Stefano Goldwurm

List of Publications by Year
in descending order

105 papers	8,767 citations	66315 42 h-index	43868 91 g-index
105 all docs	105 docs citations	105 times ranked	10217 citing authors

#	Article	IF	Citations
1	Phenotype, genotype, and worldwide genetic penetrance of LRRK2-associated Parkinson's disease: a case-control study. Lancet Neurology, The, 2008, 7, 583-590.	4.9	1,340
2	Increased hepatic iron concentration in nonalcoholic steatohepatitis is associated with increased fibrosis. Gastroenterology, 1998, 114, 311-318.	0.6	636
3	Identification of common variants influencing risk of the tauopathy progressive supranuclear palsy. Nature Genetics, 2011, 43, 699-705.	9.4	502
4	A frequent LRRK2 gene mutation associated with autosomal dominant Parkinson's disease. Lancet, The, 2005, 365, 412-415.	6.3	449
5	Early-onset parkinsonism associated with PINK1 mutations: Frequency, genotypes, and phenotypes. Neurology, 2005, 65, 87-95.	1.5	323
6	ATP13A2 missense mutations in juvenile parkinsonism and young onset Parkinson disease. Neurology, 2007, 68, 1557-1562.	1.5	312
7	Survival and dementia in <scp>GBA</scp>â€associated Parkinson's disease: <scp>T</scp>he mutation matters. Annals of Neurology, 2016, 80, 662-673.	2.8	312
8	SNCA variants are associated with increased risk for multiple system atrophy. Annals of Neurology, 2009, 65, 610-614.	2.8	257
9	Mitochondrial DNA haplogroup K is associated with a lower risk of Parkinson's disease in Italians. European Journal of Human Genetics, 2005, 13, 748-752.	1.4	197
10	The G6055A (G2019S) mutation in LRRK2 is frequent in both early and late onset Parkinson's disease and originates from a common ancestor. Journal of Medical Genetics, 2005, 42, e65-e65.	1.5	178
11	Angiogenin variants in Parkinson disease and amyotrophic lateral sclerosis. Annals of Neurology, 2011, 70, 964-973.	2.8	168
12	Mutations in the GIGYF2 (TNRC15) Gene at the PARK11 Locus in Familial Parkinson Disease. American Journal of Human Genetics, 2008, 82, 822-833.	2.6	164
13	Comprehensive analysis of the LRRK2 gene in sixty families with Parkinson's disease. European Journal of Human Genetics, 2006, 14, 322-331.	1.4	152
14	Influence of Heterozygosity for Parkin Mutation on Onset Age in Familial Parkinson Disease. Archives of Neurology, 2006, 63, 826.	4.9	147
15	Evaluation of LRRK2 G2019S penetrance: Relevance for genetic counseling in Parkinson disease. Neurology, 2007, 68, 1141-1143.	1.5	141
16	A genome-wide association study in multiple system atrophy. Neurology, 2016, 87, 1591-1598.	1.5	139
17	Lrrk2 R1441C parkinsonism is clinically similar to sporadic Parkinson disease. Neurology, 2008, 70, 1456-1460.	1.5	132
18	Penetrance estimate of LRRK2 p.G2019S mutation in individuals of nonâ€Ashkenazi Jewish ancestry. Movement Disorders, 2017, 32, 1432-1438.	2.2	126

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19	LRRK2 G2019S mutation and Parkinson's disease: A clinical, neuropsychological and neuropsychiatric study in a large Italian sample. Parkinsonism and Related Disorders, 2006, 12, 410-419.	1.1	106
20	Genomewide association study for onset age in Parkinson disease. BMC Medical Genetics, 2009, 10, 98.	2.1	104
21	Alpha-Synuclein Expression in the Oligodendrocyte Lineage: an InÂVitro and InÂVivo Study Using Rodent and Human Models. Stem Cell Reports, 2015, 5, 174-184.	2.3	104
22	Haplotypes and gene expression implicate the MAPT region for Parkinson disease. Neurology, 2008, 71, 28-34.	1.5	103
23	The Gly2019Ser mutation in LRRK2is not fully penetrant in familial Parkinson's disease: the GenePD study. BMC Medicine, 2008, 6, 32.	2.3	102
24	Swallowing disturbances in Parkinson's disease: A multivariate analysis of contributing factors. Parkinsonism and Related Disorders, 2014, 20, 1382-1387.	1.1	93
25	PINK1heterozygous rare variants: prevalence, significance and phenotypic spectrum. Human Mutation, 2008, 29, 565-565.	1.1	74
26	Creation of a library of induced pluripotent stem cells from Parkinsonian patients. Npj Parkinson's Disease, 2016, 2, 16009.	2.5	74
27	BDNF genetic variants are associated with onset age of familial Parkinson disease: GenePD Study. Neurology, 2005, 65, 1823-1825.	1.5	67
28	Whole gene deletion and splicing mutations expand thePINK1 genotypic spectrum. Human Mutation, 2007, 28, 98-98.	1.1	66
29	Glucocerebrosidase mutations in primary parkinsonism. Parkinsonism and Related Disorders, 2014, 20, 1215-1220.	1.1	63
30	The EuroBioBank Network: 10 years of hands-on experience of collaborative, transnational biobanking for rare diseases. European Journal of Human Genetics, 2015, 23, 1116-1123.	1.4	63
31	The GBAP1 pseudogene acts as a ceRNA for the glucocerebrosidase gene GBA by sponging miR-22-3p. Scientific Reports, 2017, 7, 12702.	1.6	62
32	GIGYF2 gene disruption in mice results in neurodegeneration and altered insulin-like growth factor signaling. Human Molecular Genetics, 2009, 18, 4629-4639.	1.4	61
33	Novel parkin mutations detected in patients with early-onset Parkinson's disease. Movement Disorders, 2005, 20, 424-431.	2.2	60
34	Cyclin-G-associated kinase modifies Â-synuclein expression levels and toxicity in Parkinson's disease: results from the GenePD Study. Human Molecular Genetics, 2011, 20, 1478-1487.	1.4	60
35	DNAJC12 and dopaâ€responsive nonprogressive parkinsonism. Annals of Neurology, 2017, 82, 640-646.	2.8	60
36	Nonsteroidal <scp>Antiâ€inflammatory</scp> Use and <scp>LRRK2</scp> Parkinson's Disease Penetrance. Movement Disorders, 2020, 35, 1755-1764.	2.2	57

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37	Parkinson's disease beyond 20â€...years. Journal of Neurology, Neurosurgery and Psychiatry, 2015, 86, 849-855.	0.9	55
38	Dementia in Parkinson's disease: Is male gender a risk factor?. Parkinsonism and Related Disorders, 2016, 26, 67-72.	1.1	52
39	SNCA and MAPT genes: Independent and joint effects in Parkinson disease in the Italian population. Parkinsonism and Related Disorders, 2012, 18, 257-262.	1.1	51
40	Penetrance of Glucocerebrosidase (<scp>GBA</scp>) Mutations in Parkinson's Disease: A Kin Cohort Study. Movement Disorders, 2020, 35, 2111-2114.	2.2	50
41	Singleâ€cell expression profiling of dopaminergic neurons combined with association analysis identifies pyridoxal kinase as Parkinson's disease gene. Annals of Neurology, 2009, 66, 792-798.	2.8	49
42	Kinâ€cohort analysis of LRRK2â€G2019S penetrance in Parkinson's disease. Movement Disorders, 2011, 26, 2144-2145.	2.2	49
43	Microtubule Destabilization Is Shared by Genetic and Idiopathic Parkinsonâ€™s Disease Patient Fibroblasts. PLoS ONE, 2012, 7, e37467.	1.1	43
44	Analysis of vesicular monoamine transporter 2 polymorphisms in Parkinsonâ€™s disease. Neurobiology of Aging, 2013, 34, 1712.e9-1712.e13.	1.5	43
45	Parkin absence accelerates microtubule aging in dopaminergic neurons. Neurobiology of Aging, 2018, 61, 66-74.	1.5	43
46	Parkin analysis in early onset Parkinson's disease. Parkinsonism and Related Disorders, 2008, 14, 326-333.	1.1	42
47	Striatal dopamine transporter binding in Parkinson's disease associated with theLRRK2 Gly2019Ser mutation. Movement Disorders, 2006, 21, 1144-1147.	2.2	41
48	Psychiatric symptoms in Parkinsonâ€™s disease assessed with the SCL-90R self-reported questionnaire. Neurological Sciences, 2010, 31, 35-40.	0.9	40
49	LRRK2 mutations in Parkinson's disease: Confirmation of a gender effect in the Italian population. Parkinsonism and Related Disorders, 2014, 20, 911-914.	1.1	40
50	The alliance between genetic biobanks and patient organisations: the experience of the telethon network of genetic biobanks. Orphanet Journal of Rare Diseases, 2016, 11, 142.	1.2	40
51	Telethon Network of Genetic Biobanks: a key service for diagnosis and research on rare diseases. Orphanet Journal of Rare Diseases, 2013, 8, 129.	1.2	39
52	Haplotype analysis in Australian hemochromatosis patients: evidence for a predominant ancestral haplotype exclusively associated with hemochromatosis. American Journal of Human Genetics, 1995, 56, 428-33.	2.6	39
53	Herbicide exposure modifies GSTP1 haplotype association to Parkinson onset age: The GenePD Study. Neurology, 2006, 67, 2206-2210.	1.5	38
54	Dopamine transporter imaging study in parkinsonism occurring in fragile X premutation carriers. Neurology, 2005, 65, 1971-1973.	1.5	36

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55	Î±-Synuclein multiplication analysis in Italian familial Parkinson disease. Parkinsonism and Related Disorders, 2010, 16, 228-231.	1.1	36
56	Molecular analysis of 30 mucopolysaccharidosis type I patients: evaluation of the mutational spectrum in Italian population and identification of 13 novel mutations. Human Mutation, 2002, 20, 231-231.	1.1	35
57	Biological effects of the PINK1 c.1366C>T mutation: implications in Parkinson disease pathogenesis. Neurogenetics, 2007, 8, 103-109.	0.7	35
58	TNF-Î± and Î±-synuclein fibrils differently regulate human astrocyte immune reactivity and impair mitochondrial respiration. Cell Reports, 2021, 34, 108895.	2.9	35
59	Replication of association between ELAVL4 and Parkinson disease: the GenePD study. Human Genetics, 2008, 124, 95-99.	1.8	34
60	Screen for Excess FMR1 Premutation Alleles Among Males With Parkinsonism. Archives of Neurology, 2007, 64, 1002.	4.9	33
61	The LRRK2 Variant E193K Prevents Mitochondrial Fission Upon MPP+ Treatment by Altering LRRK2 Binding to DRP1. Frontiers in Molecular Neuroscience, 2018, 11, 64.	1.4	32
62	Molybdenum(VI) salts convert the xanthine oxidoreductase apoprotein into the active enzyme in mouse L929 fibroblastic cells*. Biochemical Journal, 1994, 298, 69-77.	1.7	30
63	Insufficient evidence for pathogenicity of SNCA His50Gln (H50Q) in Parkinson's disease. Neurobiology of Aging, 2018, 64, 159.e5-159.e8.	1.5	30
64	Genomewide Association Studies of <scp>LRRK2</scp> Modifiers of Parkinson's Disease. Annals of Neurology, 2021, 90, 76-88.	2.8	30
65	Genetic, clinical, and imaging characterization of one patient with late-onset, slowly progressive, pantothenate kinase-associated neurodegeneration. Movement Disorders, 2006, 21, 417-418.	2.2	28
66	No association of GBA mutations and multiple system atrophy. European Journal of Neurology, 2013, 20, e61-2.	1.7	28
67	C9orf72 repeat expansions are restricted to the ALS-FTD spectrum. Neurobiology of Aging, 2014, 35, 936.e13-936.e17.	1.5	28
68	Biochemical and genetic defects underlying human congenital hypotransferrinemia. The Hematology Journal, 2000, 1, 390-398.	2.0	28
69	Screening for the Presence of FMR1 Premutation Alleles in Women With Parkinsonism. Archives of Neurology, 2009, 66, 244-9.	4.9	27
70	Tryptophan hydroxylase type 2 variants modulate severity and outcome of addictive behaviors in Parkinson's disease. Parkinsonism and Related Disorders, 2016, 29, 96-103.	1.1	26
71	Large-scale assessment of polyglutamine repeat expansions in Parkinson disease. Neurology, 2015, 85, 1283-1292.	1.5	25
72	A voxel-based PET study of dopamine transporters in Parkinson's disease: Relevance of age at onset. Neurobiology of Disease, 2008, 31, 102-109.	2.1	24

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73	HFE gene mutations in a population of Italian Parkinson's disease patients. Parkinsonism and Related Disorders, 2008, 14, 426-430.	1.1	24
74	Screening LRRK2 gene mutations in patients with Parkinsonâ€™s disease in Ghana. Journal of Neurology, 2012, 259, 569-570.	1.8	24
75	Analysis of ferritin genes in Parkinson disease. Clinical Chemistry and Laboratory Medicine, 2007, 45, 1450-6.	1.4	23
76	Atypical tauopathy in a patient with LRRK2â€G2019S mutation and tremorâ€dominant Parkinsonism. Neuropathology and Applied Neurobiology, 2012, 38, 382-386.	1.8	23
77	Lewy body pathology and typical Parkinson disease in a patient with a heterozygous (R275W) mutation in the Parkin gene (PARK2). Acta Neuropathologica, 2012, 123, 901-903.	3.9	22
78	Novel DYT11 gene mutation in patients without dopaminergic deficit (SWEDD) screened for dystonia. Neurology, 2014, 83, 1155-1162.	1.5	22
79	Association of nicotine dependence susceptibility gene, CHRNA5, with Parkinson's disease age at onset: Gene and smoking status interaction. Parkinsonism and Related Disorders, 2013, 19, 72-76.	1.1	21
80	The Length of SNCA Rep1 Microsatellite May Influence Cognitive Evolution in Parkinsonâ€™s Disease. Frontiers in Neurology, 2018, 9, 213.	1.1	21
81	An exome study of Parkinsonâ€™s disease in Sardinia, a Mediterranean genetic isolate. Neurogenetics, 2015, 16, 55-64.	0.7	20
82	Mutational screening and zebrafish functional analysis of GIGYF2 as a Parkinson-disease gene. Neurobiology of Aging, 2011, 32, 1994-2005.	1.5	16
83	The Asp620asn mutation in VPS35 is not a common cause of familial Parkinson's disease. Movement Disorders, 2012, 27, 800-801.	2.2	15
84	Alphaâ€synuclein repeat variants and survival in Parkinson's disease. Movement Disorders, 2014, 29, 1053-1057.	2.2	14
85	VEGF Haplotypes are Associated with Increased Risk to Progressive Supranuclear Palsy and Corticobasal Syndrome. Journal of Alzheimer's Disease, 2010, 21, 87-94.	1.2	12
86	Do Tardive Dyskinesia and l-Dopa Induced Dyskinesia Share Common Genetic Risk Factors? An Exploratory Study. Journal of Molecular Neuroscience, 2013, 51, 380-388.	1.1	12
87	DJ1 analysis in a large cohort of Italian early onset Parkinson Disease patients. Neuroscience Letters, 2013, 557, 165-170.	1.0	11
88	LRRK2â€G2019S mutation is not associated with an increased cancer risk: A kinâ€cohort study. Movement Disorders, 2014, 29, 1325-1326.	2.2	11
89	Identification of a Novel Krueppel-Related Zinc Finger Gene (ZNF184) Mapping to 6p21.3. Genomics, 1997, 40, 486-489.	1.3	10
90	Familial aggregation in Progressive Supranuclear Palsy and Corticobasal Syndrome. European Journal of Neurology, 2011, 18, 195-197.	1.7	10

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91	Multiple system atrophy and CAG repeat length: A genetic screening of polyglutamine disease genes in Italian patients. Neuroscience Letters, 2018, 678, 37-42.	1.0	10
92	Haemochromatosis after the discovery of HFE (â€œHLA-Hâ€). Gut, 1997, 41, 855-856.	6.1	9
93	Sequence Variations in Mitochondrial Ferritin: Distribution in Healthy Controls and Different Types of Patients. Genetic Testing and Molecular Biomarkers, 2010, 14, 793-796.	0.3	9
94	Huntington CAG repeat size does not modify onset age in familial Parkinson's disease: The GenePD study. Movement Disorders, 2008, 23, 1596-1601.	2.2	8
95	Beyond 35Âyears of Parkinsonâ€™s disease: a comprehensive clinical and instrumental assessment. Journal of Neurology, 2018, 265, 1989-1997.	1.8	8
96	The spinal muscular atrophy gene region at 5q13.1 has a paralogous chromosomal region at 6p21.3. Mammalian Genome, 1998, 9, 235-239.	1.0	5
97	Association analysis of PARP1 polymorphisms with Parkinsonâ€™s disease. Parkinsonism and Related Disorders, 2011, 17, 701-704.	1.1	5
98	Analysis of Nucleotide Variations in Genes of Iron Management in Patients of Parkinson's Disease and Other Movement Disorders. Parkinson's Disease, 2011, 2011, 1-6.	0.6	4
99	Generation of an induced pluripotent stem cell line (CSC-46) from a patient with Parkinson's disease carrying a novel p.R301C mutation in the GBA gene. Stem Cell Research, 2019, 34, 101373.	0.3	4
100	Later age at onset in Parkinson's disease over twenty years in an Italian tertiary clinic. Parkinsonism and Related Disorders, 2014, 20, 1181-1185.	1.1	3
101	Comment on â€œCompound heterozygosity in DJ-1 gene non-coding portion related to Parkinsonismâ€. Parkinsonism and Related Disorders, 2010, 16, 360-361.	1.1	1
102	Proteinâ€redistribution diet in a case of tyrosine hydroxylase enzyme deficiency. Movement Disorders, 2017, 32, 794-795.	2.2	1
103	Governance of Access in Biobanking: The Case of Telethon Network of Genetic Biobanks. Biopreservation and Biobanking, 2021, 19, 483-492.	0.5	1
104	Isolation and Characterisation of Cosmids to Intervals Within a 4.5Mb Region at 6p21.3. DNA Sequence, 1997, 8, 147-150.	0.7	0
105	Generation of a transcription map distal to HLA-F. European Journal of Human Genetics, 1998, 6, 475-486.	1.4	0