Laurie Ozelius Joins Mount Sinai Team

When Laurie Ozelius speaks about her new position as The Bachmann-Strauss Professor in Movement Disorders at New York’s Mount Sinai School of Medicine she describes it as a natural extension of the research she has been doing all along in human genetics. The difference is she will now have the built-in opportunity to work directly with clinicians and with patients.

Laurie Ozelius, PhD

Robert J. Desnick, PhD, MD, Professor and Chairman, Genetics and Genomic Sciences, Mount Sinai School of Medicine said, “We are delighted that Dr. Ozelius will join our faculty. This appointment brings an internationally-recognized leader in the genetics of movement disorders to Mount Sinai and represents an important collaboration between the Departments of Neurology and Genetics and Genomic Sciences.”

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Outlook WINTER 2007

New Grants Awarded to Further Research

With a sharp focus on advancing understanding of dystonia and Parkinson’s disease The Bachmann-Strauss Dystonia & Parkinson Foundation awarded $850,000 in grants in 2006. Based on recommendation of our Scientific Advisory Board, grants were made to individual researchers. (See inside.) This funding also includes support to the Dystonia Medical Research Foundation (DMRF) to augment the grants they award, and to the Michael J. Fox Foundation to continue our collaborative studies.

Among this far-reaching body of work, several individual studies are of particular note. Kathleen Sweadner, PhD at Massachusetts General Hospital and Ellen J. Hess, PhD at Johns Hopkins University, for example, are both exploring different aspects of the relationship between dystonia and Parkinson’s disease.

In order to better understand the abnormalities within the basal ganglia that result in dystonia and to develop new therapies, Dr. Hess and her team will develop a new mouse model with an inherited dystonic disorder.

They have identified L-DOPA-responsive dystonia (DRD), as a leading candidate for this study. The mutation being used is intriguing because it causes a range of disorders from dystonia to Parkinsonism and will provide a critical tool for insight into their relationship. It is expected that the generation of this model will provide the seed for a federal grant that will further our understanding of the link between dystonia and Parkinsonism.

A new clue about why the brain’s basal ganglia are vulnerable to so many disorders has come from a recently uncovered gene that causes a unique, rapid-onset dystonia and Parkinsonism (RDP).

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**Peter Feld and Brian Dugan Receive Award**

Two people were honored with the 2006 Scott M. Johnson Memorial Award for Dystonia and Parkinson’s Disease Research for their efforts to raise funds and highlight awareness. Presenting the award to Brian Dugan (left) and Peter Feld (right) was Scott’s sister, Margaret Johnson.

This award is given annually by The Bachmann-Strauss Dystonia & Parkinson Foundation to Young Professionals (YP) who are recognized as embodying Scott Johnson’s generous spirit. Scott was a member of our Young Professionals Committee who lost his life on September 11, 2001 in the World Trade Center.

Peter Feld has been actively involved in the YP group for a number of years and has served as its co-chair for the last year and a half. Brian Dugan organized a special team of New York City Marathon runners this past November that raised over $34,000 to benefit our Foundation. Brian’s motivation is his mother, Joan Dugan, who has a severe form of Parkinson’s disease.

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**Film Highlights Stories of People with Dystonia**

The Bachmann-Strauss Dystonia & Parkinson Foundation and the Dystonia Medical Research Foundation co-sponsored the New York fall premiere of “Twisted,” a film which depicts the stories of people who have dystonia.

Shown here in a panel discussion that followed the film screening are Susan Bressman, MD, Paula Schneider, a patient with dystonia, and Laurel Chiten, the film’s creator.

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**Nathan Lane Stars as “Butley”**

Bonnie Strauss with Nathan Lane following his performance as “Butley.” This limited engagement Broadway play was selected for our recent theater benefit, which raised $108,000 for research.
Dekker Foundation Gives $250K Grant

The Dekker Foundation recently awarded a multi-year grant to The Bachmann-Strauss Dystonia & Parkinson Foundation totaling $250,000. This grant for $50,000 a year over five years will begin in 2007. It was made to “support clinical research addressing therapeutic complications from Parkinson's disease and subsequent treatment methods, particularly movement disorders resulting from or exacerbated by established pharmacotherapies, medical devices and co-morbidities.”

The Dekker Foundation is a private foundation endowed by Marcel Dekker, Inc. and the Dekker family, publishers of scientific, technical, and medical information.

New Tax Benefits for IRA Gifts

The Pension Protection Act of 2006 now allows people ages 70 1/2 or older to make direct tax-free charitable transfers of up to $100,000 from their IRA before the end of 2007.

If you would like to discuss ways in which you can take advantage of this new opportunity to support dystonia and Parkinson’s disease research, please call Helen Miller, Executive Director of The Bachmann-Strauss Dystonia & Parkinson Foundation at 212.241.5614.

Gaining Momentum, Building Support

It is tremendously exciting to watch ideas gain momentum. That certainly was the backdrop at our fifth annual Think Tank. Held this past fall under the direction of our Scientific Advisory Board Chair Dr. Ted Dawson, this forum brought together nearly 40 leading clinicians and research scientists from around the globe in a highly charged, enthusiastic and positive step toward finding cures. They shared studies on the pathophysiology, imaging results, and surgical treatments. Our collective purpose: to exchange information and expand collaborative effort to accelerate understanding of dystonia.

The research grants we award and initiatives like our Think Tank are made possible by the generous funding we receive from individuals, corporations and foundations – support that often generates additional gains.

A great example was recently set by Brian Dugan, who recruited a team of people from across the country to run in the New York City Marathon in support of movement disorders research through our Foundation.

We all know how it works: one person tells another who tells another, and so forth. Thanks in large measure to that timeless principle, we began this New Year with a broader than ever base of support that now spans nearly every state across our country. We can’t thank you enough.

Collaboration has long been the keystone of Ozelius’s work. This, she said, combined with new technologies open up a broad range of possibilities to uncover new dystonia genes. “We now have the ability to look at the whole genome at once on a single chip and we can test a person’s DNA to generate a million points of data,” she said. “Here, at Mount Sinai, there are statistical geneticists integrated into the Department of Genetics and Genomic Sciences that can help me deal with this overwhelming amount of data.”

In addition to her laboratory work, Ozelius will also interact with Mount Sinai’s Neurology Department to recruit patients with the help of a genetics counselor. This newly endowed position was made possible through the Bachmann-Strauss Family Fund. “I am highly confident that Laurie’s research efforts will bring us closer to finding a cure,” said Bonnie Strauss.

After receiving a PhD in Genetics from Harvard Medical School in 1994, Ozelius carried out her postdoctoral training at Massachusetts General Hospital, eventually rising to the rank of Assistant Professor of Neurology at Harvard Medical School and Assistant Geneticist at Massachusetts General Hospital.

In 1999, she was appointed Assistant Professor in the Department of Molecular Genetics at the Albert Einstein College of Medicine in the Bronx, NY. Her laboratory focused on the genetics of movement disorders, in particular identifying genes that cause various forms of dystonia. She was involved in cloning two dystonia genes (DYT1 and DYT12) and has also mapped other dystonia genes to particular chromosomal regions.

She is looking forward most to the translational aspect of her new role. “Interacting with clinicians helps put my work in perspective because they can make observations based on my work,” she said. “But, ultimately, seeing patients is so inspiring. You want to do as much as you can do to help.”

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New Grants Awarded

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The gene with the mutations makes an enzyme that is the major consumer of energy in the brain: an ATP-hydrolyzing membrane pump called the sodium-potassium ATPase. Instead of having a slow degenerative disease, patients with RDP experience symptoms after physically stressful events or binge drinking.

Utilizing a mutant mouse that has one copy of the gene instead of two, Dr. Sweadner’s study aims to determine whether it will develop dystonia and Parkinsonism. The immediate benefit will be a mouse model of genetically-inherited rapid-onset dystonia-Parkinsonism, which can be studied for both prevention and cure.

For dystonia patients who do not respond to medical therapy, surgical treatment with Deep Brain Stimulation (Gpi DBS) can reduce symptoms. However, the precise mechanism by which this intervention works is unknown. Maren Carbon-Correll, MD at North Shore-Long Island Jewish Health System will study how brain metabolism and activation are influenced by DBS in this disease using the imaging technique positron emission tomography (PET) to measure glucose metabolism and blood flow in the brain. It is hoped that understanding which brain areas are influenced by effective DBS will help to optimize this treatment and will gain further insight into the mechanisms of the disease.

Once again, joint effort is at the fore in dystonia research. Cristopher Bragg, PhD at Massachusetts General Hospital will use a novel screening technique that identifies chemical compounds known as “small molecules” which bind directly to the human protein torsinA or to its dystonia-linked mutant form.

In a collaborative arrangement, he will send samples of compounds identified in his experiments to Dr. Xandra Breakefield at Massachusetts General Hospital and to Drs. Guy and Kim Caldwell at the University of Alabama for further testing. These scientists are studying different aspects of how mutant torsinA leads to dystonia. Dr. Breakefield’s group will determine whether Dr. Bragg’s compounds improve a functional defect which occurs in cells obtained from dystonia patients. The Caldwells will also test these compounds in an animal model based on the roundworm, c. elegans.

By looking for small molecules which prevent the effects of the mutant torsinA protein, the researchers hope to identify potential new therapeutic compounds for dystonia.

2007 GRANT FUNDING

Cristopher Bragg, PhD – Massachusetts General Hospital, Charlestown, MA
Small molecule microarray screening to identify novel ligands for torsinA

Xandra Breakefield, PhD – Massachusetts General Hospital, Charlestown, MA
Genetic intervention strategies for DYT1 dystonia

G. Caldwell, PhD & K. Caldwell, PhD – University of Alabama, Tuscaloosa, AL
Investigating small molecule therapeutics for torsion dystonia

Maren Carbon Correll, MD – North Shore-Long Island Jewish Health System, Manhasset, NY
Functional correlates of Gpi Deep Brain Stimulation in primary dystonia

Ellen J. Hess, PhD – Johns Hopkins University, Baltimore, MD
Generation of mouse model of L-DOPA-responsive dystonia (DRD)

H.A. Jinnah, PhD – Johns Hopkins University, Baltimore, MD
Why is dystonia more frequent in females than males?

Yuqing Li, PhD – University of Alabama, Birmingham, AL
Novel siRNA-based experimental therapy for DYT1 dystonia

Antonio Pisani, PhD – European Brain Research Institute, Rome, Italy
Characterization of striatal short and long term synaptic plasticity in a mouse model of DYT1 dystonia

Louis Ptacek, PhD – University of California, San Francisco, San Francisco, CA
Identification of the gene causing paroxysmal kinesigenic dyskinesia

Margaret Rice, PhD – New York University, New York, NY
Dopamine release in DYT1 transgenic mice

Kathleen Sweadner, PhD – Massachusetts General Hospital, Boston, MA
Mouse model of rapid onset dystonia-Parkinsonism and vulnerable basal ganglia neurons

Zhenyu Yue, PhD – Mount Sinai School of Medicine, New York, NY
Mouse model of human dystonia by BAC-transgenic approach
We are pleased to announce that James Marlas and John J. Pomerantz have been elected to join The Bachmann-Strauss Dystonia & Parkinson Foundation Board of Directors; Michele Tagliati, MD has been appointed to our Scientific Advisory Board.

Jim Marlas is chairman and founder of Union Capital Corporation, which targets strategic equity positions in certain private and public companies. He operates a private holding company with controlling interests in printing, packaging and marketing services. In addition, he is Vice Chairman of the New York City Opera and a member of the Chairman’s Council of the Metropolitan Museum of Art.

John Pomerantz is former Chairman and CEO of Leslie Fay. He is a founder and supporter of the Albert Einstein College of Medicine and serves on the Board of Overseers. In addition, he is a Trustee of the Fashion Institute of Technology, and is currently the Chairman of the Educational Foundation. John was the 2001 honoree of the Citymeals-on-Wheels Fashion & Beauty Tribute and is currently on the Executive Board of Citymeals-on-Wheels.

Michele Tagliati, MD is Associate Professor of Neurology at Mount Sinai School of Medicine and Director of its Parkinson’s Disease Center. A leader in the field of Deep Brain Stimulation (DBS) for Parkinson’s disease and dystonia, he currently serves as teaching faculty at the annual courses given on DBS programming at the American Academy of Neurology and the International Movement Disorders Society.

Dr. Tagliati is one of the authors of *Parkinson’s Disease for Dummies*, a new book published by Wiley. This guidebook explains how to find the right doctors, cope with daily issues, and provide care as the disease progresses.

**New Appointees to Board and SAB**

We are pleased to announce that James Marlas and John J. Pomerantz have been elected to join The Bachmann-Strauss Dystonia & Parkinson Foundation Board of Directors; Michele Tagliati, MD has been appointed to our Scientific Advisory Board.

Our annual symposium has long been regarded as the source of the most up-to-date information about dystonia and Parkinson’s disease. This past fall representatives from many of the area’s major hospitals participated in this program. They included: William Dauer, MD, Columbia University, Michele Tagliati, MD, Mount Sinai Medical Center, Mark Grove, MD, Beth Israel Medical Center, and Claire Henchliffe, MD, The New York Presbyterian Hospital.

This free event brought together more than 100 patients, families and healthcare professionals, who heard the latest about new treatment prospects and were able to ask questions of each of the presenters.

*We are grateful to Allergan and Teva Neuroscience, Inc. for sponsoring this important educational event.*
SAVE THE DATE
Monday, June 18, 2007
The Hedi Kravis Ruger Tournament
Mark your calendar now for the 15th Annual Dystonia & Parkinson’s Pro-Am Golf Invitational at the Century Country Club in Purchase, NY.
There will be a special performance at dinner by Christian Hoff – Tony Award winner for his role in the hit Broadway musical “Jersey Boys.”
For reservations or more information, call 212.241.5614 or email Bachmann.Strauss@mssm.edu

Our 5th annual Think Tank brought together an international group of noted scientists and clinicians to discuss the latest developments in dystonia research.