

Coro Paisán-Ruiz

List of Publications by Year in descending order

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Version: 2024-02-01

39
papers

2,839
citations

304743

22
h-index

315739

38
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40
all docs

40
docs citations

40
times ranked

3871
citing authors

#	ARTICLE	IF	CITATIONS
1	<scp><i>ANXA1</i></scp> with Anti-â€inflammatory Properties Might Contribute to Parkinsonism. <i>Annals of Neurology</i> , 2021, 90, 319-323.	5.3	7
2	First report of pathogenic SGCE variants in Mexican patients with myoclonus dystonia: A five-year follow-up study. <i>Parkinsonism and Related Disorders</i> , 2020, 79, 117-120.	2.2	0
3	CANVAS with cerebellar/sensory/vestibular dysfunction from RFC1 intronic pentanucleotide expansion. <i>Brain</i> , 2020, 143, 386-390.	7.6	5
4	Phenotypic and genotypic characterization of families with complex intellectual disability identified pathogenic genetic variations in known and novel disease genes. <i>Scientific Reports</i> , 2020, 10, 968.	3.3	8
5	TRAP1 chaperone protein mutations and autoinflammation. <i>Life Science Alliance</i> , 2020, 3, e201900376.	2.8	9
6	A novel <i>PUS7</i> mutation causes intellectual disability with autistic and aggressive behaviors. <i>Neurology: Genetics</i> , 2019, 5, e356.	1.9	18
7	Pla2g6 Deficiency in Zebrafish Leads to Dopaminergic Cell Death, Axonal Degeneration, Increased Î²-Synuclein Expression, and Defects in Brain Functions and Pathways. <i>Molecular Neurobiology</i> , 2018, 55, 6734-6754.	4.0	17
8	A Clinical and Molecular Genetic Study of 50 Families with Autosomal Recessive Parkinsonism Revealed Known and Novel Gene Mutations. <i>Molecular Neurobiology</i> , 2018, 55, 3477-3489.	4.0	67
9	Genetic screening in two Iranian families with early-onset Alzheimer's disease identified a novel PSEN1 mutation. <i>Neurobiology of Aging</i> , 2018, 62, 244.e15-244.e17.	3.1	9
10	Identification of a large homozygous <i>VPS13C</i> deletion in a patient with early-â€onset Parkinsonism. <i>Movement Disorders</i> , 2018, 33, 1968-1970.	3.9	38
11	Molecular characterization of <i>PRKN</i> structural variations identified through whole-â€genome sequencing. <i>Molecular Genetics & Genomic Medicine</i> , 2018, 6, 1243-1248.	1.2	6
12	Support for â€Disease-Only-â€ Genotypes and Excess of Homozygosity at the CYTH4 Primate-Specific GTTT-Repeat in Schizophrenia. <i>Genetic Testing and Molecular Biomarkers</i> , 2017, 21, 485-490.	0.7	15
13	Whole-exome sequencing associates novel <i>CSMD1</i> gene mutations with familial Parkinson disease. <i>Neurology: Genetics</i> , 2017, 3, e177.	1.9	27
14	Whole genome sequencing identifies a novel homozygous exon deletion in the NT5C2 gene in a family with intellectual disability and spastic paraplegia. <i>Npj Genomic Medicine</i> , 2017, 2, .	3.8	10
15	<i>PTRHD1</i> (C2orf79) mutations lead to autosomal-recessive intellectual disability and parkinsonism. <i>Movement Disorders</i> , 2017, 32, 287-291.	3.9	38
16	A Novel p.Glu298Lys Mutation in the ACMSD Gene in Sporadic Parkinson-â€™s Disease. <i>Journal of Parkinson's Disease</i> , 2017, 7, 459-463.	2.8	15
17	Identification of a LargeDNAJB2Deletion in a Family with Spinal Muscular Atrophy and Parkinsonism. <i>Human Mutation</i> , 2016, 37, 1180-1189.	2.5	36
18	<i>SORT1</i> Mutation Resulting in Sortilin Deficiency and p75^{NTR} Upregulation in a Family With Essential Tremor. <i>ASN Neuro</i> , 2015, 7, 175909141559829.	2.7	28

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19	GIGYF2 mutation in late-onset Parkinsonâ€™s disease with cognitive impairment. <i>Journal of Human Genetics</i> , 2015, 60, 637-640.	2.3	16
20	<i>SCN4A</i> pore mutation pathogenetically contributes to autosomal dominant essential tremor and may increase susceptibility to epilepsy. <i>Human Molecular Genetics</i> , 2015, 24, ddv410.	2.9	38
21	PLA2G6-associated Dystonia-Parkinsonism: Case Report and Literature Review. <i>Tremor and Other Hyperkinetic Movements</i> , 2015, 5, 317.	2.0	22
22	Variable phenotypic expression in families with early-onset Parkinsonism due to PRKN mutations. <i>Journal of Neurology</i> , 2014, 261, 1223-1226.	3.6	3
23	Identification of COL6A2 mutations in progressive myoclonus epilepsy syndrome. <i>Human Genetics</i> , 2013, 132, 275-283.	3.8	21
24	The Sac1 Domain of <i>SYNJ</i> 1 Identified Mutated in a Family with Earlyâ€œOnset Progressive <i>P</i> arkinsonism with Generalized Seizures. <i>Human Mutation</i> , 2013, 34, 1200-1207.	2.5	302
25	The ACMSD gene, involved in tryptophan metabolism, is mutated in a family with cortical myoclonus, epilepsy, and parkinsonism. <i>Journal of Molecular Medicine</i> , 2013, 91, 1399-1406.	3.9	111
26	LRRK2: Cause, Risk, and Mechanism. <i>Journal of Parkinson's Disease</i> , 2013, 3, 85-103.	2.8	128
27	The Use of Next-Generation Sequencing in Movement Disorders. <i>Frontiers in Genetics</i> , 2012, 3, 75.	2.3	21
28	Widespread Lewy body and tau accumulation in childhood and adult onset dystonia-parkinsonism cases with PLA2G6 mutations. <i>Neurobiology of Aging</i> , 2012, 33, 814-823.	3.1	184
29	Exome sequencing identifies GCDH (glutaryl-CoA dehydrogenase) mutations as a cause of a progressive form of early-onset generalized dystonia. <i>Human Genetics</i> , 2012, 131, 435-442.	3.8	23
30	Mutation of FA2H underlies a complicated form of hereditary spastic paraplegia (SPG35). <i>Human Mutation</i> , 2010, 31, E1251-E1260.	2.5	174
31	Defective <i>FA2H</i> leads to a novel form of neurodegeneration with brain iron accumulation (NBIA). <i>Annals of Neurology</i> , 2010, 68, 611-618.	5.3	202
32	Earlyâ€œOnset Lâ€œdopaâ€œresponsive parkinsonism with pyramidal signs due to <i>ATP13A2</i>, <i>PLA2G6</i>, <i>FBXO7</i> and <i>spatacsin</i> mutations. <i>Movement Disorders</i> , 2010, 25, 1791-1800.	3.9	287
33	Common pathogenic pathways in melanoma and Parkinson disease. <i>Neurology</i> , 2010, 75, 1653-1655.	1.1	34
34	Characterization of PLA2G6 as a locus for dystoniaâ€œparkinsonism. <i>Annals of Neurology</i> , 2009, 65, 19-23.	5.3	399
35	Homozygosity mapping through whole genome analysis identifies a <i>COL18A1</i> mutation in an Indian family presenting with an autosomal recessive neurological disorder. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2009, 150B, 993-997.	1.7	27
36	<i>LRRK2</i> gene variation and its contribution to Parkinson disease. <i>Human Mutation</i> , 2009, 30, 1153-1160.	2.5	81

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37	Parkinson's Disease and Low Frequency Alleles Found Together Throughout <i>LRRK2</i>. Annals of Human Genetics, 2009, 73, 391-403.	0.8	16
38	The genetics of Parkinson's syndromes: a critical review. Current Opinion in Genetics and Development, 2009, 19, 254-265.	3.3	195
39	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