Kristin W Baranano

List of Publications by Year
in descending order

17	397	1040056 9	839539 18
papers	citations	h-index	g-index

18	18	18	1313
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	De novo DHDDS variants cause a neurodevelopmental and neurodegenerative disorder with myoclonus. Brain, 2022, 145, 208-223.	7.6	15
2	CNS Malformations in the Newborn. Maternal Health, Neonatology and Perinatology, 2022, 8, 1.	2.2	4
3	Further Characterization of SMC1A Loss of Function Epilepsy Distinct From Cornelia de Lange Syndrome. Journal of Child Neurology, 2022, 37, 390-396.	1.4	5
4	Acceleration and plateau: two patterns and outcomes of isolated severe fetal cerebral ventricular dilation. Journal of Maternal-Fetal and Neonatal Medicine, 2021, 34, 3014-3020.	1.5	1
5	Leukoencephalopathy with calcifications and cysts: Genetic and phenotypic spectrum. American Journal of Medical Genetics, Part A, 2021, 185, 15-25.	1.2	15
6	Novel variants in KAT6B spectrum of disorders expand our knowledge of clinical manifestations and molecular mechanisms. Molecular Genetics & Genomic Medicine, 2021, 9, e1809.	1.2	4
7	Two siblings with a novel variant of EXOSC3 extended phenotypic spectrum of pontocerebellar hypoplasia 1B to an exceptionally mild form. BMJ Case Reports, 2021, 14, e236732.	0.5	2
8	De Novo Variants in the ATPase Module of MORC2 Cause a Neurodevelopmental Disorder with Growth Retardation and Variable Craniofacial Dysmorphism. American Journal of Human Genetics, 2020, 107, 352-363.	6.2	64
9	De Novo and Bi-allelic Pathogenic Variants in NARS1 Cause Neurodevelopmental Delay Due to Toxic Gain-of-Function and Partial Loss-of-Function Effects. American Journal of Human Genetics, 2020, 107, 311-324.	6.2	32
10	Posterior Neocortex-Specific Regulation of Neuronal Migration by CEP85L Identifies Maternal Centriole-Dependent Activation of CDK5. Neuron, 2020, 106, 246-255.e6.	8.1	19
11	Relapsingâ€“remitting clinical course expands the phenotype of Aicardiâ€“GoutiÃ¨res syndrome. Annals of Clinical and Translational Neurology, 2020, 7, 254-258.	3.7	2
12	Bi-allelic Variants in the GPI Transamidase Subunit PIGK Cause a Neurodevelopmental Syndrome with Hypotonia, Cerebellar Atrophy, and Epilepsy. American Journal of Human Genetics, 2020, 106, 484-495.	6.2	22
13	Intracranial calcifications and dystonia associated with a novel deletion of chromosome 8p11.2 encompassing SLC20A2 and THAP1. BMJ Case Reports, 2019, 12, e228782.	0.5	7
14	De Novo Mutations of RERE Cause a Genetic Syndrome with Features that Overlap Those Associated with Proximal 1p36 Deletions. American Journal of Human Genetics, 2016, 98, 963-970.	6.2	67
15	SCN8A Epileptic Encephalopathy: Detection of Fetal Seizures Guides Multidisciplinary Approach to Diagnosis and Treatment. Pediatric Neurology, 2016, 64, 87-91.	2.1	13
16	Mutations in SNORD118 cause the cerebral microangiopathy leukoencephalopathy with calcifications and cysts. Nature Genetics, 2016, 48, 1185-1192.	21.4	114
17	Novel KIF7 missense substitutions in two patients presenting with multiple malformations and features of acrocallosal syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 2767-2776.	1.2	9