



# Outlook

WINTER 2010

## Dystonia Center of Excellence Launched at Beth Israel

For Bonnie Strauss, the inauguration of the Bachmann-Strauss Dystonia Center of Excellence at Beth Israel Medical Center in Manhattan was an emotional moment and “a dream come true”.

“As someone with dystonia, it took me seven years to be properly diagnosed,” said the president and founder of The Bachmann-Strauss Foundation. “I know there are still many people who have it and aren’t diagnosed. There are also people with Parkinson’s disease who have dystonia and don’t know it. All share a need for proper diagnosis and treatment. Most importantly, they need a Center to turn to that combines the best clinical care, support services, and research.”

The founding of the first-of-its-kind Center, headed by Susan Bressman, MD, was celebrated at a reception at Beth Israel Medical Center on October 7, 2009. Dr. Bressman is chairman, The Alan & Barbara Mirken Department of Neurology, Beth Israel Medical Center, and Professor of Neurology, Albert Einstein College of Medicine, New York City.



At the Dystonia Center opening reception: Stanley Brezenoff, President and Chief Executive Officer, Continuum Health Partners; Lawrence S. Huntington, Chairman Board of Trustees, Continuum Health Partners, Tom and Bonnie Strauss; Susan Bressman, MD; Harris Nagler, MD, FACS, Interim President, Beth Israel Medical Center.

### One-stop shopping

This multidisciplinary Center brings together a top team of neurologists, including pediatric neurologists, surgeons, psychiatrists, genetic counselors and others, as needed, to confer about a patient’s diagnosis and to develop a comprehensive treatment plan.

Our goal is to open additional Centers across the country. Funding will be needed to support this effort.

### Where to turn

**For more information contact:**

Joan Miravite, Nurse Practitioner  
212.844.6134 or [jmiravite@chnpnet.org](mailto:jmiravite@chnpnet.org)

*Bachmann-Strauss Dystonia Center of Excellence is located at  
Beth Israel Medical Center  
Department of Neurology  
Phillips Ambulatory Care Center  
10 Union Square East, Suite 5K  
New York, NY 10003*

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# Four Areas Highlight Foundation Research Portfolio

As we move into our 15<sup>th</sup> year, our scientific portfolio has steadily grown and streamlined to accelerate the pace of new treatments and bring us closer to finding cures. In addition, our Foundation continues to be the leading organization actively looking at the connection between dystonia and Parkinson's disease.

The grants we currently award can be categorized into four broad areas:

## Expediting drug discovery

Our novel AntiDystonia Drug Development Program, initiated less than a year ago, aims at propelling the discovery of new treatments, establishing a model testing procedure to study genetic and non-genetic models of dystonia, and identifying drugs that can either move directly into clinical trial or be put forward for product development by a bio-technology or pharmaceutical company.

## Getting access to care

The groundbreaking Bachmann-Strauss Dystonia Center of Excellence was created to provide something that has been sorely lacking for people with dystonia until now: a single place to go for accurate diagnosis, treatment and support. Its built-in research component will also help to more easily translate the findings of what researchers see in the laboratory to what clinicians see in their patients.

## Finding better outcomes for long-term treatment

Physicians and patients know all too well how difficult it is to find the right balance of medications. Generous leadership grants from The Dekker Foundation are targeted to finding effective treatments and are enabling researchers to analyze the complications that occur from traditional Parkinson's therapies.

## Individual grants

The "seed money" that we provide to fund innovative projects in their earliest stages of development has shown tremendous results including new gene discovery, treatment like Deep Brain Stimulation, and has furthered our fundamental understanding of dystonia and Parkinson's disease.

*The following new grants, awarded this past fall, were the second cycle of grants made in 2009.*

**Nicole Calakos, MD, PhD**  
*Duke University Medical Center  
Durham, NC*



*Behavioral Characterization of a Mouse Model for Torsin Dysfunction Based on a Novel Human Mutation*

After identifying a novel TorsinA mutation in an individual with late-onset focal dystonia, Dr. Calakos' group will use genetically-engineered mice to study its effect on neurological function. These experiments will reveal the behavioral consequences of Torsin dysfunction, e.g. whether dystonia, parkinsonism, or other abnormal behaviors result. This animal model may later help identify the cellular and circuit abnormalities giving rise to the behaviors and to test potential therapies.

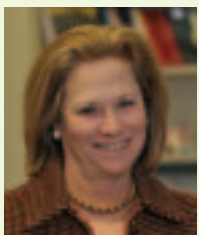
**Pedro Gonzalez-Alegre, MD**  
*University of Iowa,  
Carver College of Medicine  
Iowa City, IO*



*Exploring the role of oxidative stress on DYT1 penetrance*

This study helps explore why some DYT1 mutation carriers develop dystonia while others do not. In a process directed to maintain

## Looking to the New Decade



BONNIE STRAUSS  
FOUNDER AND PRESIDENT

2010 – a decade into the new millennium, a decade in which remarkable progress has been made in our understanding of dystonia and Parkinson's disease.

As we begin this New Year, we are celebrating the 15th anniversary of the founding of our organization. There is a great deal to be thankful for. Large and small, the support of so many people has made a tremendous imprint. Nationwide, people did whatever they could to ensure that we continue to make headway despite a very tough recession. A generous gift from an anonymous donor enabled us to establish the first-ever Dystonia Center of Excellence. This is a tremendous breakthrough for people with dystonia, who all too often search for years for proper diagnoses and care.

There is so much promising new research in the pipeline for both dystonia and Parkinson's disease that we begin this new decade with great optimism. I am so proud of all we have accomplished together and look to building on this success.

Thank you and all our very best to you for the New Year.

*Bonnie Strauss*

an adequate energy supply, neurons generate toxic molecules that can cause “oxidative stress.” TorsinA, the protein mutated in DYT1, might help maintain a balance between energy metabolism and oxidative stress, however the mutated protein could alter this process. This hypothesis will be tested by inducing oxidative stress in DYT1 mouse brain.

**Mark LeDoux, MD, PhD**  
*University of Tennessee  
Health Science Center, Memphis, TN*



*DNA binding sites for the transcription factor THAP1*

THAP1, the protein associated with DYT6 dystonia, is a transcription factor. Transcription factors regulate the expression of many genes. Continuing his work in this area, Dr. LeDoux’s studies are designed to identify DNA binding sites of THAP1 in mouse cerebellum at various stages of maturation. This work is designed to unveil dystonia-associated networks and may also identify candidate genes for dystonia.

**Laurie Ozelius, PhD**  
*Mount Sinai School of Medicine  
New York, NY*



*Application of next generation parallel sequencing to the identification of a gene for cervical dystonia*

New technology has been developed that allows for the complete sequencing of all of the coding regions in the entire genome. To assess its potential for dystonia gene discovery, a family that has eight members with cervical dystonia will be sequenced. If a mutation is identified the gene will be screened in other sporadic and familial patients with cervical dystonia.

**Antonio Pisani, MD**  
*Fondazione Santa Lucia, Rome, Italy*



*Evaluation of the role of dopamine and adenosine neurotransmission in corticostriatal bidirectional synaptic plasticity in DYT1 dystonia*

Synaptic plasticity represents the experimental paradigm of motor learning and memory. Dystonic symptoms are thought to be generated by an altered plasticity in the striatum, a brain region involved in motor control. Building on his previous research, which discovered a profound impairment in striatal synaptic plasticity, Dr. Pisani will now utilize pharmacological agents to restore normal plasticity processes in a model of DYT1 dystonia.

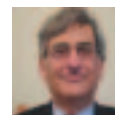
**Kristina Simonyan, MD, PhD**  
*Mount Sinai School of Medicine  
New York, NY*

*Structural biomarkers of primary focal dystonias*



Very little is known about the causes of primary focal dystonias. The aim of this study is to characterize disorder-specific brain abnormalities in people with focal hand dystonia, spasmodic dysphonia, blepharospasm and cervical dystonia using magnetic resonance imaging (MRI). This should help to better understand what brain changes might underlie the development of different types of focal dystonias.

**Amnon Sintov, PhD\***  
*Ben Gurion University of the Negev  
Beer Sheva, Israel*



*Transdermal continuous delivery of L-DOPA using a novel patch based on high-capacity, nanostructured soft micelles*

When levodopa is given orally, it is eliminated from the body quickly resulting in sharp fluctuations of its blood concentrations. These frequent changes in blood levels are considered to be responsible for the development of late motor complications. This study explores the potential clinical benefit provided by the transdermal delivery of levodopa to treat Parkinson’s disease and achieve a simple solution for the late motor complications.

*\* This grant was made possible through the generosity of The Dekker Foundation.*

## A Special Thank You

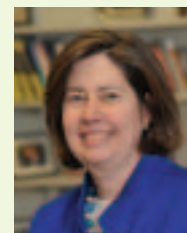
As some of you may know, I resigned in December as the Foundation’s Executive Director to become CEO of CancerCare, a national non-profit organization.

I want to thank each of you for your support of the Foundation and of me over the last five years. During this time, our scientific portfolio expanded greatly, and we broadened our outreach so donations now come in from all 50 states through a variety of new programs including Jake’s Ride, the Marathon team, and Clips for a Cure. None of the above could have happened without your support.

As I leave, please know how much I appreciate each of you. I greatly admire Bonnie Strauss for her leadership and passion to find a cure for dystonia and Parkinson’s disease. I thank the Board and SAB for their hard work and drive, and our staff for always being willing to take on one more project.

Thank you for the opportunity you have given me.

*Helen Miller*



HELEN MILLER  
EXECUTIVE DIRECTOR

# Collaborations

## Second Annual Jake's Ride Reaps Great Results



Jake Silverman at the finish line

In almost every sense of the word, it couldn't have been a better day. The second annual Jake's Ride for Dystonia Research, held Sunday, October 4 in Short Hills, NJ, raised a terrific \$205,000. This included gourmet food store chain Eden Gourmet Markets & Garden of Eden Markets' cause marketing campaign, which raised nearly \$6,400 and was instrumental in getting out word of the event.

More than 1,100 area residents rode bikes or walked to raise funds for research in honor of 13-year-old Jake Silverman and other children with dystonia. An outstanding local committee was key to making it all happen from recruiting volunteers, to making banners, and organizing food and prize donations.

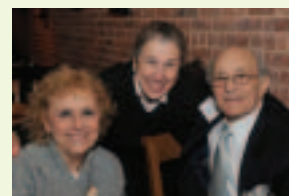
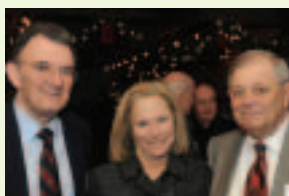
Prizes were awarded to: Ethan Stupp and Nathaniel Schwamm, the top two fundraisers; Robert Stone and Ben Caplan recruited the most team members; and Jared Seletsky and Jack Wenzel brought in the most donors.

Their efforts were truly inspiring: Twelve-year old Evan Bograd, raised \$1,300 for Jake's Ride as part of his Bar Mitzvah project. Jack Wenzel, who celebrated his 8th birthday by riding in the event, asked that contributions be made in his honor instead of getting birthday presents.



## Theater Benefit Sparkles

Theater-goers who attended our December benefit of "A Little Night Music" should take a bow for generating over \$67,000. The Broadway revival, which brought great reviews from our group, stars Angela Lansbury and Catherine Zeta-Jones.



Pictured from left: David Stone, Bonnie Strauss, and John Pomerantz; Mark Standish, Brian Bedol and Nick Patti; Alice Cohen, Patti Kenner, and Cliff Cohen

## TEAM Bachmann-Strauss Raises \$161,185 in Marathon

What a team, what an outcome! Forty-three runners gave it their all to raise funds for our organization in the ING New York City Marathon, held Sunday, November 1.

The 2009 team was chaired by Rob Lefkowitz, who ran for a second year. Seven of the participants were repeat runners, including Darwin Johnson, who ran with his father, Darwin, Sr., who has Parkinson's disease.

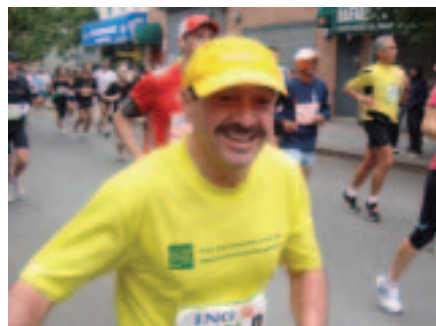
Sagar Mehta, the first to apply this year, raised more than \$35,000 alone. His grandfather, who was instrumental in

moving his family from India to the United States, had Parkinson's disease. Mehta said that being a part of TEAM Bachmann-Strauss and the marathon was "an amazing experience", and he will chair the team next year.

Our Foundation is an official Charity Partner of the world-famous marathon. Initiated four years ago by Brian Dugan, TEAM Bachmann-Strauss has grown from 10 runners that raised about \$30,000 to a team of 43 that raised more than \$160,000. This year's proceeds will fund two research grants.



Pictured above, from left: Angela Johnson and Amy Lestage; top right, Daniel and Kristen Rosenbaum; bottom right, Sagar Mehta; bottom left, Jean Magne



# Think Tank Debates Genetic Screening and Clinical Practice

*"Genetics and Biology of Dystonia" was the focus of our eighth annual Think Tank on Dystonia this past November as scientists, clinicians, and geneticists came to New York from around the world to discuss the latest research and exchange ideas.*

This preeminent, international group reviewed a wide spectrum of promising new data, but it was the discussion of whether genetic findings are shaping clinical practice that sparked the broadest range of opinion.

While anecdotal evidence and early research suggest that the different subtypes of dystonia may respond to different treatments, the discussion indicated broad variance in how doctors currently use genetic testing in their practices, e.g., if they do send dystonia patients for genetic screening, whether the findings are used to make treatment decisions or, instead, to inform a diagnosis.



**Kailash Bhatia, MD, DM, FRCP** from London, England

## Future directions

The discovery of genes that reveal the biological pathways involved in dystonia and the development of targeted treatment continue to be central to much of

the new work underway.

While discovering the differences between the subtypes of dystonia may be key to making a diagnosis, clinicians are interested in the commonalities that can be used to find therapies that treat as many patients as possible.



**Masaya Segawa, MD, PhD** from Tokyo, Japan

"Sharing patient and genetic material will be needed during the next stage of genetic analysis," said Ted Dawson, MD, PhD of Johns Hopkins University School of Medicine and Chair of our Scientific Advisory Board. Laurie Ozelius, PhD of Mount Sinai School of Medicine added that "Worldwide collaborations in genetic research will be paramount, particularly for the more common focal dystonias."

*Read more. An executive summary of our recent Think Tank on Dystonia is on [www.dystonia-parkinson.org](http://www.dystonia-parkinson.org).*

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## Dystonia Coalition Forms

A \$6 million commitment over the next five years by the National Institutes of Health (NIH) has led to the establishment of a new Dystonia Coalition, a collaboration of scientists, institutions, patient advocacy organizations and the National Institutes of Health – all united to advance clinical research for dystonia.

The NIH commitment will support a Dystonia Coalition of 34 centers in the United States, Canada and Europe. This is the largest sum ever provided for a single project devoted to clinical and translational research in the dystonias. The Coalition was established as part of a \$117 million expansion of the NIH's Rare Diseases Clinical Research Network, which will coordinate research on more than 95 rare diseases.

H.A. Jinnah, MD, PhD, professor of neurology and human genetics at Emory University School of Medicine, will direct

the Dystonia Coalition; Joel Perlmutter, MD, professor of neurology and radiology at Washington University School of Medicine, is co-director.

"I am very pleased that the NIH has recognized our program for clinical and translational research in dystonia with this big award. Lots of folks worked really hard to make it happen," said Dr. Jinnah. "We hope to be able to catalyze the development and testing of new treatments. It's an exciting time for clinical research, and I'm happy to be a part of it."



**H.A. Jinnah, MD, PhD**

Our Foundation, which has previously provided funding to Dr. Jinnah's work, is a member of the Coalition.

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Louis Bachmann (1916-2000)  
Bonnie Strauss

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## Evidence of Dystonia/Parkinson's Link Increases



*A variety of studies point to a link between dystonia and Parkinson's disease, and, according to Susan Bressman, MD at Beth Israel Medical Center, our understanding of the brain pathways that link parkinsonism and dystonia is increasing all the time.*

*"Most dystonia patients don't have parkinsonism or the signs of Parkinson's disease, although there are dystonia subtypes, some genetic, where parkinsonism and dystonia occur together," Dr. Bressman explains.*

*On the other hand, she says, most Parkinson's patients have elements of dystonia. "This is especially true in people with early-onset Parkinson's disease, and can be seen in hand posturing and toe curling that may occur before treatment is begun. It also commonly occurs as a drug-induced dyskinesia." Dyskinesia is the uncontrollable movement that is a frequent side effect of treatment for Parkinson's disease patients.*

## Symposium Provides Forum for Update and Questions

Laura Dean-Frazier came to our Symposium on Dystonia and Parkinson's disease for the first time this past October to learn more about Deep Brain Stimulation surgery and whether it could help her 14-year-old son, Dillon.

This mother of three was among more than 100 people who came to learn the latest in the field. For many, it was a unique opportunity to ask questions of the doctors and scientists and to network with others who are experiencing some of the same problems. "It has been very helpful to hear what people had to say and to get the most current information," Ms. Dean-Frazier said.



Laura Dean-Frazier

The topics focused on at this educational event included: dystonia classification, treatment and genetics, an overview of the new Bachmann-Strauss Dystonia Center of Excellence, Deep Brain Stimulation to treat dystonia and Parkinson's disease, the effects of Parkinson's disease and dopamine medications on learning and decision making, and new treatments for movement disorders. The symposium was provided free to patients, families and health care professionals.

Our thanks go to our presenters: Mark Gluck, PhD, Rutgers University – Newark; Claire Henschcliff, MD, DPhil, Weill Cornell Medical College; Nutan Sharma, MD, PhD, Massachusetts General Hospital; Vicki Shanker, MD, Deborah Raymond, MS, CGC, Joan Miravite, NP, and Lawrence Severt, MD, PhD all from Beth Israel Medical Center.

*We are grateful to Allergan for their sponsorship of this important event.*



**THE BACHMANN-STRAUSS**  
Dystonia & Parkinson Foundation, Inc.

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[www.dystonia-parkinson.org](http://www.dystonia-parkinson.org)

**SAVE THE DATE**  
**Monday, June 21, 2010**  
**The Hedi Kravis Ruger Tournament**

Mark your calendar now for the 18th annual  
Dystonia & Parkinson's Pro-Am Golf Invitational  
at Century Country Club in Purchase, NY.

*For reservations or more information, call  
212.682.9900 or email [bpfeil@bsdspf.org](mailto:bpfeil@bsdspf.org).*

*Come Celebrate the Foundation's 15th Anniversary*

*The Bachmann-Strauss Dystonia & Parkinson Foundation, Inc. was established in 1995 to find better treatments 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.*