

neuro·fibroma·tosis®

THE NATIONAL NEUROFIBROMATOSIS FOUNDATION, INC.

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neuro·fibroma·tosis® NF Marathon Team *racing for research*

We are very pleased to announce a new program that will help us expand our public awareness, increase our revenues and "pick up the pace" in our efforts to fund research towards a cure or treatment for NF.

The NF Marathon Team - Racing for Research program offers both runners and walkers from all over the globe, experienced or novice, the chance to enjoy the thrill of completing a marathon while they raise funds to help support The National Neurofibromatosis Foundation's valuable work.

With eight races around the U.S. and in Dublin, Ireland (see box p. 6), this program offers the opportunity to participate in some of the most exciting races available.

Participants receive training in fundraising and have goals to raise a certain amount for the NF cause. Each walker/runner can qualify for free transportation, lodging and more for the marathon weekend by raising the minimum amount in donations. In addi-

tion to raising much-needed funds for NF research and NNFF programs, becoming a member or donor of the NF Marathon Team helps spread NF awareness.



Anita Carter and son, Joshua

Anita Carter, the creator and coordinator of this program for the Foundation, is also the past President of the Illinois NNFF Chapter. Not only an advocate for NF, Anita is also a walker and fundraiser who has raised over \$50,000 in her own walks for NNFF and has recruited others to successfully do the same.

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NNFF Scientific Consortium Meets at MIT

While the NF1 AND NF2 genes are large and complex, their mysteries are played out at a much more specific level. The molecular biology of NF involves the exacting task of breaking down the most minute actions and reactions within and between the cells to better understand how the NF genes function.

This challenge is one that dozens of scientists from around the world take on everyday in their labs. The National Neurofibromatosis Foundation is instrumental in moving this research process along. One way the NNFF manages research in the molecular biology of NF is to host the NNFF Consortium on Molecular Biology of NF1 and NF2 so that researchers can share their up-to-the-minute findings with their colleagues. This year's meeting took place during the summer at MIT's prestigious Whitehead Institute for Biomedical Research and was

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organized by Tyler Jacks, Ph.D., MIT Center for Cancer Research; and André Bernard, Ph.D., Massachusetts General Hospital Cancer Center.

Although the NF2 gene was identified after the NF1 gene, progress in NF2 research is moving more rapidly than expected, according to Dr. Jacks. The presentations by NF2 researchers noted progress on three fronts – biochemical studies, regulation of and by Merlin (the NF2 gene product), and in modeling of NF2 in animals. Dr. James Gusella of Harvard Medical School/MGH and Dr. Gilles Thomas of the CEPH Foundation Jean Dausset in France chaired the NF2 portion of the meeting.

In addition, new researchers were brought into the consortium to expand the fields of research with potential impact on NF2.

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1999 RESEARCH AWARDS ANNOUNCED

The National Neurofibromatosis Foundation has announced the recipients of its research awards for the academic year 1999-2000. The awards are as follows:

The Joseph S. Byrka Medical Research Award

Dr. Jeffrey Field

University of Pennsylvania

One Year Research Award

Project Title: "The Role of Pak Protein Kinases in NF"

Dr. Field's study is designed to determine the cellular events that contribute to the development of neurofibrosarcomas. He has developed a new way of blocking the growth of neurofibrosarcomas in cell cultures in the lab. His experiments should pinpoint the precise signal in the cell that causes this blockage. Once the mechanism is known, the information is expected to be useful in the development of drugs that can be used to treat neurofibrosarcomas and neurofibromas.

The Feinberg and Kany Families Research Fund

Dr. Yi-Ping Hsueh

Massachusetts General Hospital

Two-Year Young Investigator Award

Project Title: "Molecular and Functional Interactions of NF1"

By determining what proteins interact with NF1, Dr. Hsueh wants to gain a better understanding of how neurofibromin controls cell growth and cell differentiation. Dr. Hsueh will study the relationship between syndecan, a cell surface protein, with the ras and neurofibromin proteins and determine the functional significance of their interaction. It is hoped that these efforts will lead to a better understanding of how NF 1 begins and thus may offer clues how to control the disorder.

The Barrie and Faith Sommerfield Family Fund

Dr. Kathryn North

New Children's Hospital, NSW, Australia

One-Year Research Award

Project Title: "Profile of the NF1 Phenotype: Natural History, Neuropsychological and Psychological Aspects"

Dr. North's proposal addresses a number of important unresolved questions concerning cognitive deficits in children with NF1. She

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GENETIC TESTING FOR NEUROFIBROMATOSIS TYPES 1 & 2

(Ed. Note: The following article is based on interviews with Margaret Wallace, PhD, and Mia MacCollin, MD, scientists involved with NF1 and NF2 research, respectively. Dr. Margaret (Peggy) Wallace is an Associate Professor of Pediatrics in the Division of Genetics at the University of Florida, Gainesville. She is a member of the NNF Research Advisory Board and the NNF Florida Chapter Board of Directors. Dr. Wallace's area of expertise is the molecular genetics of NF1. Dr. MacCollin is an Assistant Professor of Neurology at Harvard Medical School and the Director of the Neurofibromatosis Clinic at Massachusetts General Hospital. She is also a member of the NNF Research Advisory Board. Her research is focused on the genotype/phenotype relationships of NF2.)

Neurofibromatosis Type 1

Q. Who is an appropriate candidate for testing?

A. Today, the clinical diagnosis of NF1 is so good that a genetic test is typically not necessary. Appropriate candidates for testing would be those looking for a prenatal diagnosis. Or, when there is no family history and a young child doesn't have a second NF characteristic needed for diagnosis.

Q. Who is not an appropriate candidate for testing?

A. The test is not recommended when someone has already met the diagnostic criteria for NF1. If a child is suspected to have NF1, but does not meet diagnostic criteria and has no medical problems, annual follow-up exam by a physician is usually recommended rather than a genetic test.

Q. What tests are used to diagnose NF1? As an adult? Prenatal?

A. The most common test is the protein truncation test done by Lab Corp using a blood sample from the patient. It is capable of screening the NF1 gene for a serious mutation, such as when neurofibromin is either too short or missing. The protein truncation test can be done on anyone regardless of family history or age.

When there is a family history of NF1, a linkage analysis test can be done by a commercial lab. Blood is collected to get DNA from family members who have NF, and possibly, unaffected family members. Since this method detects a marker for the mutation, rather than the actual NF1-causing mutation, it is extremely accurate but not quite 100%. A pre-natal linkage analysis test can be done when there is a family history of NF1 and two other generations are available for testing. While LabCorp no longer conducts a prenatal test, there still are some research labs collaborating with clinical labs to perform a specific genetic

test. The patient's doctor can contact the labs to arrange for the test, but there are no guarantees that this can be accomplished.

Q. What are the test's limitations? Why?

A. The protein truncation test is only 60-70% sensitive. That means that a negative test result in NF1 is meaningless because the person may still have NF1 but not a detectable mutation. This is because some mutations do not cause truncation. A positive result should confirm an NF1 mutation, but it is possible that, in rare cases, a positive protein truncation test might not mean a diagnosis of NF1. Thus, diagnosis based on clinical features or a specific DNA mutation is considered the best.

None of the tests can predict the severity of the course of the disorder or the number or types of manifestations of NF1.

Q. What are the common misconceptions about these tests?

A. People mistakenly think that they need a test to validate the clinical diagnosis. It is often difficult to explain that a negative protein truncation result does not mean that someone definitely does not have NF.

Q. What does it cost? Covered by insurance?

A. The protein truncation test costs about \$500-750 and LabCorp accepts most insurance if the insurance company is willing to pay. The test should be arranged by and results discussed with a doctor. The gene linkage test may cost hundreds of dollars per sample, but the cost may vary widely depending on the lab and the number of persons sampled. All payment issues need to be coordinated with the patient's insurance company and the commercial lab.

Q. What new tests are on the horizon? Ideal test?

A. A better test, especially for cases with no family history, would be a DNA chip-based test that could screen for all possible single base mutations, although in NF1 this would still not be 100% sensitive. Currently no NF DNA chip exists and it would be very expensive to develop. Right now, chips are available for breast cancer mutation screening and cost hundreds of dollars per test. In the next 5 to 10 years there should be a better test, not necessarily cheaper, but with higher sensitivity for finding NF1 mutations.

Q. Where does someone go to be tested?

A. The tests should be arranged through a doctor. It is important that your doctor is

comfortable finding an appropriate lab and interpreting the test's results, so it is probably best to go to a NF clinic or a medical geneticist.

Neurofibromatosis Type 2

Q. Who is an appropriate candidate for testing?

A. A child whose parent is classically affected with bilateral vestibular schwannomas is an appropriate candidate for testing. Rarely, adult members of large families with NF2 who do not know the exact status of their parents, are also candidates for testing.

Q. Who is not an appropriate candidate for testing?

A. A person who already has bilateral vestibular schwannomas, the characteristic sign of NF2, is not an appropriate candidate for testing. In these cases, a blood test is unnecessary to confirm the diagnosis and may be misleading to the family.

Unfortunately, the current test is also not helpful in cases when a diagnosis is in question for the first affected person in a family because the test cannot find a mutation in 1/3 of new mutation cases. Finally, if an uncle, aunt, grandparent or sibling is known to have NF2 but a parent does not, a test is not needed because familial cases of NF2 are inherited only from an affected parent - there are no NF2 carriers and it can't skip generations.

Q. What tests are used to diagnose NF2? As an adult? Prenatal?

A. If there are two or more affected individuals, linkage analysis is done through several blood samples. Linkage analysis involves tagging the chromosome and following the pattern through the family to see if the at-risk child has an affected or non-affected chromosome. If only the parent is available, mutational analysis of a blood or tumor specimen must be done in order to detect the actual genetic change that has caused NF2 in the family. Because analysis of tumor specimens is often more informative than blood, patients may wish to consider having their surgeons freeze samples for future analysis if they think that testing may ever be a consideration for other family members.

For prenatal testing, it is essential that the family plan ahead, since initial analysis is often time consuming and cannot be completed once a pregnancy has already occurred. We often recommend that couples meet with a genetic counselor long before they are contemplating a pregnancy to discuss the many medical, social and psychological factors involved in testing. Once mutational analysis or linkage analysis has

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The test is not recommended when someone has already met the diagnostic criteria.

CHAPTER NEWS: Waging the Fight Against NF at the Grassroots

There are many ways to raise funds for NF research programs and promote awareness of neurofibromatosis with your community. Recently, several states Chapter's and individual members of the Foundation organized activities and fund raising campaigns that are excellent examples of ways to support the fight against NF.

Invent New Ways to Support NF Spin a Beetle

Crissy Ortman of St. Louis, MO was one of four finalists in a "Spin the Beetle" contest held in a local car dealership and sponsored by radio station Y98. The object was to sit in a VW Beetle for 72 hours, while being spun on a platform (10 minute breaks were given every 3 hours). The last person to remain in the car would receive a 2-year lease on the Beetle. Crissy won! She asked family and friends to donate her lucky number of \$17 to the NF Foundation to show support.



Spinning in the VW Beetle

Get a Trim



Taylor Mabardy getting a trim from her grandmother, Josephine Volpe, who hosted the Hair-Cut-A-Thon at her shop "Daniel."

A Hair-Cut-A-Thon was held in Arlington, MA in June. The salon's clients who participated in the fundraiser, organized by Teresa Mabardy, received haircuts, hair care samples, cookies and information about NF.

Sport Your NF Pride Run A Concession Stand

The Missouri NF Chapter has been chosen to manage a concession stand at Busch Stadium as the Cardinals and Mark McGwire play to their hometown fans to close out their season. The Chapter will have an opportunity to raise money and spread the word about NF at the same time as they root for the local team.

In Indianapolis, the NF Indiana Chapter has been chosen by the Indianapolis Colts Football team to manage a stand during the team's 1999-2000 season.

Capitalize on a Popular Pastime Clean Up for a Good Cause

Iowa Chapter President, Sheila Drevyanko and her family turned their driveway into a

shopping mall in August when they held a huge garage sale to benefit NF. Chapter members, their friends and families donated and purchased a wide variety of items. Announcements for the sale were placed in local publications, thereby spreading the awareness of NF throughout the region.

Other successful garage sales were held earlier this summer in Hawaii and North Carolina.

Spread the Word about NF Young Emissaries Speak Out

18-year-old Californian, Ryan Root returns to his alma mater, Camarillo High School, to give a talk about NF and learning disabilities to the health science classes.

Use Flower Power for Fundraising Plant Seeds

Volunteers from all over Illinois participated in the 6th Annual Daisy Day. Packets of daisy seeds and literature about NF were distributed to create awareness of NF.

Partner with Local Merchant

Volunteers sold cookies, muffins and pies at a bake sale at the Wal-Mart in Ardmore, OK. The store provided the space and a matching gift. Local TV and newspapers covered the story leading up to the event.

Summer Rounds of Golf Around the United States

Golf tournaments continue to be a popular means to raise funds for research programs and create public awareness of neurofibromatosis. Numerous tournaments were held during the spring and summer across the country:

The **9th Annual Friends of NF Celebrity Golf Classic with Mark Chmura** took place in June at the Ellington Ridge Country Club outside of Hartford, CT. This year's event raised record proceeds for NF research and also set a new record with 276 participants.

The event had many local and national sponsors including tournament title sponsor Prudential. More than 50 celebrities participated representing the NHL, NBA, MLB, NFL and numerous colleges. Among the celebrities in attendance were Mark Chmura, all-pro tight end *Green Bay Packers*; George Rogers, *Heisman Trophy* winner; Larry Little, *NFL Hall of Fame*; Leonard Marshall, *New York Giants*; John Davidson, *MSG Network*; Ron Greshner, *New York Rangers*; Garo Yepremian, *Super Bowl Miami Dolphins*; Jeremiah Trotter and Mike Caldwell, *Philadelphia Eagles*; Geno Auriemma, *UConn Women's Basketball* coach and *NBA* champion, Scott Burrell.

The **First Annual Gordon Schott Golf Memorial for the NF Foundation** was held in early August at the Travis Pointe Country Club in Ann Arbor, MI. The tournament was conceived and organized by Cindy Schott-Pfeifle, President of the NNFF Michigan Chapter in honor of her late father.

NF supporters were once again coupled with Gateway PGA golfers at the Missouri Chapter's **9th Annual NF Celebrity Golf Classic**. One pro played with each of the 35 foursomes, making for a very exciting round of golf at the Fox Run Golf Club in Eureka

The **Second Annual Jerry Shannon Memorial Golf Tournament Benefiting NF** was held in Rochester, NY. Funds raised at the event, which was organized by Colleen Lacy of

Webster, NY in memory of her father, have been designated for NF1 research programs.

Muttontown Country Club in East Norwich, NY was the site of the **Sixth Annual New York/New Jersey Golf Classic**. Brian Behrens again served as Chair of the event which included a full day of golf, a barbeque supper and an auction filled with great prizes. His firm, Donaldson Lufkin & Jenrette was a major sponsor. Other corporate sponsors included ABM Co. New York, Access Direct Systems, AGFA Corporation, Ambassador Construction Co., ING Baring Furman Selz LLC, Merrill Lynch, Pierce, Fenner & Smith, Morgan Stanley & Co., Inc., and RJR Nabisco.

NF



Star-studded line-up prepares to tee-off in CT.



Web News

The NF Website had over 500,000 hits in June. Was one of them from you? If not, you can be in the next count by going to <http://www.nf.org>.

NF Online Shopping Mall

The Foundation has formed an online partnership with Shop2Give.com offering web visitors the opportunity to purchase merchandise from over 60 online stores while benefiting NF **at no additional cost**. Simply click on the Shop2Give icon located on the NF Homepage (www.nf.org). As users enter an online store on the Mall a tracking device identifies the customer as having arrived via The NF Website. When a purchase is made a percentage is then donated to the Foundation from the store. Most percentages range from 3-5%, but some range as high as 15% such as Planet Rx, an online pharmacy. Each store lists how much they will donate, which can be a helpful factor when deciding between competing stores. Store categories run the gamut from books and music to travel, clothing, health, kids, pets, and many more. Several flower delivery services are offered on the Mall including 1800Flowers, a great way to mark an anniversary or a birthday. As the holiday season approaches why not do your shopping through the NF Online Shopping Mall and put a smile on your loved ones' faces while helping in the race

to find a cure. In addition to the NF Online Shopping Mall, visitors to The NF Website can purchase a variety of items in the Online Store.

E-membership Drive in Full Swing

Like all other Foundation members, e-members are very special. They are a new group of members who are signing up via The NF Website at www.nf.org/join.

How they sign up is one way they stand out. The other way is that it costs the Foundation much less to have an e-member sign up. Traditionally members respond to mailings from the Foundation. Preparing these mailings costs money, i.e. designing the mail pieces, printing, paper, hiring the mail house, adding the postage, processing incoming checks and paying bank costs. When an e-member signs up, most of these costs are not incurred and the savings can go to research and public education.

Some of the savings go, however, to the e-members themselves. The memberships of both individual e-members and family e-members received via The NF Website are 15% less than the traditional Foundation memberships. Individual e-member memberships are \$ 29.75 (instead of the traditional \$ 35.00). Family e-member memberships are \$ 42.50 (instead of the traditional \$ 50.00). Members may also renew their membership online and receive 15% off.

We Have a Winner!!!

Anneliese Bosco of New York State was recently announced as the Grand Prize Winner in the National NF Foundation's Airline Ticket Sweepstakes.



Entrants provided their email addresses to enter the sweepstakes and helped to expand the Foundation's electronic distribution list to over 1,000 recipients. The winner is the mother of a 15-year-old daughter who has NF and hopes that one day people will not only be able to treat NF's symptoms, but to cure them. She firmly believes that public education is an important part of the fight against NF. Anneliese, along with her two children, will be participating on the NF Marathon Team in the Disney World run/walk marathon to promote awareness. Anneliese also plans to volunteer her time and energy as co-President of the Schneider Children's Hospital Support Group on NY's Long Island. Keep an eye on The NF Website for another opportunity to send us your email address and a chance to win!

Bara Colodne, an intern at the National NF Foundation, draws the winning email address.

MIT Consortium

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While Dr. Elizabeth Luna of University of Massachusetts and Dr. Mitsuyoshi Nakao of Kumamoto University, Japan do not conduct research into NF2 specifically, their interests within molecular biology may provide additional clues to the behavior of the gene.

Biochemical studies explore the relationship of the NF2 protein, Merlin, to other similar proteins found in cells. Discovering the interaction between these various ERM proteins will help scientists understand why the loss of the Merlin protein that occurs in NF2 occurs and how it affects the functioning of the NF2 gene.

One of the most significant discoveries shared was the first 3-D model of ERM proteins, which interact with Merlin. The detailed atom-by-atom structure developed by Dr. Anthony Bretscher of Cornell University and is the most detailed view of this class of protein that has ever been developed. "Dr. Bretscher's model gives us a

better sense of what Merlin's structure may look like," Dr. Jacks said. "This is a monumental discovery that will help in making progress towards understanding Merlin's function."

In a related area of research, several scientists discussed their finding about the regulation of and by Merlin and the signaling pathways among cells. Some of the researchers found new proteins that interact with Merlin. These discoveries reveal more about the signaling pathways and their functions than was previously known. For example, Dr. Andrea McClatchey of the Massachusetts General Hospital suggested that NF2 might function indirectly in the Ras signaling pathway. Traditionally, only NF1 was thought to have an interaction with Ras. This finding suggests that NF2 and NF1 may have more in common than was previously thought, according to Dr. Jacks.

Other researchers focus beyond the cellular level to explore NF in the context of a traditional genetic model. The area of realistic disease modeling in fruit flies and laboratory mice is critical to the development of effective treatments. The presentations marked a significant improvement in animal models that mimic NF in humans. Dr. Marco Giovannini of the CEPH Foundation Jean Daussett in France discussed a refined mouse model for NF2 that produces Schwann cell tumors. Dr. Richard Fehon of Duke University has been able to substitute a human gene for NF2 for a fly gene. This opens up the possibility of studying human derived mutations in the fruit fly. Experiments could then be conducted on the altered flies to learn more about signaling pathways and the consequences of the loss of Merlin in the cells.

Other presentations given during the NF2 session focused on various aspects of the

NF PROFILE

Stu Wright Brings "The Rude Awakening" to South Carolina

Waking people up is Stu Wright's mission in life. As a radio professional he helps tens of thousands of South Carolina residents start off their day with his popular morning drive time radio show, "The Rude Awakening", on WORG-FM. But Stu also has devoted his spare time to waking the public up to the facts about neurofibromatosis.

For nearly twenty years, Stu has brought a combination of talk, news, and easy-listening rock music to the city of Orangeburg. His top-rated show, mellifluous voice with just a hint of a southern-drawl, and humor have made him a fixture in town.

Born in Beaufort, South Carolina, Stu was first diagnosed with NF1 when he was eighteen years old. An astute neighbor, who was a Navy dermatologist, made the diagnosis.

Having NF1 excluded Stu from military service at the height of the Vietnam War. He has mixed feelings about the automatic disqualification of individuals with NF from military service. He believes a fairer policy would be to make a case-by-case evaluation of individuals' abilities and limitations.

So, Stu pursued his lifelong interest in radio. He began his career in radio as an intern at a local station, working his way up from coffee gopher to the recording

booth. Then he landed a job as a DJ at WORG-FM, where he has been ever since.

Stu uses his radio presence as an opportunity to enlighten his listeners about NF. Stu tirelessly promotes NF awareness. He regularly plays NF public service announcements on his radio show and encourages people to distribute NF brochures, bookmarks, and other materials in local libraries, hospitals, and doctors' offices.

The radio station has also brought other rewarding experiences to Stu's life. Notably, he and his wife, Paulette, met at the radio station in the mid-1980s. They now have two sons, ages seven and twelve.

Outside of the radio arena, Stu is active in heightening awareness of NF. Stu learned of the NNFF twelve years ago and has become an active participant in the SC Chapter, and recently began serving as



Chapter Vice President. Stu also uses his office's proximity to the office of Congressman Floyd Spence (R-SC) to bolster support for the U.S. Army appropriation for NF research.

As a former professional truck driver and avid traveler, Stu dreams of taking his awareness efforts on a cross-country trek. He'd like to load up a rig with educational information and make stops to talk to people about NF.

Like many people, Stu also dreams of winning the lottery. But, Stu wouldn't use the proceeds on material goods. "I would devote my life to helping people with NF," he said.



NF2 gene product (merlin/schwannomin). The following researchers discussed their findings about the NF2 gene product: Dr. David Gutmann, Washington University; Dr. Vijaya Ramesh, Massachusetts General Hospital; Dr. Markku Sainio, University of Helsinki; Dr. Guy Rouleau, McGill University; Dr. Stefan Pulst, UCLA.

During the NF1 session, which was chaired by Dr. Bernardis, progress was reported in the areas of animal studies, the role and function of the NF1 gene product (neurofibromin), and the role of the NF1 gene in development.

Among the most important developments in the NF1 field was the news that Dr. Luis Parada of the University of Texas has developed a second-generation mouse model. "Dr. Parada has developed an elegant model that will enable scientists to better define neurofibromin's precise role in various cell types such as peripheral nerve cells," Dr. Bernardis said.

It has been known for several years that the role of neurofibromin inactivates the Ras protein, which serves as an on and off switch that controls cell growth and cell division. In a new twist to the tale of the NF1 protein and Ras relationship, Dr. Jacks reported findings that suggest the NF protein may actually play a dynamic role and promote Ras activity.

Dr. Iswar Hariharan of Massachusetts General Hospital reported on a fly model that yielded further evidence to support the theory that c-AMP is regulated by neurofibromin. "This finding is important because c-AMP is a molecule involved in learning and memory in various organisms," Dr. Bernardis noted.

Several researchers presented on growth factors in NF1: Dr. Kristine Vogel, Louisiana State University; Dr. George deVries, Loyola University; and Dr. Kevin Shannon, UCSF. Dr. Nancy Ratner, University of Cincinnati, reported on defective wound healing in

NF1 mutant mice. Dr. Wade Clapp, Indiana University, reported on a potential dominant effect of NF1 on coat color pigmentation in mice. Dr. Jeffrey DeClue, National Cancer Institute, discussed NF1 tumors and Schwann cell transformation. Dr. David Largaespada, University of Minnesota, and Dr. Camilynn Brannan, University of Florida, presented findings on myeloid leukemia. Dr. Jon Epstein, University of Pennsylvania, discussed NF1's impact on the neural crest and development of the heart. Dr. Mladen Golubic, Cleveland Clinic Foundation, presented findings on the neurofibromin's relationship with lipids and mitochondria.



DISCOVER CARD
Now Accepted for NNFF membership and donations

Pen Pals

- 44-year-old mom would love to hear from others with NF1. – Yvonne Smith; 125 Tenth St.; Baraboo, WI 53913
- Would like to correspond with other Nfers. – Shirley Yeazle; PO Box 181; Spooner, WI 54801
- My 6-year-old and I have NF1. Would like to share with someone. – Naomi; najewan@gbso.net
- 32-year-old with NF1 would like to correspond with others. – Melanie Grizzle; 899 Jordan Rd., Blairsville, GA 30512
- Mother with 2 children, both with NF, would like to correspond with somebody who's had experience. – Stacy Moinat; PO Box 2188; Alvin, TX 77512
- I am 11 years old and want to talk to others my age. – Terrdan1@aol.com
- Woman in Mexico looking for pen pals. – tamara_keller@hotmail.com or write Tamara Keller; Horacio 1805-1101; Los Morales Polanco; 11510 Mexico 5, DF
- My name is Malcolm Wiese, I am 21 and live in Australia. Would love to talk to others of any age who also have NF1. – wiese@hunterlink.net.au
- I live in France and have 9-year-old twins with NF1. One of them has pseudarthrosis of the cubitus, which is very rare. I would like to receive mail from parents or doctors who have knowledge of this and information about treatment. – Pascale Roulier; roulier@cybercable.fr
- 35-year-old mom and daughter with NF1 are looking for pen pals and friends. – Wynne Allen; 421-G Kenwood Dr., Euclid, OH 44123
- 21-year-old would like to hear from others. Will respond to all. – Shannon Murphy; YodaGirl11@hotmail.com
- I am a 47-year-old gay man with NF1. I would like correspond with other gay males and share experiences in dealing with signs of NF, such as explaining that it is not HIV-related. Please write. – carmine@gateway.net
- Lady in 50's would like to have others write. Let's become friends. – Carole Sergerson; PO Box 5442; Cheyenne, WY 82003
- 40-year-old with NF would like to share with others. – Debbie Flake; 995 S. Graham St., Martinsville, IN 46151
- My mother and I don't get a chance to meet or talk with many others about NF. I'm 26 and would like advice from others who have NF1 or children with it. – Trucompnin@aol.com
- Please write. – Terry L. Green; 5908 Bomar Dr., Cheyenne, WY 82009-2630 or tlgreen_98@yahoo.com
- I'm 34 years old and would like to correspond with others. I am the only one in my family and don't know anyone else with NF. – Robin Harper; 6824 44th Ave. East; Tacoma, WA 98443
- An adult with NF seeking friends. – Lavon Edwards; 1223 Maple St., Anderson, IN 46016

NF Marathon Team

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"My 5-year-old son Joshua has NF1," Carter said. "Many with NF suffer not only from the physical effects, but also the emotional toll created by people's uninformed reactions to those with NF. NF research can never move fast enough for me and that is why I started this program. I wanted to get people from all over the world to join my quest. The more people who participate, the more money we will generate. With all of us working together we can increase awareness which in turn will create greater understanding. I firmly believe that this program will be a major factor in helping to facilitate a treatment and possibly a cure one day."

Others share Carter's enthusiasm. In the words of Becky Deignan, Chicago Marathon NF Race for Research Coach: "The NF Marathon experience gave me so much more than I feel I put into it. I learned an incredible amount about hope, endurance, teamwork and sticking to a goal. Some do not see it as life-changing, but I do. I am changing not only my life, but truly changing the lives of those around me. How often can you say that? How often can you feel

noble and even justified in the feeling?"

If you would like information on how you can promote the NF Marathon Team program in your area, recruit participants and begin to take advantage of this great

opportunity to expand your fundraising, please feel free to call Anita Carter, at our NF Marathon Team Race Headquarters office: (toll-free) 877-NF-RACES (637-2237). Email: RaceForNF@aol.com.

NF Marathon Program Provides:

- Training Program • Training t-shirt • NF Marathon day singlet • Training Newsletter
- Nutrition information • Shoe clinics and shoe discounts • Fundraising training
- Monthly fundraising reports • Fundraising incentive prizes • Partnership with an NF Hero
- Round trip airfare to the Marathon • Hotel room, week-end of the Marathon
- Round trip ground transportation to airport, hotel, pasta dinner, Marathon
- Entry into Marathon and Pasta Dinner

October 3, 1999	Portland Marathon Portland, OR *	June 4, 2000	Rock 'N' Roll Marathon San Diego, CA
October 24, 1999	Chicago Marathon Chicago, IL *	June 17, 2000	Anchorage Marathon Anchorage, AK
January 9, 2000	Disneyworld Marathon Orlando, FL	October 23, 2000	Dublin Marathon Dublin, Ireland
March 19, 2000	Maui Marathon Maui, HI	December 10, 2000	Honolulu Marathon Honolulu, HI

* Due to the short amount of time remaining before the Portland and Chicago Marathons, it is recommended that only experienced marathoners and/or those currently in marathon training programs participate



NF Awareness in Iowa

Members of the NNFF Iowa Chapter gathered at the State Capitol in Des Moines to witness Governor Thomas Vilsack sign a proclamation declaring May as NF Awareness Month. Chapter President, Sheila Drevyanko, organized this and other activities to create awareness throughout the state. KCCI-TV aired a related story about NF and chapter members in Iowa City, Waterloo and Des Moines distributed NF educational materials to hospitals and doctors' offices.

INVESTING IN NF

PLANNED GIVING: DOING GOOD WHILE DOING WELL

Wouldn't it be great if you were able to help the causes you feel are important to support and to help yourself or your family at the same time?

We hope so, because giving generously doesn't mean you have to shortchange yourself or those you care about. Often it's possible to give in ways or at levels that may surprise you.

In the next few issues of the *Neurofibromatosis News* we'll be sharing information with you about ways that you can help yourself, your loved ones and the National Neurofibromatosis Foundation through "planned giving" or "gift planning."

If you would like more information about making a planned gift to the NNFF, please feel free to call our Development Department at 800-323-7938, extension 31.

"Planned giving" sounds difficult and can conjure up misconceptions in people's minds. Some think that it's only leaving a bequest gift in your will. While bequests are very easy and straightforward ways to make a planned gift, there are other possible gift vehicles that can do even more for you during your lifetime or for your estate after your death. There are ways that can allow you to see the benefits of your hard work and your generosity put to good use - now.

The best planned gifts fall within the context of a larger "estate plan" that looks at the entire spectrum of your goals and needs. They should also provide immediate, as well as long term financial benefits. Some think that estate planning is only for the very wealthy. Not true! Estate planning can be even more important for those that have modest assets. It starts with a signed, legal will that is appropriate for the state you live in, and should include considerations for your financial needs as well as your charitable intentions.

Planned gifts can help your estate planning by providing:

- Current income tax deductions;
- Avoidance of or minimization of long term capital gains;
- Increases in income and effective rates of return on your assets;
- Ability to give potentially more to your heirs and your favorite causes, without decreasing the size of your estate.

We understand that your personal financial goals and needs will, and should, come first. It is important that you seek the advice of your accountant or financial planner and attorney, regarding the details of making your estate plan work for you – the way you want it to.

The beauty of planned giving is that it allows you to do exactly that. We can help you to provide for your loved ones now and in the future, as well as help the Foundation achieve its goals of finding a cure and treatment for neurofibromatosis, expanding our patient services and educating the public and professionals about NF.

To add the Foundation as a bequest beneficiary in your will, add this codicil wording to your existing will. Please be sure to check with your attorney for any specifics, which may apply in your state.

"...I hereby give, devise and bequeath to The National Neurofibromatosis Foundation, Inc., a 501(c)3 not-for-profit organization, incorporated under the laws of the State of New York in 1978, having its principal address at 95 Pine Street, 16th Floor, New York, NY 10005,"


For gifts of cash or securities include the following...

..."[the sum of (\$ amount) or (% of total estate) or (# of shares of XXX corporation)]."

For gifts of real property...

..."[my real property located at (property address)]."

Please note: Through an anonymous donor,

any bequest made to the Foundation will result in an additional \$500 gift being made to the Foundation. Should you decide to include The National Neurofibromatosis Foundation in your estate, please notify our Development Office in writing so that we may process this generous additional gift. 

WAYS TO GIVE

By way of introduction to planned giving, the following is a partial list of gift vehicles which will be addressed in more detail, through future articles in this newsletter.

Bequests – Gifts from your estate, given through your will, that take effect upon your death. Allows a large gift to be made that does not have an impact on current assets or income;

Living Trusts – Revocable gifts that can be changed or updated to meet your needs over time;

Gift Annuities – Gifts that provide a fixed income for life to you or another beneficiary, with the assets going to NNFF later;

Insurance – Current policies can be transferred to the NNFF or have the NNFF named as the beneficiary;

Pooled income funds – Functions like a mutual fund and returns a dividend to you, with the assets going to NNFF later;

Deferred Gift Annuities – Functions like a retirement account, with a deferred payment coming to you later in life;

Remainder Trusts – Gifts that can provide either a fixed or variable annual income to you for life, with the assets going to NNFF later;

Lead Trusts – Provides annual income to NNFF through use of your assets now, while returning the assets to your estate/heirs later;

Real Estate – A range of gift options exist here, from outright donations to retained life gifts which allow you to live in or use property now and donate it later;

Appreciated Assets or Securities – Highly appreciated, low yielding stocks or other assets can be given to NNFF as outright gifts or used to fund various types of trusts. This can provide a greater tax advantage than selling them and then donating the proceeds.

Research Awards

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and her assistants will determine whether IQ improves with age as abnormalities seen on brain imaging tests resolve. They will also explore the controversial relationship between development abnormalities detected on brain imaging and lowering of IQ. In addition, they will examine the affect of NF on self-esteem and behavior of the child with NF1. The results of this research will provide an insight into the cause of learning disabilities in NF1, as well as providing baseline information that is essential for the development of therapies.

The Marcy and Richard Horvitz Family Fund


Dr. Jerome Ricard

University of Central Florida

Two-year Young Investigator Award

Project Title: "Study of Merlin and Paxillin Interactions Role in Tumor Suppressor Function of Merlin"

For the past 8 years, Dr. Ricard's laboratory has been studying the signaling pathways by which integrins, which regulate cell shape, movement and division, direct Schwann cells to stop divi-

sion and commence myelin formation. (Myelin is the insulating sheath around nerves.) In doing so, they have identified an important pathway by which the NF2 gene product may act to prevent uncontrolled Schwann cell growth. This proposal will continue to study how the NF2 gene product is involved in sending signals from integrin receptors. The results are expected to allow the development of rational approaches aimed at reversing the loss of cell growth control caused by NF2 mutations. 

Testing

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been done, testing of a pregnancy may be accomplished by amniocentesis or by chorionic villus sampling.

Q. What are the tests' limitations? Why?

A. Current techniques for mutational analysis are able to detect alterations in only 66% of persons known to have NF2. For the 1/3 of families in which the mutation cannot be detected, we cannot offer testing at this time.

Q. What are the common misconceptions about these tests?

A. The most common misconception is that the test is somehow better than clinical assessment. At the current time, no simple blood test exists that can substitute for evaluation by an experienced center.

Q. What new tests are on the horizon? What would the ideal test be?

A. The ideal test would be rapid, inexpensive and highly accurate, even for someone who is only under suspicion for NF2. This is a very tall order, but under a grant from the US Army we are trying to develop methods that will allow us to approach this ideal.

Q. Where does someone go to be tested?

A. People should call the National Neurofi-

bromatosis Foundation (800-323-7938) for the closest NF clinic or genetics center.

Q. What can someone expect of the testing process?

A. Here at Massachusetts General Hospital, the testing process begins with a visit to go over the ramifications of testing and to be sure that an individual is not already overtly symptomatic. For individuals who decide to follow through with the testing process, we set an appointment to discuss the test results and advise then to come with a trusted friend or family member. If the results are negative, the individual receives a copy of the report and a follow-up phone call at a later date. If the results are positive, the individual is immediately scheduled for a MRI and other tests.

Q. What does it cost? Covered by insurance?

A. The test itself costs between \$1400 and \$2000 for the first family member to be studied, and \$200 to \$500 for subsequent family members. If your family has previously participated in genetic research studies and your family's mutation has been identified the cost may be much lower. Additional costs are those of clinic visits for counseling

and to receive results. The laboratory which performs the test will not accept insurance, but will try to help families get reimbursement from their insurance companies.



neuro·fibroma·tosis
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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.



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