Alyssa L Ritter

List of Publications by Year
in descending order

25 papers	413 citations	933447 10 h-index	839539 18 g-index
25 all docs	25 docs citations	25 times ranked	944 citing authors

#	Article	IF	Citations
1	Recessive DNAH9 Loss-of-Function Mutations Cause Laterality Defects and Subtle Respiratory Ciliary-Beating Defects. American Journal of Human Genetics, 2018, 103, 995-1008.	6.2	92
2	Expanding the genotypic and phenotypic spectrum in a diverse cohort of 104 individuals with Wiedemannâ€Steiner syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 1649-1665.	1.2	34
3	A Syndromic Neurodevelopmental Disorder Caused by Mutations in SMARCD1, a Core SWI/SNF Subunit Needed for Context-Dependent Neuronal Gene Regulation in Flies. American Journal of Human Genetics, 2019, 104, 596-610.	6.2	32
4	Truncating SRCAP variants outside the Floating-Harbor syndrome locus cause a distinct neurodevelopmental disorder with a specific DNA methylation signature. American Journal of Human Genetics, 2021, 108, 1053-1068.	6.2	31
5	Disruption of cardiac thin filament assembly arising from a mutation in LMOD2 : A novel mechanism of neonatal dilated cardiomyopathy. Science Advances, 2019, 5, eaax2066.	10.3	29
6	Further delineation of the clinical spectrum of KAT6B disorders and allelic series of pathogenic variants. Genetics in Medicine, 2020, 22, 1338-1347.	2.4	25
7	Variable Clinical Manifestations of Xiaâ€Gibbs syndrome: Findings of Consecutively Identified Cases at a Single Children's Hospital. American Journal of Medical Genetics, Part A, 2018, 176, 1890-1896.	1.2	20
8	Clinical utility of exome sequencing in infantile heart failure. Genetics in Medicine, 2020, 22, 423-426.	2.4	17
9	EP300 â€related Rubinsteinâ€“Taybi syndrome: Highlighted rare phenotypic findings and a genotypeâ€“phenotype metaâ€analysis of 74 patients. American Journal of Medical Genetics, Part A, 2020, 182, 2926-2938.	1.2	16
10	DOCK3â€related neurodevelopmental syndrome: Biallelic intragenic deletion of DOCK3 in a boy with developmental delay and hypotonia. American Journal of Medical Genetics, Part A, 2018, 176, 241-245.	1.2	14
11	The variability of <scp>SMARCA4</scp>â€related <scp>Coffinâ€“Siris</scp> syndrome: Do nonsense candidate variants add to milder phenotypes?. American Journal of Medical Genetics, Part A, 2020, 182, 2058-2067.	1.2	14
12	Natural history of aortic root dilation through young adulthood in a hypermobile Ehlersâ€“Danlos syndrome cohort. American Journal of Medical Genetics, Part A, 2017, 173, 1467-1472.	1.2	12
13	Missense Mutations in NKAP Cause a Disorder of Transcriptional Regulation Characterized by Marfanoid Habitus and Cognitive Impairment. American Journal of Human Genetics, 2019, 105, 987-995.	6.2	11
14	Variants in <scp>NAA15</scp> cause pediatric hypertrophic cardiomyopathy. American Journal of Medical Genetics, Part A, 2021, 185, 228-233.	1.2	10
15	Consolidation of the clinical and genetic definition of aSOX4-related neurodevelopmental syndrome. Journal of Medical Genetics, 2022, 59, 1058-1068.	3.2	10
16	Genetic variant burden and adverse outcomes in pediatric cardiomyopathy. Pediatric Research, 2021, 89, 1470-1476.	2.3	9
17	The clinical and molecular spectrum of QRICH1 associated neurodevelopmental disorder. Human Mutation, 2022, 43, 266-282.	2.5	7
18	<scp>MYH7</scp> variants cause complex congenital heart disease. American Journal of Medical Genetics, Part A, 2022, 188, 2772-2776.	1.2	7

#	Article	IF	Citations
19	Interstitial 4q Deletion Syndrome Including <i>NR3C2</i> Causing Pseudohypoaldosteronism. Molecular Syndromology, 2019, 10, 327-331.	0.8	6
20	Genotypeâ€“phenotype association by echocardiography offers incremental value in patients with Noonan Syndrome with Multiple Lentigines. Pediatric Research, 2021, 90, 444-451.	2.3	6
21	Expanding the phenotypic spectrum of ARCN1-related syndrome. Genetics in Medicine, 2022, 24, 1227-1237.	2.4	5
22	NKX2â€6 related congenital heart disease: Biallelic homeodomainâ€disrupting variants and truncus arteriosus. American Journal of Medical Genetics, Part A, 2020, 182, 1454-1459.	1.2	3
23	Fetal cardiomyopathy in neurofibromatosis type I: Novel phenotype and review of the literature. American Journal of Medical Genetics, Part A, 2019, 179, 1042-1046.	1.2	2
24	Mosaic <scp>RAI1</scp> variant in a <scp>Smithâ€“Magenis</scp> syndrome patient with total anomalous pulmonary venous return. American Journal of Medical Genetics, Part A, 2022, 188, 3130-3134.	1.2	1
25	Fine-Tuning 3-Methylglutaconic Aciduria Cutoffs for a Patient with Infantile-Onset Barth Syndrome. Clinical Chemistry, 2022, 68, 365-367.	3.2	0