Mert Karakaya

List of Publications by Year
in descending order

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29 papers	559 citations	687363 13 h-index	677142 22 g-index
31 all docs	31 docs citations	31 times ranked	1068 citing authors

#	Article	IF	Citations
1	Novel variants broaden the phenotypic spectrum of PLEKHG5 â€associated neuropathies. European Journal of Neurology, 2021, 28, 1344-1355.	3.3	4
2	An ancestral 10-bp repeat expansion in VWA1 causes recessive hereditary motor neuropathy. Brain, 2021, 144, 584-600.	7.6	20
3	De novo DNM1L variant presenting with severe muscular atrophy, dystonia and sensory neuropathy. European Journal of Medical Genetics, 2021, 64, 104134.	1.3	9
4	Genomic variants causing mitochondrial dysfunction are common in hereditary lower motor neuron disease. Human Mutation, 2021, 42, 460-472.	2.5	6
5	The genomic and clinical landscape of fetal akinesia. Genetics in Medicine, 2020, 22, 511-523.	2.4	35
6	Clinical, electrophysiological and genetic characteristics of childhood hereditary polyneuropathies. Revue Neurologique, 2020, 176, 846-855.	1.5	4
7	De Novo and Inherited Variants in GBF1 are Associated with Axonal Neuropathy Caused by Golgi Fragmentation. American Journal of Human Genetics, 2020, 107, 763-777.	6.2	14
8	Hereditary polyneuropathy with optic atrophy due to PDXK variant leading to impaired Vitamin B6 metabolism. Neuromuscular Disorders, 2020, 30, 583-589.	0.6	7
9	Twenty-Five Years of Spinal Muscular Atrophy Research: From Phenotype to Genotype to Therapy, and What Comes Next. Annual Review of Genomics and Human Genetics, 2020, 21, 231-261.	6.2	134
10	214th ENMC International Workshop: Establishing an international consortium for gene discovery and clinical research for Congenital Muscle Disease, Heemskerk, the Netherlands, 6â€“18 October 2015. Neuromuscular Disorders, 2019, 29, 644-650.	0.6	2
11	Novel mutations in SLC6A5 with benign course in hyperekplexia. Journal of Physical Education and Sports Management, 2019, 5, a004465.	1.2	10
12	Report of a novel ATP7A mutation causing distal motor neuropathy. Neuromuscular Disorders, 2019, 29, 776-785.	0.6	15
13	Hereditary nodo-paranodopathies: genomic variants, not just autoantibodies, hit the protein. Brain, 2019, 142, 2895-2897.	7.6	0
14	Biallelic variant in AGTPBP1 causes infantile lower motor neuron degeneration and cerebellar atrophy. American Journal of Medical Genetics, Part A, 2019, 179, 1580-1584.	1.2	29
15	Giant axonal neuropathy: a differential diagnosis of consideration. Turkish Journal of Pediatrics, 2019, 61, 275.	0.6	4
16	Biallelic CHP1 mutation causes human autosomal recessive ataxia by impairing NHE1 function. Neurology: Genetics, 2018, 4, e209.	1.9	23
17	Late diagnosis of a truncating WISP3 mutation entails a severe phenotype of progressive pseudorheumatoid dysplasia. Journal of Physical Education and Sports Management, 2018, 4, a002139.	1.2	6
18	Biallelic Mutations in ADPRHL2, Encoding ADP-Ribosylhydrolase 3, Lead to a Degenerative Pediatric Stress-Induced Epileptic Ataxia Syndrome. American Journal of Human Genetics, 2018, 103, 431-439.	6.2	62

#	Article	IF	Citations
19	Targeted sequencing with expanded gene profile enables high diagnostic yield in non-5q-spinal muscular atrophies. Human Mutation, 2018, 39, 1284-1298.	2.5	42
20	Biallelic MCM3AP mutations cause Charcot-Marie-Tooth neuropathy with variable clinical presentation. Brain, 2017, 140, e65-e65.	7.6	13
21	A Novel Missense Variant in the AGRN Gene; Congenital Myasthenic Syndrome Presenting With Head Drop. Journal of Clinical Neuromuscular Disease, 2017, 18, 147-151.	0.7	26
22	P385â€…Hypotonic infant with riboflavin transporter deficiency due to slc52a2 mutations. , 2017, , .		0
23	PRUNE1: a disease-causing gene for secondary microcephaly. Brain, 2017, 140, e61-e61.	7.6	10
24	Choline transporter mutations in severe congenital myasthenic syndrome disrupt transporter localization. Brain, 2017, 140, 2838-2850.	7.6	24
25	Occipital cortex dysgenesis with white matter changes due to mutations in Laminin a2. Turkish Journal of Pediatrics, 2017, 59, 338-341.	0.6	1
26	Importance of Skin Changes in the Differential Diagnosis of Congenital Muscular Dystrophies. BioMed Research International, 2016, 2016, 1-3.	1.9	7
27	Novel Mutations in the Nonselective Sodium Leak Channel (NALCN) Lead to Distal Arthrogryposis with Increased Muscle Tone. Neuropediatrics, 2016, 47, 273-277.	0.6	18
28	Mendeliome sequencing enables differential diagnosis and treatment of neonatal lactic acidosis. Molecular and Cellular Pediatrics, 2016, 3, 22.	1.8	12
29	Whole Exome Sequencing Reveals DYSF, FKTN, and ISPD Mutations in Congenital Muscular Dystrophy Without Brain or Eye Involvement. Journal of Neuromuscular Diseases, 2015, 2, 87-92.	2.6	13