

Salmo Raskin

List of Publications by Year in descending order

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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	Phenotypic Heterogeneity of Genomic Disorders and Rare Copy-Number Variants. <i>New England Journal of Medicine</i> , 2012, 367, 1321-1331.	27.0	519
2	Parkin mutations are frequent in patients with isolated early-onset parkinsonism. <i>Brain</i> , 2003, 126, 1271-1278.	7.6	279
3	How much phenotypic variation can be attributed to parkin genotype?. <i>Annals of Neurology</i> , 2003, 54, 176-185.	5.3	271
4	The genomic landscape of balanced cytogenetic abnormalities associated with human congenital anomalies. <i>Nature Genetics</i> , 2017, 49, 36-45.	21.4	251
5	De novo SCN1A mutations are a major cause of severe myoclonic epilepsy of infancy. <i>Human Mutation</i> , 2003, 21, 615-621.	2.5	170
6	Expansion of the genotypic and phenotypic spectrum in patients with KRAS germline mutations. <i>Journal of Medical Genetics</i> , 2006, 44, 131-135.	3.2	170
7	High Incidence of Noonan Syndrome Features Including Short Stature and Pulmonic Stenosis in Patients carrying NF1 Missense Mutations Affecting p.Arg1809: Genotype-Phenotype Correlation. <i>Human Mutation</i> , 2015, 36, 1052-1063.	2.5	143
8	Inactivation of hnRNP K by Expanded Intronic AUUCU Repeat Induces Apoptosis Via Translocation of PKC β to Mitochondria in Spinocerebellar Ataxia 10. <i>PLoS Genetics</i> , 2010, 6, e1000984.	3.5	102
9	A randomized placebo-controlled phase 3 trial of an antisense oligonucleotide, drisapersen, in Duchenne muscular dystrophy. <i>Neuromuscular Disorders</i> , 2018, 28, 4-15.	0.6	102
10	A Noncoding Expansion in EIF4A3 Causes Richieri-Costa-Pereira Syndrome, a Craniofacial Disorder Associated with Limb Defects. <i>American Journal of Human Genetics</i> , 2014, 94, 120-128.	6.2	99
11	Congenital bilateral absence of the vas deferens as an atypical form of cystic fibrosis: reproductive implications and genetic counseling. <i>Andrology</i> , 2018, 6, 127-135.	3.5	94
12	Ataxia-telangiectasia – A historical review and a proposal for a new designation: ATM syndrome. <i>Journal of the Neurological Sciences</i> , 2015, 355, 3-6.	0.6	80
13	Spinocerebellar ataxia type 10 – A review. <i>Parkinsonism and Related Disorders</i> , 2011, 17, 655-661.	2.2	73
14	Incidence and expression of the N1303K mutation of the cystic fibrosis (CFTR) gene. <i>Human Genetics</i> , 1992, 89, 653-658.	3.8	69
15	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. <i>Journal of Cystic Fibrosis</i> , 2008, 7, 15-22.	0.7	57
16	Spinocerebellar ataxias – genotype-phenotype correlations in 104 Brazilian families. <i>Clinics</i> , 2012, 67, 443-449.	1.5	56
17	Repeat interruptions in spinocerebellar ataxia type 10 expansions are strongly associated with epileptic seizures. <i>Neurogenetics</i> , 2014, 15, 59-64.	1.4	51
18	<i>SLC13A5</i> is the second gene associated with Kohlschütter-Tarantino syndrome. <i>Journal of Medical Genetics</i> , 2017, 54, 54-62.	3.2	45

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19	Phenotypic consequences of genetic variation at hemizygous alleles: Sotos syndrome is a contiguous gene syndrome incorporating coagulation factor twelve (FXII) deficiency. <i>Genetics in Medicine</i> , 2005, 7, 479-483.	2.4	44
20	Reduced Penetrance in a Brazilian Family With Spinocerebellar Ataxia Type 10. <i>Archives of Neurology</i> , 2007, 64, 591.	4.5	42
21	Genetic variation at twentythree microsatellite loci in sixteen human populations. <i>Journal of Genetics</i> , 1999, 78, 99-121.	0.7	39
22	Targeted knockout of a chemokine-like gene increases anxiety and fear responses. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2018, 115, E1041-E1050.	7.1	39
23	Global genetic variation at nine short tandem repeat loci and implications on forensic genetics. <i>European Journal of Human Genetics</i> , 2003, 11, 39-49.	2.8	37
24	Spinocerebellar ataxia type 10: Frequency of epilepsy in a large sample of Brazilian patients. <i>Movement Disorders</i> , 2010, 25, 2875-2878.	3.9	36
25	Paradoxical effects of repeat interruptions on spinocerebellar ataxia type 10 expansions and repeat instability. <i>European Journal of Human Genetics</i> , 2013, 21, 1272-1276.	2.8	35
26	A New <i>ELOVL4</i> Mutation in a Case of Spinocerebellar Ataxia With Erythrokeratodermia. <i>JAMA Neurology</i> , 2015, 72, 942.	9.0	34
27	Cystic fibrosis genotyping by direct PCR analysis of Guthrie blood spots.. <i>Genome Research</i> , 1992, 2, 154-156.	5.5	33
28	Identification of eight novel NSD1 mutations in Sotos syndrome. <i>Journal of Medical Genetics</i> , 2003, 40, 126e-126.	3.2	33
29	Olfactory heterogeneity in <i>LRRK2</i> related Parkinsonism. <i>Movement Disorders</i> , 2010, 25, 2879-2883.	3.9	33
30	Bi-allelic variants in HOPS complex subunit VPS41 cause cerebellar ataxia and abnormal membrane trafficking. <i>Brain</i> , 2021, 144, 769-780.	7.6	33
31	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. <i>Clinical Chemistry</i> , 2003, 49, 319-322.	3.2	31
32	PIAS4 is associated with macro/microcephaly in the novel interstitial 19p13.3 microdeletion/microduplication syndrome. <i>European Journal of Human Genetics</i> , 2015, 23, 1615-1626.	2.8	29
33	Diffusion tensor imaging and tract-based spatial statistics analysis in Friedreich's ataxia patients. <i>Parkinsonism and Related Disorders</i> , 2015, 21, 504-508.	2.2	27
34	Kohlschütter-Tarnz Syndrome: Mutations in <i>ROGDI</i> and Evidence of Genetic Heterogeneity. <i>Human Mutation</i> , 2013, 34, 296-300.	2.5	24
35	Spinocerebellar ataxia type 3: subphenotypes in a cohort of brazilian patients. <i>Arquivos De Neuro-Psiquiatria</i> , 2014, 72, 659-662.	0.8	24
36	Nonmotor Symptoms in Patients with Spinocerebellar Ataxia Type 10. <i>Cerebellum</i> , 2017, 16, 938-944.	2.5	23

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37	Huntington disease: DNA analysis in brazilian population. Arquivos De Neuro-Psiquiatria, 2000, 58, 977-985.	0.8	22
38	Clinical and genetic study of a Brazilian family with spastic paraplegia (SPG6 locus). Movement Disorders, 2006, 21, 279-281.	3.9	22
39	Description of a Brazilian Patient Bearing the R271W<i>Pit-1</i>Gene Mutation. Thyroid, 1998, 8, 299-304.	4.5	21
40	A 1.5Mb terminal deletion of 12p associated with autism spectrum disorder. Gene, 2014, 542, 83-86.	2.2	21
41	Volumetric MRI Changes in Spinocerebellar Ataxia (SCA3 and SCA10) Patients. Cerebellum, 2020, 19, 536-543.	2.5	21
42	The G2019S <i>LRRK2</i> mutation in Brazilian patients with Parkinson's disease: Phenotype in monozygotic twins. Movement Disorders, 2008, 23, 290-294.	3.9	20
43	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
44	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
45	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
46	Clinical relevance of "bulging eyes" for the differential diagnosis of spinocerebellar ataxias. Arquivos De Neuro-Psiquiatria, 2013, 71, 428-430.	0.8	18
47	CTA/CTG expansions at the SCA 8 locus in multiple system atrophy. Clinical Neurology and Neurosurgery, 2009, 111, 208-210.	1.4	17
48	Genetics of COVID-19. Jornal De Pediatria, 2021, 97, 378-386.	2.0	17
49	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
50	Non-choreic movement disorders as initial manifestations of Huntington's disease. Arquivos De Neuro-Psiquiatria, 2007, 65, 402-405.	0.8	15
51	Spinocerebellar ataxia type 6 in Brazil. Arquivos De Neuro-Psiquiatria, 2008, 66, 691-694.	0.8	15
52	Movement Disorders in Spinocerebellar Ataxias in a Cohort of Brazilian Patients. European Neurology, 2014, 72, 360-362.	1.4	15
53	Three cases with rare interstitial rearrangements of chromosome 1 characterized by multicolor banding. Cytogenetic and Genome Research, 2005, 111, 171-174.	1.1	14
54	Juvenile Huntington's disease confirmed by genetic examination in twins. Arquivos De Neuro-Psiquiatria, 1999, 57, 867-869.	0.8	13

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55	Cystic fibrosis in a southern Brazilian population: characteristics of 90% of the alleles. <i>Clinical Genetics</i> , 2007, 72, 218-223.	2.0	13
56	Spinocerebellar ataxias in Southern Brazil: Genotypic and phenotypic evaluation of 213 families. <i>Clinical Neurology and Neurosurgery</i> , 2019, 184, 105427.	1.4	13
57	Genetic mapping of the human pituitary-specific transcriptional factor gene and its analysis in familial panhypopituitary dwarfism. <i>Human Genetics</i> , 1996, 98, 703-705.	3.8	12
58	Diastrophic dysplasia: prenatal diagnosis and review of the literature. <i>Sao Paulo Medical Journal</i> , 2013, 131, 127-132.	0.9	12
59	Tremor in X-linked recessive spinal and bulbar muscular atrophy (Kennedy's disease). <i>Clinics</i> , 2011, 66, 955-957.	1.5	12
60	When should we test patients with familial ataxias for SCA31? A misdiagnosed condition outside Japan?. <i>Journal of the Neurological Sciences</i> , 2015, 355, 206-208.	0.6	11
61	Olfactory Function in SCA10. <i>Cerebellum</i> , 2019, 18, 85-90.	2.5	11
62	Is Ataxia an Underestimated Symptom of Huntington's Disease?. <i>Frontiers in Neurology</i> , 2020, 11, 571843.	2.4	11
63	The G209A mutation in the alpha-synuclein gene in Brazilian families with Parkinson's disease. <i>Arquivos De Neuro-Psiquiatria</i> , 2001, 59, 722-724.	0.8	10
64	Cerebrotendinous xanthomatosis: report of two Brazilian brothers. <i>Arquivos De Neuro-Psiquiatria</i> , 2004, 62, 1085-1089.	0.8	10
65	<scp>SPG4</scp>-related hereditary spastic paraplegia: frequency and mutation spectrum in Brazil. <i>Clinical Genetics</i> , 2014, 86, 194-196.	2.0	10
66	Spinocerebellar ataxia type 10 in the South of Brazil: the Amerindian-Belgian connection. <i>Arquivos De Neuro-Psiquiatria</i> , 2015, 73, 725-727.	0.8	10
67	Exclusion of the Nurr1 gene in autosomal recessive Parkinson's disease. <i>Journal of Neurology</i> , 2002, 249, 1127-1129.	3.6	9
68	Symptom onset of spinocerebellar ataxia type 10 in pregnancy and puerperium. <i>Journal of Clinical Neuroscience</i> , 2011, 18, 437-438.	1.5	9
69	Normal CAG repeat variation at the DRPLA locus in world populations. <i>American Journal of Human Genetics</i> , 1995, 57, 508-11.	6.2	9
70	DNA Fingerprinting of <i>Mycobacterium tuberculosis</i> Complex Culture Isolates Collected in Brazil and Spotted onto Filter Paper. <i>Journal of Clinical Microbiology</i> , 1998, 36, 573-576.	3.9	8
71	Cãtis laxa: relato de caso. <i>Anais Brasileiros De Dermatologia</i> , 2010, 85, 684-686.	1.1	7
72	Cervical dystonia: about familial and sporadic cases in 88 patients. <i>Arquivos De Neuro-Psiquiatria</i> , 2014, 72, 107-113.	0.8	7

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73	Sleep disorders in spinocerebellar ataxia type 10. <i>Journal of Sleep Research</i> , 2018, 27, e12688.	3.2	7
74	Clinical and Genetic Evaluation of Spinocerebellar Ataxia Type 10 in 16 Brazilian Families. <i>Cerebellum</i> , 2019, 18, 849-854.	2.5	7
75	Folic acid for the prevention of neural tube defects. <i>Revista Brasileira De Ginecologia E Obstetricia</i> , 2013, 35, 287-289.	0.8	7
76	Estimation of the incidence of a rare genetic disease through a two-tier mutation survey. <i>American Journal of Human Genetics</i> , 1993, 52, 1129-38.	6.2	7
77	Congenital contractural arachnodactyly with neurogenic muscular atrophy: case report. <i>Arquivos De Neuro-Psiquiatria</i> , 2001, 59, 259-262.	0.8	6
78	Cystic fibrosis with normal sweat chloride concentration: case report. <i>Revista Do Hospital Das Clinicas</i> , 2003, 58, 260-262.	0.5	6
79	Atypical parkinsonism and SCA8. <i>Parkinsonism and Related Disorders</i> , 2006, 12, 191-192.	2.2	6
80	The history of spinocerebellar ataxia type 10 in Brazil: travels of a gene. <i>Arquivos De Neuro-Psiquiatria</i> , 2007, 65, 965-968.	0.8	6
81	Rett syndrome: clinical and molecular characterization of two Brazilian patients. <i>Arquivos De Neuro-Psiquiatria</i> , 2007, 65, 36-40.	0.8	6
82	Itajaí, Santa Catarina – Azorean ancestry and spinocerebellar ataxia type 3. <i>Arquivos De Neuro-Psiquiatria</i> , 2016, 74, 858-860.	0.8	6
83	Dystonia in Patients with Spinocerebellar Ataxia 3 - Machado-Joseph disease: An Underestimated Diagnosis?. <i>The Open Neurology Journal</i> , 2018, 12, 41-49.	0.4	6
84	Abnormal Findings in Polysomnographic Recordings of Patients with Spinocerebellar Ataxia Type 2 (SCA2). <i>Cerebellum</i> , 2019, 18, 196-202.	2.5	6
85	Cerebellar ataxia as the first manifestation of Alexander's disease. <i>Arquivos De Neuro-Psiquiatria</i> , 2012, 70, 309-310.	0.8	6
86	Analysis of diffusion tensor parameters in spinocerebellar ataxia type 3 and type 10 patients. <i>Parkinsonism and Related Disorders</i> , 2020, 78, 73-78.	2.2	5
87	A comprehensive analysis of AHRR gene as a candidate for cleft lip with or without cleft palate. <i>Mutation Research - Reviews in Mutation Research</i> , 2020, 785, 108319.	5.5	5
88	Adult onset Alexander disease presenting with progressive spastic paraplegia. <i>Parkinsonism and Related Disorders</i> , 2014, 20, 241-242.	2.2	4
89	Balance and physical functioning in Spinocerebellar ataxias 3 and 10. <i>Acta Neurologica Scandinavica</i> , 2021, 143, 458-463.	2.1	4
90	Acute onset of cerebellar ataxia in a spinocerebellar ataxia type 10 patient after use of steroids. <i>Arquivos De Neuro-Psiquiatria</i> , 2013, 71, 66-66.	0.8	4

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91	Kohlschütter-Tänz syndrome in siblings without ROGD1 mutation. <i>Oral Health and Dental Management</i> , 2014, 13, 728-30.	0.7	4
92	What can be done when asymptomatic patients discover they have Brugada syndrome? A case report of Brugada syndrome. <i>International Journal of Cardiology</i> , 2011, 150, e96-e97.	1.7	3
93	Niemann-Pick disease type C: a case series of Brazilian patients. <i>Arquivos De Neuro-Psiquiatria</i> , 2014, 72, 214-218.	0.8	3
94	Cerebellar and thalamic degeneration in spinocerebellar ataxia type 10. <i>Parkinsonism and Related Disorders</i> , 2020, 76, 76-77.	2.2	3
95	Identification of a new lesch-nyhan syndrome mutation (HPRT BRASIL) and analysis of potentially heterozygous females. <i>Arquivos De Neuro-Psiquiatria</i> , 1999, 57, 907-911.	0.8	3
96	Evidence and practices of the use of next generation sequencing in patients with undiagnosed autosomal dominant cerebellar ataxias: a review. <i>Arquivos De Neuro-Psiquiatria</i> , 2020, 78, 576-585.	0.8	3
97	Novel OPN1LW/OPN1MW Exon 3 Haplotype-Associated Splicing Defect in Patients with X-Linked Cone Dysfunction. <i>International Journal of Molecular Sciences</i> , 2022, 23, 6868.	4.1	3
98	Leber's hereditary optic neuropathy: case report and literature review. <i>Sao Paulo Medical Journal</i> , 2004, 122, 276-279.	0.9	2
99	Report of cluster headache in a pair of monozygous twins. <i>Journal of Headache and Pain</i> , 2004, 5, 140-143.	6.0	2
100	“Pseudo”Dominant™ Inheritance in Friedreich's Ataxia: Clinical and Genetic Study of a Brazilian Family. <i>Movement Disorders Clinical Practice</i> , 2014, 1, 361-363.	1.5	2
101	Spinocerebellar ataxias type 3 and 10: Onset and progression of ataxia during pregnancy and puerperium. <i>Parkinsonism and Related Disorders</i> , 2018, 52, 119-120.	2.2	2
102	Genetic evaluation for TOR1A (DYT1) in Brazilian patients with dystonia. <i>Arquivos De Neuro-Psiquiatria</i> , 2014, 72, 753-756.	0.8	2
103	Rigid spine syndrome: case report. <i>Arquivos De Neuro-Psiquiatria</i> , 1998, 56, 812-818.	0.8	1
104	Cystic Fibrosis Gene Variability in Two Southern Brazilian Amerindian Populations: Analysis of the P ^{F508} Mutation and the KM19 and XV2C Haplotypes. <i>Human Biology</i> , 2007, 79, 79-91.	0.2	1
105	Spinal muscular atrophy due to a “de novo” 1.3Mb deletion: Implication for genetic counseling. <i>Neuromuscular Disorders</i> , 2013, 23, 388-390.	0.6	1
106	Comparison of non-motors symptoms in patients with spinocerebellar ataxia type 10 and type 3. <i>Parkinsonism and Related Disorders</i> , 2016, 22, e150.	2.2	1
107	A 10.43 Mb duplication of chromosome region 5q31.2-q32 associated with a general delay in psychomotor development. <i>Meta Gene</i> , 2017, 11, 169-171.	0.6	1
108	Kennedy's disease phenotype with positive genetic study for Kugelberg-Welander's disease: case report. <i>Arquivos De Neuro-Psiquiatria</i> , 2005, 63, 330-331.	0.8	1

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109	<i>p.Phe508del</i>, <i>p.Gly542X</i>, <i>p.Arg1162X</i>, <i>p.Asn1303Lys</i>, and <i>p.Lys683serfsX38</i> mutations in CF newborn screening of Brazilian children. <i>Clinical Genetics</i> , 2017, 92, 115-116.	2.0	0
110	Cystic fibrosis in Afro-Brazilians: XK haplotypes analysis supports the European origin of p.F508del mutation. <i>Genetica</i> , 2017, 145, 19-25.	1.1	0
111	Hyposkillia and spanophilia in the movement disorders rounds. <i>Movement Disorders</i> , 2019, 34, 1399-1399.	3.9	0
112	Comment on: Diagnosis of Aicardiã€Goutiã€res Syndrome in Adults. <i>Movement Disorders Clinical Practice</i> , 2020, 7, 583-584.	1.5	0
113	Epileptic encephalopathy and atypical Rett syndrome with mutations in CDKL5: clinical and molecular characterization of two Brazilian patients. <i>Arquivos De Neuro-Psiquiatria</i> , 2013, 71, 414-415.	0.8	0
114	Utility of internal markers to improve the accuracy of cystic fibrosis genotype analysis. <i>BioTechniques</i> , 1992, 13, 372-4.	1.8	0
115	IDENTIFICAÃƒfO DE DELEÃƒfO TERMINAL 3P EM PACIENTE COM TRANSTORNO DO ESPECTRO AUTISTA. , 2022, 79, 1660.		0
116	DEFICIã€NCIA DA GLICOSE-6-FOSFATO DESIDROGENASE EM PACIENTE COM TRANSTORNO DO ESPECTRO AUTISTA. , 2022, 79, 1658.		0