

Isabel Alonso

List of Publications by Year in descending order

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82
papers

2,238
citations

218677

26
h-index

243625

44
g-index

84
all docs

84
docs citations

84
times ranked

3488
citing authors

#	ARTICLE	IF	CITATIONS
1	Trinucleotide Repeats in 202 Families With Ataxia. Archives of Neurology, 2002, 59, 623.	4.5	158
2	Mutations in PNKP Cause Recessive Ataxia with Oculomotor Apraxia Type 4. American Journal of Human Genetics, 2015, 96, 474-479.	6.2	127
3	Proinflammatory and anti-inflammatory cytokines in the CSF of patients with Alzheimer's disease and their correlation with cognitive decline. Neurobiology of Aging, 2019, 76, 125-132.	3.1	121
4	Hereditary Ataxia and Spastic Paraplegia in Portugal. JAMA Neurology, 2013, 70, 746.	9.0	106
5	DJ-1 linked parkinsonism (PARK7) is associated with Lewy body pathology. Brain, 2016, 139, 1680-1687.	7.6	89
6	A survey of spinocerebellar ataxia in South Brazil - 66 new cases with Machado-Joseph disease, SCA7, SCA8, or unidentified disease-causing mutations. Journal of Neurology, 2001, 248, 870-876.	3.6	88
7	Alteration of ornithine metabolism leads to dominant and recessive hereditary spastic paraplegia. Brain, 2015, 138, 2191-2205.	7.6	88
8	High Germinal Instability of the (CTG) _n at the SCA8 Locus of Both Expanded and Normal Alleles. American Journal of Human Genetics, 2000, 66, 830-840.	6.2	79
9	Phenotypes of Spinocerebellar Ataxia Type 6 and Familial Hemiplegic Migraine Caused by a Unique CACNA1A Missense Mutation in Patients From a Large Family. Archives of Neurology, 2003, 60, 610.	4.5	77
10	EMQN best practice guidelines for the molecular genetic diagnosis of hereditary hemochromatosis (HH). European Journal of Human Genetics, 2016, 24, 479-495.	2.8	73
11	Large normal and reduced penetrance alleles in Huntington disease: instability in families and frequency at the laboratory, at the clinic and in the population. Clinical Genetics, 2010, 78, 381-387.	2.0	60
12	Massive sequencing of 70 genes reveals a myriad of missing genes or mechanisms to be uncovered in hereditary spastic paraplegias. European Journal of Human Genetics, 2017, 25, 1217-1228.	2.8	58
13	Rare and Common Variants Conferring Risk of Tooth Agenesis. Journal of Dental Research, 2018, 97, 515-522.	5.2	52
14	Autosomal Dominant Spastic Paraplegias. JAMA Neurology, 2013, 70, 481.	9.0	48
15	EMQN Best Practice Guidelines for molecular genetic testing of SCAs. European Journal of Human Genetics, 2010, 18, 1173-1176.	2.8	41
16	BDNF and CGRP interaction: Implications in migraine susceptibility. Cephalalgia, 2010, 30, 1375-1382.	3.9	41
17	Ancestral Origin of the ATTCT Repeat Expansion in Spinocerebellar Ataxia Type 10 (SCA10). PLoS ONE, 2009, 4, e4553.	2.5	40
18	Variants in RBP4 and AR genes modulate age at onset in familial amyloid polyneuropathy (FAP) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 62	2.8	40

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19	Reduced penetrance of intermediate size alleles in spinocerebellar ataxia type 10. <i>Neurology</i> , 2006, 66, 1602-1604.	1.1	38
20	Identification of Genetic Risk Factors for Maxillary Lateral Incisor Agenesis. <i>Journal of Dental Research</i> , 2014, 93, 452-458.	5.2	36
21	A novel H101Q mutation causes PKC δ loss in spinocerebellar ataxia type 14. <i>Journal of Human Genetics</i> , 2005, 50, 523-529.	2.3	32
22	Common origin of pure and interrupted repeat expansions in spinocerebellar ataxia type 2 (SCA2). <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2010, 153B, 524-531.	1.7	30
23	Shifting the CARASIL Paradigm. <i>Stroke</i> , 2015, 46, 1110-1112.	2.0	30
24	Rare Neurodegenerative Diseases: Clinical and Genetic Update. <i>Advances in Experimental Medicine and Biology</i> , 2017, 1031, 443-496.	1.6	30
25	Motor and cognitive deficits in the heterozygous leaner mouse, a Cav2.1 voltage-gated Ca ²⁺ channel mutant. <i>Neurobiology of Aging</i> , 2008, 29, 1733-1743.	3.1	27
26	Cerebellar Ataxia, Hemiplegic Migraine, and Related Phenotypes Due to a CACNA1A Missense Mutation. <i>JAMA Neurology</i> , 2013, 70, 235.	9.0	27
27	A recessive ataxia diagnosis algorithm for the next generation sequencing era. <i>Annals of Neurology</i> , 2017, 82, 892-899.	5.3	27
28	A novel R1347Q mutation in the predicted voltage sensor segment of the P/Q-type calcium-channel α 1A-subunit in a family with progressive cerebellar ataxia and hemiplegic migraine. <i>Clinical Genetics</i> , 2003, 65, 70-72.	2.0	26
29	Population stratification may bias analysis of PGC-1 α as a modifier of age at Huntington disease motor onset. <i>Human Genetics</i> , 2012, 131, 1833-1840.	3.8	26
30	Huntington disease and Huntington disease-like in a case series from Brazil. <i>Clinical Genetics</i> , 2014, 86, 373-377.	2.0	26
31	Alu elements mediate large SPG11 gene rearrangements: further spatacsin mutations. <i>Genetics in Medicine</i> , 2012, 14, 143-151.	2.4	25
32	Spinocerebellar ataxias in 114 Brazilian families: clinical and molecular findings. <i>Clinical Genetics</i> , 2006, 70, 173-176.	2.0	24
33	Genomic mechanisms underlying <i>PARK2</i> large deletions identified in a cohort of patients with PD. <i>Neurology: Genetics</i> , 2016, 2, e73.	1.9	22
34	Genetic analyses in a cohort of Portuguese pediatric patients with congenital hypothyroidism. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2019, 32, 1265-1273.	0.9	21
35	mtDNA copy number associated with age of onset in familial amyloid polyneuropathy. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2018, 89, 300-304.	1.9	20
36	Diagnostic yield of next-generation sequencing applied to neurological disorders. <i>Journal of Clinical Neuroscience</i> , 2019, 67, 14-18.	1.5	20

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37	A Trans-acting Factor May Modify Age at Onset in Familial Amyloid Polyneuropathy ATTRV30M in Portugal. <i>Molecular Neurobiology</i> , 2018, 55, 3676-3683.	4.0	19
38	Interaction between \hat{I}^3 -Aminobutyric Acid A Receptor Genes: New Evidence in Migraine Susceptibility. <i>PLoS ONE</i> , 2013, 8, e74087.	2.5	18
39	Genetic Contributors to Intergenerational CAG Repeat Instability in Huntingtonâ€™s Disease Knock-In Mice. <i>Genetics</i> , 2017, 205, 503-516.	2.9	17
40	Assessing Risk Factors for Migraine: Differences in Gender Transmission. <i>PLoS ONE</i> , 2012, 7, e50626.	2.5	16
41	Evidence of Syntaxin 1A Involvement in Migraine Susceptibility. <i>Archives of Neurology</i> , 2010, 67, 422-7.	4.5	15
42	A role for endothelin receptor type A in migraine without aura susceptibility? A study in Portuguese patients. <i>European Journal of Neurology</i> , 2011, 18, 649-655.	3.3	15
43	Prevalence of Huntington's disease gene CAG repeat alleles in sporadic amyotrophic lateral sclerosis patients. <i>Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders</i> , 2012, 13, 265-269.	2.1	15
44	Familial hemiplegic migraine due to L263V <i>SCN1A</i> mutation: Discordance for epilepsy between two kindreds from Douro Valley. <i>Cephalalgia</i> , 2014, 34, 1015-1020.	3.9	15
45	GNAO1 mutation presenting as dyskinetic cerebral palsy. <i>Neurological Sciences</i> , 2019, 40, 2213-2216.	1.9	13
46	Intergenerational instability in Huntington disease: Extreme repeat changes among 134 transmissions. <i>Movement Disorders</i> , 2012, 27, 583-585.	3.9	12
47	Spinocerebellar ataxia type 10: common haplotype and disease progression rate in Peru and Brazil. <i>European Journal of Neurology</i> , 2017, 24, 892.	3.3	12
48	Large normal alleles of <i>ATXN2</i> decrease age at onset in transthyretin familial amyloid polyneuropathy Val30Met patients. <i>Annals of Neurology</i> , 2019, 85, 251-258.	5.3	12
49	Increasing involvement of CAPN1 variants in spastic ataxias and phenotype-genotype correlations. <i>Neurogenetics</i> , 2021, 22, 71-79.	1.4	11
50	Does DNA methylation in the promoter region of the ATXN3 gene modify age at onset in MJD (SCA3) patients?. <i>Clinical Genetics</i> , 2011, 79, 100-102.	2.0	10
51	Candidate glutamatergic and dopaminergic pathway gene variants do not influence Huntingtonâ€™s disease motor onset. <i>Neurogenetics</i> , 2013, 14, 173-179.	1.4	10
52	Clinical spectrum of C9orf72 expansion in a cohort of Huntingtonâ€™s disease phenocopies. <i>Neurological Sciences</i> , 2018, 39, 741-744.	1.9	10
53	<i>C1QA</i> and <i>C1QC</i> modify ageâ€‘atâ€‘onset in familial amyloid polyneuropathy patients. <i>Annals of Clinical and Translational Neurology</i> , 2019, 6, 748-754.	3.7	10
54	Familial amyloid polyneuropathy in Portugal: New genes modulating ageâ€‘atâ€‘onset. <i>Annals of Clinical and Translational Neurology</i> , 2017, 4, 98-105.	3.7	9

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55	Mitochondrial Encephalopathy: First Portuguese Report of a VARS2 Causative Variant. <i>JIMD Reports</i> , 2018, 42, 113-119.	1.5	8
56	Gordon Holmes syndrome due to compound heterozygosity of two new PNPLA6 variants â€“ A diagnostic challenge. <i>ENeurologicalSci</i> , 2019, 14, 9-12.	1.3	8
57	Sensory neuronopathy in ataxia with oculomotor apraxia type 2. <i>Journal of the Neurological Sciences</i> , 2010, 298, 118-120.	0.6	7
58	A Portuguese rapid-onset dystonia-parkinsonism case with atypical features. <i>Neurological Sciences</i> , 2017, 38, 1713-1714.	1.9	7
59	Determinants of age at onset in a Portuguese cohort of autosomal dominant spastic paraplegia. <i>Journal of the Neurological Sciences</i> , 2020, 410, 116646.	0.6	7
60	Perry syndrome with progressive supranuclear palsy-like phenotype in a Portuguese family â€“ Long-term clinical follow-up. <i>Parkinsonism and Related Disorders</i> , 2021, 84, 74-76.	2.2	7
61	FXTAS is rare among Portuguese patients with movement disorders: FMR1 premutations may be associated with a wider spectrum of phenotypes. <i>Behavioral and Brain Functions</i> , 2011, 7, 19.	3.3	6
62	Prevalence of Huntington's disease gene CAG trinucleotide repeat alleles in patients with bipolar disorder. <i>Bipolar Disorders</i> , 2015, 17, 403-408.	1.9	6
63	Haplotype analysis of the 4p16.3 region in Portuguese families with Huntington's disease. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2015, 168, 135-143.	1.7	6
64	Going Deep into Synaptic Vesicle Machinery Genes and Migraine Susceptibility â€“ A Caseâ€Control Association Study. <i>Headache</i> , 2020, 60, 2152-2165.	3.9	6
65	A genetic interaction of NRXN2 with GABRE, SYT1 and CASK in migraine patients: a case-control study. <i>Journal of Headache and Pain</i> , 2021, 22, 57.	6.0	6
66	The spatial learning phenotype of heterozygous leaner mice is robust to systematic variation of the housing environment. <i>Comparative Medicine</i> , 2009, 59, 129-38.	1.0	6
67	A late-onset congenital myasthenic syndrome due to a heterozygous DOK7 mutation. <i>Neuromuscular Disorders</i> , 2020, 30, 331-335.	0.6	5
68	The Prevalence of Familial Hemiplegic Migraine With Cerebellar Ataxia and Spinocerebellar Ataxia Type 6 in Portugal. <i>Headache</i> , 2014, 54, 911-915.	3.9	4
69	Chromosome substitution strain assessment of a Huntingtonâ€™s disease modifier locus. <i>Mammalian Genome</i> , 2015, 26, 119-130.	2.2	4
70	Large-Scale Functional RNAi Screen in <i>C. elegans</i> Identifies TGF-Î² and Notch Signaling Pathways as Modifiers of <i>CACNA1A</i> . <i>ASN Neuro</i> , 2016, 8, 175909141663702.	2.7	4
71	Parkin truncating variants result in a loss-of-function phenotype. <i>Scientific Reports</i> , 2019, 9, 16150.	3.3	4
72	Beyond Val30Met transthyretin (TTR): variants associated with age-at-onset in hereditary ATTRv amyloidosis. <i>Amyloid: the International Journal of Experimental and Clinical Investigation: the Official Journal of the International Society of Amyloidosis</i> , 2021, 28, 100-106.	3.0	4

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73	Complex Movement Disorders in Ataxia with Oculomotor Apraxia Type 1: Beyond the Cerebellar Syndrome. Tremor and Other Hyperkinetic Movements, 2020, 10, 39.	2.0	4
74	Paternal transmission of subcortical band heterotopia through DCX somatic mosaicism. Seizure: the Journal of the British Epilepsy Association, 2015, 25, 62-64.	2.0	3
75	Novel MAG Variant Causes Cerebellar Ataxia with Oculomotor Apraxia: Molecular Basis and Expanded Clinical Phenotype. Journal of Clinical Medicine, 2020, 9, 1212.	2.4	3
76	Monozygotic twin sisters discordant for familial hemiplegic migraine. Journal of Headache and Pain, 2013, 14, 77.	6.0	2
77	Novel <i>APTX</i> Mutation in a Hispanic Subject Affected by Ataxia with Oculomotor Apraxia Type 1. Movement Disorders Clinical Practice, 2015, 2, 90-92.	1.5	2
78	Clinical and Genetic Characterization of Brazilian Patients with Ataxia and Oculomotor Apraxia. Movement Disorders, 2022, , .	3.9	2
79	Ataxia and Progressive Encephalopathy in a 4-Year-Old Girl. Laboratory Medicine, 2010, 41, 5-9.	1.2	1
80	PRKRAP1 pseudogene complicating the diagnosis of young-onset dystonia due to PRKRA gene disease-causing variants (DYT-PRKRA). Movement Disorders Clinical Practice, 0, , .	1.5	1
81	When Decrease A β 1-42 in CSF May Not Mean Alzheimer's Disease. Alzheimer Disease and Associated Disorders, 2018, 32, 359-363.	1.3	0
82	Rett-like Syndrome in a Pediatric Patient—A Challenging Diagnosis. Journal of Pediatric Neurology, 2021, 19, 113-115.	0.2	0