

Survey of PSG Clinicians on Genetics and Genetic Testing in Parkinson's disease

Section A. Demographic information. You are:

Sex Male Female

Age <40 41-50 51-60 >60

Location of your Parkinson's clinic. For Canada, please select "Canada (all time zones)". For US, please select time zone.

Eastern

Central

Mountain

Pacific (including Alaska and Hawaii)

Canada (all time zones)

Section B. LRRK2, GBA, and other Parkinson's risk genes in your clinic population

How many patients with PD did you personally provide care for over the last 12 months? _____

How many PD patients for whom you have provided care in the last 12 months have had the following kinds of genetic tests?

a) Clinical genetic testing ordered by a health care provider: _____

b) Genetic testing performed by a direct-to-consumer (DTC) company (such as 23andMe): _____

c) Genetic test results provided to the patient as part of participation in a research study (such as Parkinson's Progression Marker Initiative [PPMI]): _____

d) Genetic testing as part of a research study where NO results were provided to the patient but results were returned to you or a collaborating investigator: _____

e) DNA banking as part of a research study where NO results were provided to the patient: _____

f) Refused genetic testing: _____

How many patients for whom you have provided care in the last 12 months have a genetically-proven

A. LRRK2 variant associated with PD? _____

B. GBA variant associated with PD? _____

- C. Parkin variant associated with PD? _____
- D. Alpha-synuclein variant associated with PD? _____
- E. DJ-1 variant associated with PD? _____
- F. PINK1 variant associated with PD? _____
- G. VPS35 variant associated with PD? _____
- H. Other (specify)_____

Section C. Genetics services and utilization

Do your PD patients have access to the services of a genetic counselor through your institution?

Yes

No

(this question branches off of the previous question, and will only be shown if participants select “yes”) Please select the ways in which your patients can access the services of a genetic counselor through your institution. You are able to check multiple boxes.

In the Neurology/Parkinson’s Clinic

Within my institution but not in the Neurology/Parkinson’s Clinic

via telephone/telemedicine while in my clinic

Patients are referred outside my institution to receive genetic counseling

Other

(this question branches off of the previous question, and will only be shown if a participant selects “other”) If you selected “other” to the previous question please provide the locations here:

If, next week, you ordered a genetic test for LRRK2 for a patient already diagnosed with PD would you: (SELECT ONE)

_____ I will provide the genetic counseling and would not involve a genetic counselor

_____ definitely involve a genetic counselor before the blood is drawn AND at the time that results are given to the patient

_____ definitely involve a genetic counselor at the time that results are given to the patient, but not before the blood draw

_____ definitely involve a genetic counselor before the blood draw, but not at the time that results are given to the patient

_____ suggest to the patient or family, before the blood draw, the option of a referral to a genetic counselor

_____ suggest to the patient or family, after results are given, the option of a referral to a genetic counselor

_____ refer the patient or family to a genetic counselor if they request it

_____ not involve a genetic counselor

_____ I would not order the test as part of clinical care

10. To what extent do any of the following keep you from ordering genetic tests for patients with PD in your clinic?

Potential barrier	No barrier..... Constant barrier 0---10---20---30---40---50---60---70---80---90---100
Insurance doesn't pay/patients can't afford	0---10---20---30---40---50---60---70---80---90---100
Patients don't want a genetic test	0---10---20---30---40---50---60---70---80---90---100
I don't know what test to order/ lab to use	0---10---20---30---40---50---60---70---80---90---100
I don't feel comfortable explaining the meaning of test results to patients	0---10---20---30---40---50---60---70---80---90---100
Genetic test results would not change my approach to therapy	0---10---20---30---40---50---60---70---80---90---100
Genetic test results would not change the patient's prognosis	0---10---20---30---40---50---60---70---80---90---100
I am concerned about the impact of a genetic test on the patient's employability/ insurability	0---10---20---30---40---50---60---70---80---90---100
I am worried about the implications of the patient's genetic test results for his/her children and other family members	0---10---20---30---40---50---60---70---80---90---100

The Parkinson's Foundation plans, through its Genetics Research Initiative, to facilitate clinical genetic testing for *GBA* and *LRRK2* in patients with PD. This initiative was established in part to familiarize the

medical and patient community with the growing importance of genetics in the diagnosis, prognosis, and management of PD, and also to facilitate the entry of appropriate patients into genotype-specific clinical trials. Genetic counseling will either be performed by a centralized genetic counseling team or in your PD clinic with compensation for phlebotomy and counseling. Knowing that information, how do you feel about the following?

YES ____ NO ____ NOT SURE ____ I think that this initiative will greatly improve my patients' access to genetic tests

YES ____ NO ____ NOT SURE ____ I would be likely to order tests on the majority of my patients

YES ____ NO ____ NOT SURE ____ I feel that I am equipped to counsel patients adequately about the clinical relevance of both normal and abnormal genetic test results

YES ____ NO ____ NOT SURE ____ I would like a genetic counselor to be involved with counseling and testing of patients at my site

YES ____ NO ____ NOT SURE ____ I have ethical concerns about such initiative

If you have concerns about this initiative, please describe:

Section D. Knowledge/attitudes about genetics and genetic testing. Please note that your responses will remain anonymous to study investigators.

Your 65-year-old male PD patient of non-Ashkenazi heritage came to your clinic today to discuss the results of a genetic test that looked for the 5 most common Gaucher disease-associated *GBA* variants. The test showed that he has one gene with a *GBA* L444P variant and one that is normal for the 5 variants assayed.

Please rely on your current knowledge and opinions in selecting your candid response to the following statements about this case:

Agree	Disagree	Uncertain	
			This patient must have some Ashkenazi background but just doesn't know it
			This patient has <i>GBA</i> -associated PD
			This patient does not have <i>GBA</i> -associated PD, because one copy of the gene is normal
			This patient is more likely to have cognitive impairment sooner than a patient with idiopathic PD
			Deep Brain Stimulation would NOT be expected to help this patient's motor symptoms
			Each of this patient's biological children has a 50% chance of developing Parkinson's disease
			Each of this patient's biological children has a 50% chance of developing Gaucher disease

A 60-year-old male PD patient of Ashkenazi background with a positive family history of Parkinson’s comes to your clinic to discuss the results of their LRRK2 genetic test, which shows a G2019S variant on one copy of the gene, with the other copy showing no variants.

Please agree or disagree with the following statements about this case:

Agree	Disagree	Uncertain	
			This patient has LRRK2-associated PD
			This patient does not have LRRK2-associated PD, because one copy of the gene is normal
			This patient is more likely to have cognitive impairment sooner than a patient with idiopathic PD
			Deep Brain Stimulation would NOT be expected to help this patient’s motor symptoms
			Each of this patient’s biological children has a 50% chance of developing Parkinson’s disease
			Each of this patient’s biological children has a 50% chance of developing Gaucher disease
			Testing for the G2019S variant is available through Direct-to-Consumer testing

Finally, we recognize that PD genetics is a new and rapidly evolving field. We want to gauge your comfort level with genetic counseling and testing. As such please, rate on the scale from 0 (not at all) to 10 (proficient)

0----10----20----30----40----50----60----70----80----90----100

Not Confident Very Confident

I am confident in my knowledge of the impact of various genetic mutations on PD phenotypic manifestations

0----10----20----30----40----50----60----70----80----90----100

Not Confident Very Confident

I am confident that I am up to date on genetically targeted experimental therapeutics on GBA and LRRK2-related trial

0----10----20----30----40----50----60----70----80----90----100

Not Comfortable

Very Comfortable

I am comfortable counseling my PD patients on GBA and LRRK2 testing

For the remaining questions, we will be asking about your knowledge and understanding of genetic factors related to Parkinson's disease.

A genetic test for Parkinson's disease can detect all genetic mutations related to Parkinson's disease. (True/False)

Environmental factors can increase the risk of Parkinson's disease. (True/False)

A negative genetic test for Parkinson's disease means a person does not have Parkinson's disease. (True/False)

Everyone with a LRRK2 or GBA gene mutation will develop Parkinson's disease. (True/False)

A genetic mutation is simply a change in a gene. (True/False)

Genetic test results can be used to determine eligibility for clinical trials. (True/False)

A person with no family history of Parkinson's disease can have an inherited Parkinson's gene mutation. (True/False)

E. Comments

Thank you for your participation in this survey. We appreciate your honesty and responsiveness to our questionnaire.

Please contact Roseanna Battista at Roseanna.Battista@chet.rochester.edu indicating that you have completed the survey to obtain your \$30 Amazon gift card.



Columbia University IRB

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