

What's Hot in Parkinson's Disease?

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Parkinson's Disease Linked to Gaucher's Disease: A Rare Disease Informs Us About a Common One

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There has been a “snowball effect” pertaining to genetic research in Parkinson's disease. Even researchers uninterested in genetics have been finding the DNA links to be fascinating, and potentially enlightening to the underlying disease pathogenesis. In the October edition of the *New England Journal of Medicine*, another genetic link has emerged—this time to a rare disorder called Gaucher's disease, a rare inherited disorder of fat metabolism that causes spleen and liver enlargement, abnormal fragility and pain of the bones, and progressive neurologic disturbances, leading to early death.

There is a gene in the body that codes for an important enzyme called glucocerebrosidase which functions to break down glucocerebroside (fat). If this fat is not broken down properly, it can damage multiple organs potentially including the brain. Interestingly, the enzyme functions in part of the cell called the lysosome, or the trash compactor. Many scientists believe part of the problem in Parkinson's disease is in the abnormal processing of protein by the body's “trash compactors.” If you have two copies of the Gaucher's gene you will develop fulminant Gaucher's disease. If you have one copy, you are referred to as a gene carrier. Gene carriers for Gaucher's disease, according to this new research study, have a five times increased risk of developing the symptoms of Parkinson's disease. It is important to point out that there is much disagreement between experts if this Gaucher's syndrome is true Parkinson's disease, or rather parkinsonism (a group of similar looking symptoms).

Several alterations of the genetic patterns in Gaucher's carriers were located by collaborating researchers within this study. Ashkenazi Jews represented the highest risk group for disease (as they did in the LRRK2 Parkinson gene studies). It is important to note that the majority of Parkinson's disease sufferers have no known genetic defect. Speculation has therefore turned to explore why some Parkinson's disease patients fail to have single gene defects. The answer may lie in a simple genetic theory—the genes load the gun and the environment pulls the trigger. More research into susceptible genetic backgrounds will be needed, as well as research into potential environmental triggers (e.g. pesticides). Studies such as this recently published article teach us that even rare diseases can inform us about common diseases.

Reference:

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