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	Effects of low-intensity training on the brain and muscle in the congenital muscular dystrophy 1D model. Neurological Sciences, 2022, , 1.	0.9	0
2	Central Core Disease: Facial Weakness Differentiating Biallelic from Monoallelic Forms. Genes, 2022, 13, 760.	1.0	3
3	Predictive factors of the contracture test for diagnosing malignant hyperthermia in a Brazilian population sample: a retrospective observational study. Brazilian Journal of Anesthesiology (Elsevier), 2022, , .	0.2	1
4	Association of Three Different Mutations in the CLCN1 Gene Modulating the Phenotype in a Consanguineous Family with Myotonia Congenita. Journal of Molecular Neuroscience, 2021, 71, 2275-2280.	1.1	0
5	Satellite cells deficiency and defective regeneration in dynamin 2â€related centronuclear myopathy. FASEB Journal, 2021, 35, e21346.	0.2	7
6	Muscle regeneration in spastic muscles of children with cerebral palsy. Developmental Medicine and Child Neurology, 2021, 63, 1137-1137.	1.1	0
7	Sarcoglycanopathies: an update. Neuromuscular Disorders, 2021, 31, 1021-1027.	0.3	19
8	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
9	Altered in vitro muscle differentiation in X-linked myopathy with excessive autophagy (XMEA). DMM Disease Models and Mechanisms, 2020, 13, .	1.2	6
10	Manifesting carriers of X-linked myotubular myopathy. Neurology: Genetics, 2020, 6, e513.	0.9	7
11	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
12	Isolation and Characterization of Muscle-Derived Stem Cells from Dystrophic Mouse Models. Methods in Molecular Biology, 2020, 2063, 171-180.	0.4	0
13	Dominant or recessive mutations in the gene causing central core myopathy in Brazilian patients. Acta Myologica, 2020, 39, 274-282.	1.5	1
14	LMNA-Related Muscular Dystrophy with Clinical Intrafamilial Variability. Journal of Molecular Neuroscience, 2019, 69, 623-627.	1.1	3
15	Muscle satellite cells and impaired late stage regeneration in different murine models for muscular dystrophies. Scientific Reports, 2019, 9, 11842.	1.6	39
16	P.54Defects in iron-sulphur cluster assembly proteins ISCU and FDX2 cause characteristic mitochondrial myopathy. Neuromuscular Disorders, 2019, 29, S56-S57.	0.3	0
17	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
18	Neurocognitive Impairment in mdx Mice. Molecular Neurobiology, 2019, 56, 7608-7616.	1.9	15

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19	233rd ENMC International Workshop:. Neuromuscular Disorders, 2018, 28, 540-549.	0.3	5
20	A novel complex neurological phenotype due to a homozygous mutation in FDX2. Brain, 2018, 141, 2289-2298.	3.7	29
21	Efficient exon skipping of SGCG mutations mediated by phosphorodiamidate morpholino oligomers. JCI Insight, 2018, 3, .	2.3	15
22	Central core myopathy with autophagy. Muscle and Nerve, 2017, 56, E8-E9.	1.0	2
23	Myocardial Fibrosis Progression in Duchenne and Becker Muscular Dystrophy. JAMA Cardiology, 2017, 2, 190.	3.0	79
24	Immune-mediated rippling muscle disease in a patient with treated hypothyroidism. Journal of the Neurological Sciences, 2017, 383, 53-55.	0.3	3
25	Idiopathic hyperCKemia and malignant hyperthermia susceptibility. Canadian Journal of Anaesthesia, 2017, 64, 1202-1210.	0.7	17
26	Congenital Muscular Dystrophy 1D Causes Matrix Metalloproteinase Activation And Blood-Brain Barrier Impairment. Current Neurovascular Research, 2017, 14, 60-64.	0.4	3
27	Neuromuscular disorders: genes, genetic counseling and therapeutic trials. Genetics and Molecular Biology, 2016, 39, 339-348.	0.6	16
28	Muscle Satellite Cells: Exploring the Basic Biology to Rule Them. Stem Cells International, 2016, 2016, 1-14.	1.2	33
29	Analysis of the autophagic pathway during in vitro muscle differentiation in X-linked myopathy with excessive autophagy. Neuromuscular Disorders, 2016, 26, S194.	0.3	0
30	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
31	Activity of Krebs cycle enzymes in mdx mice. Muscle and Nerve, 2016, 53, 91-95.	1.0	8
32	Concordant utrophin upregulation in phenotypically discordant DMD/BMD brothers. Neuromuscular Disorders, 2016, 26, 197-200.	0.3	6
33	Behavioral Responses in Animal Model of Congenital Muscular Dystrophy 1D. Molecular Neurobiology, 2016, 53, 402-407.	1.9	5
34	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
35	Next generation sequencing (NGS): A powerful tool for studying rigid spine patients and multiminicore myopathy. Neuromuscular Disorders, 2015, 25, S269.	0.3	0
36	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

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37	A normal life without muscle dystrophin. Neuromuscular Disorders, 2015, 25, 371-374.	0.3	16
38	Steroid benefit in a laminopathy-congenital muscular dystrophy patient with dropped head syndrome: A 10-year follow-up. Neuromuscular Disorders, 2015, 25, S279-S280.	0.3	0
39	The mdx mutation in the 129/Sv background results in a milder phenotype: Transcriptome comparative analysis searching for the protective factors. Neuromuscular Disorders, 2015, 25, S291.	0.3	0
40	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
41	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
42	Reviewing Large LAMA2 Deletions and Duplications in Congenital Muscular Dystrophy Patients. Journal of Neuromuscular Diseases, 2014, 1, 169-179.	1.1	14
43	G.P.319. Neuromuscular Disorders, 2014, 24, 917-918.	0.3	0
44	A.P.14. Neuromuscular Disorders, 2014, 24, 834-835.	0.3	1
45	Silent polymorphisms in the RYR1 gene do not modify the phenotype of the p.4898 I>T pathogenic mutation in central core disease: a case report. BMC Research Notes, 2014, 7, 487.	0.6	1
46	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
47	G.P.19. Neuromuscular Disorders, 2014, 24, 800.	0.3	0
48	Approach to the diagnosis of congenital myopathies. Neuromuscular Disorders, 2014, 24, 97-116.	0.3	239
49	Milder course in Duchenne patients with nonsense mutations and no muscle dystrophin. Neuromuscular Disorders, 2014, 24, 986-989.	0.3	27
50	Reviewing Large LAMA2 Deletions and Duplications in Congenital Muscular Dystrophy Patients. Journal of Neuromuscular Diseases, 2014, 1, 169-179.	1.1	6
51	Clinical and Molecular Characterization of Mcardleâ€™s Disease in Brazilian Patients. NeuroMolecular Medicine, 2013, 15, 470-475.	1.8	15
52	Muscle Phenotypic Variability in Limb Girdle Muscular Dystrophy 2 G. Journal of Molecular Neuroscience, 2013, 50, 339-344.	1.1	21
53	Central Nervous System Involvement in the Animal Model of Myodystrophy. Molecular Neurobiology, 2013, 48, 71-77.	1.9	5
54	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

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55	P.5.8 Why is LGMD2G rare?. Neuromuscular Disorders, 2013, 23, 766.	0.3	0
56	P.4.13 Central core disease (CCD): Improving the screening for mutations in RYR1 gene. Neuromuscular Disorders, 2013, 23, 763.	0.3	0
57	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
58	K7del is a common TPM2 gene mutation associated with nemaline myopathy and raised myofibre calcium sensitivity. Brain, 2013, 136, 494-507.	3.7	42
59	Mitochondrial Cardioencephalomyopathy Due to a Novel SCO2 Mutation in a Brazilian Patient. JAMA Neurology, 2013, 70, 258.	4.5	8
60	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
61	Consensus Statement on Standard of Care for Congenital Myopathies. Journal of Child Neurology, 2012, 27, 363-382.	0.7	147
62	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
63	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
64	Assessing Pathogenicity for Novel Mutation/Sequence Variants: The Value of Healthy Older Individuals. NeuroMolecular Medicine, 2012, 14, 281-284.	1.8	9
65	Renewed Avenues through Exercise Muscle Contractility and Inflammatory Status. Scientific World Journal, The, 2012, 2012, 1-7.	0.8	7
66	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
67	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
68	Reduction of acethylcolinesterase activity in the brain of mdx mice. Neuromuscular Disorders, 2011, 21, 359-362.	0.3	7
69	P1.24 Microarray analysis of two exceptional Golden Retriever Muscular Dystrophy (GRMD) dogs with no dystrophin and a mild course. Neuromuscular Disorders, 2011, 21, 648-649.	0.3	0
70	Myogenic Differentiation of ES Cells for Therapies in Neuromuscular Diseases: Progress to Date. , 2011, , .		0
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	Motor Chip: A Comparative Genomic Hybridization Microarray for Copy-Number Mutations in 245 Neuromuscular Disorders. Clinical Chemistry, 2011, 57, 1584-1596.	1.5	48

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73	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
74	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
75	Mitochondrial respiratory chain and creatine kinase activities in mdx mouse brain. Muscle and Nerve, 2010, 41, 257-260.	1.0	12
76	Ringo: Discordance between the molecular and clinical manifestation in a Golden Retriever Muscular Dystrophy dog. Neuromuscular Disorders, 2010, 20, 64-70.	0.3	33
77	EM.P.3.03 Differential expression of genes involved in muscular degeneration in four dystrophic mouse models. Neuromuscular Disorders, 2009, 19, 574-575.	0.3	0
78	Striatum brain-derived neurotrophic factor levels are decreased in dystrophin-deficient mice. Neuroscience Letters, 2009, 459, 66-68.	1.0	9
79	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
80	Oxidative variables and antioxidant enzymes activities in the mdx mouse brain. Neurochemistry International, 2009, 55, 802-805.	1.9	13
81	Mitochondrial alterations in dynamin 2-related centronuclear myopathy. Arquivos De Neuro-Psiquiatria, 2009, 67, 102-104.	0.3	13
82	Animal Models for Genetic Neuromuscular Diseases. Journal of Molecular Neuroscience, 2008, 34, 241-248.	1.1	106
83	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
84	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
85	Ringo, a Golden Retriever Muscular Dystrophy (GRMD) dog with absent dystrophin but normal strength. Neuromuscular Disorders, 2008, 18, 892-893.	0.3	41
86	T.P.2.13 Murine embryonic stem cells injected into mdx mouse â€“ In vivo myogenic capacity and immunogenic reaction. Neuromuscular Disorders, 2008, 18, 759-760.	0.3	0
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	C.P.4.08 Screening for mutations in the dynamin 2 gene in Brazilian patients with centronuclear myopathy and Charcot-Marie-Tooth neuropathy. Neuromuscular Disorders, 2007, 17, 880-881.	0.3	0
89	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
90	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

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91	A novel missense mutation in the caveolin-3 gene in rippling muscle disease. Muscle and Nerve, 2007, 36, 258-260.	1.0	9
92	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
93	Muscular Dystrophies and Protein Mutations. , 2007, , 391-407.		2
94	G.P.6 01 Utrophin in the dystrophic muscle: evaluation of the contribution of connective tissue replacement. Neuromuscular Disorders, 2006, 16, 697-698.	0.3	0
95	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
96	Sarcoglycanopathies: A Multiplex Molecular Analysis for the Most Common Mutations. Diagnostic Molecular Pathology, 2006, 15, 95-100.	2.1	11
97	Diagnosis and Molecular Characterization of Nonclassic Forms of Tay-Sachs Disease in Brazil. Journal of Child Neurology, 2006, 21, 540-544.	0.7	12
98	A family with McLeod syndrome and calpainopathy with clinically overlapping diseases. Neurology, 2005, 65, 1832-1833.	1.5	15
99	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
100	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
101	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
102	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
103	Deficiency of Muscle Î±-Actinin-3 is Compatible with High Muscle Performance. Journal of Molecular Neuroscience, 2003, 20, 39-42.	1.1	9
104	Screening for Mutations in the RYR1 Gene in Families with Malignant Hyperthermia. Journal of Molecular Neuroscience, 2003, 21, 35-42.	1.1	13
105	Calpainopathy: How Broad Is the Spectrum of Clinical Variability?. Journal of Molecular Neuroscience, 2003, 21, 233-236.	1.1	21
106	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
107	The 10 autosomal recessive limb-girdle muscular dystrophies. Neuromuscular Disorders, 2003, 13, 532-544.	0.3	122
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	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
110	Protein defects in neuromuscular diseases. Brazilian Journal of Medical and Biological Research, 2003, 36, 543-555.	0.7	27
111	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
112	Immunological Methods for the Analysis of Protein Expression in Neuromuscular Diseases. , 2003, 217, 355-378.		7
113	Telethonin protein expression in neuromuscular disorders. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2002, 1588, 33-40.	1.8	49
114	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
115	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
116	Lack of the C-terminal domain of nebulin in a patient with nemaline myopathy. Muscle and Nerve, 2002, 25, 747-752.	1.0	17
117	Clinical variability in calpainopathy: What makes the difference?. European Journal of Human Genetics, 2002, 10, 825-832.	1.4	84
118	Characterization of Human Skeletal Muscle Ankrd2. Biochemical and Biophysical Research Communications, 2001, 285, 378-386.	1.0	51
119	Nebulin expression in patients with nemaline myopathy. Neuromuscular Disorders, 2001, 11, 154-162.	0.3	39
120	Serum Creatine Kinase in Progressive Muscular Dystrophies. , 2001, , 31-49.		4
121	Mutations in the caveolin-3 gene: When are they pathogenic?. American Journal of Medical Genetics Part A, 2001, 99, 303-307.	2.4	43
122	Dysferlin Protein Analysis in Limb-Girdle Muscular Dystrophies. Journal of Molecular Neuroscience, 2001, 17, 71-80.	1.1	67
123	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
124	Brazilian family with pure autosomal dominant spastic paraplegia maps to 8q: Analysis of muscle beta 1 syntrophin. , 2000, 92, 122-127.		14
125	Partial ?-sarcoglycan deficiency with retention of the dystrophin-glycoprotein complex in a LGMD2D family. , 2000, 23, 984-988.		26
126	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

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127	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
128	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
129	Mialgias post ejercicio como forma de presentaciÃ³n de una distrofinopatÃa.: Case report.. Revista Medica De Chile, 2000, 128, .	0.1	1
130	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
131	Seven autosomal recessive limb-girdle muscular dystrophies in the Brazilian population: from LGMD2A to LGMD2G. , 1999, 82, 392-398.		90
132	Merosin-positive congenital muscular dystrophy in two siblings with cataract and slight mental retardation. Brain and Development, 1999, 21, 274-278.	0.6	28
133	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
134	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
135	Paternal inheritance or different mutations in maternally related patients occur in about 3% of Duchenne familial cases. , 1998, 78, 361-365.		9
136	Absence of correlation between skewed X inactivation in blood and serum creatine-kinase levels in Duchenne/Becker female carriers. , 1998, 80, 356-361.		31
137	Clinical diagnosis of heterozygous dystrophin gene deletions by fluorescence in situ hybridization. Neuromuscular Disorders, 1998, 8, 447-452.	0.3	13
138	Caveolin-3 in muscular dystrophy. Human Molecular Genetics, 1998, 7, 871-877.	1.4	200
139	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
140	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	1
141	Deficiency of Î±-Actinin-3 (ACTN3) Occurs in Different Forms of Muscular Dystrophy. Neuropediatrics, 1997, 28, 223-228.	0.3	23
142	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
143	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
144	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

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145	Novel point mutations in the dystrophin gene. Human Mutation, 1997, 10, 217-222.	1.1	18
146	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
147	The phenotype of chromosome 2p-linked limb-girdle muscular dystrophy. Neuromuscular Disorders, 1996, 6, 483-490.	0.3	42
148	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
149	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
150	The sarcoglycan complex in the six autosomal recessive limb-girdle muscular dystrophies. Human Molecular Genetics, 1996, 5, 1963-1969.	1.4	167
151	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
152	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
153	CTG repeat length in muscle from patients affected with myotonic dystrophy (DM). Journal of Medical Genetics, 1996, 33, 173-173.	1.5	3
154	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
155	Myotonic dystrophy: genetic, clinical, and molecular analysis of patients from 41 Brazilian families.. Journal of Medical Genetics, 1995, 32, 14-18.	1.5	27
156	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
157	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
158	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
159	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
160	Is dystrophin always altered in Becker muscular dystrophy patients?. Journal of the Neurological Sciences, 1995, 131, 99-104.	0.3	13
161	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
162	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

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163	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
164	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
165	Transposon-like element in the dystrophin gene. American Journal of Medical Genetics Part A, 1993, 46, 601-601.	2.4	2
166	Sarcolemmal distribution of abnormal dystrophin in Xp21 carriers. Neuromuscular Disorders, 1993, 3, 135-140.	0.3	3
167	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
168	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
169	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
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