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	TNF-Î± and Î±-synuclein fibrils differently regulate human astrocyte immune reactivity and impair mitochondrial respiration. Cell Reports, 2021, 34, 108895.	2.9	35
2	Genomewide Association Studies of <scp>LRRK2</scp> Modifiers of Parkinson's Disease. Annals of Neurology, 2021, 90, 76-88.	2.8	30
3	Governance of Access in Biobanking: The Case of Telethon Network of Genetic Biobanks. Biopreservation and Biobanking, 2021, 19, 483-492.	0.5	1
4	Nonsteroidal <scp>Antiâ€inflammatory</scp> Use and <scp>LRRK2</scp> Parkinson's Disease Penetrance. Movement Disorders, 2020, 35, 1755-1764.	2.2	57
5	Penetrance of Glucocerebrosidase (<scp>GBA</scp>) Mutations in Parkinson's Disease: A Kin Cohort Study. Movement Disorders, 2020, 35, 2111-2114.	2.2	50
6	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
7	Insufficient evidence for pathogenicity of SNCA His50Gln (H50Q) in Parkinson's disease. Neurobiology of Aging, 2018, 64, 159.e5-159.e8.	1.5	30
8	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
9	Parkin absence accelerates microtubule aging in dopaminergic neurons. Neurobiology of Aging, 2018, 61, 66-74.	1.5	43
10	Beyond 35Âyears of Parkinsonâ€™s disease: a comprehensive clinical and instrumental assessment. Journal of Neurology, 2018, 265, 1989-1997.	1.8	8
11	The Length of SNCA Rep1 Microsatellite May Influence Cognitive Evolution in Parkinsonâ€™s Disease. Frontiers in Neurology, 2018, 9, 213.	1.1	21
12	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
13	Proteinâ€redistribution diet in a case of tyrosine hydroxylase enzyme deficiency. Movement Disorders, 2017, 32, 794-795.	2.2	1
14	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
15	DNAJC12 and dopaâ€responsive nonprogressive parkinsonism. Annals of Neurology, 2017, 82, 640-646.	2.8	60
16	Penetrance estimate of LRRK2 p.G2019S mutation in individuals of nonâ€Ashkenazi Jewish ancestry. Movement Disorders, 2017, 32, 1432-1438.	2.2	126
17	Creation of a library of induced pluripotent stem cells from Parkinsonian patients. Npj Parkinson's Disease, 2016, 2, 16009.	2.5	74
18	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

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19	A genome-wide association study in multiple system atrophy. Neurology, 2016, 87, 1591-1598.	1.5	139
20	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
21	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
22	Dementia in Parkinson's disease: Is male gender a risk factor?. Parkinsonism and Related Disorders, 2016, 26, 67-72.	1.1	52
23	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
24	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
25	Large-scale assessment of polyglutamine repeat expansions in Parkinson disease. Neurology, 2015, 85, 1283-1292.	1.5	25
26	Parkinson's disease beyond 20â€...years. Journal of Neurology, Neurosurgery and Psychiatry, 2015, 86, 849-855.	0.9	55
27	An exome study of Parkinsonâ€™s disease in Sardinia, a Mediterranean genetic isolate. Neurogenetics, 2015, 16, 55-64.	0.7	20
28	Swallowing disturbances in Parkinson's disease: A multivariate analysis of contributing factors. Parkinsonism and Related Disorders, 2014, 20, 1382-1387.	1.1	93
29	Novel DYT11 gene mutation in patients without dopaminergic deficit (SWEDD) screened for dystonia. Neurology, 2014, 83, 1155-1162.	1.5	22
30	Alphaâ€synuclein repeat variants and survival in Parkinson's disease. Movement Disorders, 2014, 29, 1053-1057.	2.2	14
31	LRRK2â€G2019S mutation is not associated with an increased cancer risk: A kinâ€cohort study. Movement Disorders, 2014, 29, 1325-1326.	2.2	11
32	Glucocerebrosidase mutations in primary parkinsonism. Parkinsonism and Related Disorders, 2014, 20, 1215-1220.	1.1	63
33	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
34	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
35	C9orf72 repeat expansions are restricted to the ALS-FTD spectrum. Neurobiology of Aging, 2014, 35, 936.e13-936.e17.	1.5	28
36	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

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37	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
38	DJ1 analysis in a large cohort of Italian early onset Parkinson Disease patients. Neuroscience Letters, 2013, 557, 165-170.	1.0	11
39	Analysis of vesicular monoamine transporter 2 polymorphisms in Parkinsonâ€™s disease. Neurobiology of Aging, 2013, 34, 1712.e9-1712.e13.	1.5	43
40	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
41	No association of GBA mutations and multiple system atrophy. European Journal of Neurology, 2013, 20, e61-2.	1.7	28
42	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
43	Microtubule Destabilization Is Shared by Genetic and Idiopathic Parkinsonâ€™s Disease Patient Fibroblasts. PLoS ONE, 2012, 7, e37467.	1.1	43
44	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
45	Atypical tauopathy in a patient with LRRK2â€G2019S mutation and tremorâ€dominant Parkinsonism. Neuropathology and Applied Neurobiology, 2012, 38, 382-386.	1.8	23
46	The Asp620asn mutation in VPS35 is not a common cause of familial Parkinson's disease. Movement Disorders, 2012, 27, 800-801.	2.2	15
47	Screening LRRK2 gene mutations in patients with Parkinsonâ€™s disease in Ghana. Journal of Neurology, 2012, 259, 569-570.	1.8	24
48	Identification of common variants influencing risk of the tauopathy progressive supranuclear palsy. Nature Genetics, 2011, 43, 699-705.	9.4	502
49	Association analysis of PARP1 polymorphisms with Parkinsonâ€™s disease. Parkinsonism and Related Disorders, 2011, 17, 701-704.	1.1	5
50	Mutational screening and zebrafish functional analysis of GIGYF2 as a Parkinson-disease gene. Neurobiology of Aging, 2011, 32, 1994-2005.	1.5	16
51	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
52	Familial aggregation in Progressive Supranuclear Palsy and Corticobasal Syndrome. European Journal of Neurology, 2011, 18, 195-197.	1.7	10
53	Kinâ€cohort analysis of LRRK2â€G2019S penetrance in Parkinson's disease. Movement Disorders, 2011, 26, 2144-2145.	2.2	49
54	Angiogenin variants in Parkinson disease and amyotrophic lateral sclerosis. Annals of Neurology, 2011, 70, 964-973.	2.8	168

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55	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
56	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
57	Psychiatric symptoms in Parkinsonâ€™s disease assessed with the SCL-90R self-reported questionnaire. Neurological Sciences, 2010, 31, 35-40.	0.9	40
58	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
59	Î±-Synuclein multiplication analysis in Italian familial Parkinson disease. Parkinsonism and Related Disorders, 2010, 16, 228-231.	1.1	36
60	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
61	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
62	SNCA variants are associated with increased risk for multiple system atrophy. Annals of Neurology, 2009, 65, 610-614.	2.8	257
63	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
64	Genomewide association study for onset age in Parkinson disease. BMC Medical Genetics, 2009, 10, 98.	2.1	104
65	Screening for the Presence of FMR1 Premutation Alleles in Women With Parkinsonism. Archives of Neurology, 2009, 66, 244-9.	4.9	27
66	Replication of association between ELAVL4 and Parkinson disease: the GenePD study. Human Genetics, 2008, 124, 95-99.	1.8	34
67	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
68	PINK1heterozygous rare variants: prevalence, significance and phenotypic spectrum. Human Mutation, 2008, 29, 565-565.	1.1	74
69	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
70	Lrrk2 R1441C parkinsonism is clinically similar to sporadic Parkinson disease. Neurology, 2008, 70, 1456-1460.	1.5	132
71	The Gly2019Ser mutation in LRRK2is not fully penetrant in familial Parkinson's disease: the GenePD study. BMC Medicine, 2008, 6, 32.	2.3	102
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	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
74	Parkin analysis in early onset Parkinson's disease. Parkinsonism and Related Disorders, 2008, 14, 326-333.	1.1	42
75	HFE gene mutations in a population of Italian Parkinson's disease patients. Parkinsonism and Related Disorders, 2008, 14, 426-430.	1.1	24
76	Haplotypes and gene expression implicate the MAPT region for Parkinson disease. Neurology, 2008, 71, 28-34.	1.5	103
77	ATP13A2 missense mutations in juvenile parkinsonism and young onset Parkinson disease. Neurology, 2007, 68, 1557-1562.	1.5	312
78	Evaluation of LRRK2 G2019S penetrance: Relevance for genetic counseling in Parkinson disease. Neurology, 2007, 68, 1141-1143.	1.5	141
79	Analysis of ferritin genes in Parkinson disease. Clinical Chemistry and Laboratory Medicine, 2007, 45, 1450-6.	1.4	23
80	Screen for Excess FMR1 Premutation Alleles Among Males With Parkinsonism. Archives of Neurology, 2007, 64, 1002.	4.9	33
81	Whole gene deletion and splicing mutations expand thePINK1 genotypic spectrum. Human Mutation, 2007, 28, 98-98.	1.1	66
82	Biological effects of the PINK1 c.1366C>T mutation: implications in Parkinson disease pathogenesis. Neurogenetics, 2007, 8, 103-109.	0.7	35
83	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
84	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
85	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
86	Striatal dopamine transporter binding in Parkinson's disease associated with theLRRK2 Gly2019Ser mutation. Movement Disorders, 2006, 21, 1144-1147.	2.2	41
87	Influence of Heterozygosity for Parkin Mutation on Onset Age in Familial Parkinson Disease. Archives of Neurology, 2006, 63, 826.	4.9	147
88	Herbicide exposure modifies GSTP1 haplotype association to Parkinson onset age: The GenePD Study. Neurology, 2006, 67, 2206-2210.	1.5	38
89	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
90	Novel parkin mutations detected in patients with early-onset Parkinson's disease. Movement Disorders, 2005, 20, 424-431.	2.2	60

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91	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
92	Dopamine transporter imaging study in parkinsonism occurring in fragile X premutation carriers. Neurology, 2005, 65, 1971-1973.	1.5	36
93	Early-onset parkinsonism associated with PINK1 mutations: Frequency, genotypes, and phenotypes. Neurology, 2005, 65, 87-95.	1.5	323
94	A frequent LRRK2 gene mutation associated with autosomal dominant Parkinson's disease. Lancet, The, 2005, 365, 412-415.	6.3	449
95	BDNF genetic variants are associated with onset age of familial Parkinson disease: GenePD Study. Neurology, 2005, 65, 1823-1825.	1.5	67
96	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
97	Biochemical and genetic defects underlying human congenital hypotransferrinemia. The Hematology Journal, 2000, 1, 390-398.	2.0	28
98	Generation of a transcription map distal to HLA-F. European Journal of Human Genetics, 1998, 6, 475-486.	1.4	0
99	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
100	Increased hepatic iron concentration in nonalcoholic steatohepatitis is associated with increased fibrosis. Gastroenterology, 1998, 114, 311-318.	0.6	636
101	Haemochromatosis after the discovery of HFE (â€œHLA-Hâ€). Gut, 1997, 41, 855-856.	6.1	9
102	Isolation and Characterisation of Cosmids to Intervals Within a 4.5Mb Region at 6p21.3. DNA Sequence, 1997, 8, 147-150.	0.7	0
103	Identification of a Novel Krueppel-Related Zinc Finger Gene (ZNF184) Mapping to 6p21.3. Genomics, 1997, 40, 486-489.	1.3	10
104	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
105	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