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Genetic Testing and Parkinson Disease: Assessment of Patient Knowledge, Attitudes, and Interest

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Abstract

The most common genetic contributor to late-onset Parkinson disease (PD) is the *LRRK2* gene. In order to effectively integrate *LRRK2* genetic testing into clinical practice, a strategy tailored to the PD population must be developed. We assessed 168 individuals with PD for baseline knowledge of genetics, perceived risk, and interest and opinions regarding genetic counseling and testing. Most participants felt that they were familiar with general genetics terms but overall knowledge levels were low, with an average score of 55%. The majority of participants thought it was likely they inherited a PD gene (72%), believed genetic testing for PD would be useful (86%), and were interested in genetic testing (59%) and genetic counseling (56%). However, only a few participants had heard of any genetic tests for PD (29%) or *LRRK2* (10%). There appears to be a significant level of interest in genetics and genetic testing within the PD population, but a considerable deficit in genetics knowledge and an over-estimation of risk. Genetic education and counseling tools to address these needs were developed to provide patients with the ability to make informed and knowledgeable genetic testing decisions.

Keywords

Parkinson disease; Genetic testing; Genetic knowledge; Attitudes; Perceived risk

Introduction

Parkinson disease (PD) is the second most common neurodegenerative disease and is characterized by resting tremor, rigidity, bradykinesia, and postural instability (see Poewe and Mahlknecht 2009 for a review of PD). In addition, 18–41% of patients with PD develop dementia over the course of the disease (Biggins et al. 1992; de Lau et al. 2005; Mayeux et al. 1992; Tison et al. 1995). Approximately 10–16% of individuals with PD report a family history of PD in at least one first-degree relative (Elbaz et al. 1999; Marder et al. 2003; Taylor et al. 1999; Uitti et al. 2005) and research in the past decade has identified several genes and genetic loci that contribute to the development of PD. Parkinson Disease is directly caused by gene mutations in less than 10% of all cases (Tan and Jankovic 2006). Many of these genetic causes are rare and have been found only in a few families worldwide. Although variably successful symptomatic treatments for PD are available, there is no cure and no preventative measures for those individuals considered to be at an increased risk.

The most common genetic contributor to late-onset PD, both familial and sporadic, is the *LRRK2* gene encoding leucine-rich repeat kinase 2. First described in 2004, mutations in *LRRK2* were found to be associated with an autosomal dominant form of hereditary PD (Paisan-Ruiz et al. 2004; Zimprich et al. 2004), accounting for 1% of sporadic PD and 5%–6% of familial PD in the U.S (Farrer et al. 2005). The most common mutation in *LRRK2*, Gly2019Ser, exhibits reduced penetrance, with the lifetime risk of PD reported to be ~35–74% by age 80 (Goldwurm et al. 2007; Healy et al. 2008; Ozelius et al. 2006). Additional genetic and/or environmental factors that may influence penetrance have not yet been identified.

Clinical genetic testing for *LRRK2* mutations is now available for patients and at-risk family members. Several recent reports have discussed the availability of *LRRK2* genetic testing in PD; however, no publications have extensively addressed the best clinical approach and implementation of this aspect of PD clinical care (Kay et al. 2006; Klein 2006; Klein and Schlossmacher 2006; McInerney-Leo 2005; McInerney-Leo et al. 2005; Tan and Jankovic 2006). There is a general consensus that clinical *LRRK2* genetic testing for symptomatic individuals may provide a benefit by confirming the clinical diagnosis of PD, as well as providing an explanation for a significant family history of PD. However, the effects of genetic testing for PD are not well understood. Therefore, research focusing on the patient's perspective about and understanding of *LRRK2* genetic testing is essential if clinicians and genetic counselors are to provide the best level of care and support. Such research will have ongoing implications for the development of clinical genetic testing protocols for PD, including responsible approaches for predictive *LRRK2* genetic testing.

Provision of information about personal risk and education regarding genetic testing are essential, as they can have a significant impact on patients' and family members' testing decisions. To our knowledge, only one study has examined the level of baseline genetics knowledge in a PD cohort. Tan et al. (2007) interviewed a sample of 515 individuals comprised of patients with PD and their caregivers in two centers in America and Asia using a standardized questionnaire. Overall, both groups had relatively low knowledge of PD genetics. However, American patients with PD had a significantly higher level of knowledge than Asian patients with PD, with average scores of 31% and 12%, respectively. The researchers concluded that providing individuals interested in genetic testing for PD with personal risk information is a challenge as present knowledge about the pathogenicity and penetrance of some mutations is still limited (Tan and Jankovic 2006).

Understanding the motivations leading individuals to pursue or not pursue genetic testing can further help genetic counselors and physicians assist patients in weighing the benefits and limitations of testing. Studies have shown that breast cancer patients pursue genetic testing for reasons that include: wanting cancer risk information for relatives, seeking information about their own risk of recurrence, and assisting in future health care decisions (Armstrong et al. 2000; Cappelli et al. 1999). Although similar studies have yet to be conducted for patients with PD, one author predicts individuals with PD most likely would seek genetic testing not for their own benefit, but for determination of the disease risk for their family members (McInerney-Leo 2005).

The discovery of *LRRK2* as a PD disease gene has significantly changed the landscape of PD genetics—a disease that was once considered completely non-genetic in etiology is now thought to have a fairly significant genetic contribution for both familial and sporadic forms. However, the utility of *LRRK2* genetic testing in the clinical setting has not been thoroughly evaluated. In order to effectively integrate *LRRK2* genetic testing into clinical practice, an effective strategy tailored to the PD population must be developed. In this study we assessed baseline knowledge of basic genetics principles and PD genetics, interest in and opinions regarding genetic counseling and genetic testing, as well as levels of concern regarding a possible familial risk among patients with PD in order to guide the development of educational materials specific to the PD population.

Methods

Participants

Eligible participants were adult patients with a clinical diagnosis of PD followed by a University of Pennsylvania Health System neurologist. All study activities were approved by the University of Pennsylvania institutional review board. Individuals who previously participated in research studies and indicated a willingness to be recontacted for future research participation were invited by mail to participate in the study. The mailing included a letter explaining the purpose of the study, an informed consent form, and a copy of the survey. An option to complete the survey online via SurveyMonkey (www.surveymonkey.com) was also described in the letter. Participants were encouraged to contact the study coordinators if they had any questions or concerns about the study. Eligible participants were also provided with the opportunity, either by their neurologist or a member of the study team, to participate in the research study during their appointment with their neurologist.

A total of 381 individuals were contacted either by mail ($n=331$) or in person ($n=50$) regarding the study and 168 (44%) individuals enrolled. The response rate was 38% (127/331) for the mailed surveys and 82% (41/50) for the participants contacted “in person” at their neurology appointment. There were no significant differences in terms of age, $t(379)=-.02$, $p=0.98$, disease duration, $t(378)=1.13$, $p=.26$, or age of onset, $t(378)=0.52$, $p=.61$ between individuals who participated in the study and those that did not. However, a chi-square test of independence indicated that there was a significant relationship between gender and participation in the study, with women more likely to participate than men, $\chi^2(1, N=381)=8.18$, $p=.004$ (54% versus 39%, respectively).

Structured Questionnaire

A structured patient questionnaire was designed based on a literature review of common misconceptions about genetics and previously published tools used in the field of general population genetic knowledge and adult-onset genetic conditions (Bowling et al. 2008; Houfek et al. 2008; Ishiyama et al. 2008; Kelly et al. 2004; Moscarillo et al. 2007; Prakash

et al. 2006; Tan et al. 2007; Watner et al. 2002). All knowledge questions and data were collated and analyzed for the ability to assess the goals of the study. Questions were reviewed by a group of neurodegenerative disease researchers, Parkinson disease physicians, genetic counselors, and geneticists for clarity and readability, and minor modifications were made to some items. The overall readability of the questionnaire, assessed with the Flesch-Kincaid scale, was grade level 8.5, and the readability of the knowledge questions alone was grade level 11.2 (Flesch 1948; Kincaid et al. 1975).

The questionnaire included questions about demographics, general genetics knowledge, PD knowledge, and genetic testing beliefs and attitudes. Age of onset and disease duration information were collected from the patient's chart; all other demographic information was collected by self report (Table 1). Seventeen general genetics and twenty-one PD knowledge items were included. Knowledge questions were addressed using either a multiple choice or agree/disagree answer format. Knowledge questions that were presented in an agree/disagree answer format asked participants to rate a series of statements using a 5-item response scale (strongly agree, agree, neither agree nor disagree, disagree, strongly disagree). Strongly agree and agree responses were grouped and treated as answering the questions as "true." Strongly disagree and disagree responses were treated as answering the questions as "false." Participants were instructed to choose the response "neither agree or disagree" if their answer was "don't know" therefore "neither agree or disagree" responses were treated as answering the question as "don't know." Open-ended responses and yes/no/don't know formats were used to assess patients' beliefs and attitudes about genetic testing.

The questionnaire was piloted on 20 participants who completed the survey in person. The content was then reviewed for clarity and minor adjustments were made. The final version of the questionnaire was completed either by mail, online, or in person. Participants were asked not to use outside help in answering the knowledge questions. A Montreal Cognitive Assessment (MoCA) (Nasreddine et al. 2005) was obtained in person for the majority of participants in order to assess their cognitive status. The total possible score for the MoCA is 30 points; a score of 26 or above is considered normal. The MoCA is thought to be an adequate screening tool for the detection of mild cognitive impairments and dementia in patients with PD; however, further assessment is recommended for individuals who score below the normal range (Hoops et al. 2009).

Data Analysis

All data were coded and analyzed using SPSS for Windows 17.0 (SPSS, Inc. Chicago, IL). Responses for the knowledge questions were categorized as correct or incorrect, and a total score was calculated by summing up the number of correct answers. Descriptive statistics were calculated to characterize the sample in terms of demographic characteristics, PD and general genetics knowledge, and genetic testing beliefs and attitudes. The generalized estimating equation approach (Zeger et al. 1988) was used to examine the likelihood of choosing being familiar with a certain genetic term. A paired *t*-test was used to examine the difference between performance on general genetics questions vs. PD-specific genetics questions. Two-sample *t*-tests or analyses of variance (ANOVA) were used to examine associations between knowledge, demographics, and genetic testing beliefs and attitudes. Pairwise comparisons were made with Bonferroni corrections for post-hoc analysis of ANOVA results. A Pearson correlation coefficient was computed to examine the relationship between knowledge and age. Normality assumptions were checked for all continuous variables. An alpha level of .01 rather than the traditional level of .05 was used for all statistical tests to control for family-wise error rate due to the large number of univariate statistical tests performed. All statistical tests were two-sided. Open-ended questions were independently coded by the first and second authors. Each author reviewed the responses to extract major themes; the final theme categories were then jointly selected

by the authors. Each author individually coded all responses using these themes and a final consensus discussion was held to code each response. Individual responses for open-ended questions could be coded with more than one theme.

Results

Sociodemographic and Clinical Characteristics

The sociodemographic and clinical characteristics of the 168 participants are shown in Table 1. The majority of participants were Caucasian (96%), married (79%), and had a 4-year college degree or higher (67%). Thirty-two percent ($n=54$) of participants reported that they had a family history of PD, and 29% of participants reported having a family history of other neurodegenerative diseases such as amyotrophic lateral sclerosis (ALS), Alzheimer disease (AD), or other dementia. Twenty-five (15%) of the participants that reported a family history of PD reported at least one affected first degree relative, 22 (13%) reported at least one affected second-degree relative, and 7 (4%) reported at least one third-degree or more distantly related affected relative. A MoCA was obtained on 155 (92%) participants and 64 (41%) scored below the normal range.

Knowledge

Participants were asked to rate their familiarity with each of four general genetic terms (Fig. 1). The majority of participants rated all four terms as “familiar with term and know the meaning.” However, when analyzing the data using the generalized estimating equation approach, participants were significantly more likely to choose “familiar with the term and know the meaning” for the term “gene” compared to the terms “chromosome” (odds ratio (OR)=1.44, 95% confidence interval (CI)=1.18–1.76, $p=.0004$) or “genetic testing” (OR=1.33, 95% CI= 1.05–1.69, $p=.017$). Participants were also more likely to choose “familiar with the term and know the meaning” for the term “DNA” compared to the terms “chromosome” (OR=1.55, 95% CI=1.21–1.99, $p=.0005$) or “genetic testing” (OR=1.45, 95% CI=1.16–1.79, $p=.0008$). The generalized estimating equation approach is used to analyze correlated data and it takes into account correlations among repeated measurements for a participant.

On average, participants gave correct responses to 55% (range 16–95%, $SD=17\%$) of the 38 knowledge questions. While the overall difference in scores is small, participants did better on the general genetics questions (mean=59%; $SD=18\%$) compared to the PD symptoms and genetics questions (mean=54%; $SD=20\%$), $t(167)=2.82$, $p=.005$. Knowledge questions were grouped into basic concepts and participant scores for each concept were analyzed (Fig. 2). When looking at the average number of correct responses for each concept, the majority of participants demonstrated an understanding of PD symptoms and risk factor genes but did not demonstrate an understanding of the *LRRK2* gene and genetic testing. An example of the types of questions included in the knowledge section of the questionnaire is provided in Table 2.

The item most often answered correctly was “Drugs are available to treat the symptoms of Parkinson disease” (answer=agree; $n=166$, 99%). The item most often answered incorrectly was “*LRRK2* is a common genetic contributor to both familial and sporadic late-onset Parkinson disease” (answer=agree; $n=10$, 6%). The majority of respondents ($n=152$, 91%) rated the above *LRRK2* statement as “neither agree or disagree” and their responses were coded as “don’t know.” Only 16 participants (10%) reported that they had previously heard of the *LRRK2* gene. Just under half of the participants (7/16) who reported that they had heard of *LRRK2* “agreed” with the above item, accounting for the majority (7/10) of the participants who answered this item correctly. Two of the 16 participants who reported

having heard of *LRRK2* answered “disagree” to the *LRRK2* item; these two participants may have disagreed with this item because of a subjective interpretation of the word “common” or they may not have known that *LRRK2* is a significant genetic cause of PD.

Associations between participants’ total knowledge scores and their demographic characteristics were examined. All knowledge scores are presented as a percent of correct responses. While overall knowledge scores were low, higher knowledge scores were significantly associated with a number of demographic factors. An analysis of variance showed that the effect of education level on total knowledge was significant, $F(3,164)=4.76$, $p=0.003$. Pairwise comparisons were made with Bonferroni tests, holding Type I error rate at a maximum of 0.01. The average total knowledge score was significantly higher for participants who completed a postgraduate degree (mean=59%, $SD=17\%$) compared to participants who had a high school education or less (mean=45%; $SD=15\%$). There was a significant negative correlation between age and total knowledge score (Pearson correlation coefficient $r(166)=-.28$, $p<.01$), with higher knowledge scores associated with a younger age. Participants who had MoCA scores in the cognitively normal range had higher knowledge scores (mean=57%; $SD=16\%$) than participants with MoCA scores below the normal range (mean=49%; $SD=16\%$), $t(148)=2.99$, $p=.002$.

Total knowledge scores and participants’ experience with genetics as well as their awareness of PD genetics and genetic testing were examined. Average knowledge scores were significantly higher for those individuals who indicated they had studied genetics in school (mean=58%; $SD=18\%$) compared to those who had not or were unsure (mean=51%; $SD=15\%$), $t(166)=-2.74$, $p=.007$. Participants who had heard of the *LRRK2* gene (mean=68%; $SD=18\%$) had higher average knowledge scores compared to those who had not or were unsure (mean=53%; $SD=16\%$), $t(166)=-3.55$, $p<.001$. In addition, participants who had heard of a genetic test for PD (mean=61%; $SD=17\%$) had higher average knowledge scores than participants who had not heard of any genetic tests for PD or were unsure (Mean= 52%; $SD=16\%$), $t(166)=-3.58$, $p<.001$.

The association between total knowledge scores and self-rated familiarity with genetic terms was analyzed. For all four terms, participants that rated their familiarity as “familiar with term and know meaning” had significantly higher total knowledge scores compared to participants that rated their familiarity as “aware of term but unclear on meaning” or “never heard of term.” Participants that rated the term “gene” as “familiar with term and know meaning” had an average knowledge score of 59% ($SD=15\%$) compared to an average knowledge score of 48% ($SD=15\%$) for those who rated their familiarity as lower, $t(166)=-4.33$, $p<.001$. Similarly, participants that rated the term “DNA” as “familiar with term and know meaning” had an average knowledge score of 59% ($SD=15\%$), whereas participants that rated their familiarity as lower had an average knowledge score of 47% ($SD=16\%$), $t(166)=-5.00$, $p<.001$. Participants that rated the term “chromosome” as “familiar with the term and know meaning” had an average knowledge score of 62% ($SD=15\%$) compared to an average knowledge score of 47% ($SD=15\%$) for those who rated their familiarity as lower, $t(166)=-6.10$, $p<.001$. Participants that rated the term “genetic testing” as “familiar with term and know meaning” had an average knowledge score of 61% ($SD=15\%$), while participants that rated their familiarity as lower had an average score of 47% ($SD=15\%$), $t(166)=-6.00$, $p<.001$.

Perceived Risk

Forty-four participants (26%) believed they had a genetic form of PD, sixty-three participants (38%) did not believe they had a genetic form of PD, and sixty-one participants (36%) were unsure. When asked how likely they thought it was that they inherited a gene that causes PD, 57 (34%) believed it was overwhelmingly or very likely, 64 (38%) believed

it was somewhat likely, 24 (14%) thought it was unlikely, and 23 (14%) were unsure. Thirteen (8%) participants thought it was overwhelming or very likely that other family members would develop PD, 65 (39%) thought it was somewhat likely, 58 (35%) thought it was unlikely, and 32 (19%) were unsure.

Genetic Testing Attitudes and Beliefs

The majority of participants ($n=143$, 86%) believed genetic testing for PD would be useful and expressed interest in genetic testing ($n=98$, 59%) and genetic counseling ($n=93$, 56%) for PD. However, only a few participants had heard of any genetic tests for PD ($n=48$, 29%) or the gene *LRRK2* ($n=16$, 10%). Of the 48 participants that had heard of a genetic test for PD, 20 had considered obtaining a genetic testing for PD, with only 4 participants stating they had done so. When participants were asked why they were interested in pursuing genetic testing for PD the most common responses included an interest in learning more about oneself/curious about the result, to help research, and/or to provide information for family members (Table 3). Of the participants not interested in pursuing genetic testing for PD ($n=24$, 14.3%) most expressed that they were not interested because they already have PD and/or genetic testing would not directly benefit them (Table 3).

Participants were asked a number of open-ended response questions regarding the perceived benefits or risks, as well as perceived barriers for genetic testing. Most participants ($n=153$, 91%) believed that genetic testing for PD would have benefits, including helping to discover better treatments and preventions for PD as well as a cure (Table 4). Most participants ($n=105$, 63%) also believed that genetic testing for PD could have risks such as emotional or psychological distress and discrimination (Table 4). Participants were asked if they could think of any barriers that would prevent them getting a genetic test for PD assuming they wanted one. Thirty-five percent ($n=56$) believed there were potential barriers to pursuing genetic testing, particularly the cost of the test and insurance coverage (Table 4).

Participants were asked if they had any concern about the impact of genetic testing on their lives or on their loved ones. While only 20% ($n=33$) of participants responded “yes” they were concerned about the impact genetic testing may have on their lives, 43% ($n=72$) responded “yes” they were concerned about the impact genetic testing may have on their loved ones. Both questions were followed with an open-ended question asking what concerns the participants had about the impact of genetic testing. The most frequently reported concern was the impact genetic testing may have on other family members’ risk of developing PD. Participants were also concerned about how genetic testing may impact their personal and medical future, and that they would experience a negative emotional reaction (Table 5). The majority of participants that were concerned about the impact genetic testing may have on their loved ones reported that they were concerned about the possible negative emotional response their loved ones may experience from learning their risk. Participants were also concerned about their family member developing PD, confidentiality, and genetic discrimination (Table 5).

Discussion

This study presents the first report of both general genetics and PD-specific genetic knowledge in a sample of patients with PD. Most participants reported that they were familiar with and knew the meanings of general genetics terms but their overall knowledge levels were low. Interestingly, more participants self-reported a higher familiarity with the terms “gene” and “DNA” compared to the terms “chromosome” and “genetic testing” suggesting they may have more exposure to these terms. While average knowledge scores were low, participants had a higher score for general genetics knowledge compared to PD-specific genetics knowledge. Overall, participants were found to have relatively limited

knowledge of the genetics of PD, particularly the *LRRK2* gene, and genetic testing, but they demonstrated an understanding of PD symptoms, risk factor genes, familial PD risks, and penetrance. Based on the present results, there is evidence that individuals with PD may over-estimate their degree of familiarity with genetic concepts. It is apparent that the sample had an initial grasp of several important concepts, such as risk factor genes and penetrance, but they need further education not only about basic concepts such as genes, but also about the genetics of PD and genetic testing. It will be important to keep the differences in familiarity with the various genetic terms and the limited knowledge about genetic testing in mind when providing genetic counseling and education in this population.

A number of studies have been conducted to assess genetics knowledge in complex-disease populations. In a population-based study using a random digit dialing telephone survey, Rose and colleagues found that knowledge about cancer risk was generally high (Rose et al. 2005). Similar to the present results, they found that the majority of their participants demonstrated an understanding of the multifactorial nature of cancer. Lewis et al. (2009) assessed patient genetic knowledge regarding inflammatory bowel disease (IBD) and found they scored well on a 10 point genetics knowledge test administered after a 30 min. educational session on IBD and IBD genetics. However, despite high scores on the genetics knowledge test participants repeatedly offered incorrect information regarding basic genetic principles and the factors causing IBD during follow-up focus group sessions. Based on their results, the authors stressed the importance of possessing a working knowledge of both general and disease-specific genetic principles in order to comprehend the implications of genetic testing for complex genetic disease (Lewis et al. 2009).

In the present study, questions addressing concepts related to risk factor genes, penetrance, and familial risk for PD were answered correctly by the majority of participants. This finding suggests that while participants might have overall low genetics knowledge, they do have a prior understanding of important genetic concepts related to multifactorial, complex diseases. Nevertheless, patients with PD may need further education on basic genetic concepts, such as inheritance patterns and the definition of a gene, in order to have enough knowledge to truly understand genetic test results and familial PD risks. In developing patient-centered education and counseling approaches for the PD population, it will be important to provide individuals with the basic genetic information that is essential for having a working knowledge of multifactorial complex genetic disease.

Between 18 and 41% of patients with PD develop dementia over the course of the disease (Biggins et al. 1992; de Lau et al. 2005; Mayeux et al. 1992; Tison et al. 1995). Thirty-eight percent of the participants in this study scored below the normal range on the MoCA. Although further testing is required, it appears that a number of these individuals may have mild cognitive impairment or dementia. Particularly noteworthy, participants with MoCA scores below the normal range had significantly lower average knowledge scores (49%) compared to participants with MoCA scores in the normal range (57%). When offering genetic counseling and testing for PD, providers may want to consider evaluating the cognitive status of the patient and including a family member in the genetic testing and counseling process if the patient is cognitively impaired. Additionally, different education and counseling approaches may be required when the patient has dementia or mild cognitive impairment.

Studies of perceived risk are numerous in genetic testing and counseling literature. While 34% of respondents in this study thought it was overwhelmingly or very likely they had inherited a PD gene, only 8% thought it was overwhelmingly or very likely other family members would develop PD. This finding raises an interesting difference between perceived genetic risk to self and perceived risk to family members. Much of the literature on

perceived risk focuses on either the individual's perception of personal risk of disease, such as cancer, or personal risk for a genetic mutation in a predictive genetic testing setting, such as Huntington's disease. While multiple studies have shown that patients tend to overestimate risk (Croyle and Lerman 1999; Daly et al. 1996), few studies have compared risk to self vs. risk to family members. Rantala et al. (2009) used a questionnaire-based approach to investigate risk perception in a cancer genetic counseling session and found that while both risks were overestimated, the perceived risk for siblings/children was significantly lower than the individuals' perception of their personal risk (Rantala et al. 2009). In the present study, the cohort's understanding of the multifactorial nature of PD combined with the fact that they already have a diagnosis of PD may have contributed to the observed difference in perceived risk to self vs. risk to family members. Specific reasons for the difference between perceptions of personal risk and risk to family members and how this difference may impact genetic counseling and education in the PD population warrants exploration.

The findings also underscore the need for genetics education in the PD population, and they demonstrate that patients are interested in this information. The majority of participants (86%) thought genetic testing for PD would be useful, and 59% of participants were interested in genetic testing. However, only 10% of participants had heard of the gene *LRRK2* and only 29% had heard of any genetic tests for PD, revealing a possible discrepancy between patient interest and awareness of an available clinical service. Clinical genetic testing options for PD became available within the past decade; however, the first genes available for testing were applicable to only a small subset of the patient population. With the discovery of *LRRK2*, clinical genetic testing for PD entered a new era, as *LRRK2* mutations are estimated to account for 1% of sporadic PD and 5%–6% of familial PD (Farrer et al. 2005). Given that 15% of participants reported a family history of PD with at least one affected first-degree relative, there is clearly a need to improve patient education and awareness of clinical genetic testing services. While *LRRK2* will not explain the majority of the positive family histories observed in this study, patients still should be provided with the information so that they can decide whether they want genetic testing. Methods for improving patient awareness and understanding of genetic testing services in the PD population need to be addressed. The use of written materials, such as pamphlets or referrals to verified patient education websites, represent possible solutions that could be implemented to address this need.

Many participants believed genetic testing would help advance scientific knowledge of PD and possibly lead to better treatments, a cure, and/or prevention. In a recent study, 64 individuals who had or were at risk for Huntington disease, breast cancer, or alpha-1 antitrypsin deficiency were interviewed, and many believed the discovery of a gene inferred that treatment existed or would shortly be developed and that there was a potential for preventive therapies (Klitzman 2010). While these benefits are often cited as a possible long-term outcome of participation in genetic research, they are not a direct benefit of clinical genetic testing. This finding highlights a possible lack of understanding regarding the differences between clinical and research genetic testing. Therefore, while 59% of the present sample stated they were interested in genetic testing, this may not accurately reflect how many individuals are interested in clinical genetic testing for PD. Interestingly, one study assessing interest in predictive genetic testing for first-degree relatives of patients with PD found that there was only moderate interest in predictive testing since preventative therapy is currently not available (Dahodwala et al. 2007). Not surprisingly, interest in predictive genetic testing increased based on the hypothetical availability of clinical trials or a proven therapy. Participants rated assistance in making later-life decisions and the ability to help researchers as the most important reasons to get predictive genetic testing at this time (Dahodwala, et al. 2007).

When participants were asked whether or not they had concerns regarding the impact of genetic testing, 20% reported concern about the impact genetic testing may have on their own life, while 43% were concerned about the impact genetic testing may have on their loved ones. This two-fold difference raises important genetic counseling implications. Currently, genetic testing for PD has few clinical implications in terms of the care of the symptomatic patient. Arguably, the main clinical impact of a positive genetic test result is to provide insight into the biological cause of disease, and in turn use the information to evaluate genetic risks for family members. Clinicians and genetic counselors who provide genetic testing for PD need to be aware of and sensitive to patients' concerns regarding the impact of genetic testing on their loved ones. Pre-test counseling will be imperative in making sure patients have an accurate and realistic understanding of the potential impact. For example, an explicit discussion of the possible psychological impact of genetic testing, such as guilt or depression, should be initiated with patients. The purpose of such discussions is not to influence genetic testing decisions, but to best prepare individuals for all possible outcomes of the testing process. In a 2005 article, Susan McDaniel directly addresses the impact genetic testing has on family dynamics and states that "guilt over passing on a 'bad gene' to children is one of the most powerful reactions of people with genetic conditions" (McDaniel 2005, p.33). McDaniel advocates for an interdisciplinary approach to best support individuals and families facing a genetic risk of disease. Approaches for providing genetic counseling services, such as a referral network or integration of a staff genetic counselor, should be explored by PD clinical care centers.

Insurance and/or employment discrimination was the second most common open-ended response when participants were asked about their perceptions of the risk involved in genetic testing for PD. Concerns about genetic discrimination have been well documented in previous studies (Cameron et al. 2009; Lewis et al. 2009; Manne et al. 2007; Morren et al. 2007). A preliminary survey conducted on patients with PD and caregivers found that health effects and insurance and employment discrimination were major concerns regarding genetic testing for PD (Tan, et al. 2007). Given the existence of the Genetic Information Nondiscrimination Act (GINA), it is possible that many of these concerns would be lessened after patients are made aware of its existence. A few participants also expressed concern over the impact genetic testing may have on their medical futures, as well as how genetic testing was done and if there were physical risks associated with the procedure; these concerns could all be easily addressed during a standard genetic counseling session. Follow-up studies would need to be performed to investigate whether patients with PD have fewer concerns regarding the possible impact of genetic testing after receiving genetic counseling.

Study Limitations

The present study was completed within a specialty PD clinic population, in which many patients had previously participated in some form of either PD-related or genetic research activity. Thus, the nature of the sample presents an apparent limitation regarding generalizability of findings, as the participants may not represent a true cross-section of the PD patient population. In addition, participants were mostly Caucasian and highly educated. Fifteen percent reported a family history of PD in at least one first degree relative, which is at the high-end of the range reported in other studies (10–16%) (Elbaz et al. 1999; Marder et al. 2003; Taylor et al. 1999; Uitti et al. 2005). Given an overall response rate of 44%, it is possible that those individuals with a family history of PD and/or higher levels of education were more likely to respond to our survey. A response bias was observed in this study with a higher response rate for participants that were recruited in person during their appointment with their neurologist compared to those that were recruited through the mail.

The majority of questionnaires were completed at home, and it is possible participants received some assistance in answering questions. Therefore, their actual general genetic and

PD-specific knowledge levels may be lower. Conversely, 38% of the sample scored below normal on the MoCA, and thus the low knowledge scores may have been due to difficulties understanding the questions rather than to a lack of general genetics and PD-specific knowledge. It is also important to note that knowledge levels in the population may be lower than demonstrated in this sample if individuals who felt they did not know enough to answer the questions did not return the survey.

Conclusion

Patients with PD have a high level of interest in genetics and genetic testing, but as an overall group, demonstrate a low level of genetics knowledge. Furthermore, while most individuals were interested in genetic testing for PD, the majority were unaware of *LRRK2* or any other genetic tests for PD. These findings highlight a considerable need for genetics education and counseling in the PD population. The results of this study were used to guide the development of genetic education and counseling tools specific to the PD population to provide patients with the ability to make informed and knowledgeable genetic testing decisions.

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Fig. 1.
Self rating of familiarity with genetic terms ($n=168$)

Fig. 2. Average correct responses by concept category. *Black bars* correspond to general genetics knowledge categories. *Grey bars* correspond to PD-specific knowledge categories ($n=168$)

Table 1

Sociodemographic and clinical description of participants

Characteristics	Frequency (%) (n=168)
Sex	
Male	99 (58.9)
Female	69 (41.1)
Age, years [mean (range)]	66.7 (34–85)
Marital Status	
Married	133 (79.2)
Divorced/Separated/Widowed	28 (16.7)
Single	7 (4.2)
Children	
Biological	145 (86.3)
Adopted, only	8 (4.8)
None	15 (8.9)
Education	
High school or less	20 (11.9)
2 year college degree or some college	35 (20.8)
4 year college degree	55 (32.7)
Postgraduate degree	58 (34.5)
Employment Status	
Retired	98 (58.3)
Employed	42 (25.0)
Disability	23 (13.7)
Unemployed	5 (3.0)
Ethnicity	
Caucasian, non-Hispanic	161 (95.8)
Asian	3 (1.8)
Hispanic	1 (0.6)
African American	1 (0.6)
Other	2 (1.2)
Religion	
Christian, non-Catholic	68 (40.5)
Catholic	63 (37.5)
Jewish	28 (16.7)
Other	7 (4.2)
Annual Family Income	
>\$100 000	105 (62.5)
≤\$100 000	54 (32.1)
Family History of PD	54 (32.1)
Family History of Other Neurodegenerative Disease	48 (28.6)
Age of onset of PD, years [mean (range)]	58.1 (26–82)

Characteristics	Frequency (%) (<i>n</i> =168)
Disease Duration of PD, years [mean (range)]	8.8 (1–35)
MoCA (<i>n</i> =155)	
Normal (26–30 points)	91 (58.7)
Below normal (<26 points)	64 (41.3)

Table 2

Example knowledge questions for each concept category and frequency of correct responses for the 168 participants. Correct responses are italicized

Concept Category	Question	N (%)
Inheritance	Which of the following statements about chromosomes is true? a. A child inherits equal numbers of chromosomes from its mother and father b. A child inherits a random assortment of chromosomes from its mother and father c. A child inherits all their chromosomes from their father d. A child inherits all their chromosomes from their mother	108 (64.3)
Risk Factor Genes	A woman has been told that she carries a mutation associated with breast cancer. How does this influence her likelihood of developing breast cancer? a. Her risk is no different from any other healthy woman b. She is at an increased risk for breast cancer c. She will definitely get breast cancer d. She already has breast cancer since she carries the mutated gene	155 (92.3)
Gene Functions and Mutations	An individual was found to have a familial mutation in a gene associated with breast cancer. In which cells of the body can the mutation be found? a. Only in the cancer cells b. Only in the breast tissue cells c. In all cells of female individuals, but not male individuals d. In all cells of female and male individuals	94 (56.0)
Genes, DNA, and chromosomes	How are genes, DNA, and chromosomes related? a. Genes are made up of DNA units and DNA is packaged into chromosomes b. Genes are separate units from both DNA and chromosomes c. Genes are found only in chromosomes and not in DNA d. Genes are found in DNA and not in chromosomes	110 (65.5)
Genetic Testing	If a genetic cause (mutation) was identified in a family and a family member did not inherit this mutation they could never develop Parkinson disease. a. Agree b. Neither agree or disagree c. Disagree	94 (56.0)
Penetrance	Even if an individual has a mutation in a gene for Parkinson disease, they may not develop Parkinson disease. a. Agree b. Neither agree or disagree c. Disagree	112 (66.7)
LRRK2	LRRK2 is a common genetic contributor to both familial and sporadic late-onset Parkinson disease. a. Agree b. Neither agree or disagree c. Disagree	10 (6.0)

Concept Category	Question	N (%)
Familial PD Risks	If your relatives were at risk to develop Parkinson disease, when do you think they would no longer be at risk for the disease?	107 (63.7)
	a. After age 50	
	b. After age 70	
	c. After age 90	
	d. Relatives would always be at risk	
PD Genetics	Have researchers discovered a gene or genes for Parkinson disease?	43 (25.6)
	a. Yes	
	b. No	
	c. Don't know	
PD Symptoms	Drugs are available to treat the symptoms of Parkinson disease.	166 (98.8)
	a. Agree	
	b. Neither agree or disagree	
	c. Disagree	

Table 3

Summary of themes regarding reasons participants were or were not interested in pursuing genetic testing for Parkinson disease

Theme ^a	Frequency (%) ^b
Reasons why interested (n=98)	
Personal information and curiosity	30 (30.6)
Help research	26 (26.5)
Information for family	22 (22.4)
Altruism	7 (7.1)
Identify cause of Parkinson disease	5 (5.1)
Confirm diagnosis	4 (4.1)
Other	9 (9.2)
Reasons why not interested (n=24)	
Already have Parkinson disease	8 (33.3)
No personal benefit	7 (29.2)
Need more information	2 (8.3)
Don't want to know	2 (8.3)
Other	4 (16.7)

^a Although there were 168 participants, 46 participants (27.4%) answered "Don't know" when asked if interested in pursuing genetic testing for PD and were not coded

^b The frequency data adds to more than 100% because responses could be coded for multiple themes

Table 4

Summary of themes regarding participants' perceived risks and benefits of and barriers to pursuing genetic testing for PD

Theme	Frequency (%) ^a
Benefits (n=168)	
Discover better treatments and prevention	45 (26.8)
Discover a cure	43 (25.6)
Earlier detection and/or treatment	25 (14.9)
Advance scientific knowledge of disease	23 (13.7)
Define genetic risks	21 (12.5)
To make life planning decisions	12 (7.1)
None or no current benefit	6 (3.6)
Other	15 (8.9)
Don't know	17 (10.1)
Risks (n=168)	
None	64 (38.1)
Emotional and/or psychological stress	35 (20.8)
Insurance and/or employment discrimination	26 (15.5)
Confidentiality	8 (4.8)
Other	18 (10.7)
Barriers (n=161)	
Cost and/or insurance coverage	27 (16.8)
Ethical, social, or legal risks associated with genetic testing	8 (5.0)
Concern over procedure type and physical risks associated with procedure	8 (5.0)
Availability and/or convenience	5 (3.1)
Other	15 (9.3)
None	105 (65.2)

^aThe frequency data adds to more than 100% because responses could be coded for multiple themes

Table 5

Summary of themes regarding participants' concerns about the possible effects of genetic testing

Theme ^a	Frequency (%) ^b
Impact genetic testing may have on their life (<i>n</i> =33)	
Impact on and risk for family members	13 (39.4)
Negative emotional impact	5 (15.2)
Impact on medical and personal future	5 (15.2)
Discrimination and confidentiality	3 (9.1)
Unwanted information	2 (6.1)
No concern, instead listed a positive outcome	4 (12.1)
Other	4 (12.1)
Impact of genetic testing on their loved ones (<i>n</i> =72)	
Family member at risk of developing PD	18 (25.0)
Negative emotional impact on loved one	24 (33.3)
Impact on life planning and lifestyle	3 (4.2)
Discrimination and confidentiality	7 (3.7)
Unwanted information	2 (2.8)
No concern, instead listed a positive outcome	4 (5.6)
Other	7 (9.7)
Not able to interpret or don't know what concerns are	8 (11.1)

^a Four different participants for each question did not express concerns in their open-end response and instead listed perceived potential benefits of genetic testing, and thus were not in the total *n* coded for this table

^b The frequency data adds to more than 100% because responses could be coded for multiple themes