Giovanna Zorzi

List of Publications by Year in descending order

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113 papers	5,097 citations	94433 37 h-index	95266 68 g-index
118 all docs	118 docs citations	118 times ranked	7611 citing authors

#	Article	IF	Citations
1	PLA2G6, encoding a phospholipase A2, is mutated in neurodegenerative disorders with high brain iron. Nature Genetics, 2006, 38, 752-754.	21.4	497
2	Exome Sequencing Reveals De Novo WDR45 Mutations Causing a Phenotypically Distinct, X-Linked Dominant Form of NBIA. American Journal of Human Genetics, 2012, 91, 1144-1149.	6.2	309
3	Neurodegeneration associated with genetic defects in phospholipase A ₂. Neurology, 2008, 71, 1402-1409.	1.1	236
4	Beta-propeller protein-associated neurodegeneration: a new X-linked dominant disorder with brain iron accumulation. Brain, 2013, 136, 1708-1717.	7.6	203
5	Genotypic and phenotypic spectrum of PANK2 mutations in patients with neurodegeneration with brain iron accumulation. Annals of Neurology, 2006, 59, 248-256.	5.3	184
6	Exome Sequence Reveals Mutations in CoA Synthase as a Cause of Neurodegeneration with Brain Iron Accumulation. American Journal of Human Genetics, 2014, 94, 11-22.	6.2	176
7	Stimulation of the globus pallidus internus for childhoodâ€onset dystonia. Movement Disorders, 2005, 20, 1194-1200.	3.9	162
8	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
9	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.	3.9	125
10	Influence of strict, intermediate, and broad diagnostic criteria on the age- and sex-specific incidence of Parkinson's disease. Movement Disorders, 2000, 15, 819-825.	3.9	112
11	A Missense Mutation in KCTD17 Causes Autosomal Dominant Myoclonus-Dystonia. American Journal of Human Genetics, 2015, 96, 938-947.	6.2	109
12	Infantile neuroaxonal dystrophy. Neurology, 1999, 52, 1472-1472.	1.1	108
13	Metabolic consequences of mitochondrial coenzyme A deficiency in patients with PANK2 mutations. Molecular Genetics and Metabolism, 2012, 105, 463-471.	1.1	106
14	Safety and efficacy of deferiprone for pantothenate kinase-associated neurodegeneration: a randomised, double-blind, controlled trial and an open-label extension study. Lancet Neurology, The, 2019, 18, 631-642.	10.2	102
15	De Novo Mutations in PDE10A Cause Childhood-Onset Chorea with Bilateral Striatal Lesions. American Journal of Human Genetics, 2016, 98, 763-771.	6.2	96
16	Status dystonicus: Predictors of outcome and progression patterns of underlying disease. Movement Disorders, 2012, 27, 783-788.	3.9	94
17	Long-term high-frequency bilateral pallidal stimulation for neuroleptic-induced tardive dystonia. Journal of Neurosurgery, 2005, 102, 721-725.	1.6	87
18	Myoclonusâ€“dystonia syndrome: Clinical presentation, disease course, and genetic features in 11 families. Movement Disorders, 2008, 23, 28-34.	3.9	75

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19	Paroxysmal movement disorders in GLUT1 deficiency syndrome. Neurology, 2008, 71, 146-148.	1.1	73
20	Infantile neuroaxonal dystrophy: neuroradiological studies in 11 patients. Neuroradiology, 1999, 41, 376-380.	2.2	70
21	Lossâ€ofâ€Function Variants in <scp>HOPS</scp> Complex Genes <scp>VPS16</scp> and <scp>VPS41</scp> Cause Early Onset Dystonia Associated with Lysosomal Abnormalities. Annals of Neurology, 2020, 88, 867-877.	5.3	70
22	ADCY5-related movement disorders: Frequency, disease course and phenotypic variability in a cohort of paediatric patients. Parkinsonism and Related Disorders, 2017, 41, 37-43.	2.2	67
23	Consensus clinical management guideline for pantothenate kinase-associated neurodegeneration (PKAN). Molecular Genetics and Metabolism, 2017, 120, 278-287.	1.1	64
24	Neurological Disorders, other than Stroke, Associated with Antiphospholipid Antibodies in Childhood. Neuropediatrics, 1996, 27, 149-153.	0.6	63
25	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
26	Pallidal stimulation for acquired dystonia due to cerebral palsy: beyond 5Âyears. European Journal of Neurology, 2015, 22, 426.	3.3	58
27	Frequency and phenotypic spectrum of KMT2B dystonia in childhood: A singleâ€center cohort study. Movement Disorders, 2019, 34, 1516-1527.	3.9	55
28	ATP1A3-related disorders: An update. European Journal of Paediatric Neurology, 2018, 22, 257-263.	1.6	54
29	Neonatal dopa-responsive extrapyramidal syndrome in twins with recessive GTPCH deficiency. Neurology, 2003, 60, 335-337.	1.1	50
30	Neuronal ceroid-lipofuscinosis: A clinical and morphological study of 19 patients. American Journal of Medical Genetics Part A, 1995, 57, 137-141.	2.4	46
31	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
32	Deep brain stimulation for dystonia due to cerebral palsy: A review. European Journal of Paediatric Neurology, 2018, 22, 308-315.	1.6	44
33	A neurophysiological study of myoclonus in patients with DYT11 myoclonusâ€dystonia syndrome. Movement Disorders, 2008, 23, 2041-2048.	3.9	43
34	Mutation screening of the DYT6/THAP1 gene in Italy. Movement Disorders, 2009, 24, 2424-2427.	3.9	43
35	Partial Seizures Associated with Antiphospholipid Antibodies in Childhood. Neuropediatrics, 1998, 29, 249-254.	0.6	41
36	Deep brain stimulation for movement disorders. Considerations on 276 consecutive patients. Journal of Neural Transmission, 2011, 118, 1497-1510.	2.8	39

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37	Phenomenology of psychogenic movement disorders in children. Movement Disorders, 2012, 27, 1153-1157.	3.9	39
38	Detection of Sepiapterin in CSF of Patients with Sepiapterin Reductase Deficiency. Molecular Genetics and Metabolism, 2002, 75, 174-177.	1.1	38
39	C19orf12 and FA2H Mutations Are Rare in Italian Patients With Neurodegeneration With Brain Iron Accumulation. Seminars in Pediatric Neurology, 2012, 19, 75-81.	2.0	38
40	Non-DYT1 early-onset primary torsion dystonia: Comparison with DYT1 phenotype and review of the literature. Movement Disorders, 2006, 21, 1411-1418.	3.9	37
41	Frequency of DYT1 mutation in early onset primary dystonia in Italian patients. Movement Disorders, 2002, 17, 407-408.	3.9	36
42	The â€œEye-of-the-Tigerâ€•Sign may be Absent in the Early Stages of Classic Pantothenate Kinase Associated Neurodegeneration. Neuropediatrics, 2011, 42, 159-162.	0.6	34
43	Changes in Red Blood Cell membrane lipid composition: A new perspective into the pathogenesis of PKAN. Molecular Genetics and Metabolism, 2017, 121, 180-189.	1.1	34
44	Glucose transporter type 1 deficiency: Ketogenic diet in three patients with atypical phenotype. Brain and Development, 2010, 32, 404-408.	1.1	33
45	Sporadic and familial glut1ds Italian patients: A wide clinical variability. Seizure: the Journal of the British Epilepsy Association, 2015, 24, 28-32.	2.0	32
46	SETX mutations are a frequent genetic cause of juvenile and adult onset cerebellar ataxia with neuropathy and elevated serum alpha-fetoprotein. Orphanet Journal of Rare Diseases, 2013, 8, 123.	2.7	31
47	Reduced nitric oxide metabolites in CSF of patients with tetrahydrobiopterin deficiency. Journal of Neurochemistry, 2002, 80, 362-364.	3.9	30
48	Extragenetic factors and clinical penetrance of DYT1 dystonia: an exploratory study. Journal of Neurology, 2013, 260, 1081-1086.	3.6	30
49	EMG-Based Visual-Haptic Biofeedback: A Tool to Improve Motor Control in Children With Primary Dystonia. IEEE Transactions on Neural Systems and Rehabilitation Engineering, 2013, 21, 474-480.	4.9	29
50	Molecular Genetics and Interferon Signature in the Italian Aicardi GoutiÃ¨res Syndrome Cohort: Report of 12 New Cases and Literature Review. Journal of Clinical Medicine, 2019, 8, 750.	2.4	29
51	Acquired hemidystonia in childhood: A clinical and neuroradiological study of thirteen patients. Pediatric Neurology, 1996, 15, 108-113.	2.1	28
52	Targeting the brain: considerations in 332 consecutive patients treated by deep brain stimulation (DBS) for severe neurological diseases. Neurological Sciences, 2012, 33, 1285-1303.	1.9	28
53	TheCACNA1BR1389H variant is not associated with myoclonus-dystonia in a large European multicentric cohort. Human Molecular Genetics, 2015, 24, 5326-5329.	2.9	28
54	Paroxysmal dyskinesias in childhood. Pediatric Neurology, 2003, 28, 168-172.	2.1	27

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55	GTP-cyclohydrolase I gene mutations in patients with autosomal dominant and recessive GTP-CH1 deficiency: Identification and functional characterization of four novel mutations. Journal of Inherited Metabolic Disease, 2004, 27, 455-463.	3.6	27
56	Pallidal Deep Brain Stimulation in DYT6 Dystonia: Clinical Outcome and Predictive Factors for Motor Improvement. Journal of Clinical Medicine, 2019, 8, 2163.	2.4	25
57	Bilateral striatal necrosis associated withMycoplasma pneumoniae infection in an adolescent: Clinical and neuroradiologic follow up. Movement Disorders, 2000, 15, 1023-1026.	3.9	23
58	Reaching and Writing Movements: Sensitive and Reliable Tools to Measure Genetic Dystonia in Children. Journal of Child Neurology, 2011, 26, 822-829.	1.4	23
59	Neurological Disorders Associated with Striatal Lesions: Classification and Diagnostic Approach. Current Neurology and Neuroscience Reports, 2016, 16, 54.	4.2	23
60	Deep Brain Stimulation Electrode Used for Radiofrequency Lesion of the Globus Pallidus Internus in Dystonia. Stereotactic and Functional Neurosurgery, 2009, 87, 348-352.	1.5	21
61	Heterozygous KIF1A variants underlie a wide spectrum of neurodevelopmental and neurodegenerative disorders. Journal of Medical Genetics, 2021, 58, 475-483.	3.2	21
62	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
63	Fosmetpantotenate Randomized Controlled Trial in Pantothenate Kinaseâ€“Associated Neurodegeneration. Movement Disorders, 2021, 36, 1342-1352.	3.9	20
64	Error-enhancing robot therapy to induce motor control improvement in childhood onset primary dystonia. Journal of NeuroEngineering and Rehabilitation, 2012, 9, 46.	4.6	18
65	A Novel Mutation in STXBP1 Gene in a Child With Epileptic Encephalopathy and an Atypical Electroclinical Pattern. Journal of Child Neurology, 2014, 29, 249-253.	1.4	18
66	Clinical rating scale for pantothenate kinaseâ€associated neurodegeneration: A pilot study. Movement Disorders, 2017, 32, 1620-1630.	3.9	18
67	Paroxysmal nonâ€epileptic motor events in childhood: a clinical and videoâ€EEGâ€“polymyographic study. Developmental Medicine and Child Neurology, 2012, 54, 334-338.	2.1	17
68	Encephalopathies with intracranial calcification in children: clinical and genetic characterization. Orphanet Journal of Rare Diseases, 2018, 13, 135.	2.7	17
69	CANS: Childhood acute neuropsychiatric syndromes. European Journal of Paediatric Neurology, 2018, 22, 316-320.	1.6	16
70	<scp>YY1</scp>â€Related Dystonia: Clinical Aspects and Longâ€Term Response to Deep Brain Stimulation. Movement Disorders, 2021, 36, 1461-1462.	3.9	16
71	Infantile neuroaxonal dystrophy and pantothenate kinase-associated neurodegeneration. Neurology, 2004, 63, 922-924.	1.1	14
72	Clinical and genetic features of paroxysmal kinesigenic dyskinesia in Italian patients. European Journal of Paediatric Neurology, 2016, 20, 152-157.	1.6	14

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73	Deep brain stimulation versus pallidotomy for status dystonicus: a single-center case series. Journal of Neurosurgery, 2021, 134, 197-207.	1.6	14
74	Pathophysiology and Treatment of Neurodegeneration With Brain Iron Accumulation in the Pediatric Population. Current Treatment Options in Neurology, 2013, 15, 652-667.	1.8	13
75	A family with paroxysmal nonkinesigenic dyskinesias (PNKD): Evidence of mitochondrial dysfunction. European Journal of Paediatric Neurology, 2015, 19, 64-68.	1.6	13
76	DYT2 screening in early-onset isolated dystonia. European Journal of Paediatric Neurology, 2017, 21, 269-271.	1.6	13
77	A PDE10A de novo mutation causes childhoodâ€onset chorea with diurnal fluctuations. Movement Disorders, 2017, 32, 1646-1647.	3.9	13
78	Paroxysmal dystonia and paroxysmal tremor in a young patient with multiple sclerosis. Italian Journal of Neurological Sciences, 1995, 16, 315-319.	0.1	11
79	Polymyography in the diagnosis of childhood onset movement disorders. European Journal of Paediatric Neurology, 2008, 12, 480-483.	1.6	11
80	Early onset primary dystonia. European Journal of Paediatric Neurology, 2009, 13, 488-492.	1.6	11
81	Deep brain stimulation in critical care conditions. Journal of Neural Transmission, 2014, 121, 391-398.	2.8	11
82	R106C TFG variant causes infantile neuroaxonal dystrophy â€œplusâ€•syndrome. Neurogenetics, 2018, 19, 179-187.	1.4	11
83	Spontaneous MRI improvement and absence of cerebral calcification in Aicardi-GoutiÃ¨res syndrome: Diagnostic and disease-monitoring implications. Molecular Genetics and Metabolism, 2019, 126, 489-494.	1.1	10
84	Childhood-onset HAM/TSP with progressive cognitive impairment. Neurological Sciences, 2010, 31, 209-212.	1.9	9
85	Therapeutic Advances in Neurodegeneration With Brain Iron Accumulation. Seminars in Pediatric Neurology, 2012, 19, 82-86.	2.0	9
86	Therapeutic Advances in Neurodegeneration with Brain Iron Accumulation. International Review of Neurobiology, 2013, 110, 153-164.	2.0	9
87	Benign hereditary chorea and deletions outside NKX2-1: What's the role of MBIP?. European Journal of Medical Genetics, 2018, 61, 581-584.	1.3	9
88	Diagnosis and treatment of pediatric onset isolated dystonia. European Journal of Paediatric Neurology, 2018, 22, 238-244.	1.6	9
89	Persistent Fixed Torticollis due to Atlanto-Axial Rotatory Fixation: Report of 4 Pediatric Cases. Neuropediatrics, 2005, 36, 45-49.	0.6	8
90	Axonal dystrophies. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2013, 113, 1919-1924.	1.8	7

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91	Long term perceptions of illness and self after Deep Brain Stimulation in pediatric dystonia: A narrative research. European Journal of Paediatric Neurology, 2020, 26, 61-67.	1.6	7
92	SLC19A3 related disorder: Treatment implication and clinical outcome of 2 new patients. European Journal of Paediatric Neurology, 2018, 22, 332-335.	1.6	6
93	EMG-based vibro-tactile biofeedback training: effective learning accelerator for children and adolescents with dystonia? A pilot crossover trial. Journal of NeuroEngineering and Rehabilitation, 2019, 16, 150.	4.6	6
94	Restless Legs Syndrome in NKX2-1-related chorea: An expansion of the disease spectrum. Brain and Development, 2019, 41, 250-256.	1.1	6
95	Functional characterization of two novel mutations in TTF-1/NKX2.1 homeodomain in patients with benign hereditary chorea. Journal of the Neurological Sciences, 2016, 360, 78-83.	0.6	5
96	Transient paroxysmal dystonia in an infant possibly induced by cisapride. Italian Journal of Neurological Sciences, 1996, 17, 157-159.	0.1	4
97	Morphological and chemical analysis of a deep brain stimulation electrode explanted from a dystonic patient. Journal of Neural Transmission, 2013, 120, 1425-1431.	2.8	4
98	The noncoding RNA AK127244 in 2p16.3 locus: A new susceptibility region for neuropsychiatric disorders. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2018, 177, 557-562.	1.7	4
99	Status dystonicus induced by deep brain stimulation surgery. Neurological Sciences, 2020, 41, 729-730.	1.9	4
100	Pediatric Paroxysmal Exercise-Induced Neurological Symptoms: Clinical Spectrum and Diagnostic Algorithm. Frontiers in Neurology, 2021, 12, 658178.	2.4	4
101	Inherited Isolated Dystonia in Children. Journal of Pediatric Neurology, 2015, 13, 174-179.	0.2	3
102	Cerebrospinal Fluid Monoamine Metabolite Analysis in Pediatric Movement Disorders. Journal of Child Neurology, 2015, 30, 1800-1805.	1.4	3
103	Globus pallidus internus activity during simultaneous bilateral microelectrode recordings in status dystonicus. Acta Neurochirurgica, 2021, 163, 211-217.	1.7	3
104	<scp>THAP1</scp> Dystonia with Globus Pallidus <scp>T2</scp> Hypointensity: A Report of Two Cases. Movement Disorders, 2021, 36, 1463-1464.	3.9	3
105	Elevated aspartate aminotransferase and lactate dehydrogenase levels are a constant finding in <scp>PLA</scp>2G6â€associated neurodegeneration. European Journal of Neurology, 2016, 23, e24-5.	3.3	2
106	Diagnostic issues in childhood and adult dystonia. Expert Opinion on Medical Diagnostics, 2011, 5, 483-500.	1.6	1
107	Childhood onset of acquired neuromyotonia: Association with a ganglioneuroma. Muscle and Nerve, 2015, 51, 620-621.	2.2	1
108	Proteinâ€redistribution diet in a case of tyrosine hydroxylase enzyme deficiency. Movement Disorders, 2017, 32, 794-795.	3.9	1

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109	Indicazioni e trattamento chirurgico della distonia dellâ€™etÃ pediatrica. Area Pediatrica, 2012, 13, 93-100.	0.0	0
110	Global metabolic profiling reveals metabolic consequences of mitochondrial coenzyme A deficiency in patients with PANK2 mutations. Mitochondrion, 2012, 12, 577.	3.4	0
111	The first case of Cri du Chat syndrome with dystonia. Clinical Neurology and Neurosurgery, 2021, 201, 106459.	1.4	0
112	Distonie. , 2009, , 393-398.		0
113	Neurodegeneration with Brain Iron Accumulation. , 2014, , 171-198.		0