# Juliane Winkelmann

## List of Publications by Year in descending order

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

Version: 2024-02-01

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|---|---|---|---|
| 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 | The genetic etiology of periodic limb movement in sleep. Sleep, 2023, 46, . | 1.1 | 4 |
| 2 | WARS2 mutations cause dopa-responsive early-onset parkinsonism and progressive myoclonus ataxia. Parkinsonism and Related Disorders, 2022, 94, 54-61. | 2.2 | 13 |
| 3 | Variants in Mitochondrial <scp>ATP</scp> Synthase Cause Variable Neurologic Phenotypes. Annals of Neurology, 2022, 91, 225-237. | 5.3 | 12 |
| 4 | Concept of the Munich/Augsburg Consortium Precision in Mental Health for the German Center of Mental Health. Frontiers in Psychiatry, 2022, 13, 815718. | 2.6 | 2 |
| 5 | ExomeChip-based rare variant association study in restless legs syndrome. Sleep Medicine, 2022, 94, 26-30. | 1.6 | 0 |
| 6 | 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 |
| 7 | Biallelic variants in <scp>ZNF142</scp> lead to a syndromic neurodevelopmental disorder. Clinical Genetics, 2022, 102, 98-109. | 2.0 | 6 |
| 8 | Fast versus slow disease progression in amyotrophic lateral sclerosisâ€“clinical and genetic factors at the edges of the survival spectrum. Neurobiology of Aging, 2022, 119, 117-126. | 3.1 | 5 |
| 9 | Defining the genotypic and phenotypic spectrum of X-linked MSL3-related disorder. Genetics in Medicine, 2021, 23, 384-395. | 2.4 | 4 |
| 10 | De novo variants in neurodevelopmental disordersâ€”experiences from a tertiary care center. Clinical Genetics, 2021, 100, 14-28. | 2.0 | 64 |
| 11 | Clinically relevant copy-number variants in exome sequencing data of patients with dystonia. Parkinsonism and Related Disorders, 2021, 84, 129-134. | 2.2 | 15 |
| 12 | Dystonia as a prominent presenting feature in developmental and epileptic encephalopathies: A case series. Parkinsonism and Related Disorders, 2021, 90, 73-78. | 2.2 | 9 |
| 13 | Clinico-genetic findings in 509 frontotemporal dementia patients. Molecular Psychiatry, 2021, 26, 5824-5832. | 7.9 | 23 |
| 14 | Epigenome-wide association study of serum urate reveals insights into urate co-regulation and the SLC2A9 locus. Nature Communications, 2021, 12, 7173. | 12.8 | 8 |
| 15 | Meta-analyses identify DNA methylation associated with kidney function and damage. Nature Communications, 2021, 12, 7174. | 12.8 | 30 |
| 16 | Monogenic variants in dystonia: an exome-wide sequencing study. Lancet Neurology, The, 2020, 19, 908-918. | 10.2 | 139 |
| 17 | Severe paroxysmal dyskinesias without epilepsy in a RHOBTB2 mutation carrier. Parkinsonism and Related Disorders, 2020, 77, 87-88. | 2.2 | 9 |
| 18 | Recessive null-allele variants in MAG associated with spastic ataxia, nystagmus, neuropathy, and dystonia. Parkinsonism and Related Disorders, 2020, 77, 70-75. | 2.2 | 3 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | Bi-allelic variants in RNF170 are associated with hereditary spastic paraplegia. Nature Communications, 2019, 10, 4790. | 12.8 | 39 |
| 20 | Opposite microglial activation stages upon loss of <scp>PGRN</scp> or <scp>TREM</scp> 2 result in reduced cerebral glucose metabolism. EMBO Molecular Medicine, 2019, 11, . | 6.9 | 87 |
| 21 | Common Grounds for Family Maladies. Neuron, 2018, 98, 671-672. | 8.1 | 2 |
| 22 | 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 |
| 23 | Association of CLEC16A with human common variable immunodeficiency disorder and role in murine B cells. Nature Communications, 2015, 6, 6804. | 12.8 | 63 |
| 24 | Neuropeptide S Receptor Gene Variation Differentially Modulates Fronto-Limbic Effective Connectivity in Childhood and Adolescence. Cerebral Cortex, 2015, 27, bhv259. | 2.9 | 12 |
| 25 | Narcolepsy is a common phenotype in HSAN IE and ADCA-DN. Brain, 2014, 137, 1643-1655. | 7.6 | 49 |
| 26 | 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 |
| 27 | HLA DQB1*06:02 Negative Narcolepsy with Hypocretin/Orexin Deficiency. Sleep, 2014, 37, 1601-1608. | 1.1 | 59 |
| 28 | Mutations in DNMT1 cause autosomal dominant cerebellar ataxia, deafness and narcolepsy. Human Molecular Genetics, 2012, 21, 2205-2210. | 2.9 | 225 |
| 29 | Genome-Wide Association Study Identifies Novel Restless Legs Syndrome Susceptibility Loci on 2p14 and 16q12.1. PLoS Genetics, 2011, 7, e1002171. | 3.5 | 163 |