudakova will design experiments that seek to further clarify the relationship of the NF1 protein and the cyclic-AMP (c-AMP) signaling pathway in the cell. This pathway plays major roles in learning, memory and hormonal signaling. The work should lead to a better understanding of the causes of cognitive deficits seen in people with NF1.

NF

Research Provides New Insights


(continued from page 3) of several cancers. The product of NF1, a tumor suppressor gene, was thought to cooperate with the product of another tumor suppressor gene, p53, that is located on the same chromosome in the same region as NF1 in both humans (chromosome 17) and in mice (chromosome 11), and is mutated in the majority of cancers. By doing this, we have uncovered a configuration of genes that lead to the formation of neurofibrosarcomas (malignant tumors). Because we already know so much about the NF1 gene and its protein, we are developing ideas for therapies to block the tumor process in NF.

We are excited about the fact that we now know which type of genetic lesions occur in NF1 patients for cancer to develop. With this new knowledge, we can now focus on directed therapies to reverse the action of the NF1 and p53 (and related) genes in these tumors.

NNFF: What are you and your research team currently working on?

Dr. Parada: We are currently working on more sophisticated models of NF in mice that permit us to study, in isolation, the different functions of the NF gene in cells of the nervous system. We can now study cells in which the NF1 gene is lost, and why it causes tumors to grow. We contin-

ue to better understand the mechanisms by which the NF1 gene manifests itself.

In addition, my laboratory is currently collaborating with other scientists who are working on the NF1 gene. We are looking at the relationship between learning and memory and NF1; NF1 and leukemia; NF1 and bone repair; and NF1 and the normal development of the heart. 

A New NF Study Needs Your Help!

If you have any form of NF and have ever taken any of the following drugs we would like to hear from you:

- Quinidine
- Quinaglute
- Disopyramide
- Norpace
- Procainamide
- Procan
- Pronestyl

Please contact Dr. Mia MacCollin, maccollin@helix.harvard.edu or 617-726-5725. Thank you for helping with this study.

In Memoriam

John Petito *Editor of the NF2 Review*

The Foundation mourns the death of John Petito, the editor of the highly respected NF2 Review.

"We all owe John a great deal," said Foundation President Peter Bellermann. "Despite his valiant struggle against NF2, John gave and gave to others up until the very last moments of his life. He has been a source of inspiration and a hero to all who are affected by NF2. His energy, his humor and his good nature in the face of great adversities will be missed by us all."

neuro·fibroma·tosis
THE NATIONAL NEUROFIBROMATOSIS FOUNDATION, INC.

95 Pine Street, 16th Floor New York, NY 10005

Telephone: 1-800-323-7938 or 212-344-NNFF

Fax: 212-747-0004

E-mail: NNFF@nf.org

On the Web: www.nf.org

Managing Editor: Erica Mayer

The purpose of the National Neurofibromatosis Foundation, Inc. (NNFF) is to improve the well-being of patients and families affected by NF1 and NF2. The Foundation therefore sponsors scientific research aimed at finding the causes and cures for the neurofibromatoses, promotes the development of clinical activities, works to create public awareness and provides patient support services.

The National Neurofibromatosis Foundation is a founding member of the International Neurofibromatosis Association.



neuro·fibroma·tosis

THE NATIONAL NEUROFIBROMATOSIS FOUNDATION, INC.
95 Pine Street, 16th Floor • New York, New York 10005

Address Service Requested

Non-Profit
Organization
U.S. Postage
PAID
New York, NY
Permit No. 3646

Inside:

- 1 Teens with NF Become Scientists for a Day
- 3 NF Awareness Month Highlights
- 6 NF Marathon Team Going Strong

Shop ON-LINE To Benefit NF:
www.nf.org
(see page 4)