Jorge Sequeiros

List of Publications by Citations

230 papers	7,320 citations	45 h-index	75 g-index
243 ext. papers	8,203 ext. citations	5.4 avg, IF	5.16 L-index

#	Paper	IF	Citations
230	Senataxin, the ortholog of a yeast RNA helicase, is mutant in ataxia-ocular apraxia 2. *Nature Genetics*, **2004**, 36, 225-7	36.3	385
229	The gene mutated in ataxia-ocular apraxia 1 encodes the new HIT/Zn-finger protein aprataxin. *Nature Genetics*, **2001**, 29, 189-93	36.3	383
228	A dominant-negative mutation in the TRESK potassium channel is linked to familial migraine with aura. *Nature Medicine*, **2010**, 16, 1157-60	50.5	263
227	CAG repeat expansion in Huntington disease determines age at onset in a fully dominant fashion. *Neurology*, **2012**, 78, 690-5	6.5	231
226	Ataxia with oculomotor apraxia type 2: clinical, biological and genotype/phenotype correlation study of a cohort of 90 patients. *Brain*, **2009**, 132, 2688-98	11.2	173
225	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
224	Identification of genetic variants associated with Huntington□s disease progression: a genome-wide association study. *Lancet Neurology, The*, **2017**, 16, 701-711	24.1	161
223	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
222	Neurologic findings in Machado-Joseph disease: relation with disease duration, subtypes, and (CAG)n. *Archives of Neurology*, **2001**, 58, 899-904		132
221	Frataxin knockin mouse. *FEBS Letters*, **2002**, 512, 291-7	3.8	128
220	The interface between assisted reproductive technologies and genetics: technical, social, ethical and legal issues. *European Journal of Human Genetics*, **2006**, 14, 588-645	5.3	116
219	Current issues in medically assisted reproduction and genetics in Europe: research, clinical practice, ethics, legal issues and policy. European Society of Human Genetics and European Society of Human Reproduction and Embryology. *European Journal of Human Genetics*, **2013**, 21 Suppl 2, S1-21	5.3	107
218	Ancestral origins of the Machado-Joseph disease mutation: a worldwide haplotype study. *American Journal of Human Genetics*, **2001**, 68, 523-8	11	103
217	Neuroferritinopathy: missense mutation in FTL causing early-onset bilateral pallidal involvement. *Neurology*, **2005**, 65, 603-5	6.5	101
216	Improvement in the molecular diagnosis of Machado-Joseph disease. *Archives of Neurology*, **2001**, 58, 1821-7		100
215	Recessive ataxia with ocular apraxia: review of 22 Portuguese patients. *Archives of Neurology*, **2001**, 58, 201-5		96
214	Frequency of spinocerebellar ataxia type 1, dentatorubropallidoluysian atrophy, and Machado-Joseph disease mutations in a large group of spinocerebellar ataxia patients. *Neurology*, **1996**, 46, 214-8	6.5	95

213	Mutations in PNKP cause recessive ataxia with oculomotor apraxia type 4. *American Journal of Human Genetics*, **2015**, 96, 474-9	11	90
212	Ataxia rating scales--psychometric profiles, natural history and their application in clinical trials. *Cerebellum*, **2012**, 11, 488-504	4.3	88
211	Epidemiology and clinical aspects of Machado-Joseph disease. *Advances in Neurology*, **1993**, 61, 139-53		86
210	Hereditary ataxia and spastic paraplegia in Portugal: a population-based prevalence study. *JAMA Neurology*, **2013**, 70, 746-55	17.2	82
209	Massively parallel sequencing of ataxia genes after array-based enrichment. *Human Mutation*, **2010**, 31, 494-9	4.7	79
208	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
207	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
206	What is ideal genetic counselling? A survey of current international guidelines. *European Journal of Human Genetics*, **2008**, 16, 445-52	5.3	70
205	First mutation in the voltage-gated Nav1.1 subunit gene SCN1A with co-occurring familial hemiplegic migraine and epilepsy. *Cephalalgia*, **2009**, 29, 308-13	6.1	68
204	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
203	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, 501-8	11	68
202	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	11	66
201	Stereotypies in Rett syndrome: analysis of 83 patients with and without detected MECP2 mutations. *Neurology*, **2007**, 68, 1183-7	6.5	65
200	Genetic testing and common disorders in a public health framework: how to assess relevance and possibilities. Background Document to the ESHG recommendations on genetic testing and common disorders. *European Journal of Human Genetics*, **2011**, 19 Suppl 1, S6-44	5.3	60
199	Movement disorders in Rett syndrome: an analysis of 60 patients with detected MECP2 mutation and correlation with mutation type. *Movement Disorders*, **2008**, 23, 1384-90	7	57
198	PCosta da MortePataxia is spinocerebellar ataxia 36: clinical and genetic characterization. *Brain*, **2012**, 135, 1423-35	11.2	56
197	Consensus and controversies in best practices for molecular genetic testing of spinocerebellar ataxias. *European Journal of Human Genetics*, **2010**, 18, 1188-95	5.3	56
196	Inherited and acquired risk factors and their combined effects in pediatric stroke. *Pediatric Neurology*, **2003**, 28, 134-8	2.9	55

195	Nonsense mutation in TITF1 in a Portuguese family with benign hereditary chorea. *Neurogenetics*, **2005**, 6, 209-15	3	55
194	Somatic mosaicism in the central nervous system in spinocerebellar ataxia type 1 and Machado-Joseph disease. *Annals of Neurology*, **1996**, 40, 199-206	9.4	55
193	Functional genomics and biochemical characterization of the C. elegans orthologue of the Machado-Joseph disease protein ataxin-3. *FASEB Journal*, **2007**, 21, 1126-36	0.9	53
192	Asian origin for the worldwide-spread mutational event in Machado-Joseph disease. *Archives of Neurology*, **2007**, 64, 1502-8		50
191	Common SNP-based haplotype analysis of the 4p16.3 Huntington disease gene region. *American Journal of Human Genetics*, **2012**, 90, 434-44	11	48
190	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
189	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
188	NEDD8: a new ataxin-3 interactor. *Biochimica Et Biophysica Acta - Molecular Cell Research*, **2007**, 1773, 1619-27	4.9	47
187	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, 315-25	5.3	45
186	Epidemiology and population genetics of degenerative ataxias. *Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn*, **2012**, 103, 227-51	3	45
185	Autosomal dominant cerebellar ataxia: frequency analysis and clinical characterization of 45 families from Portugal. *European Journal of Neurology*, **2010**, 17, 124-8	6	45
184	CAG tract of MJD-1 may be prone to frameshifts causing polyalanine accumulation. *Human Molecular Genetics*, **2000**, 9, 1957-66	5.6	45
183	Current issues in medically assisted reproduction and genetics in Europe: research, clinical practice, ethics, legal issues and policy. *Human Reproduction*, **2014**, 29, 1603-9	5.7	42
182	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, 857-9	5.3	42
181	Familial ATTR amyloidosis: microalbuminuria as a predictor of symptomatic disease and clinical nephropathy. *Nephrology Dialysis Transplantation*, **2003**, 18, 532-8	4.3	42
180	Autosomal dominant spastic paraplegias: a review of 89 families resulting from a portuguese survey. *JAMA Neurology*, **2013**, 70, 481-7	17.2	41
179	Depressive symptoms in Machado-Joseph disease (SCA3) patients and their relatives. *Public Health Genomics*, **2007**, 10, 19-26		41
178	Genetic interaction of CTLA-4 with HLA-DR15 in multiple sclerosis patients. *Annals of Neurology*, **2003**, 54, 119-22	9.4	41

177	Analysis of SCA1, DRPLA, MJD, SCA2, and SCA6 CAG repeats in 48 Portuguese ataxia families. *American Journal of Medical Genetics Part A*, **1998**, 81, 134-8		40
176	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
175	Machado-Joseph disease in South Brazil: clinical and molecular characterization of kindreds. *Acta Neurologica Scandinavica*, **2001**, 104, 224-31	3.8	40
174	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
173	BDNF and CGRP interaction: implications in migraine susceptibility. *Cephalalgia*, **2010**, 30, 1375-82	6.1	38
172	Evaluation of CSF neurotransmitters and folate in 25 patients with Rett disorder and effects of treatment. *Brain and Development*, **2009**, 31, 46-51	2.2	38
171	Onset in the seventh decade and lack of symptoms in heterozygotes for the TTRMet30 mutation in hereditary amyloid neuropathy-type I (Portuguese, Andrade). *American Journal of Medical Genetics Part A*, **1987**, 27, 345-57		36
170	Portuguese families with dentatorubropallidoluysian atrophy (DRPLA) share a common haplotype of Asian origin. *European Journal of Human Genetics*, **2003**, 11, 808-11	5.3	35
169	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
168	Population genetics of wild-type CAG repeats in the Machado-Joseph disease gene in Portugal. *Human Heredity*, **2005**, 60, 156-63	1.1	33
167	Ancestral origin of the ATTCT repeat expansion in spinocerebellar ataxia type 10 (SCA10). *PLoS ONE*, **2009**, 4, e4553	3.7	32
166	The APOE ε2 allele increases the risk of earlier age at onset in Machado-Joseph disease. *Archives of Neurology*, **2011**, 68, 1580-3		31
165	(CAG)n loci as genetic modifiers of age-at-onset in patients with Machado-Joseph disease from mainland China. *Brain*, **2016**, 139, e41	11.2	31
164	A novel H101Q mutation causes PKCgamma loss in spinocerebellar ataxia type 14. *Journal of Human Genetics*, **2005**, 50, 523-529	4.3	30
163	Genetic linkage studies of Machado-Joseph disease with chromosome 14q STRPs in 16 Portuguese-Azorean kindreds. *Genomics*, **1994**, 21, 645-8	4.3	30
162	Modifiers of (CAG)(n) instability in Machado-Joseph disease (MJD/SCA3) transmissions: an association study with DNA replication, repair and recombination genes. *Human Genetics*, **2014**, 133, 1311-8	6.3	29
161	EMQN Best Practice Guidelines for molecular genetic testing of SCAs. *European Journal of Human Genetics*, **2010**, 18, 1173-6	5.3	29
160	Use of fluoxetine for treatment of Machado-Joseph disease: an open-label study. *Acta Neurologica Scandinavica*, **2003**, 107, 207-10	3.8	29

159	Psychological aspects of pre-symptomatic testing for Machado-Joseph disease and familial amyloid polyneuropathy type I. *Clinical Genetics*, **2006**, 69, 297-305	4	28
158	Machado-Joseph disease in a Sicilian-American family. *Journal of Neurogenetics*, **1986**, 3, 177-82	1.6	28
157	Abnormal movements in Rett syndrome are present before the regression period: a case study. *Movement Disorders*, **2007**, 22, 2284-7	7	27
156	Reduced penetrance of intermediate size alleles in spinocerebellar ataxia type 10. *Neurology*, **2006**, 66, 1602-4	6.5	26
155	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
154	EuroGentest: DNA-based testing for heritable disorders in Europe. *Public Health Genomics*, **2008**, 11, 75-120	1.9	25
153	What counts as effective genetic counselling for presymptomatic testing in late-onset disorders? A study of the consultand's perspective. *Journal of Genetic Counseling*, **2013**, 22, 437-47	2.5	24
152	Huntington disease-like 2: the first patient with apparent European ancestry. *Clinical Genetics*, **2008**, 73, 480-5	4	24
151	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
150	A multistep mutation mechanism drives the evolution of the CAG repeat at MJD/SCA3 locus. *European Journal of Human Genetics*, **2006**, 14, 932-40	5.3	24
149	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
148	Genomic structure, promoter activity, and developmental expression of the mouse homologue of the Machado-Joseph disease (MJD) gene. *Genomics*, **2004**, 84, 361-73	4.3	24
147	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
146	Limits of clinical assessment in the accurate diagnosis of Machado-Joseph disease. *Archives of Neurology*, **1996**, 53, 1168-74		24
145	Identification of genetic risk factors for maxillary lateral incisor agenesis. *Journal of Dental Research*, **2014**, 93, 452-8	8.1	23
144	Familial clustering of migraine: further evidence from a Portuguese study. *Headache*, **2009**, 49, 404-11	4.2	23
143	Two novel functional mutations in the Na+,K+-ATPase alpha2-subunit ATP1A2 gene in patients with familial hemiplegic migraine and associated neurological phenotypes. *Clinical Genetics*, **2008**, 73, 37-43	4	23
142	Spinocerebellar ataxias in 114 Brazilian families: clinical and molecular findings. *Clinical Genetics*, **2006**, 70, 173-6	4	23

141	Searching for modulating effects of SCA2, SCA6 and DRPLA CAG tracts on the Machado-Joseph disease (SCA3) phenotype. *Acta Neurologica Scandinavica*, **2003**, 107, 211-4	3.8	23
140	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
139	Frataxin overexpressing mice. *FEBS Letters*, **2004**, 572, 281-8	3.8	23
138	Linkage disequilibrium analysis in Machado-Joseph disease patients of different ethnic origins. *Human Genetics*, **1996**, 98, 620-4	6.3	23
137	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
136	Huntington disease and Huntington disease-like in a case series from Brazil. *Clinical Genetics*, **2014**, 86, 373-7	4	22
135	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
134	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	4.3	22
133	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, 153-7	5	22
132	Two pairs of proven monozygotic twins discordant for familial amyloid neuropathy (FAP) TTR Met 30. *Journal of Medical Genetics*, **1999**, 36, 629-32	5.8	22
131	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, 913-20	5.3	21
130	Regulations and practices of genetic counselling in 38 European countries: the perspective of national representatives. *European Journal of Human Genetics*, **2008**, 16, 1208-16	5.3	21
129	Communication of Information about Genetic Risks: Putting Families at the Center. *Family Process*, **2018**, 57, 836-846	3.9	20
128	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
127	Updated frequency analysis of spinocerebellar ataxia in China. *Brain*, **2018**, 141, e22	11.2	19
126	Alu elements mediate large SPG11 gene rearrangements: further spatacsin mutations. *Genetics in Medicine*, **2012**, 14, 143-51	8.1	19
125	Mutational origin of Machado-Joseph disease in the Australian Aboriginal communities of Groote Eylandt and Yirrkala. *Archives of Neurology*, **2012**, 69, 746-51		19
124	The C677T polymorphism in MTHFR is not associated with migraine in Portugal. *Disease Markers*, **2008**, 25, 107-13	3.2	19

123	Machado-Joseph disease in an American-Italian family. *Journal of Neurogenetics*, **1984**, 1, 185-8	1.6	19
122	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	11	19
121	Rett syndrome with and without detected MECP2 mutations: an attempt to redefine phenotypes. *Brain and Development*, **2011**, 33, 69-76	2.2	18
120	Cis-acting factors promoting the CAG intergenerational instability in Machado-Joseph disease. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2008**, 147B, 439-46	3.5	18
119	The challenges of incorporating genetic testing in the unified national health system in Brazil. *Genetic Testing and Molecular Biomarkers*, **2012**, 16, 651-5	1.6	17
118	TAA repeat variation in the GRIK2 gene does not influence age at onset in Huntington's disease. *Biochemical and Biophysical Research Communications*, **2012**, 424, 404-8	3.4	17
117	Report of an international survey of molecular genetic testing laboratories. *Public Health Genomics*, **2007**, 10, 123-31	1.9	16
116	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
115	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	8.1	16
114	Genetic modifiers of age-at-onset in polyglutamine diseases. *Ageing Research Reviews*, **2018**, 48, 99-108	12	16
113	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
112	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
111	Definitions of genetic testing in European legal documents. *Journal of Community Genetics*, **2012**, 3, 125-41	2.5	15
110	Machado-Joseph disease enhances genetic fitness: a comparison between affected and unaffected women and between MJD and the general population. *Annals of Human Genetics*, **2008**, 72, 57-64	2.2	15
109	Recurrent ATP1A2 mutations in Portuguese families with familial hemiplegic migraine. *Journal of Human Genetics*, **2007**, 52, 990-998	4.3	15
108	Genomic mechanisms underlying PARK2 large deletions identified in a cohort of patients with PD. *Neurology: Genetics*, **2016**, 2, e73	3.8	14
107	Genetic counseling and presymptomatic testing programs for Machado-Joseph Disease: lessons from Brazil and Portugal. *Genetics and Molecular Biology*, **2014**, 37, 263-70	2	14
106	Assessing risk factors for migraine: differences in gender transmission. *PLoS ONE*, **2012**, 7, e50626	3.7	14

105	Novel SPG3A and SPG4 mutations in dominant spastic paraplegia families. *Acta Neurologica Scandinavica*, **2009**, 119, 113-8	3.8	14
104	Prenatal diagnosis of Machado□Joseph disease by direct mutation analysis **1998**, 18, 611-617		14
103	Interaction between □aminobutyric acid A receptor genes: new evidence in migraine susceptibility. *PLoS ONE*, **2013**, 8, e74087	3.7	14
102	Rare Neurodegenerative Diseases: Clinical and Genetic Update. *Advances in Experimental Medicine and Biology*, **2017**, 1031, 443-496	3.6	13
101	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
100	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	1.6	13
99	The wide variation of definitions of genetic testing in international recommendations, guidelines and reports. *Journal of Community Genetics*, **2012**, 3, 113-24	2.5	13
98	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
97	Linear skin atrophy, scarring alopecia, anonychia, and tongue lesion: a "new" syndrome?. *American Journal of Medical Genetics Part A*, **1985**, 21, 669-80		13
96	Genomic analysis in the clinic: benefits and challenges for health care professionals and patients in Brazil. *Journal of Community Genetics*, **2015**, 6, 275-83	2.5	12
95	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
94	Gender equality in Machado-Joseph disease. *Nature Genetics*, **1995**, 11, 118-9	36.3	12
93	Twenty Years of a Pre-Symptomatic Testing Protocol for Late-Onset Neurological Diseases in Portugal. *Acta Medica Portuguesa*, **2019**, 32, 295-304	1.4	12
92	Spinocerebellar ataxia type 10: common haplotype and disease progression rate in Peru and Brazil. *European Journal of Neurology*, **2017**, 24, 892-e36	6	11
91	Genetic Counseling in Portugal: Education, Practice and a Developing Profession. *Journal of Genetic Counseling*, **2015**, 24, 548-52	2.5	11
90	Life paths of patients with transthyretin-related familial amyloid polyneuropathy Val30Met: a descriptive study. *Journal of Community Genetics*, **2018**, 9, 93-99	2.5	11
89	Discredited legacy: Stigma and familial amyloid polyneuropathy in Northwestern Portugal. *Social Science and Medicine*, **2017**, 182, 73-80	5.1	10
88	Intergenerational instability in Huntington disease: extreme repeat changes among 134 transmissions. *Movement Disorders*, **2012**, 27, 583-5	7	10

87	Genetic study of 15 STRs loci of Identifiler system in Angola population. *Forensic Science International: Genetics*, **2010**, 4, e153-7	4.3	10
86	Evidence of syntaxin 1A involvement in migraine susceptibility: a Portuguese study. *Archives of Neurology*, **2010**, 67, 422-7		10
85	A whole genome screen for association with multiple sclerosis in Portuguese patients. *Journal of Neuroimmunology*, **2003**, 143, 112-5	3.5	10
84	Genetics Health ProfessionalsPViews on Quality of Genetic Counseling Service Provision for Presymptomatic Testing in Late-Onset Neurological Diseases in Portugal: Core Components, Specific Challenges and the Need for Assessment Tools. *Journal of Genetic Counseling*, **2015**, 24, 616-25	2.5	9
83	Candidate glutamatergic and dopaminergic pathway gene variants do not influence HuntingtonP disease motor onset. *Neurogenetics*, **2013**, 14, 173-9	3	9
82	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
81	Prenatal diagnosis of Machado-Joseph disease by direct mutation analysis. *Prenatal Diagnosis*, **1998** , 18, 611-7	3.2	9
80	Origins and Spread of Machado-Joseph Disease Ancestral Mutations Events. *Advances in Experimental Medicine and Biology*, **2018**, 1049, 243-254	3.6	8
79	Quality issues concerning genetic counselling for presymptomatic testing: a European Delphi study. *European Journal of Human Genetics*, **2015**, 23, 1468-72	5.3	8
78	Haplotype diversity and somatic instability in normal and expanded SCA8 alleles. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2005**, 139B, 109-14	3.5	8
77	Mutational mechanism for DAB1 (ATTTC) insertion in SCA37: ATTTT repeat lengthening and nucleotide substitution. *Human Mutation*, **2019**, 40, 404-412	4.7	8
76	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
75	Ubiquitin-related network underlain by (CAG)n loci modulate age at onset in Machado-Joseph disease. *Brain*, **2017**, 140, e25	11.2	7
74	and modify age-at-onset in familial amyloid polyneuropathy patients. *Annals of Clinical and Translational Neurology*, **2019**, 6, 748-754	5.3	7
73	Machado-Joseph disease in a Nigerian family: mutational origin and review of the literature. *European Journal of Human Genetics*, **2015**, 23, 271-3	5.3	7
72	Long-term predictors for psychological outcome of pre-symptomatic testing for late-onset neurological diseases. *European Journal of Medical Genetics*, **2018**, 61, 575-580	2.6	7
71	Genetic screening in Europe. *Public Health Genomics*, **2010**, 13, 524-37	1.9	7
70	Scope of definitions of genetic testing: evidence from a EuroGentest survey. *Journal of Community Genetics*, **2010**, 1, 29-35	2.5	7

69	Living with Machado-Joseph disease in a small rural community of the Tagus valley. *Public Health Genomics*, **1999**, 2, 190-5	1.9	7
68	Mid- and long-term anxiety levels associated with presymptomatic testing of Huntington's disease, Machado-Joseph disease, and familial amyloid polyneuropathy. *Revista Brasileira De Psiquiatria*, **2016**, 38, 113-20	2.6	7
67	Is the High Frequency of Machado-Joseph Disease in China Due to New Mutational Origins?. *Frontiers in Genetics*, **2018**, 9, 740	4.5	7
66	Familial amyloid polyneuropathy in Portugal: New genes modulating age-at-onset. *Annals of Clinical and Translational Neurology*, **2017**, 4, 98-105	5.3	6
65	Analysis of (CAG) expansion in ATXN1, ATXN2 and ATXN3 in Chinese patients with multiple system atrophy. *Scientific Reports*, **2018**, 8, 3889	4.9	6
64	Subjects At-Risk for Genetic Diseases in Portugal: Illness Representations. *Journal of Genetic Counseling*, **2016**, 25, 79-89	2.5	6
63	Sequence analysis of 5'regulatory regions of the Machado-Joseph disease gene (ATXN3). *Cerebellum*, **2012**, 11, 1045-50	4.3	6
62	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
61	Atypical stereotypies and vocal tics in Rett syndrome: An illustrative case. *Movement Disorders*, **2008**, 23, 622-4	7	6
60	Genome-wide association study identifies genetic factors that modify age at onset in Machado-Joseph disease. *Aging*, **2020**, 12, 4742-4756	5.6	6
59	A Pipeline to Assess Disease-Associated Haplotypes in Repeat Expansion Disorders: The Example of MJD/SCA3. *Frontiers in Genetics*, **2019**, 10, 38	4.5	5
58	Investigation on modulation of DNA repair pathways in Chinese MJD patients. *Neurobiology of Aging*, **2018**, 71, 267.e5-267.e6	5.6	5
57	Investigating ANKH and ENPP1 in Slovakian families with chondrocalcinosis. *Rheumatology International*, **2012**, 32, 2745-51	3.6	5
56	Non-syndromic sensorineural prelingual deafness: the importance of genetic counseling in demystifying parents'beliefs about the cause of their children's deafness. *Journal of Genetic Counseling*, **2013**, 22, 448-54	2.5	5
55	Direct to consumer genetic tests. *European Journal of Human Genetics*, **2009**, 17, 1111	5.3	5
54	Death anxiety and symbolic immortality in relatives at risk for familial amyloid polyneuropathy type I (FAP I, ATTR V30M). *Journal of Genetic Counseling*, **2010**, 19, 585-92	2.5	5
53	Iron metabolism in mice with partial frataxin deficiency. *Cerebellum*, **2003**, 2, 146-53	4.3	5
52	Choosing not to know: accounts of non-engagement with pre-symptomatic testing for Machado-Joseph disease. *European Journal of Human Genetics*, **2019**, 27, 353-359	5.3	5

51	Polymorphisms in DNA methylation-related genes are linked to the phenotype of Machado-Joseph disease. *Neurobiology of Aging*, **2019**, 75, 225.e1-225.e8	5.6	5
50	Chromosome substitution strain assessment of a Huntington's disease modifier locus. *Mammalian Genome*, **2015**, 26, 119-30	3.2	4
49	Psychopathological dimensions in subjects with hereditary ATTR V30M amyloidosis and their relation with life events due to the disease. *Amyloid: the International Journal of Experimental and Clinical Investigation: the Official Journal of the International Society of Amyloidosis*, **2018**, 25, 26-36	2.7	4
48	Large-Scale Functional RNAi Screen in C. elegans Identifies TGF-Beta and Notch Signaling Pathways as Modifiers of CACNA1A. *ASN Neuro*, **2016**, 8,	5.3	4
47	A repeat-primed PCR assay for pentanucleotide repeat alleles in spinocerebellar ataxia type 37. *Journal of Human Genetics*, **2018**, 63, 981-987	4.3	4
46	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
45	The prevalence of familial hemiplegic migraine with cerebellar ataxia and spinocerebellar ataxia type 6 in Portugal. *Headache*, **2014**, 54, 911-5	4.2	4
44	Insufficient referral for genetic counseling in the management of hereditary haemochromatosis in portugal: a study of perceptions of health professionals requesting HFE genotyping. *Journal of Genetic Counseling*, **2014**, 23, 770-7	2.5	4
43	Spinocerebellar ataxias: an example of the challenges associated with genetic databases for dynamic mutations. *Human Mutation*, **2012**, 33, 1359-65	4.7	4
42	Y-STR haplotypes in three ethnic linguistic groups of Angola population. *Forensic Science International: Genetics*, **2011**, 5, e83-8	4.3	4
41	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, 1971-3	5.7	4
40	Identification of three novel polymorphisms in the MJD1 gene and study of their frequency in the Portuguese population. *Journal of Human Genetics*, **2002**, 47, 205-7	4.3	4
39	Spinopontine atrophy disputed as a separate entity: The first description of Machado-Joseph disease. *Neurology*, **1986**, 36, 1408-1408	6.5	4
38	Regulating Genetic Testing: The Relevance of Appropriate Definitions **2010**, 23-32		4
37	Going Deep into Synaptic Vesicle Machinery Genes and Migraine Susceptibility - A Case-Control Association Study. *Headache*, **2020**, 60, 2152-2165	4.2	4
36	Illness representations, knowledge and motivation to perform presymptomatic testing for late-onset genetic diseases. *Psychology, Health and Medicine*, **2017**, 22, 244-249	2.1	3
35	Yemenite-Jewish families with Machado-Joseph disease (MJD/SCA3) share a recent common ancestor. *European Journal of Human Genetics*, **2019**, 27, 1731-1737	5.3	3
34	The interface between medically assisted reproduction and genetics: technical, social, ethical and legal issues*. *ESHRE Monographs*, **2006**, 2006, 2-51		3

33	Machado-Joseph disease is genetically different from Holguin dominant ataxia (SCA2). *Genomics*, **1993**, 17, 556-9	4.3	3
32	Anxiety and pre-symptomatic testing for neurodegenerative disorders. *Open Journal of Genetics*, **2013**, 03, 14-26	0.2	3
31	Management of information within Portuguese families with Huntington disease: a transgenerational process for putting the puzzle together. *European Journal of Human Genetics*, **2020**, 28, 1210-1217	5.3	2
30	Family dynamics in transthyretin-related familial amyloid polyneuropathy Val30Met: Does genetic risk affect family functioning?. *Clinical Genetics*, **2018**, 94, 401-408	4	2
29	Monozygotic twin sisters discordant for familial hemiplegic migraine. *Journal of Headache and Pain*, **2013**, 14, 77	8.8	2
28	Eye of the Tiger Sign and Very Late Onset in Dentatorubral-Pallidoluysian Atrophy. *Movement Disorders Clinical Practice*, **2015**, 2, 313-315	2.2	2
27	The perceived advantages and disadvantages of presymptomatic testing for Machado-Joseph disease: development of a new self-response inventory. *Journal of Genetic Counseling*, **2006**, 15, 375-91	2.5	2
26	Corino Andrade (1906-2005): a clinical geneticist before its own time. *Clinical Genetics*, **2006**, 69, 194-6	4	2
25	Depression as the Middle- and Long-Term Impact for Pre-Symptomatic Testing of Late-Onset Neurodegenerative Disorders. *Temas Em Psicologia*, **2016**, 24, 579-594	0.3	2
24	Sex Differences and Age-Dependent Penetrance in FAP-Type I **1991**, 687-690		2
23	Between responsibility and desire: Accounts of reproductive decisions from those at risk for or affected by late-onset neurological diseases. *Journal of Genetic Counseling*, **2021**, 30, 1480-1490	2.5	2
22	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
21	Genetics and ethics in Latin America. *Journal of Community Genetics*, **2015**, 6, 185-7	2.5	1
20	Novel Variant Causes Cerebellar Ataxia with Oculomotor Apraxia: Molecular Basis and Expanded Clinical Phenotype. *Journal of Clinical Medicine*, **2020**, 9,	5.1	1
19	Paula Coutinho's outstanding contribution to the definition of Machado-Joseph disease. *Arquivos De Neuro-Psiquiatria*, **2017**, 75, 748-750	1.6	1
18	Parkin truncating variants result in a loss-of-function phenotype. *Scientific Reports*, **2019**, 9, 16150	4.9	1
17	Risk perception in subjects at-risk for Familial Amyloidotic Polyneuropathy. *Universitas Psychologica* , **2017**, 16, 1	0.5	1
16	Ataxia and Progressive Encephalopathy in a 4-Year-Old Girl. *Laboratory Medicine*, **2010**, 41, 5-9	1.6	1

15	Infection and Prematurity as the Cause of Linear Skin Atrophy, Alopecia, Anonychia, and Tongue Lesions?. *Archives of Dermatology*, **1985**, 121, 1379		1
14	Genome-wide association study identifies genetic factors that modify age at onset in Machado-Joseph disease		1
13	Role of older generations in the family adjustment to Huntington disease. *Journal of Community Genetics*, **2021**, 12, 469-477	2.5	1
12	Congenital ataxia due to novel variant in ATP8A2. *Clinical Genetics*, **2021**, 100, 79-83	4	1
11	Rare occurrence of severe blindness and deafness in Friedreich ataxia: a case report. *Cerebellum and Ataxias*, **2021**, 8, 17	1.7	1
10	Novel KMT2B mutation causes cerebellar ataxia: Expanding the clinical phenotype. *Clinical Genetics*, **2021**, 100, 743-747	4	1
9	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
8	Response to Tumas et al.. *Clinical Genetics*, **2009**, 75, 208-208	4	0
7	A new scale informed by the Reciprocal-Engagement Model for quality evaluation of genetic counselling by patients: Development and initial validation. *European Journal of Medical Genetics*, **2021**, 64, 104375	2.6	0
6	Novel Machado-Joseph disease-modifying genes and pathways identified by whole-exome sequencing. *Neurobiology of Disease*, **2021**, 162, 105578	7.5	0
5	John MacMillan, M.D. (1959-2014): an inspiring example of a community clinical geneticist. *Journal of Community Genetics*, **2015**, 6, 329-30	2.5	
4	Psychopathological Dimensions in Portuguese Subjects with Transthyretin Familial Amyloid Polyneuropathy. *Biomedicine Hub*, **2017**, 2, 1-14	1.3	
3	Expertise, empathy and ethical awareness: ideals of genetic counseling based on framing of genetic information in international guidelines. *New Genetics and Society*, **2009**, 28, 301-316	1.9	
2	Pre-symptomatic testing for neurodegenerative disorders: Middle- to long-term psychopathological impact. *Psicothema*, **2017**, 29, 446-452	2	
1	From older to younger generations: Intergenerational transmission of health-related roles in families with Huntington disease. *Journal of Aging Studies*, **2022**, 61, 101027	2.2	