Isabel Alonso

List of Publications by Citations

81 papers	1,683 citations	23 h-index	38 g-index
84 ext. papers	2,015 ext. citations	5.1 avg, IF	4.11 L-index

#	Paper	IF	Citations
81	Trinucleotide repeats in 202 families with ataxia: a small expanded (CAG)n allele at the SCA17 locus. *Archives of Neurology*, **2002**, 59, 623-9		144
80	Mutations in PNKP cause recessive ataxia with oculomotor apraxia type 4. *American Journal of Human Genetics*, **2015**, 96, 474-9	11	90
79	Hereditary ataxia and spastic paraplegia in Portugal: a population-based prevalence study. *JAMA Neurology*, **2013**, 70, 746-55	17.2	82
78	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, 870-6	5.5	79
77	High germinal instability of the (CTG)n at the SCA8 locus of both expanded and normal alleles. *American Journal of Human Genetics*, **2000**, 66, 830-40	11	75
76	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		68
75	Alteration of ornithine metabolism leads to dominant and recessive hereditary spastic paraplegia. *Brain*, **2015**, 138, 2191-205	11.2	64
74	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	5.6	64
73	DJ-1 linked parkinsonism (PARK7) is associated with Lewy body pathology. *Brain*, **2016**, 139, 1680-7	11.2	63
72	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, 381-7	4	48
71	EMQN best practice guidelines for the molecular genetic diagnosis of hereditary hemochromatosis (HH). *European Journal of Human Genetics*, **2016**, 24, 479-95	5.3	46
70	Autosomal dominant spastic paraplegias: a review of 89 families resulting from a portuguese survey. *JAMA Neurology*, **2013**, 70, 481-7	17.2	41
69	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	5.3	38
68	BDNF and CGRP interaction: implications in migraine susceptibility. *Cephalalgia*, **2010**, 30, 1375-82	6.1	38
67	Variants in RBP4 and AR genes modulate age at onset in familial amyloid polyneuropathy (FAP ATTRV30M). *European Journal of Human Genetics*, **2016**, 24, 756-60	5.3	34
66	Ancestral origin of the ATTCT repeat expansion in spinocerebellar ataxia type 10 (SCA10). *PLoS ONE*, **2009**, 4, e4553	3.7	32
65	A novel H101Q mutation causes PKCgamma loss in spinocerebellar ataxia type 14. *Journal of Human Genetics*, **2005**, 50, 523-529	4.3	30

64	EMQN Best Practice Guidelines for molecular genetic testing of SCAs. *European Journal of Human Genetics*, **2010**, 18, 1173-6	5.3	29
63	Reduced penetrance of intermediate size alleles in spinocerebellar ataxia type 10. *Neurology*, **2006**, 66, 1602-4	6.5	26
62	Population stratification may bias analysis of PGC-1 as a modifier of age at Huntington disease motor onset. *Human Genetics*, **2012**, 131, 1833-40	6.3	25
61	Shifting the CARASIL paradigm: report of a non-Asian family and literature review. *Stroke*, **2015**, 46, 1110-7	6.7	24
60	Motor and cognitive deficits in the heterozygous leaner mouse, a Cav2.1 voltage-gated Ca2+ channel mutant. *Neurobiology of Aging*, **2008**, 29, 1733-43	5.6	24
59	Identification of genetic risk factors for maxillary lateral incisor agenesis. *Journal of Dental Research*, **2014**, 93, 452-8	8.1	23
58	Spinocerebellar ataxias in 114 Brazilian families: clinical and molecular findings. *Clinical Genetics*, **2006**, 70, 173-6	4	23
57	A novel R1347Q mutation in the predicted voltage sensor segment of the P/Q-type calcium-channel alpha-subunit in a family with progressive cerebellar ataxia and hemiplegic migraine. *Clinical Genetics*, **2004**, 65, 70-2	4	23
56	Cerebellar ataxia, hemiplegic migraine, and related phenotypes due to a CACNA1A missense mutation: 12-year follow-up of a large Portuguese family. *JAMA Neurology*, **2013**, 70, 235-40	17.2	22
55	Huntington disease and Huntington disease-like in a case series from Brazil. *Clinical Genetics*, **2014**, 86, 373-7	4	22
54	Common origin of pure and interrupted repeat expansions in spinocerebellar ataxia type 2 (SCA2). *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2010**, 153B, 524-531	3.5	22
53	A recessive ataxia diagnosis algorithm for the next generation sequencing era. *Annals of Neurology*, **2017**, 82, 892-899	9.4	20
52	Rare and Common Variants Conferring Risk of Tooth Agenesis. *Journal of Dental Research*, **2018**, 97, 5158522	8.1	19
51	Alu elements mediate large SPG11 gene rearrangements: further spatacsin mutations. *Genetics in Medicine*, **2012**, 14, 143-51	8.1	19
50	A Trans-acting Factor May Modify Age at Onset in Familial Amyloid Polyneuropathy ATTRV30M in Portugal. *Molecular Neurobiology*, **2018**, 55, 3676-3683	6.2	15
49	mtDNA copy number associated with age of onset in familial amyloid polyneuropathy. *Journal of Neurology, Neurosurgery and Psychiatry*, **2018**, 89, 300-304	5.5	15
48	Genomic mechanisms underlying PARK2 large deletions identified in a cohort of patients with PD. *Neurology: Genetics*, **2016**, 2, e73	3.8	14
47	Assessing risk factors for migraine: differences in gender transmission. *PLoS ONE*, **2012**, 7, e50626	3.7	14

46	Interaction between γ-aminobutyric acid A receptor genes: new evidence in migraine susceptibility. *PLoS ONE*, **2013**, 8, e74087	3.7	14
45	Genetic Contributors to Intergenerational CAG Repeat Instability in Huntington's Disease Knock-In Mice. *Genetics*, **2017**, 205, 503-516	4	13
44	Diagnostic yield of next-generation sequencing applied to neurological disorders. *Journal of Clinical Neuroscience*, **2019**, 67, 14-18	2.2	13
43	Rare Neurodegenerative Diseases: Clinical and Genetic Update. *Advances in Experimental Medicine and Biology*, **2017**, 1031, 443-496	3.6	13
42	Familial hemiplegic migraine due to L263V SCN1A mutation: discordance for epilepsy between two kindreds from Douro Valley. *Cephalalgia*, **2014**, 34, 1015-20	6.1	13
41	Prevalence of Huntington's disease gene CAG repeat alleles in sporadic amyotrophic lateral sclerosis patients. *Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders*, **2012**, 13, 265-9		13
40	Genetic analyses in a cohort of Portuguese pediatric patients with congenital hypothyroidism. *Journal of Pediatric Endocrinology and Metabolism*, **2019**, 32, 1265-1273	1.6	12
39	A role for endothelin receptor type A in migraine without aura susceptibility? A study in Portuguese patients. *European Journal of Neurology*, **2011**, 18, 649-55	6	12
38	Spinocerebellar ataxia type 10: common haplotype and disease progression rate in Peru and Brazil. *European Journal of Neurology*, **2017**, 24, 892-e36	6	11
37	Intergenerational instability in Huntington disease: extreme repeat changes among 134 transmissions. *Movement Disorders*, **2012**, 27, 583-5	7	10
36	Evidence of syntaxin 1A involvement in migraine susceptibility: a Portuguese study. *Archives of Neurology*, **2010**, 67, 422-7		10
35	Candidate glutamatergic and dopaminergic pathway gene variants do not influence Huntington's disease motor onset. *Neurogenetics*, **2013**, 14, 173-9	3	9
34	Does DNA methylation in the promoter region of the ATXN3 gene modify age at onset in MJD (SCA3) patients?. *Clinical Genetics*, **2011**, 79, 100-2	4	9
33	Large normal alleles of ATXN2 decrease age at onset in transthyretin familial amyloid polyneuropathy Val30Met patients. *Annals of Neurology*, **2019**, 85, 251-258	9.4	8
32	and modify age-at-onset in familial amyloid polyneuropathy patients. *Annals of Clinical and Translational Neurology*, **2019**, 6, 748-754	5.3	7
31	Familial amyloid polyneuropathy in Portugal: New genes modulating age-at-onset. *Annals of Clinical and Translational Neurology*, **2017**, 4, 98-105	5.3	6
30	Mitochondrial Encephalopathy: First Portuguese Report of a VARS2 Causative Variant. *JIMD Reports*, **2018**, 42, 113-119	1.9	6
29	Clinical spectrum of C9orf72 expansion in a cohort of Huntington's disease phenocopies. *Neurological Sciences*, **2018**, 39, 741-744	3.5	6

28	FXTAS is rare among Portuguese patients with movement disorders: FMR1 premutations may be associated with a wider spectrum of phenotypes. *Behavioral and Brain Functions*, **2011**, 7, 19	4.1	6
27	Sensory neuronopathy in ataxia with oculomotor apraxia type 2. *Journal of the Neurological Sciences*, **2010**, 298, 118-20	3.2	6
26	The spatial learning phenotype of heterozygous leaner mice is robust to systematic variation of the housing environment. *Comparative Medicine*, **2009**, 59, 129-38	1.6	6
25	Prevalence of Huntington's disease gene CAG trinucleotide repeat alleles in patients with bipolar disorder. *Bipolar Disorders*, **2015**, 17, 403-8	3.8	5
24	Gordon Holmes syndrome due to compound heterozygosity of two new variants - A diagnostic challenge. *ENeurologicalSci*, **2019**, 14, 9-12	2.1	5
23	A Portuguese rapid-onset dystonia-parkinsonism case with atypical features. *Neurological Sciences*, **2017**, 38, 1713-1714	3.5	4
22	GNAO1 mutation presenting as dyskinetic cerebral palsy. *Neurological Sciences*, **2019**, 40, 2213-2216	3.5	4
21	Chromosome substitution strain assessment of a Huntington's disease modifier locus. *Mammalian Genome*, **2015**, 26, 119-30	3.2	4
20	Large-Scale Functional RNAi Screen in C. elegans Identifies TGF-β and Notch Signaling Pathways as Modifiers of CACNA1A. *ASN Neuro*, **2016**, 8,	5.3	4
19	Haplotype analysis of the 4p16.3 region in Portuguese families with Huntington's disease. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2015**, 168B, 135-43	3.5	4
18	The prevalence of familial hemiplegic migraine with cerebellar ataxia and spinocerebellar ataxia type 6 in Portugal. *Headache*, **2014**, 54, 911-5	4.2	4
17	Determinants of age at onset in a Portuguese cohort of autosomal dominant spastic paraplegia. *Journal of the Neurological Sciences*, **2020**, 410, 116646	3.2	4
16	Going Deep into Synaptic Vesicle Machinery Genes and Migraine Susceptibility - A Case-Control Association Study. *Headache*, **2020**, 60, 2152-2165	4.2	4
15	Increasing involvement of CAPN1 variants in spastic ataxias and phenotype-genotype correlations. *Neurogenetics*, **2021**, 22, 71-79	3	4
14	A late-onset congenital myasthenic syndrome due to a heterozygous DOK7 mutation. *Neuromuscular Disorders*, **2020**, 30, 331-335	2.9	3
13	Monozygotic twin sisters discordant for familial hemiplegic migraine. *Journal of Headache and Pain*, **2013**, 14, 77	8.8	2
12	Paternal transmission of subcortical band heterotopia through DCX somatic mosaicism. *Seizure: the Journal of the British Epilepsy Association*, **2015**, 25, 62-4	3.2	2
11	Complex Movement Disorders in Ataxia with Oculomotor Apraxia Type 1: Beyond the Cerebellar Syndrome. *Tremor and Other Hyperkinetic Movements*, **2020**, 10, 39	2	2

10	Perry syndrome with progressive supranuclear palsy-like phenotype in a Portuguese family - Long-term clinical follow-up. *Parkinsonism and Related Disorders*, **2021**, 84, 74-76	3.6	2
9	A genetic interaction of NRXN2 with GABRE, SYT1 and CASK in migraine patients: a case-control study. *Journal of Headache and Pain*, **2021**, 22, 57	8.8	2
8	Novel Variant Causes Cerebellar Ataxia with Oculomotor Apraxia: Molecular Basis and Expanded Clinical Phenotype. *Journal of Clinical Medicine*, **2020**, 9,	5.1	1
7	Parkin truncating variants result in a loss-of-function phenotype. *Scientific Reports*, **2019**, 9, 16150	4.9	1
6	Novel Mutation in a Hispanic Subject Affected by Ataxia with Oculomotor Apraxia Type 1. *Movement Disorders Clinical Practice*, **2015**, 2, 90-92	2.2	1
5	Ataxia and Progressive Encephalopathy in a 4-Year-Old Girl. *Laboratory Medicine*, **2010**, 41, 5-9	1.6	1
4	Beyond Val30Met transthyretin (TTR): variants associated with age-at-onset in hereditary ATTRv amyloidosis. *Amyloid: the International Journal of Experimental and Clinical Investigation: the Official Journal of the International Society of Amyloidosis*, **2021**, 28, 100-106	2.7	1
3	PRKRAP1 pseudogene c omplicating the diagnosis of young-onset dystonia due to PRKRA gene disease-causing variants (DYT- PRKRA). *Movement Disorders Clinical Practice*,	2.2	0
2	When Decrease Aß1-42 in CSF May Not Mean Alzheimer's Disease: Insights From Two Case Reports With Early Onset Dementia. *Alzheimer Disease and Associated Disorders*, **2018**, 32, 359-363	2.5	
1	Rett-like Syndrome in a Pediatric Patient: A Challenging Diagnosis. *Journal of Pediatric Neurology*, **2021**, 19, 113-115	0.2	