Patricia Maciel

List of Publications by Year
in descending order

144 papers	6,479 citations	81839 39 h-index	88593 70 g-index
159 all docs	159 docs citations	159 times ranked	9255 citing authors

#	Article	IF	Citations
1	Novel Machado-Joseph disease-modifying genes and pathways identified by whole-exome sequencing. Neurobiology of Disease, 2022, 162, 105578.	2.1	3
2	Profiling Microglia in a Mouse Model of Machadoâ€“Joseph Disease. Biomedicines, 2022, 10, 237.	1.4	3
3	Aripiprazole Offsets Mutant ATXN3-Induced Motor Dysfunction by Targeting Dopamine D2 and Serotonin 1A and 2A Receptors in C. elegans. Biomedicines, 2022, 10, 370.	1.4	6
4	Experimental modelling of Alzheimerâ€™s disease for therapeutic screening. European Journal of Medicinal Chemistry Reports, 2022, , 100044.	0.6	2
5	Modifier pathways in polyglutamine (PolyQ) diseases: from genetic screens to drug targets. Cellular and Molecular Life Sciences, 2022, 79, 274.	2.4	4
6	Microglial Depletion Has No Impact on Disease Progression in a Mouse Model of Machadoâ€“Joseph Disease. Cells, 2022, 11, 2022.	1.8	3
7	Cerebellar neuronal dysfunction accompanies early motor symptoms in spinocerebellar ataxia type 3. DMM Disease Models and Mechanisms, 2022, 15, .	1.2	5
8	IP₃R2 null mice display a normal acquisition of somatic and neurological development milestones. European Journal of Neuroscience, 2021, 54, 5673-5686.	1.2	12
9	Leading the way in the nervous system: Lipid Droplets as new players in health and disease. Biochimica Et Biophysica Acta - Molecular and Cell Biology of Lipids, 2021, 1866, 158820.	1.2	25
10	Neurotherapeutic effect of Hyptis spp. leaf extracts in Caenorhabditis elegans models of tauopathy and polyglutamine disease: Role of the glutathione redox cycle. Free Radical Biology and Medicine, 2021, 162, 202-215.	1.3	5
11	Identification of the 5-HT1A serotonin receptor as a novel therapeutic target in a C. elegans model of Machado-Joseph disease. Neurobiology of Disease, 2021, 152, 105278.	2.1	7
12	Case Report: A Novel GNB1 Mutation Causes Global Developmental Delay With Intellectual Disability and Behavioral Disorders. Frontiers in Neurology, 2021, 12, 735549.	1.1	9
13	Impact of rapeseed pomace extract on markers of oxidative stress and DNA damage in human SHâ€“SY5Y cells. Journal of Food Biochemistry, 2021, 45, e13592.	1.2	2
14	Preclinical Assessment of Mesenchymal-Stem-Cell-Based Therapies in Spinocerebellar Ataxia Type 3. Biomedicines, 2021, 9, 1754.	1.4	5
15	Loss of egli-1, the Caenorhabditis elegans Orthologue of a Downstream Target of SMN, Leads to Abnormalities in Sensorimotor Integration. Molecular Neurobiology, 2020, 57, 1553-1569.	1.9	2
16	Polyglutamine spinocerebellar ataxias: emerging therapeutic targets. Expert Opinion on Therapeutic Targets, 2020, 24, 1099-1119.	1.5	8
17	Data on the effects of Hyptis spp. and Lycium spp. plant extracts in C. elegans models of genetically determined neurodegenerative diseases. Data in Brief, 2020, 33, 106598.	0.5	1
18	Transcriptional consequences of MBD5 disruption in mouse brain and CRISPR-derived neurons. Molecular Autism, 2020, 11, 45.	2.6	11

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19	Deficiency in classical nonhomologous end-joiningâ€“mediated repair of transcribed genes is linked to SCA3 pathogenesis. Proceedings of the National Academy of Sciences of the United States of America, 2020, 117, 8154-8165.	3.3	28
20	In Vivo Molecular Signatures of Cerebellar Pathology in Spinocerebellar Ataxia Type 3. Movement Disorders, 2020, 35, 1774-1786.	2.2	23
21	Positive allosteric modulation of indoleamine 2,3-dioxygenase 1 restrains neuroinflammation. Proceedings of the National Academy of Sciences of the United States of America, 2020, 117, 3848-3857.	3.3	58
22	Large-Scale Exome Sequencing Study Implicates Both Developmental and Functional Changes in the Neurobiology of Autism. Cell, 2020, 180, 568-584.e23.	13.5	1,422
23	Unravelling the anticancer potential of functionalized chromeno[2,3-b]pyridines for breast cancer treatment. Bioorganic Chemistry, 2020, 100, 103942.	2.0	20
24	Genomic imbalances defining novel intellectual disability associated loci. Orphanet Journal of Rare Diseases, 2019, 14, 164.	1.2	3
25	From Pathogenesis to Novel Therapeutics for Spinocerebellar Ataxia Type 3: Evading Potholes on the Way to Translation. Neurotherapeutics, 2019, 16, 1009-1031.	2.1	42
26	GST-4-Dependent Suppression of Neurodegeneration in C. elegans Models of Parkinsonâ€™s and Machado-Joseph Disease by Rapeseed Pomace Extract Supplementation. Frontiers in Neuroscience, 2019, 13, 1091.	1.4	36
27	The Role of AKT3 Copy Number Changes in Brain Abnormalities and Neurodevelopmental Disorders: Four New Cases and Literature Review. Frontiers in Genetics, 2019, 10, 58.	1.1	7
28	Neurotoxic effects of MPTP on mouse cerebral cortex: Modulation of neuroinflammation as a neuroprotective strategy. Molecular and Cellular Neurosciences, 2019, 96, 1-9.	1.0	22
29	Citalopram Reduces Aggregation of ATXN3 in a YAC Transgenic Mouse Model of Machado-Joseph Disease. Molecular Neurobiology, 2019, 56, 3690-3701.	1.9	24
30	Preclinical Evidence Supporting Early Initiation of Citalopram Treatment in Machado-Joseph Disease. Molecular Neurobiology, 2019, 56, 3626-3637.	1.9	18
31	Neuroprotective Effects of Creatine in the CMVMJD135 Mouse Model of Spinocerebellar Ataxia Type 3. Movement Disorders, 2018, 33, 815-826.	2.2	26
32	Tauroursodeoxycholic Acid Improves Motor Symptoms in a Mouse Model of Parkinsonâ€™s Disease. Molecular Neurobiology, 2018, 55, 9139-9155.	1.9	55
33	Pharmacological Therapies for Machado-Joseph Disease. Advances in Experimental Medicine and Biology, 2018, 1049, 369-394.	0.8	17
34	The contribution of 7q33 copy number variations for intellectual disability. Neurogenetics, 2018, 19, 27-40.	0.7	3
35	Revalorisation of rapeseed pomace extracts: An in vitro study into its anti-oxidant and DNA protective properties. Food Chemistry, 2018, 239, 323-332.	4.2	25
36	Identification of rare de novo epigenetic variations in congenital disorders. Nature Communications, 2018, 9, 2064.	5.8	82

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37	A novel microduplication of ARID1B: Clinical, genetic, and proteomic findings. American Journal of Medical Genetics, Part A, 2017, 173, 2478-2484.	0.7	7
38	Discovery of Therapeutic Approaches for Polyglutamine Diseases: A Summary of Recent Efforts. Medicinal Research Reviews, 2017, 37, 860-906.	5.0	17
39	Whole Gene Deletion of EBF3 Supporting Haploinsufficiency of This Gene as a Mechanism of Neurodevelopmental Disease. Frontiers in Genetics, 2017, 8, 143.	1.1	17
40	Altered striatal endocannabinoid signaling in a transgenic mouse model of spinocerebellar ataxia type-3. PLoS ONE, 2017, 12, e0176521.	1.1	6
41	Study into the polyphenol content and antioxidant activity of rapeseed pomace extracts. Proceedings of the Nutrition Society, 2016, 75, .	0.4	4
42	Dysregulation of the endocannabinoid signaling system in the cerebellum and brainstem in a transgenic mouse model of spinocerebellar ataxia type-3. Neuroscience, 2016, 339, 191-209.	1.1	22
43	Combined therapy with m-TOR-dependent and -independent autophagy inducers causes neurotoxicity in a mouse model of Machadoâ€“Joseph disease. Neuroscience, 2016, 313, 162-173.	1.1	25
44	Identification of novel genetic causes of Rett syndrome-likephenotypes. Journal of Medical Genetics, 2016, 53, 190-199.	1.5	158
45	Recurrent copy number variations as risk factors for neurodevelopmental disorders: critical overview and analysis of clinical implications. Journal of Medical Genetics, 2016, 53, 73-90.	1.5	90
46	ISDN2014_0322: REMOVED: Identification of novel genetic causes of Rett syndromeâ€like phenotypes by whole exome sequencing. International Journal of Developmental Neuroscience, 2015, 47, 99-99.	0.7	0
47	Redefining the MED13L syndrome. European Journal of Human Genetics, 2015, 23, 1308-1317.	1.4	53
48	The Role of the Mammalian DNA End-processing Enzyme Polynucleotide Kinase 3â€™-Phosphatase in Spinocerebellar Ataxia Type 3 Pathogenesis. PLoS Genetics, 2015, 11, e1004749.	1.5	84
49	Inactivation of PNKP by Mutant ATXN3 Triggers Apoptosis by Activating the DNA Damage-Response Pathway in SCA3. PLoS Genetics, 2015, 11, e1004834.	1.5	69
50	Novel candidate bloodâ€based transcriptional biomarkers of machadoâ€joseph disease. Movement Disorders, 2015, 30, 968-975.	2.2	28
51	Institutionalization and indiscriminate social behavior: Differential-susceptibility versus diathesis-stress models for the 5-HTTLPR and BDNF genotypes. Physiology and Behavior, 2015, 152, 85-91.	1.0	7
52	Variant Rett syndrome in a girl with a pericentric Xâ€chromosome inversion leading to epigenetic changes and overexpression of the MECP2 gene. International Journal of Developmental Neuroscience, 2015, 46, 82-87.	0.7	11
53	Serotonergic signalling suppresses ataxin 3 aggregation and neurotoxicity in animal models of Machado-Joseph disease. Brain, 2015, 138, 3221-3237.	3.7	74
54	ISDN2014_0325: Lack of H3K4 demethylase rbrâ€2/KDM5C leads to GABAâ€related behavioral defects in C. elegans. International Journal of Developmental Neuroscience, 2015, 47, 100-100.	0.7	0

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55	Dominant negative effect of polyglutamine expansion perturbs normal function of ataxin-3 in neuronal cells. Human Molecular Genetics, 2015, 24, 100-117.	1.4	26
56	Differential mtDNA Damage Patterns in a Transgenic Mouse Model of Machadoâ€“Joseph Disease (MJD/SCA3). Journal of Molecular Neuroscience, 2015, 55, 449-453.	1.1	19
57	Limited Effect of Chronic Valproic Acid Treatment in a Mouse Model of Machado-Joseph Disease. PLoS ONE, 2015, 10, e0141610.	1.1	22
58	The disruption of proteostasis in neurodegenerative disorders. AIMS Molecular Science, 2015, 2, 259-293.	0.3	2
59	Chronic Treatment with 17-DMAG Improves Balance and Coordination in A New Mouse Model of Machado-Joseph Disease. Neurotherapeutics, 2014, 11, 433-449.	2.1	86
60	Lithium Chloride Therapy Fails to Improve Motor Function in a Transgenic Mouse Model of Machado-Joseph Disease. Cerebellum, 2014, 13, 713-727.	1.4	25
61	Phenotypic and functional consequences of haploinsufficiency of genes from exocyst and retinoic acid pathway due to a recurrent microdeletion of 2p13.2. Orphanet Journal of Rare Diseases, 2013, 8, 100.	1.2	24
62	Coffin-Siris Syndrome and the BAF Complex: Genotype-Phenotype Study in 63 Patients. Human Mutation, 2013, 34, 1519-1528.	1.1	178
63	Using C. elegans to Decipher the Cellular and Molecular Mechanisms Underlying Neurodevelopmental Disorders. Molecular Neurobiology, 2013, 48, 465-489.	1.9	32
64	Patterns of Mitochondrial DNA Damage in Blood and Brain Tissues of a Transgenic Mouse Model of Machado-Joseph Disease. Neurodegenerative Diseases, 2013, 11, 206-214.	0.8	55
65	The rs5743836 polymorphism in TLR9 confers a population-based increased risk of non-Hodgkin lymphoma. Genes and Immunity, 2012, 13, 197-201.	2.2	35
66	Sequence Analysis of 5â€² Regulatory Regions of the Machadoâ€“Joseph Disease Gene (ATXN3). Cerebellum, 2012, 11, 1045-1050.	1.4	7
67	Role of the ubiquitinâ€“proteasome system in nervous system function and disease: using C. elegans as a dissecting tool. Cellular and Molecular Life Sciences, 2012, 69, 2691-2715.	2.4	22
68	Rett syndrome with and without detected MECP2 mutations: An attempt to redefine phenotypes. Brain and Development, 2011, 33, 69-76.	0.6	24
69	An Image Processing Application for Quantification of Protein Aggregates in Caenorhabditis Elegans. Advances in Intelligent and Soft Computing, 2011, , 31-38.	0.2	2
70	Neuron-specific proteotoxicity of mutant ataxin-3 in C. elegans : rescue by the DAF-16 and HSF-1 pathways. Human Molecular Genetics, 2011, 20, 2996-3009.	1.4	101
71	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.	4.9	33
72	Absence of Ataxin-3 Leads to Enhanced Stress Response in C. elegans. PLoS ONE, 2011, 6, e18512.	1.1	26

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73	Increased transcript diversity: novel splicing variants of Machadoâ€“Joseph Disease gene (ATXN3). Neurogenetics, 2010, 11, 193-202.	0.7	37
74	Motor uncoordination and neuropathology in a transgenic mouse model of Machadoâ€“Joseph disease lacking intranuclear inclusions and ataxin-3 cleavage products. Neurobiology of Disease, 2010, 40, 163-176.	2.1	62
75	Absence of ataxin-3 leads to cytoskeletal disorganization and increased cell death. Biochimica Et Biophysica Acta - Molecular Cell Research, 2010, 1803, 1154-1163.	1.9	42
76	The (CAG)n tract of Machadoâ€“Joseph Disease gene (ATXN3): a comparison between DNA and mRNA in patients and controls. European Journal of Human Genetics, 2010, 18, 621-623.	1.4	21
77	Ataxin-3 Plays a Role in Mouse Myogenic Differentiation through Regulation of Integrin Subunit Levels. PLoS ONE, 2010, 5, e11728.	1.1	25
78	Familial Aggregation of Maxillary Lateral Incisor Agenesis. Journal of Dental Research, 2010, 89, 621-625.	2.5	26
79	Mutational analysis of MSX1 and PAX9 genes in Portuguese families with maxillary lateral incisor agenesis. European Journal of Orthodontics, 2010, 32, 582-588.	1.1	25
80	[P1.85]: Variant Rett syndrome in a girl with a pericentric Xâ€chromosome inversion leading to overexpression of the MECP2 gene. International Journal of Developmental Neuroscience, 2010, 28, 683-684.	0.7	0
81	Monoamine deficits in the brain of methyl-CpG binding protein 2 null mice suggest the involvement of the cerebral cortex in early stages of Rett syndrome. Neuroscience, 2010, 170, 453-467.	1.1	40
82	Developmental disturbances associated with agenesis of the permanent maxillary lateral incisor. British Dental Journal, 2009, 207, E25-E25.	0.3	27
83	Mutations in the MECP2 Gene Are Not a Major Cause of Rett Syndrome-Like or Related Neurodevelopmental Phenotype in Male Patients. Journal of Child Neurology, 2009, 24, 49-55.	0.7	11
84	Evaluation of CSF neurotransmitters and folate in 25 patients with Rett disorder and effects of treatment. Brain and Development, 2009, 31, 46-51.	0.6	42
85	ATX-3, CDC-48 and UBXN-5: A new trimolecular complex in Caenorhabditis elegans. Biochemical and Biophysical Research Communications, 2009, 386, 575-581.	1.0	13
86	Nucleocytoplasmic Shuttling Activity of Ataxin-3. PLoS ONE, 2009, 4, e5834.	1.1	40
87	Segregation distortion of wild-type alleles at the Machado-Joseph disease locus: a study in normal families from the Azores islands (Portugal). Journal of Human Genetics, 2008, 53, 333-339.	1.1	25
88	Atypical stereotypies and vocal tics in Rett syndrome: An illustrative case. Movement Disorders, 2008, 23, 622-624.	2.2	7
89	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.	2.2	70
90	T-1237C polymorphism of TLR9 gene is not associated with multiple sclerosis in the Portuguese population. Multiple Sclerosis Journal, 2008, 14, 550-552.	1.4	3

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91	The C677T Polymorphism inMTHFRIs Not Associated with Migraine in Portugal. Disease Markers, 2008, 25, 107-113.	0.6	25
92	Analysis of Highly Conserved Regions of the 3â€™UTR ofMECP2Gene in Patients with Clinical Diagnosis of Rett Syndrome and Other Disorders Associated with Mental Retardation. Disease Markers, 2008, 24, 319-324.	0.6	8
93	Functional genomics and biochemical characterization of the C. elegans orthologue of the Machadoâ€Joseph disease protein ataxinâ€3. FASEB Journal, 2007, 21, 1126-1136.	0.2	62
94	Neurodevelopment milestone abnormalities in rats exposed to stress in early life. Neuroscience, 2007, 147, 1022-1033.	1.1	67
95	Study of disease-relevant polymorphisms in the TLR4 and TLR9 genes: A novel method applied to the analysis of the Portuguese population. Molecular and Cellular Probes, 2007, 21, 316-320.	0.9	24
96	Stereotypies in Rett syndrome: Analysis of 83 patients with and without detected MECP2 mutations. Neurology, 2007, 68, 1183-1187.	1.5	78
97	MECP2 coding sequence and 3â€²UTR variation in 172 unrelated autistic patients. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2007, 144B, 475-483.	1.1	53
98	Abnormal movements in Rett syndrome are present before the regression period: A case study. Movement Disorders, 2007, 22, 2284-2287.	2.2	28
99	An explanation for another familial case of Rett syndrome: maternal germline mosaicism. European Journal of Human Genetics, 2007, 15, 902-904.	1.4	31
100	Evidence for abnormal early development in a mouse model of Rett syndrome. Genes, Brain and Behavior, 2007, 6, 277-286.	1.1	84
101	NEDD8: A new ataxin-3 interactor. Biochimica Et Biophysica Acta - Molecular Cell Research, 2007, 1773, 1619-1627.	1.9	55
102	Corrigendum to â€œLinkage disequilibrium screening for multiple sclerosis implicates JAG1 and POU2AF1 as susceptibility genes in Europeansâ€•[J. Neuroimmunol. 179 (2006) 108â€“116]. Journal of Neuroimmunology, 2007, 189, 175-176.	1.1	1
103	APOE epsilon variation in multiple sclerosis susceptibility and disease severity: Some answers. Neurology, 2006, 66, 1373-1383.	1.5	80
104	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.	1.1	29
105	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.	1.1	26
106	Chromatin remodeling and neuronal function: exciting links. Genes, Brain and Behavior, 2006, 5, 80-91.	1.1	8
107	Linkage disequilibrium screening for multiple sclerosis implicates JAG1 and POU2AF1 as susceptibility genes in Europeans. Journal of Neuroimmunology, 2006, 179, 108-116.	1.1	29
108	Detection of heterozygous deletions and duplications in the MECP2 gene in Rett syndrome by Robust Dosage PCR (RD-PCR). Human Mutation, 2005, 25, 505-505.	1.1	26

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109	Nonsense mutation in TITF1 in a Portuguese family with benign hereditary chorea. Neurogenetics, 2005, 6, 209-215.	0.7	59
110	Population Genetics of Wild-Type CAG Repeats in the Machado-Joseph Disease Gene in Portugal. Human Heredity, 2005, 60, 156-163.	0.4	43
111	Neuroferritinopathy: Missense mutation in FTL causing early-onset bilateral pallidal involvement. Neurology, 2005, 65, 603-605.	1.5	112
112	Developmental absence of maxillary lateral incisors in the Portuguese population. European Journal of Orthodontics, 2005, 27, 443-449.	1.1	87
113	Towards a Structural Understanding of the Fibrillization Pathway in Machado-Joseph's Disease: Trapping Early Oligomers of Non-expanded Ataxin-3. Journal of Molecular Biology, 2005, 353, 642-654.	2.0	68
114	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.	1.4	27
115	Genomic structure, promoter activity, and developmental expression of the mouse homologue of the Machadoâ€"Joseph disease (MJD) geneâ˜†. Genomics, 2004, 84, 361-373.	1.3	26
116	A whole genome screen for association with multiple sclerosis in Portuguese patients. Journal of Neuroimmunology, 2003, 143, 112-115.	1.1	13
117	Genetic interaction ofCTLA-4 with HLA-DR15 in multiple sclerosis patients. Annals of Neurology, 2003, 54, 119-122.	2.8	46
118	Molecular diagnosis of Huntington disease in Portugal: implications for genetic counselling and clinical practice. European Journal of Human Genetics, 2003, 11, 872-878.	1.4	18
119	Inherited and acquired risk factors and their combined effects in pediatric stroke. Pediatric Neurology, 2003, 28, 134-138.	1.0	64
120	Single-Tube Method for Determination of F508del Genotype in the CFTR Gene Using Bidirectional PCR Amplification of Specific Alleles. BioTechniques, 2003, 34, 460-462.	0.8	1
121	Identification of three novel polymorphisms in the MJD1 gene and study of their frequency in the Portuguese population. Journal of Human Genetics, 2002, 47, 205-207.	1.1	5
122	Ancestral Origins of the Machado-Joseph Disease Mutation: A Worldwide Haplotype Study. American Journal of Human Genetics, 2001, 68, 523-528.	2.6	118
123	The genomic structure and expression of MJD, the Machado-Joseph disease gene. Journal of Human Genetics, 2001, 46, 413-422.	1.1	66
124	Improvement in the Molecular Diagnosis of Machado-Joseph Disease. Archives of Neurology, 2001, 58, 1821.	4.9	121
125	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.	2.6	79
126	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.	1.4	31

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127	Prenatal diagnosis of Machadoâ€“Joseph disease by direct mutation analysis. , 1998, 18, 611-617.		17
128	Analysis of SCA1, DRPLA, MJD, SCA2, and SCA6 CAG repeats in 48 portuguese ataxia families. , 1998, 81, 134-138.		57
129	Restriction Map of a YAC and Cosmid Contig Encompassing the Oculopharyngeal Muscular Dystrophy Candidate Region on Chromosome 14q11.2â€“q13. Genomics, 1998, 52, 201-204.	1.3	9
130	Machadoâ€“Joseph disease gene products carrying different carboxyl termini. Neuroscience Research, 1997, 28, 373-377.	1.0	73
131	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.	2.6	20
132	No association between chromosome-18 markers and lithiumresponsive affective disorders. Psychiatry Research, 1996, 63, 17-23.	1.7	51
133	Limits of Clinical Assessment in the Accurate Diagnosis of Machado-Joseph Disease. Archives of Neurology, 1996, 53, 1168-1174.	4.9	29
134	Mutation Detection in Machado-Joseph Disease Using Repeat Expansion Detection. Molecular Medicine, 1996, 2, 77-85.	1.9	19
135	Linkage disequilibrium analysis in Machado-Joseph disease patients of different ethnic origins. Human Genetics, 1996, 98, 620-624.	1.8	24
136	Somatic mosaicism in the central nervous system in spinocerebellar ataxia type 1 and machado-joseph disease. Annals of Neurology, 1996, 40, 199-206.	2.8	59
137	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.	1.5	106
138	A familial factor independent of CAG repeat length influences age at onset of Machado-Joseph disease. American Journal of Human Genetics, 1996, 59, 119-27.	2.6	38
139	Mutation detection in Machado-Joseph disease using repeat expansion detection. Molecular Medicine, 1996, 2, 77-85.	1.9	4
140	Gender equality in Machadoâ€“Joseph disease. Nature Genetics, 1995, 11, 118-119.	9.4	12
141	Correlation between CAG repeat length and clinical features in Machado-Joseph disease. American Journal of Human Genetics, 1995, 57, 54-61.	2.6	208
142	Genetic Linkage Studies of Machado-Joseph Disease with Chromosome 14q STRPs in 16 Portuguese-Azorean Kindreds. Genomics, 1994, 21, 645-648.	1.3	34
143	Molecular Genetics of Intellectual Disability. , 0, , .		3
144	Large-Scale Exome Sequencing Study Implicates Both Developmental and Functional Changes in the Neurobiology of Autism. SSRN Electronic Journal, 0, , .	0.4	12