Miryam Carecchio

List of Publications by Year
in descending order

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Version: 2024-02-01

66 papers	2,133 citations	218677 26 h-index	254184 43 g-index

71 all docs	71 docs citations	71 times ranked	3986 citing authors

#	Article	IF	Citations
1	<scp>GPiâ€ĐBS</scp> for <scp>KMT2B</scp>â€Associated Dystonia: Systematic Review and Metaâ€Analysis. Movement Disorders Clinical Practice, 2022, 9, 31-37.	1.5	14
2	Effect of Intensive Rehabilitation Program in Thermal Water on a Group of People with Parkinsonâ€™s Disease: A Retrospective Longitudinal Study. Healthcare (Switzerland), 2022, 10, 368.	2.0	4
3	<scp>EIF2AK2</scp> Missense Variants Associated with Early Onset Generalized Dystonia. Annals of Neurology, 2021, 89, 485-497.	5.3	32
4	Expanding the genetic spectrum of primary familial brain calcification due to SLC2OA2 mutations: a case series. Neurogenetics, 2021, 22, 65-70.	1.4	4
5	<scp>NKX2</scp>.1 runâ€on mutation associated to familial brainâ€“lungâ€“thyroid syndrome. Clinical Genetics, 2021, 100, 114-116.	2.0	3
6	Cerebellar and cortical hypometabolism in progressive stimulus-sensitive limb myoclonus in celiac disease. Neurological Sciences, 2021, 42, 3453-3455.	1.9	0
7	High prolactin levels in dihydropteridine reductase deficiency: A sign of therapy failure or additional pathology?. JIMD Reports, 2021, 61, 48-51.	1.5	4
8	Impact of social and mobility restrictions in Parkinsonâ€™s disease during COVID-19 lockdown. BMC Neurology, 2021, 21, 332.	1.8	25
9	MYORG -related disease is associated with central pontine calcifications and atypical parkinsonism. Neurology: Genetics, 2020, 6, e399.	1.9	13
10	Harmful Iron-Calcium Relationship in Pantothenate kinase Associated Neurodegeneration. International Journal of Molecular Sciences, 2020, 21, 3664.	4.1	19
11	Frequency and phenotypic spectrum of KMT2B dystonia in childhood: A singleâ€center cohort study. Movement Disorders, 2019, 34, 1516-1527.	3.9	55
12	Spasmodic dysphonia as a presenting symptom of spinocerebellar ataxia type 12. Neurogenetics, 2019, 20, 161-164.	1.4	3
13	Long-term effect of subthalamic and pallidal deep brain stimulation for status dystonicus in children with methylmalonic acidemia and GNAO1 mutation. Journal of Neural Transmission, 2019, 126, 739-757.	2.8	24
14	Inborn errors of coenzyme A metabolism and neurodegeneration. Journal of Inherited Metabolic Disease, 2019, 42, 49-56.	3.6	36
15	Adult diagnosis of Cockayne syndrome. Neurology, 2019, 93, 854-855.	1.1	3
16	Parkinsonism in neurometabolic diseases. International Review of Neurobiology, 2019, 149, 355-376.	2.0	0
17	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
18	Restless Legs Syndrome in NKX2-1-related chorea: An expansion of the disease spectrum. Brain and Development, 2019, 41, 250-256.	1.1	6

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19	Recessive mutations in VPS13D cause childhood onset movement disorders. Annals of Neurology, 2018, 83, 1089-1095.	5.3	104
20	CANS: Childhood acute neuropsychiatric syndromes. European Journal of Paediatric Neurology, 2018, 22, 316-320.	1.6	16
21	The relevance of gene panels in movement disorders diagnosis: A lab perspective. European Journal of Paediatric Neurology, 2018, 22, 285-291.	1.6	32
22	Diagnosis and treatment of pediatric onset isolated dystonia. European Journal of Paediatric Neurology, 2018, 22, 238-244.	1.6	9
23	ATP1A3-related disorders: An update. European Journal of Paediatric Neurology, 2018, 22, 257-263.	1.6	54
24	Inborn errors of coenzyme A metabolism and neurodegeneration. Journal of Inherited Metabolic Disease, 2018, , .	3.6	3
25	Primary brain calcification: an international study reporting novel variants and associated phenotypes. European Journal of Human Genetics, 2018, 26, 1462-1477.	2.8	48
26	The Length of SNCA Rep1 Microsatellite May Influence Cognitive Evolution in Parkinsonâ€™s Disease. Frontiers in Neurology, 2018, 9, 213.	2.4	21
27	Partial loss-of-function of sodium channel SCN8A in familial isolated myoclonus. Human Mutation, 2018, 39, 965-969.	2.5	34
28	Encephalopathies with intracranial calcification in children: clinical and genetic characterization. Orphanet Journal of Rare Diseases, 2018, 13, 135.	2.7	17
29	DYT2 screening in early-onset isolated dystonia. European Journal of Paediatric Neurology, 2017, 21, 269-271.	1.6	13
30	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
31	Emerging Monogenic Complex Hyperkinetic Disorders. Current Neurology and Neuroscience Reports, 2017, 17, 97.	4.2	51
32	A PDE10A de novo mutation causes childhoodâ€onset chorea with diurnal fluctuations. Movement Disorders, 2017, 32, 1646-1647.	3.9	13
33	SPG5 siblings with different phenotypes showing reduction of 27-hydroxycholesterol after simvastatin-ezetimibe treatment. Journal of the Neurological Sciences, 2017, 383, 39-41.	0.6	3
34	Rare causes of early-onset dystonia-parkinsonism with cognitive impairment: a de novo PSEN-1 mutation. Neurogenetics, 2017, 18, 175-178.	1.4	23
35	Patient Affected by Beta-Propeller Protein-Associated Neurodegeneration: A Therapeutic Attempt with Iron Chelation Therapy. Frontiers in Neurology, 2017, 8, 385.	2.4	18
36	Growth Arrest Specific 6 Concentration is Increased in the Cerebrospinal Fluid of Patients with Alzheimerâ€™s Disease. Journal of Alzheimer's Disease, 2016, 55, 59-65.	2.6	41

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37	Recent advances in genetics of chorea. Current Opinion in Neurology, 2016, 29, 486-495.	3.6	25
38	Novel GNAL mutation with intra-familial clinical heterogeneity: Expanding the phenotype. Parkinsonism and Related Disorders, 2016, 23, 66-71.	2.2	35
39	A novel synonymous mutation in the MPZ gene causing an aberrant splicing pattern and Charcot-Marie-Tooth disease type 1b. Neuromuscular Disorders, 2016, 26, 516-520.	0.6	18
40	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
41	Inherited Isolated Dystonia in Children. Journal of Pediatric Neurology, 2015, 13, 174-179.	0.2	3
42	Adultâ€Onset Focal Chorea in Fahr's Disease Resulting From <scp>SLC</scp>20A2 Mutation: A Novel Phenotype. Movement Disorders Clinical Practice, 2015, 2, 79-80.	1.5	2
43	GTP cyclohydrolase 1 mutations and Parkinson's disease: New insights beyond DOPAâ€responsive dystonia. Movement Disorders, 2015, 30, 910-910.	3.9	1
44	A Missense Mutation in KCTD17 Causes Autosomal Dominant Myoclonus-Dystonia. American Journal of Human Genetics, 2015, 96, 938-947.	6.2	109
45	Identification of novel CSF biomarkers for neurodegeneration and their validation by a high-throughput multiplexed targeted proteomic assay. Molecular Neurodegeneration, 2015, 10, 64.	10.8	121
46	TheCACNA1BR1389H variant is not associated with myoclonus-dystonia in a large European multicentric cohort. Human Molecular Genetics, 2015, 24, 5326-5329.	2.9	28
47	Early onset frontotemporal dementia with psychiatric presentation due to the C9ORF72 hexanucleotide repeat expansion: a case report. BMC Neurology, 2014, 14, 228.	1.8	13
48	Predicting Cognitive Decline in ParkinsonÃ¢â‚¬â„¢s Disease: Can We Ask the Genes?. Frontiers in Neurology, 2014, 5, 224.	2.4	11
49	Peripheral nervous system involvement in Parkinson's disease: Evidence and controversies. Parkinsonism and Related Disorders, 2014, 20, 1329-1334.	2.2	64
50	Defining the Epsilonâ€Sarcoglycan (SGCE) Gene Phenotypic Signature in Myoclonusâ€Dystonia: A Reappraisal of Genetic Testing Criteria. Movement Disorders, 2013, 28, 787-794.	3.9	31
51	The syndrome of deafnessâ€dystonia: Clinical and genetic heterogeneity. Movement Disorders, 2013, 28, 795-803.	3.9	25
52	Movement Disorders in Adult Patients With Classical Galactosemia. Movement Disorders, 2013, 28, 804-810.	3.9	57
53	Evidence of Pre-Synaptic Dopaminergic Deficit in a Patient with a Novel Progranulin Mutation Presenting with Atypical Parkinsonismâ€. Journal of Alzheimer's Disease, 2013, 38, 747-752.	2.6	19
54	Immunity and inflammation in neurodegenerative diseases. American Journal of Neurodegenerative Disease, 2013, 2, 89-107.	0.1	83

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55	Phenotypic Heterogeneity of the GRN Asp22fs Mutation in a Large Italian Kindred. Journal of Alzheimer's Disease, 2011, 24, 253-259.	2.6	62
56	The Role of Osteopontin in Neurodegenerative Diseases. Journal of Alzheimer's Disease, 2011, 25, 179-185.	2.6	81
57	Cerebrospinal Fluid Biomarkers in Progranulin Mutations Carriers. Journal of Alzheimer's Disease, 2011, 27, 781-790.	2.6	45
58	Movement disorders in adult surviving patients with maple syrup urine disease. Movement Disorders, 2011, 26, 1324-1328.	3.9	46
59	Osteopontin is Increased in the Cerebrospinal Fluid of Patients with Alzheimer's Disease and Its Levels Correlate with Cognitive Decline. Journal of Alzheimer's Disease, 2010, 19, 1143-1148.	2.6	100
60	Atypical parkinsonism with apraxia and supranuclear gaze abnormalities in type 1 Gaucher disease. Expanding the spectrum: Case report and literature review. Movement Disorders, 2010, 25, 1506-1509.	3.9	21
61	Levodopaâ€induced belly dancer's dyskinesias in Parkinson's disease: Report of one case. Movement Disorders, 2010, 25, 1760-1762.	3.9	18
62	C2orf37 mutational spectrum in Woodhouseâ€“Sakati syndrome patients. Clinical Genetics, 2010, 78, 585-590.	2.0	41
63	Defective Fasâ€mediated Tâ€cell apoptosis predicts acute onset CIDP. Journal of the Peripheral Nervous System, 2009, 14, 101-106.	3.1	24
64	Complex movement disorders in primary antiphospholipid syndrome: A case report. Journal of the Neurological Sciences, 2009, 281, 101-103.	0.6	16
65	Progranulin plasma levels as potential biomarker for the identification of GRN deletion carriers. A case with atypical onset as clinical amnestic Mild Cognitive Impairment converted to Alzheimer's disease. Journal of the Neurological Sciences, 2009, 287, 291-293.	0.6	83
66	Movement disorders in metabolic diseases in adulthood. , 0, , 99-114.		0