

Ibrahim Mahjneh

List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/2755351/publications.pdf>

Version: 2024-02-01

19
papers

1,986
citations

567281

15
h-index

839539

18
g-index

20
all docs

20
docs citations

20
times ranked

2620
citing authors

#	ARTICLE	IF	CITATIONS
1	A gene related to <i>Caenorhabditis elegans</i> spermatogenesis factor <i>fer-1</i> is mutated in limb-girdle muscular dystrophy type 2B. <i>Nature Genetics</i> , 1998, 20, 37-42.	21.4	626
2	Recessive Mutations in the Putative Calcium-Activated Chloride Channel Anoctamin 5 Cause Proximal LGMD2L and Distal MMD3 Muscular Dystrophies. <i>American Journal of Human Genetics</i> , 2010, 86, 213-221.	6.2	245
3	Mutations affecting the cytoplasmic functions of the co-chaperone DNAJB6 cause limb-girdle muscular dystrophy. <i>Nature Genetics</i> , 2012, 44, 450-455.	21.4	226
4	The gene disrupted in Marinesco-Sjögren syndrome encodes SIL1, an HSPA5 cochaperone. <i>Nature Genetics</i> , 2005, 37, 1309-1311.	21.4	204
5	A gene for autosomal recessive limb-girdle muscular dystrophy maps to chromosome 2p. <i>Human Molecular Genetics</i> , 1994, 3, 455-457.	2.9	172
6	Secondary reduction in calpain 3 expression in patients with limb girdle muscular dystrophy type 2B and Miyoshi myopathy (primary dysferlinopathies). <i>Neuromuscular Disorders</i> , 2000, 10, 553-559.	0.6	138
7	Dysferlinopathy (LGMD2B): a 23-year follow-up study of 10 patients homozygous for the same frameshifting dysferlin mutations. <i>Neuromuscular Disorders</i> , 2001, 11, 20-26.	0.6	73
8	Patients with a Non-dysferlin Miyoshi Myopathy have a Novel Membrane Repair Defect. <i>Traffic</i> , 2007, 8, 77-88.	2.7	56
9	A new distal myopathy with mutation in anoctamin 5. <i>Neuromuscular Disorders</i> , 2010, 20, 791-795.	0.6	55
10	Axial myopathy - an unrecognised entity. <i>Journal of Neurology</i> , 2002, 249, 730-734.	3.6	47
11	Dysregulated calcium homeostasis prevents plasma membrane repair in Anoctamin 5/TMEM16E-deficient patient muscle cells. <i>Cell Death Discovery</i> , 2019, 5, 118.	4.7	28
12	Selective pattern of muscle involvement seen in distal muscular dystrophy associated with anoctamin 5 mutations: A follow-up muscle MRI study. <i>Neuromuscular Disorders</i> , 2012, 22, S130-S136.	0.6	27
13	DOK7 limb-girdle myasthenic syndrome mimicking congenital muscular dystrophy. <i>Neuromuscular Disorders</i> , 2013, 23, 36-42.	0.6	23
14	Four new Finnish families with LGMD1D; refinement of the clinical phenotype and the linked 7q36 locus. <i>Neuromuscular Disorders</i> , 2011, 21, 338-344.	0.6	22
15	Linkage to two separate loci in a family with a novel distal myopathy phenotype (MPD3). <i>Neuromuscular Disorders</i> , 2004, 14, 183-187.	0.6	17
16	Muscle computed tomography patterns in patients with the mitochondrial DNA mutation 3243A>G. <i>Journal of Neurology</i> , 2004, 251, 556-563.	3.6	13
17	Decreased Aerobic Capacity in ANO5-Muscular Dystrophy. <i>Journal of Neuromuscular Diseases</i> , 2016, 3, 475-485.	2.6	7
18	Dominant Distal Myopathy 3 (MPD3) Caused by a Deletion in the <i>HNRNPA1</i> Gene. <i>Neurology: Genetics</i> , 2021, 7, e632.	1.9	7

#	ARTICLE	IF	CITATIONS
19	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