

Maria Teresa Bassi

List of Publications by Year in descending order

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146
papers

12,533
citations

36203

51
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25716

108
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150
all docs

150
docs citations

150
times ranked

23617
citing authors

#	ARTICLE	IF	CITATIONS
1	Guidelines for the use and interpretation of assays for monitoring autophagy (3rd edition). <i>Autophagy</i> , 2016, 12, 1-222.	4.3	4,701
2	X-linked situs abnormalities result from mutations in ZIC3. <i>Nature Genetics</i> , 1997, 17, 305-308.	9.4	406
3	De novo mutations in ATP1A3 cause alternating hemiplegia of childhood. <i>Nature Genetics</i> , 2012, 44, 1030-1034.	9.4	345
4	Non-type I cystinuria caused by mutations in SLC7A9, encoding a subunit (bo,+AT) of rBAT. <i>Nature Genetics</i> , 1999, 23, 52-57.	9.4	280
5	SLC7A7, encoding a putative permease-related protein, is mutated in patients with lysinuric protein intolerance. <i>Nature Genetics</i> , 1999, 21, 297-301.	9.4	238
6	Cloning of the Gene Encoding a Novel Integral Membrane Protein, Muco1ipidinâ€”and Identification of the Two Major Founder Mutations Causing Muco1ipidosis Type IV. <i>American Journal of Human Genetics</i> , 2000, 67, 1110-1120.	2.6	230
7	Characterization and Mutation Analysis of Human LEFTY A and LEFTY B, Homologues of Murine Genes Implicated in Left-Right Axis Development. <i>American Journal of Human Genetics</i> , 1999, 64, 712-721.	2.6	216
8	Cloning of the gene for ocular albinism type 1 from the distal short arm of the X chromosome. <i>Nature Genetics</i> , 1995, 10, 13-19.	9.4	190
9	A complex syndrome of left-right axis, central nervous system and axial skeleton defects in <i>Zic3</i> mutant mice. <i>Development (Cambridge)</i> , 2002, 129, 2293-2302.	1.2	152
10	A novel mutation in KCNQ2 associated with BFNC, drug resistant epilepsy, and mental retardation. <i>Neurology</i> , 2004, 63, 57-65.	1.5	146
11	Identification and characterisation of human xCT that co-expresses, with 4F2 heavy chain, the amino acid transport activity system x c -. <i>Pflugers Archiv European Journal of Physiology</i> , 2001, 442, 286-296.	1.3	133
12	Agenesis of the Corpus Callosum: Clinical and Genetic Study in 63 Young Patients. <i>Pediatric Neurology</i> , 2006, 34, 186-193.	1.0	130
13	Mutations in the motor and stalk domains of KIF5A in spastic paraplegia type 10 and in axonal Charcotâ€”Marieâ€”Tooth type 2. <i>Clinical Genetics</i> , 2012, 82, 157-164.	1.0	128
14	Functional analysis of mutations in SLC7A9, and genotypeâ€”phenotype correlation in non-Type I cystinuria. <i>Human Molecular Genetics</i> , 2001, 10, 305-316.	1.4	125
15	cDNA cloning of human hnRNP protein A1 reveals the existence of multiple mRNA isoforms. <i>Nucleic Acids Research</i> , 1988, 16, 3751-3770.	6.5	120
16	Ocular albinism: evidence for a defect in an intracellular signal transduction system. <i>Nature Genetics</i> , 1999, 23, 108-112.	9.4	118
17	A gene from the Xp22.3 region shares homology with voltage-gated chloride channels. <i>Human Molecular Genetics</i> , 1994, 3, 547-552.	1.4	108
18	Nitric oxide inhibition of Drp1-mediated mitochondrial fission is critical for myogenic differentiation. <i>Cell Death and Differentiation</i> , 2010, 17, 1684-1696.	5.0	106

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19	Molecular cloning and characterization of NEU4, the fourth member of the human sialidase gene family. <i>Genomics</i> , 2004, 83, 445-453.	1.3	103
20	A high resolution deletion map of human chromosome Xp22. <i>Nature Genetics</i> , 1993, 4, 272-279.	9.4	101
21	A novel mutation in the ATP1A2 gene causes alternating hemiplegia of childhood. <i>Journal of Medical Genetics</i> , 2004, 41, 621-628.	1.5	100
22	X-Linked Late-Onset Sensorineural Deafness Caused by a Deletion Involving OA1 and a Novel Gene Containing WD-40 Repeats. <i>American Journal of Human Genetics</i> , 1999, 64, 1604-1616.	2.6	97
23	Identification and expression of NEU3, a novel human sialidase associated to the plasma membrane. <i>Biochemical Journal</i> , 2000, 349, 343.	1.7	95
24	Mutations in β - and β -tubulin encoding genes: Implications in brain malformations. <i>Brain and Development</i> , 2015, 37, 273-280.	0.6	94
25	Cornelia de Lange syndrome mutations in SMC1A or SMC3 affect binding to DNA. <i>Human Molecular Genetics</i> , 2009, 18, 418-427.	1.4	92
26	Nitric Oxide Sustains Long-Term Skeletal Muscle Regeneration by Regulating Fate of Satellite Cells Via Signaling Pathways Requiring Vangl2 and Cyclic GMP. <i>Stem Cells</i> , 2012, 30, 197-209.	1.4	91
27	The first ALS2 missense mutation associated with JPLS reveals new aspects of alsin biological function. <i>Brain</i> , 2006, 129, 1710-1719.	3.7	87
28	Hereditary spastic paraplegia type 5: natural history, biomarkers and a randomized controlled trial. <i>Brain</i> , 2017, 140, 3112-3127.	3.7	87
29	A clinical, genetic, and biochemical characterization of <i>SPG7</i> mutations in a large cohort of patients with hereditary spastic paraplegia. <i>Human Mutation</i> , 2008, 29, 522-531.	1.1	85
30	Pleiotropic effects of spastin on neurite growth depending on expression levels. <i>Journal of Neurochemistry</i> , 2009, 108, 1277-1288.	2.1	84
31	Cholestenic acids regulate motor neuron survival via liver X receptors. <i>Journal of Clinical Investigation</i> , 2014, 124, 4829-4842.	3.9	84
32	Isolation of an active gene encoding human hnRNP protein A1. <i>Journal of Molecular Biology</i> , 1989, 207, 491-503.	2.0	81
33	Analysis of the OA1 gene reveals mutations in only one-third of patients with X-linked ocular albinism. <i>Human Molecular Genetics</i> , 1995, 4, 2319-2325.	1.4	80
34	Mutated mitofusin 2 presents with intrafamilial variability and brain mitochondrial dysfunction. <i>Neurology</i> , 2008, 71, 1959-1966.	1.5	80
35	Nitric Oxide Generated by Tumor-Associated Macrophages Is Responsible for Cancer Resistance to Cisplatin and Correlated With Syntaxin 4 and Acid Sphingomyelinase Inhibition. <i>Frontiers in Immunology</i> , 2018, 9, 1186.	2.2	76
36	Defective autophagy in spastizin mutated patients with hereditary spastic paraparesis type 15. <i>Brain</i> , 2013, 136, 3119-3139.	3.7	74

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37	Overexpression of wild-type and mutant mucolipin proteins in mammalian cells: effects on the late endocytic compartment organization. <i>FEBS Letters</i> , 2004, 567, 219-224.	1.3	73
38	Identification and Characterization of AFG3L2, a Novel Paraplegin-Related Gene. <i>Genomics</i> , 1999, 59, 51-58.	1.3	70
39	FAHN/SPG35: a narrow phenotypic spectrum across disease classifications. <i>Brain</i> , 2019, 142, 1561-1572.	3.7	70
40	Structure of the SLC7A7 Gene and Mutational Analysis of Patients Affected by Lysinuric Protein Intolerance. <i>American Journal of Human Genetics</i> , 2000, 66, 92-99.	2.6	66
41	Syntaxin 4 Is Required for Acid Sphingomyelinase Activity and Apoptotic Function*. <i>Journal of Biological Chemistry</i> , 2010, 285, 40240-40251.	1.6	65
42	Defective intracellular transport and processing of OA1 is a major cause of ocular albinism type 1. <i>Human Molecular Genetics</i> , 2000, 9, 3011-3018.	1.4	63
43	Phenotypic Heterogeneity of the GRN Asp22fs Mutation in a Large Italian Kindred. <i>Journal of Alzheimer's Disease</i> , 2011, 24, 253-259.	1.2	62
44	Mutations in CYP2U1, DDHD2 and GBA2 genes are rare causes of complicated forms of hereditary spastic paraparesis. <i>Journal of Neurology</i> , 2014, 261, 373-381.	1.8	62
45	Identification and Characterization of YME1L1, a Novel Paraplegin-Related Gene. <i>Genomics</i> , 2000, 66, 48-54.	1.3	58
46	Deficient nitric oxide signalling impairs skeletal muscle growth and performance: involvement of mitochondrial dysregulation. <i>Skeletal Muscle</i> , 2014, 4, 22.	1.9	58
47	MID2, a Homologue of the Opitz Syndrome Gene MID1: Similarities in Subcellular Localization and Differences in Expression During Development. <i>Human Molecular Genetics</i> , 1999, 8, 1397-1407.	1.4	57
48	Relationship Between Migraine and Epilepsy in Pediatric Age . <i>CME. Headache</i> , 2006, 46, 413-421.	1.8	55
49	MID1 mutation screening in a large cohort of Opitz G/BBB syndrome patients: twenty-nine novel mutations identified. <i>Human Mutation</i> , 2007, 28, 206-207.	1.1	55
50	The Low-Affinity Receptor for Neurotrophins p75 ^{NTR} Plays a Key Role for Satellite Cell Function in Muscle Repair Acting via RhoA. <i>Molecular Biology of the Cell</i> , 2009, 20, 3620-3627.	0.9	55
51	Variants in KIF1A gene in dominant and sporadic forms of hereditary spastic paraparesis. <i>Journal of Neurology</i> , 2015, 262, 2684-2690.	1.8	55
52	Human hnRNP Protein A1 Gene Expression. <i>Journal of Molecular Biology</i> , 1993, 230, 77-89.	2.0	53
53	A novel <i>CLN8</i> mutation in late-infantile-onset neuronal ceroid lipofuscinosis (LINCL) reveals aspects of CLN8 neurobiological function. <i>Human Mutation</i> , 2009, 30, 1104-1116.	1.1	53
54	The genes for X-linked ocular albinism (OA1) and microphthalmia with linear skin defects (MLS): cloning and characterization of the critical regions. <i>Human Molecular Genetics</i> , 1993, 2, 947-952.	1.4	52

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55	A wide spectrum of clinical, neurophysiological and neuroradiological abnormalities in a family with a novel CACNA1A mutation. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2010, 81, 840-843.	0.9	51
56	A novel mutation in the β -tubulin gene <i>TUBB2B</i> associated with complex malformation of cortical development and deficits in axonal guidance. <i>Developmental Medicine and Child Neurology</i> , 2012, 54, 765-769.	1.1	50
57	Ears of the Lynx MRI Sign Is Associated with SPG11 and SPG15 Hereditary Spastic Paraplegia. <i>American Journal of Neuroradiology</i> , 2019, 40, 199-203.	1.2	50
58	Loss of paraplegin drives spasticity rather than ataxia in a cohort of 241 patients with <i>SPG7</i> . <i>Neurology</i> , 2019, 92, e2679-e2690.	1.5	49
59	The GST domain of GDAP1 is a frequent target of mutations in the dominant form of axonal Charcot Marie Tooth type 2K. <i>Journal of Medical Genetics</i> , 2010, 47, 712-716.	1.5	46
60	Cryptogenic Epileptic Syndromes Related to SCN1A. <i>Archives of Neurology</i> , 2008, 65, 489.	4.9	43
61	Clinical phenotype variability in patients with hereditary spastic paraplegia type 5 associated with <i>CYP7B1</i> mutations. <i>Clinical Genetics</i> , 2012, 81, 150-157.	1.0	42
62	Brain malformations and mutations in β - and β -tubulin genes: a review of the literature and description of two new cases. <i>Developmental Medicine and Child Neurology</i> , 2014, 56, 354-360.	1.1	42
63	Clinical and Paraclinical Indicators of Motor System Impairment in Hereditary Spastic Paraplegia: A Pilot Study. <i>PLoS ONE</i> , 2016, 11, e0153283.	1.1	41
64	ZFYVE26/SPASTIZIN and SPG11/SPATACSIN mutations in hereditary spastic paraplegia types AR-SPG15 and AR-SPG11 have different effects on autophagy and endocytosis. <i>Autophagy</i> , 2019, 15, 34-57.	4.3	41
65	Autophagy controls neonatal myogenesis by regulating the GH-IGF1 system through a NFE2L2- and DDIT3-mediated mechanism. <i>Autophagy</i> , 2019, 15, 58-77.	4.3	41
66	Early onset, non fluctuating spinocerebellar ataxia and a novel missense mutation in CACNA1A gene. <i>Journal of the Neurological Sciences</i> , 2006, 241, 13-17.	0.3	39
67	Eight Novel Mutations in SPG4 in a Large Sample of Patients With Hereditary Spastic Paraplegia. <i>Archives of Neurology</i> , 2006, 63, 750.	4.9	39
68	A de-novo STXBP1 gene mutation in a patient showing the Rett syndrome phenotype. <i>NeuroReport</i> , 2015, 26, 254-257.	0.6	39
69	The Fine Tuning of Drp1-Dependent Mitochondrial Remodeling and Autophagy Controls Neuronal Differentiation. <i>Frontiers in Cellular Neuroscience</i> , 2019, 13, 120.	1.8	39
70	The emerging role of Acid Sphingomyelinase in autophagy. <i>Apoptosis: an International Journal on Programmed Cell Death</i> , 2015, 20, 635-644.	2.2	36
71	Tubulin-related cerebellar dysplasia: definition of a distinct pattern of cerebellar malformation. <i>European Radiology</i> , 2017, 27, 5080-5092.	2.3	36
72	A novel de novo nonsense mutation in ATP1A2 associated with sporadic hemiplegic migraine and epileptic seizures. <i>Journal of the Neurological Sciences</i> , 2008, 273, 123-126.	0.3	35

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73	Cloning of a human homologue of the <i>Xenopus laevis</i> APX gene from the ocular albinism type 1 critical region. <i>Human Molecular Genetics</i> , 1995, 4, 373-382.	1.4	34
74	Clinical Pre-genetic Screening for Stroke Monogenic Diseases. <i>Stroke</i> , 2016, 47, 1702-1709.	1.0	34
75	A YAC-based binning strategy facilitating the rapid assembly of cosmid contigs: 1.6 Mb of overlapping cosmids in Xp22. <i>Human Molecular Genetics</i> , 1994, 3, 1155-1161.	1.4	33
76	Identification of a Novel Homolog of the <i>Drosophila</i> staufer Protein in the Chromosome 8q13-q21.1 Region. <i>Genomics</i> , 1999, 62, 113-118.	1.3	33
77	Endothelial nitric oxide synthase overexpression by neuronal cells in neurodegeneration: a link between inflammation and neuroprotection. <i>Journal of Neurochemistry</i> , 2008, 106, 193-204.	2.1	33
78	Hereditary Spastic Paraplegia: Beyond Clinical Phenotypes toward a Unified Pattern of Central Nervous System Damage. <i>Radiology</i> , 2015, 276, 207-218.	3.6	32
79	Diverse prevalence of large deletions within the OA1 gene in ocular albinism type 1 patients from Europe and North America. <i>Human Genetics</i> , 2001, 108, 51-54.	1.8	31
80	Identification of a New EGF-Repeat-Containing Gene from Human Xp22: A Candidate for Developmental Disorders. <i>Genomics</i> , 2000, 65, 16-23.	1.3	30
81	Infancy onset hereditary spastic paraplegia associated with a novel atlastin mutation. <i>Neurology</i> , 2003, 61, 580-581.	1.5	30
82	Point mutations and a large intragenic deletion in SPG11 in complicated spastic paraplegia without thin corpus callosum. <i>Journal of Medical Genetics</i> , 2009, 46, 345-351.	1.5	30
83	Novel splice-site mutations and a large intragenic deletion in <i>PLA2G6</i> associated with a severe and rapidly progressive form of infantile neuroaxonal dystrophy. <i>Clinical Genetics</i> , 2010, 78, 432-440.	1.0	30
84	Reversal of Defective Mitochondrial Biogenesis in Limb-Girdle Muscular Dystrophy 2D by Independent Modulation of Histone and PGC-1 β Acetylation. <i>Cell Reports</i> , 2016, 17, 3010-3023.	2.9	30
85	Senataxin modulates neurite growth through fibroblast growth factor 8 signalling. <i>Brain</i> , 2011, 134, 1808-1828.	3.7	28
86	A novel ATP1A2 gene mutation in familial hemiplegic migraine and epilepsy. <i>Cephalalgia</i> , 2014, 34, 68-72.	1.8	28
87	Cloning of the murine homolog of the ocular albinism type 1 (OA1) gene: sequence, genomic structure, and expression analysis in pigment cells. <i>Genome Research</i> , 1996, 6, 880-885.	2.4	27
88	Cellular expression and alternative splicing of SLC25A23, a member of the mitochondrial Ca ²⁺ -dependent solute carrier gene family. <i>Gene</i> , 2005, 345, 173-182.	1.0	27
89	Functional analysis of novel KCNQ2 and KCNQ3 gene variants found in a large pedigree with benign familial neonatal convulsions (BFNC). <i>Neurogenetics</i> , 2005, 6, 185-193.	0.7	26
90	The amino acid transporter asc-1 is not involved in cystinuria. <i>Kidney International</i> , 2004, 66, 1453-1464.	2.6	25

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91	Impairment of brain and muscle energy metabolism detected by magnetic resonance spectroscopy in hereditary spastic paraparesis type 28 patients with DDHD1 mutations. <i>Journal of Neurology</i> , 2014, 261, 1789-1793.	1.8	25
92	Deletion spanning the 5' ends of both the COL4A5 and COL4A6 genes in a patient with Alport's syndrome and leiomyomatosis. <i>Human Mutation</i> , 1994, 4, 195-198.	1.1	24
93	Characterization of two novel SETX mutations in AOA2 patients reveals aspects of the pathophysiological role of senataxin. <i>Neurogenetics</i> , 2010, 11, 91-100.	0.7	24
94	Pediatric Biobanking: A Pilot Qualitative Survey of Practices, Rules, and Researcher Opinions in Ten European Countries. <i>Biopreservation and Biobanking</i> , 2012, 10, 29-36.	0.5	22
95	Atypical adult onset complicated spastic paraparesis with thin corpus callosum in two patients carrying a novel <i>FA2H</i> mutation. <i>European Journal of Neurology</i> , 2012, 19, e127-9.	1.7	22
96	Hereditary spastic paraparesis in adults. A clinical and genetic perspective from Tuscany. <i>Clinical Neurology and Neurosurgery</i> , 2014, 120, 14-19.	0.6	22
97	The spectrum of brainstem malformations associated to mutations of the tubulin genes family: MRI and DTI analysis. <i>European Radiology</i> , 2019, 29, 770-782.	2.3	22
98	A novel mutation in KIF5A gene causing hereditary spastic paraplegia with axonal neuropathy. <i>Neurological Sciences</i> , 2011, 32, 665-668.	0.9	21
99	A New Region of Conservation Is Defined between Human and Mouse X Chromosomes. <i>Genomics</i> , 1996, 35, 244-247.	1.3	20
100	First case of compound heterozygosity in <i>ALS2</i> gene in infantile-onset ascending spastic paralysis with bulbar involvement. <i>Clinical Genetics</i> , 2008, 73, 591-593.	1.0	20
101	<i>SMC1A</i> codon 496 mutations affect the cellular response to genotoxic treatments. <i>American Journal of Medical Genetics, Part A</i> , 2012, 158A, 224-228.	0.7	19
102	Defining the clinical-genetic and neuroradiological features in SPG54: description of eight additional cases and nine novel DDHD2 variants. <i>Journal of Neurology</i> , 2019, 266, 2657-2664.	1.8	19
103	A Novel Mutation in <i>STXBP1</i> Gene in a Child With Epileptic Encephalopathy and an Atypical Electroclinical Pattern. <i>Journal of Child Neurology</i> , 2014, 29, 249-253.	0.7	18
104	SLC7A8, a Gene Mapping within the Lysinuric Protein Intolerance Critical Region, Encodes a New Member of the Glycoprotein-Associated Amino Acid Transporter Family. <i>Genomics</i> , 1999, 62, 297-303.	1.3	17
105	ZFYVE26/SPASTIZIN. <i>Autophagy</i> , 2014, 10, 374-375.	4.3	17
106	A Novel Mutation in Motor Domain of KIF5A Associated With an HSP/Axonal Neuropathy Phenotype. <i>Journal of Clinical Neuromuscular Disease</i> , 2015, 16, 153-158.	0.3	17
107	A submicroscopic deletion in a patient with isolated X-linked ocular albinism (OA1). <i>Human Molecular Genetics</i> , 1994, 3, 647-648.	1.4	16
108	Mapping of the murine Tbl1 gene reveals a new rearrangement between mouse and human X Chromosomes. <i>Mammalian Genome</i> , 1998, 9, 1062-1064.	1.0	16

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109	A 66-year-old patient with vanishing white matter disease due to the p.Ala87Val <i>EIF2B3</i> mutation. <i>Neurology</i> , 2012, 79, 2077-2078.	1.5	16
110	Clinical and genetic familial study of a large cohort of Italian children with idiopathic epilepsy. <i>Brain Research Bulletin</i> , 2009, 79, 89-96.	1.4	15
111	Loss of <i>ap4s1</i> in zebrafish leads to neurodevelopmental defects resembling spastic paraplegia 52. <i>Annals of Clinical and Translational Neurology</i> , 2020, 7, 584-589.	1.7	15
112	Assignment of the human heterogeneous nuclear ribonucleoprotein A1 gene (<i>HNRPA1</i>) to chromosome 12q13.1 by cDNA competitive in situ hybridization. <i>Genomics</i> , 1992, 12, 171-174.	1.3	14
113	Refining the mutational spectrum and geneâ€‘phenotype correlates in pontocerebellar hypoplasia: results of a multicentric study. <i>Journal of Medical Genetics</i> , 2022, 59, 399-409.	1.5	13
114	Prevalence, Characteristics, and Patterns of Health Care Use for Chronic Headache in Two Areas of Italy. Results of A Questionnaire Interview in General Practice. <i>Cephalalgia</i> , 2003, 23, 175-182.	1.8	12
115	The first Italian family with evidence of pyramidal impairment as phenotypic manifestation of Silver syndrome <i>BSCL2</i> gene mutation. <i>Neurological Sciences</i> , 2008, 29, 189-191.	0.9	12
116	Bilateral frontoparietal polymicrogyria (BFPP) syndrome secondary to a 16q12.1â€‘q21 chromosome deletion involving <i>GPR56</i> gene. <i>Clinical Genetics</i> , 2009, 76, 573-576.	1.0	12
117	The genetic features of 24 patients affected by familial and sporadic hemiplegic migraine. <i>Neurological Sciences</i> , 2011, 32, 141-142.	0.9	12
118	A novel nonsense mutation in the <i>APTX</i> gene associated with delayed DNA singleâ€‘strand break removal fails to enhance sensitivity to different genotoxic agents. <i>Human Mutation</i> , 2011, 32, E2118-33.	1.1	12
119	Atypical late-onset hereditary spastic paraplegia with thin corpus callosum due to novel compound heterozygous mutations in the <i>SPG11</i> gene. <i>Journal of Neurology</i> , 2014, 261, 1825-1827.	1.8	12
120	<i>KIF5A</i> and <i>ALS2</i> Variants in a Family With Hereditary Spastic Paraplegia and Amyotrophic Lateral Sclerosis. <i>Frontiers in Neurology</i> , 2018, 9, 1078.	1.1	12
121	Tensorâ€‘based morphometry using scalar and directional information of diffusion tensor MRI data (DTBM): Application to hereditary spastic paraplegia. <i>Human Brain Mapping</i> , 2018, 39, 4643-4651.	1.9	12
122	Novel <i>SPTBN2</i> gene mutation and first intragenic deletion in early onset spinocerebellar ataxia type 5. <i>Annals of Clinical and Translational Neurology</i> , 2021, 8, 956-963.	1.7	12
123	Monoallelic <i>KIF1A</i> -related disorders: a multicenter cross sectional study and systematic literature review. <i>Journal of Neurology</i> , 2022, 269, 437-450.	1.8	12
124	Molecular cloning of <i>Bacillus subtilis</i> genes involved in DNA metabolism. <i>Molecular Genetics and Genomics</i> , 1987, 209, 8-14.	2.4	11
125	Cerebroretinal Microangiopathy With Calcifications and Cysts Associated With <i>CTC1</i> and <i>NDP</i> Mutations. <i>Journal of Child Neurology</i> , 2013, 28, 1702-1708.	0.7	11
126	Amino acid changes in the amino terminus of the Na,Kâ€‘adenosine triphosphatase alphaâ€‘2 subunit associated to familial and sporadic hemiplegic migraine. <i>Clinical Genetics</i> , 2007, 72, 517-523.	1.0	10

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127	A novel KCNQ3 gene mutation in a child with infantile convulsions and partial epilepsy with centrotemporal spikes. <i>European Journal of Paediatric Neurology</i> , 2015, 19, 102-103.	0.7	10
128	Clinical and genetic features of a cohort of patients with MFN2-related neuropathy. <i>Scientific Reports</i> , 2022, 12, 6181.	1.6	10
129	A novel heterozygous SETX mutation in a patient presenting with chorea and motor neuron disease. <i>Amotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2014, 15, 138-140.	1.1	8
130	Novel SETX variants in a patient with ataxia, neuropathy, and oculomotor apraxia are associated with normal sensitivity to oxidative DNA damaging agents. <i>Brain and Development</i> , 2014, 36, 682-689.	0.6	8
131	Aberrant splicing due to a silent nucleotide change in <i>CCM2</i> gene in a family with cerebral cavernous malformation. <i>Clinical Genetics</i> , 2009, 75, 494-497.	1.0	7
132	Aberrant supracallosal longitudinal bundle: MR features, pathogenesis and associated clinical phenotype. <i>European Radiology</i> , 2016, 26, 2587-2596.	2.3	7
133	Charcot-Marie-Tooth type 1a in a child with Long QT syndrome. <i>European Journal of Paediatric Neurology</i> , 2009, 13, 459-462.	0.7	5
134	Broadening the Spectrum of Adulthood X-Linked Adrenoleukodystrophy: A Report of Two Atypical Cases. <i>Frontiers in Neurology</i> , 2019, 10, 70.	1.1	5
135	U-Fiber Leukoencephalopathy Due to a Novel Mutation in the TACO1 Gene. <i>Neurology: Genetics</i> , 2021, 7, e573.	0.9	5
136	SCN2A Pathogenic Variants and Epilepsy: Heterogeneous Clinical, Genetic and Diagnostic Features. <i>Brain Sciences</i> , 2022, 12, 18.	1.1	5
137	A second A1-type protein is encoded by the human hnRNP A1 gene. <i>Molecular Biology Reports</i> , 1990, 14, 83-84.	1.0	4
138	Exome sequencing reveals a novel homozygous mutation in ACP33 gene in the first Italian family with SPG21. <i>Journal of Neurology</i> , 2017, 264, 2021-2023.	1.8	4
139	Acid Sphingomyelinase Controls Early Phases of Skeletal Muscle Regeneration by Shaping the Macrophage Phenotype. <i>Cells</i> , 2021, 10, 3028.	1.8	4
140	Long-term follow-up in a cohort of children with isolated corpus callosum agenesis at fetal MRI. <i>Annals of Clinical and Translational Neurology</i> , 2021, 8, 2280-2288.	1.7	4
141	Superior Cerebellar Atrophy: An Imaging Clue to Diagnose ITPR1-Related Disorders. <i>International Journal of Molecular Sciences</i> , 2022, 23, 6723.	1.8	4
142	The Italian XLMR bank: a clinical and molecular database. <i>Human Mutation</i> , 2007, 28, 13-18.	1.1	2
143	The blurred scenario of motor neuron disorders linked to Spatacsin mutations: a case report. <i>European Journal of Neurology</i> , 2014, 21, e85-e86.	1.7	2
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145	hnRNP protein A1, molecular structure and gene expression. Molecular Biology Reports, 1987, 12, 169-170.	1.0	1
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