Interview With Dr. Gabrielle Wilson 
2015 PMDF Grant Winner

by Mary Ann Chapman, PhD

Dr. Gabrielle Wilson and her colleagues have added yet another culprit to the list of genes that cause Parkinson’s disease. This gene, known as RAB39B, is altered in a small group of people with Parkinson’s disease. The genetic alterations somehow cause nerve cells to die, and Dr. Wilson and her colleagues want to know why.

Dr. Wilson, a researcher at Murdoch Children’s Research Institute in Australia, is one of three 2016 PMDF grant award winners and I asked her, via e-mail, to tell us a little bit about her research and interest in movement disorders.

What research will you be pursuing with the PMDF grant?

Currently, very little is known about the normal role of RAB39B in the body. Our ongoing studies will investigate the normal function of RAB39B in cells and examine how the loss of RAB39B contributes to the development of Parkinson’s disease.

How did you and your colleagues discover the link between RAB39B and Parkinson’s disease?

The link was originally discovered based on a family in Australia, in which three members showed symptoms of intellectual disability in childhood, followed later by the motor symptoms that characterize Parkinson’s disease. This pattern of symptoms is different from that usually seen in Parkinson’s disease, in which the motor symptoms appear when a person is in his or her 50s or 60s, and may or may not be associated with problems in thinking. In the Australian family, blood tests showed that the affected members had a loss of DNA in the RAB39B gene. This loss of DNA prevented the gene from making a functional protein. Later we identified a family in the United States that showed a similar pattern of symptoms, and we confirmed that these members also had alterations in the RAB39B gene. These findings led to our current work, which is to investigate the normal function of the protein made from the RAB39B gene. This work will hopefully help us...
understand why people get Parkinson’s disease and lead to new treatment methods.

**What are some of the advances you have seen in movement disorder research?**

Modern genetic screening techniques have led to major advances in the identification of genetic changes that underlie movement disorders. The genetic changes have provided substantial insight into the brain pathways affected in movement disorders. Knowledge gained from genetic studies is important for understanding both inherited (ie, passed on from one generation to the next) and idiopathic (ie, of unknown cause) forms of disease because many experimental findings are common to both. This suggests that the same physiological pathways are disrupted in people with inherited and idiopathic disease.

**What are some of the questions we need to answer in the future regarding movement disorders?**

Research is currently delineating the mechanisms underlying disease progression. The information gained from these studies can then be used to determine if we can identify one or more robust biomarkers that will enable accurate diagnosis before people develop symptoms. Early diagnosis and intervention are important for the delay or prevention of disease. For example, by the time symptoms such as tremor develop in people with Parkinson’s disease, significant damage is already present within the brain. Thus, diagnosis and intervention would ideally occur prior to the onset of symptoms.

**What do you think it is important for patients and/or laypeople to know about research and/or movement disorders?**

Scientific research and subsequent discoveries would not be possible without the participation of patients, affected families and healthy individuals. The contributions of study participants can vary widely depending on the study, including completing surveys, to undergoing scans (such as an MRI), or providing biological samples. However, all of these contributions by participants are crucial to gaining insight into both normal and disease physiology. This insight will ultimately advance the development of new therapies.

**How will the PMDF money help you to pursue your research goals?**

The money kindly provided by the PMDF will contribute to the cost of resources and reagents that are essential to complete experiments that will be applied in our investigation of RAB39B function.
Dear Friends of PMDF,

I recently read an excellent book about Parkinson’s disease: *Brain Storms: The race to unlock the mysteries of Parkinson’s disease*, by Jon Palfreman. The author was diagnosed with PD in 2012. He read everything he could about Parkinson’s disease, and spoke to researchers and clinicians to understand his condition. He says, “As a lifelong science journalist who had reported on this disease, I was better placed than most to figure out the state of Parkinson’s research and ascertain what kind of future I faced.”¹ This book is the result of that research. It’s about the history of PD research and the exciting things that are being explored now. But it is also a personal memoir of a person with Parkinson’s.

In the Spring, 2012, issue of this newsletter I wrote about 23andme, a genetic testing service. They were conducting a research study on the genetics of Parkinson’s Disease, and offered free DNA testing to people with Parkinson’s disease. That study was suspended for a time, but now the offer is available again. If you have been diagnosed with Parkinson’s disease and are interested in this research, visit [http://23andme.com/pd](http://23andme.com/pd) for more information. (PMDF is not affiliated with 23andme, and makes no recommendations about them.)

Spring is here, and that means it’s time for the Zent-A-Thon, our annual 5K run/walk. Details are elsewhere in this newsletter; be sure to read about it and sign up! And don’t forget to read the other articles in this newsletter, about the Dystonia Advocacy Network and the work of one of our grant recipients.

Sincerely,

Mark Wadsworth
President

Interested in Becoming a Legislative Advocate for Dystonia? Let Us Introduce You to DAN

by April Ingram

DAN, the Dystonia Advocacy Network, is an organization that was formed in 2007, to advocate on behalf of issues that impact the dystonia community. DAN brings together individuals affected with all forms of dystonia to speak out with a single, powerful voice on government and public policy issues. This strategy works to leverage available resources to deliver maximum effect on legislative initiatives. DAN believes that we all have the power and ability to educate legislators and influence policy makers who are responsible for the areas of government that impact people with dystonia.

This devoted network of advocates has developed relationships with their legislative leaders to help them better understand the challenges of people living with all types of dystonia. These relationships have not only improved understanding and awareness, but have also created champions of the cause and a system of legislative supporters.

DAN carefully monitors and tracks legislative issues that may affect the dystonia community and implements communication and action strategies to achieve their goals. Examples of government issues have included the allocation of medical research funds and improving access to treatments. Once a particular issue has been identified and action taken, DAN members continue to evaluate the effectiveness of their approach and related activities and see if further work or a change in strategy is needed.

A fundamental part of the network’s advocacy strategy has been to encourage Congress to significantly increase funding for the National Institutes of Health (NIH) and support expansion of the dystonia research portfolio at NIH. The results of these efforts can speak for themselves, as funding for dystonia research at the NIH grew from approximately $10 million annually to just under $20 million, which also includes a $6 million, 5-year clinical research initiative, known as the “Dystonia Coalition”.

Another key area for DAN is supporting research for dystonia-affected veterans. Published medical literature has identified a link between traumatic injuries and the onset of dystonia, making this an ongoing concern for past and present military personnel and their families. DAN has worked tirelessly to ensure that the Department of Defense (DoD) and its medical research program are stakeholders in the fight to improve treatment options for these veterans. A significant step forward for this issue was having “dystonia” added to the list of conditions deemed eligible for study through the DoD Peer-Reviewed Medical Research Program in the FY 2010 Defense Appropriations Bill.

Each spring, dystonia advocates from across the country gather in Washington, DC, for a two-day advocacy event. During this time, participants hear about updates on dystonia research from leading investigators, receive comprehensive advocacy training, and have the opportunity to network with other dystonia-affected individuals from across the country. DAN members provide training and preparation for volunteers to meet with government leaders and effectively communicate key messages. Dystonia Advoca-
Dystonia Advocacy Day culminates with a trip to Capitol Hill so that participants can meet with the offices of their Members of Congress and urge their support on significant legislative issues. This event is an important opportunity for the dystonia community to make its collective voice heard throughout the halls of Congress and it is always well attended by individuals with all forms of dystonia. Without exception, volunteers who have participated in these events in the past have found Dystonia Advocacy Day to be an incredibly rewarding and empowering experience.

To learn more about the Dystonia Advocacy Network, please visit [www.dystonia-advocacy.org](http://www.dystonia-advocacy.org).

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**2016 Zent-a-thon Fundraiser**

The Parkinson’s and Movement Disorder Foundation and The National Spasmodic Torticollis Association are hosting the 6th Zent-A-Thon 5K Walk/Run Fundraiser to raise funds and help spread awareness of Parkinson’s and movement disorders. The fundraising event will take place at Mile Square Park in Fountain Valley, California, on Saturday, May 21, 2016. This is a great opportunity to get outdoors with family and friends, support the two organizations, get some health benefits, and raise awareness. This will be a fun and friendly family event that everyone can enjoy. Kids under 13 do not need to pay the registration fee.

Prizes will be awarded to the 1st, 2nd, and 3rd persons to cross the finish line. All participants who register will automatically be entered in a raffle and have the opportunity to win gift card prizes ranging from $5 to $50. There are over twenty five prizes being raffled off! In addition to winning prizes, registered participants will also receive lunch catered by Katella Deli Restaurant and Bakery located in Los Alamitos, a T-shirt, and beverages.

We would like to give a special thanks to the following companies for their generous sponsorships: Katella Deli Bakery and Restaurant, AbbVie, Avanir Pharmaceuticals, LivHome, US WorldMeds, and UCB Pharmaceuticals.

If you are in Orange County, LA County, or San Diego County, bring your family and friends and participate in the Zent-A-Thon! You can walk the 5K, run the 5K, or both.

Street parking is available around Mile Square Park, meter-free. If you park your car on the grounds, parking is $5 per vehicle.

You can register online by visiting [www.pmdf.org/events.php](http://www.pmdf.org/events.php) or by mail using the form on the last page in the newsletter.

Due to park regulations, registration cannot be processed at the park. Please register by May 18.
Zent-A-Thon 5K Walk/Run Fundraiser

Mile Square Park
Fountain Valley

Saturday, May 21, 2016
8:30 am: Sign-In
10:00 am: Race Start Time
11:00 am: Lunch Available

First Place Prize - 2 Disneyland Tickets
Second Place Prize - TRX Suspension Training Pro Pack
Third Place Prize - $100 Macy's Gift Card

Chance to win a prize in raffle drawing. Raffle prizes include: Katella Deli Gift Card, Amazon Gift Card, Starbucks Gift Card, iTunes Gift Card, Target Gift Card & More

$20 – per person to register
$5 – parking inside the park
Free – street parking
Lunch, T-shirt and beverages are provided for participants

Sponsors
NAME: _________________________________________________________________________
ADDRESS: ______________________________________________________________________
CITY: _____________________________________STATE:__________ZIP:_________________
EMAIL: ________________________________________PHONE: _________________________

5K T-SHIRT SIZE (Circle One)  Adult:  M    L    XL    XL
Minimum age of entry 13

IN CASE OF RAIN: The race will proceed as planned. We reserve the right to change the date under extreme circumstances.

ADDITIONAL DONATIONS
Donations are tax-deductible and an acknowledgement letter will be sent to the donor for tax purposes

Contributor Information

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ENTRY FEE(S) OR DONATION

Entry Fee: $_________ ($20)
Donation: $_________
Total: $_________

My Employer has a Matching Gift Program: $_________

Please make check payable to PMDF
Visa ( )  MasterCard ( )

Mail Entry Form & Payment to:
NSTA
9920 Talbert Street
Fountain Valley, CA 92708

RELEASE FORM (all registrations must be signed)

I hereby waive any and all claims against NSTA, PMDF, event sponsors, personnel, and all other persons, firms, corporations and/or entities or anyone associated with this event, their respective or successors, for any injury or claims for damages that I may suffer from participation in this event. I grant full permission for organizers to use photographs, videotapes, recordings or any other record for this event.

____________________________________________
Signature

____________________________________________
Date

____________________________________________
Signature of parent or guardian (if under 18 years old)

____________________________________________
Date
OUR MISSION

To support basic and clinical research into the causes, treatments and cures for Parkinson’s disease and other movement disorders such as dystonia, myoclonus, spasticity, and tremor.

The Parkinson’s and Movement Disorder Foundation is committed to working with other organizations that have similar philosophies in an effort to bring together expertise from both basic and clinical science perspectives.

We are dedicated to enhancing the quality of life for those who suffer from movement disorders and their families, through research, education, and community outreach.