Amy L Schneider

List of Publications by Year
in descending order

36 papers	2,162 citations	304743 22 h-index	345221 36 g-index
36 all docs	36 docs citations	36 times ranked	4118 citing authors

#	Article	IF	Citations
1	Development and Validation of a Prediction Model for Early Diagnosis of SCN1A -Related Epilepsies. Neurology, 2022, 98, .	1.1	24
2	Germline variants in tumor suppressor FBXW7 lead to impaired ubiquitination and a neurodevelopmental syndrome. American Journal of Human Genetics, 2022, 109, 601-617.	6.2	16
3	NEXMIF encephalopathy: an X-linked disorder with male and female phenotypic patterns. Genetics in Medicine, 2021, 23, 363-373.	2.4	28
4	FBXO28 causes developmental and epileptic encephalopathy with profound intellectual disability. Epilepsia, 2021, 62, e13-e21.	5.1	8
5	The clinical utility of exome sequencing and extended bioinformatic analyses in adolescents and adults with a broad range of neurological phenotypes: an Australian perspective. Journal of the Neurological Sciences, 2021, 420, 117260.	0.6	16
6	Cerebrospinal fluid liquid biopsy for detecting somatic mosaicism in brain. Brain Communications, 2021, 3, fcaa235.	3.3	42
7	The severe epilepsy syndromes of infancy: A populationâ€based study. Epilepsia, 2021, 62, 358-370.	5.1	31
8	Contribution of rare genetic variants to drug response in absence epilepsy. Epilepsy Research, 2021, 170, 106537.	1.6	9
9	Pathogenic <scp>MAST3</scp> Variants in the <scp>STK</scp> Domain Are Associated with Epilepsy. Annals of Neurology, 2021, 90, 274-284.	5.3	7
10	Defining Dravet syndrome: An essential preâ€requisite for precision medicine trials. Epilepsia, 2021, 62, 2205-2217.	5.1	50
11	Somatic IDH1 variant (p.R132C) in an adult male with Maffucci syndrome. Journal of Physical Education and Sports Management, 2021, 7, mcs.a006127.	1.2	2
12	Expanding the genetic and phenotypic relevance of KCNB1 variants in developmental and epileptic encephalopathies: 27 new patients and overview of the literature. Human Mutation, 2020, 41, 69-80.	2.5	33
13	SCN1A Variants in vaccineâ€related febrile seizures: A prospective study. Annals of Neurology, 2020, 87, 281-288.	5.3	15
14	<scp>BRAT</scp>1 encephalopathy: a recessive cause of epilepsy of infancy with migrating focal seizures. Developmental Medicine and Child Neurology, 2020, 62, 1096-1099.	2.1	18
15	Germline and Mosaic Variants in PRKACA and PRKACB Cause a Multiple Congenital Malformation Syndrome. American Journal of Human Genetics, 2020, 107, 977-988.	6.2	33
16	Cognitive, behavioral, and social functioning in children and adults with Dravet syndrome. Epilepsy and Behavior, 2020, 112, 107319.	1.7	21
17	Developmental and epilepsy spectrum of KCNB1 encephalopathy with longâ€term outcome. Epilepsia, 2020, 61, 2461-2473.	5.1	17
18	Familial adult myoclonic epilepsy type 1 SAMD12 TTTCA repeat expansion arose 17,000 years ago and is present in Sri Lankan and Indian families. European Journal of Human Genetics, 2020, 28, 973-978.	2.8	23

#	Article	IF	Citations
19	AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. Nature Communications, 2019, 10, 3094.	12.8	150
20	Intronic ATTTC repeat expansions in STARD7 in familial adult myoclonic epilepsy linked to chromosome 2. Nature Communications, 2019, 10, 4920.	12.8	99
21	The Genetic Landscape of Epilepsy of Infancy with Migrating Focal Seizures. Annals of Neurology, 2019, 86, 821-831.	5.3	96
22	Double somatic mosaicism in a child with Dravet syndrome. Neurology: Genetics, 2019, 5, e333.	1.9	7
23	Perception of impact of Dravet syndrome on children and caregivers in multiple countries: looking beyond seizures. Developmental Medicine and Child Neurology, 2019, 61, 1229-1236.	2.1	39
24	Bi-allelic Loss-of-Function CACNA1B Mutations in Progressive Epilepsy-Dyskinesia. American Journal of Human Genetics, 2019, 104, 948-956.	6.2	45
25	The epilepsy phenotypic spectrum associated with a recurrent CUX2 variant. Annals of Neurology, 2018, 83, 926-934.	5.3	20
26	Severe infantile onset developmental and epileptic encephalopathy caused by mutations in autophagy gene <scp>WDR</scp>45. Epilepsia, 2018, 59, e5-e13.	5.1	44
27	Sleep problems in Dravet syndrome: a modifiable comorbidity. Developmental Medicine and Child Neurology, 2018, 60, 192-198.	2.1	45
28	Aberrant Inclusion of a Poison Exon Causes Dravet Syndrome and Related SCN1A-Associated Genetic Epilepsies. American Journal of Human Genetics, 2018, 103, 1022-1029.	6.2	76
29	A populationâ€based costâ€effectiveness study of early genetic testing in severe epilepsies of infancy. Epilepsia, 2018, 59, 1177-1187.	5.1	77
30	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2017, 101, 664-685.	6.2	337
31	Germline De Novo Mutations in GNB1 Cause Severe Neurodevelopmental Disability, Hypotonia, and Seizures. American Journal of Human Genetics, 2016, 98, 1001-1010.	6.2	102
32	Mortality in Dravet syndrome. Epilepsy Research, 2016, 128, 43-47.	1.6	218
33	Epileptic spasms are a feature of DEPDC5 mTORopathy. Neurology: Genetics, 2015, 1, e17.	1.9	63
34	CHD2variants are a risk factor for photosensitivity in epilepsy. Brain, 2015, 138, 1198-1208.	7.6	112
35	Seizures Are Regulated by Ubiquitin-specific Peptidase 9 X-linked (USP9X), a De-Ubiquitinase. PLoS Genetics, 2015, 11, e1005022.	3.5	66
36	Mutations in the GABA Transporter SLC6A1 Cause Epilepsy with Myoclonic-Atonic Seizures. American Journal of Human Genetics, 2015, 96, 808-815.	6.2	173