

neuro·fibroma·tosis®

THE NATIONAL NEUROFIBROMATOSIS FOUNDATION, INC.

ANNUAL REPORT 1999

Connections



The National Neurofibromatosis Foundation, Inc.

FOUNDED

In 1978 by Allen E. Rubenstein, M.D., Lynne Courtemanche Shapiro, R.N. and Joel Hirschtritt, Esq.

PURPOSE

- To sponsor scientific research aimed at finding the cause and cure for both types of neurofibromatosis, NF1 and NF2;
- To promote the development of clinical activities, which assure individuals with neurofibromatosis access to the highest caliber of medical care;
- To develop programs that will increase public awareness of neurofibromatosis; and
- To provide support services for patients and families with accurate and comprehensive information about neurofibromatosis, with support group activities and referrals to qualified physicians and healthcare professionals.

MEMBERSHIP

Approximately 50,000 associates, of which approximately 14,000 are contributing members in all 50 of the United States and 51 other countries.

CHAPTERS

NNFF's 34 Chapters are central to the mission of the organization. Staffed by dedicated and trained volunteers and in some cases by professional managers, the Chapters provide grassroots resources and patient support in communities throughout the U.S. In addition, they carry out local public education campaigns and conduct fundraising activities that make a vital contribution to funding the national research efforts. NNFF Chapters are located in the following states and regions:

Alabama, California, Colorado, Connecticut, Florida, Georgia, Hawaii, Idaho, Illinois, Indiana, Iowa, Kentucky, Louisiana, Massachusetts, Michigan, Mid-Atlantic (Virginia, Maryland, Washington, DC), Mississippi, Missouri, Nevada, New York/New Jersey, Northern New England (Maine, New Hampshire, Vermont), Northern Plains (Nebraska, North Dakota, South Dakota), Ohio, Oklahoma, Oregon, Pennsylvania, Rhode Island, South Carolina, Tennessee, Utah, Washington, West Virginia, Wisconsin, Wyoming.

INTERNATIONAL AFFILIATES

63 affiliates in 32 countries:

Argentina, Austria, Australia, Belgium, Brazil, Bulgaria, Canada, Colombia, Czech Republic, Denmark, Egypt, Finland, France, Germany, Hong Kong, Hungary, India, Ireland, Israel, Italy, Japan, New Zealand, Norway, Puerto Rico, Russia, South Africa, Spain, Sweden, Switzerland, The Netherlands, Turkey, Uruguay, Ukraine, United Kingdom.

Cover: **NF Connections**

Research: **Dr. Mia MacCollin (right) and student**

Internet: **Jacqueline Media**

People: **children at the NNFF International Summer Camp for Teens**

Community: **Rebecca Stolzenburg, Julie Niemira**

standing: **Dennis Chipollini**

CHAIRMAN

Bruce Judson, Esq.
Carmel, NY

PRESIDENT

Peter R.W. Bellermann
South Orange, New Jersey

NNFF STAFF

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Andrew Phillips, Vice President of Development
Jacqueline Media, Director of Programs/Administration
Erica Mayer, Director of Public Education
Lily Loreda, Comptroller
Magda Samborska-Murgio,
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E-mail: NNFFWA@aol.com

NF Connections



Bringing together the Entire Spectrum of the NF Community

Since its founding in 1978, the NNFF has been connecting researchers, clinicians and lay people in a variety of ways, all in an effort to improve the health and well-being of individuals and families with NF.

Connecting the Scientific Community

Our medical symposia are more than opportunities for scientists to share information; they are venues where new research collaborations are formed in the interest of furthering the knowledge of NF1 and NF2. The NNFF sponsors research, helps in the development of NF clinics and works with lawmakers on Capitol Hill to secure funding for NF research each year.

Connecting People

Our network of chapters and volunteers across the country carry out the work of the Foundation at the grassroots level. Providing patients and families with information, support, and referral to clinics, the NNFF is dedicated to improving the quality of life for persons with NF and raising public awareness of NF among the general population.

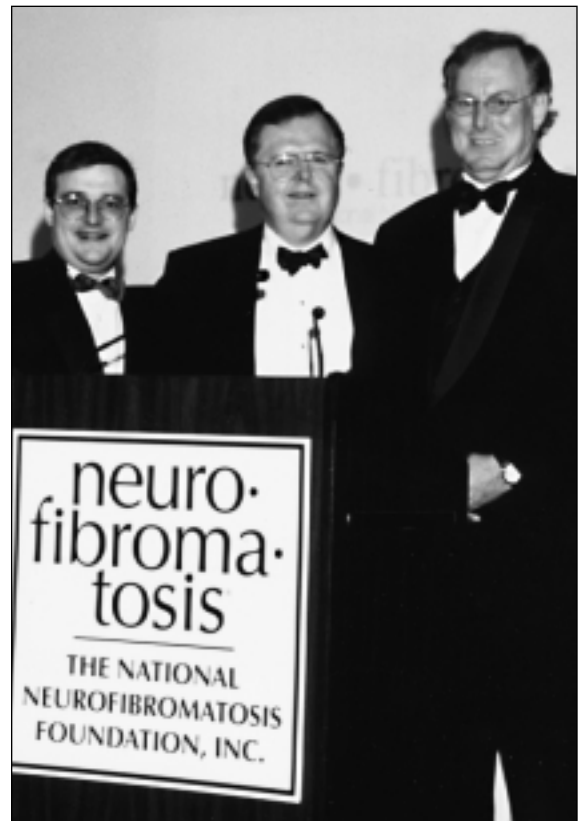
Connecting Through Technology

The NF Website provides people with up-to-date information on NF, current research and chapter/support group activities in their area. The NF chat rooms allow people to talk *live* with others about issues relating to NF. The Clinicians' Forum is a tool for clinicians to get advice and medical counsel from their peers. The NF Website keeps people connected by constantly adding features to meet the needs of our members, affected individuals and families, scientists, clinicians,

other healthcare providers, donors, the media, educators and the general public. We have created an on-line environment where people feel a part of the NF community and learn more about NF and the work of the Foundation.

During 1999 the NNFF continued to make connections all around the globe. The Foundation has helped to make great strides toward achieving its mission – finding effective treatments and a cure for NF1 and NF2. With each connection we make, we are one step closer to our goal.

(left to right):
Bruce Judson, Esq., Chairman, Peter R.W. Bellermann, President,
Allan E. Rubenstein, M.D., Medical Director



Internet Connections



Website offers new ways to communicate for patients and families

As the 20th Century came to a close, the Internet emerged as a powerful force in communications. More and more people are going on-line for their medical information and patient support. The Foundation's NF Website (www.nf.org) has become a powerful source of information and support for affiliated individuals, their families and medical professionals. Improvements to our website over the past year now enable people to keep up-to-date on the latest medical and research information, join a community that offers support and encouragement and allows them to contribute to the research effort no matter what their financial situation may be.

Connect with People Anywhere, Anytime!

Now people from all over the world can log on to the NF Website's chat rooms to discuss issues related to NF. Our live, interactive chat rooms are open 24 hours a day/7 days a week. Four different chat rooms are available – a general chat that is open to the public, the Kids Council chat room and Camp chat room are restricted to kids only, and the chat room for college students. Since the chat rooms opened in November 1999, more than 1,000 people have joined our on-line community and that number is growing every day!

Worldwide NF Clinicians Forum

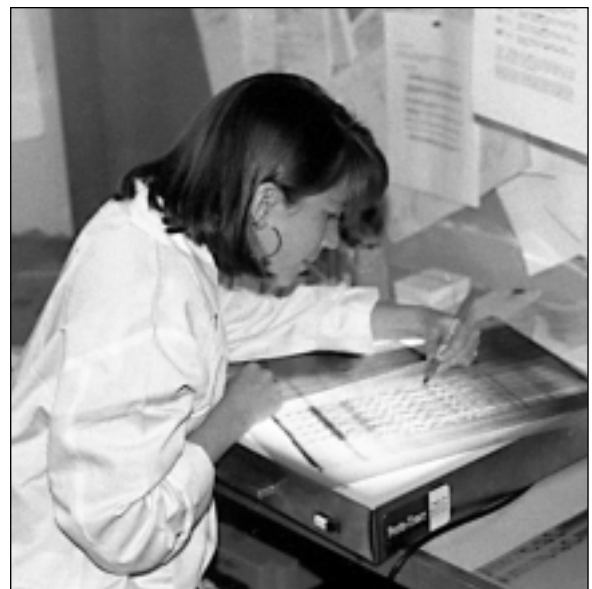
Since its launch in 1998, the Clinician's Forum (a free Internet-based service exclusively for health care professionals) has helped to raise the quality of care for patients around the world. Health professionals can post difficult case histories or pose diagnostic and/or disease management questions to NF experts.

The site is password protected to ensure that only qualified professionals are part of the discussions. The on-going dialogues remain on-line as a resource for future visitors.

NNFF International Database

The NNFF International Database is a confidential system for collecting medical, demographic and other relevant information about individuals and families with NF. The Database is a vital research tool as the data is studied to explore patterns that may emerge regarding the natural course of NF. NF clinics throughout the world regularly enter information into the Database. In 1999, data from this Database was analyzed and identified the need for further exploration in the bone abnormalities that sometimes accompany NF1 (more information on the Task Force Committee on Bone Abnormalities please

Dr. Andrea McClatchey of Massachusetts General Hospital



see [page 5](#)). The Database is funded by the NNFF and maintained by Jan M. Friedman, M.D., Ph.D. and Patricia H. Birch, M.Sc., R.N. of the Department of Medical Genetics, University of British Columbia, Vancouver, Canada. For a list of the members of the NNFF International Database, see [page 13](#).



The Gift That Keeps on Giving

On-line shopping is becoming an increasingly popular activity on the web. Realizing this, the NNFF wanted to provide their on-line community with a way to shop on-line while contributing to the Foundation at the same time. The Shop2Give program provides our on-line community with reputable on-line merchandisers who donate a percentage of each on-line purchase back to the Foundation. This is a win-win situation for all. Shoppers are not charged an additional fee for this service and they are able to donate money for NF research without having to set aside extra money from their pockets to do so! Shop2Give offers top retailers such as 1-800 Flowers, Borders Books and Music, J.C. Penney, and Office Max. Recently, Amazon.com was added to our list of on-line vendors who are giving a percentage of each purchase made through our website back to the Foundation.

A Bear For All Seasons

The NF E-Bear is a popular feature on our site. These cuddly teddy bears make a great gift and are a great way to cheer up a friend. The E-Bear has become the NNFF's official mascot. Look for him to become a more prominent and animated feature on the site that will help guide people to the information and resources they are looking for.

The NF E-Bear



Research Connections

Dr. Stuart Aaronson,
Mt. Sinai Medical Center



Connecting Research Ideas and Institutions

The mission of the Foundation is to drive research toward effective treatments and a cure for NF. We work toward this goal by funding research, bringing researchers who normally would be competitors into collaborations, hosting scientific symposia and bringing together the top minds in research to set goals and research priorities.

Task Force Committees

The NNFF Task Force on Treatment of NF1 was created in 1995 to consider the need for additional information on the natural history of the disorder and to recommend directions for development of models for experimental therapeutics for NF1. As part of the Foundation's on-going effort to advance NF research, two new Task Force committees were created in 1999—The Task Force Committee on Bone Abnormalities and NF1 and the NF1 Cardiovascular Task Force Committee.

NNFF-Sponsored Medical Symposia and Research Findings

1999 was an important year for NF research. From laying the groundwork for clinical trials to further unraveling the workings of the NF1 and NF2 genes, NNFF-sponsored research has helped to move us closer toward our goal of effective treatments and a cure for NF.

The first International Natural History Study of Plexiform Neurofibromas in NF1 meeting was held this year. The meeting was designed to establish normative data for plexiform neurofibromas and to create an infrastructure for clinical trials. The meeting was judged by all who attended to be a

successful beginning to one of the most significant current undertakings in neurofibromatosis research.

The NF gene is implicated in the development of several common cancers (breast cancer, colon cancer, and melanoma) so much of the promising research avenues in cancer treatment are relevant to NF. The annual meeting of the American Association for Cancer Research included several presentations of NNFF-sponsored research studies. Some of the most compelling presentations included a study of an angiogenesis inhibitor that caused tumor regression in mice implanted with neurogenic sarcomas from patients with NF1; information about three drugs that are approaching, or are already in, clinical trials for NF1-related tumors; a study elucidat-

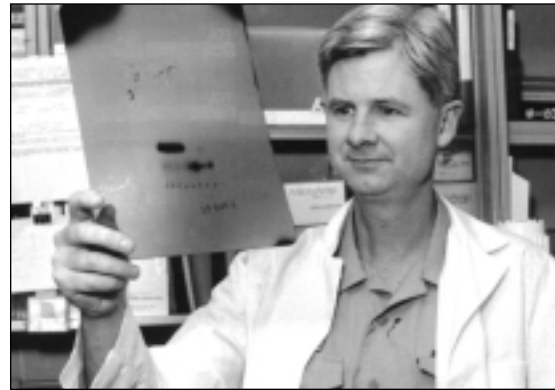
Dr. Margaret Wallace of the University of
Florida Health Science Center



ing the mechanisms by which NF1 gene mutations disrupt cell growth control which may lead to better treatments for brain tumors associated with neurofibromas; and a transgenic mouse model of NF1 that should provide a better model for the human disease.

One way the NNFF manages research in the molecular biology of NF is to host the NNFF International Consortium for Molecular Biology of NF1 and NF2 so that researchers can share their up-to-the-minute findings with their colleagues and form collaborative, multi-laboratory projects. At this year's meeting, held at MIT's Whitehead Institute for Biomedical Research, researchers agreed that NF2 research appears to be moving more rapidly than expected. NF2 researchers noted progress on three fronts—biochemical studies, regulation of and by Merlin (NF2 gene product), and modeling of NF2 in animals. One of the most significant discoveries reported on at the meeting was the first 3-D model of ERM proteins, which interact with Merlin. This breakthrough gives a better sense of what Merlin's structure may look like and that will help in understanding Merlin's function.

On the International front, NNFF hosts meetings with researchers from around the world to share information and create new collaborations. At the 8th Annual European meeting of scientists, clinicians, and lay organizations held in Ulm, Germany researchers discussed their latest findings. A promising new approach to identifying NF gene mutations was presented along with a study from the University of Mainz, which appears to be the first molecular approach to skeletal abnormalities in NF1.



*Dr. Kevin Shannon, M.D.,
of the University of California, San Francisco*

NNFF 1999 Research Awards

A critical component of the Foundation's programs are the Research Awards and Young Investigator Awards, and Contracts. Applications for these awards are submitted from researchers all over the world. The Research Advisory Board (RAB) chooses the awards through a rigorous peer review process.

New Research Awards:

The Role of Pak Protein Kinases in NF
Dr. Jeffrey Field
University of Pennsylvania
One Year Research Award

Molecular and Functional Interactions of NF1
Dr. Yi-Ping Hsueh
Massachusetts General Hospital
Two-Year Young Investigator Award

Profile of the NF1 Phenotype: Natural History, Neuropsychological and Psychological Aspects
Dr. Kathryn North
New Children's Hospital, NSW, Australia
One-Year Research Award

Study of Merlin and Paxillin Interactions Role in Tumor Suppressor Function of Merlin
Dr. Jerome Ricard
University of Central Florida
Two-Year Young Investigator Award

Clinical Care Advisory Board

The NNFF Clinical Care Advisory Board (CCAB) establishes criteria for comprehensive approaches to the diagnosis and management of persons with NF1 and NF2. This year new members were added to the CCAB with expertise in learning disabilities, pediatric oncology, and medical and consumer education.

NF Clinics

Since NF is such a highly variable disorder and can often run an unpredictable course, the NNFF encourages all persons with NF to receive regular medical care. The Foundation refers to more than 130 NF clinics in the United States and abroad for management of their disorder. Furthermore, the NNFF is instrumental in assisting in the development of NF clinics throughout the world.

Attracting Young Stars to the Field

For the first time in its history the NNFF, in cooperation with Tim Jackson of QXL.com, offered \$20,000 in prizes for research ideas in NF1 or NF2 submitted by graduate students. The purpose of the "NF Prize for Research Ideas" is to stimulate interest among young scientists in this important yet under-researched field. Entries were submitted from the top research institutions in the US and abroad. The winner will be selected by a panel of NF experts and announced in March, 2000.

\$3.5 Million Increase in US Army Funding

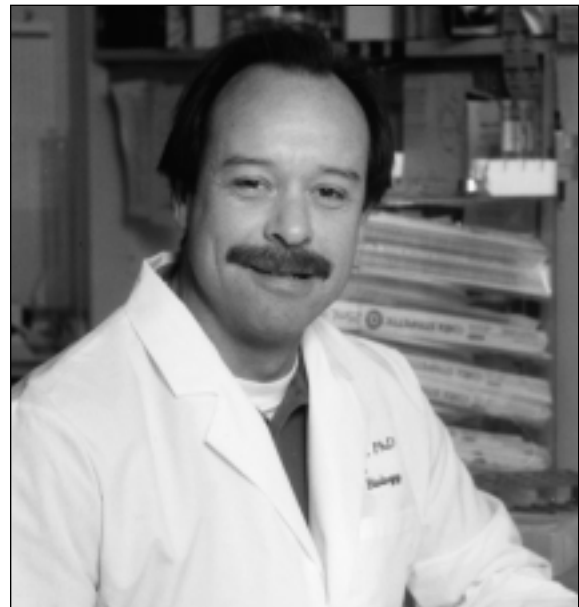
The US Army NF Research Program, that began as an initiative by the Foundation and continues to be a high priority, allocated

\$15 million towards NF for the coming year. This was an increase of \$3.5 million over the previous year. In 1999 more than 12,000 of the Foundation's members were involved in the legislative process which led to this significant result. The Army's NF Research Program, peer review-based and open to all scientists worldwide, now represents the largest source of funding for NF research.

Research Advisory Board

The NNFF Research Advisory Board (RAB) is the peer review panel for the annual review of grant applications. The RAB includes the top researchers in the fields relevant to NF1 and NF2. The RAB also provides the overall direction for other research activities sponsored by the Foundation. For a list of RAB members please see [page 13](#).

*Luis F. Parada, Ph.D.,
University of Texas, Southwestern Medical Center*



People Connections

Dennis Chipollini runs for the
NF Marathon Team



Raising Public Awareness

Although NF is the most common neurological disorder caused by a single gene and is more prevalent than Cystic Fibrosis, Huntington's Disease and Tay Sachs Disease *combined*, the general public still knows little about the disorder. Through the Foundation's public education efforts, more people are learning about NF, and turning to the Foundation as a resource.

Public Education

The Foundation provides information to the public in several ways. Our website provides up-to-date information on research and Foundation activities, our publications are disseminated to volunteers, schools, medical professionals and the media and our toll-free number allows people from all over the country to call the Foundation with specific questions or requests for referrals.

Media Outreach

An important audience to educate about NF is the media. By creating educational materials and conducting interviews with reporters, we ensure that the correct information is being relayed to the public. This year many news stories about NF have appeared in newspapers and on TV news stations nationwide.

NF Marathon Team

In addition to raising funds for NF research, the NF Marathon team provides an opportunity to raise public awareness. People of all ages and situations have joined the team. Some are running in honor of an NF hero and some are NF heroes themselves.

The Foundation has been able to generate media coverage of team members in major news markets such as Chicago, Denver, Orlando and Philadelphia. ■

Rebecca Stolzenburg (left) – the main person who put the Walk-a-thon together! Julie Niemira (right), Rebecca's helper.



The Wigmaster helps put the "freeze" on NF in Connecticut.

Community Connections

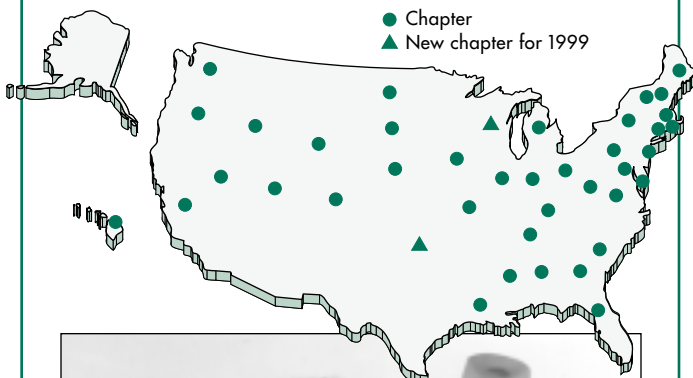


Connecting with the NF Community

The Foundation provides several ways that people with NF can connect with each other to provide support, share information and build an international community for patients and families to feel a part of.

Growing Our Chapters Nationwide

The NNFF welcomed two new chapters during 1999 — Oklahoma and Wisconsin. Our chapters provide local resources and patient support in the form of support group meetings, fundraising activities and public awareness programs. The Chapters are staffed by dedicated and trained volunteers or professional managers who have the shared goal of supporting people with NF while making a contribution through their work at the grassroots level toward funding national research activities.



Monty Roberts, "The Horse Whisperer," hosts a benefit for the Iowa Chapter.

Photo: © Christopher Dydak 1998



Members of NF Europe



International Patient Organizations

People with NF from all over the world need information and access to resources from the NNFF. To this end the Foundation directs many of these inquiries to its affiliated patient organizations in many different countries. These organizations act as an excellent resource for patients and their families for information, support and referrals to local NF experts.

In 1999, the pan-European movement took another step forward with the formation of *NF Europe* which consists of most of the NF groups on the European continent.

"The more we unite and pool our resources, the stronger each of our organizations will become, and the greater our opportunities to find effective treatments that will bring NF under control."

Peter Bellermann

Chairman of the International NF Association.

International Summer Camp for Teens

Campers came from as far away as Alaska, Belgium and Japan for the third annual International NNFF Summer Camp in Flagstaff, AZ. Adolescence can be a difficult time for any child, but a child with NF faces particular challenges. For some campers the camp was their first opportunity to meet other children with NF. For others it was the chance to renew old friendships and form new ones as they rafted down the Colorado River, played games at Camp K, and spent the day learning about genetics from an NF researcher at the Eccles Genetics Center at the University of Utah. The camp has received such a great response that an extra session has been added for the summer of 2000.

NNFF Campers rafting down the Colorado River



"I'm already counting the days until the next camp."
Camper from California



NNFF Campers become scientists for a day at the Day at the Lab Program at University of Utah.

Kids Council

Given the emotional toll of the disorder and the self-esteem problems it can create, the goal of the Council is to instill confidence and a sense of accomplishment. The Council is entirely governed by the kids (ages 6 to 17) and they produce a successful newsletter and a pen pal program! ■



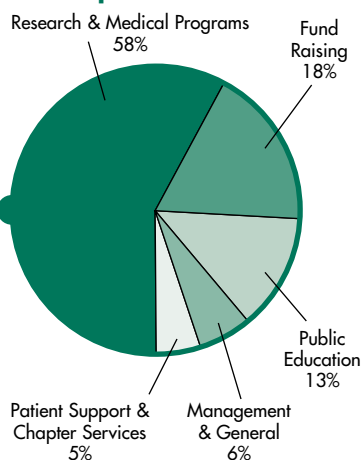
Financials



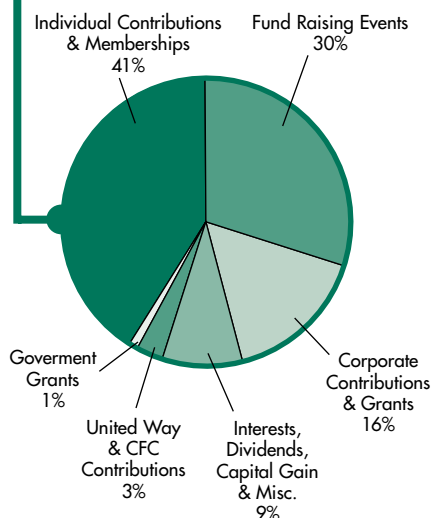
BALANCE SHEET

**NATIONAL NEUROFIBROMATOSIS FOUNDATION, INC.
STATEMENT OF FINANCIAL POSITION
SEPTEMBER 30, 1999 and 1998**

Expenditures*



Revenues



ASSETS

	1999	1998
Current Assets		
Cash and Cash Equivalents	\$ 190,900	\$ 271,566
Pledges and Other Receivables	923,599	431,573
Prepaid and Deferred Expenses	75,479	47,235
Total Current Assets	1,189,978	750,374
Furniture and Equipment, Net	86,552	110,722
Other Assets		
Investments	3,183,383	3,122,429
Pledges Receivable, Long-Term	50,000	200,000
Security Deposit	16,980	16,980
Total Other Assets	3,250,363	3,339,409
	\$4,526,893	\$4,200,505

LIABILITIES AND NET ASSETS

Current Liabilities		
Accounts Payable	\$ 34,984	\$ 74,614
Deferred Revenue	90,927	37,500
Deferred Rent Obligation, Current	2,717	2,717
Research Grants Payable	24,667	110,750
Accrued Vacation	10,505	10,302
Total Current Liabilities	163,800	235,883
Other Liabilities		
Deferred Rent Obligation, Long Term	24,450	27,166
Assets Held in Trust	173,687	184,356
Total Other Liabilities	198,137	211,522
Total Liabilities	361,937	447,405
Net Assets		
Unrestricted		
General Operating	193,006	187,429
Designated:		
Board Restricted	2,573,570	2,423,570
Operating Contingency	324,895	474,895
Total Unrestricted	3,091,471	3,085,894
Temporarily Restricted	1,073,485	667,206
Total Net Assets	4,164,956	3,753,100
	\$ 4,526,893	\$ 4,200,505

For a complete copy of the Foundation's certified financial statements, please send request to the National Neurofibromatosis Foundation, 95 Pine Street, New York, NY 10005

* exclusive of chapters

STATEMENT OF ACTIVITIES

NATIONAL NEUROFIBROMATOSIS FOUNDATION, INC. FOR THE YEAR ENDED SEPTEMBER 30, 1999 WITH COMPARATIVE TOTALS FOR 1998

	Unrestricted	Temporarily Restricted	Total 1999	Total 1998
Public Support and Revenue				
Public Support: and Reclassifications:				
Individual Contributions and Member Dues	\$583,943	\$846,503	\$1,430,446	\$649,037
Estate Bequest	—	—	—	2,120,965
Corporate Contributions and Grants	230,211	335,604	565,815	711,232
Government Grants	—	30,000	30,000	—
Fundraising Events	1,029,478	—	1,029,478	1,038,990
United Way and CFC Contributions	102,783	—	102,783	90,077
Net Assets Released from Restrictions	844,643	(844,643)	—	—
Total Public Support and Reclassifications	2,791,058	367,464	3,158,522	4,610,301
Revenue:				
Loss on Sale of Investments	(42,357)	—	(42,357)	65,421
Unrealized Holding Gain on Investments	126,258	—	126,258	2,625
Interest and Dividends	171,928	—	171,928	76,636
Publications and Miscellaneous	—	38,815	38,815	30,414
Total Revenue	255,829	38,815	294,644	175,096
Total Public Support, Reclassifications and Revenue	3,046,887	406,279	3,453,166	4,785,397
Expenses:				
National Program Services				
Research and Medical Programs	1,329,227	—	1,329,227	1,357,209
Public Education	308,234	—	308,234	202,959
Chapter Services	115,650	—	115,650	99,201
Total National Program Services	1,753,111	—	1,753,111	1,659,369
Chapter Program Services				
Research and Medical Programs	2,921	—	2,921	2,260
Public Education	233,476	—	233,476	225,931
Total Chapter Programs Services	236,397	—	236,397	228,191
Total Program Services	1,989,508	—	1,989,508	1,887,560
National Supporting Services				
Management and General	138,286	—	138,286	119,882
Fund Raising	420,340	—	420,340	287,561
Total National Supporting Services	558,626	—	558,626	407,443
Chapter Supporting Services				
Management and General	78,099	—	78,099	72,259
Fund Raising	415,077	—	415,077	469,264
Total Chapter Supporting Services	493,176	—	493,176	541,523
Total Supporting Services	1,051,802	—	1,051,802	948,966
Total Expenses	3,041,310	—	3,041,310	2,836,526
Increase in Net Assets	5,577 ¹	406,279	411,856¹	1,948,871 ²
Net Assets, Beginning of Year	3,085,894	667,206	3,753,100	1,804,229
Net Assets, End of Year	\$3,091,471	\$1,073,485	\$4,164,956	\$3,753,100

¹ Represents revenues raised for programs and activities for 2000

² Represents revenues raised for programs and activities for 1999.

Research Advisory Board

From page 7

CO-CHAIRMEN

James Gusella, Ph.D.
Harvard Medical School/Massachusetts General Hospital

Tyler Jacks, Ph.D.
MIT Center for Cancer Research/
Howard Hughes Medical Institute

MEMBERS

Stuart Aaronson, M.D.
Mount Sinai School of Medicine

Andre Bernards, Ph.D.
Harvard Medical School/
Massachusetts General Hospital

Anthony Bretscher, Ph.D.
Cornell University

John C. Carey, M.D.
University of Utah

Webster K. Cavenee, Ph.D.
Ludwig Institute for Cancer Research

Neal Copeland, Ph.D.
National Cancer Institute

Sara Courtneidge, Ph.D.
Sugen, Inc.

David Cox, Ph.D.
Stanford University

Channing Der, Ph.D.
University of North Carolina

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Hines VA Hospital

Beverly Emanuel, Ph.D.
University of Pennsylvania

Kenneth Fischbeck, Ph.D.
University of Pennsylvania

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Merck & Co./University of Pennsylvania

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University of British Columbia, Canada

Zach Hall, Ph.D.
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Massachusetts Institute of Technology

Tony Hunter, Ph.D.
The Salk Institute

William G. Johnson, M.D.
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Medical School

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Dana Farber Cancer Institute

Nikolai Kley, Ph.D.
Genome Therapeutics

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Boston Children's Hospital

David Louis, M.D.
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Massachusetts General Hospital

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Massachusetts General Hospital

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Pfizer Central Research

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University of Oklahoma

Roger Packer, M.D.
Children's National Medical Center

Luis Parada, Ph.D.
UT Southwestern Medical Center

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University of Pennsylvania

Nancy Ratner, Ph.D.
University of Cincinnati

Guy Rouleau, M.D., Ph.D.
McGill University, Canada

Allan E. Rubenstein, M.D.
Mount Sinai School of Medicine

J. Lynn Rutkowski, Ph.D.
University of Pennsylvania

Bernd Seizinger, Ph.D.
Genome Pharmaceuticals, Germany

Kevin Shannon, M.D.
University of California, San Francisco

Ira Shoulson, M.D.
University of Rochester

David Viskochil, M.D., Ph.D.
University of Utah

Margaret Wallace, Ph.D.
University of Florida

James Weston, Ph.D.
University of Oregon

Ray White, Ph.D.
University of Utah/
Howard Hughes Medical Institute

Berton Zbar, M.D.
National Cancer Institute

The NIFF International Database

From pages 3-4

Dr. Nikolay Bochkov
Moscow Medical Academy, Russia

Dr. Jose Maria de Campos
Fundacion Jimenez Diaz,
Madrid, Spain

Dr. John C. Carey
University of Utah

Dr. Alison Colley
Newcastle Western Hospital,
New South Wales, Australia

Dr. Adrian Danek
Neurologische Universitätsklinik,
Munich, Germany

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The Neurofibromatosis Symposium

Sponsored By The National Neurofibromatosis Foundation Held In Conjunction With The 49th Annual Meeting of The American Society of Human Genetics
October 19, 1999

CHAIRMAN:

Bruce Korf, MD
Harvard Medical School/Partners

Mutation analysis of the NF1 gene in 62 patients using an optimized protein truncation test (PTT), heteroduplex analysis (HA) and karyotyping allows
Ludwine Messiaen, Ph.D.
University Hospital Gent
Belgium

Spectrum of minor lesion NF1 gene mutations in 521 unrelated German in NF1 patients.

Peter Nurnberg, Ph.D.
Charite University Hospital, Germany

Illegitimate splicing of 5 of 7 Investigated in-frame Exons of the NF1 Gene.

Dieter Kaufmann, MD
University of Ulm, Germany

Application of bisulphite genomic modification to determine the methylation status of a 1.2 kb region of neurofibromatosis type 1 (NF1) promoter sequence in benign neurofibromas from 10 unrelated NF1 patients.

Martin Horan, MD
University Wales College of Medicine, UK

Premature osteoporosis in NF1

Cynthia Tiff, MD, Ph.D.
Children's National Med. Ctr.
Washington, DC

8:03-8:05PM Q&A

NF1 and overgrowth syndrome resembling Weaver syndrome in 3 sporadic patients.

Anne Moncla, MD
Hopital d'Enfants de la Timone, France

Age-related features of genetic mutations producing cognitive impairment: the fragile X mutation, NF1, and Williams Syndrome.

Gene Fisch, Ph.D.
Yale University School of Medicine

Cardiovascular malformations and other cardiac abnormalities in Neurofibromatosis 1.

Angela E. Lin, MD
Harvard Medical School

Clinical correlates of somatic mosaicism in NF2

Michael Baser, MD, Ph.D.
Los Angeles, CA

Three novel mutations in the NF2 tumor suppressor gene.

Cesar Paz-y-Mino Sr., MD
Ecuadorian Catholic University, Ecuador

Novel alternatively spliced isoforms of the NF 2 tumor suppressor demonstrate tissue-specific expression pattern and distinct subcellular localization.

Beatrice Schmucker, MD, Ph.D.
University Erlangen, Germany

Germ-line NF2 mutation type, location, and phenotype in Neurofibromatosis 2

Michael Baser, MD, Ph.D.
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Predictors of survival in neurofibromatosis 2

Michael Baser, MD, Ph.D.
Los Angeles, CA

The NNFF International Consortium for the Molecular Biology of NF1 and NF2

June 20-22, 1999
Boston, Massachusetts

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KEY NOTE SPEAKERS

"Ras-mediated signaling in disease and development"

Channing Der
University of North Carolina

"Cell signaling and the cytoskeleton"

Alan Hall
University College of London

NF1 Morning Session: André Bernards, PhD (Chair):

"EGF receptor expression in NF1 tumors and Schwann cell transformation"

Jeffrey DeClue
National Cancer Institute

"F1 +/- mice are defective in wound healing"

Nancy Ratner
University of Cincinnati

"Biological studies of Drosophila NF1"

Iswar Hariharan

Yi Zhong
Cold Spring Harbor Laboratory

"Growth factor modulation of neural crest phenotype in Neurofibromin-deficient cells"

Kristine Vogel
Louisiana State University

Altered growth factor receptors in NF1-derived Schwann cells

George de Vries
Loyola University

"Regulation of and by neurofibromin"

Tyler Jacks

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NF1 Afternoon Session: Luis Parada, U.T. Southwestern Medical Center (Chair)

"NF1 conditional knockout: a progress report"

Luis Parada (Chair)
U.T. Southwestern Medical Center

"GAPs and GEFs in myeloid leukemia development"

David Largaespada
University of Minnesota

"In vivo relevance of growth factor hypersensitivity in NF1-deficient hematopoietic cells"

Kevin Shannon
UCSF

"Identification of a candidate cooperating oncogene in progression of NF1-associated myeloid leukemia"

Camillynn Brannan
University of Florida

"Haploinsufficiency of NF1 modulates C-kit signaling in melanocytes and mast cells in vivo"

Wade Clapp
Indiana University

"NF1, neural crest and cardiac development"

Jon Epstein
University of Pennsylvania

Neurofibromin—lipids and mitochondria connection
Mladen Golubic
Cleveland Clinic Foundation

NF2 Morning Session: James Gusella, Harvard Medical School/MGH (Chair)

"Merlin as a molecular integrator"

David Gutmann
Washington University

"The involvement of calpain-dependent proteolysis of the tumor suppressor NF2 (merlin) in schwannomas and meningiomas"

Mitsuyoshi Nakao
Kumamoto University

"Inter- and intra molecular interactions of Merlin isoforms"

Vijaya Ramesh
Massachusetts General Hospital

"The NF1 message from tumor cells"

Lynn Rutkowski
University of Pennsylvania

"Actin-membrane linking proteins in transformed cells"

Elizabeth Luna
University of Massachusetts.

"Requirements for high affinity binding of cellular moesin to actin filaments"

Heinz Furthmayr
Stanford University

"Structure of the moesin N/C-ERMAD: Implications for merlin"

Anthony Bretscher
Cornell University

NF2 Afternoon Session: Gilles Thomas, Pasteur Institute (Chair)

"Mouse models for NF2"

Marco Giovannini
Pasteur Institute

"Role of the NF2 tumor suppressor, merlin in tumorigenesis and metastasis"

Andrea McClatchey
MGH Cancer Center

The COOH-terminal domain regulates functional activity of Merlin

Markku Sainio
University of Helsinki

"Searching for proteins that interact with the NF2 gene product"

Guy Rouleau
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"Effect of NF2 missense mutations on schwannomin interactions with itself and other proteins"

Stefan Pulst
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"Genetic analysis of Drosophila Merlin and human NF1 function"

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