Raj Ramesar

List of Publications by Year
in descending order

231 papers	12,360 citations	41344 49 h-index	30087 103 g-index
249 all docs	249 docs citations	249 times ranked	16924 citing authors

#	Article	IF	Citations
1	LDL Receptor-Related Protein 5 (LRP5) Affects Bone Accrual and Eye Development. Cell, 2001, 107, 513-523.	28.9	2,055
2	Long-term effect of aspirin on cancer risk in carriers of hereditary colorectal cancer: an analysis from the CAPP2 randomised controlled trial. Lancet, The, 2011, 378, 2081-2087.	13.7	849
3	Application of a 5-tiered scheme for standardized classification of 2,360 unique mismatch repair gene variants in the InSiGHT locus-specific database. Nature Genetics, 2014, 46, 107-115.	21.4	410
4	Molecular genetics of pseudoxanthoma elasticum: type and frequency of mutations inABCC6. Human Mutation, 2005, 26, 235-248.	2.5	365
5	Enabling the genomic revolution in Africa. Science, 2014, 344, 1346-1348.	12.6	361
6	Reconstructing Prehistoric African Population Structure. Cell, 2017, 171, 59-71.e21.	28.9	308
7	The molecular genetics of cognition: dopamine, COMT and BDNF. Genes, Brain and Behavior, 2006, 5, 311-328.	2.2	275
8	Effect of Aspirin or Resistant Starch on Colorectal Neoplasia in the Lynch Syndrome. New England Journal of Medicine, 2008, 359, 2567-2578.	27.0	273
9	Genetic Linkage of the Marfan Syndrome, Ectopia Lentis, and Congenital Contractural Arachnodactyly to the Fibrillin Genes on Chromosomes 15 and 5. New England Journal of Medicine, 1992, 326, 905-909.	27.0	257
10	Mutations in the pre-mRNA splicing factor gene PRPC8 in autosomal dominant retinitis pigmentosa (RP13). Human Molecular Genetics, 2001, 10, 1555-1562.	2.9	251
11	Cancer prevention with aspirin in hereditary colorectal cancer (Lynch syndrome), 10-year follow-up and registry-based 20-year data in the CAPP2 study: a double-blind, randomised, placebo-controlled trial. Lancet, The, 2020, 395, 1855-1863.	13.7	220
12	CDH23 Mutation and Phenotype Heterogeneity: A Profile of 107 Diverse Families with Usher Syndrome and Nonsyndromic Deafness. American Journal of Human Genetics, 2002, 71, 262-275.	6.2	207
13	Autosomal Dominant Craniometaphyseal Dysplasia Is Caused by Mutations in the Transmembrane Protein ANK. American Journal of Human Genetics, 2001, 68, 1321-1326.	6.2	177
14	Genomic Structure and Identification of Novel Mutations in Usherin, the Gene Responsible for Usher Syndrome Type IIa. American Journal of Human Genetics, 2000, 66, 1199-1210.	6.2	168
15	Neuropsychological dysfunction in bipolar affective disorder: a critical opinion. Bipolar Disorders, 2005, 7, 216-235.	1.9	157
16	Mutational bias provides a model for the evolution of Huntington's disease and predicts a general increase in disease prevalence. Nature Genetics, 1994, 7, 525-530.	21.4	141
17	Warriors Versus Worriers: The Role of COMT Gene Variants. CNS Spectrums, 2006, 11, 745-748.	1.2	139
18	A Mutation in the Variable Repeat Region of the Aggrecan Gene (AGC1) Causes a Form of Spondyloepiphyseal Dysplasia Associated with Severe, Premature Osteoarthritis. American Journal of Human Genetics, 2005, 77, 484-490.	6.2	137

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19	Genetic variants implicated in personality: A review of the more promising candidates. American Journal of Medical Genetics Part A, 2004, 131B, 20-32.	2.4	129
20	Mutations of the gene encoding the transmembrane transporter protein ABC-C6 cause pseudoxanthoma elasticum. Journal of Molecular Medicine, 2000, 78, 282-286.	3.9	118
21	Apoptosis-inducing signal sequence mutation in carbonic anhydrase IV identified in patients with the RP17 form of retinitis pigmentosa. Proceedings of the National Academy of Sciences of the United States of America, 2004, 101, 6617-6622.	7.1	108
22	A new locus for autosomal dominant retinitis pigmentosa on the short arm of chromosome 17. Human Molecular Genetics, 1994, 3, 915-918.	2.9	106
23	Neuropsychological task performance in bipolar spectrum illness: genetics, alcohol abuse, medication and childhood trauma. Bipolar Disorders, 2008, 10, 479-494.	1.9	98
24	X-Linked Late-Onset Sensorineural Deafness Caused by a Deletion Involving OA1 and a Novel Gene Containing WD-40 Repeats. American Journal of Human Genetics, 1999, 64, 1604-1616.	6.2	97
25	Long-term effect of resistant starch on cancer risk in carriers of hereditary colorectal cancer: an analysis from the CAPP2 randomised controlled trial. Lancet Oncology, The, 2012, 13, 1242-1249.	10.7	95
26	Genetics of schizophrenia in the South African Xhosa. Science, 2020, 367, 569-573.	12.6	93
27	Resolving the dark matter of ABCA4 for 1054 Stargardt disease probands through integrated genomics and transcriptomics. Genetics in Medicine, 2020, 22, 1235-1246.	2.4	92
28	Obesity, Aspirin, and Risk of Colorectal Cancer in Carriers of Hereditary Colorectal Cancer: A Prospective Investigation in the CAPP2 Study. Journal of Clinical Oncology, 2015, 33, 3591-3597.	1.6	91
29	Whole-genome sequencing for an enhanced understanding of genetic variation among South Africans. Nature Communications, 2017, 8, 2062.	12.8	88
30	The Co-Inheritance of Alpha-Thalassemia and Sickle Cell Anemia Is Associated with Better Hematological Indices and Lower Consultations Rate in Cameroonian Patients and Could Improve Their Survival. PLoS ONE, 2014, 9, e100516.	2.5	84
31	Surveillance colonoscopy improves survival in a cohort of subjects with a single mismatch repair gene mutation. Colorectal Disease, 2009, 11, 126-130.	1.4	80
32	Association of Variants at BCL11A and HBS1L-MYB with Hemoglobin F and Hospitalization Rates among Sickle Cell Patients in Cameroon. PLoS ONE, 2014, 9, e92506.	2.5	80
33	Genetic studies of African populations: an overview on disease susceptibility and response to vaccines and therapeutics. Human Genetics, 2008, 123, 557-598.	3.8	79
34	The hereditary adult-onset ataxias in South Africa. Journal of the Neurological Sciences, 2003, 216, 47-54.	0.6	77
35	Structural Variants Create New Topological-Associated Domains and Ectopic Retinal Enhancer-Gene Contact in Dominant Retinitis Pigmentosa. American Journal of Human Genetics, 2020, 107, 802-814.	6.2	75
36	Correlation between Waardenburg syndrome phenotype and genotype in a population of individuals with identified PAX3 mutations. Human Genetics, 1998, 102, 499-506.	3.8	69

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37	Heritable disorders of connective tissue. Best Practice and Research in Clinical Rheumatology, 2000, 14, 345-361.	3.3	68
38	A 500-kb region on chromosome 16p13.1 contains the pseudoxanthoma elasticum locus: high-resolution mapping and genomic structure. Journal of Molecular Medicine, 2000, 78, 36-46.	3.9	63
39	Genetic Heterogeneity of Usher Syndrome: Analysis of 151 Families with Usher Type I. American Journal of Human Genetics, 2000, 67, 1569-1574.	6.2	63
40	The Human Variome Project. Science, 2008, 322, 861-862.	12.6	63
41	Hypomanic, cyclothymic and hostile personality traits in bipolar spectrum illness: A family-based study. Journal of Psychiatric Research, 2008, 42, 920-929.	3.1	61
42	Genotype and Childhood Sexual Trauma Moderate Neurocognitive Performance: A Possible Role for Brain-Derived Neurotrophic Factor and Apolipoprotein E Variants. Biological Psychiatry, 2007, 62, 391-399.	1.3	59
43	Variation in the risk of colorectal cancer in families with Lynch syndrome: a retrospective cohort study. Lancet Oncology, The, 2021, 22, 1014-1022.	10.7	58
44	Neuropsychological status of bipolar I disorder: impact of psychosis. British Journal of Psychiatry, 2009, 194, 243-251.	2.8	55
45	Neurocognitive Function as an Endophenotype for Genetic Studies of Bipolar Affective Disorder. NeuroMolecular Medicine, 2005, 7, 275-286.	3.4	54
46	Evidence for paternal imprinting in familial Beckwith-Wiedemann syndrome.. Journal of Medical Genetics, 1992, 29, 221-225.	3.2	53
47	Personality: is it a viable endophenotype for genetic studies of bipolar affective disorder?. Bipolar Disorders, 2006, 8, 322-337.	1.9	52
48	Personality endophenotypes for bipolar affective disorder: a familyâ€based genetic association analysis. Genes, Brain and Behavior, 2008, 7, 869-876.	2.2	52
49	Autosomal Dominant (Beukes) Premature Degenerative Osteoarthropathy of the Hip Joint Maps to an 11-cM Region on Chromosome 4q35. American Journal of Human Genetics, 1999, 64, 904-908.	6.2	49
50	Identification of a mutation in the ubiquitin-fold modifier 1-specific peptidase 2 gene, UFSP2, in an extended South African family with Beukes hip dysplasia. South African Medical Journal, 2015, 105, 558.	0.6	49
51	Low-coverage sequencing cost-effectively detects known and novel variation in underrepresented populations. American Journal of Human Genetics, 2021, 108, 656-668.	6.2	49
52	A novel Q378X mutation exists in the transmembrane transporter protein ABCC6 and its pseudogene: implications for mutation analysis in pseudoxanthoma elasticum. Journal of Molecular Medicine, 2001, 79, 536-546.	3.9	48
53	PXR and CAR single nucleotide polymorphisms influence plasma efavirenz levels in South African HIV/AIDS patients. BMC Medical Genetics, 2012, 13, 112.	2.1	47
54	The shifting epidemiology of colorectal cancer in sub-Saharan Africa. The Lancet Gastroenterology and Hepatology, 2017, 2, 377-383.	8.1	47

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55	The relationship between childhood abuse and dissociation. Is it influenced by catechol-O-methyltransferase (COMT) activity?. International Journal of Neuropsychopharmacology, 2008, 11, 149-61.	2.1	46
56	Stickler-like syndrome due to a dominant negative mutation in the COL2A1 gene. , 1998, 80, 6-11.		45
57	Emergency Medical Services in India: The Present and Future. Prehospital and Disaster Medicine, 2014, 29, 307-310.	1.3	45
58	Nucleotide sequence and expression of a cloned Thiobacillus ferrooxidans recA gene in Escherichia coli. Gene, 1989, 78, 1-8.	2.2	44
59	An eighth locus for autosomal dominant retinitis pigmentosa is linked to chromosome 17q. Human Molecular Genetics, 1995, 4, 1459-1462.	2.9	44
60	Beyond the Caster Semenya Controversy: The Case of the Use of Genetics for Gender Testing in Sport. Journal of Genetic Counseling, 2010, 19, 545-548.	1.6	44
61	Relationship between sunlight and the age of onset of bipolar disorder: An international multisite study. Journal of Affective Disorders, 2014, 167, 104-111.	4.1	43
62	Novel presenilin 1 mutation with profound neurofibrillary pathology in an indigenous Southern African family with early-onset Alzheimer's disease. Brain, 2004, 127, 133-142.	7.6	42
63	Capturing all disease-causing mutations for clinical and research use: Toward an effortless system for the Human Variome Project. Genetics in Medicine, 2009, 11, 843-849.	2.4	42
64	Determining Ancestry Proportions in Complex Admixture Scenarios in South Africa Using a Novel Proxy Ancestry Selection Method. PLoS ONE, 2013, 8, e73971.	2.5	42
65	A Genomic Portrait of Haplotype Diversity and Signatures of Selection in Indigenous Southern African Populations. PLoS Genetics, 2015, 11, e1005052.	3.5	42
66	Lateralization of hand skill in bipolar affective disorder. Genes, Brain and Behavior, 2007, 6, 698-705.	2.2	41
67	South Africa: from species cradle to genomic applications. Nature Reviews Genetics, 2008, 9, S19-S23.	16.3	39
68	Psychosocial Burden of Sickle Cell Disease on Parents with an Affected Child in Cameroon. Journal of Genetic Counseling, 2014, 23, 192-201.	1.6	39
69	Influence of light exposure during early life on the age of onset of bipolar disorder. Journal of Psychiatric Research, 2015, 64, 1-8.	3.1	39
70	Mutation Spectrum and Founder Chromosomes for theABCA4Gene in South African Patients with Stargardt Disease. , 2004, 45, 1705.		37
71	Apolipoprotein E variants and cognition in healthy individuals: A critical opinion. Brain Research Reviews, 2006, 51, 125-135.	9.0	37
72	Whole-Exome Sequencing Identifies Biallelic IDH3A Variants as a Cause of Retinitis Pigmentosa Accompanied by Pseudocoloboma. Ophthalmology, 2017, 124, 992-1003.	5.2	37

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73	Dysthymic and anxiety-related personality traits in bipolar spectrum illness. Journal of Affective Disorders, 2008, 109, 305-311.	4.1	36
74	Spondyloepiphyseal dysplasia, mild autosomal dominant type is not due to primary defects of type II collagen. American Journal of Medical Genetics Part A, 1990, 37, 272-276.	2.4	32
75	Catechol-o-Methyltransferase Genotype and Childhood Trauma May Interact to Impact Schizotypal Personality Traits. Behavior Genetics, 2010, 40, 415-423.	2.1	32
76	Large Scale Genetic Research on Neuropsychiatric Disorders in African Populations is Needed. EBioMedicine, 2015, 2, 1259-1261.	6.1	32
77	Significant concordance of genetic variation that increases both the risk for obsessiveâ€“compulsive disorder and the volumes of the nucleus accumbens and putamen. British Journal of Psychiatry, 2018, 213, 430-436.	2.8	32
78	Trends in Suicidology: Personality as an Endophenotype for Molecular Genetic Investigations. PLoS Medicine, 2006, 3, e107.	8.4	32
79	Diverse prevalence of large deletions within the OA1 gene in ocular albinism type 1 patients from Europe and North America. Human Genetics, 2001, 108, 51-54.	3.8	31
80	Prognosis for splicing factor PRPF8 retinitis pigmentosa, novel mutations and correlation between human and yeast phenotypes. Human Mutation, 2010, 31, E1361-E1376.	2.5	31
81	Ready to Put Metadata on the Post-2015 Development Agenda? Linking Data Publications to Responsible Innovation and Science Diplomacy. OMICS A Journal of Integrative Biology, 2014, 18, 1-9.	2.0	31
82	Polygenic risk for schizophrenia and associated brain structural changes: A systematic review. Comprehensive Psychiatry, 2019, 88, 77-82.	3.1	31
83	Association of Sex With Frequent and Mild ABCA4 Alleles in Stargardt Disease. JAMA Ophthalmology, 2020, 138, 1035.	2.5	31
84	GSTM1 and GSTT1 polymorphisms as modifiers of age at diagnosis of hereditary nonpolyposis colorectal cancer (HNPCC) in a homogeneous cohort of individuals carrying a single predisposing mutation. Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis, 2006, 602, 175-181.	1.0	30
85	Retinitis pigmentosa in Southern Africa. Clinical Genetics, 1993, 44, 232-235.	2.0	30
86	Capacity-Building in Human Genetics for Developing Countries: Initiatives and Perspectives in Sub-Saharan Africa. Public Health Genomics, 2010, 13, 492-494.	1.0	29
87	Surgery for colonic cancer in HNPCC: total vs segmental colectomy. Colorectal Disease, 2011, 13, 1395-1399.	1.4	29
88	High prevalence of cisplatin-induced ototoxicity in Cape Town, South Africa. South African Medical Journal, 2014, 104, 288.	0.6	29
89	Spondyloepimetaphyseal dysplasia with joint laxity (Beighton type); mutation analysis in eight affected South African families. Clinical Genetics, 2015, 87, 492-495.	2.0	28
90	Influence of birth cohort on age of onset cluster analysis in bipolar I disorder. European Psychiatry, 2015, 30, 99-105.	0.2	28

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91	Exploring researchersâ€™ experiences of working with a researcher-driven, population-specific community advisory board in a South African schizophrenia genomics study. BMC Medical Ethics, 2015, 16, 45.	2.4	27
92	Hearing Impairment and Pigmentary Disturbance. Annals of the New York Academy of Sciences, 1991, 630, 152-166.	3.8	26
93	X-Linked Ocular Albinism and Sensorineural Deafness: Linkage to Xp22.3. Genomics, 1993, 18, 444-445.	2.9	26
94	Identification of new cases of early-onset colorectal cancer with an MLH1 epimutation in an ethnically diverse South African cohortâ€. Clinical Genetics, 2011, 80, 428-434.	2.0	26
95	Mapping of the gene for cleidocranial dysplasia in the historical Cape Town (Arnold) kindred and evidence for locus homogeneity.. Journal of Medical Genetics, 1996, 33, 511-514.	3.2	25
96	Recommendations for genetic variation data capture in developing countries to ensure a comprehensive worldwide data collection. Human Mutation, 2011, 32, 2-9.	2.5	25
97	G-protein-coupled receptor kinase 4 polymorphisms predict blood pressure response to dietary modification in Black patients with mild-to-moderate hypertension. Journal of Human Hypertension, 2012, 26, 334-339.	2.2	25
98	Association between solar insolation and a history of suicide attempts in bipolar I disorder. Journal of Psychiatric Research, 2019, 113, 1-9.	3.1	25
99	The Importance of G Protein-Coupled Receptor Kinase 4 (GRK4) in Pathogenesis of Salt Sensitivity, Salt Sensitive Hypertension and Response to Antihypertensive Treatment. International Journal of Molecular Sciences, 2015, 16, 5741-5749.	4.1	24
100	Solar insolation in springtime influences age of onset of bipolar I disorder. Acta Psychiatrica Scandinavica, 2017, 136, 571-582.	4.5	24
101	Concordance of genetic variation that increases risk for Tourette Syndrome and that influences its underlying neurocircuitry. Translational Psychiatry, 2019, 9, 120.	4.8	24
102	Expanded CAG repeats in spinocerebellar ataxia (SCA1) segregate with distinct haplotypes in South African families. Human Genetics, 1997, 100, 131-137.	3.8	23
103	The burden of sickle cell disease in Cape Town. South African Medical Journal, 2012, 102, 752.	0.6	23
104	Would you terminate a pregnancy affected by sickle cell disease? Analysis of views of patients in Cameroon. Journal of Medical Ethics, 2014, 40, 615-620.	1.8	23
105	A caseâ€“control study of risk factors for colorectal cancer in an African population. European Journal of Cancer Prevention, 2019, 28, 145-150.	1.3	22
106	Prioritization of candidate disease genes for metabolic syndrome by computational analysis of its defining phenotypes. Physiological Genomics, 2008, 35, 55-64.	2.3	21
107	Genetic variation in Otos is associated with cisplatin-induced ototoxicity. Pharmacogenomics, 2014, 15, 1667-1676.	1.3	21
108	Toward a Global Roadmap for Precision Medicine in Psychiatry: Challenges and Opportunities. OMICS A Journal of Integrative Biology, 2016, 20, 557-564.	2.0	21

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109	Promoter region variation in NFE2L2 influences susceptibility to ototoxicity in patients exposed to high cumulative doses of cisplatin. Pharmacogenomics Journal, 2017, 17, 515-520.	2.0	21
110	Clinical Application of Epilepsy Genetics in Africa: Is Now the Time?. Frontiers in Neurology, 2018, 9, 276.	2.4	21
111	Concordance of genetic variation that increases risk for anxiety disorders and posttraumatic stress disorders and that influences their underlying neurocircuitry. Journal of Affective Disorders, 2019, 245, 885-896.	4.1	21
112	Trends in Suicide Mortality in South Africa, 1997 to 2016. International Journal of Environmental Research and Public Health, 2020, 17, 1850.	2.6	21
113	Piebaldism: an autonomous autosomal dominant entity. Clinical Genetics, 1991, 39, 330-337.	2.0	20
114	A review of the optimisation of the use of formalin fixed paraffin embedded tissue for molecular analysis in a forensic post-mortem setting. Forensic Science International, 2017, 280, 181-187.	2.2	19
115	A splice junction mutation in PAX3 causes Waardenburg syndrome in a South African family. Human Molecular Genetics, 1994, 3, 197-198.	2.9	18
116	Retinitis pigmentosa locus on 17q (RP17): fine localization to 17q22 and exclusion of the PDEG and TIMP2 genes. Human Genetics, 1997, 101, 13-17.	3.8	18
117	Arrhythmogenic right ventricular cardiomyopathy type 6 (ARVC6): support for the locus assignment, narrowing of the critical region and mutation screening of three candidate genes. BMC Medical Genetics, 2006, 7, 29.	2.1	18
118	Mobile colonoscopic surveillance provides quality care for hereditary nonpolyposis colorectal carcinoma families in South Africa. Colorectal Disease, 2007, 9, 509-514.	1.4	18
119	Association of Novel Locus With Rheumatic Heart Disease in Black African Individuals. JAMA Cardiology, 2021, 6, 1000.	6.1	18
120	Refinement of the RP17 locus for autosomal dominant retinitis pigmentosa, construction of a YAC contig and investigation of the candidate gene retinal fascin. European Journal of Human Genetics, 1999, 7, 332-338.	2.8	17
121	Molecular Diagnosis of Inherited Retinal Diseases in Indigenous African Populations by Whole-Exome Sequencing. , 2016, 57, 6374.		17
122	Toward colorectal cancer control in <scp>A</scp>frica. International Journal of Cancer, 2016, 138, 1033-1034.	5.1	17
123	Human Leukocyte Antigen (HLA) Class II -DRB1 and -DQB1 Alleles and the Association with Cervical Cancer in HIV/HPV Co-Infected Women in South Africa. Journal of Cancer, 2019, 10, 2145-2152.	2.5	17
124	Molecular Investigation of Familial Beckwith-Wiedemann Syndrome: A Model for Paternal Imprinting. European Journal of Human Genetics, 1993, 1, 109-113.	2.8	17
125	A Radiation Hybrid Breakpoint Map of the Acute Myeloid Leukemia (AML) and Limb-Girdle Muscular Dystrophy 1A (LGMD1A) Regions of Chromosome 5q31 Localizing 122 Expressed Sequences. Genomics, 1999, 57, 24-35.	2.9	16
126	Psychosis and relapse in bipolar disorder are related to GRM3, DAOA, and GRIN2B genotype. African Journal of Psychiatry, 2010, 13, 297-301.	0.1	16

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127	Communicating cancer risk within an African context: Experiences, disclosure patterns and uptake rates following genetic testing for Lynch syndrome. Patient Education and Counseling, 2013, 92, 53-60.	2.2	15
128	Coinheritance of sickle cell anemia and Î±â€‘thalassemia delays disease onset and could improve survival in cameroonian's patients (Subâ€Saharan Africa). American Journal of Hematology, 2014, 89, 664-665.	4.1	15
129	The extracolonic cancer spectrum in females with the common â€˜South Africanâ€™ hMLH1 c.C1528T mutation. Familial Cancer, 2008, 7, 191-198.	1.9	14
130	Lynch syndrome: the influence of environmental factors on extracolonic cancer risk in hMLH1 c.C1528T mutation carriers and their mutation-negative sisters. Familial Cancer, 2010, 9, 357-363.	1.9	14
131	The incidence and histo-pathological characteristics of colorectal cancer in a population based cancer registry in Zimbabwe. Cancer Epidemiology, 2016, 44, 96-100.	1.9	14
132	Genetic Variation at Selected SNPs in the Leptin Gene and Association of Alleles with Markers of Kidney Disease in a Xhosa Population of South Africa. PLoS ONE, 2010, 5, e9086.	2.5	14
133	Molecular genetics improves the management of hereditary non-polyposis colorectal cancer. South African Medical Journal, 2000, 90, 709-14.	0.6	14
134	Brachydactylous dwarfs of Mseleni. American Journal of Medical Genetics Part A, 1993, 46, 636-640.	2.4	13
135	In a resource-poor country, mutation identification has the potential to reduce the cost of family management for hereditary nonpolyposis colorectal cancer. Diseases of the Colon and Rectum, 1998, 41, 1250-1253.	1.3	13
136	Immunohistochemistry detects mismatch repair gene defects in colorectal cancer. Colorectal Disease, 2006, 8, 411-417.	1.4	13
137	Cancer risk in a cohort of subjects carrying a single mismatch repair gene mutation. Familial Cancer, 2009, 8, 519-523.	1.9	13
138	Computational Analysis of Candidate Disease Genes and Variants for Salt-Sensitive Hypertension in Indigenous Southern Africans. PLoS ONE, 2010, 5, e12989.	2.5	13
139	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
140	Stargardt Disease: towards developing a model to predict phenotype. European Journal of Human Genetics, 2013, 21, 1173-1176.	2.8	12
141	Pharmacokinetics of rosuvastatin in 30 healthy Zimbabwean individuals of African ancestry. British Journal of Clinical Pharmacology, 2016, 82, 326-328.	2.4	12
142	Dietary patterns and colorectal cancer risk in Zimbabwe: A population based case-control study. Cancer Epidemiology, 2018, 57, 33-38.	1.9	12
143	Dravet syndrome in South African infants: Tools for an early diagnosis. Seizure: the Journal of the British Epilepsy Association, 2018, 62, 99-105.	2.0	12
144	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

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145	A rare homozygous rhodopsin splice-site mutation: the issue of when and whether to offer presymptomatic testing. Ophthalmic Genetics, 2003, 24, 225-232.	1.2	11
146	Beals syndrome: clinical and molecular investigations in a kindred of Indian descent. Clinical Genetics, 1991, 39, 181-188.	2.0	11
147	Advancing public health genomics in Africa through prospective cohort studies. Journal of Epidemiology and Community Health, 2010, 64, 585-586.	3.7	11
148	Direct-to-consumer genetic testing: To test or not to test, that is the question. South African Medical Journal, 2013, 103, 510.	0.6	11
149	The BDNFp.Val66Met polymorphism, childhood trauma, and brain volumes in adolescents with alcohol abuse. BMC Psychiatry, 2014, 14, 328.	2.6	11
150	Predictive Genetic Testing in Children: Constitutional Mismatch Repair Deficiency Cancer Predisposing Syndrome. Journal of Genetic Counseling, 2014, 23, 147-155.	1.6	11
151	Osteogenesis imperfecta type 3 in South Africa: Causative mutations in FKBP10. South African Medical Journal, 2017, 107, 457.	0.6	11
152	A Systematic Review of Molecular Autopsy Studies in Sudden Infant Death Cases. Journal of Pediatric Genetics, 2018, 07, 143-149.	0.7	11
153	Mismatch repair deficiency in colorectal cancer patients in a low-incidence area. South African Journal of Surgery, 2013, 51, 16-21.	0.2	11
154	Arg120stop nonsense mutation in the RP2 gene: mutational hotspot and germ line mosaicism?. Clinical Genetics, 2003, 65, 7-10.	2.0	10
155	A brain-behaviour initiative for South Africa: the time is right. Metabolic Brain Disease, 2006, 21, 266-271.	2.9	10
156	Bipolar Disorder: Emotional Dysregulation and Neuronal Vulnerability. CNS Spectrums, 2009, 14, 122-126.	1.2	10
157	Human variome project country nodes: Documenting genetic information within a country. Human Mutation, 2012, 33, 1513-1519.	2.5	10
158	Psychosocial Stressors of Sickle Cell Disease on Adult Patients in Cameroon. Journal of Genetic Counseling, 2014, 23, 948-956.	1.6	10
159	Age, absolute CD4 count, and CD4 percentage in relation to HPV infection and the stage of cervical disease in HIV-1-positive women. Medicine (United States), 2020, 99, e19273.	1.0	10
160	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
161	Preliminary evidence for linkage to chromosome 1q31-32, 10q23.3, and 16p13.3 in a South African cohort with bipolar disorder. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2007, 144B, 383-387.	1.7	9
162	UCTâ€™s contribution to medical genetics in Africa - from the past into the future. South African Medical Journal, 2012, 102, 446.	0.6	9

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163	A Mobile Colonoscopic Unit for Lynch Syndrome: Trends in Surveillance Uptake and Patient Experiences of Screening in a Developing Country. Journal of Genetic Counseling, 2013, 22, 125-137.	1.6	9
164	A Founder Mutation inMYO7AUnderlies a Significant Proportion of Usher Syndrome in Indigenous South Africans: Implications for the African Diaspora. , 2015, 56, 6671.		9
165	Ethical considerations in forensic genetics research on tissue samples collected post-mortem in Cape Town, South Africa. BMC Medical Ethics, 2017, 18, 66.	2.4	9
166	Impact of Host Molecular Genetic Variations and HIV/HPV Co-infection on Cervical Cancer Progression: A Systematic review. Oncomedicine, 2018, 3, 82-93.	1.1	9
167	De Novo Assembly-Based Analysis of RPGR Exon ORF15 in an Indigenous African Cohort Overcomes Limitations of a Standard Next-Generation Sequencing (NGS) Data Analysis Pipeline. Genes, 2020, 11, 800.	2.4	9
168	A review of systems biology research of anxiety disorders. Revista Brasileira De Psiquiatria, 2021, 43, 414-423.	1.7	9
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