GÃ¶knur HaliloÄlu

List of Publications by Year
in descending order

18 papers	591 citations	840776 11 h-index	839539 18 g-index
19 all docs	19 docs citations	19 times ranked	1225 citing authors

#	Article	IF	Citations
1	New-Onset Ocular Myasthenia after Multisystem Inflammatory Syndrome in Children. Journal of Pediatrics, 2022, 245, 213-216.	1.8	3
2	Neonatal presentations of neuromuscular disorders. European Journal of Paediatric Neurology, 2022, 38, A6-A11.	1.6	2
3	Further expanding the mutational spectrum of <scp>brain abnormalities, neurodegeneration, and dysosteosclerosis</scp>: A rare disorder with neurologic regression and skeletal features. American Journal of Medical Genetics, Part A, 2021, 185, 1888-1896.	1.2	14
4	Guidelines for the diagnosis and management of methylmalonic acidaemia and propionic acidaemia: FirstÂrevision. Journal of Inherited Metabolic Disease, 2021, 44, 566-592.	3.6	118
5	Selenoprotein Nâ€related myopathy: a retrospective natural history study to guide clinical trials. Annals of Clinical and Translational Neurology, 2020, 7, 2288-2296.	3.7	18
6	Determinants of Riboflavin Responsiveness in Multiple Acyl-CoA Dehydrogenase Deficiency. Pediatric Neurology, 2019, 99, 69-75.	2.1	22
7	Phenotypes and malignancy risk of different FUS mutations in genetic amyotrophic lateral sclerosis. Annals of Clinical and Translational Neurology, 2019, 6, 2384-2394.	3.7	49
8	Diagnostic Pathway to Nonsense Mutation Dystrophinopathy: A Tertiary-Center, Retrospective Experience. Neuropediatrics, 2019, 50, 041-045.	0.6	4
9	Expanding the phenotypic spectrum associated with mutations of DYNC1H1. Neuromuscular Disorders, 2017, 27, 607-615.	0.6	29
10	Niemann-Pick disease type C in the newborn period: a single-center experience. European Journal of Pediatrics, 2017, 176, 1669-1676.	2.7	18
11	Diagnosis of Duchenne muscular dystrophy in Italy in the last decade: Critical issues and areas for improvements. Neuromuscular Disorders, 2017, 27, 973.	0.6	4
12	Next generation sequencing in a large cohort of patients presenting with neuromuscular disease before or at birth. Orphanet Journal of Rare Diseases, 2015, 10, 148.	2.7	94
13	The Role of Electrocardiography in the Diagnosis of Spinal MuscularÂAtrophy Type III. Journal of Pediatrics, 2015, 166, 1092.	1.8	3
14	Clinical characteristics of megaconial congenital muscular dystrophy due to choline kinase beta gene defects in a series of 15 patients. Journal of Inherited Metabolic Disease, 2015, 38, 1099-1108.	3.6	24
15	Wernicke Encephalopathy Due to Thiamine Deficiency After Surgery on a Child With Duodenal Stenosis. Pediatric Neurology, 2014, 51, 840-842.	2.1	7
16	Successful treatment of cataplexy in patients with early-infantile Niemannâ€“Pick disease type C: Use of tricyclic antidepressants. European Journal of Paediatric Neurology, 2014, 18, 811-815.	1.6	10
17	SPEG Interacts with Myotubularin, and Its Deficiency Causes Centronuclear Myopathy with Dilated Cardiomyopathy. American Journal of Human Genetics, 2014, 95, 218-226.	6.2	143
18	Arthrogryposis and fetal hypomobility syndrome. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2013, 113, 1311-1319.	1.8	29