Enrico Bertini

List of Publications by Year
in descending order

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727 papers	42,990 citations	2565 99 h-index	6024 165 g-index
759 all docs	759 docs citations	759 times ranked	38800 citing authors

#	Article	IF	Citations
1	Analysis of glutathione: implication in redox and detoxification. Clinica Chimica Acta, 2003, 333, 19-39.	0.5	931
2	Consensus Statement for Standard of Care in Spinal Muscular Atrophy. Journal of Child Neurology, 2007, 22, 1027-1049.	0.7	754
3	Mutations in ADAR1 cause Aicardi-GoutiÃ¨res syndrome associated with a type I interferon signature. Nature Genetics, 2012, 44, 1243-1248.	9.4	712
4	An autosomal dominant disorder with multiple deletions of mitochondrial DNA starting at the D-loop region. Nature, 1989, 339, 309-311.	13.7	640
5	Mutations involved in Aicardi-GoutiÃ¨res syndrome implicate SAMHD1 as regulator of the innate immune response. Nature Genetics, 2009, 41, 829-832.	9.4	610
6	Mutations in genes encoding ribonuclease H2 subunits cause Aicardi-GoutiÃ¨res syndrome and mimic congenital viral brain infection. Nature Genetics, 2006, 38, 910-916.	9.4	592
7	Diagnosis and management of spinal muscular atrophy: Part 1: Recommendations for diagnosis, rehabilitation, orthopedic and nutritional care. Neuromuscular Disorders, 2018, 28, 103-115.	0.3	584
8	Mutations of SURF-1 in Leigh Disease Associated with Cytochrome c Oxidase Deficiency. American Journal of Human Genetics, 1998, 63, 1609-1621.	2.6	504
9	PLA2G6, encoding a phospholipase A2, is mutated in neurodegenerative disorders with high brain iron. Nature Genetics, 2006, 38, 752-754.	9.4	497
10	Empirical Studies in Information Visualization: Seven Scenarios. IEEE Transactions on Visualization and Computer Graphics, 2012, 18, 1520-1536.	2.9	429
11	Diagnosis and management of spinal muscular atrophy: Part 2: Pulmonary and acute care; medications, supplements and immunizations; other organ systems; and ethics. Neuromuscular Disorders, 2018, 28, 197-207.	0.3	421
12	Nusinersen initiated in infants during the presymptomatic stage of spinal muscular atrophy: Interim efficacy and safety results from the Phase 2 NURTURE study. Neuromuscular Disorders, 2019, 29, 842-856.	0.3	401
13	Mutations in INPP5E, encoding inositol polyphosphate-5-phosphatase E, link phosphatidyl inositol signaling to the ciliopathies. Nature Genetics, 2009, 41, 1032-1036.	9.4	383
14	Clinical and Molecular Phenotype of Aicardi-GoutiÃ¨res Syndrome. American Journal of Human Genetics, 2007, 81, 713-725.	2.6	375
15	Mutations in CEP290, which encodes a centrosomal protein, cause pleiotropic forms of Joubert syndrome. Nature Genetics, 2006, 38, 623-625.	9.4	368
16	Ataluren in patients with nonsense mutation Duchenne muscular dystrophy (ACT DMD): a multicentre, randomised, double-blind, placebo-controlled, phase 3 trial. Lancet, The, 2017, 390, 1489-1498.	6.3	365
17	Spinal muscular atrophy. Orphanet Journal of Rare Diseases, 2011, 6, 71.	1.2	363
18	Ataluren treatment of patients with nonsense mutation dystrophinopathy. Muscle and Nerve, 2014, 50, 477-487.	1.0	357

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19	Mutations in the gene encoding immunoglobulin Î¼-binding protein 2 cause spinal muscular atrophy with respiratory distress type 1. Nature Genetics, 2001, 29, 75-77.	9.4	317
20	Mutations in SPG11, encoding spatacsin, are a major cause of spastic paraplegia with thin corpus callosum. Nature Genetics, 2007, 39, 366-372.	9.4	303
21	COQ2 Nephropathy. Journal of the American Society of Nephrology: JASN, 2007, 18, 2773-2780.	3.0	297
22	Childhood spinal muscular atrophy: controversies and challenges. Lancet Neurology, The, 2012, 11, 443-452.	4.9	297
23	Ullrich scleroatonic muscular dystrophy is caused by recessive mutations in collagen type VI. Proceedings of the National Academy of Sciences of the United States of America, 2001, 98, 7516-7521.	3.3	288
24	Diagnostic approach to the congenital muscular dystrophies. Neuromuscular Disorders, 2014, 24, 289-311.	0.3	275
25	THE 6â€minute walk test and other endpoints in Duchenne muscular dystrophy: Longitudinal natural history observations over 48 weeks from a multicenter study. Muscle and Nerve, 2013, 48, 343-356.	1.0	258
26	De novo LMNA mutations cause a new form of congenital muscular dystrophy. Annals of Neurology, 2008, 64, 177-186.	2.8	255
27	Spinal Muscular Atrophy Associated with Progressive Myoclonic Epilepsy Is Caused by Mutations in ASAH1. American Journal of Human Genetics, 2012, 91, 5-14.	2.6	250
28	miRNAs as serum biomarkers for Duchenne muscular dystrophy. EMBO Molecular Medicine, 2011, 3, 258-265.	3.3	242
29	Spectrum of SCN1A mutations in severe myoclonic epilepsy of infancy. Neurology, 2003, 60, 1961-1967.	1.5	241
30	The 6â€minute walk test and other clinical endpoints in duchenne muscular dystrophy: Reliability, concurrent validity, and minimal clinically important differences from a multicenter study. Muscle and Nerve, 2013, 48, 357-368.	1.0	240
31	The Childrenâ€™s Hospital of Philadelphia Infant Test of Neuromuscular Disorders (CHOP INTEND): Test development and reliability. Neuromuscular Disorders, 2010, 20, 155-161.	0.3	239
32	Approach to the diagnosis of congenital myopathies. Neuromuscular Disorders, 2014, 24, 97-116.	0.3	239
33	Neurodegeneration associated with genetic defects in phospholipase A ₂. Neurology, 2008, 71, 1402-1409.	1.5	236
34	Phenylbutyrate increases SMN expression in vitro: relevance for treatment of spinal muscular atrophy. European Journal of Human Genetics, 2004, 12, 59-65.	1.4	235
35	Recessive mutations in EPG5 cause Vici syndrome, a multisystem disorder with defective autophagy. Nature Genetics, 2013, 45, 83-87.	9.4	231
36	RYR1 mutations are a common cause of congenital myopathies with central nuclei. Annals of Neurology, 2010, 68, 717-726.	2.8	230

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37	Gene expression profiling in the early phases of DMD: a constant molecular signature characterizes DMD muscle from early postnatal life throughout disease progression. FASEB Journal, 2007, 21, 1210-1226.	0.2	209
38	Infantile-Onset Ascending Hereditary Spastic Paralysis Is Associated with Mutations in the Alsin Gene. American Journal of Human Genetics, 2002, 71, 518-527.	2.6	203
39	Consensus Statement on Standard of Care for Congenital Muscular Dystrophies. Journal of Child Neurology, 2010, 25, 1559-1581.	0.7	200
40	Mutations in GDP-Mannose Pyrophosphorylase B Cause Congenital and Limb-Girdle Muscular Dystrophies Associated with Hypoglycosylation of Î±-Dystroglycan. American Journal of Human Genetics, 2013, 93, 29-41.	2.6	197
41	Cerebellar ataxia and coenzyme Q10 deficiency. Neurology, 2003, 60, 1206-1208.	1.5	195
42	Quality Metrics in High-Dimensional Data Visualization: An Overview and Systematization. IEEE Transactions on Visualization and Computer Graphics, 2011, 17, 2203-2212.	2.9	189
43	Leukoencephalopathy with thalamus and brainstem involvement and high lactate â€˜LTBLâ€™ caused by EARS2 mutations. Brain, 2012, 135, 1387-1394.	3.7	187
44	Mutations in KLHL40 Are a Frequent Cause of Severe Autosomal-Recessive Nemaline Myopathy. American Journal of Human Genetics, 2013, 93, 6-18.	2.6	186
45	SUCLA2 mutations are associated with mild methylmalonic aciduria, Leigh-like encephalomyopathy, dystonia and deafness. Brain, 2007, 130, 862-874.	3.7	180
46	Impaired Skin Fibroblast Carnitine Uptake in Primary Systemic Carnitine Deficiency Manifested by Childhood Carnitine-Responsive Cardiomyopathy. Pediatric Research, 1990, 28, 247-255.	1.1	179
47	Reliability of the North Star Ambulatory Assessment in a multicentric setting. Neuromuscular Disorders, 2009, 19, 458-461.	0.3	171
48	North Star Ambulatory Assessment, 6-minute walk test and timed items in ambulant boys with Duchenne muscular dystrophy. Neuromuscular Disorders, 2010, 20, 712-716.	0.3	171
49	Phosphomannose Isomerase Deficiency: A Carbohydrate-Deficient Glycoprotein Syndrome with Hepatic-Intestinal Presentation. American Journal of Human Genetics, 1998, 62, 1535-1539.	2.6	167
50	Congenital muscular dystrophies with defective glycosylation of dystroglycan. Neurology, 2009, 72, 1802-1809.	1.5	166
51	EPI-743 reverses the progression of the pediatric mitochondrial diseaseâ€”Genetically defined Leigh Syndrome. Molecular Genetics and Metabolism, 2012, 107, 383-388.	0.5	163
52	Molecular mechanisms and phenotypic variation in RYR1-related congenital myopathies. Brain, 2007, 130, 2024-2036.	3.7	161
53	Randomized, double-blind, placebo-controlled trial of phenylbutyrate in spinal muscular atrophy. Neurology, 2007, 68, 51-55.	1.5	159
54	Homozygosity mapping of Hallervordenâ€“Spatz syndrome to chromosome 20p12.3â€“p13. Nature Genetics, 1996, 14, 479-481.	9.4	158

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55	Phenotypic heterogeneity of the 8344A>G mtDNA â€œMERRFâ€•mutation. Neurology, 2013, 80, 2049-2054.	1.5	157
56	Glutathione in blood of patients with Friedreich's ataxia. European Journal of Clinical Investigation, 2001, 31, 1007-1011.	1.7	154
57	Genotype-phenotype correlation of paroxysmal nonkinesigenic dyskinesia. Neurology, 2007, 68, 1782-1789.	1.5	154
58	Leiomodin-3 dysfunction results in thin filament disorganization and nemaline myopathy. Journal of Clinical Investigation, 2014, 124, 4693-4708.	3.9	153
59	The effect of genotype on the natural history of eIF2B-related leukodystrophies. Neurology, 2004, 62, 1509-1517.	1.5	152
60	Functional changes in Duchenne muscular dystrophy. Neurology, 2011, 77, 250-256.	1.5	151
61	Phenylbutyrate increases SMN gene expression in spinal muscular atrophy patients. European Journal of Human Genetics, 2005, 13, 256-259.	1.4	148
62	Consensus Statement on Standard of Care for Congenital Myopathies. Journal of Child Neurology, 2012, 27, 363-382.	0.7	147
63	Infantile Alexander Disease: Spectrum of GFAP Mutations and Genotype-Phenotype Correlation. American Journal of Human Genetics, 2001, 69, 1134-1140.	2.6	146
64	Histological effects of givinostat in boys with Duchenne muscular dystrophy. Neuromuscular Disorders, 2016, 26, 643-649.	0.3	144
65	Actin Glutathionylation Increases in Fibroblasts of Patients with Friedreich's Ataxia. Journal of Biological Chemistry, 2003, 278, 42588-42595.	1.6	142
66	Patterns of disease progression in type 2 and 3 SMA: Implications for clinical trials. Neuromuscular Disorders, 2016, 26, 126-131.	0.3	142
67	Infantile spinal muscular atrophy with respiratory distress type 1 (SMARD1). Annals of Neurology, 2003, 54, 719-724.	2.8	141
68	Hemimegalencephaly and Intractable Epilepsy: Benefits of Hemispherectomy. Epilepsia, 1989, 30, 833-843.	2.6	139
69	CEP290 Mutations Are Frequently Identified in the Oculo-Renal Form of Joubert Syndromeâ€“Related Disorders. American Journal of Human Genetics, 2007, 81, 104-113.	2.6	137
70	Clinical and molecular genetic findings in COLQ-mutant congenital myasthenic syndromes. Brain, 2008, 131, 747-759.	3.7	134
71	Hypomyelinating leukodystrophies: Translational research progress and prospects. Annals of Neurology, 2014, 76, 5-19.	2.8	132
72	MEDNIK syndrome: a novel defect of copper metabolism treatable by zinc acetate therapy. Brain, 2013, 136, 872-881.	3.7	130

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73	INFUSE: Interactive Feature Selection for Predictive Modeling of High Dimensional Data. IEEE Transactions on Visualization and Computer Graphics, 2014, 20, 1614-1623.	2.9	129
74	Pilot trial of phenylbutyrate in spinal muscular atrophy. Neuromuscular Disorders, 2004, 14, 130-135.	0.3	128
75	CloudLines: Compact Display of Event Episodes in Multiple Time-Series. IEEE Transactions on Visualization and Computer Graphics, 2011, 17, 2432-2439.	2.9	127
76	AHI1gene mutations cause specific forms of Joubert syndrome-related disorders. Annals of Neurology, 2006, 59, 527-534.	2.8	125
77	GPR56-related bilateral frontoparietal polymicrogyria: further evidence for an overlap with the cobblestone complex. Brain, 2010, 133, 3194-3209.	3.7	125
78	Iron-related MRI images in patients with pantothenate kinase-associated neurodegeneration (PKAN) treated with deferiprone: Results of a phase II pilot trial. Movement Disorders, 2011, 26, 1755-1759.	2.2	125
79	Fatal infantile liver failure associated with mitochondrial DNA depletion. Journal of Pediatrics, 1992, 121, 896-901.	0.9	123
80	Mutations in GTPBP3 Cause a Mitochondrial Translation Defect Associated with Hypertrophic Cardiomyopathy, Lactic Acidosis, and Encephalopathy. American Journal of Human Genetics, 2014, 95, 708-720.	2.6	123
81	DPM2â€CDG: A muscular dystrophyâ€“dystroglycanopathy syndrome with severe epilepsy. Annals of Neurology, 2012, 72, 550-558.	2.8	121
82	Attention Deficit Hyperactivity Disorder and Cognitive Function in Duchenne Muscular Dystrophy: Phenotype-Genotype Correlation. Journal of Pediatrics, 2012, 161, 705-709.e1.	0.9	121
83	Phenotypic clustering of lamin A/C mutations in neuromuscular patients. Neurology, 2007, 69, 1285-1292.	1.5	120
84	Dominantly inherited mitochondrial myopathy with multiple deletions of mitochondrial DNA. Neurology, 1991, 41, 1053-1053.	1.5	120
85	The m.3243A>G mitochondrial DNA mutation and related phenotypes. A matter of gender?. Journal of Neurology, 2014, 261, 504-510.	1.8	119
86	Mechanisms inducing low bone density in duchenne muscular dystrophy in mice and humans. Journal of Bone and Mineral Research, 2011, 26, 1891-1903.	3.1	116
87	Collagen VI deficiency affects the organization of fibronectin in the extracellular matrix of cultured fibroblasts. Matrix Biology, 2001, 20, 475-486.	1.5	115
88	GM1 gangliosidosis and Morquio B disease: An update on genetic alterations and clinical findings. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2011, 1812, 782-790.	1.8	115
89	Mutation spectrum in the large GTPase dynamin 2, and genotype-phenotype correlation in autosomal dominant centronuclear myopathy. Human Mutation, 2012, 33, 949-959.	1.1	115
90	Mutations in the MTM1 gene implicated in X-linked myotubular myopathy. ENMC International Consortium on Myotubular Myopathy. European Neuro- Muscular Center. Human Molecular Genetics, 1997, 6, 1505-1511.	1.4	113

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91	Mutation of plasma membrane Ca ²⁺ ATPase isoform 3 in a family with X-linked congenital cerebellar ataxia impairs Ca ²⁺ homeostasis. Proceedings of the National Academy of Sciences of the United States of America, 2012, 109, 14514-14519.	3.3	113
92	Multi-minicore disease-searching for boundaries: Phenotype analysis of 38 cases. Annals of Neurology, 2000, 48, 745-757.	2.8	112
93	Periventricular heterotopia, mental retardation, and epilepsy associated with 5q14.3-q15 deletion. Neurology, 2009, 72, 784-792.	1.5	110
94	Clinical features, risk factors, and prognosis in transient global amnesia: a follow-up study. European Journal of Neurology, 2005, 12, 350-356.	1.7	109
95	Renal involvement in mitochondrial cytopathies. Pediatric Nephrology, 2012, 27, 539-550.	0.9	109
96	The expanding phenotype ofPOMT1mutations: from Walker-Warburg syndrome to congenital muscular dystrophy, microcephaly, and mental retardation. Human Mutation, 2006, 27, 453-459.	1.1	106
97	Metabolic consequences of mitochondrial coenzyme A deficiency in patients with PANK2 mutations. Molecular Genetics and Metabolism, 2012, 105, 463-471.	0.5	106
98	Affinity proteomics within rare diseases: a <scp>BIO</scp>â€<scp>NMD</scp> study for blood biomarkers of muscular dystrophies. EMBO Molecular Medicine, 2014, 6, 918-936.	3.3	105
99	Distinguishing the four genetic causes of jouberts syndrome-related disorders. Annals of Neurology, 2005, 57, 513-519.	2.8	104
100	Characterization of recessive RYR1 mutations in core myopathies. Human Molecular Genetics, 2006, 15, 2791-2803.	1.4	103
101	Somatic and germline mosaicisms in Severe Myoclonic Epilepsy of Infancy. Biochemical and Biophysical Research Communications, 2006, 341, 489-493.	1.0	102
102	Autophagy regulates satellite cell ability to regenerate normal and dystrophic muscles. Cell Death and Differentiation, 2016, 23, 1839-1849.	5.0	102
103	A randomized placebo-controlled phase 3 trial of an antisense oligonucleotide, drisapersen, in Duchenne muscular dystrophy. Neuromuscular Disorders, 2018, 28, 4-15.	0.3	102
104	24 Month Longitudinal Data in Ambulant Boys with Duchenne Muscular Dystrophy. PLoS ONE, 2013, 8, e52512.	1.1	99
105	Long Term Natural History Data in Ambulant Boys with Duchenne Muscular Dystrophy: 36-Month Changes. PLoS ONE, 2014, 9, e108205.	1.1	98
106	The inv dup(15) syndrome. Neurology, 1997, 48, 1081-1086.	1.5	97
107	Daily salbutamol in young patients with SMA type II. Neuromuscular Disorders, 2008, 18, 536-540.	0.3	97
108	Agenesis of the corpus callosum, combined immunodeficiency, bilateral cataract, and hypopigmentation in two brothers. American Journal of Medical Genetics Part A, 1988, 29, 1-8.	2.4	96

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109	MKS3/TMEM67mutations are a major cause of COACH Syndrome, a Joubert Syndrome related disorder with liver involvement. Human Mutation, 2009, 30, E432-E442.	1.1	96
110	Safety and efficacy of olesoxime in patients with type 2 or non-ambulatory type 3 spinal muscular atrophy: a randomised, double-blind, placebo-controlled phase 2 trial. Lancet Neurology, The, 2017, 16, 513-522.	4.9	95
111	Newborn screening programs for spinal muscular atrophy worldwide: Where we stand and where to go. Neuromuscular Disorders, 2021, 31, 574-582.	0.3	94
112	NPHP1 gene deletion is a rare cause of Joubert syndrome related disorders. Journal of Medical Genetics, 2005, 42, e9-e9.	1.5	93
113	Oligophrenin 1 mutations frequently cause X-linked mental retardation with cerebellar hypoplasia. Neurology, 2005, 65, 1364-1369.	1.5	93
114	Feeding problems and malnutrition in spinal muscular atrophy type II. Neuromuscular Disorders, 2008, 18, 389-393.	0.3	92
115	Mutation Update and Genotype-Phenotype Correlations of Novel and Previously Described Mutations inTPM2andTPM3Causing Congenital Myopathies. Human Mutation, 2014, 35, 779-790.	1.1	92
116	The genetic basis of undiagnosed muscular dystrophies and myopathies. Neurology, 2016, 87, 71-76.	1.5	92
117	Description, Nomenclature, and Mapping of a Novel Cerebello-Renal Syndrome with the Molar Tooth Malformation. American Journal of Human Genetics, 2003, 73, 663-670.	2.6	91
118	Congenital Muscular Dystrophies: A Brief Review. Seminars in Pediatric Neurology, 2011, 18, 277-288.	1.0	91
119	Inheritance patterns and phenotypic features of myofibrillar myopathy associated with a BAG3 mutation. Neuromuscular Disorders, 2010, 20, 438-442.	0.3	90
120	Disease-Causing SDHAF1 Mutations Impair Transfer of Fe-S Clusters to SDHB. Cell Metabolism, 2016, 23, 292-302.	7.2	89
121	Safety and efficacy of once-daily risdiplam in type 2 and non-ambulant type 3 spinal muscular atrophy (SUNFISH part 2): a phase 3, double-blind, randomised, placebo-controlled trial. Lancet Neurology, The, 2022, 21, 42-52.	4.9	89
122	Decreased guanine nucleotide exchange factor activity in eIF2B-mutated patients. European Journal of Human Genetics, 2004, 12, 561-566.	1.4	87
123	Cerebroretinal microangiopathy with calcifications and cysts (CRMCC). American Journal of Medical Genetics, Part A, 2008, 146A, 182-190.	0.7	87
124	Role of Gabapentin in Spinal Muscular Atrophy. Journal of Child Neurology, 2003, 18, 537-541.	0.7	86
125	COQ4 Mutations Cause a Broad Spectrum of Mitochondrial Disorders Associated with CoQ10 Deficiency. American Journal of Human Genetics, 2015, 96, 309-317.	2.6	86
126	Natural history of pulmonary function in collagen VI-related myopathies. Brain, 2013, 136, 3625-3633.	3.7	85

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127	POPDC1S201F causes muscular dystrophy and arrhythmia by affecting protein trafficking. Journal of Clinical Investigation, 2015, 126, 239-253.	3.9	85
128	Evaluation of alternative glyph designs for time series data in a small multiple setting. , 2013, , .		84
129	Fatal hypertrophic cardiomyopathy and nemaline myopathy associated with ACTA1 K336E mutation. Neuromuscular Disorders, 2006, 16, 548-552.	0.3	83
130	Spectrum of pontocerebellar hypoplasia in 13 girls and boys with CASK mutations: confirmation of a recognizable phenotype and first description of a male mosaic patient. Orphanet Journal of Rare Diseases, 2012, 7, 18.	1.2	83
131	Reliability of the Performance of Upper Limb assessment in Duchenne muscular dystrophy. Neuromuscular Disorders, 2014, 24, 201-206.	0.3	83
132	Nusinersen in type 1 spinal muscular atrophy: Twelveâ€month realâ€world data. Annals of Neurology, 2019, 86, 443-451.	2.8	83
133	Clinical and genetic studies in hereditary spastic paraplegia with thin corpus callosum. Neurology, 2004, 62, 262-268.	1.5	82
134	Motor function-muscle strength relationship in spinal muscular atrophy. Muscle and Nerve, 2004, 29, 548-552.	1.0	81
135	Importance of SPP1 genotype as a covariate in clinical trials in Duchenne muscular dystrophy. Neurology, 2012, 79, 159-162.	1.5	81
136	Congenital myopathies: clinical phenotypes and new diagnostic tools. Italian Journal of Pediatrics, 2017, 43, 101.	1.0	80
137	The T9176G mtDNA mutation severely affects ATP production and results in Leigh syndrome. Neurology, 2001, 56, 687-690.	1.5	79
138	Succinateâ€CoA ligase deficiency due to mutations in SUCLA2 and SUCLG1: phenotype and genotype correlations in 71 patients. Journal of Inherited Metabolic Disease, 2016, 39, 243-252.	1.7	79
139	Cardiomyopathy may be the only clinical manifestation in female carriers of Duchenne muscular dystrophy. Neurology, 1993, 43, 2342-2342.	1.5	79
140	Collagen type VI and related disorders: Bethlem myopathy and Ullrich scleroatonic muscular dystrophy. European Journal of Paediatric Neurology, 2002, 6, 193-198.	0.7	77
141	NovelTMEM67mutations and genotype-phenotype correlates in meckelin-related ciliopathies. Human Mutation, 2010, 31, n/a-n/a.	1.1	77
142	Effectiveness and safety of the tri-iodothyronine analogue Triac in children and adults with MCT8 deficiency: an international, single-arm, open-label, phase 2 trial. Lancet Diabetes and Endocrinology,the, 2019, 7, 695-706.	5.5	77
143	Subcomplexes of human ATP synthase mark mitochondrial biosynthesis disorders. Annals of Neurology, 2006, 59, 265-275.	2.8	75
144	Friedreich's ataxia: Oxidative stress and cytoskeletal abnormalities. Journal of the Neurological Sciences, 2009, 287, 111-118.	0.3	75

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145	Joubert syndrome and related disorders. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2013, 113, 1879-1888.	1.0	75
146	Frataxin Deficiency Leads to Reduced Expression and Impaired Translocation of NF-E2-Related Factor (Nrf2) in Cultured Motor Neurons. International Journal of Molecular Sciences, 2013, 14, 7853-7865.	1.8	75
147	Prevalence of congenital muscular dystrophy in Italy. Neurology, 2015, 84, 904-911.	1.5	75
148	Progressive neuropathy and recurrent myoglobinuria in a child with long-chain 3-hydroxyacyl-coenzyme A dehydrogenase deficiency. Journal of Pediatrics, 1991, 118, 744-746.	0.9	74
149	Spectrum of phenotypes in female patients with epilepsy due to protocadherin 19 mutations. Epilepsia, 2011, 52, 1251-1257.	2.6	74
150	A new syndrome with ethylmalonic aciduria and normal fatty acid oxidation in fibroblasts. Journal of Pediatrics, 1994, 124, 79-86.	0.9	73
151	GJA12 mutations are a rare cause of Pelizaeus-Merzbacher-like disease. Neurology, 2008, 70, 748-754.	1.5	73
152	Dominant and recessive COL6A1 mutations in Ullrich scleroatonic muscular dystrophy. Annals of Neurology, 2005, 58, 400-410.	2.8	72
153	Clinical and genetic characterization of Chanarinâ€“Dorfman syndrome. Biochemical and Biophysical Research Communications, 2008, 369, 1125-1128.	1.0	72
154	Unstable mutants in the peripheral endosomal membrane component ALS2 cause early-onset motor neuron disease. Proceedings of the National Academy of Sciences of the United States of America, 2003, 100, 16041-16046.	3.3	71
155	Assessing upper limb function in nonambulant SMA patients: Development of a new module. Neuromuscular Disorders, 2011, 21, 406-412.	0.3	71
156	Oxidative stress in Duchenne muscular dystrophy: focus on the NRF2 redox pathway. Human Molecular Genetics, 2017, 26, 2781-2790.	1.4	71
157	Pontocerebellar hypoplasia type 6 caused by mutations in RARS2: definition of the clinical spectrum and molecular findings in five patients. Journal of Inherited Metabolic Disease, 2013, 36, 43-53.	1.7	70
158	MYH7-related myopathies: clinical, histopathological and imaging findings in a cohort of Italian patients. Orphanet Journal of Rare Diseases, 2016, 11, 91.	1.2	70
159	New clinical phenotype of branched-chain acyl-CoA oxidation defect. Lancet, The, 1991, 338, 1522-1523.	6.3	69
160	Mutation analysis in 16 patients with mtDNA depletion. Human Mutation, 2003, 21, 453-454.	1.1	69
161	Genetic heterogeneity of megalencephalic leukoencephalopathy and subcortical cysts. Neurology, 2003, 61, 534-537.	1.5	68
162	Glutathione metabolism and antioxidant enzymes in patients affected by nonalcoholic steatohepatitis. Clinica Chimica Acta, 2005, 355, 105-111.	0.5	68

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163	Redefining phenotypes associated with mitochondrial DNA single deletion. Journal of Neurology, 2015, 262, 1301-1309.	1.8	68
164	A severe variant of childhood ataxia with central hypomyelination/vanishing white matter leukoencephalopathy related to EIF21B5 mutation. Neurology, 2002, 59, 1966-1968.	1.5	67
165	Dominant LMNA mutations can cause combined muscular dystrophy and peripheral neuropathy. Journal of Neurology, Neurosurgery and Psychiatry, 2005, 76, 1019-1021.	0.9	67
166	Nusinersen in type 1 SMA infants, children and young adults: Preliminary results on motor function. Neuromuscular Disorders, 2018, 28, 582-585.	0.3	67
167	Fulminant Leigh syndrome and sudden unexpected death in a family with the T9176C mutation of the mitochondrial ATPase 6 gene. Journal of Inherited Metabolic Disease, 1998, 21, 2-8.	1.7	66
168	Novel SACS mutations in autosomal recessive spastic ataxia of Charlevoix-Saguenay type. Neurology, 2004, 62, 103-106.	1.5	66
169	Biallelic Mutations in TBCD , Encoding the Tubulin Folding Cofactor D, Perturb Microtubule Dynamics and Cause Early-Onset Encephalopathy. American Journal of Human Genetics, 2016, 99, 962-973.	2.6	66
170	Systemic Activation of Nrf2 Pathway in Parkinson's Disease. Movement Disorders, 2020, 35, 180-184.	2.2	66
171	Megalencephalic leukoencephalopathy with subcortical cysts protein 1 functionally cooperates with the TRPV4 cation channel to activate the response of astrocytes to osmotic stress: dysregulation by pathological mutations. Human Molecular Genetics, 2012, 21, 2166-2180.	1.4	65
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173	The hyperornithinemiaâ€“hyperammonemia-homocitrullinuria syndrome. Orphanet Journal of Rare Diseases, 2015, 10, 29.	1.2	65
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