

# Alda Sousa

## List of Publications by Year in Descending Order

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**Version:** 2024-04-27

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

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	A High Methylation Level of a Novel 284 bp CpG Island in the RAMP1 Gene Promoter Is Potentially Associated with Migraine in Women. <i>Brain Sciences</i> , <b>2022</b> , 12, 526	3.4	
42	A review of migraine genetics: gathering genomic and transcriptomic factors. <i>Human Genetics</i> , <b>2021</b> , 141, 1	6.3	0
41	A genetic interaction of NRXN2 with GABRE, SYT1 and CASK in migraine patients: a case-control study. <i>Journal of Headache and Pain</i> , <b>2021</b> , 22, 57	8.8	2
40	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> , <b>2021</b> , 28, 100-106	2.7	1
39	Going Deep into Synaptic Vesicle Machinery Genes and Migraine Susceptibility - A Case-Control Association Study. <i>Headache</i> , <b>2020</b> , 60, 2152-2165	4.2	4
38	and modify age-at-onset in familial amyloid polyneuropathy patients. <i>Annals of Clinical and Translational Neurology</i> , <b>2019</b> , 6, 748-754	5.3	7
37	Large normal alleles of ATXN2 decrease age at onset in transthyretin familial amyloid polyneuropathy Val30Met patients. <i>Annals of Neurology</i> , <b>2019</b> , 85, 251-258	9.4	8
36	A Trans-acting Factor May Modify Age at Onset in Familial Amyloid Polyneuropathy ATTRV30M in Portugal. <i>Molecular Neurobiology</i> , <b>2018</b> , 55, 3676-3683	6.2	15
35	mtDNA copy number associated with age of onset in familial amyloid polyneuropathy. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , <b>2018</b> , 89, 300-304	5.5	15
34	Familial amyloid polyneuropathy in Portugal: New genes modulating age-at-onset. <i>Annals of Clinical and Translational Neurology</i> , <b>2017</b> , 4, 98-105	5.3	6
33	Variants in RBP4 and AR genes modulate age at onset in familial amyloid polyneuropathy (FAP ATTRV30M). <i>European Journal of Human Genetics</i> , <b>2016</b> , 24, 756-60	5.3	34
32	The hidden story behind gender differences in familial amyloid polyneuropathy (FAP) ATTRV30M. <i>Orphanet Journal of Rare Diseases</i> , <b>2015</b> , 10, O4	4.2	2
31	Overcoming artefact: anticipation in 284 Portuguese kindreds with familial amyloid polyneuropathy (FAP) ATTRV30M. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , <b>2014</b> , 85, 326-30	5.5	47
30	Identification of genetic risk factors for maxillary lateral incisor agenesis. <i>Journal of Dental Research</i> , <b>2014</b> , 93, 452-8	8.1	23
29	Cerebellar ataxia, hemiplegic migraine, and related phenotypes due to a CACNA1A missense mutation: 12-year follow-up of a large Portuguese family. <i>JAMA Neurology</i> , <b>2013</b> , 70, 235-40	17.2	22
28	Interaction between GABA <sub>A</sub> receptor genes: new evidence in migraine susceptibility. <i>PLoS ONE</i> , <b>2013</b> , 8, e74087	3.7	14
27	Assessing risk factors for migraine: differences in gender transmission. <i>PLoS ONE</i> , <b>2012</b> , 7, e50626	3.7	14

26	A role for endothelin receptor type A in migraine without aura susceptibility? A study in Portuguese patients. <i>European Journal of Neurology</i> , <b>2011</b> , 18, 649-55	6	12
25	Craniofacial repercussions in maxillary lateral incisors agenesis. <i>International Orthodontics</i> , <b>2011</b> , 9, 274-85	9	5
24	Large normal and reduced penetrance alleles in Huntington disease: instability in families and frequency at the laboratory, at the clinic and in the population. <i>Clinical Genetics</i> , <b>2010</b> , 78, 381-7	4	48
23	Familial aggregation of maxillary lateral incisor agenesis. <i>Journal of Dental Research</i> , <b>2010</b> , 89, 621-5	8.1	21
22	BDNF and CGRP interaction: implications in migraine susceptibility. <i>Cephalalgia</i> , <b>2010</b> , 30, 1375-82	6.1	38
21	Evidence of syntaxin 1A involvement in migraine susceptibility: a Portuguese study. <i>Archives of Neurology</i> , <b>2010</b> , 67, 422-7		10
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. <i>Journal of Genetic Counseling</i> , <b>2009</b> , 18, 483-93	2.5	20
19	Familial clustering of migraine: further evidence from a Portuguese study. <i>Headache</i> , <b>2009</b> , 49, 404-11	4.2	23
18	Usefulness of labial salivary gland biopsy in familial amyloid polyneuropathy Portuguese type. <i>Amyloid: the International Journal of Experimental and Clinical Investigation: the Official Journal of the International Society of Amyloidosis</i> , <b>2009</b> , 16, 232-8	2.7	51
17	The C677T polymorphism in MTHFR is not associated with migraine in Portugal. <i>Disease Markers</i> , <b>2008</b> , 25, 107-13	3.2	19
16	Psychological follow-up of presymptomatic genetic testing for spinocerebellar ataxia type 2 (SCA2) in Cuba. <i>Journal of Genetic Counseling</i> , <b>2007</b> , 16, 469-79	2.5	13
15	The CAG repeat at the Huntington disease gene in the Portuguese population: insights into its dynamics and to the origin of the mutation. <i>Journal of Human Genetics</i> , <b>2006</b> , 51, 189-195	4.3	24
14	Susceptibility and modifier genes in Portuguese transthyretin V30M amyloid polyneuropathy: complexity in a single-gene disease. <i>Human Molecular Genetics</i> , <b>2005</b> , 14, 543-53	5.6	99
13	Population genetics of wild-type CAG repeats in the Machado-Joseph disease gene in Portugal. <i>Human Heredity</i> , <b>2005</b> , 60, 156-63	1.1	33
12	End-stage renal disease and dialysis in hereditary amyloidosis TTR V30M: presentation, survival and prognostic factors. <i>Amyloid: the International Journal of Experimental and Clinical Investigation: the Official Journal of the International Society of Amyloidosis</i> , <b>2004</b> , 11, 27-37	2.7	40
11	Haplotypes and DNA sequence variation within and surrounding the transthyretin gene: genotype-phenotype correlations in familial amyloid polyneuropathy (V30M) in Portugal and Sweden. <i>European Journal of Human Genetics</i> , <b>2004</b> , 12, 225-37	5.3	49
10	Familial ATTR amyloidosis: microalbuminuria as a predictor of symptomatic disease and clinical nephropathy. <i>Nephrology Dialysis Transplantation</i> , <b>2003</b> , 18, 532-8	4.3	42
9	Molecular diagnosis of Huntington disease in Portugal: implications for genetic counselling and clinical practice. <i>European Journal of Human Genetics</i> , <b>2003</b> , 11, 872-8	5.3	16

8	Inherited and acquired risk factors and their combined effects in pediatric stroke. <i>Pediatric Neurology</i> , <b>2003</b> , 28, 134-8	2.9	55
7	Study of three intragenic polymorphisms in the Machado-Joseph disease gene (MJD1) in relation to genetic instability of the (CAG) <sub>n</sub> tract. <i>European Journal of Human Genetics</i> , <b>1999</b> , 7, 147-56	5.3	24
6	Genetic anticipation in Portuguese kindreds with familial amyloidotic polyneuropathy is unlikely to be caused by triplet repeat expansions. <i>Human Genetics</i> , <b>1999</b> , 104, 480-5	6.3	29
5	Genetic epidemiology of familial amyloidotic polyneuropathy (FAP)-type I in Póvoa do Varzim and Vila do Conde (north of Portugal). <i>American Journal of Medical Genetics Part A</i> , <b>1995</b> , 60, 512-21		170
4	A study of 159 Portuguese patients with familial amyloidotic polyneuropathy (FAP) whose parents were both unaffected. <i>Journal of Medical Genetics</i> , <b>1994</b> , 31, 293-9	5.8	58
3	Familial amyloidotic polyneuropathy in Sweden: a pedigree analysis. <i>Journal of Medical Genetics</i> , <b>1993</b> , 30, 388-92	5.8	58
2	Familial amyloidotic polyneuropathy in Sweden: geographical distribution, age of onset, and prevalence. <i>Human Heredity</i> , <b>1993</b> , 43, 288-94	1.1	111
1	Vitreous involvement in familial amyloidotic neuropathy: a genealogical and genetic study. <i>Clinical Genetics</i> , <b>1991</b> , 40, 452-60	4	34