Alex R Paciorkowski

List of Publications by Year
in descending order

51 papers	2,038 citations	257450 24 h-index	254184 43 g-index
51 all docs	51 docs citations	51 times ranked	4088 citing authors

#	Article	IF	Citations
1	Mendelian etiologies identified with whole exome sequencing in cerebral palsy. Annals of Clinical and Translational Neurology, 2022, 9, 193-205.	3.7	23
2	A diagnostic confidence scheme for <scp>CLN3</scp> disease. Journal of Inherited Metabolic Disease, 2021, 44, 1453-1462.	3.6	3
3	Movement disorders in children with congenital Zika virus syndrome. Brain and Development, 2020, 42, 720-729.	1.1	12
4	<scp>Kinesigenic</scp> Triggers in Episodic Ataxia Type 1. Movement Disorders Clinical Practice, 2020, 7, 723-724.	1.5	1
5	Bi-allelic Loss-of-Function CACNA1B Mutations in Progressive Epilepsy-Dyskinesia. American Journal of Human Genetics, 2019, 104, 948-956.	6.2	45
6	BioVR: a platform for virtual reality assisted biological data integration and visualization. BMC Bioinformatics, 2019, 20, 78.	2.6	33
7	Association of Severe Hydrocephalus With Congenital Zika Syndrome. JAMA Neurology, 2019, 76, 203.	9.0	28
8	Phenotypes, genotypes, and the management of paroxysmal movement disorders. Developmental Medicine and Child Neurology, 2018, 60, 559-565.	2.1	31
9	Ode to the humble Southern blot in the era of exomes. Neurology: Clinical Practice, 2018, 8, 4-5.	1.6	0
10	Depletion of transglutaminase 2 in neurons alters expression of extracellular matrix and signal transduction genes and compromises cell viability. Molecular and Cellular Neurosciences, 2018, 86, 72-80.	2.2	13
11	Expanding the neurodevelopmental phenotype of PURA syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 56-67.	1.2	26
12	Nuclear transglutaminase 2 directly regulates expression of cathepsin S in rat cortical neurons. European Journal of Neuroscience, 2018, 48, 3043-3051.	2.6	7
13	Genetic Diagnostics for Neurologists. CONTINUUM Lifelong Learning in Neurology, 2018, 24, 18-36.	0.8	5
14	Genetics and genotypeâ€“phenotype correlations in early onset epileptic encephalopathy with burst suppression. Annals of Neurology, 2017, 81, 419-429.	5.3	107
15	Developing a novel epileptic discharge localization algorithm for electroencephalogram infantile spasms during hypsarrhythmia. Medical and Biological Engineering and Computing, 2017, 55, 1659-1668.	2.8	8
16	PLXNA1 developmental encephalopathy with syndromic features: A case report and review of the literature. American Journal of Medical Genetics, Part A, 2017, 173, 1951-1954.	1.2	7
17	Epilepsy-causing sequence variations in SIK1 disrupt synaptic activity response gene expression and affect neuronal morphology. European Journal of Human Genetics, 2017, 25, 216-221.	2.8	25
18	CEDNIK. Child Neurology Open, 2017, 4, 2329048X1773321.	1.1	16

#	Article	IF	Citations
19	India Allele Finder: a web-based annotation tool for identifying common alleles in next-generation sequencing data of Indian origin. BMC Research Notes, 2017, 10, 233.	1.4	4
20	Neuroimaging findings in Mowatâ€“Wilson syndrome: a study of 54 patients. Genetics in Medicine, 2017, 19, 691-700.	2.4	45
21	Congenital Zika syndrome: an epidemic of neurologic disability. Arquivos De Neuro-Psiquiatria, 2017, 75, 605-605.	0.8	0
22	Phenotype Differentiation of FOXG1 and MECP2 Disorders: A New Method for Characterization of Developmental Encephalopathies. Journal of Pediatrics, 2016, 178, 233-240.e10.	1.8	19
23	Familial recurrences of FOXG1â€related disorder: Evidence for mosaicism. American Journal of Medical Genetics, Part A, 2015, 167, 3096-3102.	1.2	15
24	Novel mutations in ATP1A3 associated with catastrophic early life epilepsy, episodic prolonged apnea, and postnatal microcephaly. Epilepsia, 2015, 56, 422-430.	5.1	107
25	De Novo Mutations in SIK1 Cause a Spectrum of Developmental Epilepsies. American Journal of Human Genetics, 2015, 96, 682-690.	6.2	48
26	Autism spectrum disorder and epilepsy: Disorders with a shared biology. Epilepsy and Behavior, 2015, 47, 191-201.	1.7	129
27	Epilepsy and outcome in FOXG1â€related disorders. Epilepsia, 2014, 55, 1292-1300.	5.1	55
28	De Novo Mutations in the Beta-Tubulin Gene TUBB2A Cause Simplified Gyral Patterning and Infantile-Onset Epilepsy. American Journal of Human Genetics, 2014, 94, 634-641.	6.2	99
29	The Developmental Brain Disorders Database (DBDB): A curated neurogenetics knowledge base with clinical and research applications. American Journal of Medical Genetics, Part A, 2014, 164, 1503-1511.	1.2	20
30	Genetic disorders associated with postnatal microcephaly. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2014, 166, 140-155.	1.6	70
31	Introduction: Brain malformations. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2014, 166, 117-123.	1.6	13
32	Autosomal recessive mutations in nuclear transport factor KPNA7 are associated with infantile spasms and cerebellar malformation. European Journal of Human Genetics, 2014, 22, 587-593.	2.8	25
33	Mutations in CENPE define a novel kinetochore-centromeric mechanism for microcephalic primordial dwarfism. Human Genetics, 2014, 133, 1023-1039.	3.8	82
34	An Integrative Computational Approach for Prioritization of Genomic Variants. PLoS ONE, 2014, 9, e114903.	2.5	7
35	Deletion 16p13.11 uncovers NDE1 mutations on the nonâ€deleted homolog and extends the spectrum of severe microcephaly to include fetal brain disruption. American Journal of Medical Genetics, Part A, 2013, 161, 1523-1530.	1.2	66
36	CDKL5 and ARX Mutations in Males With Early-Onset Epilepsy. Pediatric Neurology, 2013, 48, 367-377.	2.1	53

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37	MEF2C Haploinsufficiency features consistent hyperkinesis, variable epilepsy, and has a role in dorsal and ventral neuronal developmental pathways. Neurogenetics, 2013, 14, 99-111.	1.4	89
38	Mutations in STAMBP, encoding a deubiquitinating enzyme, cause microcephalyâ€“capillary malformation syndrome. Nature Genetics, 2013, 45, 556-562.	21.4	94
39	Chromosome 2p15p16.1 microdeletion syndrome: 2.5ÂMb deletion in a patient with renal anomalies, intractable seizures and a choledochal cyst. European Journal of Medical Genetics, 2012, 55, 485-489.	1.3	18
40	Balance impairment in individuals with Wolfram syndrome. Gait and Posture, 2012, 36, 619-624.	1.4	14
41	Early Brain Vulnerability in Wolfram Syndrome. PLoS ONE, 2012, 7, e40604.	2.5	77
42	Genotypeâ€“phenotype correlation in interstitial 6q deletions: a report of 12 new cases. Neurogenetics, 2012, 13, 31-47.	1.4	48
43	Recurrent deletions and reciprocal duplications of 10q11.21q11.23 including CHAT and SLC18A3 are likely mediated by complex low-copy repeats. Human Mutation, 2012, 33, 165-179.	2.5	45
44	Genetic and Biologic Classification of Infantile Spasms. Pediatric Neurology, 2011, 45, 355-367.	2.1	144
45	Duplications of FOXG1 in 14q12 are associated with developmental epilepsy, mental retardation, and severe speech impairment. European Journal of Human Genetics, 2011, 19, 102-107.	2.8	104
46	Copy number variants and infantile spasms: evidence for abnormalities in ventral forebrain development and pathways of synaptic function. European Journal of Human Genetics, 2011, 19, 1238-1245.	2.8	74
47	Chromosomal Microarray Interpretation: WhatÂIs a Child Neurologist to Do?. Pediatric Neurology, 2009, 41, 391-398.	2.1	24
48	Motion analysis of a child with Niemannâ€“Pick disease type C treated with miglustat. Movement Disorders, 2008, 23, 124-128.	3.9	18
49	Juvenile-Onset GM2-Gangliosidosis in an African-American Child With Nystagmus. Pediatric Neurology, 2008, 38, 284-286.	2.1	5
50	When Is Enlargement of the Subarachnoid Spaces Not Benign? A Genetic Perspective. Pediatric Neurology, 2007, 37, 1-7.	2.1	33
51	Structureâ€“function correlations in patients with malformations of cortical development. Epilepsy and Behavior, 2002, 3, 266-274.	1.7	4