Jenny M Thies

List of Publications by Year
in descending order

16 papers	472 citations	1163117 8 h-index	940533 16 g-index
17 all docs	17 docs citations	17 times ranked	1127 citing authors

#	Article	IF	Citations
1	Expanding the genotype and phenotype spectrum of SYT1-associated neurodevelopmental disorder. Genetics in Medicine, 2022, 24, 880-893.	2.4	14
2	Tutorial: Triheptanoin and Nutrition Management for Treatment of Longâ€Chain Fatty Acid Oxidation Disorders. Journal of Parenteral and Enteral Nutrition, 2021, 45, 230-238.	2.6	9
3	Immune dysfunction in MGAT2â€CDG : A clinical report and review of the literature. American Journal of Medical Genetics, Part A, 2021, 185, 213-218.	1.2	5
4	<scp>CHRNB1</scp>â€associated congenital myasthenia syndrome: Expanding the clinical spectrum. American Journal of Medical Genetics, Part A, 2021, 185, 827-835.	1.2	6
5	Quantitative analysis of the natural history of prolidase deficiency: description of 17 families and systematic review of published cases. Genetics in Medicine, 2021, 23, 1604-1615.	2.4	10
6	Biallelic variants in KARS1 are associated with neurodevelopmental disorders and hearing loss recapitulated by the knockout zebrafish. Genetics in Medicine, 2021, 23, 1933-1943.	2.4	11
7	Delineating the molecular and phenotypic spectrum of the SETD1B-related syndrome. Genetics in Medicine, 2021, 23, 2122-2137.	2.4	16
8	Targeted long-read sequencing identifies missing disease-causing variation. American Journal of Human Genetics, 2021, 108, 1436-1449.	6.2	105
9	The Impact of Rapid Exome Sequencing on Medical Management of Critically Ill Children. Journal of Pediatrics, 2020, 226, 202-212.e1.	1.8	35
10	The coâ€occurrence of Wilson disease and Xâ€linked agammaglobulinemia in one family highlights the promising diagnostic potential of proteolytic analysis. Molecular Genetics & Genomic Medicine, 2020, 8, e1172.	1.2	3
11	Expansion of the Primrose syndrome phenotype through the comparative analysis of two new case reports with ZBTB20 variants. American Journal of Medical Genetics, Part A, 2019, 179, 2228-2232.	1.2	4
12	Rapid clinical exome sequencing in a pediatric ICU: Genetic counselor impacts and challenges. Journal of Genetic Counseling, 2019, 28, 283-291.	1.6	17
13	TANGO2: expanding the clinical phenotype and spectrum of pathogenic variants. Genetics in Medicine, 2019, 21, 601-607.	2.4	41
14	COQ2 nephropathy: a treatable cause of nephrotic syndrome in children. Pediatric Nephrology, 2018, 33, 1257-1261.	1.7	30
15	Expanding clinical phenotype in CACNA1C related disorders: From neonatal onset severe epileptic encephalopathy to lateâ€onset epilepsy. American Journal of Medical Genetics, Part A, 2018, 176, 2733-2739.	1.2	30
16	De Novo Mutations in Protein Kinase Genes CAMK2A and CAMK2B Cause Intellectual Disability. American Journal of Human Genetics, 2017, 101, 768-788.	6.2	136