Heinz Jungbluth

List of Publications by Year
in descending order

154 papers	13,228 citations	34105 52 h-index	23533 111 g-index
166 all docs	166 docs citations	166 times ranked	19760 citing authors

#	Article	IF	Citations
1	Guidelines for the use and interpretation of assays for monitoring autophagy (3rd edition). Autophagy, 2016, 12, 1-222.	9.1	4,701
2	Mutations of the Selenoprotein N Gene, Which Is Implicated in Rigid Spine Muscular Dystrophy, Cause the Classical Phenotype of Multiminicore Disease: Reassessing the Nosology of Early-Onset Myopathies. American Journal of Human Genetics, 2002, 71, 739-749.	6.2	326
3	Centronuclear (myotubular) myopathy. Orphanet Journal of Rare Diseases, 2008, 3, 26.	2.7	267
4	Approach to the diagnosis of congenital myopathies. Neuromuscular Disorders, 2014, 24, 97-116.	0.6	239
5	Recessive mutations in EPG5 cause Vici syndrome, a multisystem disorder with defective autophagy. Nature Genetics, 2013, 45, 83-87.	21.4	231
6	RYR1 mutations are a common cause of congenital myopathies with central nuclei. Annals of Neurology, 2010, 68, 717-726.	5.3	230
7	Congenital myopathies: disorders of excitationâ€“contraction coupling and muscle contraction. Nature Reviews Neurology, 2018, 14, 151-167.	10.1	212
8	A recessive form of central core disease, transiently presenting as multi-minicore disease, is associated with a homozygous mutation in the ryanodine receptor type 1 gene. Annals of Neurology, 2002, 51, 750-759.	5.3	181
9	Mutations in RYR1 are a common cause of exertional myalgia and rhabdomyolysis. Neuromuscular Disorders, 2013, 23, 540-548.	0.6	169
10	Molecular mechanisms and phenotypic variation in RYR1-related congenital myopathies. Brain, 2007, 130, 2024-2036.	7.6	161
11	Autosomal recessive inheritance of RYR1 mutations in a congenital myopathy with cores. Neurology, 2002, 59, 284-287.	1.1	157
12	Congenital muscle disorders with cores: the ryanodine receptor calcium channel paradigm. Current Opinion in Pharmacology, 2008, 8, 319-326.	3.5	149
13	Consensus Statement on Standard of Care for Congenital Myopathies. Journal of Child Neurology, 2012, 27, 363-382.	1.4	147
14	Clinical and genetic findings in a large cohort of patients with ryanodine receptor 1 gene-associated myopathies. Human Mutation, 2012, 33, 981-988.	2.5	145
15	Central core disease. Orphanet Journal of Rare Diseases, 2007, 2, 25.	2.7	140
16	Magnetic resonance imaging of muscle in congenital myopathies associated with RYR1 mutations. Neuromuscular Disorders, 2004, 14, 785-790.	0.6	135
17	Muscle histology vs MRI in Duchenne muscular dystrophy. Neurology, 2011, 76, 346-353.	1.1	134
18	Minicore myopathy with ophthalmoplegia caused by mutations in the ryanodine receptor type 1 gene. Neurology, 2005, 65, 1930-1935.	1.1	131

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19	Congenital disorders of autophagy: an emerging novel class of inborn errors of neuro-metabolism. Brain, 2016, 139, 317-337.	7.6	126
20	Core Myopathies. Seminars in Pediatric Neurology, 2011, 18, 239-249.	2.0	120
21	Principal mutation hotspot for central core disease and related myopathies in the C-terminal transmembrane region of the RYR1 gene. Neuromuscular Disorders, 2003, 13, 151-157.	0.6	118
22	Multi-minicore Disease. Orphanet Journal of Rare Diseases, 2007, 2, 31.	2.7	114
23	Congenital Myasthenic Syndromes in childhood: Diagnostic and management challenges. Journal of Neuroimmunology, 2008, 201-202, 6-12.	2.3	114
24	Congenital myopathies â€“ Clinical features and frequency of individual subtypes diagnosed over a 5-year period in the United Kingdom. Neuromuscular Disorders, 2013, 23, 195-205.	0.6	113
25	Epigenetic Allele Silencing Unveils Recessive RYR1 Mutations in Core Myopathies. American Journal of Human Genetics, 2006, 79, 859-868.	6.2	111
26	<scp>RYR</scp>1â€related myopathies: a wide spectrum of phenotypes throughout life. European Journal of Neurology, 2015, 22, 1094-1112.	3.3	111
27	The use of rituximab in myasthenia gravis and Lambert-Eaton myasthenic syndrome. Journal of Neurology, Neurosurgery and Psychiatry, 2011, 82, 671-673.	1.9	107
28	Congenital myopathies. Neurology, 2015, 84, 28-35.	1.1	106
29	Centronuclear myopathy due to a de novo dominant mutation in the skeletal muscle ryanodine receptor (RYR1) gene. Neuromuscular Disorders, 2007, 17, 338-345.	0.6	105
30	A short protocol for muscle MRI in children with muscular dystrophies. European Journal of Paediatric Neurology, 2002, 6, 305-307.	1.6	105
31	Characterization of recessive RYR1 mutations in core myopathies. Human Molecular Genetics, 2006, 15, 2791-2803.	2.9	103
32	Late-onset axial myopathy with cores due to a novel heterozygous dominant mutation in the skeletal muscle ryanodine receptor (RYR1) gene. Neuromuscular Disorders, 2009, 19, 344-347.	0.6	103
33	Selective Muscle Involvement on Magnetic Resonance Imaging in Autosomal Dominant Emery-Dreifuss Muscular Dystrophy. Neuropediatrics, 2002, 33, 10-14.	0.6	101
34	Rhabdomyolysis: a genetic perspective. Orphanet Journal of Rare Diseases, 2015, 10, 51.	2.7	101
35	EPG5-related Vici syndrome: a paradigm of neurodevelopmental disorders with defective autophagy. Brain, 2016, 139, 765-781.	7.6	99
36	Magnetic resonance imaging of muscle in nemaline myopathy. Neuromuscular Disorders, 2004, 14, 779-784.	0.6	98

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37	Kingâ€“Denborough syndrome with and without mutations in the skeletal muscle ryanodine receptor (RYR1) gene. Neuromuscular Disorders, 2011, 21, 420-427.	0.6	97
38	Impaired neuromuscular transmission and response to acetylcholinesterase inhibitors in centronuclear myopathies. Neuromuscular Disorders, 2011, 21, 379-386.	0.6	96
39	Mild phenotype of nemaline myopathy with sleep hypoventilation due to a mutation in the skeletal muscle Î±-actin (ACTA1) gene. Neuromuscular Disorders, 2001, 11, 35-40.	0.6	92
40	Muscle Magnetic Resonance Imaging in Congenital Myopathies Due to Ryanodine Receptor Type 1 Gene Mutations. Archives of Neurology, 2011, 68, 1171.	4.5	89
41	Pathogenic Mechanisms in Centronuclear Myopathies. Frontiers in Aging Neuroscience, 2014, 6, 339.	3.4	89
42	Novel Mutations Widen the Phenotypic Spectrum of Slow Skeletal/Î²-Cardiac Myosin (MYH7) Distal Myopathy. Human Mutation, 2014, 35, 868-879.	2.5	79
43	The phenotype of Charcotâ€“Marieâ€“Tooth disease type 4C due to SH3TC2 mutations and possible predisposition to an inflammatory neuropathy. Neuromuscular Disorders, 2009, 19, 264-269.	0.6	78
44	Multi-minicore disease and atypical periodic paralysis associated with novel mutations in the skeletal muscle ryanodine receptor (RYR1) gene. Neuromuscular Disorders, 2010, 20, 166-173.	0.6	78
45	Mutations in MYH7 cause Multi-minicore Disease (MmD) with variable cardiac involvement. Neuromuscular Disorders, 2012, 22, 1096-1104.	0.6	73
46	Exertional rhabdomyolysis: physiological response or manifestation of an underlying myopathy?. BMJ Open Sport and Exercise Medicine, 2016, 2, e000151.	2.9	73
47	Compound heterozygosity and nonsense mutations in the Î±1-subunit of the inhibitory glycine receptor in hyperekplexia. Human Genetics, 2001, 109, 267-270.	3.8	72
48	A novel late-onset axial myopathy associated with mutations in the skeletal muscle ryanodine receptor (RYR1) gene. Journal of Neurology, 2013, 260, 1504-1510.	3.6	71
49	MRI in DNM2-related centronuclear myopathy: Evidence for highly selective muscle involvement. Neuromuscular Disorders, 2007, 17, 28-32.	0.6	64
50	Minicore myopathy in children: a clinical and histopathological study of 19 cases. Neuromuscular Disorders, 2000, 10, 264-273.	0.6	60
51	Functional properties of ryanodine receptors carrying three amino acid substitutions identified in patients affected by multi-minicore disease and central core disease, expressed in immortalized lymphocytes. Biochemical Journal, 2006, 395, 259-266.	3.7	59
52	Pilot Trial of Salbutamol in Central Core and Multi-Minicore Diseases. Neuropediatrics, 2004, 35, 262-266.	0.6	55
53	Vici syndrome: a review. Orphanet Journal of Rare Diseases, 2016, 11, 21.	2.7	55
54	Myopathic causes of exercise intolerance with rhabdomyolysis. Developmental Medicine and Child Neurology, 2012, 54, 886-891.	2.1	54

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55	Novel deletion of lysine 7 expands the clinical, histopathological and genetic spectrum of TPM2-related myopathies. Brain, 2013, 136, 508-521.	7.6	53
56	Fetal acetylcholine receptor inactivation syndrome. Neurology: Neuroimmunology and NeuroInflammation, 2015, 2, e57.	6.0	50
57	Early and severe presentation of X-linked myotubular myopathy in a girl with skewed X-inactivation. Neuromuscular Disorders, 2003, 13, 55-59.	0.6	48
58	What's new in neuromuscular disorders? The congenital myopathies. European Journal of Paediatric Neurology, 2003, 7, 23-30.	1.6	47
59	Joint hypermobility as a distinctive feature in the differential diagnosis of myopathies. Journal of Neurology, 2009, 256, 13-27.	3.6	47
60	Stall in Canonical Autophagy-Lysosome Pathways Prompts Nucleophagy-Based Nuclear Breakdown in Neurodegeneration. Current Biology, 2017, 27, 3626-3642.e6.	3.9	47
61	MYO-MRI diagnostic protocols in genetic myopathies. Neuromuscular Disorders, 2019, 29, 827-841.	0.6	46
62	Epigenetic changes as a common trigger of muscle weakness in congenital myopathies. Human Molecular Genetics, 2015, 24, 4636-4647.	2.9	44
63	STAC3variants cause a congenital myopathy with distinctive dysmorphic features and malignant hyperthermia susceptibility. Human Mutation, 2018, 39, 1980-1994.	2.5	42
64	X-inactivation patterns in carriers of X-linked myotubular myopathy. Neuromuscular Disorders, 2003, 13, 468-471.	0.6	40
65	Vici syndrome associated with sensorineural hearing loss and evidence of neuromuscular involvement on muscle biopsy. American Journal of Medical Genetics, Part A, 2010, 152A, 741-747.	1.2	40
66	RyR1 Deficiency in Congenital Myopathies Disrupts Excitation-Contraction Coupling. Human Mutation, 2013, 34, 986-996.	2.5	40
67	Atypical periodic paralysis and myalgia. Neurology, 2018, 90, e412-e418.	1.1	39
68	CAV3 mutations causing exercise intolerance, myalgia and rhabdomyolysis: Expanding the phenotypic spectrum of caveolinopathies. Neuromuscular Disorders, 2016, 26, 504-510.	0.6	38
69	Centronuclear myopathy with cataracts due to a novel dynamin 2 (DNM2) mutation. Neuromuscular Disorders, 2010, 20, 49-52.	0.6	37
70	Congenital myopathies: not only a paediatric topic. Current Opinion in Neurology, 2016, 29, 642-650.	3.6	37
71	HeterozygousKIDINS220/ARMSnonsense variants cause spastic paraplegia, intellectual disability, nystagmus, and obesity. Human Molecular Genetics, 2016, 25, 2158-2167.	2.9	37
72	Downstream effects of plectin mutations in epidermolysis bullosa simplex with muscular dystrophy. Acta Neuropathologica Communications, 2016, 4, 44.	5.2	35

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73	Mutations in SCN4A: A Rare but Treatable Cause of Recurrent Life-Threatening Laryngospasm. Pediatrics, 2014, 134, e1447-e1450.	2.1	34
74	An RYR1 mutation associated with malignant hyperthermia is also associated with bleeding abnormalities. Science Signaling, 2016, 9, ra68.	3.6	34
75	Myopathology in times of modern imaging. Neuropathology and Applied Neurobiology, 2017, 43, 24-43.	3.2	34
76	Making sense of missense variants in TTN-related congenital myopathies. Acta Neuropathologica, 2021, 141, 431-453.	7.7	34
77	Muscle MRI findings in a three-generation family affected by Bethlem myopathy. European Journal of Paediatric Neurology, 2002, 6, 309-314.	1.6	34
78	Functional effects of mutations identified in patients with Multiminicore disease. IUBMB Life, 2007, 59, 14-20.	3.4	30
79	219th ENMC International Workshop Titinopathies International database of titin mutations and phenotypes, Heemskerk, The Netherlands, 29 Aprilâ€“1 May 2016. Neuromuscular Disorders, 2017, 27, 396-407.	0.6	29
80	Current and future therapeutic approaches to the congenital myopathies. Seminars in Cell and Developmental Biology, 2017, 64, 191-200.	5.0	29
81	Clinical utility gene card for: Centronuclear and myotubular myopathies. European Journal of Human Genetics, 2012, 20, 1101-1101.	2.8	28
82	Compound RYR1 heterozygosity resulting in a complex phenotype of malignant hyperthermia susceptibility and a core myopathy. Neuromuscular Disorders, 2015, 25, 567-576.	0.6	28
83	RYR1-related rhabdomyolysis: A common but probably underdiagnosed manifestation of skeletal muscle ryanodine receptor dysfunction. Revue Neurologique, 2016, 172, 546-558.	1.5	28
84	Early-onset movement disorder and epileptic encephalopathy due to de novo dominant SCN8A mutation. Seizure: the Journal of the British Epilepsy Association, 2015, 26, 69-71.	2.0	26
85	Transcriptional Regulation of the Glutamate/GABA/Glutamine Cycle in Adult Glia Controls Motor Activity and Seizures in Drosophila. Journal of Neuroscience, 2019, 39, 5269-5283.	3.6	26
86	The histopathological spectrum of malignant hyperthermia and rhabdomyolysis due to RYR1 mutations. Journal of Neurology, 2019, 266, 876-887.	3.6	26
87	The etiology of rhabdomyolysis: an interaction between genetic susceptibility and external triggers. European Journal of Neurology, 2021, 28, 647-659.	3.3	26
88	Impairments in contractility and cytoskeletal organisation cause nuclear defects in nemaline myopathy. Acta Neuropathologica, 2019, 138, 477-495.	7.7	25
89	Muscle MRI findings in siblings with juvenile-onset acid maltase deficiency (Pompe disease). Neuromuscular Disorders, 2008, 18, 408-409.	0.6	24
90	Outcome of children with acetylcholine receptor (AChR) antibody positive juvenile myasthenia gravis following thymectomy. Neuromuscular Disorders, 2014, 24, 25-30.	0.6	24

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91	Goldbergâ€“Shprintzen megacolon syndrome with associated sensory motor axonal neuropathy. American Journal of Medical Genetics, Part A, 2015, 167, 1300-1304.	1.2	23
92	The Vici syndrome protein EPG5 regulates intracellular nucleic acid trafficking linking autophagy to innate and adaptive immunity. Autophagy, 2018, 14, 22-37.	9.1	23
93	<scp>RBCK1</scp>â€related disease: A rare multisystem disorder with polyglucosan storage, autoâ€inflammation, recurrent infections, skeletal, and cardiac myopathyâ€”Four additional patients and a review of the current literature. Journal of Inherited Metabolic Disease, 2020, 43, 1002-1013.	3.6	23
94	Enhanced excitation-coupled Ca2+ entry induces nuclear translocation of NFAT and contributes to IL-6 release from myotubes from patients with central core disease. Human Molecular Genetics, 2011, 20, 589-600.	2.9	22
95	Recessive truncating IGHMBP2 mutations presenting as axonal sensorimotor neuropathy. Neurology, 2015, 84, 523-531.	1.1	22
96	The neuromuscular differential diagnosis of joint hypermobility. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2015, 169, 23-42.	1.6	22
97	RYR1-related malignant hyperthermia with marked cerebellar involvement â€“ A paradigm of heat-induced CNS injury?. Neuromuscular Disorders, 2015, 25, 138-140.	0.6	21
98	Cellular, biochemical and molecular changes in muscles from patients with X-linked myotubular myopathy due toMTM1mutations. Human Molecular Genetics, 2016, 26, ddw388.	2.9	20
99	Functional impairments, fatigue and quality of life in RYR1-related myopathies: A questionnaire study. Neuromuscular Disorders, 2019, 29, 30-38.	0.6	20
100	An ancestral 10-bp repeat expansion in VWA1 causes recessive hereditary motor neuropathy. Brain, 2021, 144, 584-600.	7.6	20
101	Ca2+ handling abnormalities in early-onset muscle diseases: Novel concepts and perspectives. Seminars in Cell and Developmental Biology, 2017, 64, 201-212.	5.0	19
102	The spectrum of neurodevelopmental, neuromuscular and neurodegenerative disorders due to defective autophagy. Autophagy, 2022, 18, 496-517.	9.1	18
103	A recessive ryanodine receptor 1 mutation in a CCD patient increases channel activity. Cell Calcium, 2009, 45, 192-197.	2.4	16
104	The Human 343delT HSPB5 Chaperone Associated with Early-onset Skeletal Myopathy Causes Defects in Protein Solubility. Journal of Biological Chemistry, 2016, 291, 14939-14953.	3.4	16
105	Fatal awake malignant hyperthermia episodes in a family with malignant hyperthermia susceptibility: a case series. Canadian Journal of Anaesthesia, 2019, 66, 540-545.	1.6	16
106	ECEL1 gene related contractural syndrome: Long-term follow-up and update on clinical and pathological aspects. Neuromuscular Disorders, 2018, 28, 741-749.	0.6	15
107	Ophthalmologic Features of Vici Syndrome. Journal of Pediatric Ophthalmology and Strabismus, 2014, 51, 214-220.	0.7	15
108	198th ENMC International Workshop: 7th Workshop on Centronuclear (Myotubular) myopathies, 31st May â€“ 2nd June 2013, Naarden, The Netherlands. Neuromuscular Disorders, 2013, 23, 1033-1043.	0.6	14

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109	SIL1-related Marinescoâ€“Sjoegren syndrome (MSS) with associated motor neuronopathy and bradykinetic movement disorder. Neuromuscular Disorders, 2015, 25, 585-588.	0.6	14
110	â€œHuman Stress Syndromeâ€•and the Expanding Spectrum of RYR1-Related Myopathies. Cell Biochemistry and Biophysics, 2016, 74, 85-87.	1.8	14
111	Recessive MYH7-related myopathy in two families. Neuromuscular Disorders, 2019, 29, 456-467.	0.6	14
112	Clinical utility gene card for: Vici Syndrome. European Journal of Human Genetics, 2014, 22, 435-435.	2.8	13
113	Aberrant regulation of epigenetic modifiers contributes to the pathogenesis in patients with selenoprotein N â€ related myopathies. Human Mutation, 2019, 40, 962-974.	2.5	13
114	rAAV-related therapy fully rescues myonuclear and myofilament function in X-linked myotubular myopathy. Acta Neuropathologica Communications, 2020, 8, 167.	5.2	12
115	Referral Indications for Malignant Hyperthermia Susceptibility Diagnostics in Patients without Adverse Anesthetic Events in the Era of Next-generation Sequencing. Anesthesiology, 2022, 136, 940-953.	2.5	12
116	Pre-operative exercise and pyrexia as modifying factors in malignant hyperthermia (MH). Neuromuscular Disorders, 2022, 32, 628-634.	0.6	12
117	X-linked myotubular myopathy due to a complex rearrangement involving a duplication of MTM1 exon 10. Neuromuscular Disorders, 2012, 22, 384-388.	0.6	11
118	PRUNE1 Deficiency: Expanding the Clinical and Genetic Spectrum. Neuropediatrics, 2018, 49, 330-338.	0.6	11
119	RYR1-Related Rhabdomyolysis: A Spectrum of Hypermetabolic States Due to Ryanodine Receptor Dysfunction. Current Pharmaceutical Design, 2022, 28, 2-14.	1.9	11
120	Dantrolene as a possible prophylactic treatment for RYR1â€•related rhabdomyolysis. European Journal of Neurology, 2016, 23, e56-7.	3.3	10
121	Salbutamol-responsive fetal acetylcholine receptor inactivation syndrome. Neurology, 2016, 86, 692-694.	1.1	10
122	Therapeutic Aspects in Congenital Myopathies. Seminars in Pediatric Neurology, 2019, 29, 71-82.	2.0	10
123	Severe Central Sleep Apnea in Vici Syndrome. Pediatrics, 2015, 136, e1390-e1394.	2.1	9
124	A novel case of MSTO1 gene related congenital muscular dystrophy with progressive neurological involvement. Neuromuscular Disorders, 2019, 29, 448-455.	0.6	9
125	HyperCKemia and rhabdomyolysis in the neuroleptic malignant and serotonin syndromes: A literature review. Neuromuscular Disorders, 2020, 30, 949-958.	0.6	9
126	Spectrum of Clinical Features in X-Linked Myotubular Myopathy Carriers. Neurology, 2021, 97, e501-e512.	1.1	9

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127	Polymyositis without Beneficial Response to Steroid Therapy: Should Miyoshi Myopathy be a Differential Diagnosis?. American Journal of Case Reports, 2017, 18, 17-21.	0.8	9
128	Lyme Neuroborreliosis: A Potentially Preventable Cause of Stroke. Journal of Pediatrics, 2016, 170, 334-334.e1.	1.8	8
129	The neuromuscular and multisystem features of RYR1-related malignant hyperthermia and rhabdomyolysis. Medicine (United States), 2021, 100, e26999.	1.0	8
130	An unusual case of hyperekplexia. European Journal of Paediatric Neurology, 2000, 4, 77-80.	1.6	7
131	INPP5K and SIL1 associated pathologies with overlapping clinical phenotypes converge through dysregulation of PHGDH. Brain, 2021, 144, 2427-2442.	7.6	7
132	Clinical, genetic, and histological features of centronuclear myopathy in the Netherlands. Clinical Genetics, 2021, 100, 692-702.	2.0	7
133	Autopsy findings in EPG5â€related Vici syndrome with antenatal onset. American Journal of Medical Genetics, Part A, 2017, 173, 2522-2527.	1.2	6
134	Neck-Tongue Syndrome: An Underrecognized Childhood Onset Cephalalgia. Journal of Child Neurology, 2018, 33, 347-350.	1.4	6
135	Genetic neuropathies presenting with CIDP-like features in childhood. Neuromuscular Disorders, 2021, 31, 113-122.	0.6	6
136	259th ENMC international workshop: Anaesthesia and neuromuscular disorders 11 December, 2020 and 28â€“29 May, 2021. Neuromuscular Disorders, 2022, 32, 86-97.	0.6	6
137	Parental mosaicism in RYR1 -related Central Core Disease. Neuromuscular Disorders, 2018, 28, 422-426.	0.6	5
138	Compound heterozygous RYR1 mutations in a preterm with arthrogryposis multiplex congenita and prenatal CNS bleeding. Neuromuscular Disorders, 2018, 28, 54-58.	0.6	5
139	Clinical utility gene card for: Central core disease. European Journal of Human Genetics, 2012, 20, 5-5.	2.8	4
140	Reply: Aberrant splicing induced by the most commonEPG5mutation in an individual with Vici syndrome. Brain, 2016, 139, e53-e53.	7.6	4
141	Unusual Presentations of Dystrophinopathies in Childhood. Pediatrics, 2018, 141, S510-S514.	2.1	4
142	Driving next-generation autophagy researchers towards translation (DRIVE), an international PhD training program on autophagy. Autophagy, 2019, 15, 347-351.	9.1	4
143	G.P.22. Neuromuscular Disorders, 2014, 24, 801.	0.6	3
144	Clinical utility gene card for: Multi-minicore disease. European Journal of Human Genetics, 2012, 20, 5-5.	2.8	2

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145	The Congenital Myopathies. , 2015, , 1121-1129.		2
146	Genotype-phenotype correlations in ocular manifestations of Marinescoâ€“SjÃ¶gren syndrome: Case report and literature review. European Journal of Ophthalmology, 2022, 32, NP92-NP97.	1.3	2
147	Generalized calcification in a case of dermatomyositis. Neuromuscular Disorders, 2000, 10, 150.	0.6	1
148	Congenital myopathy with focal loss of cross-striations revisited. Neuromuscular Disorders, 2013, 23, 160-164.	0.6	1
149	Autophagy â€“ a fundamental cellular mechanism on the verge of clinical translation. Neuropathology and Applied Neurobiology, 2015, 41, 598-600.	3.2	1
150	Novel phosphopantothenoylcysteine synthetase (<scp> PPCS </scp>) mutations with prominent neuromuscular features: Expanding the phenotypical spectrum of <scp> PPCS </scp> â€related disorders. American Journal of Medical Genetics, Part A, 0, , .	1.2	1
151	Congenital (Structural) Myopathies. , 2013, , 1-51.		0
152	Compound Heterozygous RYR1 Mutation in a Preterm with Arthrogryposis and Core Myopathy with Prenatal CNS Bleedings. Neuropediatrics, 2017, 48, S1-S45.	0.6	0
153	Congenital disorders of autophagy â€“ a novel class of neurological and neuromuscular disorders linking abnormal neurodevelopment and neurodegeneration. Nervenheilkunde, 2019, 38, .	0.0	0
154	The congenital myopathies. , 2020, , 451-461.		0