Amy Kritzer

List of Publications by Year
in descending order

13 papers	138 citations	1478505 6 h-index	1372567 10 g-index
13 all docs	13 docs citations	13 times ranked	199 citing authors

#	Article	IF	Citations
1	MYT1L-associated neurodevelopmental disorder: description of 40 new cases and literature review of clinical and molecular aspects. Human Genetics, 2022, 141, 65-80.	3.8	14
2	Standardizing genetic and metabolic consults for non-accidental trauma at a large pediatric academic center. Child Abuse and Neglect, 2022, 125, 105480.	2.6	0
3	Pathogenic SPTBN1 variants cause an autosomal dominant neurodevelopmental syndrome. Nature Genetics, 2021, 53, 1006-1021.	21.4	44
4	Unique variants in CLCN3, encoding an endosomal anion/proton exchanger, underlie a spectrum of neurodevelopmental disorders. American Journal of Human Genetics, 2021, 108, 1450-1465.	6.2	16
5	Quality improvement: The tools we need to improve care for patients with inborn errors of metabolism. Molecular Genetics and Metabolism, 2020, 129, 1-2.	1.1	6
6	Use of skimmed breast milk for an infant with a <scp>longâ€chain</scp> fatty acid oxidation disorder: A novel therapeutic intervention. JIMD Reports, 2020, 55, 44-50.	1.5	4
7	A phenotypically severe, biochemically â€œsilentâ€•case of HIBCH deficiency in a newborn diagnosed by rapid whole exome sequencing and enzymatic testing. American Journal of Medical Genetics, Part A, 2020, 182, 780-784.	1.2	4
8	Discontinuation of Pegvaliase therapy during maternal PKU pregnancy and postnatal breastfeeding: A case report. Molecular Genetics and Metabolism Reports, 2020, 22, 100555.	1.1	7
9	Early initiation of enzyme replacement therapy in classical Fabry disease normalizes biomarkers in clinically asymptomatic pediatric patients. Molecular Genetics and Metabolism Reports, 2019, 21, 100530.	1.1	11
10	Acute Pancreatitis in a Patient with Maple Syrup Urine Disease: A Management Paradox. Journal of Pediatrics, 2018, 198, 313-316.	1.8	0
11	Cerebellar cognitive affective syndrome: insights from Joubert syndrome. Cerebellum and Ataxias, 2018, 5, 5.	1.9	20
12	Megaloblastic Anemia Progressing to Severe Thrombotic Microangiopathy in Patients with Disordered Vitamin B12 Metabolism: Case Reports and Literature Review. Journal of Pediatrics, 2018, 202, 315-319.e2.	1.8	5
13	Reconciling newborn screening and a novel splice variant in BTD associated with partial biotinidase deficiency: a BabySeq Project case report. Journal of Physical Education and Sports Management, 2018, 4, a002873.	1.2	7