Andrea Martinuzzi

List of Publications by Year
in descending order

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160 papers	7,029 citations	61984 43 h-index	69250 77 g-index
166 all docs	166 docs citations	166 times ranked	10025 citing authors

#	Article	IF	Citations
1	Homotypic fusion of ER membranes requires the dynamin-like GTPase Atlastin. Nature, 2009, 460, 978-983.	27.8	419
2	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.	7.1	273
3	Applying the International Classification of Functioning, Disability and Health (ICF) to measure childhood disability. Disability and Rehabilitation, 2003, 25, 602-610.	1.8	268
4	Rescue of a mitochondrial deficiency causing Leber hereditary optic neuropathy. Annals of Neurology, 2002, 52, 534-542.	5.3	253
5	Oestrogens ameliorate mitochondrial dysfunction in Leberâ€™s hereditary optic neuropathy. Brain, 2011, 134, 220-234.	7.6	208
6	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.	0.9	204
7	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.	3.4	169
8	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.	6.2	148
9	Severe Impairment of Complex Iâ€“Driven Adenosine Triphosphate Synthesis in Leber Hereditary Optic Neuropathy Cybrids. Archives of Neurology, 2005, 62, 730.	4.5	144
10	Mitochondrial DNA background modulates the assembly kinetics of OXPHOS complexes in a cellular model of mitochondrial disease. Human Molecular Genetics, 2008, 17, 4001-4011.	2.9	140
11	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.	2.0	128
12	Prevalence and correlates of mental disorders among adolescents in Italy: the PrISMA study. European Child and Adolescent Psychiatry, 2009, 18, 217-226.	4.7	126
13	Cells Bearing Mutations Causing Leber's Hereditary Optic Neuropathy Are Sensitized to Fas-induced Apoptosis. Journal of Biological Chemistry, 2002, 277, 5810-5815.	3.4	122
14	MtDNA Mutations Associated with Leberâ€²s Hereditary Optic Neuropathy: Studies on Cytoplasmic Hybrid (Cybrid) Cells. Biochemical and Biophysical Research Communications, 1995, 210, 880-888.	2.1	117
15	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.	4.9	113
16	The genetic and metabolic signature of oncocytic transformation implicates HIF1Î± destabilization. Human Molecular Genetics, 2010, 19, 1019-1032.	2.9	113
17	Leber hereditary optic neuropathy mtDNA mutations disrupt glutamate transport in cybrid cell lines. Brain, 2004, 127, 2183-2192.	7.6	106
18	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.	1.0	102

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19	Phenotype modulators in myophosphorylase deficiency. Annals of Neurology, 2003, 53, 497-502.	5.3	101
20	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.	8.2	99
21	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.	1.5	97
22	Antioxidant defences in cybrids harboring mtDNA mutations associated with Leber's hereditary optic neuropathy. FEBS Journal, 2005, 272, 1124-1135.	4.7	96
23	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.	3.4	91
24	The first ALS2 missense mutation associated with JPLS reveals new aspects of alsin biological function. Brain, 2006, 129, 1710-1719.	7.6	87
25	Hereditary spastic paraplegia type 5: natural history, biomarkers and a randomized controlled trial. Brain, 2017, 140, 3112-3127.	7.6	87
26	A clinical, genetic, and biochemical characterization ofSPG7mutations in a large cohort of patients with hereditary spastic paraplegia. Human Mutation, 2008, 29, 522-531.	2.5	85
27	Cholestenoic acids regulate motor neuron survival via liver X receptors. Journal of Clinical Investigation, 2014, 124, 4829-4842.	8.2	84
28	Exploring mental status in Friedreich's ataxia: a combined neuropsychological, behavioral and neuroimaging study. European Journal of Neurology, 2006, 13, 827-835.	3.3	76
29	Defective autophagy in spastizin mutated patients with hereditary spastic paraparesis type 15. Brain, 2013, 136, 3119-3139.	7.6	74
30	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.	2.7	70
31	Determinants of Quality of Life in Ageing Populations: Results from a Cross-Sectional Study in Finland, Poland and Spain. PLoS ONE, 2016, 11, e0159293.	2.5	64
32	Aquaporin(s) Expression in Choroid Plexus Tumours. Pediatric Neurosurgery, 2006, 42, 228-233.	0.7	63
33	New Mutations in TK2 Gene Associated With Mitochondrial DNA Depletion. Pediatric Neurology, 2006, 34, 177-185.	2.1	63
34	The Italian Preadolescent Mental Health Project (PrISMA): rationale and methods. International Journal of Methods in Psychiatric Research, 2006, 15, 22-35.	2.1	63
35	Mutations in CYP2U1, DDHD2 and GBA2 genes are rare causes of complicated forms of hereditary spastic paraparesis. Journal of Neurology, 2014, 261, 373-381.	3.6	62
36	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.	3.4	59

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37	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.	3.8	55
38	Variants in KIF1A gene in dominant and sporadic forms of hereditary spastic paraparesis. Journal of Neurology, 2015, 262, 2684-2690.	3.6	55
39	Pharmacological and nutritional treatment for McArdle disease (Glycogen Storage Disease type V). The Cochrane Library, 2014, 2014, CD003458.	2.8	54
40	McArdle disease: the mutation spectrum ofPYGMin a large Italian cohort. Human Mutation, 2006, 27, 718-718.	2.5	52
41	PRIMARY OBSTRUCTION OF THE FOURTH VENTRICLE OUTLETS. Neurosurgery, 2009, 65, 1078-1086.	1.1	49
42	Loss of paraplegin drives spasticity rather than ataxia in a cohort of 241 patients with SPG7. Neurology, 2019, 92, e2679-e2690.	1.1	49
43	Peculiar combinations of individually non-pathogenic missense mitochondrial DNA variants cause low penetrance Leberâ€™s hereditary optic neuropathy. PLoS Genetics, 2018, 14, e1007210.	3.5	47
44	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.	2.9	46
45	Correlation between clinical and molecular features in two MELAS families. Journal of the Neurological Sciences, 1992, 113, 222-229.	0.6	45
46	Molecular characterization of myophosphorylase deficiency in a group of patients from Northern Italy. Journal of the Neurological Sciences, 1996, 137, 14-19.	0.6	45
47	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.	2.9	45
48	Isolation of transcriptomal changes attributable to LHON mutations and the cybridization process. Brain, 2005, 128, 1026-1037.	7.6	44
49	Novel LGI1 mutation in a family with autosomal dominant partial epilepsy with auditory features. Neurology, 2003, 60, 1687-1690.	1.1	43
50	Failure of Endoscopic Third Ventriculostomy in the Treatment of Idiopathic Normal Pressure Hydrocephalus. Minimally Invasive Neurosurgery, 2004, 47, 342-345.	0.9	43
51	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.	6.0	43
52	Clinical phenotype variability in patients with hereditary spastic paraplegia type 5 associated with CYP7B1 mutations. Clinical Genetics, 2012, 81, 150-157.	2.0	42
53	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.	3.8	41
54	Protection against Oxidant-Induced Apoptosis by Exogenous Glutathione in Leber Hereditary Optic Neuropathy Cybrids. , 2008, 49, 671.		41

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55	Clinical and Paraclinical Indicators of Motor System Impairment in Hereditary Spastic Paraplegia: A Pilot Study. PLoS ONE, 2016, 11, e0153283.	2.5	41
56	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.	6.1	40
57	Eight Novel Mutations in SPG4 in a Large Sample of Patients With Hereditary Spastic Paraplegia. Archives of Neurology, 2006, 63, 750.	4.5	39
58	A Novel in-Frame 18-bp Microdeletion inMT-CYBCauses a Multisystem Disorder with Prominent Exercise Intolerance. Human Mutation, 2014, 35, 954-958.	2.5	38
59	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.	2.1	34
60	Randomized, placeboâ€controlled, doubleâ€blind pilot trial of ramipril in McArdle's disease. Muscle and Nerve, 2008, 37, 350-357.	2.2	33
61	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.	2.1	32
62	Endoscopic anatomy of the fourth ventricle. Journal of Neurosurgery, 2008, 109, 530-535.	1.6	32
63	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.	1.1	31
64	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.	3.2	30
65	Histoenzymatic profile of human muscle cultured in monolayer and innervated de novo by fetal rat spinal cord. Muscle and Nerve, 1988, 11, 1-9.	2.2	29
66	Neuroendoscopic Aspiration of Hematocephalus Totalis: Technical Note. Operative Neurosurgery, 2005, 57, ONS-E409-ONS-E409.	0.8	29
67	Severe head injury in early infancy: analysis of causes and possible predictive factors for outcome. Child's Nervous System, 2007, 23, 873-880.	1.1	29
68	Italian ICF training programs: Describing and promoting human functioning and research. Disability and Rehabilitation, 2009, 31, S46-S49.	1.8	29
69	Defining the clinical, molecular and imaging spectrum of adaptor protein complex 4-associated hereditary spastic paraplegia. Brain, 2020, 143, 2929-2944.	7.6	29
70	SEVERE CMT TYPE 2 WITH FATAL ENCEPHALOPATHY ASSOCIATED WITH A NOVEL MFN2 SPLICING MUTATION. Neurology, 2010, 74, 1919-1921.	1.1	28
71	Multidimensional outcome measure of selective dorsal rhizotomy in spastic cerebral palsy. European Journal of Paediatric Neurology, 2014, 18, 704-713.	1.6	27
72	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.	1.6	27

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73	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.	5.3	27
74	Endoscopic Treatment of Colloid Cysts of the Third Ventricle:9 Consecutive Cases. Minimally Invasive Neurosurgery, 2000, 43, 118-123.	0.9	26
75	Aquaporin 1 expression in cystic hemangioblastomas. Neuroscience Letters, 2006, 392, 178-180.	2.1	26
76	Antioxidants partially restore glutamate transport defect in leber hereditary optic neuropathy cybrids. Journal of Neuroscience Research, 2008, 86, 3331-3337.	2.9	26
77	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.	1.8	25
78	Functional and Structural Brain Damage in Friedreich's Ataxia. Frontiers in Neurology, 2018, 9, 747.	2.4	25
79	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.	4.1	24
80	Data from the European registry for patients with McArdle disease and other muscle glycogenoses (EUROMAC). Orphanet Journal of Rare Diseases, 2020, 15, 330.	2.7	23
81	ENDOSCOPIC ANATOMY OF THE CEREBRAL AQUEDUCT. Operative Neurosurgery, 2007, 61, 1-7.	0.8	22
82	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.	1.3	22
83	Pediatric Biobanking: A Pilot Qualitative Survey of Practices, Rules, and Researcher Opinions in Ten European Countries. Biopreservation and Biobanking, 2012, 10, 29-36.	1.0	22
84	Haplogroup J mitogenomes are the most sensitive to the pesticide rotenone: Relevance for human diseases. Neurobiology of Disease, 2018, 114, 129-139.	4.4	22
85	Clinical practice guidelines for glycogen storage disease V & VII (McArdle disease and Tarui) Tj ETQq1 1 0.784314 rgBT /Overlock	0.6	22
86	ICF and ICF-CY for an innovative holistic approach to persons with chronic conditions. Disability and Rehabilitation, 2009, 31, S83-S87.	1.8	21
87	Children with disability at school: the application of ICF-CY in the Veneto region. Disability and Rehabilitation, 2009, 31, S67-S73.	1.8	21
88	A novel mutation in KIF5A gene causing hereditary spastic paraplegia with axonal neuropathy. Neurological Sciences, 2011, 32, 665-668.	1.9	21
89	Mutation Analysis of MFN2, GJB1, MPZ and PMP22 in Italian Patients with Axonal Charcotâ€“Marieâ€“Tooth Disease. NeuroMolecular Medicine, 2014, 16, 540-550.	3.4	21
90	Liver fatty acid-binding protein in two cases of human lipid storage. Molecular and Cellular Biochemistry, 1990, 98, 225-30.	3.1	20

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91	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.	2.1	20
92	McArdle's Disease. American Journal of Pathology, 1999, 154, 1893-1897.	3.8	18
93	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.	1.9	18
94	Psychological Profile in Children and Adolescents with Severe Course Juvenile Idiopathic Arthritis. Scientific World Journal, The, 2012, 2012, 1-7.	2.1	18
95	Children in Vegetative State and Minimally Conscious State: Patientsâ€™ Condition and Caregiversâ€™ Burden. Scientific World Journal, The, 2012, 2012, 1-7.	2.1	18
96	Brain White Matter Involvement in Hereditary Spastic Paraplegias: Analysis with Multiple Diffusion Tensor Indices. American Journal of Neuroradiology, 2014, 35, 1533-1538.	2.4	18
97	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.	3.8	17
98	Efficacy of a Combined Treatment of Botulinum Toxin and Intensive Physiotherapy in Hereditary Spastic Paraplegia. Frontiers in Neuroscience, 2020, 14, 111.	2.8	17
99	Two novel missense mutations (E654K, L396P) in caucasian patients with myophosphorylase deficiency (McArdle's disease). Human Mutation, 1995, 6, 276-277.	2.5	16
100	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.	0.6	16
101	Individual and group treatment for patients with acquired brain injury in comprehensive rehabilitation. Brain Injury, 2014, 28, 1102-1108.	1.2	15
102	Care pathways models and clinical outcomes in Disorders of consciousness. Brain and Behavior, 2017, 7, e00740.	2.2	15
103	Residual muscle cytochrome c oxidase activity accounts for submaximal exercise lactate threshold in chronic progressive external ophthalmoplegia. , 1996, 19, 342-349.		14
104	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.	1.8	14
105	Nonâ€convulsive status epilepticus of frontal origin in mucopolysaccharidosis type II successfully treated with ethosuximide. Developmental Medicine and Child Neurology, 2012, 54, 961-964.	2.1	14
106	Do we really need to open a classification box on personal factors in ICF?. Disability and Rehabilitation, 2016, 38, 1327-1328.	1.8	14
107	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.	2.6	14
108	The first Italian family with evidence of pyramidal impairment as phenotypic manifestation of Silver syndrome BSCL2 gene mutation. Neurological Sciences, 2008, 29, 189-191.	1.9	12

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109	Pharmacological and nutritional treatment for McArdle disease (Glycogen Storage Disease type V). , 2008, , CD003458.		12
110	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.	1.8	12
111	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.	2.5	12
112	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.	2.5	12
113	Monoallelic KIF1A-related disorders: a multicenter cross sectional study and systematic literature review. Journal of Neurology, 2022, 269, 437-450.	3.6	12
114	Disseminating the WHO International Classification of Functioning Health and Disability (ICF) in the Veneto region of Italy. Disability and Rehabilitation, 2008, 30, 71-80.	1.8	11
115	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.	1.8	11
116	Pharmacological and nutritional treatment for McArdle disease (Glycogen Storage Disease type V). , 2010, , CD003458.		11
117	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.	1.8	11
118	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.	2.8	11
119	Paralysis of Innervated Cultured Human Muscle Fibers Affects Enzymes Differentially. Journal of Neurochemistry, 1990, 54, 223-229.	3.9	10
120	Eight years of ICF in Italy: Principles, results and future perspectives. Disability and Rehabilitation, 2009, 31, S4-S7.	1.8	10
121	A Population Survey in Italy Based on the ICF Classification: Recognizing Persons with Severe Disability. Scientific World Journal, The, 2012, 2012, 1-9.	2.1	10
122	Clinical and Pulmonary Function Markers of Respiratory Exacerbation Risk in Subjects With Quadriplegic Cerebral Palsy. Respiratory Care, 2015, 60, 1431-1437.	1.6	10
123	Determinants of mobility in populations of older adults: Results from a cross-sectional study in Finland, Poland and Spain. Maturitas, 2018, 115, 84-91.	2.4	10
124	Safety and Efficacy Of Interferon Î³ in Friedreich's Ataxia. Movement Disorders, 2020, 35, 370-371.	3.9	10
125	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.	4.1	9
126	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.	2.8	8

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127	Validation of the COURAGE Built Environment Selfâ€Reported Questionnaire. Clinical Psychology and Psychotherapy, 2014, 21, 215-226.	2.7	8
128	Pediatric neurorehabilitation and the ICF. NeuroRehabilitation, 2015, 36, 31-36.	1.3	8
129	Optical Coherence Tomography in a Cohort of Genetically Defined Hereditary Spastic Paraplegia: A Brief Research Report. Frontiers in Neurology, 2019, 10, 1193.	2.4	8
130	Psychosocial impact of sport activity in neuromuscular disorders. Neurological Sciences, 2020, 41, 2561-2567.	1.9	8
131	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.	3.0	8
132	Guillainâ€“BarrÃ© syndrome with associated thrombocytopenia: prompt response to combined corticosteroid and immunoglobulin treatment. Neuromuscular Disorders, 1998, 8, 50-52.	0.6	7
133	Projecting and programming rehabilitation based on ICF-CY format in a neuropediatric hospital unit. Disability and Rehabilitation, 2009, 31, S55-S60.	1.8	7
134	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.	1.3	7
135	Sensitivity of Neuroimaging Indicators in Monitoring the Effects of Interferon Gamma Treatment in Friedreichâ€™s Ataxia. Frontiers in Neuroscience, 2020, 14, 872.	2.8	7
136	Accumulation of CK-MM is impaired in innervated and contracting cultured muscle fibers of duchenne muscular dystrophy patients. Life Sciences, 1987, 41, 927-933.	4.3	6
137	Multimodal MRI Longitudinal Assessment of White and Gray Matter in Different SPG Types of Hereditary Spastic Paraparesis. Frontiers in Neuroscience, 2020, 14, 325.	2.8	6
138	Hippocampal remodelling after MDMA neurotoxicity: A single case study. World Journal of Biological Psychiatry, 2009, 10, 961-968.	2.6	5
139	Becoming a young adult with cerebral palsy. Research in Developmental Disabilities, 2019, 92, 103450.	2.2	5
140	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.	0.2	5
141	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.	1.5	5
142	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.	1.6	4
143	Endoscopic Anatomic Features of the Triangular Recess. Neurosurgery, 2003, 52, 1491-1494.	1.1	3
144	The dystonic child treated with deep brain stimulation: ICF reading of a high-tech approach. Disability and Rehabilitation, 2009, 31, S159-S169.	1.8	3

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145	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.	1.0	3
146	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.	2.7	3
147	Editorial: Hereditary Spastic Paraplegias: At the Crossroads of Molecular Pathways and Clinical Options. Frontiers in Neuroscience, 2021, 15, 708642.	2.8	3
148	Toward a Harmonized WHO Family of International Classifications Content Model. Studies in Health Technology and Informatics, 2020, 270, 1409-1410.	0.3	3
149	Preliminary results of ICF dissemination in primary health care in Mozambique: Sharing the Italian experience. Disability and Rehabilitation, 2009, 31, S78-S82.	1.8	2
150	The blurred scenario of motor neuron disorders linked toSpatacsinmutations: a case report. European Journal of Neurology, 2014, 21, e85-e86.	3.3	2
151	Mapping SAGE questionnaire to the International Classification of Functioning, Disability and Health (ICF). Clinical Psychology and Psychotherapy, 2014, 21, 199-203.	2.7	2
152	Brain Magnetic Spectroscopy Imaging and Hereditary Spastic Paraplegia: A Focused Systematic Review on Current Landmarks and Future Perspectives. Frontiers in Neurology, 2020, 11, 515.	2.4	2
153	Functional MRI Studies in Friedreich's Ataxia: A Systematic Review. Frontiers in Neurology, 2021, 12, 802496.	2.4	2
154	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.	0.9	1
155	Interaction Between Mitochondrial DNA Variants and Mitochondria/Endoplasmic Reticulum Contact Sites: A Perspective Review. DNA and Cell Biology, 2020, 39, 1431-1443.	1.9	1
156	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.	0.9	1
157	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.	2.2	1
158	FORDYSVAR EBOOK: Best practices and technological resources for students with Specific Learning Difficulties (SpLDs). , 0, , .		1
159	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.	0.5	0
160	FORDYSVAR: Book on specific learning difficulties in reading. , 0, , .		0