Juliane Winkelmann

List of Publications by Year
in descending order

141 papers	21,124 citations	25034 57 h-index	10445 139 g-index
151 all docs	151 docs citations	151 times ranked	28617 citing authors

#	Article	IF	Citations
1	A Novel Variant of <scp>ATP5MC3</scp> Associated with Both Dystonia and Spastic Paraplegia. Movement Disorders, 2022, 37, 375-383.	3.9	10
2	AOPEP variants as a novel cause of recessive dystonia: Generalized dystonia and dystonia-parkinsonism. Parkinsonism and Related Disorders, 2022, 97, 52-56.	2.2	7
3	Intronic elements associated with insomnia and restless legs syndrome exhibit cell-type-specific epigenetic features contributing to MEIS1 regulation. Human Molecular Genetics, 2022, 31, 1733-1746.	2.9	3
4	Collagen VI Regulates Motor Circuit Plasticity and Motor Performance by Cannabinoid Modulation. Journal of Neuroscience, 2022, 42, 1557-1573.	3.6	1
5	Reassessment of candidate gene studies for idiopathic restless legs syndrome in a large genome-wide association study dataset of European ancestry. Sleep, 2022, 45, .	1.1	6
6	Myoclonic dystonia phenotype related to a novel calmodulin-binding transcription activator 1 sequence variant. Neurogenetics, 2021, 22, 137-141.	1.4	3
7	Investigation of dopaminergic signalling in Meis homeobox 1 (Meis1) deficient mice as an animal model of restless legs syndrome. Journal of Sleep Research, 2021, 30, e13311.	3.2	4
8	The genetics of restless legs syndrome. , 2021, , .		0
9	Identification of Restless Legs Syndrome Genes by Mutational Load Analysis. Annals of Neurology, 2020, 87, 184-193.	5.3	19
10	Candidate variants in TUB are associated with familial tremor. PLoS Genetics, 2020, 16, e1009010.	3.5	3
11	Private variants in PRKN are associated with late-onset Parkinson's disease. Parkinsonism and Related Disorders, 2020, 75, 24-26.	2.2	4
12	Role of MEIS1 in restless legs syndrome: From GWAS to functional studies in mice. Advances in Pharmacology, 2019, 84, 175-184.	2.0	21
13	Potential genetic modifiers of disease risk and age at onset in patients with frontotemporal lobar degeneration and GRN mutations: a genome-wide association study. Lancet Neurology, The, 2018, 17, 548-558.	10.2	97
14	Microstructural white matter abnormalities in patients with COL6A3 mutations (DYT27 dystonia). Parkinsonism and Related Disorders, 2018, 46, 74-78.	2.2	10
15	Sleep disturbance by pramipexole is modified by Meis1 in mice. Journal of Sleep Research, 2018, 27, e12557.	3.2	14
16	Restless Legs Syndrome and Other Movement Disorders of Sleepâ€"Treatment Update. Current Treatment Options in Neurology, 2018, 20, 55.	1.8	15
17	KMT2B Is Selectively Required for Neuronal Transdifferentiation, and Its Loss Exposes Dystonia Candidate Genes. Cell Reports, 2018, 25, 988-1001.	6.4	28
18	A unique de novo gain-of-function variant inCAMK4associated with intellectual disability and hyperkinetic movement disorder. Journal of Physical Education and Sports Management, 2018, 4, a003293.	1.2	16

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19	Comorbidities, treatment, and pathophysiology in restless legs syndrome. Lancet Neurology, The, 2018, 17, 994-1005.	10.2	166
20	Treatment of restless legs syndrome: Evidenceâ€based review and implications for clinical practice (Revised 2017)^{Â§}. Movement Disorders, 2018, 33, 1077-1091.	3.9	136
21	Immuneâ€Array Analysis in Sporadic Inclusion Body Myositis Reveals HLAâ€“DRB1 Amino Acid Heterogeneity Across the Myositis Spectrum. Arthritis and Rheumatology, 2017, 69, 1090-1099.	5.6	41
22	MEIS1 variant as a determinant of autonomic imbalance in Restless Legs Syndrome. Scientific Reports, 2017, 7, 46620.	3.3	22
23	Introduction: Towards a better understanding of the science of RLS/WED. Sleep Medicine, 2017, 31, 1-2.	1.6	14
24	Genome-wide association analysis of insomnia complaints identifies risk genes and genetic overlap with psychiatric and metabolic traits. Nature Genetics, 2017, 49, 1584-1592.	21.4	248
25	Genetics of restless legs syndrome. Sleep Medicine, 2017, 31, 18-22.	1.6	40
26	Identification of novel risk loci for restless legs syndrome in genome-wide association studies in individuals of European ancestry: a meta-analysis. Lancet Neurology, The, 2017, 16, 898-907.	10.2	191
27	Molecular diversity of combined and complex dystonia: insights from diagnostic exome sequencing. Neurogenetics, 2017, 18, 195-205.	1.4	37
28	Ferric carboxymaltose in patients with restless legs syndrome and nonanemic iron deficiency: A randomized trial. Movement Disorders, 2017, 32, 1478-1482.	3.9	53
29	Meis1 effects on motor phenotypes and the sensorimotor system in mice. DMM Disease Models and Mechanisms, 2017, 10, 981-991.	2.4	25
30	Animal models of RLS phenotypes. Sleep Medicine, 2017, 31, 23-28.	1.6	30
31	Clinical exome sequencing in earlyâ€onset generalized dystonia and largeâ€scale resequencing followâ€up. Movement Disorders, 2017, 32, 549-559.	3.9	94
32	Alterations in Lipid and Inositol Metabolisms in Two Dopaminergic Disorders. PLoS ONE, 2016, 11, e0147129.	2.5	31
33	The clinical phenotype of earlyâ€onset isolated dystonia caused by recessive COL6A3 mutations (DYT27). Movement Disorders, 2016, 31, 747-750.	3.9	13
34	Systematic TOR1A non-c.907_909delGAG variant analysis in isolated dystonia and controls. Parkinsonism and Related Disorders, 2016, 31, 119-123.	2.2	4
35	Haploinsufficiency of KMT2B, Encoding the Lysine-Specific Histone Methyltransferase 2B, Results in Early-Onset Generalized Dystonia. American Journal of Human Genetics, 2016, 99, 1377-1387.	6.2	135
36	Guidelines for the first-line treatment of restless legs syndrome/Willisâ€“Ekbom disease, prevention and treatment of dopaminergic augmentation: a combined task force of the IRLSSG, EURLSSG, and the RLS-foundation. Sleep Medicine, 2016, 21, 1-11.	1.6	242

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37	Restless legs syndrome associated with major diseases. Neurology, 2016, 86, 1336-1343.	1.1	276
38	Dense genotyping of immune-related loci in idiopathic inflammatory myopathies confirms HLA alleles as the strongest genetic risk factor and suggests different genetic background for major clinical subgroups. Annals of the Rheumatic Diseases, 2016, 75, 1558-1566.	0.9	127
39	Excess of rare coding variants in PLD3 in late- but not early-onset Alzheimerâ€™s disease. Human Genome Variation, 2015, 2, 14028.	0.7	18
40	TOR1A, THAP1, and GNAL mutational screening in Austrian patients with primary isolated dystonia. Movement Disorders, 2015, 30, 1853-1854.	3.9	4
41	Reply to letter: Novel compound heterozygous mutations in PRKRA cause pure dystonia. Movement Disorders, 2015, 30, 878-879.	3.9	1
42	Recessive Mutations in the Î±3 (VI) Collagen Gene COL6A3 Cause Early-Onset Isolated Dystonia. American Journal of Human Genetics, 2015, 96, 883-893.	6.2	79
43	Identification of Immune-Relevant Factors Conferring Sarcoidosis Genetic Risk. American Journal of Respiratory and Critical Care Medicine, 2015, 192, 727-736.	5.6	94
44	Rare variants in Î²-Amyloid precursor protein (APP) and Parkinsonâ€™s disease. European Journal of Human Genetics, 2015, 23, 1328-1333.	2.8	50
45	Restless legs syndromeâ€"current therapies and management of augmentation. Nature Reviews Neurology, 2015, 11, 434-445.	10.1	74
46	Association of CLEC16A with human common variable immunodeficiency disorder and role in murine B cells. Nature Communications, 2015, 6, 6804.	12.8	63
47	Large-scale TUBB4A mutational screening in isolated dystonia and controls. Parkinsonism and Related Disorders, 2015, 21, 1278-1281.	2.2	7
48	Class II HLA interactions modulate genetic risk for multiple sclerosis. Nature Genetics, 2015, 47, 1107-1113.	21.4	312
49	Genome-wide Association Analysis of Psoriatic Arthritis and Cutaneous Psoriasis Reveals Differences in Their Genetic Architecture. American Journal of Human Genetics, 2015, 97, 816-836.	6.2	245
50	Clinical Phenotype and Genetics of Restless Legs Syndrome. , 2015, , 1145-1162.		0
51	Genetics of Restless Legs Syndrome (RLS). , 2015, , 331-351.		0
52	Mutant Adenosine Deaminase 2 in a Polyarteritis Nodosa Vasculopathy. New England Journal of Medicine, 2014, 370, 921-931.	27.0	566
53	Narcolepsy is a common phenotype in HSAN IE and ADCA-DN. Brain, 2014, 137, 1643-1655.	7.6	49
54	Restless Legs Syndrome-associated intronic common variant in Meis1 alters enhancer function in the developing telencephalon. Genome Research, 2014, 24, 592-603.	5.5	102

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55	DYT16 revisited: Exome sequencing identifiesPRKRAmutations in a European dystonia family. Movement Disorders, 2014, 29, 1504-1510.	3.9	38
56	Iron in Restless Legs Syndrome. Movement Disorders Clinical Practice, 2014, 1, 161-172.	1.5	4
57	Rare variants in LRRK1 and Parkinson's disease. Neurogenetics, 2014, 15, 49-57.	1.4	33
58	Novel loci affecting iron homeostasis and their effects in individuals at risk for hemochromatosis. Nature Communications, 2014, 5, 4926.	12.8	192
59	Targeted Resequencing and Systematic InÂVivo Functional Testing Identifies Rare Variants in MEIS1 as Significant Contributors to Restless Legs Syndrome. American Journal of Human Genetics, 2014, 95, 85-95.	6.2	52
60	â€œMalignant restless legs syndromeâ€â€”A curse or a blessing?. Sleep Medicine, 2014, 15, 155-156.	1.6	0
61	Polysomnographic and neurometabolic features may mark preclinical autosomal dominant cerebellar ataxia, deafness, and narcolepsy due to a mutation in the DNA (cytosine-5-)-methyltransferase gene, DNMT1. Sleep Medicine, 2014, 15, 582-585.	1.6	6
62	Periodic Leg Movements during Sleep Are Associated with Polymorphisms in BTBD9, TOX3/BC034767, MEIS1, MAP2K5/SKOR1, and PTPRD. Sleep, 2014, 37, 1535-1542.	1.1	102
63	HLA DQB1*06:02 Negative Narcolepsy with Hypocretin/Orexin Deficiency. Sleep, 2014, 37, 1601-1608.	1.1	59
64	Rare sequence variants in ANO3 and GNAL in a primary torsion dystonia series and controls. Movement Disorders, 2014, 29, 143-147.	3.9	49
65	Blood cis-eQTL Analysis Fails to Identify Novel Association Signals among Sub-Threshold Candidates from Genome-Wide Association Studies in Restless Legs Syndrome. PLoS ONE, 2014, 9, e98092.	2.5	2
66	Analysis of immune-related loci identifies 48 new susceptibility variants for multiple sclerosis. Nature Genetics, 2013, 45, 1353-1360.	21.4	1,213
67	Association Between Variants of PRDM1 and NDP52 and Crohn's Disease, Based on Exome Sequencing and Functional Studies. Gastroenterology, 2013, 145, 339-347.	1.3	149
68	Prolonged release oxycodoneâ€“naloxone for treatment of severe restless legs syndrome after failure of previous treatment: a double-blind, randomised, placebo-controlled trial with an open-label extension. Lancet Neurology, The, 2013, 12, 1141-1150.	10.2	188
69	When restless legs syndrome turns malignant. Sleep Medicine, 2013, 14, 575-577.	1.6	10
70	Mitochondrial membrane protein associated neurodegenration: A novel variant of neurodegeneration with brain iron accumulation. Movement Disorders, 2013, 28, 224-227.	3.9	162
71	The role of <scp>SCARB2</scp> as susceptibility factor in <scp>P</scp>arkinson's disease. Movement Disorders, 2013, 28, 538-540.	3.9	41
72	High-density genotyping study identifies four new susceptibility loci for atopic dermatitis. Nature Genetics, 2013, 45, 808-812.	21.4	167

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73	Dense genotyping of immune-related disease regions identifies nine new risk loci for primary sclerosing cholangitis. Nature Genetics, 2013, 45, 670-675.	21.4	339
74	Network-Based Multiple Sclerosis Pathway Analysis with GWAS Data from 15,000 Cases and 30,000 Controls. American Journal of Human Genetics, 2013, 92, 854-865.	6.2	164
75	ImmunoChip Study Implicates Antigen Presentation to T Cells in Narcolepsy. PLoS Genetics, 2013, 9, e1003270.	3.5	206
76	Dilution of candidates: the case of iron-related genes in restless legs syndrome. European Journal of Human Genetics, 2013, 21, 410-414.	2.8	32
77	Restless legs syndrome. Current Opinion in Pulmonary Medicine, 2013, 19, 594-600.	2.6	97
78	Genetic variants in the immunoglobulin heavy chain locus are associated with the IgG index in multiple sclerosis. Annals of Neurology, 2013, 73, 86-94.	5.3	38
79	Rare Variants in PLXNA4 and Parkinsonâ€™s Disease. PLoS ONE, 2013, 8, e79145.	2.5	16
80	Niemann-Pick C Disease Gene Mutations and Age-Related Neurodegenerative Disorders. PLoS ONE, 2013, 8, e82879.	2.5	50
81	Mutations in DNMT1 cause autosomal dominant cerebellar ataxia, deafness and narcolepsy. Human Molecular Genetics, 2012, 21, 2205-2210.	2.9	225
82	Variants in eukaryotic translation initiation factor 4G1 in sporadic Parkinsonâ€™s disease. Neurogenetics, 2012, 13, 281-285.	1.4	30
83	Mutational screening of THAP1 in a German population with primary dystonia. Parkinsonism and Related Disorders, 2012, 18, 104-106.	2.2	6
84	Hostâ€“microbe interactions have shaped the genetic architecture of inflammatory bowel disease. Nature, 2012, 491, 119-124.	27.8	4,038
85	Identification of 15 new psoriasis susceptibility loci highlights the role of innate immunity. Nature Genetics, 2012, 44, 1341-1348.	21.4	848
86	Variation within the Huntington's Disease Gene Influences Normal Brain Structure. PLoS ONE, 2012, 7, e29809.	2.5	30
87	<scp>PSEA</scp>: Phenotype Set Enrichment Analysisâ€”A New Method for Analysis of Multiple Phenotypes. Genetic Epidemiology, 2012, 36, 244-252.	1.3	24
88	Genetic risk and a primary role for cell-mediated immune mechanisms in multiple sclerosis. Nature, 2011, 476, 214-219.	27.8	2,400
89	Genetics of Restless Legs Syndrome: Mendelian, Complex, and Everything in Between. Sleep Medicine Clinics, 2011, 6, 203-215.	2.6	12
90	Common variants in P2RY11 are associated with narcolepsy. Nature Genetics, 2011, 43, 66-71.	21.4	215

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91	A Mutation in VPS35, Encoding a Subunit of the Retromer Complex, Causes Late-Onset Parkinson Disease. American Journal of Human Genetics, 2011, 89, 168-175.	6.2	757
92	Absence of an Orphan Mitochondrial Protein, C19orf12, Causes a Distinct Clinical Subtype of Neurodegeneration with Brain Iron Accumulation. American Journal of Human Genetics, 2011, 89, 543-550.	6.2	224
93	When Parkinsonâ€™s disease patients go to sleep: specific sleep disturbances related to Parkinsonâ€™s disease. Journal of Neurology, 2011, 258, 328-335.	3.6	56
94	Novel SCARB2 mutation in action myoclonus-renal failure syndrome and evaluation of SCARB2mutations in isolated AMRF features. BMC Neurology, 2011, 11, 134.	1.8	22
95	Myoclonusâ€dystonia in 18p deletion syndrome. Movement Disorders, 2011, 26, 560-561.	3.9	17
96	Novel association to the proprotein convertase PCSK7 gene locus revealed by analysing soluble transferrin receptor (sTfR) levels. Human Molecular Genetics, 2011, 20, 1042-1047.	2.9	62
97	MEIS1 and BTBD9: genetic association with restless leg syndrome in end stage renal disease. Journal of Medical Genetics, 2011, 48, 462-466.	3.2	59
98	Genetics of sleep disorders. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2011, 99, 681-693.	1.8	7
99	Genome-Wide Association Study Identifies Novel Restless Legs Syndrome Susceptibility Loci on 2p14 and 16q12.1. PLoS Genetics, 2011, 7, e1002171.	3.5	163
100	Shortâ€term attention and verbal fluency is decreased in restless legs syndrome patients. Movement Disorders, 2010, 25, 2641-2648.	3.9	65
101	Recent advances in the diagnosis, genetics and treatment of restless legs syndrome. Journal of Neurology, 2009, 256, 539-553.	3.6	91
102	Narcolepsy is strongly associated with the T-cell receptor alpha locus. Nature Genetics, 2009, 41, 708-711.	21.4	445
103	Genetics of restless legs syndrome. Current Neurology and Neuroscience Reports, 2008, 8, 211-216.	4.2	55
104	Suggestive evidence for linkage for restless legs syndrome on chromosome 19p13. Neurogenetics, 2008, 9, 75-82.	1.4	61
105	Variants in the neuronal nitric oxide synthase (nNOS, NOS1) gene are associated with restless legs syndrome. Movement Disorders, 2008, 23, 350-358.	3.9	103
106	PTPRD (protein tyrosine phosphatase receptor type delta) is associated with restless legs syndrome. Nature Genetics, 2008, 40, 946-948.	21.4	252
107	Heritability of Sleep Electroencephalogram. Biological Psychiatry, 2008, 64, 344-348.	1.3	146
108	Genetics of restless legs syndrome. Neurology, 2008, 70, 664-665.	1.1	13

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109	Diagnostic Standards for Dopaminergic Augmentation of Restless Legs Syndrome: Report from a World Association of Sleep Medicine â€“ International Restless Legs Syndrome Study Group Consensus Conference at the Max Planck Institute. Sleep Medicine, 2007, 8, 520-530.	1.6	264
110	Erratum to â€œDiagnostic standards for dopaminergic augmentation of restless legs syndrome: Report from a World Association of Sleep Medicine â€“ International Restless Legs Syndrome Study Group consensus conference at the Max Planck Instituteâ€•[Sleep Med. 8 (2007) 520â€“530]. Sleep Medicine, 2007, 8, 788.	1.6	1
111	Family-based association study of the restless legs syndrome loci 2 and 3 in a European population. Movement Disorders, 2007, 22, 207-212.	3.9	31
112	Clinical trials in restless legs syndromeâ€”Recommendations of the European RLS Study Group (EURLSSG). Movement Disorders, 2007, 22 Suppl 18, S495-504.	3.9	15
113	Genetics of restless legs syndrome (RLS): State-of-the-art and future directions. Movement Disorders, 2007, 22, S449-S458.	3.9	73
114	Animal studies in restless legs syndrome. Movement Disorders, 2007, 22, S459-S465.	3.9	12
115	Augmentation as a treatment complication of restless legs syndrome: Concept and management. Movement Disorders, 2007, 22, S476-S484.	3.9	81
116	Genome-wide association study of restless legs syndrome identifies common variants in three genomic regions. Nature Genetics, 2007, 39, 1000-1006.	21.4	633
117	Paroxetine treatment improves motor symptoms in patients with multiple system atrophy. Parkinsonism and Related Disorders, 2006, 12, 432-437.	2.2	44
118	Epidemiology of restless legs syndrome: The current status. Sleep Medicine Reviews, 2006, 10, 153-167.	8.5	219
119	Genetics of restless legs syndrome. Sleep Medicine Reviews, 2006, 10, 179-183.	8.5	39
120	Olanzapine-induced Oculogyric Crisis. Journal of Clinical Psychopharmacology, 2006, 26, 431.	1.4	18
121	Evidence for further genetic locus heterogeneity and confirmation of RLSâ€1 in restless legs syndrome. Movement Disorders, 2006, 21, 28-33.	3.9	79
122	High-dose treatment with pergolide in Parkinson's disease patients with motor fluctuations and dyskinesias. Parkinsonism and Related Disorders, 2005, 11, 393-398.	2.2	17
123	Action myoclonus-renal failure syndrome: characterization of a unique cerebro-renal disorder. Brain, 2004, 127, 2173-2182.	7.6	89
124	Current treatment options for restless legs syndrome. Expert Opinion on Pharmacotherapy, 2003, 4, 1727-1738.	1.8	18
125	Das Restless-legs-Syndrom (RLS). , 2003, , 131-142.		0
126	Assessment of spontaneously occurring periodic limb movements in sleep in the rat. Journal of the Neurological Sciences, 2002, 198, 71-77.	0.6	28

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127	Sleep and periodic limb movements in corticobasal degeneration. Sleep Medicine, 2002, 3, 33-36.	1.6	61
128	Pergolide restores sleep maintenance but impairs sleep EEG synchronization in patients with restless legs syndrome. Sleep Medicine, 2002, 3, 49-54.	1.6	29
129	Complex segregation analysis of restless legs syndrome provides evidence for an autosomal dominant mode of inheritance in early age at onset families. Annals of Neurology, 2002, 52, 297-302.	5.3	229
130	Longâ€term course of restless legs syndrome in dialysis patients after kidney transplantation. Movement Disorders, 2002, 17, 1072-1076.	3.9	183
131	The Restless Legs Syndrome - A Genetic View of the Disease. Das Restless Legs Syndrom aus genetischer Sicht. Somnologie, 2002, 6, 3-6.	1.5	5
132	Opioid and dopamine antagonist drug challenges in untreated restless legs syndrome patients. Sleep Medicine, 2001, 2, 57-61.	1.6	104
133	Longâ€Term followâ€up on restless legs syndrome patients treated with opioids. Movement Disorders, 2001, 16, 1105-1109.	3.9	195
134	Indications for Performing Polysomnography in the Diagnosis and Treatment of Restless Legs Syndrome. Indikationen zur Polysomnographie in der Diagnose und Therapie des Restless Legs Syndroms. Somnologie, 2001, 5, 159-162.	1.5	16
135	Mutations in the gene encoding É›-sarcoglycan cause myoclonusâ€“dystonia syndrome. Nature Genetics, 2001, 29, 66-69.	21.4	523
136	Intrathecal baclofen for dystonia. Movement Disorders, 2001, 16, 1201.	3.9	2
137	Periodic limb movements in syringomyelia and syringobulbia. Movement Disorders, 2000, 15, 752-753.	3.9	19
138	Clinical Characteristics and Frequency of the Hereditary Restless Legs Syndrome in a Population of 300 Patients. Sleep, 2000, 23, 1-6.	1.1	299
139	Magnetic resonance imaging findings in corticobasal degeneration. Movement Disorders, 1999, 14, 669-673.	3.9	23
140	Aetiology and Treatment of Restless Legs Syndrome. CNS Drugs, 1999, 12, 9-20.	5.9	21
141	Treatment of restless leg syndrome with pergolide-an open clinical trial. Movement Disorders, 1998, 13, 566-569.	3.9	40