Providing Genetic Testing and Genetic Counseling to the Parkinson's Disease Community: The PD GENEration Pilot Study Experience

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Objective

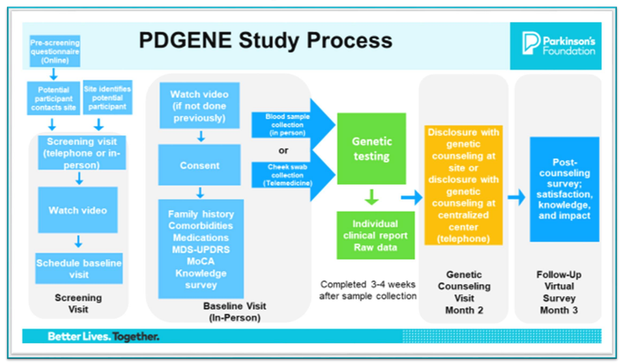
A pilot study was conducted to assess the feasibility of offering widespread genetic testing and counseling to patients with Parkinson's disease (PD) in the United States, using satisfaction and impact outcome measures to evaluate differences between centralized and study site disclosures.

Background

Genetic testing for PD is not routinely offered in neurology practice. However, knowledge of one's genetic status may have personal and clinical utilities, including the potential to enroll in gene-targeted clinical trials.

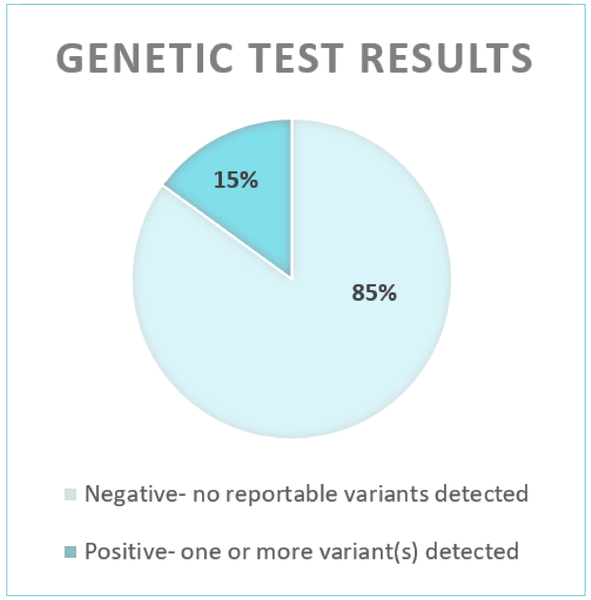
Methods

Participants were recruited at 6 study sites and underwent assessments including medical and family histories, MDS-UPDRS, MoCA, and baseline knowledge of PD genetics. Following consent, samples were obtained for sequencing of 7 PD-related genes (*SNCA, PRKN, PINK1, PARK7, LRRK2, GBA* and *VPS35*) by a CLIA-accredited laboratory. Participants were randomized to receive test results during genetic counseling conducted either at their local site (clinician or genetic counselor) or remotely by genetic counselors at Indiana University. Three-month follow-up surveys post session were sent, measuring satisfaction, knowledge, and psychological impact. Providers were also surveyed.



Results

As of 2/25/21, 650 participants were enrolled as part of the pilot study and 295 had responded to surveys. All 13 providers in the study who conducted sessions completed surveys. The majority of participants were of low-risk ancestry (not Ashkenazi Jewish, Basque, or African Berber), and the mean age of onset of PD was 58.7 years. Pathogenic genetic variants were found in 15.4% of tested participants – mostly in the genes *GBA (8%), LRRK2 (2%),* and *PRKN (2%)*. Participants were highly satisfied with the receipt of genetic test results and counseling. There were no significant differences in knowledge or satisfaction between centralized and site genetic counseling, but participants receiving results from their site reported feeling more like a "partner in care". Receiving an abnormal test result did not affect satisfaction but was associated with a longer counseling session.



Conclusion

- **Widespread genetic testing for PD paired with counseling is feasible, with high levels of satisfaction using centralized or on-site counseling.**
- **Based on these encouraging results and keen interest within the PD community, enrollment is expanding.**