

R Ramesar

List of Publications by Year in descending order

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Version: 2024-02-01

26
papers

1,390
citations

516710

16
h-index

580821

25
g-index

26
all docs

26
docs citations

26
times ranked

2190
citing authors

#	ARTICLE	IF	CITATIONS
1	The molecular genetics of cognition: dopamine, COMT and BDNF. <i>Genes, Brain and Behavior</i> , 2006, 5, 311-328.	2.2	275
2	CDH23 Mutation and Phenotype Heterogeneity: A Profile of 107 Diverse Families with Usher Syndrome and Nonsyndromic Deafness. <i>American Journal of Human Genetics</i> , 2002, 71, 262-275.	6.2	207
3	Molecular and clinical correlations in spinocerebellar ataxia type I: evidence for familial effects on the age at onset. <i>American Journal of Human Genetics</i> , 1994, 55, 244-52.	6.2	138
4	Apoptosis-inducing signal sequence mutation in carbonic anhydrase IV identified in patients with the RP17 form of retinitis pigmentosa. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2004, 101, 6617-6622.	7.1	108
5	A new locus for autosomal dominant retinitis pigmentosa on the short arm of chromosome 17. <i>Human Molecular Genetics</i> , 1994, 3, 915-918.	2.9	106
6	Surveillance colonoscopy improves survival in a cohort of subjects with a single mismatch repair gene mutation. <i>Colorectal Disease</i> , 2009, 11, 126-130.	1.4	80
7	Mutations of the KIT (mast/stem cell growth factor receptor) proto-oncogene account for a continuous range of phenotypes in human piebaldism. <i>American Journal of Human Genetics</i> , 1992, 51, 1058-65.	6.2	73
8	Genetic Heterogeneity of Usher Syndrome: Analysis of 151 Families with Usher Type I. <i>American Journal of Human Genetics</i> , 2000, 67, 1569-1574.	6.2	63
9	Autosomal Dominant (Beukes) Premature Degenerative Osteoarthropathy of the Hip Joint Maps to an 11-cM Region on Chromosome 4q35. <i>American Journal of Human Genetics</i> , 1999, 64, 904-908.	6.2	49
10	An eighth locus for autosomal dominant retinitis pigmentosa is linked to chromosome 17q. <i>Human Molecular Genetics</i> , 1995, 4, 1459-1462.	2.9	44
11	Polygenic risk for schizophrenia and associated brain structural changes: A systematic review. <i>Comprehensive Psychiatry</i> , 2019, 88, 77-82.	3.1	31
12	Retinitis pigmentosa in Southern Africa. <i>Clinical Genetics</i> , 1993, 44, 232-235.	2.0	30
13	Influence of birth cohort on age of onset cluster analysis in bipolar I disorder. <i>European Psychiatry</i> , 2015, 30, 99-105.	0.2	28
14	Solar insolation in springtime influences age of onset of bipolar I disorder. <i>Acta Psychiatrica Scandinavica</i> , 2017, 136, 571-582.	4.5	24
15	Mild spondyloepiphyseal dysplasia (Namaqualand type): genetic linkage to the type II collagen gene COL2A1. <i>American Journal of Human Genetics</i> , 1991, 48, 518-24.	6.2	22
16	Piebaldism: an autonomous autosomal dominant entity. <i>Clinical Genetics</i> , 1991, 39, 330-337.	2.0	20
17	Retinitis pigmentosa locus on 17q (RP17): fine localization to 17q22 and exclusion of the PDEG and TIMP2 genes. <i>Human Genetics</i> , 1997, 101, 13-17.	3.8	18
18	Cancer risk in a cohort of subjects carrying a single mismatch repair gene mutation. <i>Familial Cancer</i> , 2009, 8, 519-523.	1.9	13

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19	Autosomal dominant (Beukes) premature degenerative osteoarthropathy of the hip joint unlinked to COL2A1. American Journal of Medical Genetics Part A, 1994, 53, 348-351.	2.4	12
20	Identification of a locus for a form of spondyloepiphyseal dysplasia on chromosome 15q26.1: exclusion of aggrecan as a candidate gene. Journal of Medical Genetics, 2002, 39, 634-638.	3.2	11
21	Arg120stop nonsense mutation in the RP2 gene: mutational hotspot and germ line mosaicism?. Clinical Genetics, 2003, 65, 7-10.	2.0	10
22	Spondyloepiphyseal dysplasia in a cape town family: Linkage with the gene for type II collagen (COL2A1). American Journal of Medical Genetics Part A, 1992, 43, 833-838.	2.4	9
23	Low frequency of rhodopsin mutations in South African patients with autosomal dominant retinitis pigmentosa. Clinical Genetics, 2000, 58, 77-78.	2.0	7
24	Rhodopsin mutation G109R in a family with autosomal dominant retinitis pigmentosa. Human Mutation, 1998, 11, S40-S41.	2.5	6
25	No evidence of genetic anticipation in a large family with Lynch syndrome. Familial Cancer, 2014, 13, 29-34.	1.9	6
26	Immunohistochemical determination of mismatch repair gene product in colorectal carcinomas in a young indigenous African cohort.. South African Journal of Surgery, 2022, 60, 28-33.	0.2	0