Ibrahim Mahjneh

List of Publications by Year
in descending order

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19	1,986	567281 15	839539 18
papers	citations	h-index	g-index

20	20	20	2620
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Dominant Distal Myopathy 3 (MPD3) Caused by a Deletion in the HNRNPA1 Gene. Neurology: Genetics, 2021, 7, e632.	1.9	7
2	Dysregulated calcium homeostasis prevents plasma membrane repair in Anoctamin 5/TMEM16E-deficient patient muscle cells. Cell Death Discovery, 2019, 5, 118.	4.7	28
3	Decreased Aerobic Capacity inÂANO5-Muscular Dystrophy. Journal of Neuromuscular Diseases, 2016, 3, 475-485.	2.6	7
4	DOK7 limb-girdle myasthenic syndrome mimicking congenital muscular dystrophy. Neuromuscular Disorders, 2013, 23, 36-42.	0.6	23
5	Mutations affecting the cytoplasmic functions of the co-chaperone DNAJB6 cause limb-girdle muscular dystrophy. Nature Genetics, 2012, 44, 450-455.	21.4	226
6	Selective pattern of muscle involvement seen in distal muscular dystrophy associated with anoctamin 5 mutations: A follow-up muscle MRI study. Neuromuscular Disorders, 2012, 22, S130-S136.	0.6	27
7	Four new Finnish families with LGMD1D; refinement of the clinical phenotype and the linked 7q36 locus. Neuromuscular Disorders, 2011, 21, 338-344.	0.6	22
8	Recessive Mutations in the Putative Calcium-Activated Chloride Channel Anoctamin 5 Cause Proximal LGMD2L and Distal MMD3 Muscular Dystrophies. American Journal of Human Genetics, 2010, 86, 213-221.	6.2	245
9	A new distal myopathy with mutation in anoctamin 5. Neuromuscular Disorders, 2010, 20, 791-795.	0.6	55
10	Patients with a Non-dysferlin Miyoshi Myopathy have a Novel Membrane Repair Defect. Traffic, 2007, 8, 77-88.	2.7	56
11	Human Balance Estimation using a Wireless 3D Acceleration Sensor Network. Annual International Conference of the IEEE Engineering in Medicine and Biology Society, 2006, , .	0.5	0
12	The gene disrupted in Marinesco-SjÃ¶gren syndrome encodes SIL1, an HSPA5 cochaperone. Nature Genetics, 2005, 37, 1309-1311.	21.4	204
13	Muscle computed tomography patterns in patients with the mitochondrial DNA mutation 3243A>G. Journal of Neurology, 2004, 251, 556-563.	3.6	13
14	Linkage to two separate loci in a family with a novel distal myopathy phenotype (MPD3). Neuromuscular Disorders, 2004, 14, 183-187.	0.6	17
15	Axial myopathy - an unrecognised entity. Journal of Neurology, 2002, 249, 730-734.	3.6	47
16	Dysferlinopathy (LGMD2B): a 23-year follow-up study of 10 patients homozygous for the same frameshifting dysferlin mutations. Neuromuscular Disorders, 2001, 11, 20-26.	0.6	73
17	Secondary reduction in calpain 3 expression in patients with limb girdle muscular dystrophy type 2B and Miyoshi myopathy (primary dysferlinopathies). Neuromuscular Disorders, 2000, 10, 553-559.	0.6	138
18	A gene related to Caenorhabditis elegans spermatogenesis factor fer-1 is mutated in limb-girdle muscular dystrophy type 2B. Nature Genetics, 1998, 20, 37-42.	21.4	626

#	Article	IF	Citations
19	A gene for autosomal recessive limb-girdle muscular dystrophy maps to chromosome 2p. Human Molecular Genetics, 1994, 3, 455-457.	2.9	172