Birgitt SchÃ¼ele

List of Publications by Year
in descending order

75 papers	6,273 citations	81900 39 h-index	79698 73 g-index
86 all docs	86 docs citations	86 times ranked	8673 citing authors

#	Article	IF	Citations
1	LRRK2 Mutant iPSC-Derived DA Neurons Demonstrate Increased Susceptibility to Oxidative Stress. Cell Stem Cell, 2011, 8, 267-280.	11.1	668
2	Identification and Rescue of Î±-Synuclein Toxicity in Parkinson Patientâ€“Derived Neurons. Science, 2013, 342, 983-987.	12.6	416
3	Functional Impairment in Miro Degradation and Mitophagy Is a Shared Feature in Familial and Sporadic Parkinsonâ€™s Disease. Cell Stem Cell, 2016, 19, 709-724.	11.1	371
4	Phenotypic variation in a large Swedish pedigree due toSNCAduplication and triplication. Neurology, 2007, 68, 916-922.	1.1	370
5	Î²2-Adrenoreceptor is a regulator of the Î±-synuclein gene driving risk of Parkinsonâ€™s disease. Science, 2017, 357, 891-898.	12.6	341
6	Clinical Correlations With Lewy Body Pathology inLRRK2-Related Parkinson Disease. JAMA Neurology, 2015, 72, 100.	9.0	272
7	SNCA Triplication Parkinson's Patient's iPSC-derived DA Neurons Accumulate Î±-Synuclein and Are Susceptible to Oxidative Stress. PLoS ONE, 2011, 6, e26159.	2.5	257
8	LRRK2 mutations cause mitochondrial DNA damage in iPSC-derived neural cells from Parkinson's disease patients: Reversal by gene correction. Neurobiology of Disease, 2014, 62, 381-386.	4.4	235
9	Alterations in the common fragile site gene Parkin in ovarian and other cancers. Oncogene, 2003, 22, 8370-8378.	5.9	191
10	Evidence That Paternal Expression of the Îµ-Sarcoglycan Gene Accounts for Reduced Penetrance in Myoclonus-Dystonia. American Journal of Human Genetics, 2002, 71, 1303-1311.	6.2	178
11	A pathway for Parkinsonâ€™s Disease LRRK2 kinase to block primary cilia and Sonic hedgehog signaling in the brain. ELife, 2018, 7, .	6.0	170
12	Alpha-synuclein-glucocerebrosidase interactions in pharmacological Gaucher models: A biological link between Gaucher disease and parkinsonism. NeuroToxicology, 2009, 30, 1127-1132.	3.0	164
13	LRRK2 modifies Î±-syn pathology and spread in mouse models and human neurons. Acta Neuropathologica, 2019, 137, 961-980.	7.7	142
14	PINK1, Parkin, and DJ-1 mutations in Italian patients with early-onset parkinsonism. European Journal of Human Genetics, 2005, 13, 1086-1093.	2.8	132
15	Penetrance estimate of LRRK2 p.G2019S mutation in individuals of nonâ€Ashkenazi Jewish ancestry. Movement Disorders, 2017, 32, 1432-1438.	3.9	126
16	Elevated Î±-synuclein caused by SNCA gene triplication impairs neuronal differentiation and maturation in Parkinson's patient-derived induced pluripotent stem cells. Cell Death and Disease, 2015, 6, e1994-e1994.	6.3	125
17	Inactivating Mutations in ESCO2 Cause SC Phocomelia and Roberts Syndrome: No Phenotype-Genotype Correlation. American Journal of Human Genetics, 2005, 77, 1117-1128.	6.2	122
18	Skin Punch Biopsy Explant Culture for Derivation of Primary Human Fibroblasts. Journal of Visualized Experiments, 2013, , e3779.	0.3	107

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19	Parkin gene alterations in hepatocellular carcinoma. Genes Chromosomes and Cancer, 2004, 40, 85-96.	2.8	105
20	Alpha-Synuclein Physiology and Pathology: A Perspective on Cellular Structures and Organelles. Frontiers in Neuroscience, 2019, 13, 1399.	2.8	104
21	Elevated Alpha-Synuclein Impairs Innate Immune Cell Function and Provides a Potential Peripheral Biomarker for Parkinson's Disease. PLoS ONE, 2013, 8, e71634.	2.5	97
22	DICE, an efficient system for iterative genomic editing in human pluripotent stem cells. Nucleic Acids Research, 2014, 42, e34-e34.	14.5	94
23	LRRK2â€mediated Rab10 phosphorylation in immune cells from Parkinson's disease patients. Movement Disorders, 2019, 34, 406-415.	3.9	83
24	LRRK2 G2019S-induced mitochondrial DNA damage is LRRK2 kinase dependent and inhibition restores mtDNA integrity in Parkinsonâ€™s disease. Human Molecular Genetics, 2017, 26, 4340-4351.	2.9	76
25	Severe congenital encephalopathy caused by MECP2 null mutations in males: central hypoxia and reduced neuronal dendritic structure. Clinical Genetics, 2008, 74, 116-126.	2.0	75
26	Systems-Based Analyses of Brain Regions Functionally Impacted in Parkinson's Disease Reveals Underlying Causal Mechanisms. PLoS ONE, 2014, 9, e102909.	2.5	74
27	Higher Vulnerability and Stress Sensitivity of Neuronal Precursor Cells Carrying an Alpha-Synuclein Gene Triplication. PLoS ONE, 2014, 9, e112413.	2.5	73
28	Phenotypic spectrum and sex effects in eleven myoclonusâ€dystonia families with Îµâ€sarcoglycan mutations. Movement Disorders, 2008, 23, 588-592.	3.9	66
29	Can cellular models revolutionize drug discovery in Parkinson's disease?. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2009, 1792, 1043-1051.	3.8	60
30	Inflammatory profile in LRRK2-associated prodromal and clinical PD. Journal of Neuroinflammation, 2016, 13, 122.	7.2	57
31	Nonsteroidal <scp>Antiâ€inflammatory</scp> Use and <scp>LRRK2</scp> Parkinson's Disease Penetrance. Movement Disorders, 2020, 35, 1755-1764.	3.9	57
32	Small Molecules Greatly Improve Conversion of Human-Induced Pluripotent Stem Cells to the Neuronal Lineage. Stem Cells International, 2012, 2012, 1-12.	2.5	56
33	Inflammatory profile discriminates clinical subtypes in LRRK2â€associated Parkinson's disease. European Journal of Neurology, 2017, 24, 427.	3.3	56
34	Genetic heterogeneity in ten families with myoclonus-dystonia. Journal of Neurology, Neurosurgery and Psychiatry, 2004, 75, 1181-1185.	1.9	55
35	Molecular breakpoint cloning and gene expression studies of a novel translocation t(4;15)(q27;q11.2) associated with Prader-Willi syndrome. BMC Medical Genetics, 2005, 6, 18.	2.1	55
36	Myoclonusâ€“dystonia: Detection of novel, recurrent, and de novo SGCE mutations. Neurology, 2004, 62, 1229-1231.	1.1	54

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37	Parkinsonâ€™s disease associated with pure ATXN10 repeat expansion. Npj Parkinson's Disease, 2017, 3, 27.	5.3	54
38	Multisystem Lewy body disease and the other parkinsonian disorders. Nature Genetics, 2015, 47, 1378-1384.	21.4	49
39	DLX5 and DLX6 Expression Is Biallelic and Not Modulated by MeCP2 Deficiency. American Journal of Human Genetics, 2007, 81, 492-506.	6.2	48
40	The Role of Alpha-Synuclein and Other Parkinsonâ€™s Genes in Neurodevelopmental and Neurodegenerative Disorders. International Journal of Molecular Sciences, 2020, 21, 5724.	4.1	37
41	Clinical and genetic features of myoclonus-dystonia in 3 cases: A video presentation. Movement Disorders, 2004, 19, 231-234.	3.9	30
42	Genomewide Association Studies of <scp>LRRK2</scp> Modifiers of Parkinson's Disease. Annals of Neurology, 2021, 90, 76-88.	5.3	30
43	Genetic fine-mapping of the Iowan SNCA gene triplication in a patient with Parkinsonâ€™s disease. Npj Parkinson's Disease, 2018, 4, 18.	5.3	28
44	Mitochondrial Dysfunction in Skin Fibroblasts from a Parkinson's Disease Patient with an alpha-Synuclein Triplication. Journal of Parkinson's Disease, 2011, 1, 175-183.	2.8	24
45	Noradrenergic modulation of calcium currents and synaptic transmission in the olfactory bulb ofXenopus laevistadpoles. European Journal of Neuroscience, 2001, 13, 1093-1100.	2.6	22
46	Advancing Stem Cell Models of Alpha-Synuclein Gene Regulation in Neurodegenerative Disease. Frontiers in Neuroscience, 2018, 12, 199.	2.8	19
47	Heart rate variability in leucineâ€rich repeat kinase 2â€associated Parkinson's disease. Movement Disorders, 2017, 32, 610-614.	3.9	18
48	LRRK2 p.Ile1371Val Mutation in a Case with Neuropathologically Confirmed Multi-System Atrophy. Journal of Parkinson's Disease, 2018, 8, 93-100.	2.8	18
49	Effect of LRRK2 protein and activity on stimulated cytokines in human monocytes and macrophages. Npj Parkinson's Disease, 2022, 8, 34.	5.3	18
50	Role ofSCA2mutations in early- and late-onset dopa-responsive parkinsonism. Annals of Neurology, 2002, 52, 257-258.	5.3	16
51	Intrafamilial phenotypic and genetic heterogeneity of dystonia. Journal of the Neurological Sciences, 2006, 250, 92-96.	0.6	16
52	The commercial genetic testing landscape for Parkinson's disease. Parkinsonism and Related Disorders, 2021, 92, 107-111.	2.2	16
53	Genetic and Environmental Factors Influence the Pleomorphy of LRRK2 Parkinsonism. International Journal of Molecular Sciences, 2021, 22, 1045.	4.1	15
54	Novel features in a patient homozygous for the L347P mutation in the PINK1 gene. Parkinsonism and Related Disorders, 2007, 13, 359-361.	2.2	12

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55	Clustering of motor and nonmotor traits in leucineâ€rich repeat kinase 2 G2019S Parkinson's disease nonparkinsonian relatives: A multicenter family study. Movement Disorders, 2018, 33, 960-965.	3.9	12
56	Exosome/microvesicle content is altered in leucineâ€rich repeat kinase 2 mutant induced pluripotent stem cellâ€derived neural cells. Journal of Comparative Neurology, 2020, 528, 1203-1215.	1.6	11
57	Increased markers of cardiac vagal activity in leucine-rich repeat kinase 2-associated Parkinsonâ€™s disease. Clinical Autonomic Research, 2019, 29, 603-614.	2.5	10
58	Pulse-Field capillary electrophoresis of repeat-primed PCR amplicons for analysis of large repeats in Spinocerebellar Ataxia Type 10. PLoS ONE, 2020, 15, e0228789.	2.5	10
59	Slice Culture of the Olfactory Bulb of Xenopus laevis Tadpoles. Chemical Senses, 2001, 26, 399-407.	2.0	8
60	Michael J. Fox Foundation LRRK2 Consortium: geographical differences in returning genetic research data to study participants. Genetics in Medicine, 2014, 16, 644-645.	2.4	7
61	Discovery of functional non-coding conserved regions in the Î±-synuclein gene locus. F1000Research, 2014, 3, 259.	1.6	7
62	Parkingene variations and parkinsonism: Association does not imply causation. Annals of Neurology, 2007, 61, 4-6.	5.3	6
63	Call for participation in the neurogenetics consortium within the Human Variome Project. Neurogenetics, 2011, 12, 169-173.	1.4	5
64	Loss-of-Function NUBPL Mutation May Link Parkinson's Disease to Recessive Complex I Deficiency. Frontiers in Neurology, 2020, 11, 555961.	2.4	5
65	Is PARKIN parkinsonism a cancer predisposition syndrome?. Neurology: Genetics, 2015, 1, e31.	1.9	3
66	Comparative genomic hybridization solves a 14â€yearâ€old <scp>PARKIN</scp> mystery. Annals of Neurology, 2015, 78, 663-664.	5.3	3
67	Isogenic human SNCA gene dosage induced pluripotent stem cells to model Parkinsonâ€™s disease. Stem Cell Research, 2022, 60, 102733.	0.7	3
68	Short-term deceleration capacity of heart rate: a sensitive marker of cardiac autonomic dysfunction in idiopathic Parkinsonâ€™s disease. Clinical Autonomic Research, 2021, 31, 729-736.	2.5	2
69	Multiplex CRISPR/Cas9-Guided No-Amp Targeted Sequencing Panel for Spinocerebellar Ataxia Repeat Expansions. Neuromethods, 2022, , 95-120.	0.3	2
70	Multiplex imaging of human induced pluripotent stem cell-derived neurons with CO-Detection by indEXing (CODEX) technology. Journal of Neuroscience Methods, 2022, 378, 109653.	2.5	2
71	Derivation of Leptomeninges Explant Cultures from Postmortem Human Brain Donors. Journal of Visualized Experiments, 2017, , .	0.3	1
72	Report of the Phenotype of a Patient with Roberts Syndrome and a Rare ESCO2 Variant. Journal of Pediatric Genetics, 2020, 09, 058-062.	0.7	1

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73	Embryoid Body Formation from Mouse and Human Pluripotent Stem Cells for Transplantation to Study Brain Microenvironment and Cellular Differentiation. Methods in Molecular Biology, 2021, , 1.	0.9	1
74	Heart rate variability biomarkers of leucine-rich repeat kinase 2-associated Parkinson's disease. , 2020, , .		0
75	Phenotypic Heterogeneity among GBA p.R202X Carriers in Lewy Body Spectrum Disorders. Biomedicines, 2022, 10, 160.	3.2	0