Kaustuv Bhattacharya

List of Publications by Year
in descending order

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32	883	567281 15	501196 28
papers	citations	h-index	g-index

33	33	33	1632
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	FGF21 outperforms GDF15 as a diagnostic biomarker of mitochondrial disease in children. Molecular Genetics and Metabolism, 2022, 135, 63-71.	1.1	12
2	A serendipitous journey to a promoter variant: The c.â€‹<scp>106C</scp>>A variant and its role in lateâ€‹onset ornithine transcarbamylase deficiency. JIMD Reports, 2022, 63, 271-275.	1.5	3
3	Fatal Perinatal Mitochondrial Cardiac Failure Caused by Recurrent De Novo Duplications in the ATAD3 Locus. Med, 2021, 2, 49-73.e10.	4.4	33
4	Investigating paediatric hypoglycaemia: Dynamic studies at a tertiary paediatric hospital. Journal of Paediatrics and Child Health, 2021, 57, 888-893.	0.8	1
5	Purpuric, delayed child: Beyond septicaemia and into inborn errors of metabolism. Journal of Paediatrics and Child Health, 2021, 57, 1703-1706.	0.8	0
6	New insights into carnitineâ€‹acylcarnitine translocase deficiency from 23 cases: Management challenges and potential therapeutic approaches. Journal of Inherited Metabolic Disease, 2021, 44, 903-915.	3.6	8
7	Rare diseases research and policy in Australia: On the journey to equitable care. Journal of Paediatrics and Child Health, 2021, 57, 778-781.	0.8	10
8	Liver transplantation in children with inborn errors of metabolism: 30â€‰years experience in <scp>NSW</scp>, Australia. JIMD Reports, 2021, 60, 88-95.	1.5	8
9	Effectiveness of early hematopoietic stem cell transplantation in preventing neurocognitive decline in aspartylglucosaminuria: A case series. JIMD Reports, 2021, 61, 3-11.	1.5	2
10	GLRX5-associated [Fe-S] cluster biogenesis disorder: further characterisation of the neurological phenotype and long-term outcome. Orphanet Journal of Rare Diseases, 2021, 16, 465.	2.7	2
11	Efficacy and safety of D,L-3-hydroxybutyrate (D,L-3-HB) treatment in multiple acyl-CoA dehydrogenase deficiency. Genetics in Medicine, 2020, 22, 908-916.	2.4	19
12	Magnetic resonance imaging pattern recognition in childhood bilateral basal ganglia disorders. Brain Communications, 2020, 2, fcaa178.	3.3	17
13	The use of sodium DL-3-Hydroxybutyrate in severe acute neuro-metabolic compromise in patients with inherited ketone body synthetic disorders. Orphanet Journal of Rare Diseases, 2020, 15, 53.	2.7	10
14	Mutations in MAGT1 lead to a glycosylation disorder with a variable phenotype. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 9865-9870.	7.1	66
15	International best practice for the evaluation of responsiveness to sapropterin dihydrochloride in patients with phenylketonuria. Molecular Genetics and Metabolism, 2019, 127, 1-11.	1.1	44
16	Infantileâ€‹onset Pompe disease: A case series highlighting early clinical features, spectrum of disease severity and treatment response. Journal of Paediatrics and Child Health, 2018, 54, 1255-1261.	0.8	16
17	Compound heterozygous mutations in glycyl-tRNA synthetase (GARS) cause mitochondrial respiratory chain dysfunction. PLoS ONE, 2017, 12, e0178125.	2.5	16
18	Clinical course of sly syndrome (mucopolysaccharidosis type VII). Journal of Medical Genetics, 2016, 53, 403-418.	3.2	133

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19	Neurophysiological profile of peripheral neuropathy associated with childhood mitochondrial disease. Mitochondrion, 2016, 30, 162-167.	3.4	13
20	Dietary Management of the Ketogenic Glycogen Storage Diseases. FIRE Forum for International Research in Education, 2016, 4, 232640981666135.	0.7	8
21	Rare Case of Hepatic Gaucheroma in a Child on Enzyme Replacement Therapy. JIMD Reports, 2016, 32, 101-104.	1.5	8
22	Identifying the need for a multidisciplinary approach for early recognition of mucopolysaccharidosis VI (MPS VI). Molecular Genetics and Metabolism, 2015, 115, 41-47.	1.1	9
23	A pilot longitudinal study of the use of waxy maize heat modified starch in the treatment of adults with glycogen storage disease type I: a randomized double-blind cross-over study. Orphanet Journal of Rare Diseases, 2015, 10, 18.	2.7	19
24	CRIM-negative infantile Pompe disease: characterization of immune responses in patients treated with ERT monotherapy. Genetics in Medicine, 2015, 17, 912-918.	2.4	54
25	Investigation and management of the hepatic glycogen storage diseases. Translational Pediatrics, 2015, 4, 240-8.	1.2	37
26	Where will genetic research take us?. Translational Pediatrics, 2015, 4, 318-9.	1.2	0
27	Variant non ketotic hyperglycinemia is caused by mutations in LIAS, BOLA3 and the novel gene GLRX5. Brain, 2014, 137, 366-379.	7.6	187
28	Overcoming the barriers to diagnosis of Morquio A syndrome. Orphanet Journal of Rare Diseases, 2014, 9, 192.	2.7	21
29	The evolution of blood-spot newborn screening. Translational Pediatrics, 2014, 3, 63-70.	1.2	20
30	Dietary dilemmas in the management of glycogen storage disease type I. Journal of Inherited Metabolic Disease, 2011, 34, 621-629.	3.6	36
31	Use of modified cornstarch therapy to extend fasting in glycogen storage disease types Ia and Ib. American Journal of Clinical Nutrition, 2008, 88, 1272-6.	4.7	71
32	Safety and Efficacy of Elosulfase Alfa in Australian Patients with Morquio a Syndrome: A Phase 3b Study. Journal of Inborn Errors of Metabolism and Screening, 0, 8, .	0.3	0