Penn Medicine

The Transmitter

Parkinson's Disease and Movement Disorders Center (PD&MDC) Newsletter of Penn Medicine

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Research, Genetic Testing and PARKINSON'S DISEASE

This edition of The Transmitter *focuses on current research opportunities as well as genetic testing and the importance of molecular and genetic markers of Parkinson's disease (PD).*

Although only 10 to 12 percent of people with PD have a genetic cause for developing the disease, new findings offer exciting possibilities in Parkinson's research. Genetic testing can identify inherited markers of PD so patients can learn the likelihood of whether or not they – or their children and grandchildren – will develop the disease. With this knowledge, patients can prepare and start treatment early. "From A Patient Perspective" is about a patient who did just that, for herself and for her family.

In addition, this issue features a new program using music and memory (see "Tuning into Therapy") as well as upcoming events this fall.

RESEARCH OPPORTUNITIES *for PD*

By: Thomas F. Tropea, DO

Research and clinical trials teach providers about the intricacies of disease and its treatment. Were it not for ongoing studies, we would struggle to help patients. But here at Penn's Parkinson's Disease and Movement Disorders Center (PD&MDC), we have a robust clinical trial program to study the newest PD therapies currently in development. Our ongoing research helps broaden our understanding of the disease process so that we can improve the diagnosis of PD and develop better treatments.

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RESEARCH OPPORTUNITIES *for PD*

Looking at Groups of Patients

We have long recognized that no two people with PD are alike. Patients have big differences in their symptoms, responses to medications, and to the side effects of treatment. And so, going forward, we are building on our past experiences to define groups of people with PD. By understanding how one's genes or levels of different molecules, such as proteins, contribute to these differences, we can better understand PD. From this knowledge, we can bring new approaches to clinical research.

Using laboratory techniques to characterize the biology of PD, we can better define what makes people with PD differ from one another. This information is key to improving the way we make diagnoses and to developing ways of predicting the best therapy for each patient. This approach is called precision medicine because treatments are tailored to the specific patient.

The MIND Initiative

One of the most exciting new directions in PD research is made possible through a new center at the University of Pennsylvania called the Molecular Integration in Neurological Diagnosis (MIND) initiative, led by Dr. Alice Chen-Plotkin and Dr. Thomas F. Tropea. The MIND initiative is a way to bridge the gap between patient care and the cutting-edge research that is happening in the laboratory.

The MIND initiative is an observational study; we will collect medical information and a blood sample from participants, use this sample to molecularly characterize them, and watch the study group over time. This allows Penn scientists to use laboratory techniques to study genes, proteins and other molecules from patients with PD. Our study will be the largest collection of people with PD in the world to have this level of molecular characterization!

The purpose of MIND is to find out how each subject's molecular fingerprint characterizes their PD diagnosis so we can select the type of therapy each individual should receive. As we are able to target treatment more effectively, we can improve patient care and accelerate the development of newer therapies.

Join the MIND Initiative

Over the next year we will approach you at the end of your doctor visit to ask if you are interested in this study. We hope that you will be willing to participate in this exciting new research approach.

GBA Gene Research – the MOVES-PD Trial

Certain individuals with PD carry a mutation in a gene called GBA, which is considered a risk factor for PD. A new therapy is being tested that targets the GBA pathway. Our clinical trial program is enrolling people with PD who carry a mutation in the GBA gene.

The MOVES-PD trial is exciting because it is the first of its kind to target a specific group of people with PD who are defined by a genetic mutation. Some people already know their GBA status from doing genetic testing in a doctor's office or through 23andme or another direct-to-consumer genetic testing company. The PD&MDC can also perform genetic testing if you have a family history of PD or a rare neurological disorder called Gaucher's disease.

If you qualify for the MOVES-PD study, you might be selected to receive either a study drug or a placebo (a sugar pill) for up to three years to determine if the medication is effective at slowing the progression of disease. Dr. Meredith Spindler and Dr. Thomas Tropea are leading this study at the PD&MDC. Whether you already know your GBA status or want to learn your status and are interested in this study, please contact Sue Reichwein at 215.829.7273.

The only way for us to push the boundaries of our knowledge of PD is through scientific research. Conducting research requires the generosity of patients to dedicate their time and effort. Participating in research can be a truly rewarding experience for both patients as well as researchers. Through research, patients can play a central role in advancing science and in developing new therapies. Working together, we can expand our knowledge about PD and continue our march towards a cure.

Genetic Testing in PD

By: Thomas F. Tropea, DO, and Tanya Bardakjian

For many years, physicians and scientists believed that Parkinson's disease (PD) was a sporadic disorder, meaning that there were no causes or patterns that we could identify. However, about 20 years ago, pioneering research in the field of genetics shattered this idea by identifying the first gene associated with PD. This set off a cascade of new research that expanded our understanding of the role genetics plays in the risk of developing PD.

We now believe that about 10 to 12% of people with PD have a genetic cause. This number is higher for individuals with early-onset PD or disorders that mimic PD such as dementia with Lewy bodies, progressive supranuclear palsy, corticobasal degeneration or multiple system atrophy. Unfortunately, this information has not yet altered the way patients are treated, although this is likely to change in an important way in the near future.

New therapies are being developed for people who have certain genetic changes associated with PD. This is why understanding the genetics of PD has become so much more important and why you need to know the basics in order to become more aware of the possibilities for you and your family.

What Is Genetics?

Genetics is a field of medicine that studies ways in which diseases may be more likely in different generations of the same family. Inherited diseases occur by passing on a mutation, or defect, in a gene from parent to child. We inherit half of our genetic material from each of our parents. A gene is a sequence of DNA that serves as the blueprint for the various functions of our bodies.

A genetic mutation can change the way the gene functions and, in turn, can affect our health. Some genetic mutations can lead directly to a disease, such as in Huntington's disease.

However, most genetic variations associated with PD do not carry a 100% chance of getting PD, and most genetic mutations that have been identified only raise the risk of getting PD by a very small amount. This means that even if you have a genetic variation known to affect the risk of PD, you are not guaranteed to get the disease. It is important to remember that not all diseases are transmitted from one generation to the next. In order to know if you carry a genetic mutation you would need to undergo genetic testing.

What Is Genetic Testing?

Genetic testing is a medical test used by physicians, geneticists and genetic counselors to examine a person's genetic material, or DNA. This type of testing identifies changes in your DNA that are associated with specific diseases or symptoms and can be done on saliva or blood.

There are more than 1,000 genetic tests currently used in patient testing and more are being developed. A genetic test can look at a number of different aspects of your DNA. One way is to examine the structure of long lengths of DNA, called chromosomes, to identify large changes in the DNA. Another type of test, called molecular genetic testing, looks at regions of the DNA to identify mutations in the DNA sequence, or the order in which the genes are laid out. Most of the time, a handful of different tests are performed together, and a genetic counselor can help to identify the most appropriate tests for you.

What Could I Learn From Genetic Testing?

You may wish to have genetic testing for PD in order to learn whether you yourself may be at risk or whether you may carry the variation and your family members might be at risk. If you learn that you have a genetic variation associated with PD, you or your family members may be candidates to participate in research studies. In some cases, you may even be eligible to participate in clinical trials using medications for people with PD who have specific genetic variations. However, only a small percentage of people with PD have a genetic variation associated with the disease.

Should I Have Genetic Testing?

Genetic testing may be appropriate for you if you have a relative with similar symptoms. However, sometimes symptoms vary considerably between people who carry the same genetic change, and you and your family members might even be unaware of relatives who have PD symptoms. It is best to discuss testing in advance with your physician, or meet with a genetic counselor to identify the best path forward.

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Genetic testing is a medical test used by physicians, geneticists and genetic counselors to examine a person's genetic material, or DNA.

The decision to undergo genetic testing is a personal one with both risks and benefits. It is important to understand what you might gain or lose by learning your genetic status, in order to determine if genetic testing is right for you.

Some of the potential benefits of genetic testing are the possibility of learning information about your genetic status, and the opportunity to educate other family members about your status. However, this type of test is not without potential risks. Genetic testing, or the results of genetic testing, may increase anxiety and stress for some people, and may have social and financial risks. Therefore, it is important to consider all of these ramifications before pursuing genetic testing.

How Do I Get Genetic Testing?

The process of testing itself is very simple, although the results and the various consequences of them can be complex. Genetic testing can be performed through the PD&MDC, and your doctor can request a consultation with our neurogeneticist, Dr. Pedro Gonzalez-Alegre, and our genetic counselor, Tanya Bardakjian. A genetic counseling session will review the risks and benefits of genetic testing and help you understand the costs, if any, associated with these tests before you take them. These tests can be performed by drawing your blood or taking a saliva sample or cheek swab. Most commercial insurance companies currently cover clinical genetic testing.

Another approach that we cannot ignore is the explosion in direct-to-consumer genetic testing. Many people are persuaded to get this popular type of test in order to research information about where they came from and who their ancestors are. These are genetic tests that are not specific to you or your symptoms. You can purchase and mail a kit to a company such as 23andme and AncestryDNA™ that will perform a limited set of genetic tests, and some of the results may be relevant to movement disorders. If you have had this type of test, you can bring the results to your next office visit with your movement disorders doctor.

I Had Genetic Testing. What Do I Do Now?

If you have already done genetic testing and are unsure of the results or how to interpret the results, please speak with your doctor or meet with a genetic counselor. If genetic testing revealed a variation in a gene related to your PD symptoms, your physician and the genetic counselor could provide you with additional information related to this finding. Depending on the results, you may also be eligible for research studies targeting groups of people who carry mutations in specific genes.

We are still in the early days of being certain about how to use the information we get from genetic testing. If your results are negative, it does not mean you do not have a genetic variation related to your symptoms. Through further research we hope to learn more about different genetic mutations associated with your symptoms.

FROM A PATIENT PERSPECTIVE:
Why I Got Tested

By: Karen Spilks

I knew something was wrong. I'd had some odd symptoms for two years: my right foot would occasionally drag and shuffle and I had a strange sensation that my legs weren't holding me up as I walked up and down the long aisles in my workplace. And then there was my handwriting: suddenly it was so tiny and illegible that no one could read it.

I went to see a rheumatologist, who diagnosed me with fibromyalgia and put me on medicine for that diagnosis. But after nine months of continuing symptoms and no relief, I made an appointment with a neurologist. After ten minutes of watching me walk without swinging my left arm and noticing that I didn't blink very much, he blurted out, "You have Parkinson's." I felt a stab in my gut, as though he had given me a death sentence.

Now I no longer had an excuse; I had to face the clues. My mother's father and brother had PD and her sister is currently living with it. My mother, now 90, feels great guilt that it never hit her. Was it possible that I had inherited this disease?

I asked the doctor if he was sure. Although he was, he recommended getting a second opinion with a movement disorders specialist. I've been seeing Dr. Meredith Spindler at Penn for three years since to treat my PD.

I began reading, picking up anything and everything about the disease, considering carefully how it affected me.

The family prevalence of PD began to weigh heavily on me. I have a son and two grandsons. What had I passed on to them? When I learned that 23andme offered the testing to diagnosed PD patients and their families for free, I ordered the kit, spit in a tube, and waited. In six weeks, I got a call informing me that I did, in fact, carry the LRRK2 gene, very common in Ashkenazi Jews, my heritage on both sides of my family.

In my reading, I came across information on the Michael J. Fox Foundation for Parkinson's Research site about the Parkinson's Progression Markers Initiative (PPMI) study. The PPMI seemed like more than I wanted at the time (lumbar punctures! Did I have to?), so I put it on hold. Did I really want to remind myself every six months that I have a progressive illness?

I thought that way until my son, now 40, decided he should be tested, and we learned that he also has the LRRK2 mutation. I think this news hit me even harder than my own diagnosis. My child, my only one, was carrying this possibility within him. Because of me, because of his family, he would be from 35 to 50% more likely to develop PD than an average man his age.

Now I had to think differently about my own disease, because it wasn't just mine. And it wasn't like having heart disease or cancer, where sometimes if caught early, there is a better chance of survival. There is no cure for PD. Right now, all we have is research.

So, I joined the PPMI study, feeling that I had to make some lemonade out of these lemons. At this point, I have had three lumbar punctures (nowhere near as bad as I imagined!) as well as MRIs, blood draws, other scans, sleep inventories, depression/anxiety scales and cognitive testing. The PPMI staff and administration are incredible, offering meals and overnight stays in case I want to avoid traffic getting to my appointments. Whenever I go, I feel safe there. These very special people, doing this incredible work, treat me like I'm making a difference.

And I think I might be. We may not solve this problem in my lifetime, but we may be close to it for my son, and maybe my grandchildren will never have to deal with it. The Fox Foundation states, "Our single urgent goal is to cure Parkinson's disease—we can't do it without you."

I believe this with all my heart. I hope that others who have been diagnosed—especially those with a close relative or two who have PD—will jump into the research pool. It's warm and welcoming and getting clearer every year.

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The Parkinson's Progression Markers Initiative (PPMI) Update

By: Whitney Richardson

PPMI is a landmark, $80-million observational clinical study established and funded by the Michael J. Fox Foundation. The goal of PPMI is to identify and assess biomarkers, such as proteins in blood or spinal fluid, in people who may be at risk to develop PD. PPMI launched in 2010 to recruit newly diagnosed PD patients and healthy control volunteers.

In 2013, PPMI expanded to include two new groups of participants who may be at increased risk of developing PD. These individuals carry a genetic mutation in the LRRK2 and/or GBA genes, which have been associated with PD. These mutations most commonly occur among individuals of Ashkenazi Jewish descent. The PPMI is currently enrolling for this study.

Sign Up for PPMI

The Parkinson's Progression Markers Initiative (PPMI) at Penn is enrolling participants in the Genetic Cohort, and genetic testing is free to interested individuals through the Michael J. Fox Foundation. PPMI currently has 1,605 active participants at 33 sites around the globe. Penn's Parkinson's Disease and Movement Disorders Center is the second largest PPMI site in the United States with 79 active participants.

If you are interested in learning more about PPMI and free genetic testing, please contact the site coordinator, Whitney Richardson, at whitney.richardson@uphs.upenn.edu.

"TUNING INTO THERAPY": THE MUSIC AND MEMORY© PROGRAM

at the Dan Aaron Parkinson's Rehabilitation Center (DAPRC) at Penn Therapy & Fitness

By: Joellyn Fox, DPT, Lead Therapist

Have you ever had the experience of hearing a song in the car or grocery store that brings back a memory? Most everyone has. Favorite music associated with personal events can trigger specific memories, such as a teenage trip to the beach, or the song you danced to at your wedding. This personal association with music can be used as a therapeutic effect. That's what drove the therapy team at the DAPRC to become certified in Music and Memory©.

The benefits of personalized music are well documented by distinguished researchers, including Dr. Oliver Sacks, author of Musicophilia: Tales of Music and the Brain. MRIs taken while people listen to different songs indicate that their favorite music is most effective in stimulating areas of the visual, executive and cerebellar networks of the brain. This has the potential to create changes in mood and awareness in people with dementia.

The Music and Memory© Program

The role of exercise has been well documented to delay disease progression, however people with PD face many barriers. We started using the Music and Memory© program in the outpatient setting at the DAPRC to increase engagement and compliance with exercise. Dementia, attentional deficits and anxiety are other areas in which our therapists are using personalized playlists in order to make a positive impact.

If you are interested in this program, please speak with your doctor for a prescription. If you have any questions regarding our program, please email **Joellyn.fox@uphs.upenn.edu.**

MEET OUR *Research Coordinators*

This is our wonderful group of research coordinators. Whether working with the National Institutes of Health or pharmaceutical companies on new treatments for PD, or finding ways to slow the progression or halt the disease altogether, research is a crucial component of our mission at the PD&MDC. If you are interested in participating in research talk to your doctor or call 215.829.7273. You may find yourself spending some time with one of these bright and dedicated coordinators.

MEET OUR *New Staff Members*

Sarah Horn, MD, Fellow

A Texas native, I attended medical school at the University of Texas in Houston, then moved to Boston, Massachusetts, for a four-year neurology residency. I have been in Philadelphia for a movement disorders fellowship at the PD&MDC for the past year. I've really enjoyed learning the city, working with the wonderful neurology group at Penn, and meeting all of my new patients. I look forward to spending another great year here.

Caitlin Smith

Senior Practice Manager, Department of Neurology

I graduated from Kutztown University with a degree in communications and a minor in women & gender studies. After graduation, I worked for the National Multiple Sclerosis Society for five years. I just celebrated my one-year anniversary as the senior practice manager in Neurology at Penn Medicine. I am excited about the work we are doing here for patients living with movement disorders. My day-to-day responsibilities include clinical operations, overseeing secretarial and support staff, managing renovations, keeping the clinic operationally functional, and navigating patient concerns.

the parkinson council
your local partner in the movement against parkinson's disease

EVENTS

17th Annual "A Walk to Stamp Out Parkinson's

Saturday, October 13, 2018
7:45 am – 10 am
ALL-DAY access to The Philadelphia Zoo

A WALK TO STAMP OUT PARKINSON'S
THE PARKINSON COUNCIL

Join The Parkinson Council on Saturday, October 13, for the 17th annual "A Walk to Stamp Out Parkinson's" at The Philadelphia Zoo, America's first Zoo!

To get started, go to www.stampoutparkinsons.org or email walk@theparkinsoncouncil.org. As a fundraising event, Stamp Out Parkinson's has raised over $2.8 million over the last 16 years that are reinvested back into our region for research and quality of life programs and services to support thousands of families in our community.

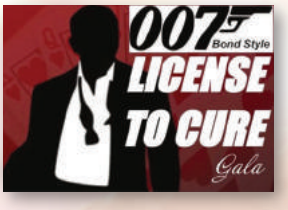
Managing Parkinson's Midlife: A Male Perspective

October 25, 2018 | 11 am – 4 pm
Morris Arboretum, 100 E. Northwestern Avenue
Philadelphia, PA

Please join us for a small group workshop for men ages 40-60 to learn how to manage the changes that PD produces. Call 215.829.2347 to reserve a spot. Space is limited to 60 participants. There is a $20 registration fee.

007 License to Cure Gala

November 17, 2018
5:30 pm – 11:30 pm
Chadds Ford, PA



For more information visit www.shakeitoff4PD.org.

Penn Medicine

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The Transmitter *is published biannually to inform the public about Parkinson's disease and other movement disorders. The information contained herein is solely to raise awareness of developments in the field of movement disorders. Nothing written in this newsletter should be used as a guideline for treatment.*

800.789.PENN
PennMedicine.org

OCTOBER 2018

Start time is 10:00 am - End time is around 1:30 pm (unless otherwise noted)

Sunday	Monday	Tuesday	Wednesday	Thursday	Friday	Saturday
	1	2	3	4	5	6
7	8	9	10	11	12	13
14	15	16	17 Friends & Family Tour 6-7 pm	18 Patterned Play 10:00-1:30	19	20
21	22	23	24 Patterned Play 5:30-8:30	25 Patterned Play 10:00-1:30	26	27
28	29	30 Patterned Play 10:00-1:30	31			

Museum Programs for individuals with Parkinson's Disease are generously supported by The Parkinson Council and the Susan O. Jaffe Quality of Life Initiative

For information and registration
Call: (215) 684-7602
Email: accessprog@philamuseum.org

Image Used Front:

Woman's Evening Ensemble: Dress, Belt, and Capelet, 1983
Designed by Pierre Cardin, French (born Italy), born 1922
Made in France, Europe
Printed silk organza over nylon horsehair, synthetic faille

The Parkinson's Disease and Movement Disorders Center is a Parkinson Foundation Center of Excellence.