Baziel van Engelen

List of Publications by Year
in descending order

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508 papers	21,037 citations	10650 74 h-index	21843 118 g-index
527 all docs	527 docs citations	527 times ranked	18162 citing authors

#	Article	IF	Citations
1	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
2	The clinical spectrum of neuralgic amyotrophy in 246 cases. Brain, 2006, 129, 438-450.	3.7	572
3	Performance of near-infrared spectroscopy in measuring local O₂ consumption and blood flow in skeletal muscle. Journal of Applied Physiology, 2001, 90, 511-519.	1.2	477
4	Skeletal Muscle Ultrasound: Correlation Between Fibrous Tissue and Echo Intensity. Ultrasound in Medicine and Biology, 2009, 35, 443-446.	0.7	456
5	Glucose transporter-1 deficiency syndrome: the expanding clinical and genetic spectrum of a treatable disorder. Brain, 2010, 133, 655-670.	3.7	356
6	Autoantibody profiles in the sera of European patients with myositis. Annals of the Rheumatic Diseases, 2001, 60, 116-123.	0.5	330
7	Anti-signal recognition particle autoantibodies: marker of a necrotising myopathy. Annals of the Rheumatic Diseases, 2006, 65, 1635-1638.	0.5	289
8	Population-based incidence and prevalence of facioscapulohumeral dystrophy. Neurology, 2014, 83, 1056-1059.	1.5	278
9	A second missense mutation in the mitochondrial ATPase 6 gene in Leigh's syndrome. Annals of Neurology, 1993, 34, 410-412.	2.8	239
10	Clinical applications of high-density surface EMG: A systematic review. Journal of Electromyography and Kinesiology, 2006, 16, 586-602.	0.7	236
11	The assessment of fatigue: Psychometric qualities and norms for the Checklist individual strength. Journal of Psychosomatic Research, 2017, 98, 40-46.	1.2	222
12	Clinical and molecular genetic characteristics of patients with cerebrotendinous xanthomatosis. Brain, 2000, 123, 908-919.	3.7	219
13	Clinical neurophysiology of fatigue. Clinical Neurophysiology, 2008, 119, 2-10.	0.7	207
14	Autoantibodies to cytosolic 5â€²â€nucleotidase 1A in inclusion body myositis. Annals of Neurology, 2013, 73, 397-407.	2.8	206
15	Serum creatine kinase as predictor of clinical course in rhabdomyolysis: a 5-year intensive care survey. Intensive Care Medicine, 2003, 29, 1121-1125.	3.9	202
16	The Epidemiology of Neuromuscular Disorders: A Comprehensive Overview of the Literature. Journal of Neuromuscular Diseases, 2015, 2, 73-85.	1.1	200
17	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
18	Epidemiology of inclusion body myositis in the Netherlands: A nationwide study. Neurology, 2000, 55, 1385-1388.	1.5	187

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19	Statin-Induced Myopathy Is Associated with Mitochondrial Complex III Inhibition. Cell Metabolism, 2015, 22, 399-407.	7.2	180
20	A weak balance: the contribution of muscle weakness to postural instability and falls. Nature Clinical Practice Neurology, 2008, 4, 504-515.	2.7	179
21	Exercise Therapy and Other Types of Physical Therapy for Patients With Neuromuscular Diseases: A Systematic Review. Archives of Physical Medicine and Rehabilitation, 2007, 88, 1452-1464.	0.5	177
22	Pain in Ehlers-Danlos Syndrome Is Common, Severe, and Associated with Functional Impairment. Journal of Pain and Symptom Management, 2010, 40, 370-378.	0.6	176
23	Common epigenetic changes of D4Z4 in contraction-dependent and contraction-independent FSHD. Human Mutation, 2009, 30, 1449-1459.	1.1	172
24	Nuclear envelope alterations in fibroblasts from LGMD1B patients carrying nonsense Y259X heterozygous or homozygous mutation in lamin A/C gene. Experimental Cell Research, 2003, 291, 352-362.	1.2	169
25	Mutations in RYR1 are a common cause of exertional myalgia and rhabdomyolysis. Neuromuscular Disorders, 2013, 23, 540-548.	0.3	169
26	A natural history study of late onset spinal muscular atrophy types 3b and 4. Journal of Neurology, 2008, 255, 1400-1404.	1.8	158
27	Causes and consequences of cerebral small vessel disease. The RUN DMC study: a prospective cohort study. Study rationale and protocol. BMC Neurology, 2011, 11, 29.	0.8	154
28	Neuralgic amyotrophy and hepatitis E virus infection. Neurology, 2014, 82, 498-503.	1.5	150
29	Cerebrotendinous Xanthomatosis: The Spectrum of Imaging Findings and the Correlation with Neuropathologic Findings. Radiology, 2000, 217, 869-876.	3.6	147
30	Successful Treatment of Dermatomyositis and Polymyositis with Anti-Tumor-Necrosis-Factor-Alpha: Preliminary Observations. European Neurology, 2003, 50, 10-15.	0.6	143
31	Dominant Mutations in KBTBD13, a Member of the BTB/Kelch Family, Cause Nemaline Myopathy with Cores. American Journal of Human Genetics, 2010, 87, 842-847.	2.6	143
32	Neuromuscular involvement in various types of Ehlersâ€“Danlos syndrome. Annals of Neurology, 2009, 65, 687-697.	2.8	141
33	Fatigue Is a Frequent and Clinically Relevant Problem in Ehlers-Danlos Syndrome. Seminars in Arthritis and Rheumatism, 2010, 40, 267-274.	1.6	131
34	Strength training and aerobic exercise training for muscle disease. The Cochrane Library, 2013, , CD003907.	1.5	130
35	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
36	Best practice guidelines and recommendations on the molecular diagnosis of myotonic dystrophy types 1 and 2. European Journal of Human Genetics, 2012, 20, 1203-1208.	1.4	129

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37	Study on the gene and phenotypic characterisation of autosomal recessive demyelinating motor and sensory neuropathy (Charcot-Marie-Tooth disease) with a gene locus on chromosome 5q23-q33. Journal of Neurology, Neurosurgery and Psychiatry, 1999, 66, 569-574.	0.9	128
38	Acceleration in the Rate of CNS Remyelination in Lysolecithin-Induced Demyelination. Journal of Neuroscience, 1998, 18, 2498-2505.	1.7	127
39	Distinct Disease Phases in Muscles of Facioscapulohumeral Dystrophy Patients Identified by MR Detected Fat Infiltration. PLoS ONE, 2014, 9, e85416.	1.1	125
40	Clinical characteristics of patients with myositis and autoantibodies to different fragments of the Mi-2Â antigen. Annals of the Rheumatic Diseases, 2006, 65, 242-245.	0.5	124
41	Protein S-100B, neuron-specific enolase (NSE), myelin basic protein (MBP) and glial fibrillary acidic protein (GFAP) in cerebrospinal fluid (CSF) and blood of neurological patients. Brain Research Bulletin, 2003, 61, 261-264.	1.4	122
42	Inclusion body myositis. Journal of Neurology, 2005, 252, 1448-1454.	1.8	118
43	In vivo quantitative near-infrared spectroscopy in skeletal muscle during incremental isometric handgrip exercise. Clinical Physiology and Functional Imaging, 2002, 22, 210-217.	0.5	117
44	Nucleoplasmic LAP2Î±â€“lamin A complexes are required to maintain a proliferative state in human fibroblasts. Journal of Cell Biology, 2007, 176, 163-172.	2.3	117
45	Disease specificity of autoantibodies to cytosolic 5â€²-nucleotidase 1A in sporadic inclusion body myositis versus known autoimmune diseases. Annals of the Rheumatic Diseases, 2016, 75, 696-701.	0.5	116
46	Consensus-based care recommendations for adults with myotonic dystrophy type 1. Neurology: Clinical Practice, 2018, 8, 507-520.	0.8	115
47	MSH3 modifies somatic instability and disease severity in Huntingtonâ€™s and myotonic dystrophy type 1. Brain, 2019, 142, 1876-1886.	3.7	114
48	Clinical features and predictors for disease natural progression in adults with Pompe disease: a nationwide prospective observational study. Orphanet Journal of Rare Diseases, 2012, 7, 88.	1.2	112
49	Amyloid deposits and inflammatory infiltrates in sporadic inclusion body myositis: the inflammatory egg comes before the degenerative chicken. Acta Neuropathologica, 2015, 129, 611-624.	3.9	112
50	<scp>RYR</scp>1â€related myopathies: a wide spectrum of phenotypes throughout life. European Journal of Neurology, 2015, 22, 1094-1112.	1.7	111
51	Comparison of weakness progression in inclusion body myositis during treatment with methotrexate or placebo. Annals of Neurology, 2002, 51, 369-372.	2.8	108
52	Both aerobic exercise and cognitive-behavioral therapy reduce chronic fatigue in FSHD. Neurology, 2014, 83, 1914-1922.	1.5	106
53	The development of a model of fatigue in neuromuscular disorders: A longitudinal study. Journal of Psychosomatic Research, 2007, 62, 571-579.	1.2	102
54	Improved vision after intravenous immunoglobulin in stable demyelinating optic neuritis. Annals of Neurology, 1992, 32, 834-835.	2.8	98

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55	The relative prevalence of dermatomyositis and polymyositis in Europe exhibits a latitudinal gradient. Annals of the Rheumatic Diseases, 2000, 59, 141-142.	0.5	97
56	Difference in distribution of muscle weakness between myasthenia gravis and the Lambert-Eaton myasthenic syndrome. Journal of Neurology, Neurosurgery and Psychiatry, 2002, 73, 766-768.	0.9	97
57	Measurement of the Energy-Generating Capacity of Human Muscle Mitochondria: Diagnostic Procedure and Application to Human Pathology. Clinical Chemistry, 2006, 52, 860-871.	1.5	96
58	Intrinsic Epigenetic Regulation of the D4Z4 Macrosatellite Repeat in a Transgenic Mouse Model for FSHD. PLoS Genetics, 2013, 9, e1003415.	1.5	95
59	Cognitive behavioural therapy with optional graded exercise therapy in patients with severe fatigue with myotonic dystrophy type 1: a multicentre, single-blind, randomised trial. Lancet Neurology, The, 2018, 17, 671-680.	4.9	95
60	Relation between muscle fiber conduction velocity and fiber size in neuromuscular disorders. Journal of Applied Physiology, 2006, 100, 1837-1841.	1.2	93
61	Open-Label Trial of Anti-TNF-Î± in Dermato- and Polymyositis Treated Concomitantly with Methotrexate. European Neurology, 2008, 59, 159-163.	0.6	92
62	Oculopharyngeal muscular dystrophy with limb girdle weakness as major complaint. Journal of Neurology, 2003, 250, 1307-1312.	1.8	91
63	Living with myotonic dystrophy; what can be learned from couples? a qualitative study. BMC Neurology, 2011, 11, 86.	0.8	91
64	Localization of 4q35.2 to the nuclear periphery: is FSHD a nuclear envelope disease?. Human Molecular Genetics, 2004, 13, 1857-1871.	1.4	90
65	Effect of simvastatin in addition to chenodeoxycholic acid in patients with cerebrotendinous xanthomatosis. Metabolism: Clinical and Experimental, 1999, 48, 233-238.	1.5	89
66	The Epidemiology of Neuromuscular Disorders: A Comprehensive Overview of the Literature. Journal of Neuromuscular Diseases, 2015, 2, 73-85.	1.1	89
67	Clinical and serological characteristics of 125 Dutch myositis patients. Journal of Neurology, 2002, 249, 69-75.	1.8	88
68	Experienced and physiological fatigue in neuromuscular disorders. Clinical Neurophysiology, 2007, 118, 292-300.	0.7	88
69	171st ENMC International Workshop: Standards of care and management of facioscapulohumeral muscular dystrophy. Neuromuscular Disorders, 2010, 20, 471-475.	0.3	88
70	Rituximab treatment in patients with refractory inflammatory myopathies. Rheumatology, 2011, 50, 2206-2213.	0.9	88
71	Treatment for idiopathic and hereditary neuralgic amyotrophy (brachial neuritis). The Cochrane Library, 2009, , CD006976.	1.5	87
72	Presence of Diarrhea and Absence of Tendon Xanthomas in Patients With Cerebrotendinous Xanthomatosis. Archives of Neurology, 2000, 57, 520.	4.9	86

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73	Effect of enzyme therapy and prognostic factors in 69 adults with Pompe disease: an open-label single-center study. Orphanet Journal of Rare Diseases, 2012, 7, 73.	1.2	86
74	The cognitive profile of myotonic dystrophy type 1:ÂA systematic review and meta-analysis. Cortex, 2017, 95, 143-155.	1.1	82
75	Autoantibodies directed to novel components of the PM/Scl complex, the human exosome. Arthritis Research, 2002, 4, 134.	2.0	81
76	What's in a name? The clinical features of facioscapulohumeral muscular dystrophy. Practical Neurology, 2016, 16, 201-207.	0.5	81
77	Effect of Mexiletine on Muscle Stiffness in Patients With Nondystrophic Myotonia Evaluated Using Aggregated N-of-1 Trials. JAMA - Journal of the American Medical Association, 2018, 320, 2344.	3.8	81
78	Hepatitis E virus infection and acute non-traumatic neurological injury: A prospective multicentre study. Journal of Hepatology, 2017, 67, 925-932.	1.8	80
79	Protein Complexes in the Archaeon Methanothermobacter thermautotrophicus Analyzed by Blue Native/SDS-PAGE and Mass Spectrometry. Molecular and Cellular Proteomics, 2005, 4, 1653-1663.	2.5	79
80	Is fatigue a disease-specific or generic symptom in chronic medical conditions?. Health Psychology, 2018, 37, 530-543.	1.3	79
81	Clinical and molecular overlap between myopathies and inherited connective tissue diseases. Neuromuscular Disorders, 2008, 18, 843-856.	0.3	76
82	Brain imaging in myotonic dystrophy type 1. Neurology, 2017, 89, 960-969.	1.5	76
83	Treatment of Dermatomyositis and Polymyositis with Anti-Tumor Necrosis Factor-Î±: Long-Term Follow-Up. European Neurology, 2004, 52, 61-63.	0.6	75
84	Clinical phenotype and outcome of hepatitis E virusâ€“associated neuralgic amyotrophy. Neurology, 2017, 89, 909-917.	1.5	75
85	Adding quantitative muscle MRI to the FSHD clinical trial toolbox. Neurology, 2017, 89, 2057-2065.	1.5	72
86	Leukoencephalopathy with swelling in children and adolescents: MRI patterns and differential diagnosis. Neuroradiology, 1995, 37, 679-686.	1.1	71
87	A novel late-onset axial myopathy associated with mutations in the skeletal muscle ryanodine receptor (RYR1) gene. Journal of Neurology, 2013, 260, 1504-1510.	1.8	71
88	Cytosolic 5â€²-nucleotidase 1A autoantibody profile and clinical characteristics in inclusion body myositis. Annals of the Rheumatic Diseases, 2017, 76, 862-868.	0.5	71
89	Genetic determinants of disease severity in the myotonic dystrophy type 1 OPTIMISTIC cohort. Neurology, 2019, 93, e995-e1009.	1.5	71
90	Effects of training and albuterol on pain and fatigue in facioscapulohumeral muscular dystrophy. Journal of Neurology, 2007, 254, 931-940.	1.8	69

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91	Long-Term Pain, Fatigue, and Impairment in Neuralgic Amyotrophy. Archives of Physical Medicine and Rehabilitation, 2009, 90, 435-439.	0.5	69
92	In tandem analysis of CLCN1 and SCN4A greatly enhances mutation detection in families with non-dystrophic myotonia. European Journal of Human Genetics, 2008, 16, 921-929.	1.4	68
93	Ocular and Systemic Manifestations of Cerebrotendinous Xanthomatosis. American Journal of Ophthalmology, 1995, 120, 597-604.	1.7	67
94	Drug treatment for myotonia. The Cochrane Library, 2006, , CD004762.	1.5	66
95	Increased plasticity of the nuclear envelope and hypermobility of telomeres due to the loss of Aâ€“type lamins. Biochimica Et Biophysica Acta - General Subjects, 2010, 1800, 448-458.	1.1	65
96	Acquired neuromyotonia: superiority of plasma exchange over high-dose intravenous human immunoglobulin. Journal of Neurology, 1999, 246, 623-625.	1.8	64
97	Generation of Isogenic D4Z4 Contracted and Noncontracted Immortal Muscle Cell Clones from a Mosaic Patient. American Journal of Pathology, 2012, 181, 1387-1401.	1.9	63
98	Propagation disturbance of motor unit action potentials during transient paresis in generalized myotonia: A high-density surface EMG study. Brain, 2001, 124, 352-360.	3.7	62
99	Balance control in patients with distal versus proximal muscle weakness. Neuroscience, 2009, 164, 1876-1886.	1.1	61
100	Quantitative MRI reveals decelerated fatty infiltration in muscles of active FSHD patients. Neurology, 2016, 86, 1700-1707.	1.5	61
101	Identifying deficits in balance control following vestibular or proprioceptive loss using posturographic analysis of stance tasks. Clinical Neurophysiology, 2008, 119, 2338-2346.	0.7	60
102	High-dose intravenous immunoglobulin treatment in cryptogenic West and Lennox-Gastaut syndrome; an add-on study. European Journal of Pediatrics, 1994, 153, 762-769.	1.3	59
103	TDP-43 accumulation is common in myopathies with rimmed vacuoles. Acta Neuropathologica, 2009, 117, 209-211.	3.9	59
104	Variability in fibre properties in paralysed human quadriceps muscles and effects of training. Pflugers Archiv European Journal of Physiology, 2003, 445, 734-740.	1.3	58
105	Effects of Mindfulness-Based Stress Reduction on the Mental Health of Clinical Clerkship Students: A Cluster-Randomized Controlled Trial. Academic Medicine, 2017, 92, 1012-1021.	0.8	56
106	The mitochondrial 13513G>A mutation is most frequent in Leigh syndrome combined with reduced complex I activity, optic atrophy and/or Wolffâ€“Parkinsonâ€“White. European Journal of Human Genetics, 2007, 15, 155-161.	1.4	55
107	A frameshift mutation in LRSAM1 is responsible for a dominant hereditary polyneuropathy. Human Molecular Genetics, 2012, 21, 358-370.	1.4	55
108	MRI in sarcoglycanopathies: a large international cohort study. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 72-77.	0.9	55

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109	A locus on chromosome 15q for a dominantly inherited nemaline myopathy with core-like lesions. Brain, 2003, 126, 1545-1551.	3.7	54
110	Mitochondrial enzymes discriminate between mitochondrial disorders and chronic fatigue syndrome. Mitochondrion, 2011, 11, 735-738.	1.6	54
111	Mutationâ€specific effects on thin filament length in thin filament myopathy. Annals of Neurology, 2016, 79, 959-969.	2.8	54
112	Quantitative muscle MRI and ultrasound for facioscapulohumeral muscular dystrophy: complementary imaging biomarkers. Journal of Neurology, 2018, 265, 2646-2655.	1.8	54
113	Benzodiazepine Withdrawal Reaction in Two Children following Discontinuation of Sedation with Midazolam. Annals of Pharmacotherapy, 1993, 27, 579-581.	0.9	53
114	Fuel utilization in patients with very long-chain acyl-coa dehydrogenase deficiency. Annals of Neurology, 2004, 56, 279-283.	2.8	53
115	Fatigue is associated with muscle weakness in Ehlers-Danlos syndrome: an explorative study. Physiotherapy, 2011, 97, 170-174.	0.2	53
116	Guidelines on clinical presentation and management of nondystrophic myotonias. Muscle and Nerve, 2020, 62, 430-444.	1.0	53
117	Calcium regulation and muscle disease. Journal of Muscle Research and Cell Motility, 2002, 23, 59-63.	0.9	51
118	Diminished central activation during maximal voluntary contraction in chronic fatigue syndrome. Clinical Neurophysiology, 2004, 115, 2518-2524.	0.7	51
119	Myositis-specific autoantibodies: overview and recent developments. Current Opinion in Rheumatology, 2001, 13, 476-482.	2.0	50
120	Transferrin and Apolipoprotein C-III Isofocusing Are Complementary in the Diagnosis of N- and O-Glycan Biosynthesis Defects. Clinical Chemistry, 2007, 53, 180-187.	1.5	50
121	Gastrointestinal involvement is frequent in Myotonic Dystrophy type 2. Neuromuscular Disorders, 2008, 18, 646-649.	0.3	50
122	Cognitive behaviour therapy plus aerobic exercise training to increase activity in patients with myotonic dystrophy type 1 (DM1) compared to usual care (OPTIMISTIC): study protocol for randomised controlled trial. Trials, 2015, 16, 224.	0.7	49
123	A decline in PABPN1 induces progressive muscle weakness in Oculopharyngeal muscle dystrophy and in muscle aging. Aging, 2013, 5, 412-426.	1.4	49
124	Falls in patients with neuromuscular disorders. Journal of the Neurological Sciences, 2006, 251, 87-90.	0.3	48
125	Sensory Nerve Conduction Studies in Neuralgic Amyotrophy. American Journal of Physical Medicine and Rehabilitation, 2009, 88, 941-946.	0.7	48
126	Correlation analysis of clinical parameters with epigenetic modifications in the DUX4 promoter in FSHD. Epigenetics, 2012, 7, 579-584.	1.3	48

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127	Fatigue, not self-rated motor symptom severity, affects quality of life in functional motor disorders. Journal of Neurology, 2018, 265, 1803-1809.	1.8	48
128	Treatment of the inflammatory myopathies: update and practical recommendations. Expert Opinion on Pharmacotherapy, 2009, 10, 1183-1190.	0.9	47
129	Joint hypermobility as a distinctive feature in the differential diagnosis of myopathies. Journal of Neurology, 2009, 256, 13-27.	1.8	47
130	Quantitative muscle ultrasound versus quantitative magnetic resonance imaging in facioscapulohumeral dystrophy. Muscle and Nerve, 2014, 50, 968-975.	1.0	47
131	Recognizing the tenascinâ€X deficient type of Ehlersâ€“Danlos syndrome: a crossâ€sectional study in 17 patients. Clinical Genetics, 2017, 91, 411-425.	1.0	46
132	Single-cell RNA sequencing in facioscapulohumeral muscular dystrophy disease etiology and development. Human Molecular Genetics, 2019, 28, 1064-1075.	1.4	46
133	Effect of aerobic exercise training and cognitive behavioural therapy on reduction of chronic fatigue in patients with facioscapulohumeral dystrophy: protocol of the FACTS-2-FSHD trial. BMC Neurology, 2010, 10, 56.	0.8	45
134	Scleroderma-polymyositis overlap syndrome versus idiopathic polymyositis and systemic sclerosis: a descriptive study on clinical features and myopathology. Arthritis Research and Therapy, 2014, 16, R111.	1.6	45
135	Possible mechanisms of muscle cramp from temporal and spatial surface EMG characteristics. Journal of Applied Physiology, 2000, 88, 1698-1706.	1.2	44
136	Strength training and aerobic exercise training for muscle disease. The Cochrane Library, 2019, 2019, CD003907.	1.5	44
137	Permanent Loss of Cervical Spinal Cord Function Associated with the Posterior Approach. Anesthesia and Analgesia, 2006, 102, 330-331.	1.1	43
138	Vestibular and proprioceptive influences on trunk movements during quiet standing. Neuroscience, 2009, 161, 904-914.	1.1	43
139	Only fat infiltrated muscles in resting lower leg of FSHD patients show disturbed energy metabolism. NMR in Biomedicine, 2010, 23, 563-568.	1.6	43
140	Poor sleep quality and fatigue but no excessive daytime sleepiness in myotonic dystrophy type 2. Journal of Neurology, Neurosurgery and Psychiatry, 2010, 81, 963-967.	0.9	43
141	Immunoglobulin treatment in epilepsy, a review of the literature. Epilepsy Research, 1994, 19, 181-190.	0.8	42
142	Quantitative near-infrared spectroscopy discriminates between mitochondrial myopathies and normal muscle. Annals of Neurology, 1999, 46, 667-670.	2.8	42
143	Disease Course of Charcot-Marie-Tooth Disease Type 2. Archives of Neurology, 2003, 60, 823.	4.9	42
144	Fasciculation Potentials in High-Density Surface EMG. Journal of Clinical Neurophysiology, 2007, 24, 301-307.	0.9	42

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145	Dysarthria and dysphagia are highly prevalent among various types of neuromuscular diseases. Disability and Rehabilitation, 2014, 36, 1285-1289.	0.9	42
146	HLA class I and II in Lambert-Eaton myasthenic syndrome without associated tumor. Human Immunology, 2001, 62, 809-813.	1.2	40
147	Rhabdomyolysis Caused by an Inherited Metabolic Disease: Very Long-chain Acyl-CoA Dehydrogenase Deficiency. American Journal of Medicine, 2006, 119, 176-179.	0.6	40
148	Needle Electromyographic Findings in 98 Patients with Myositis. European Neurology, 2006, 55, 183-188.	0.6	40
149	Referral of patients with neuromuscular disease to occupational therapy, physical therapy and speech therapy: Usual practice versus multidisciplinary advice. Disability and Rehabilitation, 2007, 29, 717-726.	0.9	40
150	Facioscapulohumeral muscular dystrophy. Current Opinion in Neurology, 2009, 22, 539-542.	1.8	40
151	Postural instability in Charcot-Marie-Tooth type 1A patients is strongly associated with reduced somatosensation. Gait and Posture, 2010, 31, 483-488.	0.6	40
152	Deregulation of the ubiquitin-proteasome system is the predominant molecular pathology in OPMD animal models and patients. Skeletal Muscle, 2011, 1, 15.	1.9	40
153	Sporadic late-onset nemaline myopathy with MGUS. Neurology, 2014, 83, 2133-2139.	1.5	40
154	Prevalence and mutation spectrum of skeletal muscle channelopathies in the Netherlands. Neuromuscular Disorders, 2018, 28, 402-407.	0.3	40
155	Consequences of epigenetic derepression in facioscapulohumeral muscular dystrophy. Clinical Genetics, 2020, 97, 799-814.	1.0	40
156	The Dutch neuromuscular database CRAMP (Computer Registry of All Myopathies and) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 302 Td (Po	0.3	39
157	Different types of fatigue in patients with facioscapulohumeral dystrophy, myotonic dystrophy and HMSN-I. Experienced fatigue and physiological fatigue. Neurological Sciences, 2008, 29, 238-240.	0.9	39
158	Molecular therapy in myotonic dystrophy: focus on RNA gain-of-function. Human Molecular Genetics, 2010, 19, R90-R97.	1.4	39
159	Early onset facioscapulohumeral dystrophy â€“ a systematic review using individual patient data. Neuromuscular Disorders, 2017, 27, 1077-1083.	0.3	39
160	Muscle ultrasound measurements and functional muscle parameters in non-dystrophic myotonias suggest structural muscle changes. Neuromuscular Disorders, 2009, 19, 462-467.	0.3	38
161	Research priorities of patients with neuromuscular disease. Disability and Rehabilitation, 2013, 35, 405-412.	0.9	38
162	Muscle MRI in a large cohort of patients with oculopharyngeal muscular dystrophy. Journal of Neurology, Neurosurgery and Psychiatry, 2019, 90, 576-585.	0.9	38

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163	High specificity of myositis specific autoantibodies for myositis compared with other neuromuscular disorders. Journal of Neurology, 2005, 252, 534-537.	1.8	36
164	Psychiatric disorders appear equally in patients with myotonic dystrophy, facioscapulohumeral dystrophy, and hereditary motor and sensory neuropathy type I. Acta Neurologica Scandinavica, 2007, 115, 265-270.	1.0	36
165	Sarcomeric dysfunction contributes to muscle weakness in facioscapulohumeral muscular dystrophy. Neurology, 2013, 80, 733-737.	1.5	36
166	Second IVIg course in Guillainâ€BarrÃ© syndrome patients with poor prognosis (SIDâ€GBS trial): Protocol for a doubleâ€blind randomized, placeboâ€controlled clinical trial. Journal of the Peripheral Nervous System, 2018, 23, 210-215.	1.4	36
167	Decreased immunoglobulin class switching in nijmegen breakage syndrome due to the DNA repair defect. Human Immunology, 2001, 62, 1324-1327.	1.2	35
168	Brody syndrome: A clinically heterogeneous entity distinct from Brody disease. Neuromuscular Disorders, 2012, 22, 944-954.	0.3	35
169	The Radboud Dysarthria Assessment: Development and Clinimetric Evaluation. Folia Phoniatrica Et Logopaedica, 2017, 69, 143-153.	0.5	35
170	Myositis specific autoantibodies: changing insights in pathophysiology and clinical associations. Current Opinion in Rheumatology, 2004, 16, 692-9.	2.0	35
171	Idiopathic Neuralgic Amyotrophy in Children. A Distinct Phenotype Compared to the Adult Form. Neuropediatrics, 2000, 31, 328-332.	0.3	34
172	Lower extremity muscle pathology in myotonic dystrophy type 1 assessed by quantitative MRI. Neurology, 2019, 92, e2803-e2814.	1.5	34
173	Second intravenous immunoglobulin dose in patients with Guillain-BarrÃ© syndrome with poor prognosis (SID-GBS): a double-blind, randomised, placebo-controlled trial. Lancet Neurology, The, 2021, 20, 275-283.	4.9	34
174	Muscle-fiber conduction velocity and electromyography as diagnostic tools in patients with suspected inflammatory myopathy: A prospective study. Muscle and Nerve, 2004, 29, 46-50.	1.0	33
175	Titin-based stiffening of muscle fibers in Ehlers-Danlos Syndrome. Journal of Applied Physiology, 2012, 112, 1157-1165.	1.2	33
176	Skeletal muscle imaging in facioscapulohumeral muscular dystrophy, pattern and asymmetry of individual muscle involvement. Neuromuscular Disorders, 2014, 24, 1087-1096.	0.3	33
177	A family-based study into penetrance in facioscapulohumeral muscular dystrophy type 1. Neurology, 2018, 91, e444-e454.	1.5	33
178	Central adaptations during repetitive contractions assessed by the readiness potential. European Journal of Applied Physiology, 2006, 97, 521-526.	1.2	32
179	Reduced quantitative muscle function in tenascin-X deficient Ehlers-Danlos patients. Neuromuscular Disorders, 2007, 17, 597-602.	0.3	32
180	Autoantibodies to Cytosolic 5â€²-Nucleotidase 1A in Primary SjÃ¶grenâ€™s Syndrome and Systemic Lupus Erythematosus. Frontiers in Immunology, 2018, 9, 1200.	2.2	32

#	Article	IF	Citations
181	Panel-Based Exome Sequencing for Neuromuscular Disorders as a Diagnostic Service. Journal of Neuromuscular Diseases, 2019, 6, 241-258.	1.1	32
182	Proteasome-mediated degradation of integral inner nuclear membrane protein emerin in fibroblasts lacking A-type lamins. Biochemical and Biophysical Research Communications, 2006, 351, 1011-1017.	1.0	31
183	Health status in non-dystrophic myotonias: close relation with pain and fatigue. Journal of Neurology, 2009, 256, 939-947.	1.8	31
184	PGM1 deficiency: Substrate use during exercise and effect of treatment with galactose. Neuromuscular Disorders, 2017, 27, 370-376.	0.3	31
185	Llama-derived phage display antibodies in the dissection of the human disease oculopharyngeal muscular dystrophy. Journal of Immunological Methods, 2003, 279, 149-161.	0.6	30
186	Eosinophilic fasciitis in a child mimicking a myopathy. Neuromuscular Disorders, 2006, 16, 144-148.	0.3	30
187	Recurrent neuropathy associated with Ehlersâ€“Danlos syndrome. Journal of Neurology, 2006, 253, 670-671.	1.8	30
188	Strength training and aerobic exercise training for muscle disease. , 2010, , CD003907.		30
189	Increased fascial thickness of the deltoid muscle in dermatomyositis and polymyositis: An ultrasound study. Muscle and Nerve, 2015, 52, 534-539.	1.0	30
190	Genetic defects in the hexosamine and sialic acid biosynthesis pathway. Biochimica Et Biophysica Acta - General Subjects, 2016, 1860, 1640-1654.	1.1	30
191	Early onset as a marker for disease severity in facioscapulohumeral muscular dystrophy. Neurology, 2019, 92, e378-e385.	1.5	30
192	The Effect of Coenzyme Q10 Therapy in Parkinson Disease Could Be Symptomatic. Archives of Neurology, 2003, 60, 1170.	4.9	30
193	A new phenotype of autosomal dominant nemaline myopathy. Neuromuscular Disorders, 2002, 12, 13-18.	0.3	29
194	Statin-disclosed acid maltase deficiency. Journal of Internal Medicine, 2005, 258, 196-197.	2.7	29
195	BSCL2 mutations in two Dutch families with overlapping Silver syndrome-distal hereditary motor neuropathy. Neuromuscular Disorders, 2006, 16, 122-125.	0.3	29
196	Dysphagia is present but mild in myotonic dystrophy type 2. Neuromuscular Disorders, 2009, 19, 196-198.	0.3	29
197	Disease impact in chronic progressive external ophthalmoplegia: More than meets the eye. Neuromuscular Disorders, 2011, 21, 272-278.	0.3	29
198	SERCA1 protein expression in muscle of patients with Brody disease and Brody syndrome and in cultured human muscle fibers. Molecular Genetics and Metabolism, 2013, 110, 162-169.	0.5	29

#	Article	IF	Citations
199	Reference values of maximum performance tests of speech production. International Journal of Speech-Language Pathology, 2019, 21, 56-64.	0.6	29
200	Employment status of patients with neuromuscular diseases in relation to personal factors, fatigue and health status: A secondary analysis. Journal of Rehabilitation Medicine, 2010, 42, 60-65.	0.8	28
201	Muscle characteristics and altered myofascial force transmission in tenascin-X-deficient mice, a mouse model of Ehlers-Danlos syndrome. Journal of Applied Physiology, 2010, 109, 986-995.	1.2	28
202	High Frequency of Neuropathic Pain in Ehlers-Danlos Syndrome: An Association with Axonal Polyneuropathy and Compression Neuropathy?. Journal of Pain and Symptom Management, 2011, 41, e4-e6.	0.6	28
203	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
204	No effect of folic acid and methionine supplementation on D4Z4 methylation in patients with facioscapulohumeral muscular dystrophy. Neuromuscular Disorders, 2006, 16, 766-769.	0.3	27
205	Cis D4Z4 repeat duplications associated with facioscapulohumeral muscular dystrophy type 2. Human Molecular Genetics, 2018, 27, 3488-3497.	1.4	27
206	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
207	Frequent occurrence of anti-tRNAHis autoantibodies that recognize a conformational epitope in sera of patients with myositis. Arthritis and Rheumatism, 1998, 41, 1428-1437.	6.7	26
208	Molecular Analysis of Myophosphorylase Deficiency in Dutch Patients with McArdle's Disease. Annals of Human Genetics, 2004, 68, 17-22.	0.3	26
209	High disease impact of myotonic dystrophy type 2 on physical and mental functioning. Journal of Neurology, 2011, 258, 1820-1826.	1.8	26
210	Mild Muscular Features in Tenascin-X Knockout Mice, A Model of Ehlersâ€“Danlos Syndrome. Connective Tissue Research, 2011, 52, 422-432.	1.1	26
211	Paediatric cerebrotendinous xanthomatosis. Journal of Inherited Metabolic Disease, 1992, 15, 374-376.	1.7	25
212	Oculopharyngeal muscular dystrophy (OPMD) due to a small duplication in the PABPN1 gene. Human Mutation, 2003, 21, 553-553.	1.1	25
213	The relation between daytime sleepiness, fatigue, and reduced motivation in patients with adult onset myotonic dystrophy. Journal of Neurology, Neurosurgery and Psychiatry, 2003, 74, 138-139.	0.9	25
214	Polymyositis, invasion of non-necrotic muscle fibres, and the art of repetition. BMJ: British Medical Journal, 2004, 329, 1464-1467.	2.4	25
215	Muscle 3243Aâ†'G mutation load and capacity of the mitochondrial energyâ€generating system. Annals of Neurology, 2008, 63, 473-481.	2.8	25
216	Symptomatic lipid storage in carriers for the PNPLA2 gene. European Journal of Human Genetics, 2013, 21, 807-815.	1.4	25

#	Article	IF	Citations
217	Shared medical appointments improve QOL in neuromuscular patients. Neurology, 2014, 83, 240-246.	1.5	25
218	Phenotypeâ€genotype relations in facioscapulohumeral muscular dystrophy type 1. Clinical Genetics, 2018, 94, 521-527.	1.0	25
219	Effects of weakness of orofacial muscles on swallowing and communication in FSHD. Neurology, 2019, 92, e957-e963.	1.5	25
220	Correlation Between Quantitative MRI and Muscle Histopathology in Muscle Biopsies from Healthy Controls and Patients with IBM, FSHD and OPMD. Journal of Neuromuscular Diseases, 2020, 7, 495-504.	1.1	25
221	KBTBD13 is an actin-binding protein that modulates muscle kinetics. Journal of Clinical Investigation, 2020, 130, 754-767.	3.9	25
222	Classification of employment factors according to the International Classification of Functioning, Disability and Health in patients with neuromuscular diseases: A systematic review. Disability and Rehabilitation, 2009, 31, 2150-2163.	0.9	24
223	Feverâ€induced recurrent rhabdomyolysis due to a novel mutation in the ryanodine receptor type 1 gene. Internal Medicine Journal, 2014, 44, 819-820.	0.5	24
224	FSHD type 2 and Bosma arhinia microphthalmia syndrome. Neurology, 2018, 91, e562-e570.	1.5	24
225	MRI-Guided Biopsy as a Tool for Diagnosis and Research of Muscle Disorders. Journal of Neuromuscular Diseases, 2018, 5, 315-319.	1.1	24
226	Skeletal muscle of mice with a mutation in slow Î±-tropomyosin is weaker at lower lengths. Neuromuscular Disorders, 2002, 12, 952-957.	0.3	23
227	Concomitant dermatomyositis and myasthenia gravis presenting with respiratory insufficiency. Muscle and Nerve, 2002, 25, 293-296.	1.0	23
228	Comparison of CMT1A and CMT2: similarities and differences. Journal of Neurology, 2006, 253, 1572-1580.	1.8	23
229	Skeletal muscle involvement in myotonic dystrophy type 2. A comparative muscle ultrasound study. Neuromuscular Disorders, 2012, 22, 492-499.	0.3	23
230	Trunk muscle involvement is most critical for the loss of balance control in patients with facioscapulohumeral muscular dystrophy. Clinical Biomechanics, 2014, 29, 855-860.	0.5	23
231	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
232	Affective symptoms and apathy in myotonic dystrophy type 1 a systematic review and meta-analysis. Journal of Affective Disorders, 2019, 250, 260-269.	2.0	23
233	Self-management program improves participation in patients with neuromuscular disease. Neurology, 2019, 93, e1720-e1731.	1.5	23
234	Structural white matter networks in myotonic dystrophy type 1. NeuroImage: Clinical, 2019, 21, 101615.	1.4	23

#	Article	IF	Citations
235	Muscle ultrasound is a responsive biomarker in facioscapulohumeral dystrophy. Neurology, 2020, 94, e1488-e1494.	1.5	23
236	Neuromuscular features in Marfan syndrome. Clinical Genetics, 2009, 76, 25-37.	1.0	22
237	Isolated eyelid closure myotonia in two families with sodium channel myotonia. Neurogenetics, 2010, 11, 257-260.	0.7	22
238	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
239	Clinical, morphological and genetic characterization of Brody disease: an international study of 40 patients. Brain, 2020, 143, 452-466.	3.7	22
240	Overweight Is an Independent Risk Factor for Reduced Lung Volumes in Myotonic Dystrophy Type 1. PLoS ONE, 2016, 11, e0152344.	1.1	22
241	Sensorineural Hearing Impairment in Patients with Pmp22 Duplication, Deletion, and Frameshift Mutations. Otology and Neurotology, 2005, 26, 405-414.	0.7	21
242	Early diagnosis of ALS: The search for signs of denervation in clinically normal muscles. Journal of the Neurological Sciences, 2007, 263, 154-157.	0.3	21
243	Contribution of central and peripheral factors to residual fatigue in Guillainâ€“BarrÃ© syndrome. Muscle and Nerve, 2007, 36, 93-99.	1.0	21
244	Cell Membrane Integrity in Myotonic Dystrophy Type 1: Implications for Therapy. PLoS ONE, 2015, 10, e0121556.	1.1	21
245	Patients with Systemic Sclerosis/polymyositis Overlap Have a Worse Survival Rate Than Patients Without It. Journal of Rheumatology, 2016, 43, 1838-1843.	1.0	21
246	Lifetime endogenous estrogen exposure and disease severity in female patients with facioscapulohumeral muscular dystrophy. Neuromuscular Disorders, 2018, 28, 508-511.	0.3	21
247	Facioscapulohumeral Dystrophy in Childhood: A Nationwide Natural History Study. Annals of Neurology, 2018, 84, 627-637.	2.8	21
248	Human brain pathology in myotonic dystrophy type 1: A systematic review. Neuropathology, 2021, 41, 3-20.	0.7	21
249	<title>Validation of measurement protocols to assess oxygen consumption and blood flow in the human forearm by near-infrared spectroscopy</title>. , 1998, 3194, 133.		20
250	225th ENMC international workshop:. Neuromuscular Disorders, 2017, 27, 782-790.	0.3	20
251	Functional impairments, fatigue and quality of life in RYR1-related myopathies: A questionnaire study. Neuromuscular Disorders, 2019, 29, 30-38.	0.3	20
252	Phase 1 clinical trial of losmapimod in facioscapulohumeral dystrophy: Safety, tolerability, pharmacokinetics, and target engagement. British Journal of Clinical Pharmacology, 2021, 87, 4658-4669.	1.1	20

#	Article	IF	Citations
253	Involuntary painful muscle contractions in Satoyoshi syndrome: A surface electromyographic study. Movement Disorders, 2006, 21, 2015-2018.	2.2	19
254	Falls and resulting fractures in Myotonic Dystrophy: Results from a multinational retrospective survey. Neuromuscular Disorders, 2018, 28, 229-235.	0.3	19
255	Qualitative and Quantitative Aspects of Pain in Patients With Myotonic Dystrophy Type 2. Journal of Pain, 2018, 19, 920-930.	0.7	19
256	Eight years after an international workshop on myotonic dystrophy patient registries: case study of a global collaboration for a rare disease. Orphanet Journal of Rare Diseases, 2018, 13, 155.	1.2	19
257	Serologie HLA typing in cryptogenic Lennox-Gastaut syndrome. Epilepsy Research, 1994, 17, 43-47.	0.8	18
258	A double missense mutation in the ATM gene of a Dutch family with ataxia telangiectasia. Human Genetics, 1998, 102, 187-191.	1.8	18
259	Guillainâ€“BarrÃ© syndrome as presenting feature in a patient with lupus nephritis, with complete resolution after cyclophosphamide treatment. Nephrology Dialysis Transplantation, 2001, 16, 840-842.	0.4	18
260	Proteomics and neuromuscular diseases: theoretical concept and first results. Annals of Clinical Biochemistry, 2003, 40, 9-15.	0.8	18
261	Na+-K+-ATPase is not involved in the warming-up phenomenon in generalized myotonia. Muscle and Nerve, 2006, 33, 514-523.	1.0	18
262	Development of a tool to guide referral of patients with neuromuscular disorders to allied health services. Part one. Disability and Rehabilitation, 2008, 30, 855-862.	0.9	18
263	Increased plasma amyloid-Î²42 protein in sporadic inclusion body myositis. Acta Neuropathologica, 2009, 118, 429-431.	3.9	18
264	Child Neurology: Differential diagnosis of a low CSF glucose in children and young adults. Neurology, 2013, 81, e178-81.	1.5	18
265	N-of-1 Trials: Evidence-Based Clinical Care or Medical Research that Requires IRB Approval? A Practical Flowchart Based on an Ethical Framework. Healthcare (Switzerland), 2020, 8, 49.	1.0	18
266	Electrocardiographic predictors of infrahissian conduction disturbances in myotonic dystrophy type 1. Europace, 2021, 23, 298-304.	0.7	18
267	Chromosome 10q-linked FSHD identifies DUX4 as principal disease gene. Journal of Medical Genetics, 2022, 59, 180-188.	1.5	18
268	High prevalence of incomplete right bundle branch block in facioscapulohumeral muscular dystrophy without cardiac symptoms. Functional Neurology, 2014, 29, 159-65.	1.3	18
269	Muscle Uridine Diphosphate-Hexosamines Do Not Decrease Despite Correction of Hyperglycemia-Induced Insulin Resistance in Type 2 Diabetes. Journal of Clinical Endocrinology and Metabolism, 2002, 87, 5179-5184.	1.8	17
270	Muscle slowness in a family with nemaline myopathy. Neuromuscular Disorders, 2006, 16, 477-480.	0.3	17

#	Article	IF	Citations
271	Sleep disturbances in chronic progressive external ophthalmoplegia. European Journal of Neurology, 2012, 19, 176-178.	1.7	17
272	Combined N-of-1 trials to investigate mexiletine in non-dystrophic myotonia using a Bayesian approach; study rationale and protocol. BMC Neurology, 2015, 15, 43.	0.8	17
273	The epidemiology of neuromuscular disorders: Age at onset and gender in the Netherlands. Neuromuscular Disorders, 2016, 26, 447-452.	0.3	17
274	Involvement of pelvic girdle and proximal leg muscles in early oculopharyngeal muscular dystrophy. Neuromuscular Disorders, 2017, 27, 1099-1105.	0.3	17
275	Insulin Signaling as a Key Moderator in Myotonic Dystrophy Type 1. Frontiers in Neurology, 2019, 10, 1229.	1.1	17
276	Short fatigue questionnaire: Screening for severe fatigue.. Journal of Psychosomatic Research, 2020, 137, 110229.	1.2	17
277	White matter abnormalities in congenital muscular dystrophy. Journal of the Neurological Sciences, 1995, 129, 162-169.	0.3	16
278	Nature and frequency of respiratory involvement in chronic progressive external ophthalmoplegia. Journal of Neurology, 2011, 258, 2020-2025.	1.8	16
279	Autoantibody testing in idiopathic inflammatory myopathies. Practical Neurology, 2019, 19, 284-294.	0.5	16
280	Deep phenotyping of facioscapulohumeral muscular dystrophy type 2 by magnetic resonance imaging. European Journal of Neurology, 2020, 27, 2604-2615.	1.7	16
281	Congenital fibre type disproportion a time-locked diagnosis: A clinical and morphological follow-up study. Clinical Neurology and Neurosurgery, 2000, 102, 97-101.	0.6	15
282	Diagnosis and Differential Diagnosis of Muscle Cramps: A Clinical Approach. Journal of Clinical Neuromuscular Disease, 2002, 4, 89-94.	0.3	15
283	Nuclear entrapment and extracellular depletion of PCOLCE is associated with muscle degeneration in oculopharyngeal muscular dystrophy. BMC Neurology, 2013, 13, 70.	0.8	15
284	How Persons with a Neuromuscular Disease Perceive Employment Participation: A Qualitative Study. Journal of Occupational Rehabilitation, 2014, 24, 52-67.	1.2	15
285	Effectiveness and cost-effectiveness of a self-management group program to improve social participation in patients with neuromuscular disease and chronic fatigue: protocol of the Energetic study. BMC Neurology, 2015, 15, 58.	0.8	15
286	Facioscapulohumeral dystrophy in children: design of a prospective, observational study on natural history, predictors and clinical impact (iFocus FSHD). BMC Neurology, 2016, 16, 138.	0.8	15
287	The Dutch patients' perspective on oculopharyngeal muscular dystrophy: A questionnaire study on fatigue, pain and impairments. Neuromuscular Disorders, 2016, 26, 221-226.	0.3	15
288	Electrical impedance myography in facioscapulohumeral muscular dystrophy: A 1â€year followâ€up study. Muscle and Nerve, 2018, 58, 213-218.	1.0	15

#	Article	IF	Citations
289	Continued misuse of orphan drug legislation: a life-threatening risk for mexiletine. European Heart Journal, 2020, 41, 614-617.	1.0	15
290	Nijmegen Breakage Syndrome: A Neuropathological Study. Neuropediatrics, 2003, 34, 189-193.	0.3	14
291	A case of neuromuscular mimicry. Neuromuscular Disorders, 2006, 16, 510-513.	0.3	14
292	Development of a tool to guide referral of patients with neuromuscular disorders to allied health services. Part two. Disability and Rehabilitation, 2008, 30, 863-870.	0.9	14
293	Neuromuscular properties of the thigh muscles in patients with ehlersâ€“danlos syndrome. Muscle and Nerve, 2013, 47, 96-104.	1.0	14
294	Respiratory function in facioscapulohumeral muscular dystrophy 1. Neuromuscular Disorders, 2017, 27, 526-530.	0.3	14
295	Ophthalmological findings in facioscapulohumeral dystrophy. Brain Communications, 2019, 1, fcz023.	1.5	14
296	Quantitative near-infrared spectroscopy discriminates between mitochondrial myopathies and normal muscle. Annals of Neurology, 1999, 46, 667-70.	2.8	14
297	Facioscapulohumeral dystrophy transcriptome signatures correlate with different stages of disease and are marked by different MRI biomarkers. Scientific Reports, 2022, 12, 1426.	1.6	14
298	Light chain ratios and concentrations of serum immunoglobulins in children with epilepsy. Epilepsy Research, 1992, 13, 255-260.	0.8	13
299	Warm-up phenomenon in myotonia associated with the V445M sodium channel mutation. Journal of Neurology, 2007, 254, 257-258.	1.8	13
300	Implementation of multidisciplinary advice to allied health care professionals regarding the management of their patients with neuromuscular diseases. Disability and Rehabilitation, 2011, 33, 787-795.	0.9	13
301	184th ENMC international workshop: Pain and fatigue in neuromuscular disorders. Neuromuscular Disorders, 2013, 23, 1028-1032.	0.3	13
302	Characterizing the face in facioscapulohumeral muscular dystrophy. Journal of Neurology, 2021, 268, 1342-1350.	1.8	13
303	Rasch analysis to evaluate the motor function measure for patients with facioscapulohumeral muscular dystrophy. International Journal of Rehabilitation Research, 2021, 44, 38-44.	0.7	13
304	Enhancement of central nervous system remyelination in immune and non-immune experimental models of demyelination. Multiple Sclerosis Journal, 1997, 3, 76-79.	1.4	12
305	Familial inclusion body myositis with histologically confirmed sensorimotor axonal neuropathy. Journal of Neurology, 2000, 247, 882-884.	1.8	12
306	Proteomics Approaches to Study Genetic and Metabolic Disorders. Journal of Proteome Research, 2007, 6, 506-512.	1.8	12

#	Article	IF	Citations
307	Optimizing referral of patients with neuromuscular disorders to allied health care. European Journal of Neurology, 2009, 16, 562-568.	1.7	12
308	Screening for antecedent Campylobacter jejuni infections and antiâ€ganglioside antibodies in idiopathic neuralgic amyotrophy. Journal of the Peripheral Nervous System, 2011, 16, 153-156.	1.4	12
309	Speech Pathology Interventions in Patients with Neuromuscular Diseases: A Systematic Review. Folia Phoniatrica Et Logopaedica, 2011, 63, 15-20.	0.5	12
310	Determining the role of sarcomeric proteins in facioscapulohumeral muscular dystrophy: a study protocol. BMC Neurology, 2013, 13, 144.	0.8	12
311	The individualized neuromuscular quality of life questionnaire: Cultural translation and psychometric validation for the Dutch population. Muscle and Nerve, 2015, 51, 496-500.	1.0	12
312	Diagnostics of short tandem repeat expansion variants using massively parallel sequencing and componential tools. European Journal of Human Genetics, 2019, 27, 400-407.	1.4	12
313	Swallowing, Chewing and Speaking: Frequently Impaired in Oculopharyngeal Muscular Dystrophy. Journal of Neuromuscular Diseases, 2020, 7, 1-12.	1.1	12
314	Natural history, outcome measures and trial readiness in LAMA2-related muscular dystrophy and SELENON-related myopathy in children and adults: protocol of the LAST STRONG study. BMC Neurology, 2021, 21, 313.	0.8	12
315	Muscle function in a patient with Brody's disease. , 1999, 22, 704-711.		11
316	The Regulation of Fine Movements in Patients with Charcot Marie Tooth, Type la: Some Ideas about Continuous Adaptation. Motor Control, 2001, 5, 200-214.	0.3	11
317	Influence of Relatives on Fatigue Experienced by Patients with Facioscapulohumeral Dystrophy, Myotonic Dystrophy and HMSN-I. European Neurology, 2006, 56, 24-30.	0.6	11
318	The wrong end of the telescope: neuromuscular mimics of movement disorders (and vice versa). Practical Neurology, 2016, 16, 264-269.	0.5	11
319	Hearing impairment in patients with myotonic dystrophy type 2. Neurology, 2018, 90, e615-e622.	1.5	11
320	Specific muscle strength is reduced in facioscapulohumeral dystrophy: An MRI based musculoskeletal analysis. Neuromuscular Disorders, 2018, 28, 238-245.	0.3	11
321	Health-Related Quality of Life in Patients with Adult-Onset Myotonic Dystrophy Type 1: A Systematic Review. Patient, 2019, 12, 365-373.	1.1	11
322	Quantitative Muscle MRI Depicts Increased Muscle Mass after a Behavioral Change in Myotonic Dystrophy Type 1. Radiology, 2020, 297, 132-142.	3.6	11
323	Systemic cell therapy for muscular dystrophies. Stem Cell Reviews and Reports, 2021, 17, 878-899.	1.7	11
324	Longitudinal Assessment of Strength, Functional Capacity, Oropharyngeal Function, and Quality of Life in Oculopharyngeal Muscular Dystrophy. Neurology, 2021, 97, e1475-e1483.	1.5	11

#	Article	IF	Citations
325	Familial adult-onset muscular dystrophy with leukoencephalopathy. Annals of Neurology, 1992, 32, 577-580.	2.8	10
326	The effect of dantrolene sodium in Very Long Chain Acyl-CoA Dehydrogenase Deficiency. Neuromuscular Disorders, 2005, 15, 844-846.	0.3	10
327	Radicular dysfunction due to spinal deformities in Marfan syndrome at older age: Three case reports. European Journal of Medical Genetics, 2010, 53, 35-39.	0.7	10
328	Effects of shared medical appointments on quality of life and cost-effectiveness for patients with a chronic neuromuscular disease. Study protocol of a randomized controlled trial. BMC Neurology, 2011, 11, 106.	0.8	10
329	Integrating clinical and genetic observations in facioscapulohumeral muscular dystrophy. Current Opinion in Neurology, 2016, 29, 606-613.	1.8	10
330	The yield of diagnostic work-up of patients presenting with myalgia, exercise intolerance, or fatigue. Neuromuscular Disorders, 2017, 27, 243-250.	0.3	10
331	High incidence of falls in patients with myotonic dystrophy type 1 and 2: A prospective study. Neuromuscular Disorders, 2019, 29, 758-765.	0.3	10
332	Altered sensorimotor representations after recovery from peripheral nerve damage in neuralgic amyotrophy. Cortex, 2020, 127, 180-190.	1.1	10
333	Associations Between Variant Repeat Interruptions and Clinical Outcomes in Myotonic Dystrophy Type 1. Neurology: Genetics, 2021, 7, e572.	0.9	10
334	Cerebrospinal fluid examinations in cryptogenic West and Lennox-Gastaut syndrome before and after intravenous immunoglobulin administration. Epilepsy Research, 1994, 18, 139-147.	0.8	9
335	Quadriceps weakness in a family with nemaline myopathy: influence of knee angle. Clinical Science, 2003, 105, 585-589.	1.8	9
336	Warming up improves speech production in patients with adult onset myotonic dystrophy. Journal of Communication Disorders, 2007, 40, 185-195.	0.8	9
337	The Evidence for Occupational Therapy for Adults with Neuromuscular Diseases: A Systematic Review. OTJR Occupation, Participation and Health, 2008, 28, 12-18.	0.4	9
338	TDP-43 plasma levels do not differentiate sporadic inclusion body myositis from other inflammatory myopathies. Acta Neuropathologica, 2010, 120, 825-826.	3.9	9
339	Primary Cataract as a Key to Recognition of Myotonic Dystrophy Type 1. European Journal of Ophthalmology, 2015, 25, e46-e49.	0.7	9
340	Myotonic discharges discriminate chloride from sodium muscle channelopathies. Neuromuscular Disorders, 2015, 25, 73-80.	0.3	9
341	NA-CONTROL: a study protocol for a randomised controlled trial to compare specific outpatient rehabilitation that targets cerebral mechanisms through relearning motor control and uses self-management strategies to improve functional capability of the upper extremity, to usual care in patients with neuralgic amyotrophy. Trials, 2019, 20, 482.	0.7	9
342	Age-Associated Salivary MicroRNA Biomarkers for Oculopharyngeal Muscular Dystrophy. International Journal of Molecular Sciences, 2020, 21, 6059.	1.8	9

#	Article	IF	Citations
343	Reduced specific force in patients with mild and severe facioscapulohumeral muscular dystrophy. Muscle and Nerve, 2021, 63, 60-67.	1.0	9
344	Quantitative Measurement of Oxygen Consumption and Forearm Blood Flow in Patients with Mitochondrial Myopathies. Advances in Experimental Medicine and Biology, 1999, 471, 313-319.	0.8	9
345	Postural changes, dysphagia, and systemic sclerosis. Annals of the Rheumatic Diseases, 1998, 57, 331-338.	0.5	8
346	Upregulation of Ca²⁺removal in human skeletal muscle: a possible role for Ca²⁺-dependent priming of mitochondrial ATP synthesis. American Journal of Physiology - Cell Physiology, 2003, 285, C1263-C1269.	2.1	8
347	Gait propulsion in patients with facioscapulohumeral muscular dystrophy and ankle plantarflexor weakness. Gait and Posture, 2015, 41, 476-481.	0.6	8
348	A 22-year follow-up reveals a variable disease severity in early-onset facioscapulohumeral dystrophy. European Journal of Paediatric Neurology, 2018, 22, 782-785.	0.7	8
349	Swallowing, Chewing and Speaking: Frequently Impaired in Oculopharyngeal Muscular Dystrophy. Journal of Neuromuscular Diseases, 2020, 7, 483-494.	1.1	8
350	Preserved single muscle fiber specific force in facioscapulohumeral muscular dystrophy. Neurology, 2020, 94, e1157-e1170.	1.5	8
351	The facioscapulohumeral muscular dystrophy Raschâ€built overall disability scale (FSHDâ€RODS). European Journal of Neurology, 2021, 28, 2339-2348.	1.7	8
352	The neuromuscular and multisystem features of RYR1-related malignant hyperthermia and rhabdomyolysis. Medicine (United States), 2021, 100, e26999.	0.4	8
353	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
354	Are muscle cramps in Isaacs' syndrome triggered by human immunoglobulin?. Journal of Neurology, Neurosurgery and Psychiatry, 1995, 58, 393-393.	0.9	7
355	Maintaining Constant Voluntary Force in Generalized Myotonia Despite Muscle Membrane Disturbances: Insights from a High-Density Surface EMG Study. Journal of Clinical Neurophysiology, 2004, 21, 114-123.	0.9	7
356	Fast Responses to Stepping on an Unexpected Surface Height Depend on Intact Large-Diameter Nerve Fibers: A Study on Charcotâ€“Marieâ€“Tooth Type 1A Disease. Journal of Neurophysiology, 2009, 102, 1684-1698.	0.9	7
357	Dutch myotonic dystrophy type 2 patients and a North-African DM2 family carry the common European founder haplotype. European Journal of Human Genetics, 2011, 19, 567-570.	1.4	7
358	Cost-effectiveness of shared medical appointments for neuromuscular patients. Neurology, 2015, 85, 619-625.	1.5	7
359	Clinical practice considerations in facioscapulohumeral muscular dystrophy Sydney, Australia, 21 September 2015. Neuromuscular Disorders, 2016, 26, 462-471.	0.3	7
360	The Position of Neuromuscular Patients in Shared Decision Making. Report from the 235th ENMC Workshop: Milan, Italy, January 19-20, 2018. Journal of Neuromuscular Diseases, 2019, 6, 161-172.	1.1	7

#	Article	IF	Citations
361	Clinical, genetic, and histological features of centronuclear myopathy in the Netherlands. Clinical Genetics, 2021, 100, 692-702.	1.0	7
362	N-of-1 Trials in Neurology. Neurology, 2022, 98, .	1.5	7
363	Natural History of Facioscapulohumeral Dystrophy in Children. Neurology, 2021, 97, e2103-e2113.	1.5	7
364	Respiratory muscle function in patients with nemaline myopathy. Neuromuscular Disorders, 2022, 32, 654-663.	0.3	7
365	Tubular Aggregates. Archives of Neurology, 1999, 56, 1410.	4.9	6
366	Diagnostic yield of muscle fibre conduction velocity in myopathies. Journal of the Neurological Sciences, 2011, 309, 40-44.	0.3	6
367	Dynamic stability during level walking and obstacle crossing in persons with facioscapulohumeral muscular dystrophy. Gait and Posture, 2015, 42, 295-300.	0.6	6
368	Characterization of sarcoplasmic reticulum Ca2+ ATPase pumps in muscle of patients with myotonic dystrophy and with hypothyroid myopathy. Neuromuscular Disorders, 2016, 26, 378-385.	0.3	6
369	New Insights in Adherence and Survival in Myotonic Dystrophy Patients Using Home Mechanical Ventilation. Respiration, 2021, 100, 154-163.	1.2	6
370	Profiling Serum Antibodies Against Muscle Antigens in Facioscapulohumeral Muscular Dystrophy Finds No Disease-Specific Autoantibodies. Journal of Neuromuscular Diseases, 2021, 8, 801-814.	1.1	6
371	Near-infrared spectroscopy in chronic progressive external ophthalmoplegia: Adipose tissue thickness confounds decreased muscle oxygen consumption. Annals of Neurology, 2002, 51, 272-273.	2.8	5
372	Persistent increased risk for thymoma in myasthenia gravis associated with myositis. Muscle and Nerve, 2006, 34, 251-252.	1.0	5
373	Ambulatory disabilities and the use of walking aids in patients with Hereditary Motor and Sensory Neuropathy type I (HMSN I). Disability and Rehabilitation: Assistive Technology, 2007, 2, 35-41.	1.3	5
374	Caffeine and Muscle Cramps: A Stimulating Connection. American Journal of Medicine, 2007, 120, e1-e2.	0.6	5
375	Chronic progressive external ophthalmoplegia caused by an m.4267A > G mutation in the mitochondrial tRNAIle. Journal of Neurology, 2007, 254, 1614-1615.	1.8	5
376	The phenotype of the Gly94fsX222 PMP22 insertion. Journal of the Peripheral Nervous System, 2011, 16, 113-118.	1.4	5
377	No relevant excess prevalence of myotonic dystrophy type 2 in patients with suspected fibromyalgia syndrome. Neuromuscular Disorders, 2016, 26, 370-373.	0.3	5
378	Ultrasound: A Potential Tool for Detecting of Fasciitis in Dermatomyositis and Polymyositis. Journal of Rheumatology, 2018, 45, 441.1-442.	1.0	5

#	Article	IF	Citations
379	Repeatability and reliability of muscle relaxation properties induced by motor cortical stimulation. Journal of Applied Physiology, 2018, 124, 1597-1604.	1.2	5
380	Noninvasive Home Mechanical Ventilation in Adult Myotonic Dystrophy Type 1: A Systematic Review. Respiration, 2021, 100, 816-825.	1.2	5
381	Respiratory muscle imaging by ultrasound and MRI in neuromuscular disorders. European Respiratory Journal, 2021, 58, 2100137.	3.1	5
382	The socioeconomic burden of facioscapulohumeral muscular dystrophy. Journal of Neurology, 2021, 268, 4778-4788.	1.8	5
383	Facioscapulohumeral muscular dystrophyâ€"Reproductive counseling, pregnancy, and delivery in a complex multigenetic disease. Clinical Genetics, 2022, 101, 149-160.	1.0	5
384	Scheie Syndrome Presenting as Myopathy. Neuropediatrics, 2001, 32, 93-96.	0.3	4
385	NIR spectroscopic measurement of local muscle metabolism during rhythmic, sustained, and intermittent handgrip exercise. , 2003, , .		4
386	Auditory Processing in Patients With Charcot-Marie-Tooth Disease Type 1A. Otology and Neurotology, 2003, 24, 872-877.	0.7	4
387	Contractile properties of knee-extensors in one single family with nemaline myopathy: central and peripheral aspects of muscle activation. Clinical Physiology and Functional Imaging, 2007, 27, 217-224.	0.5	4
388	Computer-aided visualization of muscle weakness distribution. Journal of Neurology, 2008, 255, 1670-1678.	1.8	4
389	Problems of Adults with a Mitochondrial Disease â€“ The Patientsâ€™ Perspective: Focus on Loss. JIMD Reports, 2012, 6, 85-94.	0.7	4
390	Exploring Employment in Consultation Reports of Patients With Neuromuscular Diseases. Archives of Physical Medicine and Rehabilitation, 2012, 93, 2276-2280.	0.5	4
391	The European NeuroMuscular Centre (ENMC): 20 years on â€¦. Neuromuscular Disorders, 2013, 23, 375-376.	0.3	4
392	LAMA2 mutations in adult-onset muscular dystrophy with leukoencephalopathy. Muscle and Nerve, 2014, 49, 616-617.	1.0	4
393	Both aerobic exercise training and cognitive behavior therapy reduce chronic fatigue in patients with facioscapulohumeral muscular dystrophy: A randomized controlled trial. Annals of Physical and Rehabilitation Medicine, 2014, 57, e96.	1.1	4
394	Clinical Functional Capacity Testing in Patients With Facioscapulohumeral Muscular Dystrophy: Construct Validity and Interrater Reliability of Antigravity Tests. Archives of Physical Medicine and Rehabilitation, 2015, 96, 2201-2206.	0.5	4
395	Monitoring creatine and phosphocreatine by 13C MR spectroscopic imaging during and after 13C4 creatine loading: a feasibility study. Amino Acids, 2016, 48, 1857-1866.	1.2	4
396	Muscle ultrasonography is a potential tool for detecting fasciitis in dermatomyositis and polymyositis: comment on the article by Yoshida etÂal. Arthritis and Rheumatology, 2017, 69, 2248-2249.	2.9	4

#	Article	IF	Citations
397	NEM6, KBTBD13-Related Congenital Myopathy: Myopathological Analysis in 18 Dutch Patients Reveals Ring Rods Fibers, Cores, Nuclear Clumps, and Granulo-Filamentous Protein Material. Journal of Neuropathology and Experimental Neurology, 2021, 80, 366-376.	0.9	4
398	Clinical Outcome Evaluations and CBT Response Prediction in Myotonic Dystrophy. Journal of Neuromuscular Diseases, 2021, 8, 1031-1046.	1.1	4
399	Serial Isoelectric Focusing as an Effective and Economic Way to Obtain Maximal Resolution and High-Throughput in 2D-Based Comparative Proteomics of Scarce Samples:Â Proof-of-Principle. Journal of Proteome Research, 2005, 4, 2364-2368.	1.8	3
400	Sensorimotor Axonal Polyneuropathy Without Hepatic Failure in Erythropoietic Protoporphyria. Journal of Clinical Neuromuscular Disease, 2009, 11, 72-76.	0.3	3
401	The neglected brain in myotonic dystrophy types 1 and 2. Neurology, 2010, 74, 1090-1091.	1.5	3
402	The astrologist's posture: a useful clinical observation. Journal of Neurology, Neurosurgery and Psychiatry, 2011, 82, 164-164.	0.9	3
403	Compound heterozygous mutations of the TNXB gene cause primary myopathy. Neuromuscular Disorders, 2014, 24, 88-89.	0.3	3
404	Characteristics and natural history of oculopharyngeal muscular dystrophy (OPMD): The study protocol of â€˜OPMD Forteâ€™. Neuromuscular Disorders, 2016, 26, S139-S140.	0.3	3
405	Cytokine genes as potential biomarkers for muscle weakness in OPMD. Human Molecular Genetics, 2016, 25, 4282-4287.	1.4	3
406	Bilateral Vestibulopathy Aggravates Balance and Gait Disturbances in Sensory Ataxic Neuropathy, Dysarthria, and Ophthalmoparesis: A Case Report. Journal of Clinical Neuromuscular Disease, 2016, 18, 34-36.	0.3	3
407	Oculopharyngeal muscular dystrophy with frontotemporal dementia. European Geriatric Medicine, 2017, 8, 81-83.	1.2	3
408	Muscle fiber dysfunction contributes to weakness in inclusion body myositis. Neuromuscular Disorders, 2019, 29, 468-476.	0.3	3
409	Semiâ€automated Rasch analysis using inâ€plusâ€outâ€ofâ€questionnaire log likelihood. British Journal of Mathematical and Statistical Psychology, 2021, 74, 313-339.	1.0	3
410	Mixed methods evaluation of a self-management group programme for patients with neuromuscular disease and chronic fatigue. BMJ Open, 2021, 11, e048890.	0.8	3
411	Experiences of patients with facioscapulohumeral dystrophy with facial weakness: a qualitative study. Disability and Rehabilitation, 2022, 44, 6775-6782.	0.9	3
412	Neurological features of Noonan syndrome and related <scp>RASopathies</scp> : Pain and nerve enlargement characterized by nerve ultrasound. American Journal of Medical Genetics, Part A, 2022, , .	0.7	3
413	Blood Transcriptome Profiling Links Immunity to Disease Severity in Myotonic Dystrophy Type 1 (DM1). International Journal of Molecular Sciences, 2022, 23, 3081.	1.8	3
414	Halothane-induced Calcium Release in Cultured Human Skeletal Muscle Cells from a Family Susceptible to Malignant Hyperthermia with an Unidentified Mutation in Chromosome 19. Anesthesiology, 2002, 97, 272-274.	1.3	2

#	Article	IF	Citations
415	Less is more: treatment of aggravating behaviour in myasthenia gravis patients with dysphagia. European Journal of Neurology, 2002, 9, 688-689.	1.7	2
416	G.P.15. Neuromuscular Disorders, 2014, 24, 798-799.	0.3	2
417	Effect of Suboptimal Sampling and Handling Conditions on Urinary Metabolic Profiles. Chromatographia, 2015, 78, 429-434.	0.7	2
418	Clinical phenotype and outcome of hepatitis E virus associated neuralgic amyotrophy; an international retrospective comparative cohort study. Journal of Hepatology, 2017, 66, S59.	1.8	2
419	Inclusion body myositis in patients with spinocerebellar ataxia types 3 and 6. Journal of Neurology, Neurosurgery and Psychiatry, 2020, 91, 876-878.	0.9	2
420	Antiâ€“Cytosolic 5â€²â€Nucleotidase 1A Autoantibodies Are Absent in Juvenile Dermatomyositis. Arthritis and Rheumatology, 2021, 73, 1329-1333.	2.9	2
421	Long-term follow-up of respiratory function in facioscapulohumeral muscular dystrophy. Journal of Neurology, 2022, 269, 3682-3689.	1.8	2
422	Visuomotor processing is altered after peripheral nerve damage in neuralgic amyotrophy. Brain Communications, 2022, 4, fcac034.	1.5	2
423	Development and validation of the patient-reported â€œFacial Function Scaleâ€• for facioscapulohumeral muscular dystrophy. Disability and Rehabilitation, 2023, 45, 1530-1535.	0.9	2
424	Reachable workspace analysis is a potential measurement for impairment of the upper extremity in neuralgic amyotrophy. Muscle and Nerve, 2022, 66, 282-288.	1.0	2
425	Can human polyclonal immunoglobulin raise the threshold for convulsions in rats?. Seizure: the Journal of the British Epilepsy Association, 1996, 5, 171-174.	0.9	1
426	Intravenous immunoglobulin preparation increases myoplasmic calcium concentration by activating the dihydropyridineâ€“ryanodine receptor complex. Journal of the Neurological Sciences, 1998, 156, 35-40.	0.3	1
427	Autoantibody Testing in the Evaluation of Idiopathic Inflammatory Myopathies. Journal of Clinical Neuromuscular Disease, 2000, 2, 1-2.	0.3	1
428	O.15 Mutations in the skeletal muscle ryanodine receptor (RYR1) gene presenting with exertional myalgia and rhabdomyolysis. Neuromuscular Disorders, 2011, 21, 748.	0.3	1
429	Severe Dejerineâ€Sottas disease with respiratory failure and dysmorphic features in association with a PMP22 point mutation and a 3q23 microdeletion. Journal of the Peripheral Nervous System, 2012, 17, 223-225.	1.4	1
430	IMPLICATIONS OF ANTI-CN1A SEROTYPE IN INCLUSION BODY MYOSITIS. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, e1.160-e1.	0.9	1
431	Adding quantitative muscle MRI to the FSHD clinical trial toolbox. Neuromuscular Disorders, 2017, 27, S122.	0.3	1
432	Evidence of ER stress and UPR activation in patients with Brody disease and Brody syndrome. Neuropathology and Applied Neurobiology, 2018, 44, 533-536.	1.8	1

#	Article	IF	Citations
433	O.25Phase 1 clinical trial of losmapimod in FSHD: safety, tolerability and target engagement. Neuromuscular Disorders, 2019, 29, S123.	0.3	1
434	Scapular dyskinesis in myotonic dystrophy type 1: clinical characteristics and genetic investigations. Journal of Neurology, 2019, 266, 2987-2996.	1.8	1
435	Characterization of EEG-based functional brain networks in myotonic dystrophy type 1. Clinical Neurophysiology, 2020, 131, 1886-1895.	0.7	1
436	248th ENMC International Workshop: Myotonic dystrophies: Molecular approaches for clinical purposes, framing a European molecular research network, Hoofddorp, the Netherlands, 11â€“13 October 2019. Neuromuscular Disorders, 2020, 30, 521-531.	0.3	1
437	N-of-1 trial of salbutamol in hyperkalaemic periodic paralysis. Journal of Neurology, Neurosurgery and Psychiatry, 2021, 92, jnnp-2021-326347.	0.9	1
438	Behavioural Impairment and Frontotemporal Dementia in Oculopharyngeal Muscular Dystrophy. Journal of Neuromuscular Diseases, 2021, , 1-7.	1.1	1
439	Exploring the influence of smoking and alcohol consumption on clinical severity in patients with facioscapulohumeral muscular dystrophy. Neuromuscular Disorders, 2021, 31, 824-828.	0.3	1
440	Care for capabilities: Implementing the capability approach in rehabilitation of patients with neuromuscular diseases. Study protocol of the controlled before-after ReCap-NMD study. PLoS ONE, 2021, 16, e0261475.	1.1	1
441	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
442	An evaluation of 24â€...h Holter monitoring in patients with myotonic dystrophy type 1. Europace, 0, , .	0.7	1
443	Polyclonal Ig: an immunopharmacon?. Trends in Immunology, 1994, 15, 341-342.	7.5	0
444	Disease Course of Charcot-Marie-Tooth Disease Type 2 and Comorbidityâ€”Reply. Archives of Neurology, 2004, 61, 1470.	4.9	0
445	Safety of the Posterior Approach to the Brachial Plexus. Anesthesia and Analgesia, 2006, 103, 1046.	1.1	0
446	G.P.7.13 Gene expression profiling in a skeletal muscle cell model of oculopharyngeal muscular dystrophy reveals an extracellular matrix defect. Neuromuscular Disorders, 2007, 17, 809.	0.3	0
447	G.P.10.13 Genotype-phenotype correlations for the skeletal muscle chloride- and sodium-channelopathies. Neuromuscular Disorders, 2007, 17, 821-822.	0.3	0
448	C.P.2.02 Neuromuscular involvement in Ehlers-Danlos syndrome. Neuromuscular Disorders, 2007, 17, 843-844.	0.3	0
449	G.P.14.13 Gastrointestinal symptoms in myotonic dystrophy type 2. Neuromuscular Disorders, 2007, 17, 857-858.	0.3	0
450	M.P.4.08 Are oxidative capacity and glycolysis affected in X-linked phosphorylase b kinase deficiency?. Neuromuscular Disorders, 2007, 17, 861.	0.3	0

#	Article	IF	Citations
451	Cervical myelopathy caused by retrograde intraneural dissection of anesthetic solution. Muscle and Nerve, 2008, 37, 546-547.	1.0	0
452	D.P.4.02 Dysphagia in Myotonic Dystrophy type 2. Neuromuscular Disorders, 2008, 18, 796-797.	0.3	0
453	73 Problems of Children and Adolescents with a Mitochondrial Disease, Their Parents and Siblings. The Patients Perspective.. Pediatric Research, 2010, 68, 40-40.	1.1	0
454	O.12 A new member of the BTB/Kelch family of proteins is mutated in nemaline myopathy type 6 (NEM6). Neuromuscular Disorders, 2010, 20, 638.	0.3	0
455	P2.44 Accelerated skeletal muscle ageing is a molecular signature in OPMD. Neuromuscular Disorders, 2011, 21, 673.	0.3	0
456	Coffee and muscle cramps. Neuromuscular Disorders, 2011, 21, 832.	0.3	0
457	Dominant Mutations in KBTBD13, a Member of the BTB/Kelch Family, Cause Nemaline Myopathy with Cores. American Journal of Human Genetics, 2011, 88, 122.	2.6	0
458	C.P.3 Genetic and clinical heterogeneity of RYR1-related myopathies in a cohort of 60 Dutch families with identification of 40 novel mutations. Neuromuscular Disorders, 2012, 22, 841.	0.3	0
459	G.P.108 Sarcomeric dysfunction contributes to muscle weakness in facioscapulohumeral muscular dystrophy. Neuromuscular Disorders, 2012, 22, 900-901.	0.3	0
460	Exertional hyperckemia might be the first manifestation of a genetic disorder. Muscle and Nerve, 2013, 48, 461-462.	1.0	0
461	G.O.28. Neuromuscular Disorders, 2014, 24, 923-924.	0.3	0
462	G.P.131. Neuromuscular Disorders, 2014, 24, 840.	0.3	0
463	Histological abnormalities induced by the electromyography needle. Neuromuscular Disorders, 2014, 24, 77-78.	0.3	0
464	Muscle MRI correlates with histology and clinical features in patients with FSHD, OPMD and sIBM. Neuromuscular Disorders, 2015, 25, S295-S296.	0.3	0
465	Computerized quantification of facial weakness in facioscapulohumeral muscular dystrophy. Neuromuscular Disorders, 2015, 25, S214.	0.3	0
466	Overlap laminopathy with mild neurogenic atrophy and overt muscular dystrophy. Neuromuscular Disorders, 2015, 25, S279.	0.3	0
467	A2.4â€...Subcellular localisation of cytosolic 5â€™-nucleotidase 1A in cell lines and skeletal muscle: implications for inclusion body myositis. Annals of the Rheumatic Diseases, 2015, 74, A17.1-A17.	0.5	0
468	Disease modifying factors in facioscapulohumeral muscular dystrophy: Protocol of the FSHD-FOCUS study. Neuromuscular Disorders, 2015, 25, S214-S215.	0.3	0

#	Article	IF	Citations
469	PGM1 deficiency â€“ A heterogeneous myopathy with opportunities for treatment. Neuromuscular Disorders, 2015, 25, S188-S189.	0.3	0
470	Why are FSHD muscles weak? A novel role for sarcomeric proteins. Neuromuscular Disorders, 2015, 25, S213.	0.3	0
471	Axial myopathy in a patient with a heterozygous MSTN mutation. Neuromuscular Disorders, 2015, 25, S216.	0.3	0
472	The energetic study: Effectiveness of a self-management group programme to improve social participation in patients with neuromuscular disease and chronic fatigue. Neuromuscular Disorders, 2016, 26, S147.	0.3	0
473	Contractile dysfunction in permeabilized muscle fibers of NEM6 patients with the Dutch founder mutation in KBTBD13. Neuromuscular Disorders, 2016, 26, S134.	0.3	0
474	Getting the picture of facioscapulohumeral muscular dystrophy: Muscle magnetic resonance imaging and ultrasound. Neuromuscular Disorders, 2016, 26, S101.	0.3	0
475	Implementation and process evaluation of the energetic study. Neuromuscular Disorders, 2016, 26, S147-S148.	0.3	0
476	Gene variants in SMCHD1 and DNMT3B modify the risk for FSHD. Neuromuscular Disorders, 2016, 26, S152.	0.3	0
477	Changes in sarcomeric contractile function influence force generation in facioscapulohumeral muscular dystrophy. Neuromuscular Disorders, 2016, 26, S167.	0.3	0
478	Respiratory pattern in a FSHD pediatric population. Respiratory Medicine, 2017, 126, 130-131.	1.3	0
479	Muscle fiber dysfunction contributes to clinical muscle weakness in inclusion body myositis. Neuromuscular Disorders, 2017, 27, S154.	0.3	0
480	The Brody disease cohort study: clarification of the phenotype. Neuromuscular Disorders, 2017, 27, S164.	0.3	0
481	Specific strength is reduced in facioscapulohumeral dystrophy muscles. An MRI-based musculoskeletal analysis. Neuromuscular Disorders, 2017, 27, S200.	0.3	0
482	Immunemediated necrotizing autoimmune myopathy: Dutch and Belgian experience. Neuromuscular Disorders, 2017, 27, S151.	0.3	0
483	Muscular fat fraction correlates with functionality in myotonic dystrophy type 1. Neuromuscular Disorders, 2017, 27, S180.	0.3	0
484	Retinal abnormalities in FSHD. Neuromuscular Disorders, 2017, 27, S201.	0.3	0
485	Slow relaxation kinetics of sarcomeres contribute to muscle slowness in NEM6 patients. Neuromuscular Disorders, 2017, 27, S230.	0.3	0
486	CONGENITAL MYOPATHIES: NEMALINE AND TITINOPATHIES. Neuromuscular Disorders, 2018, 28, S100-S101.	0.3	0

#	Article	IF	Citations
487	NEW GENES, FUNCTIONS AND BIOMARKERS. Neuromuscular Disorders, 2018, 28, S31.	0.3	0
488	P.40Ophthalmological findings in facioscapulohumeral dystrophy. Neuromuscular Disorders, 2019, 29, S52-S53.	0.3	0
489	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.	0.3	0
490	P.63Chronic progressive external ophthalmoplegia (CPEO) and CPEO-plus cohort of 54 patients from the Netherlands. Neuromuscular Disorders, 2019, 29, S59.	0.3	0
491	P.162Novel Kbtbd13R408C-knockin mouse model phenocopies NEM6 myopathy. Neuromuscular Disorders, 2019, 29, S95.	0.3	0
492	O.13Nemaline myopathy patients with mutations in KBTBD13 display a cardiac phenotype. Neuromuscular Disorders, 2019, 29, S118.	0.3	0
493	P.247Capturing disease progression in oculopharyngeal muscular dystrophy (OPMD). Neuromuscular Disorders, 2019, 29, S139.	0.3	0
494	P.306Multicentric MRI study in a cohort of FSHD2 patients: pattern definition and differences between FSHD1 and FSHD2. Neuromuscular Disorders, 2019, 29, S156.	0.3	0
495	Group medical appointments for people with physical illness. The Cochrane Library, 0, , .	1.5	0
496	CONGENITAL MYOPATHIES 1 â€“ NEMALINE. Neuromuscular Disorders, 2020, 30, S53.	0.3	0
497	CONGENITAL MYOPATHIES 1 â€“ NEMALINE. Neuromuscular Disorders, 2020, 30, S55.	0.3	0
498	FSHD / OPMD / MYOTONIC DYSTROPHY. Neuromuscular Disorders, 2020, 30, S112-S113.	0.3	0
499	FSHD / OPMD / MYOTONIC DYSTROPHY. Neuromuscular Disorders, 2020, 30, S113.	0.3	0
500	FSHD / OPMD / MYOTONIC DYSTROPHY. Neuromuscular Disorders, 2020, 30, S113.	0.3	0
501	MYOTONIC DYSTROPHY. Neuromuscular Disorders, 2021, 31, S121.	0.3	0
502	FSHD. Neuromuscular Disorders, 2021, 31, S99-S100.	0.3	0
503	FSHD. Neuromuscular Disorders, 2021, 31, S100.	0.3	0
504	CONGENITAL MUSCULAR DYSTROPHIES. Neuromuscular Disorders, 2021, 31, S70.	0.3	0

#	Article	IF	Citations
505	CONGENITAL MYOPATHIES. Neuromuscular Disorders, 2021, 31, S65.	0.3	0
506	9a Addendum: neuralgische amyotrofie. , 2007, , 111-123.		0
507	Addendum: neuralgische amyotrofie Een deel van de tekst van dit addendum is een bewerking van een artikel dat is gepubliceerd in het â€˜Neurologen Vademecumâ€™, uitgegeven door Bohn Stafleu van Loghum, Houten. Meer specifieke informatie is te vinden in eerder gepubliceerde Orthopedische CasuÃ¯stiek, 2002; addendum: neuralgische amyotrofie (Nens van Alfen, Baziel van Engelen). , 2010, , 391-397.		0
508	AB0776â€…Muscle ultrasonography: a potential new diagnostic tool for inflammatory myopathies. , 2018, , .		0