MEDGENOME | ACTIA Inherited Disease Genetics

CAP ACCREDITED COLLEGE of AMERICAN PATHOLOGISTS

Parkinson's Disease (PD)

- One of the most common neurodegenerative disorder affecting ~ 1-2% of the population above 60 years of age
- Monogenic mutations contributes to ~30% of familial cases and remaining are primararily polygenic in nature.

Genes (monogenic) Implicated in PD Etiology

Autosomal Dominant

SNCA, CHCHD2, VPS35, LRRK2

Autosomal Recessive

PLA2G6, FBXO7, DNAJC6, SYNJ1, VPS13C, PINK1, DJ1, PRKN, ATP13A2, DNAJC6

Evolving Evidence

ADORA1, PODXL, GBA, CSMD1, DNAJC13, TMEM230, LRP10, NR4A2, PANK2, PARL, PNXNA4, RIC3, TNK2, TNR, SNCAIP, CSF1R, EIF4G1, UCHL1, GIGYF2, HTRA2, NUS1*

*Risk

Test Details

Test Code	Test Name	Methodology	TAT
MGM126	Early-onset juvenile parkinsonism gene panel	NGS	19 days
MGM2436	*PRKN* gene deletion/duplication analysis	MLPA	14 days
MGM3325	(Combo) Monogenic Parkinson Disease Panel	NGS & MLPA	14 days
GES008	Polygenic Risk Score for **PD**	WGGA	14 days

WGGA - Whole Genome Genotyping Array | NGS - Next Generation Sequencing
MLPA - Multiplex ligation-dependent probe amplification

RESEARCH ARTICLE

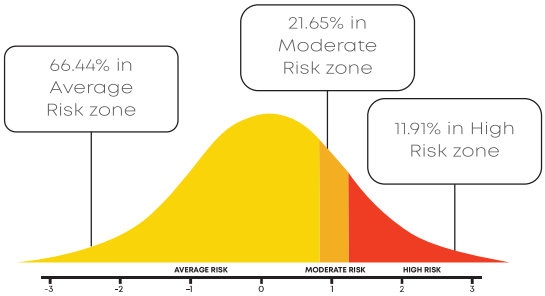
The Genetic Drivers of Juvenile, Young, and Early-Onset Parkinson's Disease in India

"In our study of 576 subjects with PD, almost all pathogenic PRKN variants were deletions (22/25 cases), most of them homozygous (20/22) involving 3rd and/or 4th exon"

Scan for full Research Paper

Polygenic Risk Score (PRS) Test

This is a screening test that helps estimate the genetic predisposition of an individual to develop PD. The risk is calculated from ~2000 genetic markers implicated in the disease and is given as Polygenic Risk Score (PRS).



Parkinson's RISK SCORE (PDRS)

Indicates if the individual is at high, moderate or average genetic risk.
Assay is based on study in Indian cohort

Talk to the Experts:

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