PINK1 GENE INFORMATION

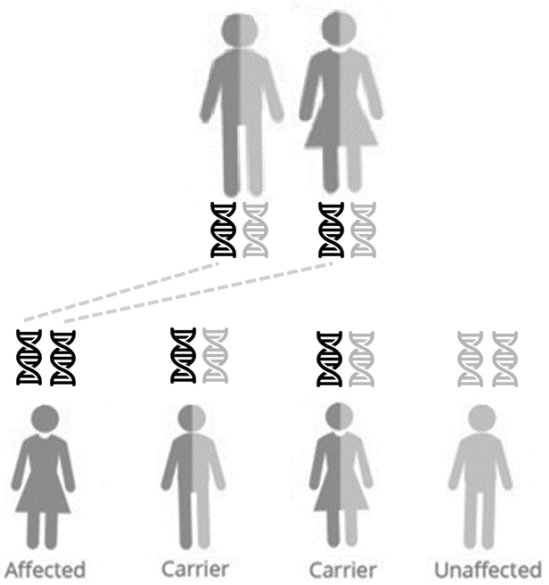
PARKINSON'S DISEASE FACT SHEET

Parkinson's disease is multifactorial in most families; it is likely caused by an interplay of both genetic and environmental factors. A number of genes have been identified as risk factors for Parkinson's disease, with many others likely unknown. This fact sheet outlines basic information about the *PINK1* gene as well as risk to individuals and their relatives. If you have a *PINK1* variant and/or are concerned about your risk, speaking with a genetic counselor can be helpful.

PINK1 Variants

Gene variants in *PINK1* are associated with features of classic Parkinson's disease, but symptoms are usually slower in progression, and may have less non-motor features.

PINK1 variants are inherited in an autosomal recessive pattern (see Figure). This means that a person has to inherit two gene variants, one from each parent, to have the disease. Individuals with only one variant are sometimes called carriers and typically will not have the disease. If a person has two recessive variants, we can assume that one variant came from each of their parents. Individuals with two recessive variants will automatically pass one variant on to each child. Both males and females can pass on and inherit these variants.



Having two variants in the *PINK1* gene almost always causes Parkinson's disease and usually at a much younger age, such as before age 40 years. Both parents and all children of the affected person will have at least one copy of the variant (see Figure). Individuals with just one gene variant will usually not have Parkinson's disease, however there is evidence to suggest they might be at a slightly increased risk.

Research Opportunities

Many research studies are interested in enrolling people with *PINK1* variants. This is true whether or not that person or anyone else in their family has Parkinson's disease. Some studies are trying to understand more about the causes of Parkinson's disease. Other studies are working to develop new treatments. Trials of new medications for people with Parkinson's disease and *PINK1* variants are ongoing.

Various research projects at the Indiana University School of Medicine include genetic testing and genetic counseling for Parkinson's disease. These projects enroll volunteers with and without Parkinson's disease. To learn more, please contact our team at wrd@iu.edu or call 888-830-6299.

References

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