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	Genetic and Biologic Classification of Infantile Spasms. Pediatric Neurology, 2011, 45, 355-367.	2.1	144
2	Autism spectrum disorder and epilepsy: Disorders with a shared biology. Epilepsy and Behavior, 2015, 47, 191-201.	1.7	129
3	Novel mutations in ATP1A3 associated with catastrophic early life epilepsy, episodic prolonged apnea, and postnatal microcephaly. Epilepsia, 2015, 56, 422-430.	5.1	107
4	Genetics and genotypeâ€“phenotype correlations in early onset epileptic encephalopathy with burst suppression. Annals of Neurology, 2017, 81, 419-429.	5.3	107
5	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
6	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
7	Mutations in STAMBP, encoding a deubiquitinating enzyme, cause microcephalyâ€“capillary malformation syndrome. Nature Genetics, 2013, 45, 556-562.	21.4	94
8	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
9	Mutations in CENPE define a novel kinetochore-centromeric mechanism for microcephalic primordial dwarfism. Human Genetics, 2014, 133, 1023-1039.	3.8	82
10	Early Brain Vulnerability in Wolfram Syndrome. PLoS ONE, 2012, 7, e40604.	2.5	77
11	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
12	Genetic disorders associated with postnatal microcephaly. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2014, 166, 140-155.	1.6	70
13	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
14	Epilepsy and outcome in FOXG1â€related disorders. Epilepsia, 2014, 55, 1292-1300.	5.1	55
15	CDKL5 and ARX Mutations in Males With Early-Onset Epilepsy. Pediatric Neurology, 2013, 48, 367-377.	2.1	53
16	Genotypeâ€“phenotype correlation in interstitial 6q deletions: a report of 12 new cases. Neurogenetics, 2012, 13, 31-47.	1.4	48
17	De Novo Mutations in SIK1 Cause a Spectrum of Developmental Epilepsies. American Journal of Human Genetics, 2015, 96, 682-690.	6.2	48
18	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

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19	Neuroimaging findings in Mowatâ€“Wilson syndrome: a study of 54 patients. Genetics in Medicine, 2017, 19, 691-700.	2.4	45
20	Bi-allelic Loss-of-Function CACNA1B Mutations in Progressive Epilepsy-Dyskinesia. American Journal of Human Genetics, 2019, 104, 948-956.	6.2	45
21	When Is Enlargement of the Subarachnoid Spaces Not Benign? A Genetic Perspective. Pediatric Neurology, 2007, 37, 1-7.	2.1	33
22	BioVR: a platform for virtual reality assisted biological data integration and visualization. BMC Bioinformatics, 2019, 20, 78.	2.6	33
23	Phenotypes, genotypes, and the management of paroxysmal movement disorders. Developmental Medicine and Child Neurology, 2018, 60, 559-565.	2.1	31
24	Association of Severe Hydrocephalus With Congenital Zika Syndrome. JAMA Neurology, 2019, 76, 203.	9.0	28
25	Expanding the neurodevelopmental phenotype of PURA syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 56-67.	1.2	26
26	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
27	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
28	Chromosomal Microarray Interpretation: WhatÂIs a Child Neurologist to Do?. Pediatric Neurology, 2009, 41, 391-398.	2.1	24
29	Mendelian etiologies identified with whole exome sequencing in cerebral palsy. Annals of Clinical and Translational Neurology, 2022, 9, 193-205.	3.7	23
30	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
31	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
32	Motion analysis of a child with Niemannâ€“Pick disease type C treated with miglustat. Movement Disorders, 2008, 23, 124-128.	3.9	18
33	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
34	CEDNIK. Child Neurology Open, 2017, 4, 2329048X1773321.	1.1	16
35	Familial recurrences of FOXG1â€related disorder: Evidence for mosaicism. American Journal of Medical Genetics, Part A, 2015, 167, 3096-3102.	1.2	15
36	Balance impairment in individuals with Wolfram syndrome. Gait and Posture, 2012, 36, 619-624.	1.4	14

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37	Introduction: Brain malformations. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2014, 166, 117-123.	1.6	13
38	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
39	Movement disorders in children with congenital Zika virus syndrome. Brain and Development, 2020, 42, 720-729.	1.1	12
40	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
41	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
42	Nuclear transglutaminase 2 directly regulates expression of cathepsin S in rat cortical neurons. European Journal of Neuroscience, 2018, 48, 3043-3051.	2.6	7
43	An Integrative Computational Approach for Prioritization of Genomic Variants. PLoS ONE, 2014, 9, e114903.	2.5	7
44	Juvenile-Onset GM2-Gangliosidosis in an African-American Child With Nystagmus. Pediatric Neurology, 2008, 38, 284-286.	2.1	5
45	Genetic Diagnostics for Neurologists. CONTINUUM Lifelong Learning in Neurology, 2018, 24, 18-36.	0.8	5
46	Structureâ€“function correlations in patients with malformations of cortical development. Epilepsy and Behavior, 2002, 3, 266-274.	1.7	4
47	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
48	A diagnostic confidence scheme for <scp>CLN3</scp> disease. Journal of Inherited Metabolic Disease, 2021, 44, 1453-1462.	3.6	3
49	<scp>Kinesigenic</scp> Triggers in Episodic Ataxia Type 1. Movement Disorders Clinical Practice, 2020, 7, 723-724.	1.5	1
50	Ode to the humble Southern blot in the era of exomes. Neurology: Clinical Practice, 2018, 8, 4-5.	1.6	0
51	Congenital Zika syndrome: an epidemic of neurologic disability. Arquivos De Neuro-Psiquiatria, 2017, 75, 605-605.	0.8	0