Juliane Winkelmann

List of Publications by Year
in descending order

109	19,637	31976 53	26613 107
papers	citations	h-index	g-index

113	113	113	28198
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Clinical exome sequencing in earlyâ€onset generalized dystonia and largeâ€scale resequencing followâ€up. Movement Disorders, 2017, 32, 549-559.	3.9	94
2	SOX5-Null Heterozygous Mutation in a Family with Adult-Onset Hyperkinesia and Behavioral Abnormalities. Case Reports in Genetics, 2017, 2017, 1-6.	0.2	4
3	Alterations in Lipid and Inositol Metabolisms in Two Dopaminergic Disorders. PLoS ONE, 2016, 11, e0147129.	2.5	31
4	The clinical phenotype of earlyâ€onset isolated dystonia caused by recessive COL6A3 mutations (DYT27). Movement Disorders, 2016, 31, 747-750.	3.9	13
5	Systematic TOR1A non-c.907_909delGAG variant analysis in isolated dystonia and controls. Parkinsonism and Related Disorders, 2016, 31, 119-123.	2.2	4
6	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
7	Restless legs syndrome associated with major diseases. Neurology, 2016, 86, 1336-1343.	1.1	276
8	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
9	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
10	TOR1A, THAP1, and GNAL mutational screening in Austrian patients with primary isolated dystonia. Movement Disorders, 2015, 30, 1853-1854.	3.9	4
11	Reply to letter: Novel compound heterozygous mutations in PRKRA cause pure dystonia. Movement Disorders, 2015, 30, 878-879.	3.9	1
12	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
13	Identification of Immune-Relevant Factors Conferring Sarcoidosis Genetic Risk. American Journal of Respiratory and Critical Care Medicine, 2015, 192, 727-736.	5.6	94
14	Rare variants in Î²-Amyloid precursor protein (APP) and Parkinsonâ€™s disease. European Journal of Human Genetics, 2015, 23, 1328-1333.	2.8	50
15	Restless legs syndromeâ€”current therapies and management of augmentation. Nature Reviews Neurology, 2015, 11, 434-445.	10.1	74
16	Association of CLEC16A with human common variable immunodeficiency disorder and role in murine B cells. Nature Communications, 2015, 6, 6804.	12.8	63
17	Large-scale TUBB4A mutational screening in isolated dystonia and controls. Parkinsonism and Related Disorders, 2015, 21, 1278-1281.	2.2	7
18	Class II HLA interactions modulate genetic risk for multiple sclerosis. Nature Genetics, 2015, 47, 1107-1113.	21.4	312

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19	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
20	Clinical Phenotype and Genetics of Restless Legs Syndrome. , 2015, , 1145-1162.		0
21	Genetics of Restless Legs Syndrome (RLS). , 2015, , 331-351.		0
22	Mutant Adenosine Deaminase 2 in a Polyarteritis Nodosa Vasculopathy. New England Journal of Medicine, 2014, 370, 921-931.	27.0	566
23	Narcolepsy is a common phenotype in HSAN IE and ADCA-DN. Brain, 2014, 137, 1643-1655.	7.6	49
24	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
25	DYT16 revisited: Exome sequencing identifiesPRKRAmutations in a European dystonia family. Movement Disorders, 2014, 29, 1504-1510.	3.9	38
26	Iron in Restless Legs Syndrome. Movement Disorders Clinical Practice, 2014, 1, 161-172.	1.5	4
27	Rare variants in LRRK1 and Parkinson's disease. Neurogenetics, 2014, 15, 49-57.	1.4	33
28	Novel loci affecting iron homeostasis and their effects in individuals at risk for hemochromatosis. Nature Communications, 2014, 5, 4926.	12.8	192
29	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
30	â€œMalignant restless legs syndromeâ€â€”A curse or a blessing?. Sleep Medicine, 2014, 15, 155-156.	1.6	0
31	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
32	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
33	HLA DQB1*06:02 Negative Narcolepsy with Hypocretin/Orexin Deficiency. Sleep, 2014, 37, 1601-1608.	1.1	59
34	Rare sequence variants in ANO3 and GNAL in a primary torsion dystonia series and controls. Movement Disorders, 2014, 29, 143-147.	3.9	49
35	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
36	Analysis of immune-related loci identifies 48 new susceptibility variants for multiple sclerosis. Nature Genetics, 2013, 45, 1353-1360.	21.4	1,213

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37	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
38	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
39	When restless legs syndrome turns malignant. Sleep Medicine, 2013, 14, 575-577.	1.6	10
40	Mitochondrial membrane protein associated neurodegenration: A novel variant of neurodegeneration with brain iron accumulation. Movement Disorders, 2013, 28, 224-227.	3.9	162
41	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
42	High-density genotyping study identifies four new susceptibility loci for atopic dermatitis. Nature Genetics, 2013, 45, 808-812.	21.4	167
43	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
44	ImmunoChip Study Implicates Antigen Presentation to T Cells in Narcolepsy. PLoS Genetics, 2013, 9, e1003270.	3.5	206
45	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
46	Restless legs syndrome. Current Opinion in Pulmonary Medicine, 2013, 19, 594-600.	2.6	97
47	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
48	Rare Variants in PLXNA4 and Parkinsonâ€™s Disease. PLoS ONE, 2013, 8, e79145.	2.5	16
49	Niemann-Pick C Disease Gene Mutations and Age-Related Neurodegenerative Disorders. PLoS ONE, 2013, 8, e82879.	2.5	50
50	Mutations in DNMT1 cause autosomal dominant cerebellar ataxia, deafness and narcolepsy. Human Molecular Genetics, 2012, 21, 2205-2210.	2.9	225
51	Variants in eukaryotic translation initiation factor 4G1 in sporadic Parkinsonâ€™s disease. Neurogenetics, 2012, 13, 281-285.	1.4	30
52	Mutational screening of THAP1 in a German population with primary dystonia. Parkinsonism and Related Disorders, 2012, 18, 104-106.	2.2	6
53	Network-based SNP meta-analysis identifies joint and disjoint genetic features across common human diseases. BMC Genomics, 2012, 13, 490.	2.8	1
54	Hostâ€“microbe interactions have shaped the genetic architecture of inflammatory bowel disease. Nature, 2012, 491, 119-124.	27.8	4,038

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55	Identification of 15 new psoriasis susceptibility loci highlights the role of innate immunity. Nature Genetics, 2012, 44, 1341-1348.	21.4	848
56	Variation within the Huntington's Disease Gene Influences Normal Brain Structure. PLoS ONE, 2012, 7, e29809.	2.5	30
57	<scp>PSEA</scp>: Phenotype Set Enrichment Analysisâ€"A New Method for Analysis of Multiple Phenotypes. Genetic Epidemiology, 2012, 36, 244-252.	1.3	24
58	Genetic risk and a primary role for cell-mediated immune mechanisms in multiple sclerosis. Nature, 2011, 476, 214-219.	27.8	2,400
59	Genetics of Restless Legs Syndrome: Mendelian, Complex, and Everything in Between. Sleep Medicine Clinics, 2011, 6, 203-215.	2.6	12
60	Common variants in P2RY11 are associated with narcolepsy. Nature Genetics, 2011, 43, 66-71.	21.4	215
61	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
62	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
63	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
64	Parkinsonâ€™s disease risk score: moving to a premotor diagnosis. Journal of Neurology, 2011, 258, 311-315.	3.6	46
65	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
66	Myoclonusâ€dystonia in 18p deletion syndrome. Movement Disorders, 2011, 26, 560-561.	3.9	17
67	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
68	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
69	Genetics of sleep disorders. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2011, 99, 681-693.	1.8	7
70	Genome-Wide Association Study Identifies Novel Restless Legs Syndrome Susceptibility Loci on 2p14 and 16q12.1. PLoS Genetics, 2011, 7, e1002171.	3.5	163
71	Shortâ€term attention and verbal fluency is decreased in restless legs syndrome patients. Movement Disorders, 2010, 25, 2641-2648.	3.9	65
72	Common Variants at 10 Genomic Loci Influence Hemoglobin A1C Levels via Glycemic and Nonglycemic Pathways. Diabetes, 2010, 59, 3229-3239.	0.6	387

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73	Recent advances in the diagnosis, genetics and treatment of restless legs syndrome. Journal of Neurology, 2009, 256, 539-553.	3.6	91
74	Narcolepsy is strongly associated with the T-cell receptor alpha locus. Nature Genetics, 2009, 41, 708-711.	21.4	445
75	Genetics of restless legs syndrome. Current Neurology and Neuroscience Reports, 2008, 8, 211-216.	4.2	55
76	Suggestive evidence for linkage for restless legs syndrome on chromosome 19p13. Neurogenetics, 2008, 9, 75-82.	1.4	61
77	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
78	PTPRD (protein tyrosine phosphatase receptor type delta) is associated with restless legs syndrome. Nature Genetics, 2008, 40, 946-948.	21.4	252
79	Heritability of Sleep Electroencephalogram. Biological Psychiatry, 2008, 64, 344-348.	1.3	146
80	Genetics of restless legs syndrome. Neurology, 2008, 70, 664-665.	1.1	13
81	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
82	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
83	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
84	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
85	Genetics of restless legs syndrome (RLS): State-of-the-art and future directions. Movement Disorders, 2007, 22, S449-S458.	3.9	73
86	Animal studies in restless legs syndrome. Movement Disorders, 2007, 22, S459-S465.	3.9	12
87	Augmentation as a treatment complication of restless legs syndrome: Concept and management. Movement Disorders, 2007, 22, S476-S484.	3.9	81
88	Genome-wide association study of restless legs syndrome identifies common variants in three genomic regions. Nature Genetics, 2007, 39, 1000-1006.	21.4	633
89	Paroxetine treatment improves motor symptoms in patients with multiple system atrophy. Parkinsonism and Related Disorders, 2006, 12, 432-437.	2.2	44
90	Epidemiology of restless legs syndrome: The current status. Sleep Medicine Reviews, 2006, 10, 153-167.	8.5	219

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91	Genetics of restless legs syndrome. Sleep Medicine Reviews, 2006, 10, 179-183.	8.5	39
92	Olanzapine-induced Oculogyric Crisis. Journal of Clinical Psychopharmacology, 2006, 26, 431.	1.4	18
93	Evidence for further genetic locus heterogeneity and confirmation of RLSâ€1 in restless legs syndrome. Movement Disorders, 2006, 21, 28-33.	3.9	79
94	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
95	Action myoclonus-renal failure syndrome: characterization of a unique cerebro-renal disorder. Brain, 2004, 127, 2173-2182.	7.6	89
96	Current treatment options for restless legs syndrome. Expert Opinion on Pharmacotherapy, 2003, 4, 1727-1738.	1.8	18
97	Das Restless-legs-Syndrom (RLS). , 2003, , 131-142.		0
98	Assessment of spontaneously occurring periodic limb movements in sleep in the rat. Journal of the Neurological Sciences, 2002, 198, 71-77.	0.6	28
99	Sleep and periodic limb movements in corticobasal degeneration. Sleep Medicine, 2002, 3, 33-36.	1.6	61
100	Pergolide restores sleep maintenance but impairs sleep EEG synchronization in patients with restless legs syndrome. Sleep Medicine, 2002, 3, 49-54.	1.6	29
101	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
102	Longâ€term course of restless legs syndrome in dialysis patients after kidney transplantation. Movement Disorders, 2002, 17, 1072-1076.	3.9	183
103	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
104	Opioid and dopamine antagonist drug challenges in untreated restless legs syndrome patients. Sleep Medicine, 2001, 2, 57-61.	1.6	104
105	Longâ€Term followâ€up on restless legs syndrome patients treated with opioids. Movement Disorders, 2001, 16, 1105-1109.	3.9	195
106	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
107	Mutations in the gene encoding É›-sarcoglycan cause myoclonusâ€“dystonia syndrome. Nature Genetics, 2001, 29, 66-69.	21.4	523
108	Aetiology and Treatment of Restless Legs Syndrome. CNS Drugs, 1999, 12, 9-20.	5.9	21

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109	Treatment of restless leg syndrome with pergolide-an open clinical trial. Movement Disorders, 1998, 13, 566-569.	3.9	40