

Alyssa L Ritter

List of Publications by Year in descending order

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Version: 2024-02-01

25
papers

413
citations

933447

10
h-index

839539

18
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25
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25
docs citations

25
times ranked

944
citing authors

#	ARTICLE	IF	CITATIONS
1	Fine-Tuning 3-Methylglutaconic Aciduria Cutoffs for a Patient with Infantile-Onset Barth Syndrome. <i>Clinical Chemistry</i> , 2022, 68, 365-367.	3.2	0
2	The clinical and molecular spectrum of <i>QRICH1</i> associated neurodevelopmental disorder. <i>Human Mutation</i> , 2022, 43, 266-282.	2.5	7
3	Expanding the phenotypic spectrum of <i>ARCNI</i> -related syndrome. <i>Genetics in Medicine</i> , 2022, 24, 1227-1237.	2.4	5
4	Consolidation of the clinical and genetic definition of a <i>SOX4</i> -related neurodevelopmental syndrome. <i>Journal of Medical Genetics</i> , 2022, 59, 1058-1068.	3.2	10
5	<i>MYH7</i> variants cause complex congenital heart disease. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 2772-2776.	1.2	7
6	Mosaic <i>RAI1</i> variant in a Smith-Magenis syndrome patient with total anomalous pulmonary venous return. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 3130-3134.	1.2	1
7	Genotype-phenotype association by echocardiography offers incremental value in patients with Noonan Syndrome with Multiple Lentigines. <i>Pediatric Research</i> , 2021, 90, 444-451.	2.3	6
8	Genetic variant burden and adverse outcomes in pediatric cardiomyopathy. <i>Pediatric Research</i> , 2021, 89, 1470-1476.	2.3	9
9	Variants in <i>NAA15</i> cause pediatric hypertrophic cardiomyopathy. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 228-233.	1.2	10
10	Expanding the genotypic and phenotypic spectrum in a diverse cohort of 104 individuals with Wiedemann-Steiner syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 1649-1665.	1.2	34
11	Truncating <i>SRCAP</i> variants outside the Floating-Harbor syndrome locus cause a distinct neurodevelopmental disorder with a specific DNA methylation signature. <i>American Journal of Human Genetics</i> , 2021, 108, 1053-1068.	6.2	31
12	Clinical utility of exome sequencing in infantile heart failure. <i>Genetics in Medicine</i> , 2020, 22, 423-426.	2.4	17
13	<i>EP300</i> -related Rubinstein-Taybi syndrome: Highlighted rare phenotypic findings and a genotype-phenotype meta-analysis of 74 patients. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 2926-2938.	1.2	16
14	The variability of <i>SMARCA4</i> -related Coffin-Siris syndrome: Do nonsense candidate variants add to milder phenotypes?. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 2058-2067.	1.2	14
15	Further delineation of the clinical spectrum of <i>KAT6B</i> disorders and allelic series of pathogenic variants. <i>Genetics in Medicine</i> , 2020, 22, 1338-1347.	2.4	25
16	<i>NKX2-6</i> related congenital heart disease: Biallelic homeodomain-disrupting variants and truncus arteriosus. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 1454-1459.	1.2	3
17	Missense Mutations in <i>NKAP</i> Cause a Disorder of Transcriptional Regulation Characterized by Marfanoid Habitus and Cognitive Impairment. <i>American Journal of Human Genetics</i> , 2019, 105, 987-995.	6.2	11
18	Disruption of cardiac thin filament assembly arising from a mutation in <i>LMOD2</i> : A novel mechanism of neonatal dilated cardiomyopathy. <i>Science Advances</i> , 2019, 5, eaax2066.	10.3	29

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19	Fetal cardiomyopathy in neurofibromatosis type I: Novel phenotype and review of the literature. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 1042-1046.	1.2	2
20	A Syndromic Neurodevelopmental Disorder Caused by Mutations in SMARCD1, a Core SWI/SNF Subunit Needed for Context-Dependent Neuronal Gene Regulation in Flies. <i>American Journal of Human Genetics</i> , 2019, 104, 596-610.	6.2	32
21	Interstitial 4q Deletion Syndrome Including <i>NR3C2</i> ; Causing Pseudohypoaldosteronism. <i>Molecular Syndromology</i> , 2019, 10, 327-331.	0.8	6
22	<i>DOCK3</i> -related neurodevelopmental syndrome: Biallelic intragenic deletion of <i>DOCK3</i> in a boy with developmental delay and hypotonia. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 241-245.	1.2	14
23	Recessive DNAH9 Loss-of-Function Mutations Cause Laterality Defects and Subtle Respiratory Ciliary-Beating Defects. <i>American Journal of Human Genetics</i> , 2018, 103, 995-1008.	6.2	92
24	Variable Clinical Manifestations of Xia-Gibbs syndrome: Findings of Consecutively Identified Cases at a Single Children's Hospital. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1890-1896.	1.2	20
25	Natural history of aortic root dilation through young adulthood in a hypermobile Ehlers-Danlos syndrome cohort. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 1467-1472.	1.2	12