Salmo Raskin

List of Publications by Year
in descending order

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Version: 2024-02-01

116 papers	3,885 citations	159585 30 h-index	138484 58 g-index
118 all docs	118 docs citations	118 times ranked	6696 citing authors

#	Article	IF	Citations
1	Novel OPN1LW/OPN1MW Exon 3 Haplotype-Associated Splicing Defect in Patients with X-Linked Cone Dysfunction. International Journal of Molecular Sciences, 2022, 23, 6868.	4.1	3
2	IDENTIFICAÃ‡ÃƒO DE DELEÃ‡ÃƒO TERMINAL 3P EM PACIENTE COM TRANSTORNO DO ESPECTRO AUTISTA. , 2022, 79, 1660.		0
3	DEFICIÃŠNCIA DA GLICOSE-6-FOSFATO DESIDROGENASE EM PACIENTE COM TRANSTORNO DO ESPECTRO AUTISTA. , 2022, 79, 1658.		0
4	Genetics of COVID-19. Jornal De Pediatria, 2021, 97, 378-386.	2.0	17
5	Balance and physical functioning in Spinocerebellar ataxias 3 and 10. Acta Neurologica Scandinavica, 2021, 143, 458-463.	2.1	4
6	Bi-allelic variants in HOPS complex subunit VPS41 cause cerebellar ataxia and abnormal membrane trafficking. Brain, 2021, 144, 769-780.	7.6	33
7	Analysis of diffusion tensor parameters in spinocerebellar ataxia type 3 and type 10 patients. Parkinsonism and Related Disorders, 2020, 78, 73-78.	2.2	5
8	Is Ataxia an Underestimated Symptom of Huntington's Disease?. Frontiers in Neurology, 2020, 11, 571843.	2.4	11
9	Volumetric MRI Changes in Spinocerebellar Ataxia (SCA3 and SCA10) Patients. Cerebellum, 2020, 19, 536-543.	2.5	21
10	Comment on: Diagnosis of Aicardiâ€GoutiÃ¨res Syndrome in Adults. Movement Disorders Clinical Practice, 2020, 7, 583-584.	1.5	0
11	A comprehensive analysis of AHRR gene as a candidate for cleft lip with or without cleft palate. Mutation Research - Reviews in Mutation Research, 2020, 785, 108319.	5.5	5
12	Cerebellar and thalamic degeneration in spinocerebellar ataxia type 10. Parkinsonism and Related Disorders, 2020, 76, 76-77.	2.2	3
13	Evidence and practices of the use of next generation sequencing in patients with undiagnosed autosomal dominant cerebellar ataxias: a review. Arquivos De Neuro-Psiquiatria, 2020, 78, 576-585.	0.8	3
14	Olfactory Function in SCA10. Cerebellum, 2019, 18, 85-90.	2.5	11
15	Clinical and Genetic Evaluation of Spinocerebellar Ataxia Type 10 in 16 Brazilian Families. Cerebellum, 2019, 18, 849-854.	2.5	7
16	Spinocerebellar ataxias in Southern Brazil: Genotypic and phenotypic evaluation of 213 families. Clinical Neurology and Neurosurgery, 2019, 184, 105427.	1.4	13
17	Hyposkillia and spanophilia in the movement disorders rounds. Movement Disorders, 2019, 34, 1399-1399.	3.9	0
18	Abnormal Findings in Polysomnographic Recordings of Patients with Spinocerebellar Ataxia Type 2 (SCA2). Cerebellum, 2019, 18, 196-202.	2.5	6

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19	Spinocerebellar ataxias type 3 and 10: Onset and progression of ataxia during pregnancy and puerperium. Parkinsonism and Related Disorders, 2018, 52, 119-120.	2.2	2
20	Sleep disorders in spinocerebellar ataxia type 10. Journal of Sleep Research, 2018, 27, e12688.	3.2	7
21	Targeted knockout of a chemokine-like gene increases anxiety and fear responses. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, E1041-E1050.	7.1	39
22	Different Cerebellar Ataxia Phenotypes Associated with Mutations of the PNPLA6 Gene in Brazilian Patients with Recessive Ataxias. Cerebellum, 2018, 17, 380-385.	2.5	20
23	A randomized placebo-controlled phase 3 trial of an antisense oligonucleotide, drisapersen, in Duchenne muscular dystrophy. Neuromuscular Disorders, 2018, 28, 4-15.	0.6	102
24	Congenital bilateral absence of the vas deferens as an atypical form of cystic fibrosis: reproductive implications and genetic counseling. Andrology, 2018, 6, 127-135.	3.5	94
25	Dystonia in Patients with Spinocerebellar Ataxia 3 - Machado-Joseph disease: An Underestimated Diagnosis?. The Open Neurology Journal, 2018, 12, 41-49.	0.4	6
26	p.Phe508del, p.Gly542X, p.Arg1162X, p.Asn1303Lys, and p.Lys683serfsX38 mutations in CF newborn screening of Brazilian children. Clinical Genetics, 2017, 92, 115-116.	2.0	0
27	SLC13A5is the second gene associated with KohlschÃ¼tterâ€“TÃ¶nz syndrome. Journal of Medical Genetics, 2017, 54, 54-62.	3.2	45
28	Cystic fibrosis in Afro-Brazilians: XK haplotypes analysis supports the European origin of p.F508del mutation. Genetica, 2017, 145, 19-25.	1.1	0
29	Nonmotor Symptoms in Patients with Spinocerebellar Ataxia Type 10. Cerebellum, 2017, 16, 938-944.	2.5	23
30	The genomic landscape of balanced cytogenetic abnormalities associated with human congenital anomalies. Nature Genetics, 2017, 49, 36-45.	21.4	251
31	A 10.43 Mb duplication of chromosome region 5q31.2-q32 associated with a general delay in psychomotor development. Meta Gene, 2017, 11, 169-171.	0.6	1
32	ItajaÃ; Santa Catarina â€“ Azorean ancestry and spinocerebellar ataxia type 3. Arquivos De Neuro-Psiquiatria, 2016, 74, 858-860.	0.8	6
33	Comparison of non-motors symptoms in patients with spinocerebellar ataxia type 10 and type 3. Parkinsonism and Related Disorders, 2016, 22, e150.	2.2	1
34	High Incidence of Noonan Syndrome Features Including Short Stature and Pulmonic Stenosis in Patients carrying NF1 Missense Mutations Affecting p.Arg1809: Genotypeâ€“Phenotype Correlation. Human Mutation, 2015, 36, 1052-1063.	2.5	143
35	Spinocerebellar ataxia type 10 in the South of Brazil: the Amerindian-Belgian connection. Arquivos De Neuro-Psiquiatria, 2015, 73, 725-727.	0.8	10
36	Diffusion tensor imaging and tract-based spatial statistics analysis in Friedreich's ataxia patients. Parkinsonism and Related Disorders, 2015, 21, 504-508.	2.2	27

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37	Ataxia-telangiectasia â€" A historical review and a proposal for a new designation: ATM syndrome. Journal of the Neurological Sciences, 2015, 355, 3-6.	0.6	80
38	PIAS4 is associated with macro/microcephaly in the novel interstitial 19p13.3 microdeletion/microduplication syndrome. European Journal of Human Genetics, 2015, 23, 1615-1626.	2.8	29
39	A NewELOVL4Mutation in a Case of Spinocerebellar Ataxia With Erythrokeratodermia. JAMA Neurology, 2015, 72, 942.	9.0	34
40	When should we test patients with familial ataxias for SCA31? A misdiagnosed condition outside Japan?. Journal of the Neurological Sciences, 2015, 355, 206-208.	0.6	11
41	Spinocerebellar ataxia type 3: subphenotypes in a cohort of brazilian patients. Arquivos De Neuro-Psiquiatria, 2014, 72, 659-662.	0.8	24
42	Cervical dystonia: about familial and sporadic cases in 88 patients. Arquivos De Neuro-Psiquiatria, 2014, 72, 107-113.	0.8	7
43	Niemann-Pick disease type C: a case series of Brazilian patients. Arquivos De Neuro-Psiquiatria, 2014, 72, 214-218.	0.8	3
44	â€˜Pseudoâ€Dominantâ€™ Inheritance in Friedreich's Ataxia: Clinical and Genetic Study of a Brazilian Family. Movement Disorders Clinical Practice, 2014, 1, 361-363.	1.5	2
45	Movement Disorders in Spinocerebellar Ataxias in a Cohort of Brazilian Patients. European Neurology, 2014, 72, 360-362.	1.4	15
46	<scp>SPG4</scp>â€related hereditary spastic paraplegia: frequency and mutation spectrum in Brazil. Clinical Genetics, 2014, 86, 194-196.	2.0	10
47	Repeat interruptions in spinocerebellar ataxia type 10 expansions are strongly associated with epileptic seizures. Neurogenetics, 2014, 15, 59-64.	1.4	51
48	A 1.5Mb terminal deletion of 12p associated with autism spectrum disorder. Gene, 2014, 542, 83-86.	2.2	21
49	A Noncoding Expansion in EIF4A3 Causes Richieri-Costa-Pereira Syndrome, a Craniofacial Disorder Associated with Limb Defects. American Journal of Human Genetics, 2014, 94, 120-128.	6.2	99
50	Adult onset Alexander disease presenting with progressive spastic paraplegia. Parkinsonism and Related Disorders, 2014, 20, 241-242.	2.2	4
51	Genetic evaluation for TOR1-A (DYT1) in Brazilian patients with dystonia. Arquivos De Neuro-Psiquiatria, 2014, 72, 753-756.	0.8	2
52	KohlschÃ¼tter-TÃ¶nz syndrome in siblings without ROGDI mutation. Oral Health and Dental Management, 2014, 13, 728-30.	0.7	4
53	KohlschÃ¼tter-TÃ¶nz Syndrome: Mutations inROGDIand Evidence of Genetic Heterogeneity. Human Mutation, 2013, 34, 296-300.	2.5	24
54	Spinal muscular atrophy due to a â€œde novoâ€•1.3Mb deletion: Implication for genetic counseling. Neuromuscular Disorders, 2013, 23, 388-390.	0.6	1

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55	Paradoxical effects of repeat interruptions on spinocerebellar ataxia type 10 expansions and repeat instability. European Journal of Human Genetics, 2013, 21, 1272-1276.	2.8	35
56	Wilson's disease in Southern Brazil: genotype-phenotype correlation and description of two novel mutations in ATP7B gene. Arquivos De Neuro-Psiquiatria, 2013, 71, 503-507.	0.8	19
57	Diastrophic dysplasia: prenatal diagnosis and review of the literature. Sao Paulo Medical Journal, 2013, 131, 127-132.	0.9	12
58	Clinical relevance of "bulging eyes" for the differential diagnosis of spinocerebellar ataxias. Arquivos De Neuro-Psiquiatria, 2013, 71, 428-430.	0.8	18
59	Acute onset of cerebellar ataxia in a spinocerebellar ataxia type 10 patient after use of steroids. Arquivos De Neuro-Psiquiatria, 2013, 71, 66-66.	0.8	4
60	Folic acid for the prevention of neural tube defects. Revista Brasileira De Ginecologia E Obstetricia, 2013, 35, 287-289.	0.8	7
61	Epileptic encephalopathy and atypical Rett syndrome with mutations in CDKL5: clinical and molecular characterization of two Brazilian patients. Arquivos De Neuro-Psiquiatria, 2013, 71, 414-415.	0.8	0
62	Phenotypic Heterogeneity of Genomic Disorders and Rare Copy-Number Variants. New England Journal of Medicine, 2012, 367, 1321-1331.	27.0	519
63	Spinocerebellar ataxias â€“ genotype-phenotype correlations in 104 Brazilian families. Clinics, 2012, 67, 443-449.	1.5	56
64	Cerebellar ataxia as the first manifestation of Alexander's disease. Arquivos De Neuro-Psiquiatria, 2012, 70, 309-310.	0.8	6
65	What can be done when asymptomatic patients discover they have Brugada syndrome? A case report of Brugada syndrome. International Journal of Cardiology, 2011, 150, e96-e97.	1.7	3
66	Symptom onset of spinocerebellar ataxia type 10 in pregnancy and puerperium. Journal of Clinical Neuroscience, 2011, 18, 437-438.	1.5	9
67	Spinocerebellar ataxia type 10 â€“ A review. Parkinsonism and Related Disorders, 2011, 17, 655-661.	2.2	73
68	Tremor in X-linked recessive spinal and bulbar muscular atrophy (Kennedy's disease). Clinics, 2011, 66, 955-957.	1.5	12
69	Spinocerebellar ataxia type 10: Frequency of epilepsy in a large sample of Brazilian patients. Movement Disorders, 2010, 25, 2875-2878.	3.9	36
70	Olfactory heterogeneity in LRRK2 related Parkinsonism. Movement Disorders, 2010, 25, 2879-2883.	3.9	33
71	CÃªtis laxa: relato de caso. Anais Brasileiros De Dermatologia, 2010, 85, 684-686.	1.1	7
72	Inactivation of hnRNP K by Expanded Intronic AUUCU Repeat Induces Apoptosis Via Translocation of PKCÎ´ to Mitochondria in Spinocerebellar Ataxia 10. PLoS Genetics, 2010, 6, e1000984.	3.5	102

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73	CFTR allelic heterogeneity in Brazil: historical and geographical perspectives and implications for screening and counseling for cystic fibrosis in this country. Journal of Human Genetics, 2010, 55, 71-76.	2.3	16
74	CTA/CTG expansions at the SCA 8 locus in multiple system atrophy. Clinical Neurology and Neurosurgery, 2009, 111, 208-210.	1.4	17
75	The G2019S LRRK2 mutation in Brazilian patients with Parkinson's disease: Phenotype in monozygotic twins. Movement Disorders, 2008, 23, 290-294.	3.9	20
76	Incidence of cystic fibrosis in five different states of Brazil as determined by screening of p.F508del, mutation at the CFTR gene in newborns and patients. Journal of Cystic Fibrosis, 2008, 7, 15-22.	0.7	57
77	Spinocerebellar ataxia type 6 in Brazil. Arquivos De Neuro-Psiquiatria, 2008, 66, 691-694.	0.8	15
78	Reduced Penetrance in a Brazilian Family With Spinocerebellar Ataxia Type 10. Archives of Neurology, 2007, 64, 591.	4.5	42
79	Cystic Fibrosis Gene Variability in Two Southern Brazilian Amerindian Populations: Analysis of the Î”F508 Mutation and the KM19 and XV2C Haplotypes. Human Biology, 2007, 79, 79-91.	0.2	1
80	The history of spinocerebellar ataxia type 10 in Brazil: travels of a gene. Arquivos De Neuro-Psiquiatria, 2007, 65, 965-968.	0.8	6
81	Rett syndrome: clinical and molecular characterization of two Brazilian patients. Arquivos De Neuro-Psiquiatria, 2007, 65, 36-40.	0.8	6
82	Non-choreic movement disorders as initial manifestations of Huntington's disease. Arquivos De Neuro-Psiquiatria, 2007, 65, 402-405.	0.8	15
83	Cystic fibrosis in a southern Brazilian population: characteristics of 90% of the alleles. Clinical Genetics, 2007, 72, 218-223.	2.0	13
84	Atypical parkinsonism and SCA8. Parkinsonism and Related Disorders, 2006, 12, 191-192.	2.2	6
85	Clinical and genetic study of a Brazilian family with spastic paraplegia (SPG6 locus). Movement Disorders, 2006, 21, 279-281.	3.9	22
86	Expansion of the genotypic and phenotypic spectrum in patients with KRAS germline mutations. Journal of Medical Genetics, 2006, 44, 131-135.	3.2	170
87	Phenotypic consequences of genetic variation at hemizygous alleles: Sotos syndrome is a contiguous gene syndrome incorporating coagulation factor twelve (FXII) deficiency. Genetics in Medicine, 2005, 7, 479-483.	2.4	44
88	Three cases with rare interstitial rearrangements of chromosome 1 characterized by multicolor banding. Cytogenetic and Genome Research, 2005, 111, 171-174.	1.1	14
89	Kennedy's disease phenotype with positive genetic study for Kugelberg-Welander's disease: case report. Arquivos De Neuro-Psiquiatria, 2005, 63, 330-331.	0.8	1
90	Leber's hereditary optic neuropathy: case report and literature review. Sao Paulo Medical Journal, 2004, 122, 276-279.	0.9	2

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91	Cerebrotendinous xanthomatosis: report of two Brazilian brothers. Arquivos De Neuro-Psiquiatria, 2004, 62, 1085-1089.	0.8	10
92	Report of cluster headache in a pair of monozygous twins. Journal of Headache and Pain, 2004, 5, 140-143.	6.0	2
93	De novoSCN1Amutations are a major cause of severe myoclonic epilepsy of infancy. Human Mutation, 2003, 21, 615-621.	2.5	170
94	How much phenotypic variation can be attributed toparkingenotype?. Annals of Neurology, 2003, 54, 176-185.	5.3	271
95	Global genetic variation at nine short tandem repeat loci and implications on forensic genetics. European Journal of Human Genetics, 2003, 11, 39-49.	2.8	37
96	High Allelic Heterogeneity Between Afro-Brazilians and Euro-Brazilians Impacts Cystic Fibrosis Genetic Testing. Genetic Testing and Molecular Biomarkers, 2003, 7, 213-218.	1.7	18
97	Identification of eight novel NSD1 mutations in Sotos syndrome. Journal of Medical Genetics, 2003, 40, 126e-126.	3.2	33
98	Deletion of the C4-CYP21 Repeat Module Leading to the Formation of a Chimeric CYP21P/CYP21 Gene in a 9.3-kb Fragment as a Cause of Steroid 21-Hydroxylase Deficiency. Clinical Chemistry, 2003, 49, 319-322.	3.2	31
99	Parkin mutations are frequent in patients with isolated earlyâ€onset parkinsonism. Brain, 2003, 126, 1271-1278.	7.6	279
100	Cystic fibrosis with normal sweat chloride concentration: case report. Revista Do Hospital Das Clinicas, 2003, 58, 260-262.	0.5	6
101	Exclusion of the Nurr1 gene in autosomal recessive Parkinson's disease. Journal of Neurology, 2002, 249, 1127-1129.	3.6	9
102	Congenital contractural arachnodactyly with neurogenic muscular atrophy: case report. Arquivos De Neuro-Psiquiatria, 2001, 59, 259-262.	0.8	6
103	The G209A mutation in the alpha-synuclein gene in Brazilian families with Parkinson's disease. Arquivos De Neuro-Psiquiatria, 2001, 59, 722-724.	0.8	10
104	Huntington disease: DNA analysis in brazilian population. Arquivos De Neuro-Psiquiatria, 2000, 58, 977-985.	0.8	22
105	Juvenile Huntington's disease confirmed by genetic examination in twins. Arquivos De Neuro-Psiquiatria, 1999, 57, 867-869.	0.8	13
106	Genetic variation at twentythree microsatellite loci in sixteen human populations. Journal of Genetics, 1999, 78, 99-121.	0.7	39
107	Identification of a new lesch-nyhan syndrome mutation (HPRT BRASIL) and analysis of potentially heterozygous females. Arquivos De Neuro-Psiquiatria, 1999, 57, 907-911.	0.8	3
108	Description of a Brazilian Patient Bearing the R271WPit-1Gene Mutation. Thyroid, 1998, 8, 299-304.	4.5	21

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109	Rigid spine syndrome: case report. Arquivos De Neuro-Psiquiatria, 1998, 56, 812-818.	0.8	1
110	DNA Fingerprinting of Mycobacterium tuberculosis Complex Culture Isolates Collected in Brazil and Spotted onto Filter Paper. Journal of Clinical Microbiology, 1998, 36, 573-576.	3.9	8
111	Genetic mapping of the human pituitary-specific transcriptional factor gene and its analysis in familial panhypopituitary dwarfism. Human Genetics, 1996, 98, 703-705.	3.8	12
112	Normal CAG repeat variation at the DRPLA locus in world populations. American Journal of Human Genetics, 1995, 57, 508-11.	6.2	9
113	Estimation of the incidence of a rare genetic disease through a two-tier mutation survey. American Journal of Human Genetics, 1993, 52, 1129-38.	6.2	7
114	Cystic fibrosis genotyping by direct PCR analysis of Guthrie blood spots.. Genome Research, 1992, 2, 154-156.	5.5	33
115	Incidence and expression of the N1303K mutation of the cystic fibrosis (CFTR) gene. Human Genetics, 1992, 89, 653-658.	3.8	69
116	Utility of internal markers to improve the accuracy of cystic fibrosis genotype analysis. BioTechniques, 1992, 13, 372-4.	1.8	0