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	Hostâ€“microbe interactions have shaped the genetic architecture of inflammatory bowel disease. Nature, 2012, 491, 119-124.	27.8	4,038
2	Genetic risk and a primary role for cell-mediated immune mechanisms in multiple sclerosis. Nature, 2011, 476, 214-219.	27.8	2,400
3	Analysis of immune-related loci identifies 48 new susceptibility variants for multiple sclerosis. Nature Genetics, 2013, 45, 1353-1360.	21.4	1,213
4	Identification of 15 new psoriasis susceptibility loci highlights the role of innate immunity. Nature Genetics, 2012, 44, 1341-1348.	21.4	848
5	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
6	Genome-wide association study of restless legs syndrome identifies common variants in three genomic regions. Nature Genetics, 2007, 39, 1000-1006.	21.4	633
7	Mutant Adenosine Deaminase 2 in a Polyarteritis Nodosa Vasculopathy. New England Journal of Medicine, 2014, 370, 921-931.	27.0	566
8	Mutations in the gene encoding É›-sarcoglycan cause myoclonusâ€“dystonia syndrome. Nature Genetics, 2001, 29, 66-69.	21.4	523
9	Narcolepsy is strongly associated with the T-cell receptor alpha locus. Nature Genetics, 2009, 41, 708-711.	21.4	445
10	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
11	Class II HLA interactions modulate genetic risk for multiple sclerosis. Nature Genetics, 2015, 47, 1107-1113.	21.4	312
12	Clinical Characteristics and Frequency of the Hereditary Restless Legs Syndrome in a Population of 300 Patients. Sleep, 2000, 23, 1-6.	1.1	299
13	Restless legs syndrome associated with major diseases. Neurology, 2016, 86, 1336-1343.	1.1	276
14	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
15	PTPRD (protein tyrosine phosphatase receptor type delta) is associated with restless legs syndrome. Nature Genetics, 2008, 40, 946-948.	21.4	252
16	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
17	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
18	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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19	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
20	Mutations in DNMT1 cause autosomal dominant cerebellar ataxia, deafness and narcolepsy. Human Molecular Genetics, 2012, 21, 2205-2210.	2.9	225
21	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
22	Epidemiology of restless legs syndrome: The current status. Sleep Medicine Reviews, 2006, 10, 153-167.	8.5	219
23	Common variants in P2RY11 are associated with narcolepsy. Nature Genetics, 2011, 43, 66-71.	21.4	215
24	ImmunoChip Study Implicates Antigen Presentation to T Cells in Narcolepsy. PLoS Genetics, 2013, 9, e1003270.	3.5	206
25	Longâ€Term followâ€up on restless legs syndrome patients treated with opioids. Movement Disorders, 2001, 16, 1105-1109.	3.9	195
26	Novel loci affecting iron homeostasis and their effects in individuals at risk for hemochromatosis. Nature Communications, 2014, 5, 4926.	12.8	192
27	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
28	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
29	Longâ€term course of restless legs syndrome in dialysis patients after kidney transplantation. Movement Disorders, 2002, 17, 1072-1076.	3.9	183
30	High-density genotyping study identifies four new susceptibility loci for atopic dermatitis. Nature Genetics, 2013, 45, 808-812.	21.4	167
31	Comorbidities, treatment, and pathophysiology in restless legs syndrome. Lancet Neurology, The, 2018, 17, 994-1005.	10.2	166
32	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
33	Genome-Wide Association Study Identifies Novel Restless Legs Syndrome Susceptibility Loci on 2p14 and 16q12.1. PLoS Genetics, 2011, 7, e1002171.	3.5	163
34	Mitochondrial membrane protein associated neurodegenration: A novel variant of neurodegeneration with brain iron accumulation. Movement Disorders, 2013, 28, 224-227.	3.9	162
35	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
36	Heritability of Sleep Electroencephalogram. Biological Psychiatry, 2008, 64, 344-348.	1.3	146

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37	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
38	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
39	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
40	Opioid and dopamine antagonist drug challenges in untreated restless legs syndrome patients. Sleep Medicine, 2001, 2, 57-61.	1.6	104
41	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
42	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
43	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
44	Restless legs syndrome. Current Opinion in Pulmonary Medicine, 2013, 19, 594-600.	2.6	97
45	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
46	Identification of Immune-Relevant Factors Conferring Sarcoidosis Genetic Risk. American Journal of Respiratory and Critical Care Medicine, 2015, 192, 727-736.	5.6	94
47	Clinical exome sequencing in earlyâ€onset generalized dystonia and largeâ€scale resequencing followâ€up. Movement Disorders, 2017, 32, 549-559.	3.9	94
48	Recent advances in the diagnosis, genetics and treatment of restless legs syndrome. Journal of Neurology, 2009, 256, 539-553.	3.6	91
49	Action myoclonus-renal failure syndrome: characterization of a unique cerebro-renal disorder. Brain, 2004, 127, 2173-2182.	7.6	89
50	Augmentation as a treatment complication of restless legs syndrome: Concept and management. Movement Disorders, 2007, 22, S476-S484.	3.9	81
51	Evidence for further genetic locus heterogeneity and confirmation of RLSâ€1 in restless legs syndrome. Movement Disorders, 2006, 21, 28-33.	3.9	79
52	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
53	Restless legs syndromeâ€"current therapies and management of augmentation. Nature Reviews Neurology, 2015, 11, 434-445.	10.1	74
54	Genetics of restless legs syndrome (RLS): State-of-the-art and future directions. Movement Disorders, 2007, 22, S449-S458.	3.9	73

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55	Shortâ€term attention and verbal fluency is decreased in restless legs syndrome patients. Movement Disorders, 2010, 25, 2641-2648.	3.9	65
56	Association of CLEC16A with human common variable immunodeficiency disorder and role in murine B cells. Nature Communications, 2015, 6, 6804.	12.8	63
57	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
58	Sleep and periodic limb movements in corticobasal degeneration. Sleep Medicine, 2002, 3, 33-36.	1.6	61
59	Suggestive evidence for linkage for restless legs syndrome on chromosome 19p13. Neurogenetics, 2008, 9, 75-82.	1.4	61
60	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
61	HLA DQB1*06:02 Negative Narcolepsy with Hypocretin/Orexin Deficiency. Sleep, 2014, 37, 1601-1608.	1.1	59
62	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
63	Genetics of restless legs syndrome. Current Neurology and Neuroscience Reports, 2008, 8, 211-216.	4.2	55
64	Ferric carboxymaltose in patients with restless legs syndrome and nonanemic iron deficiency: A randomized trial. Movement Disorders, 2017, 32, 1478-1482.	3.9	53
65	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
66	Rare variants in Î²-Amyloid precursor protein (APP) and Parkinsonâ€™s disease. European Journal of Human Genetics, 2015, 23, 1328-1333.	2.8	50
67	Niemann-Pick C Disease Gene Mutations and Age-Related Neurodegenerative Disorders. PLoS ONE, 2013, 8, e82879.	2.5	50
68	Narcolepsy is a common phenotype in HSAN IE and ADCA-DN. Brain, 2014, 137, 1643-1655.	7.6	49
69	Rare sequence variants in ANO3 and GNAL in a primary torsion dystonia series and controls. Movement Disorders, 2014, 29, 143-147.	3.9	49
70	Paroxetine treatment improves motor symptoms in patients with multiple system atrophy. Parkinsonism and Related Disorders, 2006, 12, 432-437.	2.2	44
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	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

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73	Treatment of restless leg syndrome with pergolide-an open clinical trial. Movement Disorders, 1998, 13, 566-569.	3.9	40
74	Genetics of restless legs syndrome. Sleep Medicine, 2017, 31, 18-22.	1.6	40
75	Genetics of restless legs syndrome. Sleep Medicine Reviews, 2006, 10, 179-183.	8.5	39
76	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
77	DYT16 revisited: Exome sequencing identifiesPRKRAmutations in a European dystonia family. Movement Disorders, 2014, 29, 1504-1510.	3.9	38
78	Molecular diversity of combined and complex dystonia: insights from diagnostic exome sequencing. Neurogenetics, 2017, 18, 195-205.	1.4	37
79	Rare variants in LRRK1 and Parkinson's disease. Neurogenetics, 2014, 15, 49-57.	1.4	33
80	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
81	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
82	Alterations in Lipid and Inositol Metabolisms in Two Dopaminergic Disorders. PLoS ONE, 2016, 11, e0147129.	2.5	31
83	Variants in eukaryotic translation initiation factor 4G1 in sporadic Parkinsonâ€™s disease. Neurogenetics, 2012, 13, 281-285.	1.4	30
84	Variation within the Huntington's Disease Gene Influences Normal Brain Structure. PLoS ONE, 2012, 7, e29809.	2.5	30
85	Animal models of RLS phenotypes. Sleep Medicine, 2017, 31, 23-28.	1.6	30
86	Pergolide restores sleep maintenance but impairs sleep EEG synchronization in patients with restless legs syndrome. Sleep Medicine, 2002, 3, 49-54.	1.6	29
87	Assessment of spontaneously occurring periodic limb movements in sleep in the rat. Journal of the Neurological Sciences, 2002, 198, 71-77.	0.6	28
88	KMT2B Is Selectively Required for Neuronal Transdifferentiation, and Its Loss Exposes Dystonia Candidate Genes. Cell Reports, 2018, 25, 988-1001.	6.4	28
89	Meis1 effects on motor phenotypes and the sensorimotor system in mice. DMM Disease Models and Mechanisms, 2017, 10, 981-991.	2.4	25
90	<scp>PSEA</scp>: Phenotype Set Enrichment Analysisâ€”A New Method for Analysis of Multiple Phenotypes. Genetic Epidemiology, 2012, 36, 244-252.	1.3	24

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91	Magnetic resonance imaging findings in corticobasal degeneration. Movement Disorders, 1999, 14, 669-673.	3.9	23
92	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
93	MEIS1 variant as a determinant of autonomic imbalance in Restless Legs Syndrome. Scientific Reports, 2017, 7, 46620.	3.3	22
94	Aetiology and Treatment of Restless Legs Syndrome. CNS Drugs, 1999, 12, 9-20.	5.9	21
95	Role of MEIS1 in restless legs syndrome: From GWAS to functional studies in mice. Advances in Pharmacology, 2019, 84, 175-184.	2.0	21
96	Periodic limb movements in syringomyelia and syringobulbia. Movement Disorders, 2000, 15, 752-753.	3.9	19
97	Identification of Restless Legs Syndrome Genes by Mutational Load Analysis. Annals of Neurology, 2020, 87, 184-193.	5.3	19
98	Current treatment options for restless legs syndrome. Expert Opinion on Pharmacotherapy, 2003, 4, 1727-1738.	1.8	18
99	Olanzapine-induced Oculogyric Crisis. Journal of Clinical Psychopharmacology, 2006, 26, 431.	1.4	18
100	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
101	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
102	Myoclonusâ€dystonia in 18p deletion syndrome. Movement Disorders, 2011, 26, 560-561.	3.9	17
103	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
104	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
105	Rare Variants in PLXNA4 and Parkinsonâ€™s Disease. PLoS ONE, 2013, 8, e79145.	2.5	16
106	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
107	Restless Legs Syndrome and Other Movement Disorders of Sleepâ€"Treatment Update. Current Treatment Options in Neurology, 2018, 20, 55.	1.8	15
108	Introduction: Towards a better understanding of the science of RLS/WED. Sleep Medicine, 2017, 31, 1-2.	1.6	14

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109	Sleep disturbance by pramipexole is modified by Meis1 in mice. Journal of Sleep Research, 2018, 27, e12557.	3.2	14
110	Genetics of restless legs syndrome. Neurology, 2008, 70, 664-665.	1.1	13
111	The clinical phenotype of earlyâ€onset isolated dystonia caused by recessive COL6A3 mutations (DYT27). Movement Disorders, 2016, 31, 747-750.	3.9	13
112	Animal studies in restless legs syndrome. Movement Disorders, 2007, 22, S459-S465.	3.9	12
113	Genetics of Restless Legs Syndrome: Mendelian, Complex, and Everything in Between. Sleep Medicine Clinics, 2011, 6, 203-215.	2.6	12
114	When restless legs syndrome turns malignant. Sleep Medicine, 2013, 14, 575-577.	1.6	10
115	Microstructural white matter abnormalities in patients with COL6A3 mutations (DYT27 dystonia). Parkinsonism and Related Disorders, 2018, 46, 74-78.	2.2	10
116	A Novel Variant of <scp>ATP5MC3</scp> Associated with Both Dystonia and Spastic Paraplegia. Movement Disorders, 2022, 37, 375-383.	3.9	10
117	Genetics of sleep disorders. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2011, 99, 681-693.	1.8	7
118	Large-scale TUBB4A mutational screening in isolated dystonia and controls. Parkinsonism and Related Disorders, 2015, 21, 1278-1281.	2.2	7
119	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
120	Mutational screening of THAP1 in a German population with primary dystonia. Parkinsonism and Related Disorders, 2012, 18, 104-106.	2.2	6
121	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
122	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
123	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
124	Iron in Restless Legs Syndrome. Movement Disorders Clinical Practice, 2014, 1, 161-172.	1.5	4
125	TOR1A, THAP1, and GNAL mutational screening in Austrian patients with primary isolated dystonia. Movement Disorders, 2015, 30, 1853-1854.	3.9	4
126	Systematic TOR1A non-c.907_909delGAG variant analysis in isolated dystonia and controls. Parkinsonism and Related Disorders, 2016, 31, 119-123.	2.2	4

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127	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
128	Private variants in PRKN are associated with late-onset Parkinson's disease. Parkinsonism and Related Disorders, 2020, 75, 24-26.	2.2	4
129	Candidate variants in TUB are associated with familial tremor. PLoS Genetics, 2020, 16, e1009010.	3.5	3
130	Myoclonic dystonia phenotype related to a novel calmodulin-binding transcription activator 1 sequence variant. Neurogenetics, 2021, 22, 137-141.	1.4	3
131	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
132	Intrathecal baclofen for dystonia. Movement Disorders, 2001, 16, 1201.	3.9	2
133	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
134	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
135	Reply to letter: Novel compound heterozygous mutations in PRKRA cause pure dystonia. Movement Disorders, 2015, 30, 878-879.	3.9	1
136	Collagen VI Regulates Motor Circuit Plasticity and Motor Performance by Cannabinoid Modulation. Journal of Neuroscience, 2022, 42, 1557-1573.	3.6	1
137	â€œMalignant restless legs syndromeâ€â€”A curse or a blessing?. Sleep Medicine, 2014, 15, 155-156.	1.6	0
138	Clinical Phenotype and Genetics of Restless Legs Syndrome. , 2015, , 1145-1162.		0
139	Das Restless-legs-Syndrom (RLS). , 2003, , 131-142.		0
140	Genetics of Restless Legs Syndrome (RLS). , 2015, , 331-351.		0
141	The genetics of restless legs syndrome. , 2021, , .		0