Isabel Alonso

List of Publications by Year
in descending order

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	Clinical and Genetic Characterization of Brazilian Patients with Ataxia and Oculomotor Apraxia. Movement Disorders, 2022, , .	3.9	2
2	Rett-like Syndrome in a Pediatric Patientâ€"A Challenging Diagnosis. Journal of Pediatric Neurology, 2021, 19, 113-115.	0.2	0
3	Increasing involvement of CAPN1 variants in spastic ataxias and phenotype-genotype correlations. Neurogenetics, 2021, 22, 71-79.	1.4	11
4	Perry syndrome with progressive supranuclear palsy-like phenotype in a Portuguese family â€“ Long-term clinical follow-up. Parkinsonism and Related Disorders, 2021, 84, 74-76.	2.2	7
5	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.	6.0	6
6	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.	3.0	4
7	Determinants of age at onset in a Portuguese cohort of autosomal dominant spastic paraplegia. Journal of the Neurological Sciences, 2020, 410, 116646.	0.6	7
8	Going Deep into Synaptic Vesicle Machinery Genes and Migraine Susceptibility â€“ A Caseâ€Control Association Study. Headache, 2020, 60, 2152-2165.	3.9	6
9	A late-onset congenital myasthenic syndrome due to a heterozygous DOK7 mutation. Neuromuscular Disorders, 2020, 30, 331-335.	0.6	5
10	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
11	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
12	Diagnostic yield of next-generation sequencing applied to neurological disorders. Journal of Clinical Neuroscience, 2019, 67, 14-18.	1.5	20
13	Parkin truncating variants result in a loss-of-function phenotype. Scientific Reports, 2019, 9, 16150.	3.3	4
14	Genetic analyses in a cohort of Portuguese pediatric patients with congenital hypothyroidism. Journal of Pediatric Endocrinology and Metabolism, 2019, 32, 1265-1273.	0.9	21
15	GNAO1 mutation presenting as dyskinetic cerebral palsy. Neurological Sciences, 2019, 40, 2213-2216.	1.9	13
16	C1<scp>QA</scp> and C1<scp>QC</scp> modify ageâ€atâ€onset in familial amyloid polyneuropathy patients. Annals of Clinical and Translational Neurology, 2019, 6, 748-754.	3.7	10
17	Gordon Holmes syndrome due to compound heterozygosity of two new PNPLA6 variants â€“ A diagnostic challenge. ENeurologicalSci, 2019, 14, 9-12.	1.3	8
18	Large normal alleles of ATXN2 decrease age at onset in transthyretin familial amyloid polyneuropathy Val30Met patients. Annals of Neurology, 2019, 85, 251-258.	5.3	12

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19	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
20	A Trans-acting Factor May Modify Age at Onset in Familial Amyloid Polyneuropathy ATTRV30M in Portugal. Molecular Neurobiology, 2018, 55, 3676-3683.	4.0	19
21	Mitochondrial Encephalopathy: First Portuguese Report of a VARS2 Causative Variant. JIMD Reports, 2018, 42, 113-119.	1.5	8
22	Clinical spectrum of C9orf72 expansion in a cohort of Huntingtonâ€™s disease phenocopies. Neurological Sciences, 2018, 39, 741-744.	1.9	10
23	Rare and Common Variants Conferring Risk of Tooth Agenesis. Journal of Dental Research, 2018, 97, 515-522.	5.2	52
24	When Decrease AÎ²1-42 in CSF May Not Mean Alzheimer's Disease. Alzheimer Disease and Associated Disorders, 2018, 32, 359-363.	1.3	0
25	mtDNA copy number associated with age of onset in familial amyloid polyneuropathy. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 300-304.	1.9	20
26	Genetic Contributors to Intergenerational CAG Repeat Instability in Huntingtonâ€™s Disease Knock-In Mice. Genetics, 2017, 205, 503-516.	2.9	17
27	A Portuguese rapid-onset dystonia-parkinsonism case with atypical features. Neurological Sciences, 2017, 38, 1713-1714.	1.9	7
28	Spinocerebellar ataxia type 10: common haplotype and disease progression rate in Peru and Brazil. European Journal of Neurology, 2017, 24, 892.	3.3	12
29	Familial amyloid polyneuropathy in Portugal: New genes modulating ageâ€•atâ€•onset. Annals of Clinical and Translational Neurology, 2017, 4, 98-105.	3.7	9
30	A recessive ataxia diagnosis algorithm for the next generation sequencing era. Annals of Neurology, 2017, 82, 892-899.	5.3	27
31	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
32	Rare Neurodegenerative Diseases: Clinical and Genetic Update. Advances in Experimental Medicine and Biology, 2017, 1031, 443-496.	1.6	30
33	DJ-1 linked parkinsonism (PARK7) is associated with Lewy body pathology. Brain, 2016, 139, 1680-1687.	7.6	89
34	Large-Scale Functional RNAi Screen in C. elegans Identifies TGF-Î² and Notch Signaling Pathways as Modifiers of CACNA1A. ASN Neuro, 2016, 8, 175909141663702.	2.7	4
35	Genomic mechanisms underlying PARK2 large deletions identified in a cohort of patients with PD. Neurology: Genetics, 2016, 2, e73.	1.9	22
36	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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37	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
38	Novel <scp>APTX</scp> Mutation in a Hispanic Subject Affected by Ataxia with Oculomotor Apraxia Type 1. Movement Disorders Clinical Practice, 2015, 2, 90-92.	1.5	2
39	Prevalence of Huntington's disease gene CAG trinucleotide repeat alleles in patients with bipolar disorder. Bipolar Disorders, 2015, 17, 403-408.	1.9	6
40	Haplotype analysis of the 4p16.3 region in Portuguese families with Huntington's disease. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2015, 168, 135-143.	1.7	6
41	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
42	Mutations in PNKP Cause Recessive Ataxia with Oculomotor Apraxia Type 4. American Journal of Human Genetics, 2015, 96, 474-479.	6.2	127
43	Alteration of ornithine metabolism leads to dominant and recessive hereditary spastic paraplegia. Brain, 2015, 138, 2191-2205.	7.6	88
44	Shifting the CARASIL Paradigm. Stroke, 2015, 46, 1110-1112.	2.0	30
45	Chromosome substitution strain assessment of a Huntingtonâ€™s disease modifier locus. Mammalian Genome, 2015, 26, 119-130.	2.2	4
46	The Prevalence of Familial Hemiplegic Migraine With Cerebellar Ataxia and Spinocerebellar Ataxia Type 6 in <scp>P</scp>ortugal. Headache, 2014, 54, 911-915.	3.9	4
47	Familial hemiplegic migraine due to L263V SCN1A mutation: Discordance for epilepsy between two kindreds from Douro Valley. Cephalalgia, 2014, 34, 1015-1020.	3.9	15
48	Huntington disease and Huntington diseaseâ€like in a case series from Brazil. Clinical Genetics, 2014, 86, 373-377.	2.0	26
49	Identification of Genetic Risk Factors for Maxillary Lateral Incisor Agenesis. Journal of Dental Research, 2014, 93, 452-458.	5.2	36
50	Hereditary Ataxia and Spastic Paraplegia in Portugal. JAMA Neurology, 2013, 70, 746.	9.0	106
51	Monozygotic twin sisters discordant for familial hemiplegic migraine. Journal of Headache and Pain, 2013, 14, 77.	6.0	2
52	Candidate glutamatergic and dopaminergic pathway gene variants do not influence Huntingtonâ€™s disease motor onset. Neurogenetics, 2013, 14, 173-179.	1.4	10
53	Cerebellar Ataxia, Hemiplegic Migraine, and Related Phenotypes Due to a CACNA1A Missense Mutation. JAMA Neurology, 2013, 70, 235.	9.0	27
54	Autosomal Dominant Spastic Paraplegias. JAMA Neurology, 2013, 70, 481.	9.0	48

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55	Interaction between Î³-Aminobutyric Acid A Receptor Genes: New Evidence in Migraine Susceptibility. PLoS ONE, 2013, 8, e74087.	2.5	18
56	Alu elements mediate large SPG11 gene rearrangements: further spatacsin mutations. Genetics in Medicine, 2012, 14, 143-151.	2.4	25
57	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, 265-269.	2.1	15
58	Population stratification may bias analysis of PGC-1Î± as a modifier of age at Huntington disease motor onset. Human Genetics, 2012, 131, 1833-1840.	3.8	26
59	Assessing Risk Factors for Migraine: Differences in Gender Transmission. PLoS ONE, 2012, 7, e50626.	2.5	16
60	Intergenerational instability in Huntington disease: Extreme repeat changes among 134 transmissions. Movement Disorders, 2012, 27, 583-585.	3.9	12
61	Does DNA methylation in the promoter region of the ATXN3 gene modify age at onset in MJD (SCA3) patients?. Clinical Genetics, 2011, 79, 100-102.	2.0	10
62	A role for endothelin receptor type A in migraine without aura susceptibility? A study in Portuguese patients. European Journal of Neurology, 2011, 18, 649-655.	3.3	15
63	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.	3.3	6
64	Evidence of Syntaxin 1A Involvement in Migraine Susceptibility. Archives of Neurology, 2010, 67, 422-7.	4.5	15
65	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.	1.7	30
66	EMQN Best Practice Guidelines for molecular genetic testing of SCAs. European Journal of Human Genetics, 2010, 18, 1173-1176.	2.8	41
67	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
68	Ataxia and Progressive Encephalopathy in a 4-Year-Old Girl. Laboratory Medicine, 2010, 41, 5-9.	1.2	1
69	Sensory neuronopathy in ataxia with oculomotor apraxia type 2. Journal of the Neurological Sciences, 2010, 298, 118-120.	0.6	7
70	BDNF and CGRP interaction: Implications in migraine susceptibility. Cephalalgia, 2010, 30, 1375-1382.	3.9	41
71	Ancestral Origin of the ATTCT Repeat Expansion in Spinocerebellar Ataxia Type 10 (SCA10). PLoS ONE, 2009, 4, e4553.	2.5	40
72	The spatial learning phenotype of heterozygous leaner mice is robust to systematic variation of the housing environment. Comparative Medicine, 2009, 59, 129-38.	1.0	6

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73	Motor and cognitive deficits in the heterozygous leaner mouse, a Cav2.1 voltage-gated Ca2+ channel mutant. Neurobiology of Aging, 2008, 29, 1733-1743.	3.1	27
74	Spinocerebellar ataxias in 114 Brazilian families: clinical and molecular findings. Clinical Genetics, 2006, 70, 173-176.	2.0	24
75	Reduced penetrance of intermediate size alleles in spinocerebellar ataxia type 10. Neurology, 2006, 66, 1602-1604.	1.1	38
76	A novel H101Q mutation causes PKCÎ³ loss in spinocerebellar ataxia type 14. Journal of Human Genetics, 2005, 50, 523-529.	2.3	32
77	A novel R1347Q mutation in the predicted voltage sensor segment of the P/Q-type calcium-channel Î±1A-subunit in a family with progressive cerebellar ataxia and hemiplegic migraine. Clinical Genetics, 2003, 65, 70-72.	2.0	26
78	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
79	Trinucleotide Repeats in 202 Families With Ataxia. Archives of Neurology, 2002, 59, 623.	4.5	158
80	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
81	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
82	PRKRAP1 pseudogene c omplicating the diagnosis of youngâ€onset dystonia due to PRKRA gene diseaseâ€causing variants (DYTâ€PRKRA). Movement Disorders Clinical Practice, 0, , .	1.5	1