Katja Lohmann

List of Publications by Year
in descending order

64	3,030	218677	175258
papers	citations	26 h-index	52 g-index

66	66	66	3816
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Mutations in the gene encoding PDGF-B cause brain calcifications in humans and mice. Nature Genetics, 2013, 45, 1077-1082.	21.4	273
2	<scp>N</scp>omenclature of genetic movement disorders: <scp>R</scp>ecommendations of the international <scp>P</scp>arkinson and movement disorder society task force. Movement Disorders, 2016, 31, 436-457.	3.9	228
3	Genotypeâ€Phenotype Relations for the Parkinson's Disease Genes Parkin, PINK1, DJ1: MDSGene Systematic Review. Movement Disorders, 2018, 33, 730-741.	3.9	215
4	Mutations in THAP1 (DYT6) and generalised dystonia with prominent spasmodic dysphonia: a genetic screening study. Lancet Neurology, The, 2009, 8, 447-452.	10.2	194
5	Nonmotor and diagnostic findings in subjects with de novo Parkinson disease of the DeNoPa cohort. Neurology, 2013, 81, 1226-1234.	1.1	153
6	Mutant Parkin Impairs Mitochondrial Function and Morphology in Human Fibroblasts. PLoS ONE, 2010, 5, e12962.	2.5	140
7	Mutations in VPS13D lead to a new recessive ataxia with spasticity and mitochondrial defects. Annals of Neurology, 2018, 83, 1075-1088.	5.3	122
8	Novel Dystonia Genes: Clues on Disease Mechanisms and the Complexities of Highâ€Throughput Sequencing. Movement Disorders, 2016, 31, 471-477.	3.9	121
9	Genotypeâ€phenotype relations for the Parkinson's disease genes SNCA, LRRK2, VPS35: MDSGene systematic review. Movement Disorders, 2018, 33, 1857-1870.	3.9	120
10	Short- and long-term outcome of chronic pallidal neurostimulation in monogenic isolated dystonia. Neurology, 2015, 84, 895-903.	1.1	117
11	Update on the Genetics of Dystonia. Current Neurology and Neuroscience Reports, 2017, 17, 26.	4.2	98
12	Genotypeâ€“Phenotype Relations for Isolated Dystonia Genes: <scp>MDSGene</scp> Systematic Review. Movement Disorders, 2021, 36, 1086-1103.	3.9	74
13	Fixing the broken system of genetic locus symbols. Neurology, 2012, 78, 1016-1024.	1.1	70
14	Mutations inGNAL. JAMA Neurology, 2014, 71, 490.	9.0	70
15	A hexanucleotide repeat modifies expressivity of Xâ€linked dystonia parkinsonism. Annals of Neurology, 2019, 85, 812-822.	5.3	67
16	<scp>GBA</scp> Variants in Parkinson's Disease: Clinical, Metabolomic, and Multimodal Neuroimaging Phenotypes. Movement Disorders, 2020, 35, 2201-2210.	3.9	55
17	Launching the movement disorders society genetic mutation database (MDSGene). Movement Disorders, 2016, 31, 607-609.	3.9	54
18	Genomeâ€wide association study in musician's dystonia: A risk variant at the arylsulfatase G locus?. Movement Disorders, 2014, 29, 921-927.	3.9	53

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19	Nomenclature of Genetic Movement Disorders: Recommendations of the International Parkinson and Movement Disorder Society Task Force â€“ An Update. Movement Disorders, 2022, 37, 905-935.	3.9	49
20	Identification and functional analysis of novel THAP1 mutations. European Journal of Human Genetics, 2012, 20, 171-175.	2.8	48
21	Recessive dystonia-ataxia syndrome in a Turkish family caused by a COX20 (FAM36A) mutation. Journal of Neurology, 2014, 261, 207-212.	3.6	40
22	Clinical neuroimaging and electrophysiological assessment of three DYT6 dystonia families. Movement Disorders, 2010, 25, 2405-2412.	3.9	38
23	Rare Variants in Specific Lysosomal Genes Are Associated With Parkinson's Disease. Movement Disorders, 2020, 35, 1245-1248.	3.9	37
24	Genotypeâ€“Phenotype Relations in Primary Familial Brain Calcification: Systematic <scp>MDSGene</scp> Review. Movement Disorders, 2021, 36, 2468-2480.	3.9	35
25	Unraveling Cellular Phenotypes of NovelTorsinA/TOR1AMutations. Human Mutation, 2014, 35, 1114-1122.	2.5	34
26	Novel GNB1 missense mutation in a patient with generalized dystonia, hypotonia, and intellectual disability. Neurology: Genetics, 2016, 2, e106.	1.9	33
27	Clinical spectrum of the pentanucleotide repeat expansion in the RFC1 gene in ataxia syndromes. Neurology, 2020, 95, e2912-e2923.	1.1	32
28	<scp>EIF2AK2</scp> Missense Variants Associated with Early Onset Generalized Dystonia. Annals of Neurology, 2021, 89, 485-497.	5.3	32
29	A Novel OPA3 Mutation Revealed by Exome Sequencing. JAMA Neurology, 2013, 70, 783.	9.0	26
30	Pallidal Deep Brain Stimulation in DYT6 Dystonia: Clinical Outcome and Predictive Factors for Motor Improvement. Journal of Clinical Medicine, 2019, 8, 2163.	2.4	25
31	Role of ANO3 mutations in dystonia: A large-scale mutational screening study. Parkinsonism and Related Disorders, 2019, 62, 196-200.	2.2	25
32	Genotypeâ€“Phenotype Relations for the Atypical Parkinsonism Genes: MDSGene Systematic Review. Movement Disorders, 2021, 36, 1499-1510.	3.9	22
33	Multiâ€omic landscaping of human midbrains identifies diseaseâ€relevant molecular targets and pathways in advancedâ€stage Parkinson's disease. Clinical and Translational Medicine, 2022, 12, e692.	4.0	22
34	A nonsense mutation in CHCHD2 in a patient with Parkinson disease. Neurology, 2016, 86, 577-579.	1.1	20
35	THAP1, the gene mutated in DYT6 dystonia, autoregulates its own expression. Biochimica Et Biophysica Acta - Gene Regulatory Mechanisms, 2014, 1839, 1196-1204.	1.9	19
36	Relationship of Genotype, Phenotype, and Treatment in Dopaâ€Responsive Dystonia: <scp>MDSGene</scp> Review. Movement Disorders, 2022, 37, 237-252.	3.9	19

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37	Faithful SGCE imprinting in iPSC-derived cortical neurons: an endogenous cellular model of myoclonus-dystonia. Scientific Reports, 2017, 7, 41156.	3.3	18
38	A novel, inâ€frame KMT2B deletion in a patient with apparently isolated, generalized dystonia. Movement Disorders, 2017, 32, 1495-1497.	3.9	18
39	The commercial genetic testing landscape for Parkinson's disease. Parkinsonism and Related Disorders, 2021, 92, 107-111.	2.2	16
40	The role of mutations in COL6A3 in isolated dystonia. Journal of Neurology, 2016, 263, 730-734.	3.6	15
41	Validity and Prognostic Value of a Polygenic Risk Score for Parkinsonâ€™s Disease. Genes, 2021, 12, 1859.	2.4	15
42	Truncating <scp>VPS16</scp> Mutations Are Rare in Early Onset Dystonia. Annals of Neurology, 2021, 89, 625-626.	5.3	14
43	Linking Penetrance and Transcription in <scp>DYTâ€THAP1</scp>: Insights From a Human <scp>iPSC</scp>â€Derived Cortical Model. Movement Disorders, 2021, 36, 1381-1391.	3.9	14
44	Genetic association study of the Pâ€type ATPase ATP13A2 in lateâ€onset Parkinson's disease. Movement Disorders, 2009, 24, 429-433.	3.9	13
45	A recurrent de-novo ANO3 mutation causes early-onset generalized dystonia. Journal of the Neurological Sciences, 2019, 396, 199-201.	0.6	13
46	Discordant Monozygotic Parkinson Disease Twins: Role of Mitochondrial Integrity. Annals of Neurology, 2021, 89, 158-164.	5.3	10
47	Zonisamideâ€responsive myoclonus in SEMA6Bâ€associated progressive myoclonic epilepsy. Annals of Clinical and Translational Neurology, 2021, 8, 1524-1527.	3.7	10
48	Munchausen syndrome by genetics: Next-generation challenges for clinicians. Neurology, 2017, 88, 1000-1001.	1.1	9
49	Field synopsis and systematic meta-analyses of genetic association studies in isolated dystonia. Parkinsonism and Related Disorders, 2018, 57, 50-57.	2.2	9
50	De Novo and Dominantly Inherited <scp>SPTAN1</scp> Mutations Cause Spastic Paraplegia and Cerebellar Ataxia. Movement Disorders, 2022, 37, 1175-1186.	3.9	9
51	Mutations in VPS26A are not a frequent cause of Parkinson's disease. Neurobiology of Aging, 2014, 35, 1512.e1-1512.e2.	3.1	7
52	Reduced Expression of GABAA Receptor Alpha2 Subunit Is Associated With Disinhibition of DYT-THAP1 Dystonia Patient-Derived Striatal Medium Spiny Neurons. Frontiers in Cell and Developmental Biology, 2021, 9, 650586.	3.7	7
53	Altered homodimer formation and increased iron accumulation in VAC14-related disease: Case report and review of the literature. Parkinsonism and Related Disorders, 2020, 80, 41-46.	2.2	5
54	A Multiâ€center Genomeâ€wide Association Study of Cervical Dystonia. Movement Disorders, 2021, 36, 2795-2801.	3.9	5

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55	Discordance in monozygotic Parkinson's disease twins â€“ continuum or dichotomy?. Annals of Clinical and Translational Neurology, 2019, 6, 1102-1105.	3.7	4
56	Parkin Deficiency Appears Not to Be Associated with Cardiac Damage in Parkinson's Disease. Movement Disorders, 2021, 36, 271-273.	3.9	4
57	Generation and characterization of eight human-derived iPSC lines from affected and unaffected THAP1 mutation carriers. Stem Cell Research, 2018, 33, 60-64.	0.7	3
58	Multimodal Longitudinal Neurophysiological Investigations in Dopaâ€Responsive Dystonia. Movement Disorders, 2021, 36, 1986-1987.	3.9	3
59	Familial idiopathic basal ganglia calcification: Unraveling the first genetic cause. Movement Disorders, 2012, 27, 963-963.	3.9	1
60	Compound Heterozygous DARS2 Mutations as a Mimic of Hereditary Spastic Paraplegia. Movement Disorders Clinical Practice, 2021, 8, 972-976.	1.5	1
61	Linking Huntington's Disease and Xâ€linked Dystonia Parkinsonism on the Molecular Level. Movement Disorders, 2020, 35, 1752-1753.	3.9	0
62	Importance of Tissue Selection for Genetic Testing: Detection of a Terminal 18q Deletion after Stem Cell Transplantation. Movement Disorders Clinical Practice, 2020, 7, 453-455.	1.5	0
63	Importance of Methylation Pattern: Episignatures as a Novel Instrument in Diagnostics. Movement Disorders, 2022, 37, 38-38.	3.9	0
64	PRKRAP1 and Other Pseudogenes in Movement Disorders: The Troublemakers in Genetic Analyses Are More Than Genomic Fossils. Movement Disorders Clinical Practice, 0, , .	1.5	0