

Disease causing mutation found in French-Canadians

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Dr. Ziv Gan-Or is a genetics researcher at The Neuro, and the study's lead author. Credit: Ziv Gan-Or

A team of Canadian scientists, including researchers at the Montreal Neurological Institute and Hospital (The Neuro) has discovered the first French-Canadian founder mutation gene linked to synucleinopathies, a group of neurodegenerative diseases that includes Parkinson's disease (PD), dementia with Lewy-Bodies (DLB) and multiple system atrophy (MSA).

The mutation, known as p.Trp378Gly, is in the GBA gene, one of the most prominent [genes](#) involved in PD. The most common GBA mutations create a small risk of developing synucleinopathies, but in combination with other [mutations](#) the risk can increase dramatically.

The study found that one per cent of French Canadians with synucleinopathies have the GBA mutation. This mutation is specific to French Canadians and was probably inherited from a single common ancestor.

People who inherit this gene mutation from one parent become carriers of the mutation and are at risk of developing synucleinopathies. Those who inherit this gene mutation in addition to another mutation in GBA from both parents will develop Gaucher's [disease](#) (GD). GD symptoms include enlargement of the spleen and liver, low red blood cell level, lung disease, arthritis, bone pain and fractures, but symptoms vary among [patients](#).

"We don't know how frequent this mutation is in the general French-Canadian population, but it doesn't appear to be very common. We're

going to test additional subjects so we can determine its exact frequency," says Dr. Ziv Gan-Or, a genetics researcher at The Neuro and the study's lead author. "This mutation causes Gaucher's disease, so it has clinical importance for couples who want to have children. They can request genetic screening for this specific [gene mutation](#)."

The study involved genetic examination of 891 control patients, 436 PD patients, and 189 REM-sleep Behaviour Disorder (RBD) patients whose condition has a strong link to synucleinopathies. All PD and RBD patients were of French Canadian ancestry. The results were published in the print version of the journal *Clinical Genetics* on Oct. 1, 2018.

More information: J.A. Ruskey et al, The GBA p.Trp378Gly mutation is a probable French-Canadian founder mutation causing Gaucher disease and synucleinopathies, *Clinical Genetics* (2018). [DOI: 10.1111/cge.13405](https://doi.org/10.1111/cge.13405)

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