The Genetic Testing Experience of Individuals with Parkinson Disease

UNIVERSITY of MARYLAND SCHOOL OF MEDICINE

Sydney Richards[1], Weiyi Mu[2], Rachel Nusbaum[3], Katherine Lincoln[4], Julie Solimine[5]

[1]Master's in Genetic Counseling Training Program, University of Maryland School of Medicine; [2]Department of Genetic Medicine, Johns Hopkins University School of Medicine;
[3]Department of Pediatrics, University of Maryland School of Medicine; [4]Center for Human Genetics, University Hospitals Cleveland Medical Center;
[5]Department of Obstetrics, Gynecology, and Reproductive Sciences, University of Maryland School of Medicine

INTRODUCTION:

The patient experience with genetic testing is essential to understand, yet the perspective of patients with Parkinson Disease (PD) following genetic testing and results disclosure is lacking.

In the literature, previous studies have explored the attitudes, interest, and knowledge of patients with PD about genetic testing for their diagnosis prior to pursuing genetic testing,[1-4] as well as attitudes toward genetic testing for PD among clinicians.[5] Two studies have found the majority of participants with PD to be interested in genetic testing.[1,2] Research exploring patients' attitudes toward testing has also identified anticipated positive and negative consequences that participants believe might occur after genetic testing.[1-3]

A recent study found individuals to have increased adverse psychological reactions of distress and uncertainty among those who carry a pathogenic variant for PD.[6] Further research is needed to determine if these findings can be reproduced among a more diverse population of individuals who underwent testing.

This current study addresses this gap in knowledge in an effort to better understand the experience of genetic testing within this patient population.

AIMS:

- **Aim 1:** Determine if previously anticipated positive and negative consequences of genetic testing for PD have occurred for individuals with PD who have had genetic testing.
- **Aim 2:** Identify reasons why individuals with PD have not had genetic testing for PD.

METHODS:

- Participants were recruited through 15 patient/disease support and advocacy organizations and seven private PD Facebook groups nationwide.
- Eligible participants had a diagnosis of PD, were at least 18 years or older, and currently live in the US.
- Participants were excluded if they were non-English speaking or if they currently needed another individual to act as a proxy for medical care.
- Demographic and genetic testing information, as well as positive and negative consequences experienced or anticipated by the genetic testing and non-genetic testing groups, respectively, were collected via an online-based survey.
- Quantitative data analysis was performed using IBM SPSS statistical software.
- Descriptive statistics, Pearson correlation coefficient, ANOVA, and independent sample t-test were utilized as appropriate. Tukey's HSD test for post-hoc analysis of statistically significant ANOVA findings was also used.

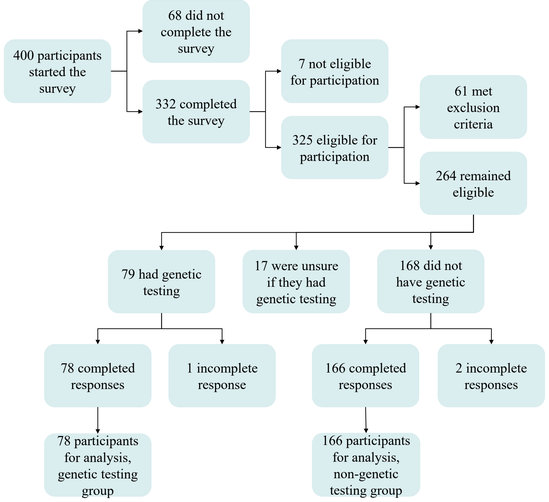

Figure 1. Participant selection for analysis

RESULTS:

	All Participants	Participants with genetic testing	Participants without genetic testing
	n = 244	n = 78	n = 166
Average age*, mean (range)	65.6 years (23-85)	62.4 years (23-80)	67.2 years (39-85)
Average age range at diagnosis, mean	55-65 years	50-59 years	60-69 years
Gender			
Male	116 (47.5%)	35 (44.9%)	81 (48.8%)
Female	127 (52.0%)	43 (55.1%)	84 (50.6)
Self-Describe**	1 (0.4%)		1 (0.6%)
Ethnicity			
Hispanic or Latino	7 (2.0%)	3 (3.8%)	4 (2.4%)
Non-Hispanic or Latino	228 (93.4%)	74 (94.9%)	154 (92.8%)
Prefer not to answer	9 (3.7%)	1 (1.3%)	8 (4.8%)
Race			
Asian	5 (2.0%)	1 (1.3%)	4 (2.4%)
Black or African American	2 (0.8%)	1 (1.3%)	1 (0.6%)
White	233 (95.5%)	74 (94.9%)	159 (95.8%)
Prefer not to answer	4 (1.6%)	2 (2.6%)	2 (1.2%)

*Two participants did not complete this question, from those without genetic testing, and therefore, the "all participants" category; **The one participant who selected this response did not submit a written response for this question.

Table 1. Demographic and participant characteristics

Figure 2. Method of how genetic test was received among participants who underwent genetic testing for PD

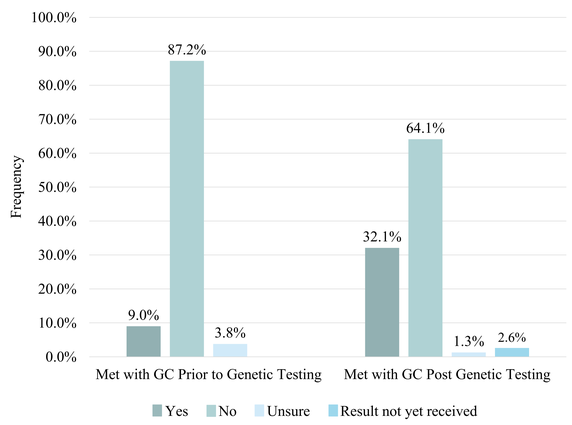

Figure 3. Participants' response to meeting with a genetic counselor (GC) prior to or post genetic testing

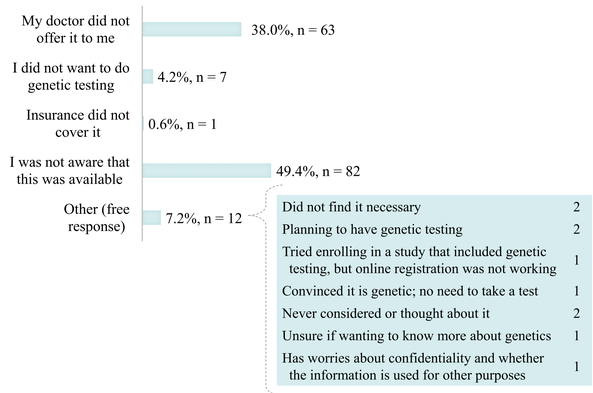

Figure 4. Selected and/or annotated reasons why participants have not had genetic testing

Of the 166 participants who did not have genetic testing, one participant did not select a reason why they have not had genetic testing. Two participants who selected 'other' did not enter their free response.

	Positive Consequences of Genetic Testing Statements	Negative Consequences of Genetic Testing Statements
Personal consequences	• Allowing patients to learn more about themselves and their condition • Helping patients adapt to their diagnosis of PD • Changing patients' futures	• Affecting the patient and/or family in getting disability and life insurance • Preventing patients from continuing or finding a job • Providing no personal benefit
Consequences related to diagnosis of PD	• Helping patients' providers better manage their condition • Allowing patients to identify the cause of their PD • Helping patients discuss resources and research for PD	• Affecting financial planning • Not changing clinical management • Causing patients to face emotional and/or psychological distress
Family consequences	• Providing information for patients' families • Helping patients discuss the impact of genetic testing on the family • Benefiting patients' families	• Causing patients' loved ones to face discrimination and confidentiality issues • Giving patients' loved ones unwanted information • Inducing anxiety among family members

Table 2. Subgroups of positive and negative consequences and respective statements utilized in study

	Participants who have had genetic testing n = 78		Participants who have not had genetic testing n = 166		*p*
	M	*SD*	*M*	*SD*	
Positive personal consequences	16.2	3.6	12.9	3.2	<.001*
Negative personal consequences	20.6	3.3	18.0	3.8	<.001*
Positive consequences related to diagnosis of PD	21.0	5.2	14.3	3.7	<.001*
Negative consequences related to diagnosis of PD	18.5	3.0	16.7	3.0	<.001*
Positive family consequences	10.6	3.4	7.4	2.7	<.001*
Negative family consequences	16.5	3.1	12.7	3.4	<.001*

*indicates statistical significance; *M* = mean, *SD* = standard deviation

Table 3. Positive and Negative Consequences Experienced versus Anticipated

This table describes the comparisons between participants who have and have not had genetic testing for each positive and negative subgroup of consequences. Positive and negative personal consequences have a total possible score of 25. Positive consequences related to diagnosis of PD has a total possible score of 35. Negative consequences related to diagnosis of PD has a total possible score of 25. Positive and negative family consequences have a total possible score of 20. Higher scores suggest an increased selection of 'disagree' or 'strongly disagree' among participants' responses on the Likert-scale questions.

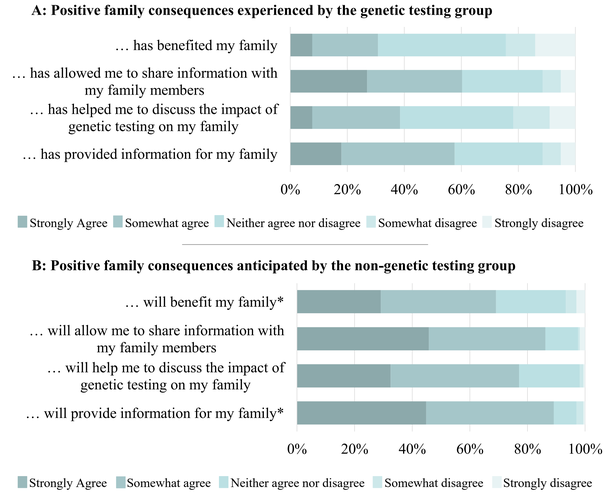

Figure 5. Experienced and Anticipated Positive Family Consequences

*One participant did not complete the respective question.

This figure details the participants' responses to Likert-scale statements regarding positive family consequences of genetic testing. Possible response options ranged from strongly agree to strongly disagree. The specific statements about genetic testing answered by the participants are annotated to the left of each figure. Of note, similar visual trends between both groups were observed among negative family consequences, positive and negative personal consequences, and positive and negative consequences related to diagnosis of PD.

DISCUSSION:

Conclusions

- Few participants who underwent genetic testing for PD obtained genetic testing through their healthcare provider, and most individuals did not meet with a genetic counselor prior to or following genetic testing.
- The most common barriers that led participants to not having had genetic testing included not being aware of genetic testing for PD and not being offered genetic testing options by their healthcare providers.
- The genetic testing group experienced fewer positive consequences and fewer negative consequences than anticipated by the non-genetic testing group. This suggests that these consequences are not occurring to the extent of what is anticipated by those who have not had genetic testing.
- The study's findings emphasize the need for neurologists to have genetic testing discussions and to collaborate with genetic counselors, for genetic counselors to work in practice with the PD population, and for anticipatory guidance to be provided in the genetic testing discussion to better set expectations.

Limitations

- Study population is lacking in diversity and representation
- Sample size was relatively small
- Only individuals with internet access were able to participate
- Possible ascertainment bias in individuals who participate in disease advocacy groups

Acknowledgements

We thank all participants for their time and effort, as well as the patient/disease support and advocacy organizations and private Facebook groups for their support and assistance with study recruitment. We also thank Rebecca Thompson, PhD for guidance in conducting statistical analyses.

References

1. Falcone DC, et al. Genetic testing and Parkinson disease: Assessment of patient knowledge, attitudes, and interest. *J Genet Couns*. 2011;20(4):384-395.
2. Maloney KA, et al. Parkinson's Disease: Patients' Knowledge, Attitudes, and Interest in Genetic Counseling. *J Genet Couns*. 2018;27(5):1200-1209.
3. Tan EK, et al. Comparing knowledge and attitudes towards genetic testing in Parkinson's disease in an American and Asian population. *J Neurol Sci*. 2007;252(2):113-120.
4. Sakanaka K, et al. Knowledge of and interest in genetic results among Parkinson disease patients and caregivers. *J Genet Couns*. 2014;23(1):114-120.
5. Alcalay RN, et al. Genetic testing for Parkinson disease: current practice, knowledge, and attitudes among US and Canadian movement disorders specialists. *Genet Med*. 2020;22(3):574-580.
6. Verbrugge J, et al. Outcomes of genetic test disclosure and genetic counseling in a large Parkinson's disease research study. *J Genet Couns*. 2021;30(3):755-765.