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	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
2	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
3	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
4	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
5	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
6	Expanding the phenotypic spectrum associated with mutations of DYNC1H1. Neuromuscular Disorders, 2017, 27, 607-615.	0.6	29
7	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
8	Determinants of Riboflavin Responsiveness in Multiple Acyl-CoA Dehydrogenase Deficiency. Pediatric Neurology, 2019, 99, 69-75.	2.1	22
9	Niemann-Pick disease type C in the newborn period: a single-center experience. European Journal of Pediatrics, 2017, 176, 1669-1676.	2.7	18
10	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
11	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
12	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
13	Wernicke Encephalopathy Due to Thiamine Deficiency After Surgery on a Child With Duodenal Stenosis. Pediatric Neurology, 2014, 51, 840-842.	2.1	7
14	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
15	Diagnostic Pathway to Nonsense Mutation Dystrophinopathy: A Tertiary-Center, Retrospective Experience. Neuropediatrics, 2019, 50, 041-045.	0.6	4
16	The Role of Electrocardiography in the Diagnosis of Spinal MuscularÂAtrophy Type III. Journal of Pediatrics, 2015, 166, 1092.	1.8	3
17	New-Onset Ocular Myasthenia after Multisystem Inflammatory Syndrome in Children. Journal of Pediatrics, 2022, 245, 213-216.	1.8	3
18	Neonatal presentations of neuromuscular disorders. European Journal of Paediatric Neurology, 2022, 38, A6-A11.	1.6	2