Alda Sousa

List of Publications by Citations

43 papers	1,316 citations	22 h-index	36 g-index
43 ext. papers	1,448 ext. citations	4.8 avg, IF	3.65 L-index

#	Paper	IF	Citations
43	Genetic epidemiology of familial amyloidotic polyneuropathy (FAP)-type I in P□oa do Varzim and Vila do Conde (north of Portugal). *American Journal of Medical Genetics Part A*, **1995**, 60, 512-21		170
42	Familial amyloidotic polyneuropathy in Sweden: geographical distribution, age of onset, and prevalence. *Human Heredity*, **1993**, 43, 288-94	1.1	111
41	Susceptibility and modifier genes in Portuguese transthyretin V30M amyloid polyneuropathy: complexity in a single-gene disease. *Human Molecular Genetics*, **2005**, 14, 543-53	5.6	99
40	Familial amyloidotic polyneuropathy in Sweden: a pedigree analysis. *Journal of Medical Genetics*, **1993**, 30, 388-92	5.8	58
39	A study of 159 Portuguese patients with familial amyloidotic polyneuropathy (FAP) whose parents were both unaffected. *Journal of Medical Genetics*, **1994**, 31, 293-9	5.8	58
38	Inherited and acquired risk factors and their combined effects in pediatric stroke. *Pediatric Neurology*, **2003**, 28, 134-8	2.9	55
37	Usefulness of labial salivary gland biopsy in familial amyloid polyneuropathy Portuguese type. *Amyloid: the International Journal of Experimental and Clinical Investigation: the Official Journal of the International Society of Amyloidosis*, **2009**, 16, 232-8	2.7	51
36	Haplotypes and DNA sequence variation within and surrounding the transthyretin gene: genotype-phenotype correlations in familial amyloid polyneuropathy (V30M) in Portugal and Sweden. *European Journal of Human Genetics*, **2004**, 12, 225-37	5.3	49
35	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
34	Overcoming artefact: anticipation in 284 Portuguese kindreds with familial amyloid polyneuropathy (FAP) ATTRV30M. *Journal of Neurology, Neurosurgery and Psychiatry*, **2014**, 85, 326-30	5.5	47
33	Familial ATTR amyloidosis: microalbuminuria as a predictor of symptomatic disease and clinical nephropathy. *Nephrology Dialysis Transplantation*, **2003**, 18, 532-8	4.3	42
32	End-stage renal disease and dialysis in hereditary amyloidosis TTR V30M: presentation, survival and prognostic factors. *Amyloid: the International Journal of Experimental and Clinical Investigation: the Official Journal of the International Society of Amyloidosis*, **2004**, 11, 27-37	2.7	40
31	BDNF and CGRP interaction: implications in migraine susceptibility. *Cephalalgia*, **2010**, 30, 1375-82	6.1	38
30	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
29	Vitreous involvement in familial amyloidotic neuropathy: a genealogical and genetic study. *Clinical Genetics*, **1991**, 40, 452-60	4	34
28	Population genetics of wild-type CAG repeats in the Machado-Joseph disease gene in Portugal. *Human Heredity*, **2005**, 60, 156-63	1.1	33
27	Genetic anticipation in Portuguese kindreds with familial amyloidotic polyneuropathy is unlikely to be caused by triplet repeat expansions. *Human Genetics*, **1999**, 104, 480-5	6.3	29

26	The CAG repeat at the Huntington disease gene in the Portuguese population: insights into its dynamics and to the origin of the mutation. *Journal of Human Genetics*, **2006**, 51, 189-195	4.3	24
25	Study of three intragenic polymorphisms in the Machado-Joseph disease gene (MJD1) in relation to genetic instability of the (CAG)n tract. *European Journal of Human Genetics*, **1999**, 7, 147-56	5.3	24
24	Identification of genetic risk factors for maxillary lateral incisor agenesis. *Journal of Dental Research*, **2014**, 93, 452-8	8.1	23
23	Familial clustering of migraine: further evidence from a Portuguese study. *Headache*, **2009**, 49, 404-11	4.2	23
22	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
21	Familial aggregation of maxillary lateral incisor agenesis. *Journal of Dental Research*, **2010**, 89, 621-5	8.1	21
20	Role of the disease in the psychological impact of pre-symptomatic testing for SCA2 and FAP ATTRV30M: Experience with the disease, kinship and gender of the transmitting parent. *Journal of Genetic Counseling*, **2009**, 18, 483-93	2.5	20
19	The C677T polymorphism in MTHFR is not associated with migraine in Portugal. *Disease Markers*, **2008**, 25, 107-13	3.2	19
18	Molecular diagnosis of Huntington disease in Portugal: implications for genetic counselling and clinical practice. *European Journal of Human Genetics*, **2003**, 11, 872-8	5.3	16
17	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
16	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
15	Assessing risk factors for migraine: differences in gender transmission. *PLoS ONE*, **2012**, 7, e50626	3.7	14
14	Interaction between □aminobutyric acid A receptor genes: new evidence in migraine susceptibility. *PLoS ONE*, **2013**, 8, e74087	3.7	14
13	Psychological follow-up of presymptomatic genetic testing for spinocerebellar ataxia type 2 (SCA2) in Cuba. *Journal of Genetic Counseling*, **2007**, 16, 469-79	2.5	13
12	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
11	Evidence of syntaxin 1A involvement in migraine susceptibility: a Portuguese study. *Archives of Neurology*, **2010**, 67, 422-7		10
10	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
9	and modify age-at-onset in familial amyloid polyneuropathy patients. *Annals of Clinical and Translational Neurology*, **2019**, 6, 748-754	5.3	7

8	Familial amyloid polyneuropathy in Portugal: New genes modulating age-at-onset. *Annals of Clinical and Translational Neurology*, **2017**, 4, 98-105	5.3	6
7	Craniofacial repercussions in maxillary lateral incisors agenesis. *International Orthodontics*, **2011**, 9, 274-85	0.9	5
6	Going Deep into Synaptic Vesicle Machinery Genes and Migraine Susceptibility - A Case-Control Association Study. *Headache*, **2020**, 60, 2152-2165	4.2	4
5	The hidden story behind gender differences in familial amyloid polyneuropathy (FAP) ATTRV30M. *Orphanet Journal of Rare Diseases*, **2015**, 10, O4	4.2	2
4	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
3	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
2	A review of migraine genetics: gathering genomic and transcriptomic factors. *Human Genetics*, **2021** , 141, 1	6.3	0
1	A High Methylation Level of a Novel 284 bp CpG Island in the RAMP1 Gene Promoter Is Potentially Associated with Migraine in Women. *Brain Sciences*, **2022**, 12, 526	3.4	