Dagmara KabziÅ„ska

List of Publications by Year
in descending order

19	279	1040056 9	940533 16
papers	citations	h-index	g-index

19	19	19	576
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Truncating and Missense Mutations in IGHMBP2 Cause Charcot-Marie Tooth Disease Type 2. American Journal of Human Genetics, 2014, 95, 590-601.	6.2	75
2	Characterization of HNRNPA1 mutations defines diversity in pathogenic mechanisms and clinical presentation. JCI Insight, 2021, 6, .	5.0	38
3	The genetic landscape of axonal neuropathies in the middle-aged and elderly. Neurology, 2020, 95, e3163-e3179.	1.1	19
4	Lateâ€onset Charcotâ€Marieâ€Tooth type 2 disease with hearing impairment associated with a novel Pro105Thr mutation in the MPZ gene. American Journal of Medical Genetics, Part A, 2007, 143A, 2196-2199.	1.2	16
5	L239F founder mutation in GDAP1 is associated with a mild Charcotâ€“Marieâ€“Tooth type 4C4 (CMT4C4) phenotype. Neurogenetics, 2010, 11, 357-366.	1.4	15
6	A new missense GDAP1 mutation disturbing targeting to the mitochondrial membrane causes a severe form of AR-CMT2C disease. Neurogenetics, 2011, 12, 145-153.	1.4	15
7	The Frequency of c.550delA Mutation of the CANP3 Gene in the Polish LGMD2A Population. Genetic Testing and Molecular Biomarkers, 2015, 19, 637-640.	0.7	14
8	Early-Onset Facioscapulohumeral Muscular Dystrophy Type 1 With Some Atypical Features. Journal of Child Neurology, 2015, 30, 580-587.	1.4	14
9	A novel TPM2 gene splice-site mutation causes severe congenital myopathy with arthrogryposis and dysmorphic features. Journal of Applied Genetics, 2017, 58, 199-203.	1.9	13
10	Pathogenic Effect of GDAP1 Gene Mutations in a Yeast Model. Genes, 2020, 11, 310.	2.4	11
11	A Yeast-Based Model for Hereditary Motor and Sensory Neuropathies: A Simple System for Complex, Heterogeneous Diseases. International Journal of Molecular Sciences, 2020, 21, 4277.	4.1	9
12	A novel Met116Thr mutation in the GDAP1 gene in a Polish family with the axonal recessive Charcotâ€“Marieâ€“Tooth type 4 disease. Journal of the Neurological Sciences, 2006, 241, 7-11.	0.6	8
13	Mutations in GDAP1 Influence Structure and Function of the Trans-Golgi Network. International Journal of Molecular Sciences, 2021, 22, 914.	4.1	8
14	Is a novel I214M substitution in the NEFL gene a cause of Charcot-Marie-Tooth disease? Functional analysis using cell culture models. Journal of the Peripheral Nervous System, 2006, 11, 225-231.	3.1	7
15	Two pathogenic mutations located within the 5'-regulatory sequence of the GJB1 gene affecting initiation of transcription and translation. Acta Biochimica Polonica, 2011, 58, 359-63.	0.5	5
16	A severe recessive and a mild dominant form of Charcot-Marie-Tooth disease associated with a newly identified Glu222Lys GDAP1 gene mutation. Acta Biochimica Polonica, 2014, 61, 739-44.	0.5	5
17	Molecular pathogenesis, experimental therapy and genetic counseling in hereditary sensory neuropathies. Acta Neurobiologiae Experimentalis, 2015, 75, 126-43.	0.7	5
18	A late-onset and mild form of Charcot-Marie-Tooth disease type 2 caused by a novel splice-site mutation within the Mitofusin-2 gene. Acta Myologica, 2013, 32, 166-9.	1.5	2

#	Article	IF	Citations
19	Phylogenetic Relatedness within the Internally Brooding Sea Anemones from the Arctic-Boreal Region. Biology, 2021, 10, 81.	2.8	0