Kaustuv Bhattacharya

List of Publications by Year
in descending order

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Version: 2024-02-01

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	Variant non ketotic hyperglycinemia is caused by mutations in LIAS, BOLA3 and the novel gene GLRX5. Brain, 2014, 137, 366-379.	7.6	187
2	Clinical course of sly syndrome (mucopolysaccharidosis type VII). Journal of Medical Genetics, 2016, 53, 403-418.	3.2	133
3	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
4	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
5	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
6	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
7	Investigation and management of the hepatic glycogen storage diseases. Translational Pediatrics, 2015, 4, 240-8.	1.2	37
8	Dietary dilemmas in the management of glycogen storage disease type I. Journal of Inherited Metabolic Disease, 2011, 34, 621-629.	3.6	36
9	Fatal Perinatal Mitochondrial Cardiac Failure Caused by Recurrent De Novo Duplications in the ATAD3 Locus. Med, 2021, 2, 49-73.e10.	4.4	33
10	Overcoming the barriers to diagnosis of Morquio A syndrome. Orphanet Journal of Rare Diseases, 2014, 9, 192.	2.7	21
11	The evolution of blood-spot newborn screening. Translational Pediatrics, 2014, 3, 63-70.	1.2	20
12	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
13	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
14	Magnetic resonance imaging pattern recognition in childhood bilateral basal ganglia disorders. Brain Communications, 2020, 2, fcaa178.	3.3	17
15	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
16	Compound heterozygous mutations in glycyl-tRNA synthetase (GARS) cause mitochondrial respiratory chain dysfunction. PLoS ONE, 2017, 12, e0178125.	2.5	16
17	Neurophysiological profile of peripheral neuropathy associated with childhood mitochondrial disease. Mitochondrion, 2016, 30, 162-167.	3.4	13
18	FGF21 outperforms GDF15 as a diagnostic biomarker of mitochondrial disease in children. Molecular Genetics and Metabolism, 2022, 135, 63-71.	1.1	12

#	Article	IF	Citations
19	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
20	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
21	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
22	Dietary Management of the Ketogenic Glycogen Storage Diseases. FIRE Forum for International Research in Education, 2016, 4, 232640981666135.	0.7	8
23	Rare Case of Hepatic Gaucheroma in a Child on Enzyme Replacement Therapy. JIMD Reports, 2016, 32, 101-104.	1.5	8
24	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
25	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
26	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
27	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
28	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
29	Investigating paediatric hypoglycaemia: Dynamic studies at a tertiary paediatric hospital. Journal of Paediatrics and Child Health, 2021, 57, 888-893.	0.8	1
30	Purpuric, delayed child: Beyond septicaemia and into inborn errors of metabolism. Journal of Paediatrics and Child Health, 2021, 57, 1703-1706.	0.8	0
31	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
32	Where will genetic research take us?. Translational Pediatrics, 2015, 4, 318-9.	1.2	0