

Documents

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The LRRK2 p.L1795F variant causes Parkinson's disease in the European population

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Abstract

LRRK2-PD represents the most common form of autosomal dominant Parkinson's disease. We identified the LRRK2 p.L1795F variant in three families and six additional unrelated cases using genetic data from over 50,000 individuals. Carriers with available genotyping data shared a common haplotype. The clinical presentation resembles other LRRK2-PD forms. Combined with published functional evidence showing strongly enhanced LRRK2 kinase activity, we provide evidence that LRRK2 p.L1795F is pathogenic. © The Author(s) 2025.

References

- Westenberger, A.
Relevance of genetic testing in the gene-targeted trial era: the Rostock Parkinson's disease study
(2024) *Brain*, 147, pp. 2652-2667.
39087914, 11292909
- Cook, L.
Parkinson's disease variant detection and disclosure: PD GENERation, a North American study
(2024) *Brain*, 147, pp. 2668-2679.
39074992, 11292896
- Taylor, M., Alessi, D.R.
Advances in elucidating the function of leucine-rich repeat protein kinase-2 in normal cells and Parkinson's disease
(2020) *Curr. Opin. Cell Biol*, 63, pp. 102-113.
1:CAS:528:DC%2BB3cXitFeqtL8%3D, 32036294, 7262585
- Kalogeropoulou, A.F.
Impact of 100 LRRK2 variants linked to Parkinson's disease on kinase activity and microtubule binding
(2022) *Biochem. J*, 479, pp. 1759-1783.
1:CAS:528:DC%2BB38XisV2qu7vK, 35950872
- Nichols, W.C.
LRRK2 mutation analysis in Parkinson disease families with evidence of linkage to PARK8
(2007) *Neurology*, 69, pp. 1737-1744.
1:CAS:528:DC%2BD2sXht1Shu7jL, 17804834

- Benitez, B.A.
Resequencing analysis of five Mendelian genes and the top genes from genome-wide association studies in Parkinson's Disease
(2016) *Mol. Neurodegener*, 11, p. 29.
27094865, 4837564
- Illés, A.
The role of genetic testing in the clinical practice and research of early-onset parkinsonian disorders in a hungarian cohort: increasing challenge in genetic counselling, improving chances in stratification for clinical trials
(2019) *Front. Genet*, 10, p. 1061.
31737044, 6837163
- Ostrozovicova, M.
L1795F LRRK2 variant is a common cause of Parkinson's disease in Central Europe
(2024) *Res. Sq*,
p.,., <https://>
- Pitz, V.
Analysis of rare Parkinson's disease variants in millions of people
(2024) *NPJ Parkinsons Dis*, 10, p. 11.
1:CAS:528:DC%2BB2cXpsVOmug%3D%3D, 38191580, 10774311
- McFarthing, K.
Parkinson's Disease Drug Therapies in the Clinical Trial Pipeline: 2022 Update
(2022) *J. Parkinsons. Dis*, 12, pp. 1073-1082.
1:CAS:528:DC%2BB38XhtlCkt7%2FN, 35527571, 9198738
- Kluss, J.H., Lewis, P.A., Greggio, E.
Leucine-rich repeat kinase 2 (LRRK2): an update on the potential therapeutic target for Parkinson's disease
(2022) *Expert Opin. Ther. Targets*, 26, pp. 537-546.
1:CAS:528:DC%2BB38Xhslams7fN, 35642531
- Trujillano, D.
A comprehensive global genotype-phenotype database for rare diseases
(2017) *Mol. Genet. Genomic Med*, 5, pp. 66-75.
28116331
- Sun, K.Y.
A deep catalog of protein-coding variation in 985,830 individuals
(2023) *bioRxiv*,
38234819, 10793442
- Sudlow, C.
UK biobank: an open access resource for identifying the causes of a wide range of complex diseases of middle and old age
(2015) *PLoS Med*, 12.
25826379, 4380465
- *Movement Disorder Society Genetic mutation database (MDSGene)*,
- Saunders-Pullman, R., Raymond, D. & Elango, S. LRRK2 Parkinson Disease. (eds. Adam, M. P. et al.) (University of Washington, Seattle, Seattle (WA), 2006
- Zimprich, A.
Mutations in LRRK2 cause autosomal-dominant parkinsonism with pleomorphic pathology
(2004) *Neuron*, 44, pp. 601-607.
1:CAS:528:DC%2BD2cXhtVKhurjL, 15541309

- Borsche, M.
The New p.F1700L LRRK2 variant causes Parkinson's disease by extensively increasing kinase activity
(2023) *Mov. Disord*, 38, pp. 1105-1107.
36971062, 10947214
- Kim, J.S.
A Korean Parkinson's disease family with the LRRK2 p.Tyr1699Cys mutation showing clinical heterogeneity
(2012) *Mov. Disord*, 27, pp. 320-324.
1:CAS:528:DC%2BC38Xhs1Glur%2FE, 22162019
- Wszolek, Z.K.
German-Canadian family (family A) with parkinsonism, amyotrophy, and dementia - Longitudinal observations
(1997) *Parkinsonism Relat. Disord*, 3, pp. 125-139.
1:STN:280:DC%2BD1cvitFyisQ%3D%3D, 18591067
- Khan, N.L.
Mutations in the gene LRRK2 encoding dardarin (PARK8) cause familial Parkinson's disease: clinical, pathological, olfactory and functional imaging and genetic data
(2005) *Brain*, 128, pp. 2786-2796.
16272164
- **GP2: The Global Parkinson's Genetics Program**
(2021) *Mov. Disord*, 36, pp. 842-851.
Global Parkinson's Genetics Program
- Lange, L.M.
Elucidating causative gene variants in hereditary Parkinson's disease in the Global Parkinson's Genetics Program (GP2)
(2023) *NPJ Parkinsons Dis*, 9, p. 100.
37369645, 10300084
- Towns, C.
Defining the causes of sporadic Parkinson's disease in the global Parkinson's genetics program (GP2)
(2023) *NPJ Parkinsons Dis*, 9, p. 131.
37699923, 10497609
- Regier, A.A.
Functional equivalence of genome sequencing analysis pipelines enables harmonized variant calling across human genetics projects
(2018) *Nat. Commun*, 9, pp. 1-8.
1:CAS:528:DC%2BC1cXhvVKnsbrE
- Poplin, R.
A universal SNP and small-indel variant caller using deep neural networks
(2018) *Nat. Biotechnol*, 36, pp. 983-987.
1:CAS:528:DC%2BC1cXhsIOhtbjE, 30247488
- Yun, T.
Accurate, scalable cohort variant calls using DeepVariant and GLnexus
(2021) *Bioinformatics*, 36, pp. 5582-5589.
33399819, 8023681
- Iwaki, H.
Accelerating medicines partnership: Parkinson's disease
(2021) *Genetic Resource. Mov. Disord*, 36, pp. 1795-1804.
1:CAS:528:DC%2BB3MXhs1CmsLbO, 33960523

- McLaren, W.
The Ensembl Variant Effect Predictor
(2016) *Genome Biol*, 17.
27268795, 4893825
- Manichaikul, A.
Robust relationship inference in genome-wide association studies
(2010) *Bioinformatics*, 26, pp. 2867-2873.
1:CAS:528:DC%2BC3cXhsVSlt7bK, 20926424, 3025716
- Vitale, D.
(2024) *Genotools: An Open-Source Python Package for Efficient Genotype Data Quality Control and Analysis*,
bioRxiv
- Leonard, H.
(2024) *Global Parkinson's Genetics Program Data Release 7*,
Zenodo
- Bandres-Ciga, S.
NeuroBooster Array: a genome-wide genotyping platform to study neurological disorders across diverse populations
(2023) *medRxiv*,
38076854, 10659467
- Browning, B.L., Tian, X., Zhou, Y., Browning, S.R.
Fast two-stage phasing of large-scale sequence data
(2021) *Am. J. Hum. Genet*, 108, pp. 1880-1890.
1:CAS:528:DC%2BB3MXhvFGlsr%2FL, 34478634, 8551421
- Zhou, Y., Browning, S.R., Browning, B.L.
A fast and simple method for detecting identity-by-descent segments in large-scale data
(2020) *Am. J. Hum. Genet*, 106, pp. 426-437.
1:CAS:528:DC%2BB3cXltV2qsr0%3D, 32169169, 7118582

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