Sabrina Sacconi

List of Publications by Year
in descending order

168 papers	6,878 citations	61984 43 h-index	71685 76 g-index
181 all docs	181 docs citations	181 times ranked	6572 citing authors

#	Article	IF	Citations
1	Identification of Serum Interleukin 6 Levels as a Disease Severity Biomarker in Facioscapulohumeral Muscular Dystrophy. Journal of Neuromuscular Diseases, 2022, 9, 83-93.	2.6	15
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.	2.9	8
3	Meeting report: the 2021 FSHD International Research Congress. Skeletal Muscle, 2022, 12, 1.	4.2	12
4	The Splicing of the Mitochondrial Calcium Uniporter Genuine Activator MICU1 Is Driven by RBFOX2 Splicing Factor during Myogenic Differentiation. International Journal of Molecular Sciences, 2022, 23, 2517.	4.1	2
5	Convergence of patient- and physician-reported outcomes in the French National Registry of Facioscapulohumeral Dystrophy. Orphanet Journal of Rare Diseases, 2022, 17, 96.	2.7	2
6	Ultraâ€“highâ€frequency ultrasound imaging of sural nerve: A comparative study with nerve biopsy in progressive neuropathies. Muscle and Nerve, 2021, 63, 46-51.	2.2	9
7	Motor axonal neuropathy associated with <scp>GNE</scp> mutations. Muscle and Nerve, 2021, 63, 396-401.	2.2	12
8	The facioscapulohumeral muscular dystrophy Raschâ€built overall disability scale (FSHDâ€RODS). European Journal of Neurology, 2021, 28, 2339-2348.	3.3	8
9	Genotypeâ€“phenotype correlation in French patients with myelin protein zero geneâ€related inherited neuropathy. European Journal of Neurology, 2021, 28, 2913-2921.	3.3	6
10	Efficacy and safety of mexiletine in non-dystrophic myotonias: A randomised, double-blind, placebo-controlled, cross-over study. Neuromuscular Disorders, 2021, 31, 1124-1135.	0.6	14
11	A case of <scp>ASAH1</scp>â€related pure SMA evolving into adultâ€onset Farber disease. Clinical Genetics, 2021, 100, 234-235.	2.0	3
12	E-Health & Innovation to Overcome Barriers in Neuromuscular Diseases. Report from the 1st eNMD Congress: Nice, France, March 22-23, 2019. Journal of Neuromuscular Diseases, 2021, 8, 743-754.	2.6	2
13	1st FSHD European Trial Network workshop:Working towards trial readiness across Europe. Neuromuscular Disorders, 2021, 31, 907-918.	0.6	9
14	A recurrent RYR1 mutation associated with early-onset hypotonia and benign disease course. Acta Neuropathologica Communications, 2021, 9, 155.	5.2	1
15	SMA â€“ OUTCOME MEASURES AND REGISTRIES. Neuromuscular Disorders, 2021, 31, S130-S131.	0.6	1
16	CHANNELOPATHIES AND RELATED DISORDERS. Neuromuscular Disorders, 2021, 31, S116-S117.	0.6	0
17	CHANNELOPATHIES AND RELATED DISORDERS. Neuromuscular Disorders, 2021, 31, S118.	0.6	0
18	CHANNELOPATHIES AND RELATED DISORDERS. Neuromuscular Disorders, 2021, 31, S116.	0.6	0

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19	Safety and efficacy of cipaglucosidase alfa plus miglustat versus alglucosidase alfa plus placebo in late-onset Pompe disease (PROPEL): an international, randomised, double-blind, parallel-group, phase 3 trial. Lancet Neurology, The, 2021, 20, 1027-1037.	10.2	42
20	AUTOPHAGIC MYOPATHIES / MYOFIBRILLAR MYOPATHIES / DISTAL MYOPATHIES / POMPE DISEASE. Neuromuscular Disorders, 2020, 30, S49.	0.6	0
21	SMA: REGISTRIES, BIOMARKERS & OUTCOME MEASURES. Neuromuscular Disorders, 2020, 30, S101.	0.6	0
22	FSHD / OPMD / MYOTONIC DYSTROPHY. Neuromuscular Disorders, 2020, 30, S112.	0.6	0
23	FSHD / OPMD / MYOTONIC DYSTROPHY. Neuromuscular Disorders, 2020, 30, S112.	0.6	0
24	Creation and implementation of a European registry for patients with McArdle disease and other muscle glycogenoses (EUROMAC registry). Orphanet Journal of Rare Diseases, 2020, 15, 187.	2.7	3
25	Data from the European registry for patients with McArdle disease and other muscle glycogenoses (EUROMAC). Orphanet Journal of Rare Diseases, 2020, 15, 330.	2.7	23
26	Deep phenotyping of facioscapulohumeral muscular dystrophy type 2 by magnetic resonance imaging. European Journal of Neurology, 2020, 27, 2604-2615.	3.3	16
27	Diagnostic challenges in metabolic myopathies. Expert Review of Neurotherapeutics, 2020, 20, 1287-1298.	2.8	7
28	Singleâ€fiber studies for assigning pathogenicity of eight mitochondrial DNA variants associated with mitochondrial diseases. Human Mutation, 2020, 41, 1394-1406.	2.5	4
29	Inclusion body myositis in patients with spinocerebellar ataxia types 3 and 6. Journal of Neurology, Neurosurgery and Psychiatry, 2020, 91, 876-878.	1.9	2
30	Longâ€term benefit of enzyme replacement therapy with alglucosidase alfa in adults with Pompe disease: Prospective analysis from the French Pompe Registry. Journal of Inherited Metabolic Disease, 2020, 43, 1219-1231.	3.6	21
31	Type 1 FSHD with 6â€“10 Repeated Units: Factors Underlying Severity in Index Cases and Disease Penetrance in Their Relatives Attention. International Journal of Molecular Sciences, 2020, 21, 2221.	4.1	2
32	Bilateral scapulothoracic arthrodesis for facioscapulohumeral muscular dystrophy: function, fusion, and respiratory consequences. Journal of Shoulder and Elbow Surgery, 2020, 29, 931-940.	2.6	8
33	Mitochondrial function in skeletal myofibers is controlled by a TRF2â€SIRT3 axis over lifetime. Aging Cell, 2020, 19, e13097.	6.7	31
34	Mannose 6â€phosphonate labelling: A key for processing the therapeutic enzyme in Pompe disease. Journal of Cellular and Molecular Medicine, 2019, 23, 6499-6503.	3.6	4
35	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.	3.2	27
36	P.46Clinical trial readiness to solve barriers to drug development in FSHD (ReSolve): protocol of a large, multi-center prospective study. Neuromuscular Disorders, 2019, 29, S54.	0.6	0

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37	P.69NEO1 and NEO-EXT studies: exploratory efficacy of repeat avalglucosidase alfa dosing for up to 5 years in participants with late-onset Pompe disease (LOPD). Neuromuscular Disorders, 2019, 29, S60-S61.	0.6	0
38	P.120EUROMAC: A European registry for patients with McArdle disease and other very rare muscle glycogenoses. Neuromuscular Disorders, 2019, 29, S83.	0.6	0
39	P.306Multicentric MRI study in a cohort of FSHD2 patients: pattern definition and differences between FSHD1 and FSHD2. Neuromuscular Disorders, 2019, 29, S156.	0.6	0
40	P.398Usefulness of R-Pact scale for the follow-up of patients with late-onset Pompe disease: results from the French Pompe registry. Neuromuscular Disorders, 2019, 29, S198.	0.6	0
41	Deep morphological analysis of muscle biopsies from type III glycogenesis (GSDIII), debranching enzyme deficiency, revealed stereotyped vacuolar myopathy and autophagy impairment. Acta Neuropathologica Communications, 2019, 7, 167.	5.2	17
42	Intronic SMCHD1 variants in FSHD: testing the potential for CRISPR-Cas9 genome editing. Journal of Medical Genetics, 2019, 56, 828-837.	3.2	27
43	Scapular dyskinesis in myotonic dystrophy type 1: clinical characteristics and genetic investigations. Journal of Neurology, 2019, 266, 2987-2996.	3.6	1
44	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.	1.8	28
45	Expanding the importance of HMERF titinopathy: new mutations and clinical aspects. Journal of Neurology, 2019, 266, 680-690.	3.6	31
46	Comparison of high-frequency and ultrahigh-frequency probes in chronic inflammatory demyelinating polyneuropathy. Journal of Neurology, 2019, 266, 2277-2285.	3.6	16
47	A multicenter retrospective study of charcotâ€marieâ€tooth disease type 4B (CMT4B) associated with mutations in myotubularinâ€related proteins (MTMRs). Annals of Neurology, 2019, 86, 55-67.	5.3	35
48	FSHD1 and FSHD2 form a disease continuum. Neurology, 2019, 92, e2273-e2285.	1.1	50
49	NEO1 and NEO-EXT studies: Long-term safety of repeat avalglucosidase alfa dosing for 4.5 years in late-onset Pompe disease patients. Molecular Genetics and Metabolism, 2019, 126, S115-S116.	1.1	0
50	Deciphering the complexity of the 4q and 10q subtelomeres by molecular combing in healthy individuals and patients with facioscapulohumeral dystrophy. Journal of Medical Genetics, 2019, 56, 590-601.	3.2	24
51	Dynamic transcriptomic analysis reveals suppression of PGC1Î±/ERRÎ± drives perturbed myogenesis in facioscapulohumeral muscular dystrophy. Human Molecular Genetics, 2019, 28, 1244-1259.	2.9	52
52	COX16 is required for assembly of cytochrome c oxidase in human cells and is involved in copper delivery to COX2. Biochimica Et Biophysica Acta - Bioenergetics, 2018, 1859, 244-252.	1.0	25
53	Monosomy 18p is a risk factor for facioscapulohumeral dystrophy. Journal of Medical Genetics, 2018, 55, 469-478.	3.2	11
54	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.	2.8	22

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55	MT-CYB deletion in an encephalomyopathy with hyperintensity of middle cerebellar peduncles. Neurology: Genetics, 2018, 4, e268.	1.9	6
56	The French National Registry of patients with Facioscapulohumeral muscular dystrophy. Orphanet Journal of Rare Diseases, 2018, 13, 218.	2.7	11
57	Lateâ€onset Pompe disease in France: molecular features and epidemiology from a nationwide study. Journal of Inherited Metabolic Disease, 2018, 41, 937-946.	3.6	27
58	New variant of necklace fibres display peculiar lysosomal structures and mitophagy. Neuromuscular Disorders, 2018, 28, 846-856.	0.6	4
59	Cis D4Z4 repeat duplications associated with facioscapulohumeral muscular dystrophy type 2. Human Molecular Genetics, 2018, 27, 3488-3497.	2.9	27
60	Miller Fisher syndrome, Bickerstaff brainstem encephalitis and Guillain-BarrÃ© syndrome overlap with persistent non-demyelinating conduction blocks: a case report. BMC Neurology, 2018, 18, 101.	1.8	9
61	Inflammatory facioscapulohumeral muscular dystrophy type 2 in 18p deletion syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 1760-1763.	1.2	6
62	FSHD type 2 and Bosma arhinia microphthalmia syndrome. Neurology, 2018, 91, e562-e570.	1.1	24
63	Long term follow-up of cerebrovascular abnormalities in late onset Pompe disease (LOPD). Journal of Neurology, 2017, 264, 589-590.	3.6	6
64	The quality of life in genetic neuromuscular disease questionnaire: Rasch validation of the French version. Muscle and Nerve, 2017, 56, 1085-1091.	2.2	14
65	Cytosolic 5â€²-nucleotidase 1A autoantibody profile and clinical characteristics in inclusion body myositis. Annals of the Rheumatic Diseases, 2017, 76, 862-868.	0.9	71
66	225th ENMC international workshop:. Neuromuscular Disorders, 2017, 27, 782-790.	0.6	20
67	Short-TERM Neuromuscular Electrical Stimulation Training of the Tibialis Anterior Did Not Improve Strength and Motor Function in Facioscapulohumeral Muscular Dystrophy Patients. American Journal of Physical Medicine and Rehabilitation, 2017, 96, e56-e63.	1.4	6
68	A novel gainâ€ofâ€function mutation in ORAI1 causes lateâ€onset tubular aggregate myopathy and congenital miosis. Clinical Genetics, 2017, 91, 780-786.	2.0	50
69	Estrogens as a potential disease modifier in FSHD: a retrospective clinical study. Neuromuscular Disorders, 2017, 27, S200.	0.6	3
70	Safety and efficacy of eculizumab in anti-acetylcholine receptor antibody-positive refractory generalised myasthenia gravis (REGAIN): a phase 3, randomised, double-blind, placebo-controlled, multicentre study. Lancet Neurology, The, 2017, 16, 976-986.	10.2	472
71	Effect of enzyme replacement therapy with alglucosidase alfa (MyozymeÂ®) in 12 patients with advanced late-onset Pompe disease. Molecular Genetics and Metabolism, 2017, 122, 80-85.	1.1	21
72	The EUROMAC registry for rare glycogen storage diseases: preliminary report. Neuromuscular Disorders, 2017, 27, S203-S204.	0.6	0

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73	Autophagy impairment in muscle biopsies from debranching enzyme deficiency (GSDIII) patients: pinpointing novel therapeutic perspectives. Neuromuscular Disorders, 2017, 27, S205-S206.	0.6	1
74	Expanding importance of HMERF titinopathy: new mutations and clinical aspects. Neuromuscular Disorders, 2017, 27, S237.	0.6	0
75	Morphological spectrum of RYR1 recessive myopathies: Clinical and genetic correlation.. Neuromuscular Disorders, 2017, 27, S239.	0.6	0
76	Bent spine syndrome as the initial symptom of lateâ€onset Pompe disease. Muscle and Nerve, 2017, 56, 167-170.	2.2	9
77	Severe defect in mitochondrial complex I assembly with mitochondrial DNA deletions in ACAD9â€deficient mild myopathy. Muscle and Nerve, 2017, 55, 919-922.	2.2	3
78	Estrogens enhance myoblast differentiation in facioscapulohumeral muscular dystrophy by antagonizing DUX4 activity. Journal of Clinical Investigation, 2017, 127, 1531-1545.	8.2	46
79	Long-term exposure to Myozyme results in a decrease of anti-drug antibodies in late-onset Pompe disease patients. Scientific Reports, 2016, 6, 36182.	3.3	22
80	Modeling Andersen's Syndrome in Human Induced Pluripotent Stem Cells. Stem Cells and Development, 2016, 25, 151-159.	2.1	14
81	The motor function measure (MFM) in the facio scapulo humeral dystrophy (FSHD) population: Description and responsiveness. Annals of Physical and Rehabilitation Medicine, 2016, 59, e84-e85.	2.3	3
82	TheCOQ2genotype predicts the severity of coenzyme Q₁₀deficiency. Human Molecular Genetics, 2016, 25, 4256-4265.	2.9	53
83	Mini-Exome Coupled to Read-Depth Based Copy Number Variation Analysis in Patients with Inherited Ataxias. Human Mutation, 2016, 37, 1340-1353.	2.5	33
84	GNE myopathy: Characteristics of affected patients diagnosed in mainland France. Neuromuscular Disorders, 2016, 26, S169.	0.6	0
85	The genetic basis of undiagnosed muscular dystrophies and myopathies. Neurology, 2016, 87, 71-76.	1.1	92
86	Clinical practice considerations in facioscapulohumeral muscular dystrophy Sydney, Australia, 21 September 2015. Neuromuscular Disorders, 2016, 26, 462-471.	0.6	7
87	Design, construction, and monitoring of a building with deep basements in Rome. Canadian Geotechnical Journal, 2016, 53, 210-224.	2.8	7
88	Gender as a Modifying Factor Influencing Myotonic Dystrophy Type 1 Phenotype Severity and Mortality: A Nationwide Multiple Databases Cross-Sectional Observational Study. PLoS ONE, 2016, 11, e0148264.	2.5	113
89	Correlation between low FAT1 expression and early affected muscle in FSHD. Neuromuscular Disorders, 2015, 25, S312.	0.6	0
90	The impact of enzyme replacement therapy on the progression of Pompe disease. Neuromuscular Disorders, 2015, 25, S189.	0.6	1

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91	CHCHD10 mutations are not a common cause of SMN1â€negative type III/IV spinal motor atrophy. Annals of Neurology, 2015, 78, 831-831.	5.3	3
92	Correlation between low <scp>FAT</scp>1 expression and early affected muscle in facioscapulohumeral muscular dystrophy. Annals of Neurology, 2015, 78, 387-400.	5.3	32
93	Letter to the Editor on a paper by Hsiao C-T, Tsai P-C, Liao Y-C, Lee Y-C, Soong B-W. C9ORF72 repeat expansion is not a significant cause of late-onset cerebellar ataxia syndrome. J Neurol Sci 2014;347:322â€“324.. Journal of the Neurological Sciences, 2015, 351, 196-197.	0.6	1
94	Next generation sequencing on patients with LGMD and nonspecific myopathies: Findings associated with ANO5 mutations. Neuromuscular Disorders, 2015, 25, 533-541.	0.6	65
95	Low penetrance in facioscapulohumeral muscular dystrophy type 1 with large pathological D4Z4 alleles: a cross-sectional multicenter study. Orphanet Journal of Rare Diseases, 2015, 10, 2.	2.7	32
96	Construction of a Quality of Life Questionnaire for slowly progressive neuromuscular disease. Quality of Life Research, 2015, 24, 2615-2623.	3.1	19
97	Large screening of patients diagnosed as limb girdle muscular dystrophy or congenital myopathy using Motorplex. Neuromuscular Disorders, 2015, 25, S297.	0.6	0
98	Facioscapulohumeral muscular dystrophy. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2015, 1852, 607-614.	3.8	47
99	Inter-individual differences in CpG methylation at D4Z4 correlate with clinical variability in FSHD1 and FSHD2. Human Molecular Genetics, 2015, 24, 659-669.	2.9	130
100	The inward rectifier potassium channel Kir2.1 is required for osteoblastogenesis. Human Molecular Genetics, 2015, 24, 471-479.	2.9	19
101	Long-Term Mechanical Ventilation Equipment for Neuromuscular Patients: Meeting the Expectations of Patients and Prescribers. Respiratory Care, 2014, 59, 97-106.	1.6	28
102	Atrio-ventricular block requiring pacemaker in patients with late onset Pompe disease. Neuromuscular Disorders, 2014, 24, 648-650.	0.6	15
103	Quantitative multiplex PCR of short fluorescent fragments for the detection of large intragenic POLG rearrangements in a large French cohort. European Journal of Human Genetics, 2014, 22, 542-550.	2.8	27
104	DUX4 and DUX4 downstream target genes are expressed in fetal FSHD muscles. Human Molecular Genetics, 2014, 23, 171-181.	2.9	61
105	Unusual multisystemic involvement and a novel BAG3 mutation revealed by NGS screening in a large cohort of myofibrillar myopathies. Orphanet Journal of Rare Diseases, 2014, 9, 121.	2.7	38
106	G.O.7. Neuromuscular Disorders, 2014, 24, 851.	0.6	0
107	A.P.3. Neuromuscular Disorders, 2014, 24, 831.	0.6	0
108	G.P.245. Neuromuscular Disorders, 2014, 24, 890.	0.6	1

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109	Development and validation of a motor function classification in patients with neuromuscular disease: The NM-Score. Annals of Physical and Rehabilitation Medicine, 2013, 56, 673-686.	2.3	6
110	The FSHD2 Gene SMCHD1 Is a Modifier of Disease Severity in Families Affected by FSHD1. American Journal of Human Genetics, 2013, 93, 744-751.	6.2	154
111	The French Pompe registry. Baseline characteristics of a cohort of 126 patients with adult Pompe disease. Revue Neurologique, 2013, 169, 595-602.	1.5	48
112	P.16.3 DUX4 and DUX4 downstream target genes are expressed in fetal FSHD muscles. Neuromuscular Disorders, 2013, 23, 823.	0.6	0
113	A Novel Role for the RNAâ€“Binding Protein FXR1P in Myoblasts Cell-Cycle Progression by Modulating p21/Cdkn1a/Cip1/Waf1 mRNA Stability. PLoS Genetics, 2013, 9, e1003367.	3.5	67
114	Coats syndrome in facioscapulohumeral dystrophy type 1. Neurology, 2013, 80, 1247-1250.	1.1	63
115	<scp>EFNS</scp> review on the role of muscle biopsy in the investigation of myalgia. European Journal of Neurology, 2013, 20, 997-1005.	3.3	27
116	Haploinsufficiency ofCOQ4causes coenzyme Q₁₀deficiency. Journal of Medical Genetics, 2012, 49, 187-191.	3.2	95
117	Patients with a phenotype consistent with facioscapulohumeral muscular dystrophy display genetic and epigenetic heterogeneity. Journal of Medical Genetics, 2012, 49, 41-46.	3.2	55
118	The Potassium Channel Kir2.1 Activity is Required for Osteoblastogenesis. Biophysical Journal, 2012, 102, 538a.	0.5	1
119	Digenic inheritance of an SMCHD1 mutation and an FSHD-permissive D4Z4 allele causes facioscapulohumeral muscular dystrophy type 2. Nature Genetics, 2012, 44, 1370-1374.	21.4	582
120	Pain assessment in Charcot-Marie-Tooth (CMT) disease. Annals of Physical and Rehabilitation Medicine, 2012, 55, 160-173.	2.3	49
121	A novel CRYAB mutation resulting in multisystemic disease. Neuromuscular Disorders, 2012, 22, 66-72.	0.6	84
122	Copper and bezafibrate cooperate to rescue cytochrome c oxidase deficiency in cells of patients with sco2 mutations. Orphanet Journal of Rare Diseases, 2012, 7, 21.	2.7	29
123	Motor and respiratory heterogeneity in Duchenne patients: Implication for clinical trials. European Journal of Paediatric Neurology, 2012, 16, 149-160.	1.6	112
124	Distrofia muscolare facio-scapolo-omerale. EMC - Neurologia, 2011, 11, 1-11.	0.0	0
125	O.19 Facioscapulohumeral muscular dystrophy: Muscle running out of control. Neuromuscular Disorders, 2011, 21, 749.	0.6	0
126	P3.52 Risk of dysrythmic cardiomyopathy may be considered in patients with adult onset Pompe diasease. Neuromuscular Disorders, 2011, 21, 698.	0.6	0

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127	Current French Pompe Prevalence Study (French PoPS). Clinical Therapeutics, 2011, 33, S21.	2.5	1
128	A Unifying Genetic Model for Facioscapulohumeral Muscular Dystrophy. Science, 2010, 329, 1650-1653.	12.6	638
129	Abnormalities of cerebral arteries are frequent in patients with late-onset Pompe disease. Journal of Neurology, 2010, 257, 1730-1733.	3.6	68
130	Isolation of a Highly Myogenic CD34-Negative Subset of Human Skeletal Muscle Cells Free of Adipogenic Potential. Stem Cells, 2010, 28, 753-764.	3.2	60
131	Hierarchization of Myogenic and Adipogenic Progenitors Within Human Skeletal Muscle. Stem Cells, 2010, 28, 2182-2194.	3.2	48
132	EFNS guidelines on the diagnostic approach to pauciâ€•or asymptomatic hyperCKemia. European Journal of Neurology, 2010, 17, 767-773.	3.3	157
133	Clinical features of facioscapulohumeral muscular dystrophy 2. Neurology, 2010, 75, 1548-1554.	1.1	215
134	Coenzyme Q10 is frequently reduced in muscle of patients with mitochondrial myopathy. Neuromuscular Disorders, 2010, 20, 44-48.	0.6	84
135	Neuromuscular Electrical Stimulation Training: A Safe and Effective Treatment for Facioscapulohumeral Muscular Dystrophy Patients. Archives of Physical Medicine and Rehabilitation, 2010, 91, 697-702.	0.9	29
136	Mutation analysis of COX18 in 29 patients with isolated cytochrome c oxidase deficiency. Journal of Human Genetics, 2009, 54, 419-421.	2.3	7
137	The D4Z4 Macrosatellite Repeat Acts as a CTCF and A-Type Lamins-Dependent Insulator in Facio-Scapulo-Humeral Dystrophy. PLoS Genetics, 2009, 5, e1000394.	3.5	99
138	Mechanisms underlying Andersen's syndrome pathology in skeletal muscle are revealed in human myotubes. American Journal of Physiology - Cell Physiology, 2009, 297, C876-C885.	4.6	17
139	Common epigenetic changes of D4Z4 in contraction-dependent and contraction-independent FSHD. Human Mutation, 2009, 30, 1449-1459.	2.5	172
140	Do patients having a decrease in SNAP amplitude during the course of MMN present with a different condition?. Journal of Neurology, 2009, 256, 1876-1880.	3.6	32
141	G.O.3 Contraction-dependent (FSHD1) and independent (FSHD2) epigenetic changes of D4Z4 unify FSHD. Neuromuscular Disorders, 2009, 19, 545.	0.6	0
142	Null mutations causing depletion of the type 1 ryanodine receptor (RYR1) are commonly associated with recessive structural congenital myopathies with cores. Human Mutation, 2008, 29, 670-678.	2.5	89
143	G.P.11.11 Phenotypes of Pompe disease siblings. Neuromuscular Disorders, 2008, 18, 803.	0.6	0
144	Optimal reconfiguration policy to react to product changes. International Journal of Production Research, 2008, 46, 2651-2673.	7.5	22

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145	Alteration of expression of muscle specific isoforms of the fragile X related protein 1 (FXR1P) in facioscapulohumeral muscular dystrophy patients. Journal of Medical Genetics, 2008, 45, 679-685.	3.2	54
146	Dehydroepiandrosterone for myotonic dystrophy type 1. Neurology, 2008, 71, 407-412.	1.1	18
147	A functionally dominant mitochondrial DNA mutation. Human Molecular Genetics, 2008, 17, 1814-1820.	2.9	104
148	M.P.3.12 Characterization of COX16, a novel human gene required for cytochrome c oxidase assembly. Neuromuscular Disorders, 2007, 17, 830.	0.6	0
149	Argininosuccinate lyase deficiency: mutational spectrum in Italian patients and identification of a novelASLpseudogene. Human Mutation, 2007, 28, 694-702.	2.5	46
150	Diagnostic challenges in facioscapulohumeral muscular dystrophy. Neurology, 2006, 67, 1464-1466.	1.1	21
151	Unusual Clinical Presentations in Four Cases of Leigh Disease, Cytochrome C Oxidase Deficiency, and SURF1 Gene Mutations. Journal of Child Neurology, 2005, 20, 670-674.	1.4	32
152	Infantile encephalomyopathy and nephropathy with CoQ10 deficiency: A CoQ10-responsive condition. Neurology, 2005, 65, 606-608.	1.1	184
153	Normal growth and regenerating ability of myoblasts from unaffected muscles of facioscapulohumeral muscular dystrophy patients. Gene Therapy, 2005, 12, 1651-1662.	4.5	72
154	hCOX18 and hCOX19: Two human genes involved in cytochrome c oxidase assembly. Biochemical and Biophysical Research Communications, 2005, 337, 832-839.	2.1	28
155	Peut-on envisager une thÃ©rapie cellulaire autologue de la dystrophie musculaire facio-scapulo-humÃ©rale ?. Bulletin De L'Academie Nationale De Medecine, 2005, 189, 697-714.	0.0	2
156	Congenital cardiomyopathy and pulmonary hypertension: Another fatal variant of cytochrome-c oxidase deficiency. Journal of Inherited Metabolic Disease, 2004, 27, 735-739.	3.6	20
157	NovelSURF1 mutation in a child with subacute encephalopathy and without the radiological features of Leigh Syndrome. American Journal of Medical Genetics Part A, 2004, 128A, 195-198.	2.4	29
158	Hepatocerebral Mitochondrial DNA Depletion Syndrome: Clinical and Morphologic Features of a Nuclear Gene Mutation. Journal of Pediatric Gastroenterology and Nutrition, 2004, 38, 216-220.	1.8	25
159	Mutation Screening in Patients With Isolated Cytochrome c Oxidase Deficiency. Pediatric Research, 2003, 53, 224-230.	2.3	62
160	Mutation Screening in Patients With Isolated Cytochrome c Oxidase Deficiency. Pediatric Research, 2003, 53, 224-230.	2.3	44
161	Cytochrome c Oxidase Deficiency Due to a Novel SCO2 Mutation Mimics Werdnig-Hoffmann Disease. Archives of Neurology, 2002, 59, 862-5.	4.5	95
162	Mitochondrial DNA depletion. Neurology, 2002, 59, 1197-1202.	1.1	156

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163	Complex Neurologic Syndrome Associated With the G1606A Mutation of Mitochondrial DNA. Archives of Neurology, 2002, 59, 1013.	4.5	32
164	Copper supplementation restores cytochrome c oxidase activity in cultured cells from patients with SCO2 mutations. Biochemical Journal, 2002, 363, 321.	3.7	55
165	Copper supplementation restores cytochrome c oxidase activity in cultured cells from patients with SCO2 mutations. Biochemical Journal, 2002, 363, 321-327.	3.7	66
166	Mitochondrial myopathy and ophthalmoplegia in a sporadic patient with the G12315A mutation in mitochondrial DNA. Neuromuscular Disorders, 2002, 12, 865-868.	0.6	29
167	Mitochondrial DNA depletion and dGK gene mutations. Annals of Neurology, 2002, 52, 311-317.	5.3	152
168	Acute Disseminated Encephalomyelitis Associated With Hepatitis C Virus Infection. Archives of Neurology, 2001, 58, 1679.	4.5	57