

Precision Medicine

Genetics of Parkinson's Disease

What is Parkinson's disease and what's my risk?

Parkinson's disease (PD) is a progressive movement disorder. The main features are tremor, slowness, stiffness, and walking/balance problems, as well as memory problems and other non-movement symptoms. In the general population, the lifetime risk of developing PD is 4-9%. Age is the strongest risk factor for developing the disease. Men are diagnosed with PD at a higher rate than women.

Is Parkinson's disease genetic?

There are three types of PD:

- Juvenile-onset PD diagnosed before age 20 accounts for 2% of PD cases.
- Early onset Parkinson's disease (EOPD) diagnosed between ages 20-50 accounts for 10-20% of PD cases.
- Late onset Parkinson's disease (LOPD) diagnosed after age 50 the predominant form of PD.

Family history of PD may be consistent with sporadic, familial, or monogenic PD.

- Sporadic PD (75% of all cases) is characterized by an isolated case in the family. The PD occurred sporadically or randomly.
- Familial PD (15-20% of all cases) is characterized by disease that occurs in more than 1 individual in a family. The PD in the family happened due to an interaction between environmental and minor genetic factors (multifactorial).
- Monogenic PD (5-10% of all cases) is characterized by disease that occurs due to a pathogenic variant or mutation in a gene linked to PD: GBA, LRRK2, PRKN, PINK1, SNCA, and others.

What are my options for genetic testing for Parkinson's disease?

<u>For unaffected individuals:</u> Genetic testing for sporadic or familial PD provides limited information about a person's chance of developing or preventing the disease. Many of the common genes and variants linked to PD are considered risk factors and do not guarantee one will develop PD. Genetic testing is not recommended due to limited usefulness in medical care. If you have a relative with a known mutation in a gene linked to PD, predictive genetic testing can be considered only after pre-test counseling in a specialized clinic that includes a neurologic evaluation.

<u>For individuals concerned they have Parkinson-like symptoms:</u> In order to consider genetic testing for PD, you must have a diagnosis. Please ask your primary care physician to refer you to a neurologist for an evaluation.

<u>For individuals diagnosed with Parkinson's disease:</u> Genetic testing for PD can be considered and discussed with a genetic counselor. Please ask your neurologist about receiving a referral.

References:

https://www.ncbi.nlm.nih.gov/pmc/articles/PMC8101316/ https://www.ncbi.nlm.nih.gov/pmc/articles/PMC8304082/ https://www.michaeljfox.org/

Additional Resources

For additional resources, please visit our website: https://www.TriHealth.com/Institutes-And-Services/Precision-Medicine

