Jorge Sequeiros

List of Publications by Year in descending order

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231 papers	8,963 citations	38738 50 h-index	58576 82 g-index
243 all docs	243 docs citations	243 times ranked	8876 citing authors

#	Article	IF	Citations
1	Senataxin, the ortholog of a yeast RNA helicase, is mutant in ataxia-ocular apraxia 2. Nature Genetics, 2004, 36, 225-227.	21.4	454
2	The gene mutated in ataxia-ocular apraxia 1 encodes the new HIT/Zn-finger protein aprataxin. Nature Genetics, 2001, 29, 189-193.	21.4	424
3	A dominant-negative mutation in the TRESK potassium channel is linked to familial migraine with aura. Nature Medicine, 2010, 16, 1157-1160.	30.7	312
4	CAG repeat expansion in Huntington disease determines age at onset in a fully dominant fashion. Neurology, 2012, 78, 690-695.	1.1	303
5	Identification of genetic variants associated with Huntington's disease progression: a genome-wide association study. Lancet Neurology, The, 2017, 16, 701-711.	10.2	248
6	Ataxia with oculomotor apraxia type 2: clinical, biological and genotype/phenotype correlation study of a cohort of 90 patients. Brain, 2009, 132, 2688-2698.	7.6	218
7	Genetic epidemiology of familial amyloidotic polyneuropathy (FAP)-type I in PÃ³voa do Varzim and Vila do Conde (north of Portugal). American Journal of Medical Genetics Part A, 1995, 60, 512-521.	2.4	195
8	Trinucleotide Repeats in 202 Families With Ataxia. Archives of Neurology, 2002, 59, 623.	4.5	158
9	Frataxin knockin mouse. FEBS Letters, 2002, 512, 291-297.	2.8	155
10	Neurologic Findings in Machado-Joseph Disease. Archives of Neurology, 2001, 58, 899.	4.5	154
11	The interface between assisted reproductive technologies and genetics: technical, social, ethical and legal issues. European Journal of Human Genetics, 2006, 14, 588-645.	2.8	137
12	Mutations in PNKP Cause Recessive Ataxia with Oculomotor Apraxia Type 4. American Journal of Human Genetics, 2015, 96, 474-479.	6.2	127
13	Improvement in the Molecular Diagnosis of Machado-Joseph Disease. Archives of Neurology, 2001, 58, 1821.	4.5	121
14	Current issues in medically assisted reproduction and genetics in Europe: research, clinical practice, ethics, legal issues and policy. European Journal of Human Genetics, 2013, 21, S1-S21.	2.8	120
15	Ancestral Origins of the Machado-Joseph Disease Mutation: A Worldwide Haplotype Study. American Journal of Human Genetics, 2001, 68, 523-528.	6.2	118
16	Neuroferritinopathy: Missense mutation in FTL causing early-onset bilateral pallidal involvement. Neurology, 2005, 65, 603-605.	1.1	112
17	A Pentanucleotide ATTTC Repeat Insertion in the Non-coding Region of DAB1, Mapping to SCA37, Causes Spinocerebellar Ataxia. American Journal of Human Genetics, 2017, 101, 87-103.	6.2	112
18	Recessive Ataxia With Ocular Apraxia. Archives of Neurology, 2001, 58, 201.	4.5	109

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19	Frequency of spinocerebellar ataxia type 1, dentatorubropallidoluysian atrophy, and Machado-Joseph disease mutations in a large group of spinocerebellar ataxia patients. Neurology, 1996, 46, 214-218.	1.1	106
20	Hereditary Ataxia and Spastic Paraplegia in Portugal. JAMA Neurology, 2013, 70, 746.	9.0	106
21	Ataxia Rating Scalesâ€"Psychometric Profiles, Natural History and Their Application in Clinical Trials. Cerebellum, 2012, 11, 488-504.	2.5	101
22	Epidemiology and clinical aspects of Machado-Joseph disease. Advances in Neurology, 1993, 61, 139-53.	0.8	94
23	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
24	What is ideal genetic counselling? A survey of current international guidelines. European Journal of Human Genetics, 2008, 16, 445-452.	2.8	86
25	Massively parallel sequencing of ataxia genes after array-based enrichment. Human Mutation, 2010, 31, 494-499.	2.5	86
26	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
27	Stereotypies in Rett syndrome: Analysis of 83 patients with and without detected MECP2 mutations. Neurology, 2007, 68, 1183-1187.	1.1	78
28	â€˜Costa da Morteâ€™ ataxia is spinocerebellar ataxia 36: clinical and genetic characterization. Brain, 2012, 135, 1423-1435.	7.6	78
29	Homozygosity Mapping of Portuguese and Japanese Forms of Ataxia-Oculomotor Apraxia to 9p13, and Evidence for Genetic Heterogeneity. American Journal of Human Genetics, 2001, 68, 501-508.	6.2	77
30	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
31	First Mutation in the Voltage-Gated Na_v1.1 Subunit Gene SCN1A with Co-Occurring Familial Hemiplegic Migraine and Epilepsy. Cephalalgia, 2009, 29, 308-313.	3.9	77
32	Genetic testing and common disorders in a public health framework: how to assess relevance and possibilities. European Journal of Human Genetics, 2011, 19, S6-S44.	2.8	75
33	Consensus and controversies in best practices for molecular genetic testing of spinocerebellar ataxias. European Journal of Human Genetics, 2010, 18, 1188-1195.	2.8	74
34	Movement disorders in Rett syndrome: An analysis of 60 patients with detected MECP2 mutation and correlation with mutation type. Movement Disorders, 2008, 23, 1384-1390.	3.9	70
35	How communication of genetic information within the family is addressed in genetic counselling: a systematic review of research evidence. European Journal of Human Genetics, 2016, 24, 315-325.	2.8	66
36	Asian Origin for the Worldwide-Spread Mutational Event in Machado-Joseph Disease. Archives of Neurology, 2007, 64, 1502.	4.5	65

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37	Inherited and acquired risk factors and their combined effects in pediatric stroke. Pediatric Neurology, 2003, 28, 134-138.	2.1	64
38	Functional genomics and biochemical characterization of the C. elegans orthologue of the Machadoâ€Joseph disease protein ataxinâ€3. FASEB Journal, 2007, 21, 1126-1136.	0.5	62
39	CAG tract of MJD-1 may be prone to frameshifts causing polyalanine accumulation. Human Molecular Genetics, 2000, 9, 1957-1966.	2.9	61
40	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
41	Common SNP-Based Haplotype Analysis of the 4p16.3 Huntington Disease Gene Region. American Journal of Human Genetics, 2012, 90, 434-444.	6.2	60
42	Overcoming artefact: anticipation in 284 Portuguese kindreds with familial amyloid polyneuropathy (FAP) ATTRV30M. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, 326-330.	1.9	60
43	Somatic mosaicism in the central nervous system in spinocerebellar ataxia type 1 and machado-joseph disease. Annals of Neurology, 1996, 40, 199-206.	5.3	59
44	Nonsense mutation in TITF1 in a Portuguese family with benign hereditary chorea. Neurogenetics, 2005, 6, 209-215.	1.4	59
45	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
46	Analysis of SCA1, DRPLA, MJD, SCA2, and SCA6 CAG repeats in 48 portuguese ataxia families. American Journal of Medical Genetics Part A, 1998, 81, 134-138.	2.4	57
47	Epidemiology and population genetics of degenerative ataxias. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2012, 103, 227-251.	1.8	57
48	Current issues in medically assisted reproduction and genetics in Europe: research, clinical practice, ethics, legal issues and policy. Human Reproduction, 2014, 29, 1603-1609.	0.9	57
49	NEDD8: A new ataxin-3 interactor. Biochimica Et Biophysica Acta - Molecular Cell Research, 2007, 1773, 1619-1627.	4.1	55
50	Familial ATTR amyloidosis: microalbuminuria as a predictor of symptomatic disease and clinical nephropathy. Nephrology Dialysis Transplantation, 2003, 18, 532-538.	0.7	54
51	Autosomal dominant cerebellar ataxia: frequency analysis and clinical characterization of 45 families from Portugal. European Journal of Neurology, 2010, 17, 124-128.	3.3	54
52	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.	3.0	52
53	Genetic horoscopes: is it all in the genes? Points for regulatory control of direct-to-consumer genetic testing. European Journal of Human Genetics, 2009, 17, 857-859.	2.8	50
54	Autosomal Dominant Spastic Paraplegias. JAMA Neurology, 2013, 70, 481.	9.0	48

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55	Genetic interaction of CTLAâ€4 with HLAâ€DR15 in multiple sclerosis patients. Annals of Neurology, 2003, 54, 119-122.	5.3	46
56	Depressive Symptoms in Machado-Joseph Disease (SCA3) Patients and Their Relatives. Public Health Genomics, 2007, 10, 19-26.	1.0	46
57	Machado-Joseph disease in South Brazil: clinical and molecular characterization of kindreds. Acta Neurologica Scandinavica, 2001, 104, 224-231.	2.1	44
58	Population Genetics of Wild-Type CAG Repeats in the Machado-Joseph Disease Gene in Portugal. Human Heredity, 2005, 60, 156-163.	0.8	43
59	Evaluation of CSF neurotransmitters and folate in 25 patients with Rett disorder and effects of treatment. Brain and Development, 2009, 31, 46-51.	1.1	42
60	EMQN Best Practice Guidelines for molecular genetic testing of SCAs. European Journal of Human Genetics, 2010, 18, 1173-1176.	2.8	41
61	BDNF and CGRP interaction: Implications in migraine susceptibility. Cephalalgia, 2010, 30, 1375-1382.	3.9	41
62	Ancestral Origin of the ATTCT Repeat Expansion in Spinocerebellar Ataxia Type 10 (SCA10). PLoS ONE, 2009, 4, e4553.	2.5	40
63	Variants in RBP4 and AR genes modulate age at onset in familial amyloid polyneuropathy (FAP) Tj ETQq1 1 0.784314 rgBT /Overlock 10	2.8	40
64	Reduced penetrance of intermediate size alleles in spinocerebellar ataxia type 10. Neurology, 2006, 66, 1602-1604.	1.1	38
65	Onset in the seventh decade and lack of symptoms in heterozygotes for the TTRMet30 mutation in hereditary amyloid neuropathyâ€"type I (Portuguese, Andrade). American Journal of Medical Genetics Part A, 1987, 27, 345-357.	2.4	37
66	Portuguese families with dentatorubropallidoluysian atrophy (DRPLA) share a common haplotype of Asian origin. European Journal of Human Genetics, 2003, 11, 808-811.	2.8	37
67	(CAG) _n loci as genetic modifiers of age-at-onset in patients with Machado-Joseph disease from mainland China. Brain, 2016, 139, e41-e41.	7.6	37
68	Communication of Information about Genetic Risks: Putting Families at the Center. Family Process, 2018, 57, 836-846.	2.6	37
69	Identification of Genetic Risk Factors for Maxillary Lateral Incisor Agenesis. Journal of Dental Research, 2014, 93, 452-458.	5.2	36
70	Genetic Linkage Studies of Machado-Joseph Disease with Chromosome 14q STRPs in 16 Portuguese-Azorean Kindreds. Genomics, 1994, 21, 645-648.	2.9	34
71	Psychological aspects of pre-symptomatic testing for Machado-Joseph disease and familial amyloid polyneuropathy type I. Clinical Genetics, 2006, 69, 297-305.	2.0	33
72	The <emph type="ital">APOE</emph> Îµ2 Allele Increases the Risk of Earlier Age at Onset in Machado-Joseph Disease. Archives of Neurology, 2011, 68, 1580.	4.5	33

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73	Modifiers of (CAG)n instability in Machadoâ€"Joseph disease (MJD/SCA3) transmissions: an association study with DNA replication, repair and recombination genes. Human Genetics, 2014, 133, 1311-1318.	3.8	33
74	Updated frequency analysis of spinocerebellar ataxia in China. Brain, 2018, 141, e22-e22.	7.6	33
75	Use of fluoxetine for treatment of Machado-Joseph disease: an open-label study. Acta Neurologica Scandinavica, 2003, 107, 207-210.	2.1	32
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	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, 147-156.	2.8	31
78	A multistep mutation mechanism drives the evolution of the CAG repeat at MJD/SCA3 locus. European Journal of Human Genetics, 2006, 14, 932-940.	2.8	31
79	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
80	Rare Neurodegenerative Diseases: Clinical and Genetic Update. Advances in Experimental Medicine and Biology, 2017, 1031, 443-496.	1.6	30
81	Clinical, neuropathological, and genetic characterization of STUB1 variants in cerebellar ataxias: a frequent cause of predominant cognitive impairment. Genetics in Medicine, 2020, 22, 1851-1862.	2.4	30
82	Machado-Joseph Disease in a Sicilian-American Family. Journal of Neurogenetics, 1986, 3, 177-182.	1.4	29
83	Limits of Clinical Assessment in the Accurate Diagnosis of Machado-Joseph Disease. Archives of Neurology, 1996, 53, 1168-1174.	4.5	29
84	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.	2.3	29
85	Huntington diseaseâ€like 2: the first patient with apparent European ancestry. Clinical Genetics, 2008, 73, 480-485.	2.0	29
86	Abnormal movements in Rett syndrome are present before the regression period: A case study. Movement Disorders, 2007, 22, 2284-2287.	3.9	28
87	EuroGentest: DNA-Based Testing for Heritable Disorders in Europe. Public Health Genomics, 2008, 11, 75-120.	1.0	28
88	What Counts as Effective Genetic Counselling for Presymptomatic Testing in Lateâ€Onset Disorders? A Study of the Consultand's Perspective. Journal of Genetic Counseling, 2013, 22, 437-447.	1.6	28
89	Genotypes at the APOE and SCA2 loci do not predict the course of multiple sclerosis in patients of Portuguese origin. Multiple Sclerosis Journal, 2004, 10, 153-157.	3.0	27
90	Frataxin overexpressing mice. FEBS Letters, 2004, 572, 281-288.	2.8	27

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91	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
92	Cerebellar Ataxia, Hemiplegic Migraine, and Related Phenotypes Due to a CACNA1A Missense Mutation. JAMA Neurology, 2013, 70, 235.	9.0	27
93	Searching for modulating effects of SCA2, SCA6 and DRPLA CAG tracts on the Machado-Joseph disease (SCA3) phenotype. Acta Neurologica Scandinavica, 2003, 107, 211-214.	2.1	26
94	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
95	Genomic structure, promoter activity, and developmental expression of the mouse homologue of the Machadoâ€“Joseph disease (MJD) geneâ˜†. Genomics, 2004, 84, 361-373.	2.9	26
96	Exclusion of mutations in the PRNP, JPH3, TBP, ATN1, CREBBP, POU3F2 and FTL genes as a cause of disease in Portuguese patients with a Huntington-like phenotype. Journal of Human Genetics, 2006, 51, 645-651.	2.3	26
97	Two novel functional mutations in the Na⁺,K⁺â€ATPase Î±2â€subunit ATP1A2 gene in patients with familial hemiplegic migraine and associated neurological phenotypes. Clinical Genetics, 2008, 73, 37-43.	2.0	26
98	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
99	Huntington disease and Huntington diseaseâ€like in a case series from Brazil. Clinical Genetics, 2014, 86, 373-377.	2.0	26
100	The C677T Polymorphism inMTHFRIs Not Associated with Migraine in Portugal. Disease Markers, 2008, 25, 107-113.	1.3	25
101	Familial Clustering of Migraine: Further Evidence From a Portuguese Study. Headache, 2009, 49, 404-411.	3.9	25
102	Mutational Origin of Machado-Joseph Disease in the Australian Aboriginal Communities of Groote Eylandt and Yirrkala. Archives of Neurology, 2012, 69, 746-51.	4.5	25
103	Alu elements mediate large SPG11 gene rearrangements: further spatacsin mutations. Genetics in Medicine, 2012, 14, 143-151.	2.4	25
104	Linkage disequilibrium analysis in Machado-Joseph disease patients of different ethnic origins. Human Genetics, 1996, 98, 620-624.	3.8	24
105	Spinocerebellar ataxias in 114 Brazilian families: clinical and molecular findings. Clinical Genetics, 2006, 70, 173-176.	2.0	24
106	Cisâ€acting factors promoting the CAG intergenerational instability in Machadoâ€“Joseph disease. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2008, 147B, 439-446.	1.7	24
107	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, 483-493.	1.6	24
108	Rett syndrome with and without detected MECP2 mutations: An attempt to redefine phenotypes. Brain and Development, 2011, 33, 69-76.	1.1	24

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109	Experience and outcome of 3 years of a European EQA scheme for genetic testing of the spinocerebellar ataxias. European Journal of Human Genetics, 2008, 16, 913-920.	2.8	23
110	Regulations and practices of genetic counselling in 38 European countries: the perspective of national representatives. European Journal of Human Genetics, 2008, 16, 1208-1216.	2.8	23
111	The Challenges of Incorporating Genetic Testing in the Unified National Health System in Brazil. Genetic Testing and Molecular Biomarkers, 2012, 16, 651-655.	0.7	23
112	Two pairs of proven monozygotic twins discordant for familial amyloid neuropathy (FAP) TTR Met 30. Journal of Medical Genetics, 1999, 36, 629-32.	3.2	23
113	Machado-Joseph Disease Enhances Genetic Fitness: A Comparison Between Affected and Unaffected Women and Between MJD and the General Population. Annals of Human Genetics, 2007, 72, 070807042352001-???.	0.8	22
114	Genomic mechanisms underlying PARK2 large deletions identified in a cohort of patients with PD. Neurology: Genetics, 2016, 2, e73.	1.9	22
115	Mutational mechanism for DAB1 (ATTTC) _n insertion in SCA37: ATTTT repeat lengthening and nucleotide substitution. Human Mutation, 2019, 40, 404-412.	2.5	21
116	Report of an International Survey of Molecular Genetic Testing Laboratories. Public Health Genomics, 2007, 10, 123-131.	1.0	20
117	TAA repeat variation in the GRIK2 gene does not influence age at onset in Huntingtonâ€™s disease. Biochemical and Biophysical Research Communications, 2012, 424, 404-408.	2.1	20
118	Definitions of genetic testing in European legal documents. Journal of Community Genetics, 2012, 3, 125-141.	1.2	20
119	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
120	Genetic modifiers of age-at-onset in polyglutamine diseases. Ageing Research Reviews, 2018, 48, 99-108.	10.9	20
121	Mosaicism of the CAG repeat in CNS tissue in relation to age at death in spinocerebellar ataxia type 1 and Machado-Joseph disease patients. American Journal of Human Genetics, 1997, 60, 993-6.	6.2	20
122	Machado-Joseph Disease in an American-Italian Family. Journal of Neurogenetics, 1984, 1, 185-188.	1.4	19
123	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
124	Molecular diagnosis of Huntington disease in Portugal: implications for genetic counselling and clinical practice. European Journal of Human Genetics, 2003, 11, 872-878.	2.8	18
125	Interaction between Î³-Aminobutyric Acid A Receptor Genes: New Evidence in Migraine Susceptibility. PLoS ONE, 2013, 8, e74087.	2.5	18
126	Prenatal diagnosis of Machadoâ€“Joseph disease by direct mutation analysis. , 1998, 18, 611-617.		17

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127	Origins and Spread of Machado-Joseph Disease Ancestral Mutations Events. Advances in Experimental Medicine and Biology, 2018, 1049, 243-254.	1.6	17
128	Is the High Frequency of Machado-Joseph Disease in China Due to New Mutational Origins?. Frontiers in Genetics, 2018, 9, 740.	2.3	17
129	Linear skin atrophy, scarring alopecia, anonychia, and tongue lesion: A â€œnewâ€•syndrome?. American Journal of Medical Genetics Part A, 1985, 21, 669-680.	2.4	16
130	NovelSPG3AandSPG4mutations in dominant spastic paraplegia families. Acta Neurologica Scandinavica, 2009, 119, 113-118.	2.1	16
131	Assessing Risk Factors for Migraine: Differences in Gender Transmission. PLoS ONE, 2012, 7, e50626.	2.5	16
132	The wide variation of definitions of genetic testing in international recommendations, guidelines and reports. Journal of Community Genetics, 2012, 3, 113-124.	1.2	16
133	[NO TITLE AVAILABLE]. Genetics and Molecular Biology, 2014, 37, 263-270.	1.3	16
134	Genomic analysis in the clinic: benefits and challenges for health care professionals and patients in Brazil. Journal of Community Genetics, 2015, 6, 275-283.	1.2	16
135	Genetic Counseling in Portugal: Education, Practice and a Developing Profession. Journal of Genetic Counseling, 2015, 24, 548-552.	1.6	16
136	Recurrent ATP1A2 mutations in Portuguese families with familial hemiplegic migraine. Journal of Human Genetics, 2007, 52, 990-998.	2.3	15
137	Evidence of Syntaxin 1A Involvement in Migraine Susceptibility. Archives of Neurology, 2010, 67, 422-7.	4.5	15
138	Genetic study of 15 STRs loci of Identifiler system in Angola population. Forensic Science International: Genetics, 2010, 4, e153-e157.	3.1	15
139	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
140	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
141	Life paths of patients with transthyretin-related familial amyloid polyneuropathy Val30Met: a descriptive study. Journal of Community Genetics, 2018, 9, 93-99.	1.2	14
142	Twenty Years of a Pre-Symptomatic Testing Protocol for Late-Onset Neurological Diseases in Portugal. Acta Medica Portuguesa, 2019, 32, 295.	0.4	14
143	A whole genome screen for association with multiple sclerosis in Portuguese patients. Journal of Neuroimmunology, 2003, 143, 112-115.	2.3	13
144	Psychological Follow-up of Presymptomatic Genetic Testing for Spinocerebellar Ataxia Type 2 (SCA2) in Cuba. Journal of Genetic Counseling, 2007, 16, 469-479.	1.6	13

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145	Quality Assessment of Genetic Counseling Process in the Context of Presymptomatic Testing for Late-Onset Disorders: A Thematic Analysis of Three Review Articles. Genetic Testing and Molecular Biomarkers, 2012, 16, 36-45.	0.7	13
146	Gender equality in Machadoâ€“Joseph disease. Nature Genetics, 1995, 11, 118-119.	21.4	12
147	Intergenerational instability in Huntington disease: Extreme repeat changes among 134 transmissions. Movement Disorders, 2012, 27, 583-585.	3.9	12
148	Genetics Health Professionalsâ€™ Views on Quality of Genetic Counseling Service Provision for Presymptomatic Testing in Lateâ€Onset Neurological Diseases in Portugal: Core Components, Specific Challenges and the Need for Assessment Tools. Journal of Genetic Counseling, 2015, 24, 616-625.	1.6	12
149	Discredited legacy: Stigma and familial amyloid polyneuropathy in Northwestern Portugal. Social Science and Medicine, 2017, 182, 73-80.	3.8	12
150	Spinocerebellar ataxia type 10: common haplotype and disease progression rate in Peru and Brazil. European Journal of Neurology, 2017, 24, 892.	3.3	12
151	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
152	Analysis of (CAG)n expansion in ATXN1, ATXN2 and ATXN3 in Chinese patients with multiple system atrophy. Scientific Reports, 2018, 8, 3889.	3.3	11
153	Living with Machado-Joseph Disease in a Small Rural Community of the Tagus Valley. Public Health Genomics, 1999, 2, 190-195.	1.0	10
154	The need for interaction between assisted reproduction technology and genetics: recommendations of the European Societies of Human Genetics and Human Reproduction and Embryology*. Human Reproduction, 2006, 21, 1971-1973.	0.9	10
155	Scope of definitions of genetic testing: evidence from a EuroGentest survey. Journal of Community Genetics, 2010, 1, 29-35.	1.2	10
156	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
157	Candidate glutamatergic and dopaminergic pathway gene variants do not influence Huntingtonâ€™s disease motor onset. Neurogenetics, 2013, 14, 173-179.	1.4	10
158	Machadoâ€“Joseph disease in a Nigerian family: mutational origin and review of the literature. European Journal of Human Genetics, 2015, 23, 271-273.	2.8	10
159	Ubiquitin-related network underlain by (CAG)n loci modulate age at onset in Machado-Joseph disease. Brain, 2017, 140, e25-e25.	7.6	10
160	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
161	A Pipeline to Assess Disease-Associated Haplotypes in Repeat Expansion Disorders: The Example of MJD/SCA3 Locus. Frontiers in Genetics, 2019, 10, 38.	2.3	10
162	Genome-wide association study identifies genetic factors that modify age at onset in Machado-Joseph disease. Aging, 2020, 12, 4742-4756.	3.1	10

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163	Quality issues concerning genetic counselling for presymptomatic testing: a European Delphi study. European Journal of Human Genetics, 2015, 23, 1468-1472.	2.8	9
164	Subjects Atâ€Risk for Genetic Diseases in Portugal: Illness Representations. Journal of Genetic Counseling, 2016, 25, 79-89.	1.6	9
165	Familial amyloid polyneuropathy in Portugal: New genes modulating ageâ€atâ€onset. Annals of Clinical and Translational Neurology, 2017, 4, 98-105.	3.7	9
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