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	Pathogenic SPTBN1 variants cause an autosomal dominant neurodevelopmental syndrome. Nature Genetics, 2021, 53, 1006-1021.	21.4	44
2	Cerebellar cognitive affective syndrome: insights from Joubert syndrome. Cerebellum and Ataxias, 2018, 5, 5.	1.9	20
3	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
4	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
5	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
6	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
7	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
8	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
9	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
10	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
11	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
12	Acute Pancreatitis in a Patient with Maple Syrup Urine Disease: A Management Paradox. Journal of Pediatrics, 2018, 198, 313-316.	1.8	0
13	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