

# Outlook

The Newsletter of The Bachmann-Strauss Dystonia & Parkinson Foundation, Inc. • Volume 2, Number 2 • Fall 2001

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## Landmark Dystonia Research Featured in *Brain Research Magazine*

We are proud to report that the work of Drs. Ruth Walker and P. Shashidharan, funded by the Bachmann-Strauss Dystonia & Parkinson Foundation (BSDPF), recently made the cover of *Brain Research* magazine.

Working together at Mount Sinai, and using an antibody developed by Dr. Shashidharan, the researchers searched an entire rat's brain for the protein torsinA (which is produced by the DYT1 gene, a mutation of which has been identified as the cause of one form of dystonia). They were hoping to find the protein only in specific neurons, in order to pinpoint the parts of the brain affected in dystonia.

Instead, they found that torsinA appears to be present in virtually all of the rat brain's neurons. In other words while the function of torsinA is still not known, we now know that whatever it does, it does

throughout the brain. This is an important finding, implying that additional factors, as yet unknown, are responsible for the development of dystonia.

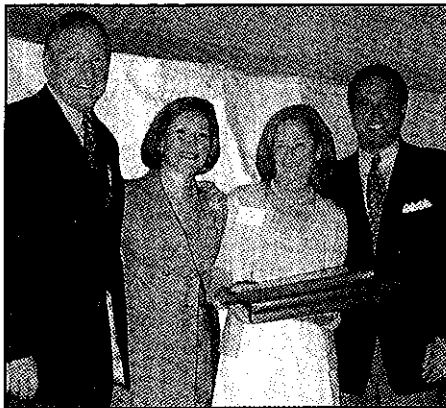
The *Brain Research* article, entitled "Distribution and Immunohistochemical Characterization of torsinA Immunoreactivity in Rat Brain," also covers other aspects of the study in which the researchers used different antibodies to identify neurochemicals in different subtypes of neurons.

Further research is needed to determine the function of torsinA, and to determine how this function is affected by the DYT1 mutation. We also need to learn why the resulting neurological symptoms are limited to a movement disorder, and why symptoms develop only in 30-40% of those who carry the mutated gene. ☆



Dr. Ruth Walker

## Golf, Tennis & Record-Breaking Fundraising: The 9th Annual Dystonia Invitational



Bonnie Strauss (second from right) congratulates the Dystonia Invitational's Corporate Chairs (l. to r.) Jay Goldberg, Mary Cirillo Goldberg and Loren Katzovitz on their record achievement.

It just keeps getting better! This year's Dystonia Invitational, held on June 18, raised over \$1.3 million to support movement disorder research. That's the largest sum we've raised to date from our annual golf outing. Over 250 amateur golfers (plus 70 pros) took part in the Hedi Kravis Ruger Memorial Tournament, playing at Century Country Club, The Golf Club of Purchase and The Blind Brook Club, all located in Purchase, New York.

Mitchell F. Brin, M.D., a member of our Board, a longtime Director of the Movement Disorders Program at Mount Sinai, and currently a Vice President at Allergan, Inc.

(continued on page 5)

### The Bachmann-Strauss Dystonia & Parkinson Foundation, Inc.

The Bachmann-Strauss Dystonia & Parkinson Foundation, Inc., was established in 1995 to raise funds to support research, provide treatment and promote medical and patient education. These efforts are designed to "make a difference" for those with dystonia and other movement disorders.

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THE BACHMANN-STRAUSS  
DYSTONIA & PARKINSON  
FOUNDATION, INC.

The Mount Sinai Medical Center  
One Gustave L. Levy Place, Box 1490  
New York, NY 10029

**In Support of  
Movement Disorders  
Research**

*Address Service Requested*

**Visit Us on the Web:**  
*[www.dystonia-parkinsons.org](http://www.dystonia-parkinsons.org)*

In addition to learning more about the Bachmann-Strauss Dystonia & Parkinson Foundation in this newsletter, you can find us on the web at the address above.

Now, information about the Foundation's mission, events such as the Dystonia Golf Invitational and Young Professionals are only a few mouse clicks away. If you have questions about a dystonia or Parkinson's disease diagnosis, or want to know more about the direction of current research, you'll find answers on the Bachmann-Strauss website, as well as literature in our publications section. Also featured are summaries of the most recently held patient and family symposia on dystonia and Parkinson's, as well as "Bonnie's Story," the inspiring account of the Foundation's founder and her determination, first to have her own dystonia properly diagnosed and treated, and then to support researchers looking for treatments and cures for both dystonia and Parkinson's disease.

Please visit our website soon — and then use our "Contact Us" section to give us your feedback on this new communications tool. ☆

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## Dystonia Medical Research Foundation Hosts First-Ever Dystonia Family Symposium



*Guest speakers at the Symposium included (l. to r.): Zach Haney, Dr. Cynthia Comella and Laurie Zaimanowitz.*

Families gathered in Chicago on Memorial Day weekend for the inaugural Dystonia Family Symposium sponsored by the Dystonia Medical Research Foundation, specifically developed for children, parents and families.

"What a wonderful weekend filled with the honesty and innocence of children — and full of information for adults and siblings," said Darlene Moser, Leader of the Lincoln Nebraska Dystonia Support Group and Southwest Regional Coordinator. Darlene participated and spoke on the "Genetic Counseling and Pregnancy" panel.

The symposium was designed to address dystonia as it affects the whole family. Sessions addressed medical topics, family dynamics, sibling issues, education and disability rights, genetic counseling and pregnancy, assistive technology, assistance dogs, recreation and sports, career counseling, and choosing a school.

As Meghan Fell reflected, a member of the 18-40 age group, "The symposium was a huge success and a lot of fun. I know that all of us are looking forward to the next one." ☆

## The President's View



First, I want to express my appreciation to my staff for their tireless work in organizing the Dystonia Invitational and the Young Professionals' events this year, and my gratitude to the many others who supported the two events by purchasing tickets, journal ads, raffle tickets and auction items. Special thanks go to Mary Cirillo Goldberg, Jay N. Goldberg and Loren M. Katzovitz for chairing the Dystonia Invitational, to Lisa Borowitz who has helped with each Invitational, and to Emily Kanders and Elizabeth Kohn, Co-Chairs of this year's Young Professionals' event. Their generosity of spirit and ample organizing talents created two wonderful events enjoyed by all who attended, and raised over \$1.4 million for research. Thanks also to C. Hugh Hildesley, Executive Vice President of Sotheby's, for conducting our auction and especially Dr. Mitchell F. Brin our honoree, for his ongoing commitment to research in movement disorders.

For readers who live with dystonia or Parkinson's disease, and for their families, I call your attention to two upcoming symposia: on dystonia (October 15th) and Parkinson's (November 1). Part of the BSDPF's mission is keeping patients informed about the latest research developments and treatment options. These symposia will provide valuable information you won't want to miss. Times and locations are listed on page 3 (Save These Dates).

The BSDPF supports research in both dystonia and Parkinson's disease because both disorders entail involuntary muscle activity characterized by a malfunction in the brain's basal ganglia. The study of these disorders by scientists working together has increased the understanding and treatment of both disorders. For example, the use of Deep Brain Stimulation (DBS) for Parkinson's disease is now being used to treat some dystonia patients. Moreover, some families have a history of Parkinson's disease and/or dystonia appearing in different generations. Further study of the genetics involved in both disorders could prove to be very important in finding a cure. Scientists, like those at Mount Sinai, often conduct research on both of these diseases to more fully understand the function of the basal ganglia in movement disorders. The BSDPF will again be supporting both dystonia and Parkinson's researchers at the Mount Sinai Medical Center and through the Dystonia Medical Research Foundation in the coming year to bring us closer to finding a cure for these devastating disorders.

I close this column with a salute to all the researchers who work with such dedication to uncover the causes of dystonia and Parkinson's disease, and to find treatments. Their efforts already have yielded both a better understanding of movement disorders and a marked improvement in the quality of life for people with these disorders. And all of us at the BSDPF congratulate Drs. Walker and Shashidharan for the publication of their paper as the cover story of a recent issue of *Brain Research* magazine (see page 1). We are proud to support their work. ☆

*Bonnie Strauss*

Bonnie Strauss  
President and Founder

## Dystonia: A Parent's Story

When Ashley Friedman was six, she started to limp on one foot. Her mother, thinking Ashley had injured herself, took her to the pediatrician.

After a year of multiple misdiagnoses — including visits to an orthopedist, a pediatric neurologist and even a child psychiatrist (who firmly believed the problem was psychosomatic), Ashley was finally diagnosed with dystonia by Dr. Fabio Danisi of Mount Sinai's Movement Disorders Program.

Ironically, after visiting so many specialists, it was a physical therapist who first suggested to the family that Ashley might have dystonia, and recommended that she seek help at Mount Sinai.



Ashley Friedman

"It took an emotional toll on Ashley," her mom says, "because we didn't believe her. The psychiatrist asked her, 'Why are you acting like a baby?' When she finally got her diagnosis, she was angry with me and with everybody."

Ashley wasn't the only one who suffered because of the misdiagnosis. A school social worker reported Ashley's case to the N.J. Division of Youth and Family Services, believing that the cause of Ashley's supposed psychological problems must be abuse. A state social worker wanted her committed to an institution. Fortunately, knowing these suspicions were absolutely wrong, the Friedmans refused.

Ashley is in a wheelchair now. It's hard for her to watch other children doing all the things she used to love to do — like riding a bike, dancing, roller-skating. "But she's been a trouper," Mrs. Friedman says. "She goes to school every day, she goes to the Girls and Boys Club after school and plays basketball with the other kids. To be honest, if it happened to me, I don't think I could handle it as well."

It hurts Mrs. Friedman that her daughter has had to miss out on "so much of the good part of being a child." But the family has high hopes for the Botox® treatments that Ashley is about to begin. "We try to keep a positive attitude. I hope the treatments will work well enough so she can stand again, and take steps with assistance. There's nothing to lose, and everything to gain."

We wish Ashley and her family the best of luck, and hope that she is among the many dystonia patients who have so far responded well to Botox®. ☆

## Executive Director's Report



It has been a very full eight months since I joined the Bachmann-Strauss Dystonia & Parkinson Foundation in January. Our office is now fully staffed and I want to extend a big welcome to Shari Berg serving as our Events Coordinator, and Karina Rustia serving as our Events Manager. Shari and Karina's excellent skills helped us raise additional funds for dystonia and Parkinson's disease medical research. Our success also could not have been possible without the assistance of over 15 volunteers who worked really hard in helping us surpass our goals.

We had a very successful Fourth Annual Young Professionals' event at the Central Park Boathouse (see story on page 3). We extend our appreciation to the Co-Chairs Elizabeth Kohn and Emily Kanders; as well as the Steering and Event Committees: Marilyn Ostro, Journal Chair; Robin Fried, Favor Chair; and Dan Flynn and Tom Hess, Raffle Chairs.

With two very successful events, the Dystonia Invitational and the Young Professionals' Central Park outing behind

us, we are now very busy planning the Parkinson's Theater Benefit on November 14 at the Plymouth Theater. Most importantly, we are sending out requests for proposals for medical research grants that will receive funding in 2002. The Bachmann-Strauss Dystonia & Parkinson Foundation's Scientific Advisory Board will review all proposals in October.

We hope to see you in the coming months at one of our events or in our new office. We also want to wish you a wonderful fall and holiday season. Next year we will be celebrating the 10th anniversary of the Dystonia Invitational and we will plan something really special. Thanks again for joining our team effort in finding a cure for dystonia and Parkinson's disease. Please contact our office at (212) 241-5614 or [Bachmann.Strauss@mssm.edu](mailto:Bachmann.Strauss@mssm.edu) to receive additional information or to volunteer your time.

We always look forward to hearing from you. ☆

Margie J. Walden  
Executive Director

## Golf, Tennis & Record-Breaking Fundraising

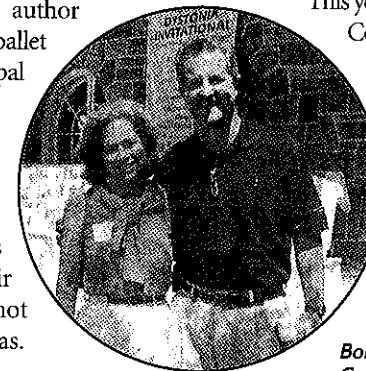
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(manufacturer of BOTOX®) was this year's honoree. Board member and award-winning composer Billy Goldenberg wrote a song called "A Doctor" to honor Dr. Brin on this very special occasion. The song was performed by Broadway legend Penny Fuller, who is currently starring on Broadway in *The Dinner Party*.

Keynote speaker Dr. Max Gomez, the Health and Science Editor for NewsChannel 4, said, "With increased funding for research, a cure can be found within the next ten years."

More than 320 BSDPF friends and supporters attended the evening dinner and auction. Among the nearly two dozen spectacular items auctioned off were a four-night stay at a 17th Century chateau in Bordeaux, France, a cocktail party with celebrated author Tom Wolfe, and a child's private ballet lesson with NYC Ballet Principal Dancer Kathleen Tracey.

We are deeply indebted to Mary Cirillo Goldberg, Jay N. Goldberg and Loren Katzovitz, who served as Chairs for this very special day. Without their hard work, this event would not have been the huge success it was.

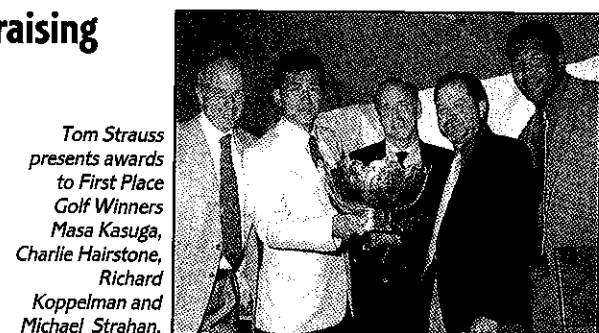


Bonnie Strauss welcomes keynote speaker, Dr. Max Gomez, NewsChannel 4's Health & Science Editor.

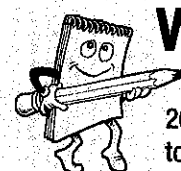
We also are grateful to Allergan, Inc., and Bank of America Private Bank, our corporate sponsors. Allergan, Inc., will contribute \$250,000 over the next five years to establish the Mitchell F. Brin Fellowship in Movement Disorders at Mount Sinai.

This year's tournament was dedicated in memory of John M. Corrigan (1942-2000), a beloved board member who was with us from the very beginning of the Foundation, and who is sorely missed.

Proceeds from the Dystonia Invitational will fund research projects for the Movement Disorders Program at the Mount Sinai Medical Center. In addition, funds will support research projects in collaboration with the Dystonia Medical Research Foundation. ☆



Tom Strauss presents awards to First Place Golf Winners Masa Kasuga, Charlie Hairstone, Richard Koppelman and Michael Strahan.

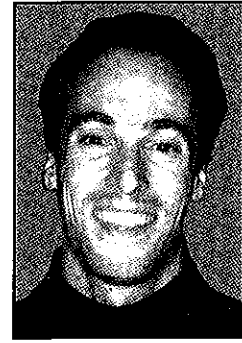


## We've Moved Up! Same Building — New Office

The new BSDPF office has become a reality! Last month, we moved "upstairs" to Room 2010 (20th Floor) in the Annenberg building. It's a much larger space — giving us more room to grow! Our phone number and mailing address remain the same. ☆

## The First Brin Fellow: Dr. Matt Brodsky

Dr. Matt Brodsky has been selected as the first recipient of the Mitchell F. Brin Fellowship in Movement Disorders. The fellowship was established by Allergan in cooperation with the BSDPF and the Mount Sinai Medical Center Department of Neurology. It honors Dr. Brin's exceptional commitment and research in the field of movement disorders. Dr. Brodsky will use the fellowship to continue his research at the Mount Sinai Medical Center's Department of Neurology, where he completed his residency a year ago.



Dr. Matt Brodsky

Dr. Brodsky's recent clinical research has focused mainly on treatments for Parkinson's Disease and dystonia. He is the co-author of the article on Parkinson's Disease to be published in the *Encyclopedia of the Brain*, as well as co-author of the soon-to-be-published "Neurologic Drug and Psychotropic Drug Interactions: Significance and Recommendations." ☆

## Young Professionals Make a Difference — Once Again!

The Fourth Annual Young Professionals' Benefit was held on Wednesday, July 18, at The Boathouse in Central Park. Benefit Co-Chairs Emily Kanders and Elizabeth Kohn did a fabulous job putting together this evening of cocktails, hors d'oeuvres, and dancing under the stars. Of the more than 300 people who attended, 39 won raffle prizes, including a weekend in South Beach at the Greenview Hotel (donated by the Rubell family), gift certificates to ARK restaurants, the Russian Tea Room, Lenox Restaurant, Blue Hill Restaurant, candlesticks from Tiffany, riding lessons from the North Shore Equestrian Center, and more. To top it all off, attendees received "goody" bags filled with items donated by Atlantic Records, Bloom, Robin and Keven Brennan, Colgate-Palmolive Company, GlaxoSmithKline, Conde Nast Traveler, Michael C. Fina, North Shore Bottling Company, Smith & Wollensky, Taste of Nature, and Zagat.



In all, the event raised \$50,000, which will be used to fund movement disorders research at the Mount Sinai Medical Center. ☆

Young Professionals Benefit Co-Chairs (left to right) Elizabeth Kohn and Emily Kanders.

# save these dates!

## Dystonia Symposium

Monday, October 15, 2001, 2-4 pm

Hatch Auditorium, The Mount Sinai Medical Center, Guggenheim Pavilion (5th Avenue and 100th Street).

Speakers, including Dr. C. Warren Olanow, Dr. Jose Obeso, Dr. Fabio Danisi and Margie J. Walden will discuss cutting-edge research, advances in surgical procedures, the latest in clinical procedures and the educational benefits of non-profits for patients.

## Parkinson's Symposium

Thursday, November 1, 2001, 2-4 pm

Goldwurm Auditorium, The Mount Sinai Medical Center, 1425 Madison Avenue (northeast corner of 98th Street).

Speakers will include Dr. C. Warren Olanow, Dr. Jean-Michel Gracies, Dr. Dan Perl and Susan Dolan. Topics to be discussed include current research from around the world and advances in surgical procedures.

## ... AND SAVE THIS DATE!



Parkinson's Theatre Benefit  
The Bachmann-Strauss Dystonia & Parkinson Foundation

Wednesday, Nov. 14, 2001

**THOU SHALT NOT**  
a new musical

David Thompson  
Author

Susan Stroman  
Director & Choreographer

Harry Connick, Jr.  
Music & Lyrics

Pre-Theater Dinner

PLYMOUTH THEATRE 236 West 45th Street  
A LINCOLN CENTER THEATER PRODUCTION

Proceeds from this event will support Parkinson's Disease research

# research notes

Each year, the BSDPF supports cutting-edge research projects at Mount Sinai. Grantees for the year 2001 include:

## DYSTONIA

**Stuart Sealfon, M.D.:** "D2 Receptor Mutation Linked to Myoclonus Dystonia." In order to determine whether the D2 receptor mutation (linked to myoclonus dystonia) may disrupt this neuroprotective pathway, the research team is establishing stable cell lines with the dystonia-linked D2 receptor mutations. The team will evaluate the capacity of this mutant receptor to signal via atypical signal pathways and to mediate neuroprotection. People with myoclonus dystonia may have myoclonus (fast, lightning-like jerks) with or without dystonia. There are a few families described with this disorder and the mutation of the dopamine D2 receptor. Although the effect of this mutation is not yet known, this will be studied in cell cultures with the mutation.

**P. Shashidharan, Ph.D.:** "A Transgenic Mouse Model of DYT1 Dystonia." Significant progress has been made in this team's efforts: A colony of mice carrying the transgene has been generated, and two of the transgenic mice developed abnormal movements and postures at the age of one month.

**Ruth Kornreich, Ph.D., Jianli Dong, M.D., Ph.D., Robert J. Desnick, Ph.D., M.D.:** "Determination of the Carrier Frequency and Penetrance of the 946delGAG Mutation in the Torsion Dystonia (DYT1) Gene in the Ashkenazi Jewish Population." Having screened 2,000 Ashkenazi Jewish individuals for this mutation (the major mutation responsible for torsion dystonia in the Ashkenazi Jewish population), the team has found a carrier frequency of 1 in 400. Preparations are now underway to analyze penetrance, which means to understand why some people with the DYT1 gene develop the symptoms of dystonia and some do not.

**Colum D. MacKinnon, M.D., Ph.D.:** "Cortical Control of Agonist-Antagonist Muscle Activity in Patients with Primary Dystonia." Nerve cells from the brain control the activity of neurons in the spinal cord and play a role in controlling movements. Sensory feedback from the limbs is known to be abnormal in dystonia and is modulated by their descending pathways. The team will use magnetic stimulation to non-invasively study nerve pathways from the brain to the spinal cord and from there to the wrist in control subjects and people with dystonia. This provides an avenue for further investigation, as well as a way to test the efficacy of treatment regimens.

**Jeremy M. Silverman, Ph.D.:** "The Phenotypic Expression of Myoclonic Dystonia." Working with families with individuals who have myoclonic dystonia (MD), the team's aim has been to conduct detailed psychiatric and cognitive examinations of both affected and unaffected members. Assessments are now in progress. The team has found an interesting association between MD and obsessive-compulsive traits; depression, anxiety and substance abuse are also often present in the study families. However, the current number of participants is too small for meaningful statistical comparison, making the study of additional families necessary.

**Fabio Danisi, M.D.:** "Genetics and Phenotypic Expression of Idiopathic Dystonia, Parkinson's Disease, and Other Movement Disorders." A genetics research team has been coordinating and implementing studies, with the goals of finding the gene(s) responsible for dystonia and Parkinson's, as well as better defining the clinical pictures of various types of dystonia. The team and its collaborators have published or submitted a dozen papers on this work.

**Jean-Michel Gracies, M.D., Ph.D. and Donald Weisz, Ph.D.:** "Rapid Alternating Movements in Human Subjects: Establishing Normative Data for Quantitative Testing in Dystonia and Parkinson's Disease." It is known that hand movements are slowed in Parkinson's and abnormal in dystonia, however it has not been easy to measure this objectively in the clinic. The Hand Tapper device has been developed to measure hand tapping speed in a convenient manner, to assist with diagnosis and monitoring of patients' response to treatment. Initial studies need to determine normal values and those in Parkinson's and dystonia. Sixty-six control subjects so far have been studied, and their movements quantified using the Hand Tapper. Fifteen Parkinson's patients also have been studied. In the future, Hand Tapper evaluation may be used as a diagnostic tool, allowing objective study of patients' active function over time.

## PARKINSON'S DISEASE

**Catherine Mytilineou, Ph.D.:** "The Role of Arachidonic Acid in Neurodegeneration." This study looks at a mechanism by which dopamine cells may be damaged in Parkinson's disease. This may lead to ways to protect dopamine cells from oxidative stress. Initial experiments have supported their theory that arachidonic acid plays an important role in this mechanism.

**P. Shashidharan, Ph.D.:** "Torsin-A Overexpression in Dopamine Neurons and Sporadic Parkinson's Disease in a Mouse Model." In preparation for further study, the team has cloned DNA for torsinA from human postmortem tissue. ☆