

# Juliane Winkelmann

## List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/3867774/publications.pdf>

Version: 2024-02-01

29  
papers

1,249  
citations

687363

13  
h-index

501196

28  
g-index

29  
all docs

29  
docs citations

29  
times ranked

2751  
citing authors

#	ARTICLE	IF	CITATIONS
1	Mutations in DNMT1 cause autosomal dominant cerebellar ataxia, deafness and narcolepsy. <i>Human Molecular Genetics</i> , 2012, 21, 2205-2210.	2.9	225
2	Identification of novel risk loci for restless legs syndrome in genome-wide association studies in individuals of European ancestry: a meta-analysis. <i>Lancet Neurology</i> , The, 2017, 16, 898-907.	10.2	191
3	Genome-Wide Association Study Identifies Novel Restless Legs Syndrome Susceptibility Loci on 2p14 and 16q12.1. <i>PLoS Genetics</i> , 2011, 7, e1002171.	3.5	163
4	Monogenic variants in dystonia: an exome-wide sequencing study. <i>Lancet Neurology</i> , The, 2020, 19, 908-918.	10.2	139
5	Opposite microglial activation stages upon loss of <sc>PGRN</sc> or <sc>TREM</sc> 2 result in reduced cerebral glucose metabolism. <i>EMBO Molecular Medicine</i> , 2019, 11, .	6.9	87
6	<i>De novo</i> variants in neurodevelopmental disordersâ€™ experiences from a tertiary care center. <i>Clinical Genetics</i> , 2021, 100, 14-28.	2.0	64
7	Association of CLEC16A with human common variable immunodeficiency disorder and role in murine B cells. <i>Nature Communications</i> , 2015, 6, 6804.	12.8	63
8	HLA DQB1*06:02 Negative Narcolepsy with Hypocretin/Orexin Deficiency. <i>Sleep</i> , 2014, 37, 1601-1608.	1.1	59
9	Narcolepsy is a common phenotype in HSAN IE and ADCA-DN. <i>Brain</i> , 2014, 137, 1643-1655.	7.6	49
10	Bi-allelic variants in RNF170 are associated with hereditary spastic paraplegia. <i>Nature Communications</i> , 2019, 10, 4790.	12.8	39
11	Meta-analyses identify DNA methylation associated with kidney function and damage. <i>Nature Communications</i> , 2021, 12, 7174.	12.8	30
12	Clinico-genetic findings in 509 frontotemporal dementia patients. <i>Molecular Psychiatry</i> , 2021, 26, 5824-5832.	7.9	23
13	Clinically relevant copy-number variants in exome sequencing data of patients with dystonia. <i>Parkinsonism and Related Disorders</i> , 2021, 84, 129-134.	2.2	15
14	WARS2 mutations cause dopa-responsive early-onset parkinsonism and progressive myoclonus ataxia. <i>Parkinsonism and Related Disorders</i> , 2022, 94, 54-61.	2.2	13
15	Neuropeptide S Receptor Gene Variation Differentially Modulates Fronto-Limbic Effective Connectivity in Childhood and Adolescence. <i>Cerebral Cortex</i> , 2015, 27, bhv259.	2.9	12
16	Variants in Mitochondrial <sc>ATP</sc> Synthase Cause Variable Neurologic Phenotypes. <i>Annals of Neurology</i> , 2022, 91, 225-237.	5.3	12
17	Severe paroxysmal dyskinesias without epilepsy in a RHOTB2 mutation carrier. <i>Parkinsonism and Related Disorders</i> , 2020, 77, 87-88.	2.2	9
18	Dystonia as a prominent presenting feature in developmental and epileptic encephalopathies: A case series. <i>Parkinsonism and Related Disorders</i> , 2021, 90, 73-78.	2.2	9

#	ARTICLE	IF	CITATIONS
19	Epigenome-wide association study of serum urate reveals insights into urate co-regulation and the SLC2A9 locus. <i>Nature Communications</i> , 2021, 12, 7173.	12.8	8
20	AOPEP variants as a novel cause of recessive dystonia: Generalized dystonia and dystonia-parkinsonism. <i>Parkinsonism and Related Disorders</i> , 2022, 97, 52-56.	2.2	7
21	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. <i>Sleep Medicine</i> , 2014, 15, 582-585.	1.6	6
22	Biallelic variants in <i>ZNF142</i> lead to a syndromic neurodevelopmental disorder. <i>Clinical Genetics</i> , 2022, 102, 98-109.	2.0	6
23	Fast versus slow disease progression in amyotrophic lateral sclerosis—clinical and genetic factors at the edges of the survival spectrum. <i>Neurobiology of Aging</i> , 2022, 119, 117-126.	3.1	5
24	Defining the genotypic and phenotypic spectrum of X-linked MSL3-related disorder. <i>Genetics in Medicine</i> , 2021, 23, 384-395.	2.4	4
25	The genetic etiology of periodic limb movement in sleep. <i>Sleep</i> , 2023, 46, .	1.1	4
26	Recessive null-allele variants in MAG associated with spastic ataxia, nystagmus, neuropathy, and dystonia. <i>Parkinsonism and Related Disorders</i> , 2020, 77, 70-75.	2.2	3
27	Common Grounds for Family Maladies. <i>Neuron</i> , 2018, 98, 671-672.	8.1	2
28	Concept of the Munich/Augsburg Consortium Precision in Mental Health for the German Center of Mental Health. <i>Frontiers in Psychiatry</i> , 2022, 13, 815718.	2.6	2
29	ExomeChip-based rare variant association study in restless legs syndrome. <i>Sleep Medicine</i> , 2022, 94, 26-30.	1.6	0