



# Outlook

FALL 2003

## Tracking the Mystery of Adult Onset Dystonia

*"Most dystonia should be curable because there's no obvious brain degeneration."*

At this point no one really knows how many thousands of men and women suddenly get dystonia in their adult years, partially because it is a disease that is often undiagnosed or misdiagnosed. While it begins at varying ages and takes different forms, adult onset dystonia is often "focal" – attacking only one area of the body. One form of the disease, torticollis or cervical dystonia, which causes neck spasms, usually occurs between 35-45 years of age while blepharospasm, which causes blinking, may occur in one's 50s or 60s.



Susan Bressman, MD

A team of two scientists are now working with other researchers to unravel this mystery. Funded by a 2003 grant from The Bachmann-Strauss Dystonia & Parkinson Foundation to conduct "Genetic Association Studies in Focal Dystonia Patients", Susan Bressman, MD, and Laurie Ozelius, PhD, are co-principal investigators who approach the puzzle from two different perspectives. Dr. Ozelius runs her own laboratory in human genetics at the Albert Einstein College of Medicine in the Bronx and Dr. Bressman is a clinician and Chairman of The Alan and Barbara Mirken Department of Neurology at Beth Israel Medical Center in New York.



Laurie Ozelius, PhD

Working collaboratively, Bressman and Ozelius will compare differences in groups of adults affected by dystonia versus those of a control population. They will look at the various types of the disease that affect specific areas of the body including blepharospasm, torticollis, laryngeal or spasmodic dystonia, which affects the vocal cords, and writer's cramp. Their hope is to find at least 100 patients affected with each variety.

In a related study funded by the National Institutes of Health, Bressman and Ozelius are specifically studying large families with many affected individuals. "We're scouring the planet, especially looking all over the U.S. and Canada, and we're collaborating with a group in Ireland," says Dr. Bressman. Finding participants is a challenge. "The problem is that many people who have dystonia are still not diagnosed for it," says Dr. Ozelius. "People with writer's cramp may not seek treatment and those with a blinking problem may go to an eye doctor instead of a neurologist. It's an education problem. Many doctors still don't know about dystonia."

The study will also look at the types of dystonia that occur among different ethnic groups and among the sexes. Right now there is some evidence that cervical dystonia affects a large number of people of Irish and German descent, while African-Americans tend to get spasmodic dystonia. Focal dystonia also appears to be more prevalent in women than in men. Studies to obtain solid data are also needed to confirm the incidence of these diseases among different groups.

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## MARK YOUR CALENDAR

### Key Information for Patients and Families

Held annually, these important free events bring patients, families and caregivers up-to-date on the latest research breakthroughs.

#### Thursday, October 9, 2003

Join us in the Hatch Auditorium, Guggenheim Pavilion, Mount Sinai Medical Center, Fifth Avenue and 100th Street, New York City.

#### • Dystonia Symposium 11:00 a.m. - 12:00 p.m.

#### • Keynote Speaker and Discussion for Dystonia and Parkinson's Patients 12:30 - 1:30 p.m.

Presented by Daniel Moros, MD, Associate Clinical Professor, Department of Neurology, Mount Sinai Medical Center.

#### • Parkinson's Disease Symposium 2:00 - 3:00 p.m.

## SAVE THE DATE

Don't miss out on a terrific night of theater to benefit Parkinson's disease research.

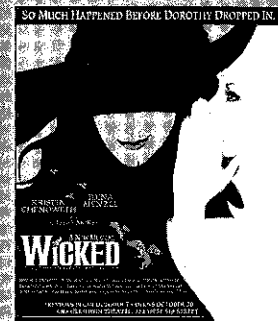
#### Thursday, October 16, 2003

Join us for dinner and the Broadway performance of **Wicked**

#### The Untold Story of the Witches of Oz

Gershwin Theater  
222 West 51st Street

Dinner at 6 p.m. at Victor's Cafe and Theater at 8 p.m.



For more information on these events or to register, please call 212.241.5614 or email [Bachmann.Strauss@mssm.edu](mailto:Bachmann.Strauss@mssm.edu).

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## Tracking the Mystery of Adult Onset Dystonia

So far, researchers have seen some evidence that some adult onset dystonia may be gene-related but Dr. Bressman, a specialist in dystonia genetics for the past 22 years, doesn't believe that is the entire cause. "Because there are so many different subtypes of the disease, we think there could be multiple factors and causes," she said. "Genes are certainly a major piece of the puzzle but they're not the only explanation. Perhaps it's environmental or perhaps it's the interaction between several genes. That's why we're doing this study."

According to Dr. Ozelius, only 10 to 35 percent of people say there is another relative in their family with symptoms, but some people may not find an exact match in their family because the other person may have a different form of the disease. Dr. Bressman can ask the key questions to probe: Does anyone in your family blink a lot? Does anyone talk in a whisper or complain of a stiff neck?

By isolating DNA – the blueprint that makes each of us unique – from people with different types of dystonia, Bressman and Ozelius will be able to look at the small gene changes that may occur to try and pinpoint a cause and to see how different genes may interact with each other. Along the way, they will also look at the commonalities between dystonia and Parkinson's disease to see if there is a link between the two. "We don't know where the problem is yet but there's reason to believe the same pathways in the brain are involved," says Dr. Ozelius.

Dr. Bressman believes that the key to a cure will be to find a way to alter the system. "Most dystonia should be curable because there's no obvious brain degeneration," she said. "We're all looking for a cure. It's going to take research and a lot of people working on it but we're definitely making headway."

To see if you are a candidate for this genetic study, please contact Beth Israel Medical Center at 888.228.1688.

## The Key to Support and Progress



BONNIE STRAUSS  
FOUNDER AND PRESIDENT



MARGIE J. WALDEN  
EXECUTIVE DIRECTOR

We move toward the fall with a very full calendar of activities for scientists, patients and family members. For the second year, this November we are bringing together medical professionals and researchers for a think tank on dystonia. This intensive two-day meeting will enable them to share progress, form collaborations, and make the kind of knowledge gains that will one day lead to a cure.

Our annual symposia on Parkinson's disease and dystonia, to be held Thursday, October 9, will provide patients, families and their caregivers with the most up-to-date information. Our theater benefit – the new Broadway play "Wicked" – will follow on Thursday, October 16. This event enables more people to support Parkinson's disease research through our organization.

Our highly successful Annual Dystonia Invitational, held earlier this summer, and the fun events organized by our Young Professionals produced tangible results, enabling us to make grants and develop key educational programs and initiatives. The generosity of the volunteers who help to organize events like these and the support of so many individuals and corporations make it possible for us to fund some of the most promising work in the field of movement disorders.

We are proud of the headway we are making and prouder still of those who support this important work. Many, many thanks.

## A Gift of Love and Hope

### Eighth graders raise funds to fight dystonia

When Lauren Von Der Ahe's school launched a drive to raise money for charity, the eighth grader told her classmates about dystonia and its devastating affect on her sister, Kristin. The students selected The Bachmann-Strauss Dystonia & Parkinson Foundation as a beneficiary and raised \$1,000. Lauren got matching gifts from her family, bringing the total gift to \$4,000 for dystonia research.

Kristin's life and her battle with dystonia is quite a story. Shortly before she was to start kindergarten this normal, active, and very bright child began having severe headaches. She was diagnosed with a benign brain tumor. During surgery both optic nerves were damaged and Kristin had a stroke. Afterward, with a continuing high fever and some sporadic movement, doctors kept increasing her medication. No one saw that she was having an allergic reaction.

"It was a very frustrating period because no one knew what was really happening to her," says her mother, Carolyn Von Der Ahe. Finally, an attending neurologist diagnosed Kristin with secondary dystonia.

As Kristin began to get her strength back, the dystonic symptoms became more pronounced. She began to lose her speech as her tongue increasingly went into spasm. Now 10 years old, Kristin is blind as a result of the surgery and unable to speak because her tongue and her mouth are in constant spasm. Special tutors help her to learn Braille, math and English, and she attends third grade in a regular neighborhood school in the Los Angeles area, where she's keeping up at grade level.

"Blindness is the least of her disability," says her mother. "She has never slept through the night because the dystonic movements wake her. She can't sit when she eats. She can sit in school but she can't use a pencil because it sets off movements. When she walks her arm will go into spasm, which throws off her balance."

The Von Der Ahe family continues to search for ways to help their daughter. "Dystonia doesn't just affect a child, it affects a family," says Mrs. Von Der Ahe. "She's gone farther than anyone ever thought she would, but the key comes down to what can be done for her dystonia."

They are now exploring whether Kristin is a candidate for Deep Brain Stimulation. Recently approved by the Food and Drug Administration, the surgery has shown some success in helping people with hereditary dystonia.

Lauren remains closely involved. She works as a volunteer helping Kristin to go horseback riding. Like many 14-year olds, she baby sits when her parents go out. But capping it off is the highly successful fundraising effort that she led to help her sister.

"My teacher knew about Parkinson's disease but no one at school had ever heard of dystonia. My mom came to our school assembly," says Lauren. "We gave her a check and she told the school about dystonia and about Bachmann-Strauss. Kristin was there too. She programmed her communications device and told the assembly, "Thank you for all you have done."



Lauren Von Der Ahe with her sister, Kristin

*"Dystonia doesn't just affect a child, it affects a family."*

# Grant Awards

## Initiative Launched to Study Dyskinesia in Parkinson's Disease Patients

### Bachmann-Strauss in collaborative effort with Michael J. Fox Foundation

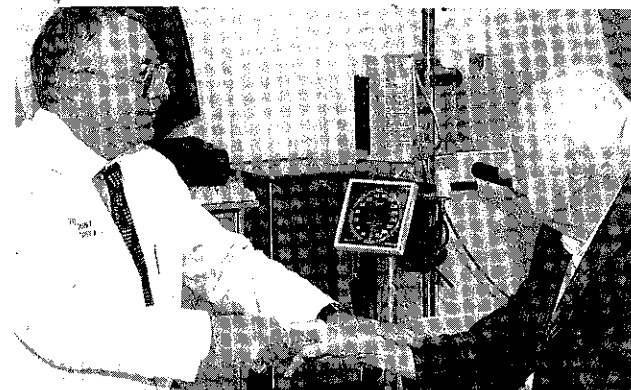
The Bachmann-Strauss Dystonia & Parkinson Foundation is partnering with the Michael J. Fox Foundation for Parkinson's Research to study dyskinesia, the uncontrollable movements that often characterize one of the side-effects of Parkinson's disease treatment. These movements are similar to the muscle spasms seen in dystonia.

This \$2 million research initiative will look at the molecular mechanisms of dyskinesia. Request for Proposals, sent to the research community, elicited 36 proposals from scientists in eight different countries. Our Foundation provided a \$100,000 grant for this study. Grantees will be announced shortly on our Web site—[www.dystonia-parkinsons.org](http://www.dystonia-parkinsons.org).

The presence of dyskinesia is a major limiting factor in achieving adequate long-term control of Parkinson's disease symptoms. The most commonly administered drug to treat Parkinson's symptoms is Levodopa (also called L-dopa), which helps restore levels of dopamine, a chemical in the brain responsible for smooth, coordinated movement and other motor functions. After five to ten years of treatment with Levodopa, approximately 80 percent of patients will develop uncontrollable movements or dyskinesias.

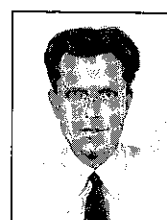
"Ours is the only organization actively looking at the interface of dystonia and Parkinson's disease," says Margie J. Walden, executive director of The Bachmann-Strauss Dystonia & Parkinson Foundation. "Dystonia and dyskinesia are both hyperkinetic movement disorders, so this collaborative effort makes tremendous sense and, we hope, will enable us to expand the knowledge we already have."

Currently there are approximately one million Americans living with Parkinson's disease.



Roger Mahey, right, is one of thousands of Parkinson's disease patients who suffer from dyskinesia. Here Dr. William Koller, director of Mount Sinai Medical Center's Movement Disorders Program, examines Mr. Mahey's symptoms.

### Thomas Haelbig Awarded Mitchell F. Brin Fellowship



Thomas Dino Haelbig, MD, a neurologist from Berlin, Germany, has been awarded The Mitchell F. Brin Fellowship for his work in movement disorders.

Dr. Haelbig is a Scientific Assistant on the medical faculty of the Humboldt University of Berlin, Germany, and a Resident in Neurology at the Department of Neurology, Charité-Humboldt University of Berlin. Specializing in movement disorders, he attended Charité-Humboldt University Medical School in Berlin, and the Université René Descartes, in Paris, France. His clinical education was at the Free University of Berlin Medical School, Berlin, Germany, and his preclinical education at the University of Heidelberg Medical School, Heidelberg, Germany.

Established by Allergan in cooperation with The Bachmann-Strauss Dystonia & Parkinson Foundation and Mount Sinai Medical Center's Department of Neurology, the fellowship pays tribute to Dr. Brin's commitment and research in the field of movement disorders.



Practice session for the Invitational

### 11th Annual Golf Event Shines Bright

After weeks of rain that soaked the northeast, the air was crisp and the sun was bright for our 11th Annual Dystonia Invitational. Playing at three courses in Westchester County, 215 golfers and 55 pros participated in this Hedi Kravis Ruger Memorial Tournament. The evening's cocktail reception, dinner and very lively auction was attended by 220 people.

The show of support for our organization was outstanding, raising more than \$1.2 million for movement disorders research.

Our dinner honoree was Loren M. Katzovitz, president and managing partner, Guggenheim Partners and member of our Board of Trustees, who was recognized for his commitment and dedication to raising funds for medical research. L.H. Hollier, MD, president, the Mount Sinai Hospital, gave the keynote speech about the important work done by young researchers. The guest who stole the show, captivated the audience, and received a standing ovation was 14 year old Lauren Von Der Ahe, who spoke about her younger sister Kristin's battle with dystonia. (See article in this issue.)

Our thanks go to the Corporate Chairs, Patrick Hughes, J. Todd Morley and Jonathan Spitalny, to the Vice Chairs, to the Benefit Committee, and to the 15 volunteers who worked tirelessly to make this a truly successful event.

Save the date: 12th Annual Dystonia Invitational, June 21, 2004

### Young Professionals Spring into Action

With great energy, commitment and spirit of fun, our Young Professionals organized two outstanding events this year – one in February and one in July – raising over \$52,000 to support research for dystonia and Parkinson's disease.

Led by Co-chairs (from left below) Dan Flynn, Liz Strauss and Tommy Hess, more than 260 people came to our evening of dancing under the stars at Tavern on the Green at the end of July. There was also a raffle and silent auction with impressive items ranging from a weekend in South Beach to tickets to a Yankees' game.

If you are between 25-35 years old and would like to join our Young Professionals, please contact 212.241.5614 or email [Bachmann.Strauss@mssm.edu](mailto:Bachmann.Strauss@mssm.edu).



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The Bachmann-Strauss Dystonia & Parkinson Foundation, Inc. was established in 1995 to find better treatment and cures for the movement disorders dystonia and Parkinson's disease, and to provide medical and patient information. An independent, nonprofit, 501(c)3 organization, its funding is made possible through the generosity of individual and corporate contributors.



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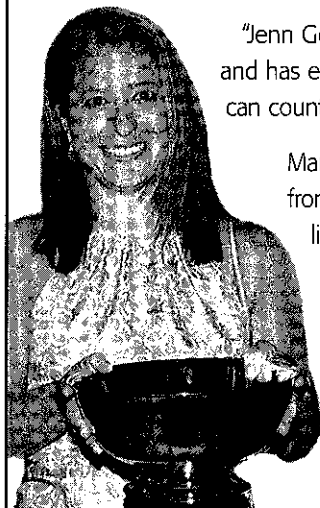
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### **Jennifer Geller Receives First Annual Scott M. Johnson Memorial Award**

After 26-year-old Scott Johnson tragically lost his life in the September 11th terrorist attack on the World Trade Center, his colleagues on our Young Professionals Committee wanted to find a way to honor his life, his spirit and his dedication. The result was the creation of The Scott M. Johnson Memorial Award for Dystonia & Parkinson's Disease Research.

Jennifer Meltzer Geller, the first recipient of this award, became involved with our organization four years ago after her mother-in-law was diagnosed with Parkinson's disease. Since then, she has been actively involved in various fundraising activities including serving as a member of our Young Professional Benefit Committee and a Co-chair of our Silent Auction Committee.



"Jenn Geller has attended meetings, worked at getting Journal ads, raffle and silent auction items, and has encouraged her friends to get more involved and come to our events. She is someone you can count on to get the job done," says Bonnie Strauss, our founder and president.

Manager of Human Resources for Tiffany & Co, Jennifer received her undergraduate degree from George Washington University and holds an MA and EdM from Columbia University. She lives in Manhattan with her husband, Richard Geller, and their 1-year-old daughter Alexis.

*The Scott M. Johnson Memorial Award for Dystonia & Parkinson's Disease Research is given annually to a member of our Young Professionals Committee who demonstrates tremendous enthusiasm and dedication in helping our organization raise funds and public awareness about these neuromuscular diseases.*

*Jennifer Geller receiving her award*