

Katja Lohmann

List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/4847432/publications.pdf>

Version: 2024-02-01

64
papers

3,030
citations

218677

26
h-index

175258

52
g-index

66
all docs

66
docs citations

66
times ranked

3816
citing authors

#	ARTICLE	IF	CITATIONS
1	Importance of Methylation Pattern: Episignatures as a Novel Instrument in Diagnostics. <i>Movement Disorders</i> , 2022, 37, 38-38.	3.9	0
2	Multi-omic landscaping of human midbrains identifies disease-relevant molecular targets and pathways in advanced-stage Parkinson's disease. <i>Clinical and Translational Medicine</i> , 2022, 12, e692.	4.0	22
3	De Novo and Dominantly Inherited <i>SPTAN1</i> Mutations Cause Spastic Paraplegia and Cerebellar Ataxia. <i>Movement Disorders</i> , 2022, 37, 1175-1186.	3.9	9
4	Relationship of Genotype, Phenotype, and Treatment in Dopa-Responsive Dystonia: MDSGene Review. <i>Movement Disorders</i> , 2022, 37, 237-252.	3.9	19
5	Nomenclature of Genetic Movement Disorders: Recommendations of the International Parkinson and Movement Disorder Society Task Force – An Update. <i>Movement Disorders</i> , 2022, 37, 905-935.	3.9	49
6	<i>EIF2AK2</i> Missense Variants Associated with Early Onset Generalized Dystonia. <i>Annals of Neurology</i> , 2021, 89, 485-497.	5.3	32
7	Truncating <i>VPS16</i> Mutations Are Rare in Early Onset Dystonia. <i>Annals of Neurology</i> , 2021, 89, 625-626.	5.3	14
8	Discordant Monozygotic Parkinson Disease Twins: Role of Mitochondrial Integrity. <i>Annals of Neurology</i> , 2021, 89, 158-164.	5.3	10
9	Parkin Deficiency Appears Not to Be Associated with Cardiac Damage in Parkinson's Disease. <i>Movement Disorders</i> , 2021, 36, 271-273.	3.9	4
10	Genotype-Phenotype Relations for Isolated Dystonia Genes: MDSGene Systematic Review. <i>Movement Disorders</i> , 2021, 36, 1086-1103.	3.9	74
11	Linking Penetrance and Transcription in <i>DYT-THAP1</i> : Insights From a Human iPSC-Derived Cortical Model. <i>Movement Disorders</i> , 2021, 36, 1381-1391.	3.9	14
12	Genotype-Phenotype Relations for the Atypical Parkinsonism Genes: MDSGene Systematic Review. <i>Movement Disorders</i> , 2021, 36, 1499-1510.	3.9	22
13	Reduced Expression of GABAA Receptor Alpha2 Subunit Is Associated With Disinhibition of <i>DYT-THAP1</i> Dystonia Patient-Derived Striatal Medium Spiny Neurons. <i>Frontiers in Cell and Developmental Biology</i> , 2021, 9, 650586.	3.7	7
14	Multimodal Longitudinal Neurophysiological Investigations in Dopa-Responsive Dystonia. <i>Movement Disorders</i> , 2021, 36, 1986-1987.	3.9	3
15	Compound Heterozygous <i>DARS2</i> Mutations as a Mimic of Hereditary Spastic Paraplegia. <i>Movement Disorders Clinical Practice</i> , 2021, 8, 972-976.	1.5	1
16	Zonisamide-responsive myoclonus in <i>SEMA6B</i> -associated progressive myoclonic epilepsy. <i>Annals of Clinical and Translational Neurology</i> , 2021, 8, 1524-1527.	3.7	10
17	A Multi-center Genome-wide Association Study of Cervical Dystonia. <i>Movement Disorders</i> , 2021, 36, 2795-2801.	3.9	5
18	Genotype-Phenotype Relations in Primary Familial Brain Calcification: Systematic MDSGene Review. <i>Movement Disorders</i> , 2021, 36, 2468-2480.	3.9	35

#	ARTICLE	IF	CITATIONS
19	The commercial genetic testing landscape for Parkinson's disease. <i>Parkinsonism and Related Disorders</i> , 2021, 92, 107-111.	2.2	16
20	Validity and Prognostic Value of a Polygenic Risk Score for Parkinson's Disease. <i>Genes</i> , 2021, 12, 1859.	2.4	15
21	Linking Huntington's Disease and X-linked Dystonia Parkinsonism on the Molecular Level. <i>Movement Disorders</i> , 2020, 35, 1752-1753.	3.9	0
22	Altered homodimer formation and increased iron accumulation in VAC14-related disease: Case report and review of the literature. <i>Parkinsonism and Related Disorders</i> , 2020, 80, 41-46.	2.2	5
23	Clinical spectrum of the pentanucleotide repeat expansion in the <i>RFC1</i> gene in ataxia syndromes. <i>Neurology</i> , 2020, 95, e2912-e2923.	1.1	32
24	<i>GBA</i> Variants in Parkinson's Disease: Clinical, Metabolomic, and Multimodal Neuroimaging Phenotypes. <i>Movement Disorders</i> , 2020, 35, 2201-2210.	3.9	55
25	Importance of Tissue Selection for Genetic Testing: Detection of a Terminal 18q Deletion after Stem Cell Transplantation. <i>Movement Disorders Clinical Practice</i> , 2020, 7, 453-455.	1.5	0
26	Rare Variants in Specific Lysosomal Genes Are Associated With Parkinson's Disease. <i>Movement Disorders</i> , 2020, 35, 1245-1248.	3.9	37
27	Discordance in monozygotic Parkinson's disease twins – continuum or dichotomy?. <i>Annals of Clinical and Translational Neurology</i> , 2019, 6, 1102-1105.	3.7	4
28	A hexanucleotide repeat modifies expressivity of X-linked dystonia parkinsonism. <i>Annals of Neurology</i> , 2019, 85, 812-822.	5.3	67
29	Pallidal Deep Brain Stimulation in DYT6 Dystonia: Clinical Outcome and Predictive Factors for Motor Improvement. <i>Journal of Clinical Medicine</i> , 2019, 8, 2163.	2.4	25
30	Role of ANO3 mutations in dystonia: A large-scale mutational screening study. <i>Parkinsonism and Related Disorders</i> , 2019, 62, 196-200.	2.2	25
31	A recurrent de-novo ANO3 mutation causes early-onset generalized dystonia. <i>Journal of the Neurological Sciences</i> , 2019, 396, 199-201.	0.6	13
32	Mutations in <i>VPS13D</i> lead to a new recessive ataxia with spasticity and mitochondrial defects. <i>Annals of Neurology</i> , 2018, 83, 1075-1088.	5.3	122
33	Genotype-Phenotype Relations for the Parkinson's Disease Genes <i>Parkin</i> , <i>PINK1</i> , <i>DJ1</i> : MDSGene Systematic Review. <i>Movement Disorders</i> , 2018, 33, 730-741.	3.9	215
34	Generation and characterization of eight human-derived iPSC lines from affected and unaffected THAP1 mutation carriers. <i>Stem Cell Research</i> , 2018, 33, 60-64.	0.7	3
35	Genotype-phenotype relations for the Parkinson's disease genes SNCA, LRRK2, VPS35: MDSGene systematic review. <i>Movement Disorders</i> , 2018, 33, 1857-1870.	3.9	120
36	Field synopsis and systematic meta-analyses of genetic association studies in isolated dystonia. <i>Parkinsonism and Related Disorders</i> , 2018, 57, 50-57.	2.2	9

#	ARTICLE	IF	CITATIONS
37	Munchausen syndrome by genetics: Next-generation challenges for clinicians. <i>Neurology</i> , 2017, 88, 1000-1001.	1.1	9
38	Update on the Genetics of Dystonia. <i>Current Neurology and Neuroscience Reports</i> , 2017, 17, 26.	4.2	98
39	Faithful SGCE imprinting in iPSC-derived cortical neurons: an endogenous cellular model of myoclonus-dystonia. <i>Scientific Reports</i> , 2017, 7, 41156.	3.3	18
40	A novel, in-frame <i>KMT2B</i> deletion in a patient with apparently isolated, generalized dystonia. <i>Movement Disorders</i> , 2017, 32, 1495-1497.	3.9	18
41	Novel Dystonia Genes: Clues on Disease Mechanisms and the Complexities of High-Throughput Sequencing. <i>Movement Disorders</i> , 2016, 31, 471-477.	3.9	121
42	Launching the movement disorders society genetic mutation database (MDSGene). <i>Movement Disorders</i> , 2016, 31, 607-609.	3.9	54
43	Nomenclature of genetic movement disorders: Recommendations of the international Parkinson and movement disorder society task force. <i>Movement Disorders</i> , 2016, 31, 436-457.	3.9	228
44	Novel <i>GNB1</i> missense mutation in a patient with generalized dystonia, hypotonia, and intellectual disability. <i>Neurology: Genetics</i> , 2016, 2, e106.	1.9	33
45	A nonsense mutation in <i>CHCHD2</i> in a patient with Parkinson disease. <i>Neurology</i> , 2016, 86, 577-579.	1.1	20
46	The role of mutations in <i>COL6A3</i> in isolated dystonia. <i>Journal of Neurology</i> , 2016, 263, 730-734.	3.6	15
47	Short- and long-term outcome of chronic pallidal neurostimulation in monogenic isolated dystonia. <i>Neurology</i> , 2015, 84, 895-903.	1.1	117
48	Genome-wide association study in musician's dystonia: A risk variant at the arylsulfatase G locus?. <i>Movement Disorders</i> , 2014, 29, 921-927.	3.9	53
49	Mutations in <i>GNAL</i> . <i>JAMA Neurology</i> , 2014, 71, 490.	9.0	70
50	Mutations in <i>VPS26A</i> are not a frequent cause of Parkinson's disease. <i>Neurobiology of Aging</i> , 2014, 35, 1512.e1-1512.e2.	3.1	7
51	Recessive dystonia-ataxia syndrome in a Turkish family caused by a <i>COX20</i> (<i>FAM36A</i>) mutation. <i>Journal of Neurology</i> , 2014, 261, 207-212.	3.6	40
52	THAP1, the gene mutated in DYT6 dystonia, autoregulates its own expression. <i>Biochimica Et Biophysica Acta - Gene Regulatory Mechanisms</i> , 2014, 1839, 1196-1204.	1.9	19
53	Unraveling Cellular Phenotypes of Novel <i>TorsinA/TOR1A</i> Mutations. <i>Human Mutation</i> , 2014, 35, 1114-1122.	2.5	34
54	Mutations in the gene encoding PDGF-B cause brain calcifications in humans and mice. <i>Nature Genetics</i> , 2013, 45, 1077-1082.	21.4	273

#	ARTICLE	IF	CITATIONS
55	A Novel OPA3 Mutation Revealed by Exome Sequencing. <i>JAMA Neurology</i> , 2013, 70, 783.	9.0	26
56	Nonmotor and diagnostic findings in subjects with de novo Parkinson disease of the DeNoPa cohort. <i>Neurology</i> , 2013, 81, 1226-1234.	1.1	153
57	Fixing the broken system of genetic locus symbols. <i>Neurology</i> , 2012, 78, 1016-1024.	1.1	70
58	Identification and functional analysis of novel THAP1 mutations. <i>European Journal of Human Genetics</i> , 2012, 20, 171-175.	2.8	48
59	Familial idiopathic basal ganglia calcification: Unraveling the first genetic cause. <i>Movement Disorders</i> , 2012, 27, 963-963.	3.9	1
60	Clinical neuroimaging and electrophysiological assessment of three <i>DYT6</i> dystonia families. <i>Movement Disorders</i> , 2010, 25, 2405-2412.	3.9	38
61	Mutant Parkin Impairs Mitochondrial Function and Morphology in Human Fibroblasts. <i>PLoS ONE</i> , 2010, 5, e12962.	2.5	140
62	Mutations in THAP1 (DYT6) and generalised dystonia with prominent spasmodic dysphonia: a genetic screening study. <i>Lancet Neurology</i> , The, 2009, 8, 447-452.	10.2	194
63	Genetic association study of the Pâ€type ATPase <i>ATP13A2</i> in lateâ€onset Parkinson's disease. <i>Movement Disorders</i> , 2009, 24, 429-433.	3.9	13
64	<i>PRKRAP1</i> and Other Pseudogenes in Movement Disorders: The Troublemakers in Genetic Analyses Are More Than Genomic Fossils. <i>Movement Disorders Clinical Practice</i> , 0, , .	1.5	0