Coro PaisÃ¡n-Ruiz

List of Publications by Year
in descending order

39 papers	2,839 citations	304743 22 h-index	315739 38 g-index
40 all docs	40 docs citations	40 times ranked	3871 citing authors

#	Article	IF	Citations
1	Characterization of PLA2G6 as a locus for dystoniaâ€parkinsonism. Annals of Neurology, 2009, 65, 19-23.	5.3	399
2	The Sac1 Domain of <scp>SYNJ</scp> 1 Identified Mutated in a Family with Earlyâ€Onset Progressive <scp>P</scp> arkinsonism with Generalized Seizures. Human Mutation, 2013, 34, 1200-1207.	2.5	302
3	Earlyâ€onset Lâ€dopaâ€responsive parkinsonism with pyramidal signs due to ATP13A2, PLA2G6, FBXO7 and spatacsin mutations. Movement Disorders, 2010, 25, 1791-1800.	3.9	287
4	DYT16, a novel young-onset dystonia-parkinsonism disorder: identification of a segregating mutation in the stress-response protein PRKRA. Lancet Neurology, The, 2008, 7, 207-215.	10.2	202
5	Defective FA2H leads to a novel form of neurodegeneration with brain iron accumulation (NBIA). Annals of Neurology, 2010, 68, 611-618.	5.3	202
6	The genetics of Parkinson's syndromes: a critical review. Current Opinion in Genetics and Development, 2009, 19, 254-265.	3.3	195
7	Widespread Lewy body and tau accumulation in childhood and adult onset dystonia-parkinsonism cases with PLA2G6 mutations. Neurobiology of Aging, 2012, 33, 814-823.	3.1	184
8	Mutation of FA2H underlies a complicated form of hereditary spastic paraplegia (SPG35). Human Mutation, 2010, 31, E1251-E1260.	2.5	174
9	LRRK2: Cause, Risk, and Mechanism. Journal of Parkinson's Disease, 2013, 3, 85-103.	2.8	128
10	The ACMSD gene, involved in tryptophan metabolism, is mutated in a family with cortical myoclonus, epilepsy, and parkinsonism. Journal of Molecular Medicine, 2013, 91, 1399-1406.	3.9	111
11	LRRK2gene variation and its contribution to Parkinson disease. Human Mutation, 2009, 30, 1153-1160.	2.5	81
12	A Clinical and Molecular Genetic Study of 50 Families with Autosomal Recessive Parkinsonism Revealed Known and Novel Gene Mutations. Molecular Neurobiology, 2018, 55, 3477-3489.	4.0	67
13	SCN4Apore mutation pathogenetically contributes to autosomal dominant essential tremor and may increase susceptibility to epilepsy. Human Molecular Genetics, 2015, 24, ddv410.	2.9	38
14	PTRHD1(C2orf79) mutations lead to autosomal-recessive intellectual disability and parkinsonism. Movement Disorders, 2017, 32, 287-291.	3.9	38
15	Identification of a large homozygous VPS13C deletion in a patient with earlyâ€onset Parkinsonism. Movement Disorders, 2018, 33, 1968-1970.	3.9	38
16	Identification of a LargeDNAJB2Deletion in a Family with Spinal Muscular Atrophy and Parkinsonism. Human Mutation, 2016, 37, 1180-1189.	2.5	36
17	Common pathogenic pathways in melanoma and Parkinson disease. Neurology, 2010, 75, 1653-1655.	1.1	34
18	SORT1 Mutation Resulting in Sortilin Deficiency and p75^{NTR} Upregulation in a Family With Essential Tremor. ASN Neuro, 2015, 7, 175909141559829.	2.7	28

#	Article	IF	Citations
19	Homozygosity mapping through whole genome analysis identifies a COL18A1 mutation in an Indian family presenting with an autosomal recessive neurological disorder. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2009, 150B, 993-997.	1.7	27
20	Whole-exome sequencing associates novel CSMD1 gene mutations with familial Parkinson disease. Neurology: Genetics, 2017, 3, e177.	1.9	27
21	Exome sequencing identifies GCDH (glutaryl-CoA dehydrogenase) mutations as a cause of a progressive form of early-onset generalized dystonia. Human Genetics, 2012, 131, 435-442.	3.8	23
22	PLA2G6-associated Dystonia-Parkinsonism: Case Report and Literature Review. Tremor and Other Hyperkinetic Movements, 2015, 5, 317.	2.0	22
23	The Use of Next-Generation Sequencing in Movement Disorders. Frontiers in Genetics, 2012, 3, 75.	2.3	21
24	Identification of COL6A2 mutations in progressive myoclonus epilepsy syndrome. Human Genetics, 2013, 132, 275-283.	3.8	21
25	A novel PUS7 mutation causes intellectual disability with autistic and aggressive behaviors. Neurology: Genetics, 2019, 5, e356.	1.9	18
26	Pla2g6 Deficiency in Zebrafish Leads to Dopaminergic Cell Death, Axonal Degeneration, Increased Î²-Synuclein Expression, and Defects in Brain Functions and Pathways. Molecular Neurobiology, 2018, 55, 6734-6754.	4.0	17
27	Parkinson's Disease and Low Frequency Alleles Found Together Throughout LRRK2. Annals of Human Genetics, 2009, 73, 391-403.	0.8	16
28	GIGYF2 mutation in late-onset Parkinsonâ€™s disease with cognitive impairment. Journal of Human Genetics, 2015, 60, 637-640.	2.3	16
29	Support for â€œDisease-Onlyâ€•Genotypes and Excess of Homozygosity at the CYTH4 Primate-Specific GTTT-Repeat in Schizophrenia. Genetic Testing and Molecular Biomarkers, 2017, 21, 485-490.	0.7	15
30	A Novel p.Glu298Lys Mutation in the ACMSD Gene in Sporadic Parkinsonâ€™s Disease. Journal of Parkinson's Disease, 2017, 7, 459-463.	2.8	15
31	Whole genome sequencing identifies a novel homozygous exon deletion in the NT5C2 gene in a family with intellectual disability and spastic paraplegia. Npj Genomic Medicine, 2017, 2, .	3.8	10
32	Genetic screening in two Iranian families with early-onset Alzheimer's disease identified a novel PSEN1 mutation. Neurobiology of Aging, 2018, 62, 244.e15-244.e17.	3.1	9
33	TRAP1 chaperone protein mutations and autoinflammation. Life Science Alliance, 2020, 3, e201900376.	2.8	9
34	Phenotypic and genotypic characterization of families with complex intellectual disability identified pathogenic genetic variations in known and novel disease genes. Scientific Reports, 2020, 10, 968.	3.3	8
35	<scp>ANXA1</scp> with Antiâ€inflammatory Properties Might Contribute to Parkinsonism. Annals of Neurology, 2021, 90, 319-323.	5.3	7
36	Molecular characterization of PRKN structural variations identified through wholeâ€genome sequencing. Molecular Genetics & Genomic Medicine, 2018, 6, 1243-1248.	1.2	6

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37	CANVAS with cerebellar/sensory/vestibular dysfunction from RFC1 intronic pentanucleotide expansion. Brain, 2020, 143, 386-390.	7.6	5
38	Variable phenotypic expression in families with early-onset Parkinsonism due to PRKN mutations. Journal of Neurology, 2014, 261, 1223-1226.	3.6	3
39	First report of pathogenic SGCE variants in Mexican patients with myoclonus dystonia: A five-year follow-up study. Parkinsonism and Related Disorders, 2020, 79, 117-120.	2.2	0