



# COLUMBIA UNIVERSITY IRVING MEDICAL CENTER PARKINSON NEWSLETTER

Summer 2019

## The Genetics of Parkinson's Disease

By Roy Alcalay, MD, MS Alfred and Minnie Bressler Associate Professor of Neurology

**What is the role of genetics and PD?** Our understanding of the mechanisms that lead to PD has significantly improved in the past two decades. While we still do not know exactly what causes PD, we believe that a combination of genetic and environmental factors are the cause.

The extent to which each genetic or environmental factor is involved varies from person to person. In some families, changes (or mutations) in certain genes are inherited or passed down from generation to generation. Mutations in a few of these genes may cause familial PD (in a recessive or dominant inheritance), but these are relatively rare and are often present in people with young-onset PD. In addition, there are genetic risk factors that increase PD risk. Carriers of these risk factors may be at higher risk for PD (e.g., 5 fold increased risk). Still, we estimate that most of those who carry these risk factors will not develop PD. The most important genes that are linked to increased risk for PD are glucocerebrosidase (*GBA*) and *LRRK2*. Our ability to identify these risk factors varies by ethnic groups, and in selected populations, such as Ashkenazi Jewish and North African Arab Berber populations, we can identify a *GBA* or *LRRK2* mutation in over 1/3 of people with PD.

Researchers around the world, including at Columbia University, try to better define the PD symptoms of people with PD who carry such mutations. Based on large scale studies comparing motor symptoms of PD patients with the *LRRK2* G2019S mutation and non-mutation carriers with PD, data showed slower progression of cognitive and motor symptoms in the *LRRK2* G2019S group. In contrast, recent studies on *GBA* demonstrate that non-motor symptoms appear to be more prominent among *GBA* PD patients than in PD patients without *GBA* mutations. Like other people with PD, carriers of *LRRK2* and *GBA*, will receive motor-symptom benefit from carbidopa-levodopa or other dopaminergic medications. Columbia University was among the leading centers in researching these genes.

**What is genetic testing?** Genetic testing, which involves a simple blood draw, is a type of medical test that identifies changes, or variants, in genes.

Genetic testing can inform whether you carry known gene changes linked to the disease. Up to 10% of people with PD have a genetic form of the disease, so by better understanding how those with genetic forms of PD experience symptoms related to Parkinson's and respond to treatment, scientists can begin to develop improved treatments and personalized medicine.

**What is the importance of genetics and PD?** Genetic research has made great strides to help better understand the biology of Parkinson's and guide the development of treatments for all people with PD. In the near future, knowing a person's genetic background may help predict the most effective treatments precisely tailored to an individual's unique experience with PD.

**How do I get genetic testing for PD?** While there are many clinical research studies and at-home testing kits it is critical to consult your physician if you are interested in learning more about genetic testing for PD.

For more questions or to learn more about genetic testing opportunities at Columbia, contact Amanda Chan at: [akc2177@cumc.columbia.edu](mailto:akc2177@cumc.columbia.edu) or 212-305-4233.



## Support Groups!

The Department of Neurology currently hosts two support groups: **PD Patient Support Group.** Meets the last Tuesday of every month. For more information and to register please contact Elizabeth Delaney, LMSW at 212-305-5779.

**Deep Brain Stimulation (DBS) support group.** For information regarding the next DBS meeting, please contact Linda Winfield, RN at 212-305-1303.



## Center Spotlight! : New PD Study on Efficacy of Haptic Wearable Device for Tremor

### What is this study?

This study evaluates the use of a wrist-worn device (also known as the Emma watch) developed to improve handwriting and hand function for individuals with action tremor. The device is lightweight (similar to a Fitbit or Apple watch) and delivers vibrations through six small electromagnetic mechanical stimulators, three on each side of the device.

### What is involved?

The study involves two visits to the Neurorehabilitation Research Lab at Teachers College, Columbia University (525 W 120th St), for up to 90 minutes each visit. During each visit, handwriting and hand function are evaluated with and without the wrist-worn device.

### Who can participate?

We are recruiting individuals who have Parkinson's disease or essential tremor who have action tremor. An action tremor is one that occurs during active voluntary movement and people with action tremor commonly report tremors that interfere with tasks such as eating, drinking, and writing.



If you are interested in volunteering to participate in the study or would like more information, please contact the **Neurorehabilitation Research Lab** at **212-678-3916** or by email at [neurorehablab@tc.columbia.edu](mailto:neurorehablab@tc.columbia.edu)

### Division of Movement Disorders

Columbia University  
Irving Medical Center  
710 West 168th Street  
New York, NY 10032  
212-305-1303

[www.columbianeurology.org/patient-care/movement-disorders](http://www.columbianeurology.org/patient-care/movement-disorders)

### Wish to Donate?...

Should you be interested in discussing how you can help support our clinical, research, and patient care activities, please contact Matt Reals, Senior Director of Development, at 212-304-7203 or [mr3134@columbia.edu](mailto:mr3134@columbia.edu)

## The Doctor Is In....Tarrytown!

We would like to invite all who are interested to a FREE upcoming event in Tarrytown, NY. Come join an open discussion with experts in the treatment of Parkinson's disease. Speakers include Dr. Cheryl Waters, Dr. Nora Vanegas-Arroyave, Dr. Blair Ford, and neurosurgeon Dr. Guy McKhann II. We will be discussing Parkinson's disease treatments, how diet can help Parkinson's disease, and deep brain stimulation.

There will be light refreshments served. Registration is required. Below is more information about the event:

**Date:** Thursday, August 1, 2019

**Time:** 6:00PM-8:00PM

**Location:** Westchester Mariott, 670 White Plains Rd., Tarrytown, NY 10591

**RSVP to Elizabeth** by e-mailing [ead2179@cumc.columbia.edu](mailto:ead2179@cumc.columbia.edu) or **212-305-5779**



Cheryl Waters, MD  
Movement Disorders  
Neurologist

Nora Vanegas, MD  
Movement Disorders  
Neurologist

Blair Ferd, MD  
Movement Disorders  
Neurologist

Guy M. McKhann II, MD  
DBS Neurosurgeon

If you have a question regarding Parkinson's and its treatment that you would like featured in the next newsletter, please e-mail your question to Elizabeth Delaney, LMSW at [movementdisorders@columbia.edu](mailto:movementdisorders@columbia.edu)

The information published in this newsletter is not intended to replace, and should not be interpreted or relied upon, as professional advice, whether medical or otherwise. Please refer to your own professional for all advice concerning legal, medical, or other matters published in connection with this article.

*"Don't count the days, make the days count."*

*-Muhammad Ali, diagnosed with PD in 1984*