Berna Seker Yilmaz

List of Publications by Year
in descending order

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25 papers	189 citations	1478505 6 h-index	1125743 13 g-index
28 all docs	28 docs citations	28 times ranked	262 citing authors

#	Article	IF	Citations
1	Evaluation of bone health in patients with mucopolysaccharidosis. Journal of Bone and Mineral Metabolism, 2022, , 1.	2.7	1
2	Novel therapies for mucopolysaccharidosis type <scp>III</scp>. Journal of Inherited Metabolic Disease, 2021, 44, 129-147.	3.6	31
3	The outcome of 41 Late-Diagnosed Turkish GA-1 Patients: A Candidate for the Turkish NBS. Neuropediatrics, 2021, 52, 358-369.	0.6	4
4	Mucopolysaccharidosis Type-II with Pathognomonic Skin Appearance: A Case with Pebbling Sign. Journal of Pediatric Research, 2021, 8, 206-208.	0.2	0
5	Clinical and molecular findings in 37 Turkish patients with isolated methylmalonic acidemia. Turkish Journal of Medical Sciences, 2021, 51, 1220-1228.	0.9	1
6	Safety and efficacy of an engineered hepatotropic AAV gene therapy for ornithine transcarbamylase deficiency in cynomolgus monkeys. Molecular Therapy - Methods and Clinical Development, 2021, 23, 135-146.	4.1	21
7	Niemannâ€“Pick type C disease with a novel intronic mutation: three Turkish cases from the same family. Journal of Pediatric Endocrinology and Metabolism, 2021, .	0.9	2
8	Clinical and Molecular Features of Early Infantile Niemann Pick Type C Disease. International Journal of Molecular Sciences, 2020, 21, 5059.	4.1	21
9	Gene therapy for inherited metabolic diseases. Medycyna Wieku Rozwojowego, 2020, 24, 53-64.	0.2	6
10	GP49â€...Munchausen by proxy syndrome in three siblings diagnosed as isovaleric acidemia. , 2019, , .		0
11	P433â€...An interesting case diagnosed as both phenylketonuria and maternal phenylketonuria. , 2019, , .		0
12	Twenty-seven mutations with three novel pathologenic variants causing biotinidase deficiency: a report of 203 patients from the southeastern part of Turkey. Journal of Pediatric Endocrinology and Metabolism, 2018, 31, 339-343.	0.9	12
13	Early onset alpha-mannosidosis: A Turkish case. Molecular Genetics and Metabolism, 2018, 123, S100-S101.	1.1	0
14	Hypercarotenemia. Cukurova Medical Journal, 2018, 43, 500-501.	0.2	0
15	An ignored cause of red urine in children: rhabdomyolysis due to carnitine palmitoyltransferase II (CPT-II) deficiency. Journal of Pediatric Endocrinology and Metabolism, 2017, 30, 237-239.	0.9	3
16	Improved metabolic control in tetrahydrobiopterin (BH4), responsive phenylketonuria with sapropterin administered in two divided doses vs. a single daily dose. Journal of Pediatric Endocrinology and Metabolism, 2017, 30, 713-718.	0.9	6
17	Homozygous familial hypobetalipoproteinemia: A Turkish case carrying a missense mutation in apolipoprotein B. Clinica Chimica Acta, 2016, 452, 185-190.	1.1	11
18	A 17-Year-Old Girl with Chronic Intermittent Abdominal Pain. Pediatric Annals, 2015, 44, 139-141.	0.8	3

#	Article	IF	Citations
19	Two Novel Missense Mutations in Nonketotic Hyperglycinemia. Journal of Child Neurology, 2015, 30, 789-792.	1.4	5
20	An asymptomatic mother diagnosed with 3-methylcrotonyl-CoA carboxylase deficiency after newborn screening. Journal of Pediatric Endocrinology and Metabolism, 2015, 28, 669-71.	0.9	5
21	Primary systemic carnitine deficiency: a Turkish case with a novel homozygous SLC22A5 mutation and 14 years follow-up. Journal of Pediatric Endocrinology and Metabolism, 2015, 28, 1179-81.	0.9	5
22	Predictors of Intractable Childhood Epilepsy. Pediatric Neurology, 2013, 48, 52-55.	2.1	39
23	Symptomatic and asymptomatic candidiasis in a pediatric intensive care unit. Italian Journal of Pediatrics, 2011, 37, 56.	2.6	9
24	Demographic, Phenotypic and Genotypic Features of Alkaptonuria Patients: A Single Centre Experience. Journal of Pediatric Research, 0, , 7-11.	0.2	4
25	A 6-Month-Old Boy with Reddish, Scaly Skin: Netherton Syndrome. Journal of Pediatric Research, 0, , 54-56.	0.2	0