Mariz Vainzof

List of Publications by Year
in descending order

199 papers	6,179 citations	71061 41 h-index	88593 70 g-index
201 all docs	201 docs citations	201 times ranked	5340 citing authors

#	Article	IF	Citations
1	Autosomal recessive limbgirdle muscular dystrophy, LGMD2F, is caused by a mutation in the Î'â€"sarcoglycan gene. Nature Genetics, 1996, 14, 195-198.	9.4	417
2	Limb-girdle muscular dystrophy type 2G is caused by mutations in the gene encoding the sarcomeric protein telethonin. Nature Genetics, 2000, 24, 163-166.	9.4	312
3	Approach to the diagnosis of congenital myopathies. Neuromuscular Disorders, 2014, 24, 97-116.	0.3	239
4	Caveolin-3 in muscular dystrophy. Human Molecular Genetics, 1998, 7, 871-877.	1.4	200
5	The sarcoglycan complex in the six autosomal recessive limb-girdle muscular dystrophies. Human Molecular Genetics, 1996, 5, 1963-1969.	1.4	167
6	Consensus Statement on Standard of Care for Congenital Myopathies. Journal of Child Neurology, 2012, 27, 363-382.	0.7	147
7	Serum creatine-kinase (CK) and pyruvate-kinase (PK) activities in Duchenne (DMD) as compared with Becker (BMD) muscular dystrophy. Journal of the Neurological Sciences, 1991, 102, 190-196.	0.3	136
8	The Seventh Form of Autosomal Recessive Limb-Girdle Muscular Dystrophy Is Mapped to 17q11-12. American Journal of Human Genetics, 1997, 61, 151-159.	2.6	136
9	The facioscapulohumeral muscular dystrophy (FSHD1) gene affects males more severely and more frequently than females. American Journal of Medical Genetics Part A, 1998, 77, 155-161.	2.4	123
10	The 10 autosomal recessive limb-girdle muscular dystrophies. Neuromuscular Disorders, 2003, 13, 532-544.	0.3	122
11	Genomic screening for beta-sarcoglycan gene mutations: missense mutations may cause severe limb-girdle muscular dystrophy type 2E (LGMD 2E). Human Molecular Genetics, 1996, 5, 1953-1961.	1.4	111
12	Animal Models for Genetic Neuromuscular Diseases. Journal of Molecular Neuroscience, 2008, 34, 241-248.	1.1	106
13	A defect in the RNA-processing protein HNRPDL causes limb-girdle muscular dystrophy 1G (LGMD1G). Human Molecular Genetics, 2014, 23, 4103-4110.	1.4	101
14	Limb-girdle muscular dystrophy: one gene with different phenotypes, one phenotype with different genes. Current Opinion in Neurology, 2000, 13, 511-517.	1.8	93
15	Human multipotent adiposeâ€derived stem cells restore dystrophin expression of Duchenne skeletalâ€muscle cells in vitro. Biology of the Cell, 2008, 100, 231-241.	0.7	93
16	Linkage analysis in autosomal recessive limb-girdle muscular dystrophy (AR LGMD) maps a sixth form to 5q33-34 (LGMD2F) and indicates that there is at least one more subtype of AR LGMD. Human Molecular Genetics, 1996, 5, 815-820.	1.4	92
17	Seven autosomal recessive limb-girdle muscular dystrophies in the Brazilian population: from LGMD2A to LGMD2G. , 1999, 82, 392-398.		90
18	Clinical variability in calpainopathy: What makes the difference?. European Journal of Human Genetics, 2002, 10, 825-832.	1.4	84

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19	Sarcoglycanopathies are responsible for 68% of severe autosomal recessive limb-girdle muscular dystrophy in the Brazilian population. Journal of the Neurological Sciences, 1999, 164, 44-49.	0.3	81
20	Analysis of the CTG repeat in skeletal muscle of young and adult myotonic dystrophy patients: when does the expansion occur?. Human Molecular Genetics, 1995, 4, 401-406.	1.4	79
21	Myocardial Fibrosis Progression in Duchenne and Becker Muscular Dystrophy. JAMA Cardiology, 2017, 2, 190.	3.0	79
22	A common missense mutation in the adhalin gene in three unrelated Brazilian families with a relatively mild form of autosomal recessive limb-girdle muscular dystrophy. Human Molecular Genetics, 1995, 4, 1163-1167.	1.4	75
23	SJL Dystrophic Mice Express a Significant Amount of Human Muscle Proteins Following Systemic Delivery of Human Adipose-Derived Stromal Cells Without Immunosuppression. Stem Cells, 2008, 26, 2391-2398.	1.4	68
24	Dysferlin Protein Analysis in Limb-Girdle Muscular Dystrophies. Journal of Molecular Neuroscience, 2001, 17, 71-80.	1.1	67
25	Apparent association of mental retardation and specific patterns of deletions screened with probes cf56a and cf23a in Duchenne muscular dystrophy. American Journal of Medical Genetics Part A, 1991, 39, 437-441.	2.4	66
26	Asymptomatic carriers for homozygous novel mutations in the FKRP gene: the other end of the spectrum. European Journal of Human Genetics, 2003, 11, 923-930.	1.4	66
27	Mutation in the Scyl1 gene encoding aminoâ€terminal kinaseâ€like protein causes a recessive form of spinocerebellar neurodegeneration. EMBO Reports, 2007, 8, 691-697.	2.0	63
28	A novel locus for late onset amyotrophic lateral sclerosis/motor neurone disease variant at 20q13. Journal of Medical Genetics, 2004, 41, 315-320.	1.5	61
29	A new form of autosomal dominant limb-girdle muscular dystrophy (LGMD1G) with progressive fingers and toes flexion limitation maps to chromosome 4p21. European Journal of Human Genetics, 2004, 12, 1033-1040.	1.4	61
30	Human Adipose-Derived Mesenchymal Stromal Cells Injected Systemically into GRMD Dogs without Immunosuppression are Able to Reach the Host Muscle and Express Human Dystrophin. Cell Transplantation, 2012, 21, 1407-1417.	1.2	56
31	Immunofluorescence dystrophin study in Duchenne dystrophy through the concomitant use of two antibodies directed against the carboxy-terminal and the amino-terminal region of the protein. Journal of the Neurological Sciences, 1991, 101, 141-147.	0.3	52
32	Characterization of Human Skeletal Muscle Ankrd2. Biochemical and Biophysical Research Communications, 2001, 285, 378-386.	1.0	51
33	Central core disease due to recessive mutations inRYR1 gene: Is it more common than described?. Muscle and Nerve, 2007, 35, 670-674.	1.0	51
34	Genetic heterogeneity for Duchenne-like muscular dystrophy (DLMD) based on linkage and 50 DAG analysis. Human Molecular Genetics, 1993, 2, 1945-1947.	1.4	50
35	Telethonin protein expression in neuromuscular disorders. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2002, 1588, 33-40.	1.8	49
36	Deficiency of Merosin (Laminin M or Î±2) in Congenital Muscular Dystrophy Associated with Cerebral White Matter Alterations. Neuropediatrics, 1995, 26, 293-297.	0.3	48

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37	A first missense mutation in the delta sarcoglycan gene associated with a severe phenotype and frequency of limb-girdle muscular dystrophy type 2F (LGMD2F) in Brazilian sarcoglycanopathies.. Journal of Medical Genetics, 1998, 35, 951-953.	1.5	48
38	Motor Chip: A Comparative Genomic Hybridization Microarray for Copy-Number Mutations in 245 Neuromuscular Disorders. Clinical Chemistry, 2011, 57, 1584-1596.	1.5	48
39	Knobloch syndrome in a large Brazilian consanguineous family: Confirmation of autosomal recessive inheritance. American Journal of Medical Genetics Part A, 1994, 52, 170-173.	2.4	47
40	Main clinical features of the three mapped autosomal recessive limb-girdle muscular dystrophies and estimated proportion of each form in 13 Brazilian families.. Journal of Medical Genetics, 1996, 33, 97-102.	1.5	45
41	Mutations in the caveolin-3 gene: When are they pathogenic?. American Journal of Medical Genetics Part A, 2001, 99, 303-307.	2.4	43
42	The phenotype of chromosome 2p-linked limb-girdle muscular dystrophy. Neuromuscular Disorders, 1996, 6, 483-490.	0.3	42
43	Human Multipotent Mesenchymal Stromal Cells from Distinct Sources Show Different In Vivo Potential to Differentiate into Muscle Cells When Injected in Dystrophic Mice. Stem Cell Reviews and Reports, 2010, 6, 560-566.	5.6	42
44	K7del is a common TPM2 gene mutation associated with nemaline myopathy and raised myofibre calcium sensitivity. Brain, 2013, 136, 494-507.	3.7	42
45	Confirmation of the 2p Locus for the Mild Autosomal Recessive Limb-Girdle Muscular Dystrophy Gene (LGMD2B) in Three Families Allows Refinement of the Candidate Region. Genomics, 1995, 27, 192-195.	1.3	41
46	Absence of calpain 3 in a form of limb-girdle muscular dystrophy (LGMD2A). Journal of the Neurological Sciences, 1997, 146, 173-178.	0.3	41
47	Ringo, a Golden Retriever Muscular Dystrophy (GRMD) dog with absent dystrophin but normal strength. Neuromuscular Disorders, 2008, 18, 892-893.	0.3	41
48	Nebulin expression in patients with nemaline myopathy. Neuromuscular Disorders, 2001, 11, 154-162.	0.3	39
49	Muscle satellite cells and impaired late stage regeneration in different murine models for muscular dystrophies. Scientific Reports, 2019, 9, 11842.	1.6	39
50	Dystrophin immunostaining in muscles from patients with different types of muscular dystrophy: a Brazilian study. Journal of the Neurological Sciences, 1990, 98, 221-233.	0.3	38
51	A deletion including the brain promoter of the Duchenne muscular dystrophy gene is not associated with mental retardation. Neuromuscular Disorders, 1992, 2, 117-120.	0.3	38
52	Half the dystrophin gene is apparently enough for a mild clinical course: confirmation of its potential use for gene therapy. Human Molecular Genetics, 1994, 3, 919-922.	1.4	36
53	Prenatal diagnosis in laminin Î±2 chain (merosin)-deficient congenital muscular dystrophy: A collective experience of five international centers. Neuromuscular Disorders, 2005, 15, 588-594.	0.3	35
54	Ringo: Discordance between the molecular and clinical manifestation in a Golden Retriever Muscular Dystrophy dog. Neuromuscular Disorders, 2010, 20, 64-70.	0.3	33

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55	Muscle Satellite Cells: Exploring the Basic Biology to Rule Them. Stem Cells International, 2016, 2016, 1-14.	1.2	33
56	Is the maintainance of the C-terminus domain of dystrophin enough to ensure a milder Becker muscular dystrophy phenotype?. Human Molecular Genetics, 1993, 2, 39-42.	1.4	32
57	Absence of correlation between utrophin localization and quantity and the clinical severity in Duchenne/Becker dystrophies. American Journal of Medical Genetics Part A, 1995, 58, 305-309.	2.4	32
58	Absence of correlation between skewed X inactivation in blood and serum creatine-kinase levels in Duchenne/Becker female carriers. , 1998, 80, 356-361.		31
59	Metabolic profile of dystrophic mdx mouse muscles analyzed with in vitro magnetic resonance spectroscopy (MRS). Magnetic Resonance Imaging, 2012, 30, 1167-1176.	1.0	31
60	Evidence of genetic heterogeneity in the autosomal recessive adult forms of limb-girdle muscular dystrophy following linkage analysis with 15q probes in Brazilian families.. Journal of Medical Genetics, 1993, 30, 385-387.	1.5	30
61	A novel complex neurological phenotype due to a homozygous mutation in FDX2. Brain, 2018, 141, 2289-2298.	3.7	29
62	Relation between height and clinical course in Duchenne muscular dystrophy. American Journal of Medical Genetics Part A, 1988, 29, 405-410.	2.4	28
63	Point mutation in a Becker muscular dystrophy patient. Human Molecular Genetics, 1993, 2, 75-77.	1.4	28
64	Merosin-positive congenital muscular dystrophy in two siblings with cataract and slight mental retardation. Brain and Development, 1999, 21, 274-278.	0.6	28
65	Myotonic dystrophy: genetic, clinical, and molecular analysis of patients from 41 Brazilian families.. Journal of Medical Genetics, 1995, 32, 14-18.	1.5	27
66	Protein defects in neuromuscular diseases. Brazilian Journal of Medical and Biological Research, 2003, 36, 543-555.	0.7	27
67	Systemic Delivery of Human Mesenchymal Stromal Cells Combined with IGF-1 Enhances Muscle Functional Recovery in LAMA2dy/2j Dystrophic Mice. Stem Cell Reviews and Reports, 2013, 9, 93-109.	5.6	27
68	Milder course in Duchenne patients with nonsense mutations and no muscle dystrophin. Neuromuscular Disorders, 2014, 24, 986-989.	0.3	27
69	Partial ?-sarcoglycan deficiency with retention of the dystrophin-glycoprotein complex in a LGMD2D family. , 2000, 23, 984-988.		26
70	The effect of calpain 3 deficiency on the pattern of muscle degeneration in the earliest stages of LGMD2A. Journal of Clinical Pathology, 2003, 56, 624-626.	1.0	26
71	Preclinical Studies with Umbilical Cord Mesenchymal Stromal Cells in Different Animal Models for Muscular Dystrophy. Journal of Biomedicine and Biotechnology, 2011, 2011, 1-9.	3.0	26
72	Differential Expression of Genes Involved in the Degeneration and Regeneration Pathways in Mouse Models for Muscular Dystrophies. NeuroMolecular Medicine, 2012, 14, 74-83.	1.8	25

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73	Human Adipose Tissue Derived Pericytes Increase Life Span in Utrn tm1Ked Dmd mdx /J Mice. Stem Cell Reviews and Reports, 2014, 10, 830-840.	5.6	25
74	Severe nonspecific X-linked mental retardation caused by a proximally Xp located gene: Intragenic heterogeneity or a new form of X-linked mental retardation?. American Journal of Medical Genetics Part A, 1993, 46, 172-175.	2.4	24
75	Cosegregation of schizophrenia with Becker muscular dystrophy: susceptibility locus for schizophrenia at Xp21 or an effect of the dystrophin gene in the brain?. Journal of Medical Genetics, 1993, 30, 131-134.	1.5	24
76	No evidence of genetic heterogeneity in Brazilian facioscapulohumeral muscular dystrophy familes (FSHD) with 4q markers. Human Molecular Genetics, 1993, 2, 557-562.	1.4	23
77	Congenital muscular dystrophy with cerebral white matter hypodensity. Correlation of clinical features and merosin deficiency. Brain and Development, 1996, 18, 53-58.	0.6	23
78	Deficiency of Î±-Actinin-3 (ACTN3) Occurs in Different Forms of Muscular Dystrophy. Neuropediatrics, 1997, 28, 223-228.	0.3	23
79	Intrafamilial variability in dystrophin abundance correlated with difference in the severity of the phenotype. Journal of the Neurological Sciences, 1993, 119, 38-42.	0.3	22
80	Further evidence for the organisation of the four sarcoglycans proteins within the dystrophinâ€“glycoprotein complex. European Journal of Human Genetics, 1999, 7, 251-254.	1.4	22
81	Calpainopathy: How Broad Is the Spectrum of Clinical Variability?. Journal of Molecular Neuroscience, 2003, 21, 233-236.	1.1	21
82	Muscle Phenotypic Variability in Limb Girdle Muscular Dystrophy 2 G. Journal of Molecular Neuroscience, 2013, 50, 339-344.	1.1	21
83	Limb girdle muscular dystrophy type 2G with myopathic-neurogenic motor unit potentials and a novel muscle image pattern. BMC Clinical Pathology, 2014, 14, 41.	1.8	19
84	Quantitative T2 Combined with Texture Analysis of Nuclear Magnetic Resonance Images Identify Different Degrees of Muscle Involvement in Three Mouse Models of Muscle Dystrophy: mdx, Largemyd and mdx/Largemyd. PLoS ONE, 2015, 10, e0117835.	1.1	19
85	Sarcoglycanopathies: an update. Neuromuscular Disorders, 2021, 31, 1021-1027.	0.3	19
86	Novel point mutations in the dystrophin gene. Human Mutation, 1997, 10, 217-222.	1.1	18
87	Muscle Protein Alterations in LGMD2I Patients With Different Mutations in the Fukutin-related Protein Gene. Journal of Histochemistry and Cytochemistry, 2008, 56, 995-1001.	1.3	18
88	Assessment of the 50-kDa dystrophin-associated glycoprotein in Brazilian patients with severe childhood autosomal recessive muscular dystrophy. Journal of the Neurological Sciences, 1994, 123, 122-128.	0.3	17
89	Lack of the C-terminal domain of nebulin in a patient with nemaline myopathy. Muscle and Nerve, 2002, 25, 747-752.	1.0	17
90	Idiopathic hyperCKemia and malignant hyperthermia susceptibility. Canadian Journal of Anaesthesia, 2017, 64, 1202-1210.	0.7	17

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91	A normal life without muscle dystrophin. Neuromuscular Disorders, 2015, 25, 371-374.	0.3	16
92	Neuromuscular disorders: genes, genetic counseling and therapeutic trials. Genetics and Molecular Biology, 2016, 39, 339-348.	0.6	16
93	Screening of male patients with autosomal recessive Duchenne dystrophy through dystrophin and DNA studies. American Journal of Medical Genetics Part A, 1991, 39, 38-41.	2.4	15
94	A family with McLeod syndrome and calpainopathy with clinically overlapping diseases. Neurology, 2005, 65, 1832-1833.	1.5	15
95	Stem cells from umbilical cord blood do have myogenic potential, with and without differentiation induction in vitro. Journal of Translational Medicine, 2009, 7, 6.	1.8	15
96	Necklace fibers as histopathological marker in a patient with severe form of X-linked myotubular myopathy. Neuromuscular Disorders, 2012, 22, 541-545.	0.3	15
97	Clinical and Molecular Characterization of Mcardleâ€™s Disease in Brazilian Patients. NeuroMolecular Medicine, 2013, 15, 470-475.	1.8	15
98	Neurocognitive Impairment in mdx Mice. Molecular Neurobiology, 2019, 56, 7608-7616.	1.9	15
99	Efficient exon skipping of SGCG mutations mediated by phosphorodiamidate morpholino oligomers. JCI Insight, 2018, 3, .	2.3	15
100	Brazilian family with pure autosomal dominant spastic paraplegia maps to 8q: Analysis of muscle beta 1 syntrophin. , 2000, 92, 122-127.		14
101	Reviewing Large LAMA2 Deletions and Duplications in Congenital Muscular Dystrophy Patients. Journal of Neuromuscular Diseases, 2014, 1, 169-179.	1.1	14
102	The mdx Mutation in the 129/Sv Background Results in a Milder Phenotype: Transcriptome Comparative Analysis Searching for the Protective Factors. PLoS ONE, 2016, 11, e0150748.	1.1	14
103	Familial occurrence of Duchenne dystrophy through paternal lines in four families. American Journal of Medical Genetics Part A, 1991, 38, 80-84.	2.4	13
104	Is dystrophin always altered in Becker muscular dystrophy patients?. Journal of the Neurological Sciences, 1995, 131, 99-104.	0.3	13
105	Segregation distortion of the CTG repeats at the myotonic dystrophy (DM) locus: new data from Brazilian DM families.. Journal of Medical Genetics, 1997, 34, 790-791.	1.5	13
106	Clinical diagnosis of heterozygous dystrophin gene deletions by fluorescence in situ hybridization. Neuromuscular Disorders, 1998, 8, 447-452.	0.3	13
107	Screening for Mutations in the RYR1 Gene in Families with Malignant Hyperthermia. Journal of Molecular Neuroscience, 2003, 21, 35-42.	1.1	13
108	Rod Distribution and Muscle Fiber Type Modification in the Progression of Nemaline Myopathy. Journal of Child Neurology, 2003, 18, 235-240.	0.7	13

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109	Oxidative variables and antioxidant enzymes activities in the mdx mouse brain. Neurochemistry International, 2009, 55, 802-805.	1.9	13
110	Mitochondrial alterations in dynamin 2-related centronuclear myopathy. Arquivos De Neuro-Psiquiatria, 2009, 67, 102-104.	0.3	13
111	Effect of mazindol on growth hormone levels in patients with Duchenne muscular dystrophy. American Journal of Medical Genetics Part A, 1988, 31, 821-833.	2.4	12
112	Diagnosis and Molecular Characterization of Nonclassic Forms of Tay-Sachs Disease in Brazil. Journal of Child Neurology, 2006, 21, 540-544.	0.7	12
113	A new evidence for the maintenance of the sarcoglycan complex in muscle sarcolemma in spite of the primary absence of Î´-SG protein. Journal of Molecular Medicine, 2007, 85, 415-420.	1.7	12
114	Mitochondrial respiratory chain and creatine kinase activities in mdx mouse brain. Muscle and Nerve, 2010, 41, 257-260.	1.0	12
115	Nocturnal rhythm of growth hormone in Duchenne patients: Effect of different doses of mazindol and/or cyproheptadine. American Journal of Medical Genetics Part A, 1989, 33, 457-467.	2.4	11
116	Limb-girdle syndrome: a genetic study of 22 large Brazilian families comparison with X-linked Duchenne and Becker dystrophies. Journal of the Neurological Sciences, 1991, 103, 65-75.	0.3	11
117	Sarcoglycanopathies: A Multiplex Molecular Analysis for the Most Common Mutations. Diagnostic Molecular Pathology, 2006, 15, 95-100.	2.1	11
118	Linkage analysis in families with autosomal recessive limb-girdle muscular dystrophy (LGMD) and 6q probes flanking the dystrophin-related sequence. American Journal of Medical Genetics Part A, 1991, 38, 140-146.	2.4	10
119	Comparative transcriptome analysis of muscular dystrophy models Largemyd, Dmdmdx/Largemyd and Dmdmdx: what makes them different?. European Journal of Human Genetics, 2016, 24, 1301-1309.	1.4	10
120	Additional dystrophin fragment in Becker muscular dystrophy patients: Correlation with the pattern of DNA deletion. American Journal of Medical Genetics Part A, 1992, 44, 382-384.	2.4	9
121	Paternal inheritance or different mutations in maternally related patients occur in about 3% of Duchenne familial cases. , 1998, 78, 361-365.		9
122	Does the A3333G Mutation in theCACNL1A3Gene, Detected in Malignant Hyperthermia, Also Occur in Central Core Disease?. Genetic Testing and Molecular Biomarkers, 2000, 4, 383-386.	1.7	9
123	Heterogeneity of Classic Congenital Muscular Dystrophy With Involvement of the Central Nervous System: Report of Five Atypical Cases. Journal of Child Neurology, 2000, 15, 172-178.	0.7	9
124	Deficiency of Muscle Î±-Actinin-3 is Compatible with High Muscle Performance. Journal of Molecular Neuroscience, 2003, 20, 39-42.	1.1	9
125	A novel missense mutation in the caveolin-3 gene in rippling muscle disease. Muscle and Nerve, 2007, 36, 258-260.	1.0	9
126	Striatum brain-derived neurotrophic factor levels are decreased in dystrophin-deficient mice. Neuroscience Letters, 2009, 459, 66-68.	1.0	9

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127	Assessing Pathogenicity for Novel Mutation/Sequence Variants: The Value of Healthy Older Individuals. NeuroMolecular Medicine, 2012, 14, 281-284.	1.8	9
128	Dmdmdx/Largemyd: a new mouse model of neuromuscular diseases useful for studying physiopathological mechanisms and testing therapies. DMM Disease Models and Mechanisms, 2013, 6, 1167-74.	1.2	9
129	Facioscapulohumeral (FSHD1) and other forms of muscular dystrophy in the same family: is there more in muscular dystrophy than meets the eye?. Neuromuscular Disorders, 2002, 12, 554-557.	0.3	8
130	Mitochondrial Cardioencephalomyopathy Due to a Novel SCO2 Mutation in a Brazilian Patient. JAMA Neurology, 2013, 70, 258.	4.5	8
131	Activity of Krebs cycle enzymes in mdx mice. Muscle and Nerve, 2016, 53, 91-95.	1.0	8
132	CorrelaÃ§Ã£o entre o tempo de realizaÃ§Ã£o de diferentes atividades fÃsicas por portadores de distrofia muscular de Duchenne. Revista De Terapia Ocupacional Da Universidade De SÃ£o Paulo, 2003, 14, 133.	0.1	7
133	Mutation analysis in the FKRP gene provides an explanation for a rare cause of intrafamilial clinical variability in LGMD2I. Neuromuscular Disorders, 2006, 16, 870-873.	0.3	7
134	Reduction of acethylcolinesterase activity in the brain of mdx mice. Neuromuscular Disorders, 2011, 21, 359-362.	0.3	7
135	Renewed Avenues through Exercise Muscle Contractility and Inflammatory Status. Scientific World Journal, The, 2012, 2012, 1-7.	0.8	7
136	Thomsen or Becker myotonia? A novel autosomal recessive nonsense mutation in the CLCN1 gene associated with a mild phenotype. Muscle and Nerve, 2012, 45, 279-283.	1.0	7
137	Manifesting carriers of X-linked myotubular myopathy. Neurology: Genetics, 2020, 6, e513.	0.9	7
138	Satellite cells deficiency and defective regeneration in dynamin 2â€related centronuclear myopathy. FASEB Journal, 2021, 35, e21346.	0.2	7
139	Skeletal Muscle Injury by Electroporation: A Model to Study Degeneration/Regeneration Pathways in Muscle. Methods in Molecular Biology, 2020, 2063, 157-169.	0.4	7
140	Immunological Methods for the Analysis of Protein Expression in Neuromuscular Diseases. , 2003, 217, 355-378.		7
141	Hypothesis: The existence of embryonic and adult isoforms of mRNA dystrophin provides an explanation for unusual clinical findings. American Journal of Medical Genetics Part A, 1989, 32, 438-441.	2.4	6
142	Exclusion of the 15q locus as a candidate gene for severe childhood autosomal recessive Duchenne-like muscular dystrophy in Brazilian families. Human Molecular Genetics, 1993, 2, 201-202.	1.4	6
143	Protein and DNA Analysis for the Prenatal Diagnosis of Î±2-Lamininâ€“Deficient Congenital Muscular Dystrophy. Diagnostic Molecular Pathology, 2004, 13, 167-171.	2.1	6
144	Concordant utrophin upregulation in phenotypically discordant DMD/BMD brothers. Neuromuscular Disorders, 2016, 26, 197-200.	0.3	6

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145	Altered in vitro muscle differentiation in X-linked myopathy with excessive autophagy (XMEA). DMM Disease Models and Mechanisms, 2020, 13, .	1.2	6
146	Reviewing Large LAMA2 Deletions and Duplications in Congenital Muscular Dystrophy Patients. Journal of Neuromuscular Diseases, 2014, 1, 169-179.	1.1	6
147	Serum CK-MB activity in progressive muscular dystrophy: Is it of nosologic value?. American Journal of Medical Genetics Part A, 1985, 22, 81-87.	2.4	5
148	Central Nervous System Involvement in the Animal Model of Myodystrophy. Molecular Neurobiology, 2013, 48, 71-77.	1.9	5
149	Behavioral Responses in Animal Model of Congenital Muscular Dystrophy 1D. Molecular Neurobiology, 2016, 53, 402-407.	1.9	5
150	233rd ENMC International Workshop:. Neuromuscular Disorders, 2018, 28, 540-549.	0.3	5
151	Mazindol and growth hormone inhibition in Duchenne muscular dystrophy. American Journal of Medical Genetics Part A, 1987, 27, 993-995.	2.4	4
152	Serum Creatine Kinase in Progressive Muscular Dystrophies. , 2001, , 31-49.		4
153	Faster regeneration associated to high expression of Fam65b and Hdac6 in dysferlin-deficient mouse. Journal of Molecular Histology, 2019, 50, 375-387.	1.0	4
154	Sarcolemmal distribution of abnormal dystrophin in Xp21 carriers. Neuromuscular Disorders, 1993, 3, 135-140.	0.3	3
155	CTG repeat length in muscle from patients affected with myotonic dystrophy (DM). Journal of Medical Genetics, 1996, 33, 173-173.	1.5	3
156	CaracterizaÃ§Ã£o da passagem da postura de bipedestaÃ§Ã£o para a de sedestaÃ§Ã£o no solo, em crianÃ§as portadoras de Distrofia Muscular de Duchenne. Revista De Terapia Ocupacional Da Universidade De SÃ£o Paulo, 2002, 13, 31.	0.1	3
157	Immune-mediated rippling muscle disease in a patient with treated hypothyroidism. Journal of the Neurological Sciences, 2017, 383, 53-55.	0.3	3
158	LMNA-Related Muscular Dystrophy with Clinical Intrafamilial Variability. Journal of Molecular Neuroscience, 2019, 69, 623-627.	1.1	3
159	A Novel SPEG mutation causing congenital myopathy with fiber size disproportion and dilated cardiomyopathy with heart transplantation. Neuromuscular Disorders, 2021, 31, 1199-1206.	0.3	3
160	Congenital Muscular Dystrophy 1D Causes Matrix Metalloproteinase Activation And Blood-Brain Barrier Impairment. Current Neurovascular Research, 2017, 14, 60-64.	0.4	3
161	Central Core Disease: Facial Weakness Differentiating Biallelic from Monoallelic Forms. Genes, 2022, 13, 760.	1.0	3
162	Steroids in duchenne muscular dystrophy. Neuromuscular Disorders, 1992, 2, 59.	0.3	2

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163	Transposon-like element in the dystrophin gene. American Journal of Medical Genetics Part A, 1993, 46, 601-601.	2.4	2
164	Muscular dystrophies presenting with proximal muscle weakness. , 0, , 230-256.		2
165	Central core myopathy with autophagy. Muscle and Nerve, 2017, 56, E8-E9.	1.0	2
166	Muscular Dystrophies and Protein Mutations. , 2007, , 391-407.		2
167	Exclusion of the gene responsible for facioscapulohumeral muscular dystrophy (FSH) at 6q23-q27. Journal of the Neurological Sciences, 1991, 102, 206-208.	0.3	1
168	Genetic variability in the myostatin gene does not explain the muscle hypertrophy and clinical penetrance in myotonia congenita. Muscle and Nerve, 2010, 41, 427-428.	1.0	1
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