Barbara Castellotti

List of Publications by Year
in descending order

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86	5,350	109321 35	88630 70
papers	citations	h-index	g-index

86	86	86	8301
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Exome sequencing in amyotrophic lateral sclerosis identifies risk genes and pathways. Science, 2015, 347, 1436-1441.	12.6	823
2	Exome-wide Rare Variant Analysis Identifies TUBA4A Mutations Associated with Familial ALS. Neuron, 2014, 84, 324-331.	8.1	308
3	Mutations in the mitochondrial protease gene AFG3L2 cause dominant hereditary ataxia SCA28. Nature Genetics, 2010, 42, 313-321.	21.4	291
4	Phenotypic variability in friedreich ataxia: Role of the associated GAA triplet repeat expansion. Annals of Neurology, 1997, 41, 675-682.	5.3	249
5	Ultra-Rare Genetic Variation in the Epilepsies: A Whole-Exome Sequencing Study of 17,606 Individuals. American Journal of Human Genetics, 2019, 105, 267-282.	6.2	237
6	NEK1 variants confer susceptibility to amyotrophic lateral sclerosis. Nature Genetics, 2016, 48, 1037-1042.	21.4	218
7	High frequency ofTARDBPgene mutations in Italian patients with amyotrophic lateral sclerosis. Human Mutation, 2009, 30, 688-694.	2.5	184
8	Mutations of FUS gene in sporadic amyotrophic lateral sclerosis. Journal of Medical Genetics, 2010, 47, 190-194.	3.2	152
9	Overlapping phenotypes in complex spastic paraplegias SPG11, SPG15, SPG35 and SPG48. Brain, 2014, 137, 1907-1920.	7.6	133
10	Ataxia with isolated vitamin E deficiency: neurological phenotype, clinical follow-up and novel mutations in TTPAgene in Italian families. Neurological Sciences, 2004, 25, 130-137.	1.9	131
11	A genome-wide association meta-analysis identifies a novel locus at 17q11.2 associated with sporadic amyotrophic lateral sclerosis. Human Molecular Genetics, 2014, 23, 2220-2231.	2.9	123
12	Phenotypic manifestations associated with CAG-repeat expansion in the androgen receptor gene in male patients and heterozygous females: a clinical and molecular study of 30 families. Neuromuscular Disorders, 2000, 10, 391-397.	0.6	112
13	Mapping of genes predisposing to idiopathic generalized epilepsy. Human Molecular Genetics, 1995, 4, 1201-1207.	2.9	109
14	Identification of new ANG gene mutations in a large cohort of Italian patients with amyotrophic lateral sclerosis. Neurogenetics, 2008, 9, 33-40.	1.4	102
15	HCN1mutation spectrum: from neonatal epileptic encephalopathy to benign generalized epilepsy and beyond. Brain, 2018, 141, 3160-3178.	7.6	96
16	FMR1 gene premutation is a frequent genetic cause of late-onset sporadic cerebellar ataxia. Neurology, 2005, 64, 145-147.	1.1	90
17	Novel optineurin mutations in patients with familial and sporadic amyotrophic lateral sclerosis. Journal of Neurology, Neurosurgery and Psychiatry, 2011, 82, 1239-1243.	1.9	86
18	C9ORF72 repeat expansion in a large Italian ALS cohort: evidence of a founder effect. Neurobiology of Aging, 2012, 33, 2528.e7-2528.e14.	3.1	74

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19	Ubiquilin 2mutations in Italian patients with amyotrophic lateral sclerosis and frontotemporal dementia. Journal of Neurology, Neurosurgery and Psychiatry, 2013, 84, 183-187.	1.9	74
20	Paroxysmal movement disorders in GLUT1 deficiency syndrome. Neurology, 2008, 71, 146-148.	1.1	73
21	Atypical movement disorders in the early stages of Huntington's disease: clinical and genetic analysis. Clinical Genetics, 2001, 58, 50-56.	2.0	72
22	Frataxin gene point mutations in Italian Friedreich ataxia patients. Neurogenetics, 2007, 8, 289-299.	1.4	71
23	Clinical and molecular studies of 73 Italian families with autosomal dominant cerebellar ataxia type I: SCA1 and SCA2 are the most common genotypes. Journal of Neurology, 1999, 246, 389-393.	3.6	63
24	Identification of novel and recurrent CACNA1A gene mutations in fifteen patients with episodic ataxia type 2. Journal of the Neurological Sciences, 2010, 291, 30-36.	0.6	63
25	Association of a Locus in theCAMTA1Gene With Survival in Patients With Sporadic Amyotrophic Lateral Sclerosis. JAMA Neurology, 2016, 73, 812.	9.0	57
26	Early Treatment with Quinidine in 2 Patients with Epilepsy of Infancy with Migrating Focal Seizures (EIMFS) Due to Gain-of-Function KCNT1 Mutations: Functional Studies, Clinical Responses, and Critical Issues for Personalized Therapy. Neurotherapeutics, 2018, 15, 1112-1126.	4.4	56
27	Pathogenic Huntingtin Repeat Expansions in Patients with Frontotemporal Dementia and Amyotrophic Lateral Sclerosis. Neuron, 2021, 109, 448-460.e4.	8.1	56
28	Expanding the phenotypic spectrum of Allanâ€“Herndonâ€“Dudley syndrome in patients with <scp>SLC</scp>16A2 mutations. Developmental Medicine and Child Neurology, 2019, 61, 1439-1447.	2.1	53
29	Disease characteristics of MCT8 deficiency: an international, retrospective, multicentre cohort study. Lancet Diabetes and Endocrinology,the, 2020, 8, 594-605.	11.4	50
30	Superoxide dismutase gene mutations in Italian patients with familial and sporadic amyotrophic lateral sclerosis: identification of three novel missense mutations. Neuromuscular Disorders, 2001, 11, 404-410.	0.6	47
31	A novel de novo HCN1 loss-of-function mutation in genetic generalized epilepsy causing increased neuronal excitability. Neurobiology of Disease, 2018, 118, 55-63.	4.4	47
32	Epilepsy subtype-specific copy number burden observed in a genome-wide study of 17â€‰458 subjects. Brain, 2020, 143, 2106-2118.	7.6	47
33	Ataxia with oculomotor apraxia type1 (AOA1): novel and recurrent aprataxin mutations, coenzyme Q10 analyses, and clinical findings in Italian patients. Neurogenetics, 2011, 12, 193-201.	1.4	46
34	Association of Variants in the SPTLC1 Gene With Juvenile Amyotrophic Lateral Sclerosis. JAMA Neurology, 2021, 78, 1236.	9.0	46
35	Mutational Analysis of EFHC1 Gene in Italian Families with Juvenile Myoclonic Epilepsy. Epilepsia, 2007, 48, 1686-1690.	5.1	44
36	TUBA4A gene analysis in sporadic amyotrophic lateral sclerosis: identification of novel mutations. Journal of Neurology, 2015, 262, 1376-1378.	3.6	44

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37	Progressive myoclonus epilepsiesâ€"Residual unsolved cases have marked genetic heterogeneity including dolichol-dependent protein glycosylation pathway genes. American Journal of Human Genetics, 2021, 108, 722-738.	6.2	41
38	Family and molecular data for a fine analysis of age at onset in Huntington disease. American Journal of Medical Genetics Part A, 2000, 95, 366-373.	2.4	40
39	Very late onset Friedreich's ataxia without cardiomyopathy is associated with limited GAA expansion in the X25 gene. Neurology, 1997, 49, 1153-1155.	1.1	35
40	Screening of the PFN1 gene in sporadic amyotrophic lateral sclerosis and in frontotemporal dementia. Neurobiology of Aging, 2013, 34, 1517.e9-1517.e10.	3.1	35
41	Sub-genic intolerance, ClinVar, and the epilepsies: A whole-exome sequencing study of 29,165 individuals. American Journal of Human Genetics, 2021, 108, 965-982.	6.2	35
42	HCN ion channels and accessory proteins in epilepsy: genetic analysis of a large cohort of patients and review of the literature. Epilepsy Research, 2019, 153, 49-58.	1.6	32
43	Clinical and Molecular Characteristics of SLC16A2 (MCT8) Mutations in Three Families with the Allan-Herndon-Dudley Syndrome. Human Mutation, 2017, 38, 260-264.	2.5	31
44	Neonatal developmental and epileptic encephalopathy due to autosomal recessive variants in SLC13A5 gene. Epilepsia, 2020, 61, 2474-2485.	5.1	31
45	Analysis of hnRNPA1, A2/B1, and A3 genes in patients with amyotrophic lateral sclerosis. Neurobiology of Aging, 2013, 34, 2695.e11-2695.e12.	3.1	30
46	Preferential expression of mutant ABCD1 allele is common in adrenoleukodystrophy female carriers but unrelated to clinical symptoms. Orphanet Journal of Rare Diseases, 2012, 7, 10.	2.7	29
47	No association of DPP6 with amyotrophic lateral sclerosis in an Italian population. Neurobiology of Aging, 2011, 32, 966-967.	3.1	28
48	Kufs disease due to mutation ofCLN6: clinical, pathological and molecular genetic features. Brain, 2019, 142, 59-69.	7.6	28
49	ATAXIN2 CAG-repeat length in Italian patients with amyotrophic lateral sclerosis: risk factor or variant phenotype? Implication for genetic testing and counseling. Neurobiology of Aging, 2012, 33, 1847.e15-1847.e21.	3.1	27
50	Autosomal dominant lateral temporal epilepsy: Absence of mutations in ADAM22 and Kv1 channel genes encoding LGI1-associated proteins. Epilepsy Research, 2008, 80, 1-8.	1.6	26
51	A Loss-of-Function HCN4 Mutation Associated With Familial Benign Myoclonic Epilepsy in Infancy Causes Increased Neuronal Excitability. Frontiers in Molecular Neuroscience, 2018, 11, 269.	2.9	25
52	Lack of aprataxin impairs mitochondrial functions via downregulation of the APE1/NRF1/NRF2 pathway. Human Molecular Genetics, 2015, 24, 4516-4529.	2.9	23
53	Ataxia With Oculomotor Apraxia Type 1 (AOA1): Clinical and Neuropsychological Features in 2 New Patients and Differential Diagnosis. Journal of Child Neurology, 2008, 23, 895-900.	1.4	22
54	Refractory Absence Epilepsy and Glut1 Deficiency Syndrome: A New Case Report and Literature Review. Neuropediatrics, 2014, 45, 328-332.	0.6	22

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55	Kv7.3 Compound Heterozygous Variants in Early Onset Encephalopathy Reveal Additive Contribution of C-Terminal Residues to PIP2-Dependent K+ Channel Gating. Molecular Neurobiology, 2018, 55, 7009-7024.	4.0	21
56	Substantia Nigra Swelling and Dentate Nucleus T2 Hyperintensity May Be Early Magnetic Resonance Imaging Signs of Î²â€Propeller Proteinâ€Associated Neurodegeneration. Movement Disorders Clinical Practice, 2019, 6, 51-56.	1.5	20
57	ASAH1 variant causing a mild SMA phenotype with no myoclonic epilepsy: a clinical, biochemical and molecular study. European Journal of Human Genetics, 2016, 24, 1578-1583.	2.8	18
58	Screening of SLC2A1 in a large cohort of patients suspected for Glut1 deficiency syndrome: identification of novel variants and associated phenotypes. Journal of Neurology, 2019, 266, 1439-1448.	3.6	18
59	Unusual EEG pattern linked to chromosome 3p in a family with idiopathic generalized epilepsy. Neurology, 1998, 51, 493-498.	1.1	17
60	Mutational analysis of VCP gene in familial amyotrophic lateral sclerosis. Neurobiology of Aging, 2012, 33, 630.e1-630.e2.	3.1	17
61	Paroxysmal exercise-induced dyskinesia with self-limiting partial epilepsy: A novel GLUT-1Âmutation with benign phenotype. Parkinsonism and Related Disorders, 2011, 17, 479-481.	2.2	16
62	Alternating Hemiplegia and Epilepsia Partialis Continua: A new phenotype for a novel compound TBC1D24 mutation. Seizure: the Journal of the British Epilepsy Association, 2017, 47, 71-73.	2.0	16
63	Progressive myoclonus epilepsy caused by a gain-of-function KCNA2 mutation. Seizure: the Journal of the British Epilepsy Association, 2019, 65, 106-108.	2.0	16
64	Clinical and molecular findings in the first identified Italian family with dentatorubral-pallidoluysian atrophy. Acta Neurologica Scandinavica, 1998, 98, 324-327.	2.1	15
65	The role of de novo mutations in the development of amyotrophic lateral sclerosis. Human Mutation, 2017, 38, 1534-1541.	2.5	13
66	Novel mutations in SLC16A2 associated with a less severe phenotype of MCT8 deficiency. Metabolic Brain Disease, 2019, 34, 1565-1575.	2.9	12
67	Do the functional properties of HCN1 mutants correlate with the clinical features in epileptic patients?. Progress in Biophysics and Molecular Biology, 2021, 166, 147-155.	2.9	11
68	Riboflavin-responsive multiple acyl-CoA dehydrogenase deficiency with unknown genetic defect. Neurological Sciences, 2012, 33, 1383-1387.	1.9	10
69	Hyperargininemia: 7-Month Follow-Up Under Sodium Benzoate Therapy in an Italian Child Presenting Progressive Spastic Paraparesis, Cognitive Decline, and Novel Mutation in ARG1 Gene. Pediatric Neurology, 2014, 51, 430-433.	2.1	9
70	In-vivo brain H1-MR-Spectroscopy identification and quantification of 2-hydroxyglutarate in L-2-Hydroxyglutaric aciduria. Brain Research, 2016, 1648, 506-511.	2.2	9
71	Gabapentin treatment in a patient with KCNQ2 developmental epileptic encephalopathy. Pharmacological Research, 2020, 160, 105200.	7.1	7
72	Diagnosis and Management of Type 1 Sialidosis: Clinical Insights from Long-Term Care of Four Unrelated Patients. Brain Sciences, 2020, 10, 506.	2.3	7

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73	Saposin B deficiency as a cause of adult-onset metachromatic leukodystrophy. Neurology, 2019, 93, 310-312.	1.1	6
74	Clinical and molecular report of novel GALC mutations in Moroccan patient with Krabbe disease: case report. BMC Pediatrics, 2015, 15, 182.	1.7	5
75	Severe epilepsy in CNTNAP2-related Pitt-Hopkins-like syndrome successfully treated with stiripentol. Seizure: the Journal of the British Epilepsy Association, 2021, 88, 143-145.	2.0	4
76	Early Parkinsonism in a Senegalese girl with Lafora disease. Epileptic Disorders, 2020, 22, 233-236.	1.3	4
77	Granny trips down: is she carrying the big bad wolf?. Neurological Sciences, 2013, 34, 2077-2079.	1.9	2
78	Epilepsy and NREM-parasomnia caused by novel hemizygous ARHGEF9 mutation. Sleep Medicine, 2020, 76, 158-159.	1.6	2
79	Peripheral nerve enlargement on nerve ultrasound parallels neuropathological changes in adultâ€onset Krabbe disease. Muscle and Nerve, 2021, 63, E33-E35.	2.2	2
80	Paroxysmal tonic upgaze in a child with SCN8A-related encephalopathy. Epileptic Disorders, 2021, 23, 643-647.	1.3	2
81	Successful use of perampanel in GABRA1-related myoclonic epilepsy with photosensitivity. Epilepsy and Behavior Reports, 2022, 19, 100544.	1.0	2
82	Kennedy's disease: clinical and molecular study of two Italian families. Italian Journal of Neurological Sciences, 1995, 16, 467-471.	0.1	1
83	Biopsy-proven multiple sclerosis in an adult patient with atypical craniometaphyseal dysplasia. BMJ Case Reports, 2018, 2018, bcr-2017-223390.	0.5	1
84	Clinical and genetic study of a family with spinocerebellar ataxia type 1 (SCA1) and beta-thalassemia. Italian Journal of Neurological Sciences, 1998, 19, 345-350.	0.1	0
85	SCN8A splicing mutation causing skipping of the exon 15 associated with intellectual disability and cortical myoclonus. Seizure: the Journal of the British Epilepsy Association, 2020, 82, 56-58.	2.0	0
86	Functional Characterization of Two Variants at the Intron 6â€”Exon 7 Boundary of the KCNQ2 Potassium Channel Gene Causing Distinct Epileptic Phenotypes. Frontiers in Pharmacology, 0, 13, .	3.5	0