

Kristin W Baranano

List of Publications by Year in descending order

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Version: 2024-02-01

17
papers

397
citations

1040056

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839539

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docs citations

18
times ranked

1313
citing authors

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