Rabi Tawil

List of Publications by Year
in descending order

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85 papers	5,739 citations	117571 34 h-index	85498 71 g-index
91 all docs	91 docs citations	91 times ranked	3326 citing authors

#	Article	IF	Citations
1	Chromosome 10q-linked FSHD identifies DUX4 as principal disease gene. Journal of Medical Genetics, 2022, 59, 180-188.	1.5	18
2	High-resolution breakpoint junction mapping of proximally extended D4Z4 deletions in FSHD1 reveals evidence for a founder effect. Human Molecular Genetics, 2022, 31, 748-760.	1.4	8
3	Cytosolic adaptation to mitochondria-induced proteostatic stress causes progressive muscle wasting. IScience, 2022, 25, 103715.	1.9	6
4	Reply: Wheelchair use in genetically-confirmed FSHD1 from a large cohort study in Chinese population. Brain, 2022, , .	3.7	0
5	Effect of Different Corticosteroid Dosing Regimens on Clinical Outcomes in Boys With Duchenne Muscular Dystrophy. JAMA - Journal of the American Medical Association, 2022, 327, 1456.	3.8	43
6	Elevated plasma complement components in facioscapulohumeral dystrophy. Human Molecular Genetics, 2022, 31, 1821-1829.	1.4	10
7	Quantitative Muscle Analysis in Facioscapulohumeral Muscular Dystrophy Using <scp>Wholeâ€Body Fatâ€Referenced MRI</scp> : Protocol Development, Multicenter Feasibility, and Repeatability. Muscle and Nerve, 2022, , .	1.0	1
8	Understanding the Perseverance of the Muscular Dystrophy Community One-Year into the COVID-19 Pandemic. Journal of Neuromuscular Diseases, 2022, 9, 517-523.	1.1	4
9	Mexiletine in Myotonic Dystrophy Type 1. Neurology, 2021, 96, e228-e240.	1.5	27
10	Relationship of DUX4 and target gene expression in FSHD myocytes. Human Mutation, 2021, 42, 421-433.	1.1	9
11	A Roadmap to Patient Engagement. Neurology: Clinical Practice, 2021, 11, e722-e726.	0.8	1
12	Current Therapeutic Approaches in FSHD. Journal of Neuromuscular Diseases, 2021, 8, 441-451.	1.1	18
13	The facioscapulohumeral muscular dystrophy Raschâ€built overall disability scale (FSHDâ€RODS). European Journal of Neurology, 2021, 28, 2339-2348.	1.7	8
14	A patientâ€focused survey to assess the effects of the <scp>COVID</scp>â€19 pandemic and social guidelines on people with muscular dystrophy. Muscle and Nerve, 2021, 64, 321-327.	1.0	6
15	Predictors of functional outcomes in patients with facioscapulohumeral muscular dystrophy. Brain, 2021, 144, 3451-3460.	3.7	9
16	Magnetic resonance imaging correlates with electrical impedance myography in facioscapulohumeral muscular dystrophy. Muscle and Nerve, 2020, 61, 644-649.	1.0	10
17	Case Studies on the Genetic and Clinical Diagnosis of Facioscapulohumeral Muscular Dystrophy. Neurologic Clinics, 2020, 38, 529-540.	0.8	2
18	Spinal Cord and Motor Neuron TDP-43 Pathology in a Sporadic Inclusion Body Myositis Patient. Journal of Neuropathology and Experimental Neurology, 2020, 79, 1130-1133.	0.9	0

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19	Single-nucleus RNA-seq identifies divergent populations of FSHD2 myotube nuclei. PLoS Genetics, 2020, 16, e1008754.	1.5	27
20	Long Exercise Test in Periodic Paralysis: A Bayesian Analysis. Muscle and Nerve, 2019, 59, 47-54.	1.0	9
21	SMCHD1 mutation spectrum for facioscapulohumeral muscular dystrophy type 2 (FSHD2) and Bosma arhinia microphthalmia syndrome (BAMS) reveals disease-specific localisation of variants in the ATPase domain. Journal of Medical Genetics, 2019, 56, 693-700.	1.5	27
22	Intronic SMCHD1 variants in FSHD: testing the potential for CRISPR-Cas9 genome editing. Journal of Medical Genetics, 2019, 56, 828-837.	1.5	27
23	Patient-Reported Symptoms in Facioscapulohumeral Muscular Dystrophy (PRISM-FSHD). Neurology, 2019, 93, e1180-e1192.	1.5	43
24	Clinical trial readiness to solve barriers to drug development in FSHD (ReSolve): protocol of a large, international, multi-center prospective study. BMC Neurology, 2019, 19, 224.	0.8	28
25	Generation of genetically matched hiPSC lines from two mosaic facioscapulohumeral dystrophy type 1 patients. Stem Cell Research, 2019, 40, 101560.	0.3	6
26	Clinically Advanced p38 Inhibitors Suppress DUX4 Expression in Cellular and Animal Models of Facioscapulohumeral Muscular Dystrophy. Journal of Pharmacology and Experimental Therapeutics, 2019, 370, 219-230.	1.3	58
27	Single-cell RNA sequencing in facioscapulohumeral muscular dystrophy disease etiology and development. Human Molecular Genetics, 2019, 28, 1064-1075.	1.4	46
28	Facioscapulohumeral muscular dystrophy functional composite outcome measure. Muscle and Nerve, 2018, 58, 72-78.	1.0	21
29	Smchd1 haploinsufficiency exacerbates the phenotype of a transgenic FSHD1 mouse model. Human Molecular Genetics, 2018, 27, 716-731.	1.4	23
30	Facioscapulohumeral muscular dystrophy. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2018, 148, 541-548.	1.0	40
31	Electrical impedance myography in facioscapulohumeral muscular dystrophy: A 1â€year followâ€up study. Muscle and Nerve, 2018, 58, 213-218.	1.0	15
32	Monosomy 18p is a risk factor for facioscapulohumeral dystrophy. Journal of Medical Genetics, 2018, 55, 469-478.	1.5	11
33	Deep characterization of a common D4Z4 variant identifies biallelic DUX4 expression as a modifier for disease penetrance in FSHD2. European Journal of Human Genetics, 2018, 26, 94-106.	1.4	22
34	Variable penetrance of Andersen-Tawil syndrome in a family with a rare missense KCNJ2 mutation. Neurology: Genetics, 2018, 4, e284.	0.9	6
35	Facioscapulohumeral Muscular Dystrophy: Update on Pathogenesis and Future Treatments. Neurotherapeutics, 2018, 15, 863-871.	2.1	72
36	NuRD and CAF-1-mediated silencing of the D4Z4 array is modulated by DUX4-induced MBD3L proteins. ELife, 2018, 7, .	2.8	47

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37	Cis D4Z4 repeat duplications associated with facioscapulohumeral muscular dystrophy type 2. Human Molecular Genetics, 2018, 27, 3488-3497.	1.4	27
38	A checklist for clinical trials in rare disease: obstacles and anticipatory actionsâ€"lessons learned from the FOR-DMD trial. Trials, 2018, 19, 291.	0.7	26
39	Small noncoding RNAs in FSHD2 muscle cells reveal both DUX4- and SMCHD1-specific signatures. Human Molecular Genetics, 2018, 27, 2644-2657.	1.4	6
40	Developing standardized corticosteroid treatment for Duchenne muscular dystrophy. Contemporary Clinical Trials, 2017, 58, 34-39.	0.8	56
41	Episodic weakness and Charcotâ€"marieâ€"tooth disease due to a mitochondrial MTâ€ATP6 mutation. Muscle and Nerve, 2017, 55, 922-927.	1.0	11
42	High frequency of gastrointestinal manifestations in myotonic dystrophy type 1 and type 2. Neurology, 2017, 89, 1348-1354.	1.5	52
43	SMCHD1 regulates a limited set of gene clusters on autosomal chromosomes. Skeletal Muscle, 2017, 7, 12.	1.9	32
44	Validity of the 6 minute walk test in facioscapulohumeral muscular dystrophy. Muscle and Nerve, 2017, 55, 333-337.	1.0	37
45	BET bromodomain inhibitors and agonists of the beta-2 adrenergic receptor identified in screens for compounds that inhibit DUX4 expression in FSHD muscle cells. Skeletal Muscle, 2017, 7, 16.	1.9	46
46	Electrical impedance myography in facioscapulohumeral muscular dystrophy. Muscle and Nerve, 2016, 54, 696-701.	1.0	21
47	A cross sectional study of two independent cohorts identifies serum biomarkers for facioscapulohumeral muscular dystrophy (FSHD). Neuromuscular Disorders, 2016, 26, 405-413.	0.3	36
48	Mutations in DNMT3B Modify Epigenetic Repression of the D4Z4 Repeat and the Penetrance of Facioscapulohumeral Dystrophy. American Journal of Human Genetics, 2016, 98, 1020-1029.	2.6	188
49	Facioscapulohumeral Dystrophy. Current Neurology and Neuroscience Reports, 2016, 16, 66.	2.0	83
50	Clinical practice considerations in facioscapulohumeral muscular dystrophy Sydney, Australia, 21 September 2015. Neuromuscular Disorders, 2016, 26, 462-471.	0.3	7
51	Editorial by concerned physicians: Unintended effect of the orphan drug act on the potential cost of 3,4-diaminopyridine. Muscle and Nerve, 2016, 53, 165-168.	1.0	24
52	Randomized, placebo-controlled trials of dichlorphenamide in periodic paralysis. Neurology, 2016, 86, 1408-1416.	1.5	53
53	Clinical trial preparedness in facioscapulohumeral muscular dystrophy: Clinical, tissue, and imaging outcome measures 29â€"30 May 2015, Rochester, New York. Neuromuscular Disorders, 2016, 26, 181-186.	0.3	43
54	Double SMCHD1 variants in FSHD2: the synergistic effect of two SMCHD1 variants on D4Z4 hypomethylation and disease penetrance in FSHD2. European Journal of Human Genetics, 2016, 24, 78-85.	1.4	23

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55	Immunohistochemical Characterization ofÂFacioscapulohumeralMuscular DystrophyÂMuscle Biopsies. Journal of Neuromuscular Diseases, 2015, 2, 291-299.	1.1	26
56	A feedback loop between nonsense-mediated decay and the retrogene DUX4 in facioscapulohumeral muscular dystrophy. ELife, 2015, 4, .	2.8	97
57	Muscle pathology grade for facioscapulohumeral muscular dystrophy biopsies. Muscle and Nerve, 2015, 52, 521-526.	1.0	50
58	Milder phenotype in facioscapulohumeral dystrophy with 7â€“10 residual D4Z4 repeats. Neurology, 2015, 85, 2147-2150.	1.5	44
59	DICER/AGO-dependent epigenetic silencing of D4Z4 repeats enhanced by exogenous siRNA suggests mechanisms and therapies for FSHD. Human Molecular Genetics, 2015, 24, 4817-4828.	1.4	37
60	Evidence-based guideline summary: Evaluation, diagnosis, and management of facioscapulohumeral muscular dystrophy. Neurology, 2015, 85, 357-364.	1.5	157
61	Hemizygosity forSMCHD1in Facioscapulohumeral Muscular Dystrophy Type 2: Consequences for 18p Deletion Syndrome. Human Mutation, 2015, 36, 679-683.	1.1	32
62	Increased DUX4 expression during muscle differentiation correlates with decreased SMCHD1 protein levels at D4Z4. Epigenetics, 2015, 10, 1133-1142.	1.3	52
63	Inter-individual differences in CpG methylation at D4Z4 correlate with clinical variability in FSHD1 and FSHD2. Human Molecular Genetics, 2015, 24, 659-669.	1.4	130
64	Multiplex Screen of Serum Biomarkers in Facioscapulohumeral Muscular Dystrophy. Journal of Neuromuscular Diseases, 2014, 1, 181-190.	1.1	38
65	DUX4-induced gene expression is the major molecular signature in FSHD skeletal muscle. Human Molecular Genetics, 2014, 23, 5342-5352.	1.4	170
66	DUX4 promotes transcription of FRG2 by directly activating its promoter in facioscapulohumeral muscular dystrophy. Skeletal Muscle, 2014, 4, 19.	1.9	19
67	Facioscapulohumeral Muscular Dystrophy. Neurologic Clinics, 2014, 32, 721-728.	0.8	61
68	Post-exercise increment in compound muscle action potential amplitude in hyperkalemic periodic paralysis. Clinical Neurophysiology, 2014, 125, 2134-2135.	0.7	2
69	Genetic and Epigenetic Characteristics of FSHD-Associated 4q and 10q D4Z4 that are Distinct from Non-4q/10q D4Z4 Homologs. Human Mutation, 2014, 35, 998-1010.	1.1	42
70	Facioscapulohumeral dystrophy: the path to consensus on pathophysiology. Skeletal Muscle, 2014, 4, 12.	1.9	144
71	The FSHD2 Gene SMCHD1 Is a Modifier of Disease Severity in Families Affected by FSHD1. American Journal of Human Genetics, 2013, 93, 744-751.	2.6	154
72	DUX4 Binding to Retroelements Creates Promoters That Are Active in FSHD Muscle and Testis. PLoS Genetics, 2013, 9, e1003947.	1.5	151

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73	Digenic inheritance of an SMCHD1 mutation and an FSHD-permissive D4Z4 allele causes facioscapulohumeral muscular dystrophy type 2. Nature Genetics, 2012, 44, 1370-1374.	9.4	582
74	DUX4 Activates Germline Genes, Retroelements, and Immune Mediators: Implications for Facioscapulohumeral Dystrophy. Developmental Cell, 2012, 22, 38-51.	3.1	384
75	A Unifying Genetic Model for Facioscapulohumeral Muscular Dystrophy. Science, 2010, 329, 1650-1653.	6.0	638
76	Facioscapulohumeral Dystrophy: Incomplete Suppression of a Retrotransposed Gene. PLoS Genetics, 2010, 6, e1001181.	1.5	394
77	171st ENMC International Workshop: Standards of care and management of facioscapulohumeral muscular dystrophy. Neuromuscular Disorders, 2010, 20, 471-475.	0.3	88
78	Common epigenetic changes of D4Z4 in contraction-dependent and contraction-independent FSHD. Human Mutation, 2009, 30, 1449-1459.	1.1	172
79	Facioscapulohumeral Muscular Dystrophy. Neurotherapeutics, 2008, 5, 601-606.	2.1	106
80	Neurologic channelopathies: Evolving concepts and therapeutic challenges. Neurotherapeutics, 2007, 4, 173-173.	2.1	1
81	Facioscapulohumeral muscular dystrophy. Muscle and Nerve, 2006, 34, 1-15.	1.0	302
82	Inclusion body myositis. Current Opinion in Rheumatology, 2002, 14, 653-657.	2.0	56
83	Reply. Annals of Neurology, 1994, 36, 253-253.	2.8	6
84	Facioscapulohumeral Dystrophy Natural History Study: Standardization of Testing Procedures and Reliability of Measurements. Physical Therapy, 1994, 74, 253-263.	1.1	121
85	Losmapimod for facioscapulohumeral muscular dystrophy. , 0, , .		0