

Weiyi Mu

List of Publications by Year in descending order

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Version: 2024-02-01

21
papers

555
citations

932766

10
h-index

713013

21
g-index

21
all docs

21
docs citations

21
times ranked

1327
citing authors

#	ARTICLE	IF	CITATIONS
1	Bioinformatics-Based Identification of Expanded Repeats: A Non-reference Intronic Pentamer Expansion in RFC1 Causes CANVAS. <i>American Journal of Human Genetics</i> , 2019, 105, 151-165.	2.6	170
2	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.	2.6	64
3	Genetic testing for the epilepsies: A systematic review. <i>Epilepsia</i> , 2022, 63, 375-387.	2.6	53
4	Genotype-phenotype correlation of congenital anomalies in multiple congenital anomalies hypotonia seizures syndrome (MCAHS1)/PIGN-related epilepsy. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 77-86.	0.7	41
5	De novo missense variants in PPP1CB are associated with intellectual disability and congenital heart disease. <i>Human Genetics</i> , 2016, 135, 1399-1409.	1.8	40
6	Factors affecting quality of life in children and adolescents with hypermobile Ehlers-Danlos syndrome/hypermobility spectrum disorders. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 561-569.	0.7	39
7	Linkage-specific deubiquitylation by OTUD5 defines an embryonic pathway intolerant to genomic variation. <i>Science Advances</i> , 2021, 7, .	4.7	25
8	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.	2.6	22
9	Expansion of the clinical spectrum associated with AARS2-related disorders. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 1556-1564.	0.7	20
10	Characterization of pulmonary arteriovenous malformations in ACVRL1 versus ENG mutation carriers in hereditary hemorrhagic telangiectasia. <i>Genetics in Medicine</i> , 2018, 20, 639-644.	1.1	14
11	Pain and sleep quality in children with non-vascular Ehlers-Danlos syndromes. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1858-1864.	0.7	12
12	The utility of whole exome sequencing in diagnosing neurological disorders in adults from a highly consanguineous population. <i>Journal of Neurogenetics</i> , 2019, 33, 21-26.	0.6	10
13	A structured genetics rotation for pediatric residents: an important educational opportunity. <i>Genetics in Medicine</i> , 2020, 22, 793-796.	1.1	9
14	An atypical 0.73 MB microduplication of 22q11.21 and a novel SALL4 missense mutation associated with thumb agenesis and radioulnar synostosis. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 1644-1649.	0.7	8
15	Eighteen-year-old man with autism, obsessive compulsive disorder and a SHANK2 variant presents with severe anorexia that responds to high-dose fluoxetine. <i>BMJ Case Reports</i> , 2018, 2018, bcr-2018-225119.	0.2	7
16	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.	0.2	7
17	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.	0.6	4
18	Incontinentia Pigmenti with Persistent Hypercalcemia: Case Report. <i>Pediatric Dermatology</i> , 2016, 33, e315-7.	0.5	3

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19	Adult patients with undiagnosed conditions and their responses to unresolved uncertainty from exome sequencing. <i>Journal of Genetic Counseling</i> , 2020, 29, 992-1003.	0.9	3
20	Relapsingâ€“remitting clinical course expands the phenotype of Aicardiâ€“GoutiÃˆres syndrome. <i>Annals of Clinical and Translational Neurology</i> , 2020, 7, 254-258.	1.7	2
21	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.	0.2	2