Andrea Martinuzzi

List of Publications by Year
in descending order

160 papers	7,029 citations	61857 43 h-index	69108 77 g-index
166 all docs	166 docs citations	166 times ranked	10025 citing authors

#	Article	IF	Citations
1	Monoallelic KIF1A-related disorders: a multicenter cross sectional study and systematic literature review. Journal of Neurology, 2022, 269, 437-450.	1.8	12
2	Trends observed in bilateral cerebral palsy during a thirty-year period: A cohort study with an ICF-based overview. Pediatrics and Neonatology, 2021, 62, 284-291.	0.3	1
3	Editorial: Hereditary Spastic Paraplegias: At the Crossroads of Molecular Pathways and Clinical Options. Frontiers in Neuroscience, 2021, 15, 708642.	1.4	3
4	Brain Structure and Degeneration Staging in Friedreich Ataxia: <scp>Magnetic Resonance Imaging</scp> Volumetrics from the <scp>ENIGMAâ€Ataxia</scp> Working Group. Annals of Neurology, 2021, 90, 570-583.	2.8	27
5	Clinical practice guidelines for glycogen storage disease V & VII (McArdle disease and Tarui) Tj ETQq1 1 0.784314 rgBT /Overlock	0.3	22
6	Functional MRI Studies in Friedreich's Ataxia: A Systematic Review. Frontiers in Neurology, 2021, 12, 802496.	1.1	2
7	Towards Consensus on Good Practices for the Use of New Technologies for Intervention and Support in Developmental Dyslexia: A Delphi Study Conducted among Italian Specialized Professionals. Children, 2021, 8, 1126.	0.6	5
8	Ontological modeling of the International Classification of Functioning, Disabilities and Health (ICF): Activities&Participation and Environmental Factors components. BMC Medical Informatics and Decision Making, 2021, 21, 367.	1.5	8
9	Adaptor protein complex 4 deficiency: a paradigm of childhood-onset hereditary spastic paraplegia caused by defective protein trafficking. Human Molecular Genetics, 2020, 29, 320-334.	1.4	45
10	Defining the clinical, molecular and imaging spectrum of adaptor protein complex 4-associated hereditary spastic paraplegia. Brain, 2020, 143, 2929-2944.	3.7	29
11	Creation and implementation of a European registry for patients with McArdle disease and other muscle glycogenoses (EUROMAC registry). Orphanet Journal of Rare Diseases, 2020, 15, 187.	1.2	3
12	Brain Magnetic Spectroscopy Imaging and Hereditary Spastic Paraplegia: A Focused Systematic Review on Current Landmarks and Future Perspectives. Frontiers in Neurology, 2020, 11, 515.	1.1	2
13	Data from the European registry for patients with McArdle disease and other muscle glycogenoses (EUROMAC). Orphanet Journal of Rare Diseases, 2020, 15, 330.	1.2	23
14	Sensitivity of Neuroimaging Indicators in Monitoring the Effects of Interferon Gamma Treatment in Friedreichâ€™s Ataxia. Frontiers in Neuroscience, 2020, 14, 872.	1.4	7
15	Changes in Psychiatric Diagnoses During the Transition Phase from Childhood to Adulthood in a Group of Patients with Intellectual Disability. Adolescent Psychiatry (Hilversum, Netherlands), 2020, 10, 41-47.	0.1	5
16	Multimodal MRI Longitudinal Assessment of White and Gray Matter in Different SPG Types of Hereditary Spastic Paraparesis. Frontiers in Neuroscience, 2020, 14, 325.	1.4	6
17	Efficacy of a Combined Treatment of Botulinum Toxin and Intensive Physiotherapy in Hereditary Spastic Paraplegia. Frontiers in Neuroscience, 2020, 14, 111.	1.4	17
18	Interaction Between Mitochondrial DNA Variants and Mitochondria/Endoplasmic Reticulum Contact Sites: A Perspective Review. DNA and Cell Biology, 2020, 39, 1431-1443.	0.9	1

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19	Safety and Efficacy Of Interferon Î³ in Friedreich's Ataxia. Movement Disorders, 2020, 35, 370-371.	2.2	10
20	Psychosocial impact of sport activity in neuromuscular disorders. Neurological Sciences, 2020, 41, 2561-2567.	0.9	8
21	Toward a Harmonized WHO Family of International Classifications Content Model. Studies in Health Technology and Informatics, 2020, 270, 1409-1410.	0.2	3
22	Reduction in respiratory exacerbation rate in patients with severe bilateral cerebral palsy following daily PEP-mask therapy: a retrospective study. European Journal of Physical and Rehabilitation Medicine, 2020, 56, 68-72.	1.1	1
23	Becoming a young adult with cerebral palsy. Research in Developmental Disabilities, 2019, 92, 103450.	1.2	5
24	Loss of paraplegin drives spasticity rather than ataxia in a cohort of 241 patients with SPG7. Neurology, 2019, 92, e2679-e2690.	1.5	49
25	Optical Coherence Tomography in a Cohort of Genetically Defined Hereditary Spastic Paraplegia: A Brief Research Report. Frontiers in Neurology, 2019, 10, 1193.	1.1	8
26	Haplogroup J mitogenomes are the most sensitive to the pesticide rotenone: Relevance for human diseases. Neurobiology of Disease, 2018, 114, 129-139.	2.1	22
27	Functional and Structural Brain Damage in Friedreich's Ataxia. Frontiers in Neurology, 2018, 9, 747.	1.1	25
28	Factors Related to Unemployment in Europe. A Cross-Sectional Study from the COURAGE Survey in Finland, Poland and Spain. International Journal of Environmental Research and Public Health, 2018, 15, 722.	1.2	14
29	Determinants of mobility in populations of older adults: Results from a cross-sectional study in Finland, Poland and Spain. Maturitas, 2018, 115, 84-91.	1.0	10
30	Peculiar combinations of individually non-pathogenic missense mitochondrial DNA variants cause low penetrance Leberâ€™s hereditary optic neuropathy. PLoS Genetics, 2018, 14, e1007210.	1.5	47
31	Benefits of Centralized Scheduling in a Postacute Residential Rehabilitation Program for People With Acquired Brain Lesions: A Pilot Study. Archives of Physical Medicine and Rehabilitation, 2017, 98, 746-750.	0.5	1
32	Care pathways models and clinical outcomes in Disorders of consciousness. Brain and Behavior, 2017, 7, e00740.	1.0	15
33	Hereditary spastic paraplegia type 5: natural history, biomarkers and a randomized controlled trial. Brain, 2017, 140, 3112-3127.	3.7	87
34	Determinants of Quality of Life in Ageing Populations: Results from a Cross-Sectional Study in Finland, Poland and Spain. PLoS ONE, 2016, 11, e0159293.	1.1	64
35	Clinical and Paraclinical Indicators of Motor System Impairment in Hereditary Spastic Paraplegia: A Pilot Study. PLoS ONE, 2016, 11, e0153283.	1.1	41
36	Safety profile of incobotulinum toxin A [XeominÂ®] in gastrocnemious muscles injections in children with cerebral palsy: Randomized double-blind clinical trial. European Journal of Paediatric Neurology, 2016, 20, 532-537.	0.7	27

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37	Do we really need to open a classification box on personal factors in ICF?. Disability and Rehabilitation, 2016, 38, 1327-1328.	0.9	14
38	Clinical and Pulmonary Function Markers of Respiratory Exacerbation Risk in Subjects With Quadriplegic Cerebral Palsy. Respiratory Care, 2015, 60, 1431-1437.	0.8	10
39	Pediatric neurorehabilitation and the ICF. NeuroRehabilitation, 2015, 36, 31-36.	0.5	8
40	Variants in KIF1A gene in dominant and sporadic forms of hereditary spastic paraparesis. Journal of Neurology, 2015, 262, 2684-2690.	1.8	55
41	A Long Term Effects of a New Onset Psychosis after DBS Treated with Quetiapine in a Patient with Parkinson's Disease. Psychiatry Investigation, 2015, 12, 146.	0.7	4
42	Pharmacological and nutritional treatment for McArdle disease (Glycogen Storage Disease type V). The Cochrane Library, 2014, 2014, CD003458.	1.5	54
43	Individual and group treatment for patients with acquired brain injury in comprehensive rehabilitation. Brain Injury, 2014, 28, 1102-1108.	0.6	15
44	Cholestenoic acids regulate motor neuron survival via liver X receptors. Journal of Clinical Investigation, 2014, 124, 4829-4842.	3.9	84
45	The blurred scenario of motor neuron disorders linked toSpatacsinmutations: a case report. European Journal of Neurology, 2014, 21, e85-e86.	1.7	2
46	Determinants of Health and Disability in Ageing Population: The COURAGE in Europe Project (Collaborative Research on Ageing in Europe). Clinical Psychology and Psychotherapy, 2014, 21, 193-198.	1.4	70
47	Mapping SAGE questionnaire to the International Classification of Functioning, Disability and Health (ICF). Clinical Psychology and Psychotherapy, 2014, 21, 199-203.	1.4	2
48	Validation of the COURAGE Built Environment Selfâ€Reported Questionnaire. Clinical Psychology and Psychotherapy, 2014, 21, 215-226.	1.4	8
49	Sociodemographic features and diagnoses as predictors of severe disability in a sample of adults applying for disability certification. International Journal of Rehabilitation Research, 2014, 37, 180-186.	0.7	7
50	Mutations in CYP2U1, DDHD2 and GBA2 genes are rare causes of complicated forms of hereditary spastic paraparesis. Journal of Neurology, 2014, 261, 373-381.	1.8	62
51	Mutation Analysis of MFN2, GJB1, MPZ and PMP22 in Italian Patients with Axonal Charcotâ€“Marieâ€“Tooth Disease. NeuroMolecular Medicine, 2014, 16, 540-550.	1.8	21
52	Multidimensional outcome measure of selective dorsal rhizotomy in spastic cerebral palsy. European Journal of Paediatric Neurology, 2014, 18, 704-713.	0.7	27
53	A Novel in-Frame 18-bp Microdeletion inMT-CYBCauses a Multisystem Disorder with Prominent Exercise Intolerance. Human Mutation, 2014, 35, 954-958.	1.1	38
54	A survey on feasibility of ICF-CY use to describe persisting difficulties in executing tasks and activities of children and adolescent with disability in Italy. Disability and Health Journal, 2014, 7, 433-441.	1.6	11

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55	Brain White Matter Involvement in Hereditary Spastic Paraplegias: Analysis with Multiple Diffusion Tensor Indices. American Journal of Neuroradiology, 2014, 35, 1533-1538.	1.2	18
56	Cybrid studies establish the causal link between the mtDNA m.3890G>A/MT-ND1 mutation and optic atrophy with bilateral brainstem lesions. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2013, 1832, 445-452.	1.8	17
57	The cytochrome b p.278Y>C mutation causative of a multisystem disorder enhances superoxide production and alters supramolecular interactions of respiratory chain complexes. Human Molecular Genetics, 2013, 22, 2141-2151.	1.4	46
58	Implementation of an ICF-based project/program in a pediatric neuro-rehabiltation hospital: follow-up evaluation by stakeholders. Disability and Rehabilitation, 2013, 35, 1059-1064.	0.9	11
59	Defective autophagy in spastizin mutated patients with hereditary spastic paraparesis type 15. Brain, 2013, 136, 3119-3139.	3.7	74
60	Functioning and disability of children and adolescents in a vegetative state and a minimally conscious state. International Journal of Rehabilitation Research, 2012, 35, 352-359.	0.7	22
61	Pediatric Biobanking: A Pilot Qualitative Survey of Practices, Rules, and Researcher Opinions in Ten European Countries. Biopreservation and Biobanking, 2012, 10, 29-36.	0.5	22
62	Nonâ€convulsive status epilepticus of frontal origin in mucopolysaccharidosis type II successfully treated with ethosuximide. Developmental Medicine and Child Neurology, 2012, 54, 961-964.	1.1	14
63	Psychological Profile in Children and Adolescents with Severe Course Juvenile Idiopathic Arthritis. Scientific World Journal, The, 2012, 2012, 1-7.	0.8	18
64	Children in Vegetative State and Minimally Conscious State: Patientsâ€™ Condition and Caregiversâ€™ Burden. Scientific World Journal, The, 2012, 2012, 1-7.	0.8	18
65	A Population Survey in Italy Based on the ICF Classification: Recognizing Persons with Severe Disability. Scientific World Journal, The, 2012, 2012, 1-9.	0.8	10
66	Clinical phenotype variability in patients with hereditary spastic paraplegia type 5 associated with CYP7B1 mutations. Clinical Genetics, 2012, 81, 150-157.	1.0	42
67	Mutations in the motor and stalk domains of KIF5A in spastic paraplegia type 10 and in axonal Charcotâ€“Marieâ€“Tooth type 2. Clinical Genetics, 2012, 82, 157-164.	1.0	128
68	Oestrogens ameliorate mitochondrial dysfunction in Leberâ€™s hereditary optic neuropathy. Brain, 2011, 134, 220-234.	3.7	208
69	The role of pH on the thermodynamics and kinetics of muscle biochemistry: An in vivo study by 31P-MRS in patients with myo-phosphorylase deficiency. Biochimica Et Biophysica Acta - Bioenergetics, 2011, 1807, 1244-1249.	0.5	3
70	A novel mutation in KIF5A gene causing hereditary spastic paraplegia with axonal neuropathy. Neurological Sciences, 2011, 32, 665-668.	0.9	21
71	A novel nonsense mutation in the APTX gene associated with delayed DNA singleâ€strand break removal fails to enhance sensitivity to different genotoxic agents. Human Mutation, 2011, 32, E2118-33.	1.1	12
72	Pharmacological and nutritional treatment for McArdle disease (Glycogen Storage Disease type V). , 2010, , CD003458.		11

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73	The International Classification of Functioning Disability and Health, version for children and youth as a roadmap for projecting and programming rehabilitation in a neuropaediatric hospital unit. Journal of Rehabilitation Medicine, 2010, 42, 49-55.	0.8	31
74	SEVERE CMT TYPE 2 WITH FATAL ENCEPHALOPATHY ASSOCIATED WITH A NOVEL MFN2 SPLICING MUTATION. Neurology, 2010, 74, 1919-1921.	1.5	28
75	The genetic and metabolic signature of oncocytic transformation implicates HIF1Î± destabilization. Human Molecular Genetics, 2010, 19, 1019-1032.	1.4	113
76	Respiratory Complex I Dysfunction Due to Mitochondrial DNA Mutations Shifts the Voltage Threshold for Opening of the Permeability Transition Pore toward Resting Levels. Journal of Biological Chemistry, 2009, 284, 2045-2052.	1.6	91
77	Point mutations and a large intragenic deletion in SPG11 in complicated spastic paraplegia without thin corpus callosum. Journal of Medical Genetics, 2009, 46, 345-351.	1.5	30
78	The ICF and Labour Policies Project: The first Italian nation-wide experience of ICF implementation in the labour sector. Disability and Rehabilitation, 2009, 31, S16-S21.	0.9	11
79	Projecting and programming rehabilitation based on ICF-CY format in a neuropediatric hospital unit. Disability and Rehabilitation, 2009, 31, S55-S60.	0.9	7
80	Moving towards ICF use for monitoring the UN Convention on the rights of persons with disabilities: The Italian experience. Disability and Rehabilitation, 2009, 31, S74-S77.	0.9	14
81	Preliminary results of ICF dissemination in primary health care in Mozambique: Sharing the Italian experience. Disability and Rehabilitation, 2009, 31, S78-S82.	0.9	2
82	ICF and ICF-CY for an innovative holistic approach to persons with chronic conditions. Disability and Rehabilitation, 2009, 31, S83-S87.	0.9	21
83	Towards a common disability assessment framework: theoretical and methodological issues for providing public services and benefits using ICF. Disability and Rehabilitation, 2009, 31, S8-S15.	0.9	25
84	The dystonic child treated with deep brain stimulation: ICF reading of a high-tech approach. Disability and Rehabilitation, 2009, 31, S159-S169.	0.9	3
85	Functioning and disability in patients with Angelman syndrome: utility of the International Classification of functioning disability and health, children and youth adaptation framework. Disability and Rehabilitation, 2009, 31, S121-S127.	0.9	12
86	Eight years of ICF in Italy: Principles, results and future perspectives. Disability and Rehabilitation, 2009, 31, S4-S7.	0.9	10
87	Italian ICF training programs: Describing and promoting human functioning and research. Disability and Rehabilitation, 2009, 31, S46-S49.	0.9	29
88	Children with disability at school: the application of ICF-CY in the Veneto region. Disability and Rehabilitation, 2009, 31, S67-S73.	0.9	21
89	Cytosolic pH buffering during exercise and recovery in skeletal muscle of patients with McArdleâ€™s disease. European Journal of Applied Physiology, 2009, 105, 687-694.	1.2	12
90	Prevalence and correlates of mental disorders among adolescents in Italy: the PrISMA study. European Child and Adolescent Psychiatry, 2009, 18, 217-226.	2.8	126

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91	Homotypic fusion of ER membranes requires the dynamin-like GTPase Atlastin. Nature, 2009, 460, 978-983.	13.7	419
92	Hippocampal remodelling after MDMA neurotoxicity: A single case study. World Journal of Biological Psychiatry, 2009, 10, 961-968.	1.3	5
93	Respiratory Complex I Dysfunction Due to Mitochondrial DNA Mutations Shifts the Voltage Threshold for Opening of the Permeability Transition Pore toward Resting Levels. Biophysical Journal, 2009, 96, 529a.	0.2	0
94	PRIMARY OBSTRUCTION OF THE FOURTH VENTRICLE OUTLETS. Neurosurgery, 2009, 65, 1078-1086.	0.6	49
95	The first Italian family with evidence of pyramidal impairment as phenotypic manifestation of Silver syndrome BSCL2 gene mutation. Neurological Sciences, 2008, 29, 189-191.	0.9	12
96	Antioxidants partially restore glutamate transport defect in leber hereditary optic neuropathy cybrids. Journal of Neuroscience Research, 2008, 86, 3331-3337.	1.3	26
97	Randomized, placeboâ€controlled, doubleâ€blind pilot trial of ramipril in McArdle's disease. Muscle and Nerve, 2008, 37, 350-357.	1.0	33
98	A clinical, genetic, and biochemical characterization ofSPG7mutations in a large cohort of patients with hereditary spastic paraplegia. Human Mutation, 2008, 29, 522-531.	1.1	85
99	Pharmacological and nutritional treatment for McArdle disease (Glycogen Storage Disease type V). , 2008, , CD003458.		12
100	Disseminating the WHO International Classification of Functioning Health and Disability (ICF) in the Veneto region of Italy. Disability and Rehabilitation, 2008, 30, 71-80.	0.9	11
101	Protection against Oxidant-Induced Apoptosis by Exogenous Glutathione in Leber Hereditary Optic Neuropathy Cybrids. , 2008, 49, 671.		41
102	Mitochondrial DNA background modulates the assembly kinetics of OXPHOS complexes in a cellular model of mitochondrial disease. Human Molecular Genetics, 2008, 17, 4001-4011.	1.4	140
103	Endoscopic anatomy of the fourth ventricle. Journal of Neurosurgery, 2008, 109, 530-535.	0.9	32
104	ENDOSCOPIC ANATOMY OF THE CEREBRAL AQUEDUCT. Operative Neurosurgery, 2007, 61, 1-7.	0.4	22
105	Severe head injury in early infancy: analysis of causes and possible predictive factors for outcome. Child's Nervous System, 2007, 23, 873-880.	0.6	29
106	Aquaporin(s) Expression in Choroid Plexus Tumours. Pediatric Neurosurgery, 2006, 42, 228-233.	0.4	63
107	Mitochondrial disease activates transcripts of the unfolded protein response and cell cycle and inhibits vesicular secretion and oligodendrocyte-specific transcripts. Mitochondrion, 2006, 6, 161-175.	1.6	59
108	Aquaporin 1 expression in cystic hemangioblastomas. Neuroscience Letters, 2006, 392, 178-180.	1.0	26

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109	New Mutations in TK2 Gene Associated With Mitochondrial DNA Depletion. Pediatric Neurology, 2006, 34, 177-185.	1.0	63
110	Exploring mental status in Friedreich's ataxia: a combined neuropsychological, behavioral and neuroimaging study. European Journal of Neurology, 2006, 13, 827-835.	1.7	76
111	McArdle disease: the mutation spectrum ofPYGMin a large Italian cohort. Human Mutation, 2006, 27, 718-718.	1.1	52
112	The Italian Preadolescent Mental Health Project (PrISMA): rationale and methods. International Journal of Methods in Psychiatric Research, 2006, 15, 22-35.	1.1	63
113	The first ALS2 missense mutation associated with JPLS reveals new aspects of alsin biological function. Brain, 2006, 129, 1710-1719.	3.7	87
114	Coiling and neuroendoscopy: a new perspective in the treatment of intraventricular haemorrhages due to bleeding aneurysms. Journal of Neurology, Neurosurgery and Psychiatry, 2006, 77, 1354-1358.	0.9	18
115	Eight Novel Mutations in SPG4 in a Large Sample of Patients With Hereditary Spastic Paraplegia. Archives of Neurology, 2006, 63, 750.	4.9	39
116	Defective Oxidative Phosphorylation in Thyroid Oncocytic Carcinoma Is Associated with Pathogenic Mitochondrial DNA Mutations Affecting Complexes I and III. Cancer Research, 2006, 66, 6087-6096.	0.4	204
117	Neuroendoscopic Aspiration of Hematocephalus Totalis: Technical Note. Operative Neurosurgery, 2005, 57, ONS-E409-ONS-E409.	0.4	29
118	Antioxidant defences in cybrids harboring mtDNA mutations associated with Leber's hereditary optic neuropathy. FEBS Journal, 2005, 272, 1124-1135.	2.2	96
119	Caspase-independent death of Leberâ€™s hereditary optic neuropathy cybrids is driven by energetic failure and mediated by AIF and Endonuclease G. Apoptosis: an International Journal on Programmed Cell Death, 2005, 10, 997-1007.	2.2	113
120	Training on the International Classification of Functioning, Disability and Health (ICF): the ICFâ€“DIN Basic and the ICFâ€“DIN Advanced Course developed by the Disability Italian Network. Journal of Headache and Pain, 2005, 6, 159-164.	2.5	43
121	Complex I deficiency primes Bax-dependent neuronal apoptosis through mitochondrial oxidative damage. Proceedings of the National Academy of Sciences of the United States of America, 2005, 102, 19126-19131.	3.3	273
122	Free Mg2+ concentration in the calf muscle of glycogen phosphorylase and phosphofructokinase deficiency patients assessed in different metabolic conditions by 31P MRS. Dynamic Medicine: DM, 2005, 4, 7.	2.7	8
123	Isolation of transcriptomal changes attributable to LHON mutations and the cybridization process. Brain, 2005, 128, 1026-1037.	3.7	44
124	Severe Impairment of Complex Iâ€“Driven Adenosine Triphosphate Synthesis in Leber Hereditary Optic Neuropathy Cybrids. Archives of Neurology, 2005, 62, 730.	4.9	144
125	Disease-related phenotypes in a Drosophila model of hereditary spastic paraplegia are ameliorated by treatment with vinblastine. Journal of Clinical Investigation, 2005, 115, 3026-3034.	3.9	99
126	Failure of Endoscopic Third Ventriculostomy in the Treatment of Idiopathic Normal Pressure Hydrocephalus. Minimally Invasive Neurosurgery, 2004, 47, 342-345.	0.9	43

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127	Leber hereditary optic neuropathy mtDNA mutations disrupt glutamate transport in cybrid cell lines. Brain, 2004, 127, 2183-2192.	3.7	106
128	Bioenergetics shapes cellular death pathways in Leber's hereditary optic neuropathy: a model of mitochondrial neurodegeneration. Biochimica Et Biophysica Acta - Bioenergetics, 2004, 1658, 172-179.	0.5	102
129	International Classification of Functioning, Disability and Health in a cohort of children with cognitive, motor, and complex disabilities. Developmental Medicine and Child Neurology, 2004, 46, 98-106.	1.1	20
130	International Classification of Functioning, Disability and Health in a cohort of children with cognitive, motor, and complex disabilities. Developmental Medicine and Child Neurology, 2004, 46, 98-106.	1.1	34
131	Apoptotic Cell Death of Cybrid Cells Bearing Leber's Hereditary Optic Neuropathy Mutations Is Caspase Independent. Annals of the New York Academy of Sciences, 2003, 1010, 213-217.	1.8	41
132	Phenotype modulators in myophosphorylase deficiency. Annals of Neurology, 2003, 53, 497-502.	2.8	101
133	Leber's Hereditary Optic Neuropathy (LHON) Pathogenic Mutations Induce Mitochondrial-dependent Apoptotic Death in Transmitochondrial Cells Incubated with Galactose Medium. Journal of Biological Chemistry, 2003, 278, 4145-4150.	1.6	169
134	Applying the International Classification of Functioning, Disability and Health (ICF) to measure childhood disability. Disability and Rehabilitation, 2003, 25, 602-610.	0.9	268
135	Novel LGI1 mutation in a family with autosomal dominant partial epilepsy with auditory features. Neurology, 2003, 60, 1687-1690.	1.5	43
136	Endoscopic Anatomic Features of the Triangular Recess. Neurosurgery, 2003, 52, 1491-1494.	0.6	3
137	Cells Bearing Mutations Causing Leber's Hereditary Optic Neuropathy Are Sensitized to Fas-induced Apoptosis. Journal of Biological Chemistry, 2002, 277, 5810-5815.	1.6	122
138	Respiratory function in cybrid cell lines carrying European mtDNA haplogroups: implications for Leber's hereditary optic neuropathy. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2002, 1588, 7-14.	1.8	55
139	Rescue of a mitochondrial deficiency causing Leber hereditary optic neuropathy. Annals of Neurology, 2002, 52, 534-542.	2.8	253
140	Endoscopic Treatment of Colloid Cysts of the Third Ventricle:9 Consecutive Cases. Minimally Invasive Neurosurgery, 2000, 43, 118-123.	0.9	26
141	A Stop-Codon Mutation in the Human mtDNA Cytochrome c Oxidase I Gene Disrupts the Functional Structure of Complex IV. American Journal of Human Genetics, 1999, 65, 611-620.	2.6	148
142	McArdle's Disease. American Journal of Pathology, 1999, 154, 1893-1897.	1.9	18
143	Guillainâ€“BarrÃ© syndrome with associated thrombocytopenia: prompt response to combined corticosteroid and immunoglobulin treatment. Neuromuscular Disorders, 1998, 8, 50-52.	0.3	7
144	Molecular characterization of myophosphorylase deficiency in a group of patients from Northern Italy. Journal of the Neurological Sciences, 1996, 137, 14-19.	0.3	45

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145	Residual muscle cytochrome c oxidase activity accounts for submaximal exercise lactate threshold in chronic progressive external ophthalmoplegia. , 1996, 19, 342-349.		14
146	Two novel missense mutations (E654K, L396P) in caucasian patients with myophosphorylase deficiency (McArdle's disease). Human Mutation, 1995, 6, 276-277.	1.1	16
147	MtDNA Mutations Associated with Leberâ€²s Hereditary Optic Neuropathy: Studies on Cytoplasmic Hybrid (Cybrid) Cells. Biochemical and Biophysical Research Communications, 1995, 210, 880-888.	1.0	117
148	Myophosphorylase deficiency affects muscle mitochondrial respiration as shown by 31P-MR spectroscopy in a case with associated multifocal encephalopathy. Journal of the Neurological Sciences, 1995, 128, 84-91.	0.3	16
149	Clinical and Biochemical Aspects of Carnitine Deficiency and Insufficiency: Transport Defects and Inborn Errors of Î²-Oxidation. Critical Reviews in Clinical Laboratory Sciences, 1992, 29, 217-242.	2.7	40
150	Correlation between clinical and molecular features in two MELAS families. Journal of the Neurological Sciences, 1992, 113, 222-229.	0.3	45
151	Paralysis of Innervated Cultured Human Muscle Fibers Affects Enzymes Differentially. Journal of Neurochemistry, 1990, 54, 223-229.	2.1	10
152	Liver fatty acid-binding protein in two cases of human lipid storage. Molecular and Cellular Biochemistry, 1990, 98, 225-30.	1.4	20
153	Histoenzymatic profile of human muscle cultured in monolayer and innervated de novo by fetal rat spinal cord. Muscle and Nerve, 1988, 11, 1-9.	1.0	29
154	Asynchronous regulation of muscle specific isozymes of creatine kinase, glycogen phosphorylase, lactic dehydrogenase and phosphoglycerate mutase in innervated and non-innervated cultured human muscle. Neuroscience Letters, 1988, 89, 216-222.	1.0	32
155	Developmental expression of the muscle-specific isozyme of phosphoglycerate mutase in human muscle cultured in monolayer and innervated by fetal rat spinal cord. Experimental Neurology, 1987, 96, 365-375.	2.0	24
156	Effects of electrical stimulation and tetrodotoxin paralysis on expression of muscle-specific isozymes of four enzymes in aneurally cultured embryonic rat muscle. Experimental Neurology, 1987, 97, 739-745.	2.0	9
157	Accumulation of CK-MM is impaired in innervated and contracting cultured muscle fibers of duchenne muscular dystrophy patients. Life Sciences, 1987, 41, 927-933.	2.0	6
158	De novo neuromuscular junction formation on human muscle fibres cultured in monolayer and innervated by foetal rat spinal cord: Ultrastructural and ultrastructural-cytochemical studies. Journal of Neurocytology, 1987, 16, 523-537.	1.6	97
159	FORDYSVAR EBOOK: Best practices and technological resources for students with Specific Learning Difficulties (SpLDs). , 0, , .		1
160	FORDYSVAR: Book on specific learning difficulties in reading. , 0, , .		0