Martin Krenn

List of Publications by Year in descending order

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686830 676716 38 618 13 22 citations h-index g-index papers 38 38 38 1349 citing authors docs citations times ranked all docs

#	Article	IF	CITATIONS
1	Monogenic variants in dystonia: an exome-wide sequencing study. Lancet Neurology, The, 2020, 19, 908-918.	4.9	139
2	Rare coding variants in genes encoding GABAA receptors in genetic generalised epilepsies: an exome-based case-control study. Lancet Neurology, The, 2018, 17, 699-708.	4.9	67
3	Genetic variation in <i>CFH</i> predicts phenytoin-induced maculopapular exanthema in European-descent patients. Neurology, 2018, 90, e332-e341.	1.5	43
4	Mutations outside the N-terminal part of RBCK1 may cause polyglucosan body myopathy with immunological dysfunction: expanding the genotype–phenotype spectrum. Journal of Neurology, 2018, 265, 394-401.	1.8	36
5	The TGFâ€b/SOX4 axis and ROSâ€driven autophagy coâ€mediate CD39 expression in regulatory Tâ€cells. FASEB Journal, 2020, 34, 8367-8384.	0.2	28
6	Hereditary spastic paraplegia caused by compound heterozygous mutations outside the motor domain of the <i>KIF1A</i> gene. European Journal of Neurology, 2017, 24, 741-747.	1.7	25
7	Retinal and Corneal Neurodegeneration and Their Association with Systemic Signs of Peripheral Neuropathy in Type 2 Diabetes. American Journal of Ophthalmology, 2020, 209, 197-205.	1.7	23
8	Frequency and clinical features of treatment-refractory myasthenia gravis. Journal of Neurology, 2020, 267, 1004-1011.	1.8	22
9	Testing association of rare genetic variants with resistance to three common antiseizure medications. Epilepsia, 2020, 61, 657-666.	2.6	22
10	Genotypeâ€guided diagnostic reassessment after exome sequencing in neuromuscular disorders: experiences with a twoâ€step approach. European Journal of Neurology, 2020, 27, 51-61.	1.7	21
11	Pharmacoresponse in genetic generalized epilepsy: a genome-wide association study. Pharmacogenomics, 2020, 21, 325-335.	0.6	21
12	Pathomechanisms and Clinical Implications of Myasthenic Syndromes Exacerbated and Induced by Medical Treatments. Frontiers in Molecular Neuroscience, 2020, 13, 156.	1.4	16
13	Diagnostic exome sequencing in non-acquired focal epilepsies highlights a major role of GATOR1 complex genes. Journal of Medical Genetics, 2020, 57, 624-633.	1.5	16
14	Assessing the role of rare genetic variants in drugâ€resistant, nonâ€lesional focal epilepsy. Annals of Clinical and Translational Neurology, 2021, 8, 1376-1387.	1.7	16
15	Nerve conduction studies in Guillain-Barr \tilde{A} © syndrome: Influence of timing and value of repeated measurements. Journal of the Neurological Sciences, 2021, 420, 117267.	0.3	13
16	Cerebrospinal fluid analysis in Guillain–Barré syndrome: value of albumin quotients. Journal of Neurology, 2021, 268, 3294-3300.	1.8	10
17	ATP1A3-related epilepsy: Report of seven cases and literature-based analysis of treatment response. Journal of Clinical Neuroscience, 2020, 72, 31-38.	0.8	8
18	Mutational and phenotypic expansion of ATP1A3-related disorders: Report of nine cases. Gene, 2020, 749, 144709.	1.0	8

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19	Association of ultraâ€rare coding variants with genetic generalized epilepsy: A case–control whole exome sequencing study. Epilepsia, 2022, 63, 723-735.	2.6	8
20	Scoring Algorithmâ€Based Genomic Testing in Dystonia: A Prospective Validation Study. Movement Disorders, 2021, 36, 1959-1964.	2.2	7
21	Clonal hematopoiesis as a pitfall in germline variant interpretation in the context of Mendelian disorders. Human Molecular Genetics, 2022, 31, 2386-2395.	1.4	7
22	Shortâ€ŧerm and sustained clinical response following thymectomy in patients with myasthenia gravis. European Journal of Neurology, 2022, 29, 2453-2462.	1.7	7
23	Rare Human Missense Variants can affect the Function of Disease-Relevant Proteins by Loss and Gain of Peroxisomal Targeting Motifs. International Journal of Molecular Sciences, 2019, 20, 4609.	1.8	6
24	Delineation of epileptic and neurodevelopmental phenotypes associated with variants in STX1B. Seizure: the Journal of the British Epilepsy Association, 2021, 87, 25-29.	0.9	6
25	Real-world treatment of adult patients with Guillain-Barr \tilde{A} © syndrome over the last two decades. Scientific Reports, 2021, 11, 19170.	1.6	6
26	CANOMAD responding to weekly treatment with intravenous immunoglobulin (IVIg). BMJ Case Reports, 2014, 2014, bcr2013202545-bcr2013202545.	0.2	6
27	A de novo missense variant in <i>GABRA4</i> alters receptor function in an epileptic and neurodevelopmental phenotype. Epilepsia, 2022, 63, .	2.6	6
28	Childhoodâ€onset progressive dystonia associated with pathogenic truncating variants in <i>CHD8</i> Annals of Clinical and Translational Neurology, 2021, 8, 1986-1990.	1.7	5
29	Adult-onset variant ataxia-telangiectasia diagnosed by exome and cDNA sequencing. Neurology: Genetics, 2019, 5, e346.	0.9	4
30	Phenotypic variability of <i>GABRA1</i> â€related epilepsy in monozygotic twins. Annals of Clinical and Translational Neurology, 2019, 6, 2317-2322.	1.7	4
31	Clinicoâ€genetic spectrum of limbâ€girdle muscular weakness in Austria: A multicentre cohort study. European Journal of Neurology, 2022, , .	1.7	4
32	Estimation of patent foramen ovale size using transcranial Doppler ultrasound in patients with ischemic stroke. Journal of Neuroimaging, 2021, , .	1.0	3
33	Connectome Analysis in an Individual with SETD1B-Related Neurodevelopmental Disorder and Epilepsy. Journal of Developmental and Behavioral Pediatrics, 2022, 43, e419-e422.	0.6	3
34	Incidence and clinical spectrum of rhabdomyolysis in general neurology: a retrospective cohort study. Neuromuscular Disorders, 2021, , .	0.3	1
35	A de novo truncating variant in CSDE1 in an adult-onset neuropsychiatric phenotype without intellectual disability. European Journal of Medical Genetics, 2022, 65, 104423.	0.7	1
36	The relationship between electrical stimulus and viscoelastic parameters of the denervated anterior thigh muscles. , 2009, , .		0

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37	Reply to Comment on: Retinal and Corneal Neurodegeneration and Its Association to Systemic Signs of Peripheral Neuropathy in Type 2 Diabetes. American Journal of Ophthalmology, 2020, 216, 287-288.	1.7	0
38	ATP1A3-related Epilepsy: Report of Six Cases and Literature-based Analysis of Treatment Response. Neuropediatrics, 2019, 50, .	0.3	0