

Martin Krenn

List of Publications by Year in descending order

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Version: 2024-02-01

38
papers

618
citations

686830

13
h-index

676716

22
g-index

38
all docs

38
docs citations

38
times ranked

1349
citing authors

| # | ARTICLE | IF | CITATIONS |
|----|---|-----|-----------|
| 1 | Association of ultra-rare coding variants with genetic generalized epilepsy: A case-control whole exome sequencing study. <i>Epilepsia</i> , 2022, 63, 723-735. | 2.6 | 8 |
| 2 | A de novo truncating variant in CSDE1 in an adult-onset neuropsychiatric phenotype without intellectual disability. <i>European Journal of Medical Genetics</i> , 2022, 65, 104423. | 0.7 | 1 |
| 3 | A de novo missense variant in <i>GABRA4</i> alters receptor function in an epileptic and neurodevelopmental phenotype. <i>Epilepsia</i> , 2022, 63, . | 2.6 | 6 |
| 4 | Clonal hematopoiesis as a pitfall in germline variant interpretation in the context of Mendelian disorders. <i>Human Molecular Genetics</i> , 2022, 31, 2386-2395. | 1.4 | 7 |
| 5 | Clinico-genetic spectrum of limb-girdle muscular weakness in Austria: A multicentre cohort study. <i>European Journal of Neurology</i> , 2022, , . | 1.7 | 4 |
| 6 | Connectome Analysis in an Individual with SETD1B-Related Neurodevelopmental Disorder and Epilepsy. <i>Journal of Developmental and Behavioral Pediatrics</i> , 2022, 43, e419-e422. | 0.6 | 3 |
| 7 | Short-term and sustained clinical response following thymectomy in patients with myasthenia gravis. <i>European Journal of Neurology</i> , 2022, 29, 2453-2462. | 1.7 | 7 |
| 8 | Nerve conduction studies in Guillain-Barré syndrome: Influence of timing and value of repeated measurements. <i>Journal of the Neurological Sciences</i> , 2021, 420, 117267. | 0.3 | 13 |
| 9 | Cerebrospinal fluid analysis in Guillain-Barré syndrome: value of albumin quotients. <i>Journal of Neurology</i> , 2021, 268, 3294-3300. | 1.8 | 10 |
| 10 | Delineation of epileptic and neurodevelopmental phenotypes associated with variants in STX1B. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2021, 87, 25-29. | 0.9 | 6 |
| 11 | Assessing the role of rare genetic variants in drug-resistant, non-lesional focal epilepsy. <i>Annals of Clinical and Translational Neurology</i> , 2021, 8, 1376-1387. | 1.7 | 16 |
| 12 | Scoring Algorithm-Based Genomic Testing in Dystonia: A Prospective Validation Study. <i>Movement Disorders</i> , 2021, 36, 1959-1964. | 2.2 | 7 |
| 13 | Childhood-onset progressive dystonia associated with pathogenic truncating variants in <i>CHD8</i> . <i>Annals of Clinical and Translational Neurology</i> , 2021, 8, 1986-1990. | 1.7 | 5 |
| 14 | Real-world treatment of adult patients with Guillain-Barré syndrome over the last two decades. <i>Scientific Reports</i> , 2021, 11, 19170. | 1.6 | 6 |
| 15 | Estimation of patent foramen ovale size using transcranial Doppler ultrasound in patients with ischemic stroke. <i>Journal of Neuroimaging</i> , 2021, , . | 1.0 | 3 |
| 16 | Incidence and clinical spectrum of rhabdomyolysis in general neurology: a retrospective cohort study. <i>Neuromuscular Disorders</i> , 2021, , . | 0.3 | 1 |
| 17 | Retinal and Corneal Neurodegeneration and Their Association with Systemic Signs of Peripheral Neuropathy in Type 2 Diabetes. <i>American Journal of Ophthalmology</i> , 2020, 209, 197-205. | 1.7 | 23 |
| 18 | Genotype-guided diagnostic reassessment after exome sequencing in neuromuscular disorders: experiences with a two-step approach. <i>European Journal of Neurology</i> , 2020, 27, 51-61. | 1.7 | 21 |

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|----|--|-----|-----------|
| 19 | Frequency and clinical features of treatment-refractory myasthenia gravis. <i>Journal of Neurology</i> , 2020, 267, 1004-1011. | 1.8 | 22 |
| 20 | Monogenic variants in dystonia: an exome-wide sequencing study. <i>Lancet Neurology</i> , The, 2020, 19, 908-918. | 4.9 | 139 |
| 21 | Pathomechanisms and Clinical Implications of Myasthenic Syndromes Exacerbated and Induced by Medical Treatments. <i>Frontiers in Molecular Neuroscience</i> , 2020, 13, 156. | 1.4 | 16 |
| 22 | Pharmacoresponse in genetic generalized epilepsy: a genome-wide association study. <i>Pharmacogenomics</i> , 2020, 21, 325-335. | 0.6 | 21 |
| 23 | Testing association of rare genetic variants with resistance to three common antiseizure medications. <i>Epilepsia</i> , 2020, 61, 657-666. | 2.6 | 22 |
| 24 | Reply to Comment on: Retinal and Corneal Neurodegeneration and Its Association to Systemic Signs of Peripheral Neuropathy in Type 2 Diabetes. <i>American Journal of Ophthalmology</i> , 2020, 216, 287-288. | 1.7 | 0 |
| 25 | Diagnostic exome sequencing in non-acquired focal epilepsies highlights a major role of GATOR1 complex genes. <i>Journal of Medical Genetics</i> , 2020, 57, 624-633. | 1.5 | 16 |
| 26 | ATP1A3-related epilepsy: Report of seven cases and literature-based analysis of treatment response. <i>Journal of Clinical Neuroscience</i> , 2020, 72, 31-38. | 0.8 | 8 |
| 27 | Mutational and phenotypic expansion of ATP1A3-related disorders: Report of nine cases. <i>Gene</i> , 2020, 749, 144709. | 1.0 | 8 |
| 28 | The TGF β /SOX4 axis and ROS-driven autophagy co-mediate CD39 expression in regulatory T cells. <i>FASEB Journal</i> , 2020, 34, 8367-8384. | 0.2 | 28 |
| 29 | Adult-onset variant ataxia-telangiectasia diagnosed by exome and cDNA sequencing. <i>Neurology: Genetics</i> , 2019, 5, e346. | 0.9 | 4 |
| 30 | Phenotypic variability of <i>GABRA1</i> -related epilepsy in monozygotic twins. <i>Annals of Clinical and Translational Neurology</i> , 2019, 6, 2317-2322. | 1.7 | 4 |
| 31 | Rare Human Missense Variants can affect the Function of Disease-Relevant Proteins by Loss and Gain of Peroxisomal Targeting Motifs. <i>International Journal of Molecular Sciences</i> , 2019, 20, 4609. | 1.8 | 6 |
| 32 | ATP1A3-related Epilepsy: Report of Six Cases and Literature-based Analysis of Treatment Response. <i>Neuropediatrics</i> , 2019, 50, . | 0.3 | 0 |
| 33 | Mutations outside the N-terminal part of RBCK1 may cause polyglucosan body myopathy with immunological dysfunction: expanding the genotype-phenotype spectrum. <i>Journal of Neurology</i> , 2018, 265, 394-401. | 1.8 | 36 |
| 34 | Genetic variation in <i>CFH</i> predicts phenytoin-induced maculopapular exanthema in European-descent patients. <i>Neurology</i> , 2018, 90, e332-e341. | 1.5 | 43 |
| 35 | Rare coding variants in genes encoding GABAA receptors in genetic generalised epilepsies: an exome-based case-control study. <i>Lancet Neurology</i> , The, 2018, 17, 699-708. | 4.9 | 67 |
| 36 | Hereditary spastic paraplegia caused by compound heterozygous mutations outside the motor domain of the <i>KIF1A</i> gene. <i>European Journal of Neurology</i> , 2017, 24, 741-747. | 1.7 | 25 |

| # | ARTICLE | IF | CITATIONS |
|----|---|-----|-----------|
| 37 | CANOMAD responding to weekly treatment with intravenous immunoglobulin (IVIg). BMJ Case Reports, 2014, 2014, bcr2013202545-bcr2013202545. | 0.2 | 6 |
| 38 | The relationship between electrical stimulus and viscoelastic parameters of the denervated anterior thigh muscles. , 2009, , . | | 0 |