

Neuropsychological Testing Is More Useful Than Genetic Testing In The Baseline PD Evaluation

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Case Study 1

- 65yo physician presents to consultation because of asymmetric left hand rest tremor
- In further questioning he reports forgetfulness and word finding difficulty.
- His office manager hinted he may benefit from early retirement, but he thought his tremor is too mild

In this specific case, neuropsychological testing is clearly key

Case Study 2

- 42yo lawyer presents to consultation because of asymmetric left hand rest tremor
- Cognitive functioning reportedly intact. MoCA score 30/30.
- She is not bothered by the tremor, but would like to enroll in the most appropriate cutting edge studies for her

In this specific case, she would extremely benefit from genetic testing

Genetic Testing in PD

- Rarely offered to patients in the USA
- In a survey of PD clinicians, 41% had not referred any PD patients for genetic testing in the last year and >80% reported referring fewer than 11 patients (Alcalay, 2019)
- Most common reasons for not referring:
 - Lack of insurance coverage/cost
 - Lack of perceived utility
 - Level of comfort in clinicians' own ability to genetically counsel PD patients on *GBA* and *LRRK2* (score 52 out 100)

Reasons for Testing in PD

- Genetic data may determine clinical trials eligibility
- Patients' interest: 23andMe >10k PD participants (Blauwendraat, 2019)
- Genetic data can help infer on prognosis (e.g., *GBA*, faster progression; Cilia, 2016)
- In early-onset PD, diagnostic testing and family planning

Press Releases

Sanofi Initiates Phase 2 Clinical Trial to Evaluate Therapy for Genetic Form of Parkinson's Disease

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LTI-291

OVERVIEW

Phase 2 Clinical Trial of PRO1A in Patients With Parkinson's Disease With at Least One GBA1 Mutation (PROPEL)

ClinicalTrials.gov

ClinicalTrials.gov

A Study to Evaluate the Safety, Tolerability, and Pharmacokinetics of BSB04 in Adults With Parkinson's Disease (REASON)

GBA

The Science of Parkinson's disease

Plain English information about Parkinson's disease provided by scientists doing the research.

ABOUT | ALPHA SYNUCLEIN | BIOLOGY OF THE BLOOD | DIAGNOSIS | GENETICS OF PD | LECTURES | PARKINSON'S DISEASE | SITE MAP | THE BREATHER ANALYSIS STUDY


DECEMBER 21, 2017

Inhibiting LRRK2: The Denali Phase I results

SEARCH

LRRK2

Should We Offer Genetic Testing for People with PD?



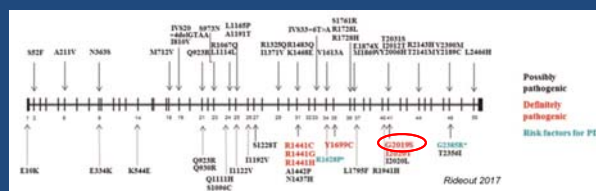
PD GENERation

- Funded by the Parkinson's Foundation
- **Primary goal:** to make genetic testing accessible for people with PD and test:
 1. Feasibility of delivery to large number of participants
 2. The effect (impact and satisfaction) from receiving the results
 3. Compare delivery of the genetic testing results delivered by clinician vs genetic counselor

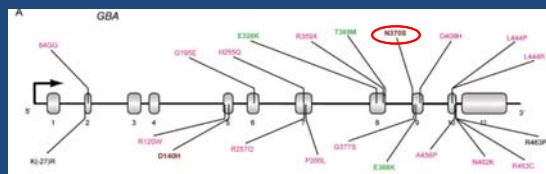


23&Me Reports Two PD Variants

LRRK2



GBA

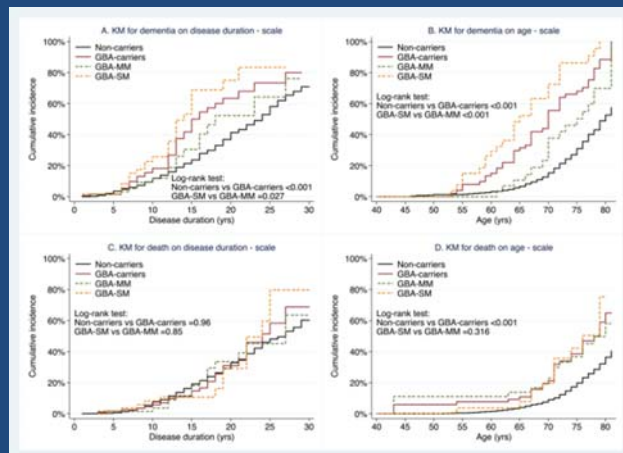


Case Study 3

- 75yo retired teacher presents to consultation because of asymmetric left hand rest tremor
- Mild cognitive complaints. Do not interfere with work.
- He is very curious to know why he developed PD. He is considering in participating in 23andMe to find out if he carries any genetic mutations

In this specific case, genetic counseling and testing may be superior to direct to consumer testing.

GBA Carriers: Rate of Progression



← Dementia

← Death

Cilia, 2017

- Similar findings by Winder-Rhodes, 2013; Thaler, 2016 (Gaucher versus heterozygotes); Liu, 2017

Case Study 4

- 68yo retired architect presents to consultation because of asymmetric left hand rest tremor
- Mild cognitive complaints. Do not interfere with activities of daily living.
- Of Ashkenazi Jewish ancestry. Would like to know what is her prognosis for disease progression and cognitive dysfunction

In this specific case, genetic testing for *LRRK2* and *GBA* may help predict long term outcomes

Case Study 5

- 38yo IT developer present with left hand tremor. His brother has PD and successful DBS at age 32.
- He and his spouse are going through fertility treatments and would like to know the risk for an offspring
- He tests positive for *PRKN*. His wife is a non-carrier

In this specific case, genetic testing for may help with family planning

Cases in which Genetic Testing May Be Superior to Neuropsychological Testing

- In those interested in novel clinical trials
- In those who want to know why they developed PD
- Genetic data can help infer on prognosis
- In early-onset PD, diagnostic testing and family planning



Should we offer genetic testing in Parkinson's Disease?