CSMD1 gene mutations could lead to familial Parkinson disease

Mutation of the CSMD1 gene - which encodes a protein that participates in complement activation and inflammation in the CNS – leads to familial Parkinson disease (PD) in the absence of other gene mutations, new research published in *Neurology: Genetics* suggests. The team performed whole-exome sequencing in two unrelated Spanish families with PD, in which known gene mutations known to cause PD were excluded. Single nucleotide substitutions and short insertions or deletions were sought after sequencing. Mutations of the CSMD1 gene have already been implicated in schizophrenia and Alzheimer disease. Taken together, the researchers conclude that the complement pathway may offer an important therapeutic target in PD and other neurodegenerative conditions.

Original article Ruiz-Martinez, J. et al. Whole-exome sequencing associates novel CSMD1 gene mutations with familial Parkinson disease. Neurology: Genetics. 2017. Doi: 10.1212/NXG.0000000000000177.