Raj Ramesar

List of Publications by Year
in descending order

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230 papers	12,360 citations	41258 49 h-index	30010 103 g-index
249 all docs	249 docs citations	249 times ranked	16924 citing authors

#	Article	IF	Citations
1	Massively Parallel Sequencing of 43 Arrhythmia Genes in a Selected SUDI Cohort from Cape Town. Journal of Pediatric Genetics, 2022, 11, 292-297.	0.3	1
2	Correspondence on â€œCancer risks by gene, age, and gender in 6350 carriers of pathogenic mismatch repair variants: findings from the Prospective Lynch Syndrome Databaseâ€•by Dominguez-Valentin etÂal. Genetics in Medicine, 2022, , .	1.1	2
3	Global Globin Network Consensus Paper: Classification and Stratified Roadmaps for Improved Thalassaemia Care and Prevention in 32 Countries. Journal of Personalized Medicine, 2022, 12, 552.	1.1	6
4	A Scoring Model and Protocol to Adapt Universal Screening for Lynch Syndrome to Identify Germline Pathogenic Variants by Next Generation Sequencing from Colorectal Cancer Patients and Cascade Screening. Cancers, 2022, 14, 2901.	1.7	1
5	Overlap in genetic risk for cross-disorder vulnerability to mental disorders and genetic risk for altered subcortical brain volumes. Journal of Affective Disorders, 2021, 282, 740-756.	2.0	6
6	Low-coverage sequencing cost-effectively detects known and novel variation in underrepresented populations. American Journal of Human Genetics, 2021, 108, 656-668.	2.6	49
7	Variation in the risk of colorectal cancer in families with Lynch syndrome: a retrospective cohort study. Lancet Oncology, The, 2021, 22, 1014-1022.	5.1	58
8	A review of systems biology research of anxiety disorders. Revista Brasileira De Psiquiatria, 2021, 43, 414-423.	0.9	9
9	Association of Novel Locus With Rheumatic Heart Disease in Black African Individuals. JAMA Cardiology, 2021, 6, 1000.	3.0	18
10	Variations in seasonal solar insolation are associated with a history of suicide attempts in bipolar I disorder. International Journal of Bipolar Disorders, 2021, 9, 26.	0.8	6
11	PROTEA, A Southern African Multicenter Congenital Heart Disease Registry and Biorepository: Rationale, Design, and Initial Results. Frontiers in Pediatrics, 2021, 9, 763060.	0.9	6
12	Organophosphate pesticide exposure as a risk factor for attempted suicide in Cape Town, South Africa: A case-control study. Archives of Environmental and Occupational Health, 2021, , 1-11.	0.7	0
13	Mseleni joint disease: an endemic arthritis of unknown cause. Lancet Rheumatology, The, 2020, 2, e8-e9.	2.2	3
14	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.	2.6	75
15	Whole-exome sequencing in an Afrikaner family with bipolar disorder. Journal of Affective Disorders, 2020, 276, 69-75.	2.0	4
16	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.	1.0	9
17	Renal dysfunction, rodâ€cone dystrophy, and sensorineural hearing loss caused by a mutation in RRM2B. Human Mutation, 2020, 41, 1871-1876.	1.1	3
18	Association of Sex With Frequent and Mild ABCA4 Alleles in Stargardt Disease. JAMA Ophthalmology, 2020, 138, 1035.	1.4	31

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19	Resolving the dark matter of ABCA4 for 1054 Stargardt disease probands through integrated genomics and transcriptomics. Genetics in Medicine, 2020, 22, 1235-1246.	1.1	92
20	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.	6.3	220
21	Trends in Suicide Mortality in South Africa, 1997 to 2016. International Journal of Environmental Research and Public Health, 2020, 17, 1850.	1.2	21
22	Assessment of candidate variants causative of inborn metabolic diseases in SUDI cases in South Africa, and a case report. International Journal of Legal Medicine, 2020, 134, 1639-1645.	1.2	4
23	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.	0.4	10
24	Genetics of schizophrenia in the South African Xhosa. Science, 2020, 367, 569-573.	6.0	93
25	Developing a Road Map to Spread Genomic Knowledge in Africa: 10th Conference of the African Society of Human Genetics, Cairo, Egypt. American Journal of Tropical Medicine and Hygiene, 2020, 102, 719-723.	0.6	6
26	Building Skills and Resources for Genomics, Epigenetics, and Bioinformatics Research for Africa: Report of the Joint 11th Conference of the African Society of Human Genetics and 12th H3Africa Consortium, 2018. American Journal of Tropical Medicine and Hygiene, 2020, 102, 1417-1424.	0.6	7
27	The impact of the c.5603A>T hypomorphic variant on founder mutation screening of for Stargardt disease in South Africa. Molecular Vision, 2020, 26, 613-622.	1.1	2
28	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.	1.2	17
29	35 INVESTIGATION OF THE GENETIC INFLUENCES ON BIPOLAR DISORDER AND SUBCORTICAL BRAIN VOLUMES. European Neuropsychopharmacology, 2019, 29, S79.	0.3	0
30	Investigation on the hereditary basis of colorectal cancers in an African population with frequent early onset cases. PLoS ONE, 2019, 14, e0224023.	1.1	6
31	Association between solar insolation and a history of suicide attempts in bipolar I disorder. Journal of Psychiatric Research, 2019, 113, 1-9.	1.5	25
32	Concordance of genetic variation that increases risk for Tourette Syndrome and that influences its underlying neurocircuitry. Translational Psychiatry, 2019, 9, 120.	2.4	24
33	The ITHANET-Human Variome Project: Moving Functional Annotation Forward. Hemoglobin, 2019, 43, 327-327.	0.4	1
34	Investigation of Cervical Tumor Biopsies for Chromosomal Loss of Heterozygosity (LOH) and Microsatellite Instability (MSI) at the HLA II Locus in HIV-1/HPV Co-infected Women. Frontiers in Oncology, 2019, 9, 951.	1.3	6
35	Massively parallel sequencing in sudden unexpected death in infants: A case report in South Africa. Forensic Science International: Genetics Supplement Series, 2019, 7, 459-461.	0.1	2
36	Polygenic risk for schizophrenia and associated brain structural changes: A systematic review. Comprehensive Psychiatry, 2019, 88, 77-82.	1.5	31

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37	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.	2.0	21
38	Detecting genetic modifiers of spondyloepimetaphyseal dysplasia with joint laxity in the Caucasian Afrikaner community. Human Molecular Genetics, 2019, 28, 1053-1063.	1.4	1
39	A caseâ€“control study of risk factors for colorectal cancer in an African population. European Journal of Cancer Prevention, 2019, 28, 145-150.	0.6	22
40	Update on Inherited Retinal Disease in South Africa: Encouraging Diversity in Molecular Genetics. Advances in Experimental Medicine and Biology, 2019, 1185, 257-261.	0.8	2
41	DNA variants and organophosphate neurotoxicity among emerging farmers in the Western Cape of South Africa. American Journal of Industrial Medicine, 2018, 61, 11-20.	1.0	8
42	ASSAf consensus study on the ethical, legal and social implications of genetics and genomics in South Africa. South African Journal of Science, 2018, 114, .	0.3	6
43	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
44	Dietary patterns and colorectal cancer risk in Zimbabwe: A population based case-control study. Cancer Epidemiology, 2018, 57, 33-38.	0.8	12
45	Dravet syndrome in South African infants: Tools for an early diagnosis. Seizure: the Journal of the British Epilepsy Association, 2018, 62, 99-105.	0.9	12
46	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.	1.7	32
47	Clinical Application of Epilepsy Genetics in Africa: Is Now the Time?. Frontiers in Neurology, 2018, 9, 276.	1.1	21
48	A Systematic Review of Molecular Autopsy Studies in Sudden Infant Death Cases. Journal of Pediatric Genetics, 2018, 07, 143-149.	0.3	11
49	Whole-Exome Sequencing Identifies Biallelic IDH3A Variants as a Cause of Retinitis Pigmentosa Accompanied by Pseudocoloboma. Ophthalmology, 2017, 124, 992-1003.	2.5	37
50	The shifting epidemiology of colorectal cancer in sub-Saharan Africa. The Lancet Gastroenterology and Hepatology, 2017, 2, 377-383.	3.7	47
51	Genetic variation within GRIN2B in adolescents with alcohol use disorder may be associated with larger left posterior cingulate cortex volume. Acta Neuropsychiatrica, 2017, 29, 252-258.	1.0	7
52	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.	1.3	19
53	Reconstructing Prehistoric African Population Structure. Cell, 2017, 171, 59-71.e21.	13.5	308
54	Solar insolation in springtime influences age of onset of bipolar I disorder. Acta Psychiatrica Scandinavica, 2017, 136, 571-582.	2.2	24

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55	Promoter region variation in NFE2L2 influences susceptibility to ototoxicity in patients exposed to high cumulative doses of cisplatin. Pharmacogenomics Journal, 2017, 17, 515-520.	0.9	21
56	Whole-genome sequencing for an enhanced understanding of genetic variation among South Africans. Nature Communications, 2017, 8, 2062.	5.8	88
57	New-onset diabetes after transplant: Incidence, risk factors and outcome. South African Medical Journal, 2017, 107, 791.	0.2	8
58	Strengthening human genetics research in Africa: report of the 9th meeting of the African Society of Human Genetics in Dakar in May 2016. Global Health, Epidemiology and Genomics, 2017, 2, e10.	0.2	8
59	African Dawn. Historical Reflections, 2017, 43, .	0.0	0
60	Ethical considerations in forensic genetics research on tissue samples collected post-mortem in Cape Town, South Africa. BMC Medical Ethics, 2017, 18, 66.	1.0	9
61	Osteogenesis imperfecta type 3 in South Africa: Causative mutations in FKBP10. South African Medical Journal, 2017, 107, 457.	0.2	11
62	Digitotalar dysmorphism: Molecular elucidation. South African Medical Journal, 2016, 106, 253.	0.2	1
63	Molecular Diagnosis of Inherited Retinal Diseases in Indigenous African Populations by Whole-Exome Sequencing. , 2016, 57, 6374.		17
64	Inherited retinal disorders in South Africa and the clinical impact of evolving technologies. South African Medical Journal, 2016, 106, 33.	0.2	6
65	Toward colorectal cancer control in <scp>A</scp>frica. International Journal of Cancer, 2016, 138, 1033-1034.	2.3	17
66	The incidence and histo-pathological characteristics of colorectal cancer in a population based cancer registry in Zimbabwe. Cancer Epidemiology, 2016, 44, 96-100.	0.8	14
67	Minimum information required for a DMET experiment reporting. Pharmacogenomics, 2016, 17, 1533-1545.	0.6	6
68	Toward a Global Roadmap for Precision Medicine in Psychiatry: Challenges and Opportunities. OMICS A Journal of Integrative Biology, 2016, 20, 557-564.	1.0	21
69	Pharmacokinetics of rosuvastatin in 30 healthy Zimbabwean individuals of African ancestry. British Journal of Clinical Pharmacology, 2016, 82, 326-328.	1.1	12
70	Possible involvement of the circadian pathway in alcohol use disorder in a South African adolescent cohort. Metabolic Brain Disease, 2016, 31, 75-80.	1.4	2
71	Glutamatergic and HPA-axis pathway genes in bipolar disorder comorbid with alcohol- and substance use disorders. Metabolic Brain Disease, 2016, 31, 183-189.	1.4	6
72	Large Scale Genetic Research on Neuropsychiatric Disorders in African Populations is Needed. EBioMedicine, 2015, 2, 1259-1261.	2.7	32

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73	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.	0.2	49
74	A Founder Mutation inMYO7AUnderlies a Significant Proportion of Usher Syndrome in Indigenous South Africans: Implications for the African Diaspora. , 2015, 56, 6671.		9
75	Spondyloepimetaphyseal dysplasia with joint laxity (Beighton type); mutation analysis in eight affected South African families. Clinical Genetics, 2015, 87, 492-495.	1.0	28
76	Influence of birth cohort on age of onset cluster analysis in bipolar I disorder. European Psychiatry, 2015, 30, 99-105.	0.1	28
77	A Genomic Portrait of Haplotype Diversity and Signatures of Selection in Indigenous Southern African Populations. PLoS Genetics, 2015, 11, e1005052.	1.5	42
78	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.	1.8	24
79	Influence of light exposure during early life on the age of onset of bipolar disorder. Journal of Psychiatric Research, 2015, 64, 1-8.	1.5	39
80	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.	0.8	91
81	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.	1.0	27
82	African dawn. Nature, 2015, 517, 276-277.	13.7	4
83	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.	1.1	84
84	High prevalence of cisplatin-induced ototoxicity in Cape Town, South Africa. South African Medical Journal, 2014, 104, 288.	0.2	29
85	Genetic variation in Otos is associated with cisplatin-induced ototoxicity. Pharmacogenomics, 2014, 15, 1667-1676.	0.6	21
86	The BDNFp.Val66Met polymorphism, childhood trauma, and brain volumes in adolescents with alcohol abuse. BMC Psychiatry, 2014, 14, 328.	1.1	11
87	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.	1.0	31
88	Emergency Medical Services in India: The Present and Future. Prehospital and Disaster Medicine, 2014, 29, 307-310.	0.7	45
89	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.	1.0	23
90	Psychosocial Burden of Sickle Cell Disease on Parents with an Affected Child in Cameroon. Journal of Genetic Counseling, 2014, 23, 192-201.	0.9	39

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91	No evidence of genetic anticipation in a large family with Lynch syndrome. Familial Cancer, 2014, 13, 29-34.	0.9	6
92	Haplotype-based study of the association of alcohol and acetaldehyde-metabolising genes with alcohol dependence (with or without comorbid anxiety symptoms) in a Cape Mixed Ancestry population. Metabolic Brain Disease, 2014, 29, 333-340.	1.4	5
93	Psychosocial Stressors of Sickle Cell Disease on Adult Patients in Cameroon. Journal of Genetic Counseling, 2014, 23, 948-956.	0.9	10
94	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.	9.4	410
95	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.	2.0	15
96	Enabling the genomic revolution in Africa. Science, 2014, 344, 1346-1348.	6.0	361
97	A mutation in a splicing factor that causes retinitis pigmentosa has a transcriptome-wide effect on mRNA splicing. BMC Research Notes, 2014, 7, 401.	0.6	6
98	Fertility and apparent genetic anticipation in Lynch syndrome. Familial Cancer, 2014, 13, 369-374.	0.9	3
99	Predictive Genetic Testing in Children: Constitutional Mismatch Repair Deficiency Cancer Predisposing Syndrome. Journal of Genetic Counseling, 2014, 23, 147-155.	0.9	11
100	Relationship between sunlight and the age of onset of bipolar disorder: An international multisite study. Journal of Affective Disorders, 2014, 167, 104-111.	2.0	43
101	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.	1.1	80
102	Stargardt Disease: towards developing a model to predict phenotype. European Journal of Human Genetics, 2013, 21, 1173-1176.	1.4	12
103	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.	0.9	9
104	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.	1.0	15
105	Lynch Syndrome: Genetic Counselling of At-Risk Individuals and Families. , 2013, , 171-190.		0
106	Direct-to-consumer genetic testing: To test or not to test, that is the question. South African Medical Journal, 2013, 103, 510.	0.2	11
107	The value of genetic testing for inherited retinal disease caused by mutations in the ABCA4 gene in South Africans. South African Medical Journal, 2013, 103, 702.	0.2	0
108	Determining Ancestry Proportions in Complex Admixture Scenarios in South Africa Using a Novel Proxy Ancestry Selection Method. PLoS ONE, 2013, 8, e73971.	1.1	42

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109	Mismatch repair deficiency in colorectal cancer patients in a low-incidence area. South African Journal of Surgery, 2013, 51, 16-21.	0.1	11
110	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.	1.0	25
111	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
112	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.	5.1	95
113	UCTâ€™s contribution to medical genetics in Africa - from the past into the future. South African Medical Journal, 2012, 102, 446.	0.2	9
114	The burden of sickle cell disease in Cape Town. South African Medical Journal, 2012, 102, 752.	0.2	23
115	Human variome project country nodes: Documenting genetic information within a country. Human Mutation, 2012, 33, 1513-1519.	1.1	10
116	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.	6.3	849
117	The A142V Polymorphism of the G Protein Coupled Receptor Kinase 4 Gene Predicts Natriuretic Response to Saline Challenge in Young Normotensive Lean Black and White South African Men. Nephrology Research & Reviews, 2011, 3, 49-53.	0.2	5
118	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.	1.0	26
119	Surgery for colonic cancer in HNPCC: total vs segmental colectomy. Colorectal Disease, 2011, 13, 1395-1399.	0.7	29
120	Recommendations for genetic variation data capture in developing countries to ensure a comprehensive worldwide data collection. Human Mutation, 2011, 32, 2-9.	1.1	25
121	UCT and CPGR join forces with international Pharmacogenomics initiative focussing on African diseases. EMBnet Journal, 2011, 17, 3.	0.2	0
122	Catechol-o-Methyltransferase Genotype and Childhood Trauma May Interact to Impact Schizotypal Personality Traits. Behavior Genetics, 2010, 40, 415-423.	1.4	32
123	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.	0.9	44
124	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.	0.9	14
125	Prognosis for splicing factor PRPF8 retinitis pigmentosa, novel mutations and correlation between human and yeast phenotypes. Human Mutation, 2010, 31, E1361-E1376.	1.1	31
126	Cellâ€specific differences in the processing of the R14W CAIV mutant associated with retinitis pigmentosa 17. Journal of Cellular Biochemistry, 2010, 111, 735-741.	1.2	5

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127	Cancer occurrence during follow-up of the CAPP2 study -aspirin use for up to four years significantly reduces Lynch syndrome cancers for up to several years after completion of therapy. Hereditary Cancer in Clinical Practice, 2010, 8, O5.	0.6	2
128	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
129	Advancing public health genomics in Africa through prospective cohort studies. Journal of Epidemiology and Community Health, 2010, 64, 585-586.	2.0	11
130	Capacity-Building in Human Genetics for Developing Countries: Initiatives and Perspectives in Sub-Saharan Africa. Public Health Genomics, 2010, 13, 492-494.	0.6	29
131	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.	1.1	14
132	Computational Analysis of Candidate Disease Genes and Variants for Salt-Sensitive Hypertension in Indigenous Southern Africans. PLoS ONE, 2010, 5, e12989.	1.1	13
133	Across Culture and Health Systems: Africa. , 2010, , 471-486.		0
134	Clinical Utility of the ABCR400 Microarray. JAMA Ophthalmology, 2009, 127, 549.	2.6	6
135	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.	1.1	42
136	Cancer risk in a cohort of subjects carrying a single mismatch repair gene mutation. Familial Cancer, 2009, 8, 519-523.	0.9	13
137	Surveillance colonoscopy improves survival in a cohort of subjects with a single mismatch repair gene mutation. Colorectal Disease, 2009, 11, 126-130.	0.7	80
138	Bipolar Disorder: Emotional Dysregulation and Neuronal Vulnerability. CNS Spectrums, 2009, 14, 122-126.	0.7	10
139	Neuropsychological status of bipolar I disorder: impact of psychosis. British Journal of Psychiatry, 2009, 194, 243-251.	1.7	55
140	The extracolonic cancer spectrum in females with the common â€˜South Africanâ€™ hMLH1 c.C1528T mutation. Familial Cancer, 2008, 7, 191-198.	0.9	14
141	Management of a South African family with retinitis pigmentosaâ€”should potential therapy influence translational research protocols?. Journal of Ocular Biology, Diseases, and Informatics, 2008, 1, 55-8.	0.2	1
142	Genetic studies of African populations: an overview on disease susceptibility and response to vaccines and therapeutics. Human Genetics, 2008, 123, 557-598.	1.8	79
143	Infrequent and low expression of cancerâ€testis antigens located on the X chromosome in colorectal cancer: Implications for immunotherapy in South African populations. Biotechnology Journal, 2008, 3, 1417-1423.	1.8	8
144	Personality endophenotypes for bipolar affective disorder: a familyâ€based genetic association analysis. Genes, Brain and Behavior, 2008, 7, 869-876.	1.1	52

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145	South Africa: from species cradle to genomic applications. Nature Reviews Genetics, 2008, 9, S19-S23.	7.7	39
146	Neuropsychological task performance in bipolar spectrum illness: genetics, alcohol abuse, medication and childhood trauma. Bipolar Disorders, 2008, 10, 479-494.	1.1	98
147	Dysthymic and anxiety-related personality traits in bipolar spectrum illness. Journal of Affective Disorders, 2008, 109, 305-311.	2.0	36
148	Hypomanic, cyclothymic and hostile personality traits in bipolar spectrum illness: A family-based study. Journal of Psychiatric Research, 2008, 42, 920-929.	1.5	61
149	The Human Variome Project. Science, 2008, 322, 861-862.	6.0	63
150	Effect of Aspirin or Resistant Starch on Colorectal Neoplasia in the Lynch Syndrome. New England Journal of Medicine, 2008, 359, 2567-2578.	13.9	273
151	The relationship between childhood abuse and dissociation. Is it influenced by catechol-O-methyltransferase (COMT) activity?. International Journal of Neuropsychopharmacology, 2008, 11, 149-61.	1.0	46
152	Prioritization of candidate disease genes for metabolic syndrome by computational analysis of its defining phenotypes. Physiological Genomics, 2008, 35, 55-64.	1.0	21
153	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.	0.7	59
154	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.	1.1	9
155	Mobile colonoscopic surveillance provides quality care for hereditary nonpolyposis colorectal carcinoma families in South Africa. Colorectal Disease, 2007, 9, 509-514.	0.7	18
156	Lateralization of hand skill in bipolar affective disorder. Genes, Brain and Behavior, 2007, 6, 698-705.	1.1	41
157	A linkage and family-based association analysis of a potential neurocognitive endophenotype of bipolar disorder. NeuroMolecular Medicine, 2007, 9, 101-116.	1.8	7
158	A linkage and family-based association analysis of a potential neurocognitive endophenotype of bipolar disorder. NeuroMolecular Medicine, 2007, 9, 101-116.	1.8	0
159	Carbonic Anhydrase Inhibitors as a Possible Therapy for RP17, an Autosomal Dominant Retinitis Pigmentosa Associated With the R14W Mutation, Apoptosis, and the Unfolded Protein Response. , 2007, , 455-458.		0
160	Neurocognitive function in an extended Afrikaner-ancestry family with affective illness. Journal of Psychiatry and Neuroscience, 2007, 32, 116-20.	1.4	5
161	Warriors Versus Worriers: The Role of COMT Gene Variants. CNS Spectrums, 2006, 11, 745-748.	0.7	139
162	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

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163	Personality: is it a viable endophenotype for genetic studies of bipolar affective disorder?. Bipolar Disorders, 2006, 8, 322-337.	1.1	52
164	Immunohistochemistry detects mismatch repair gene defects in colorectal cancer. Colorectal Disease, 2006, 8, 411-417.	0.7	13
165	The molecular genetics of cognition: dopamine, COMT and BDNF. Genes, Brain and Behavior, 2006, 5, 311-328.	1.1	275
166	A brain-behaviour initiative for South Africa: the time is right. Metabolic Brain Disease, 2006, 21, 266-271.	1.4	10
167	Apolipoprotein E variants and cognition in healthy individuals: A critical opinion. Brain Research Reviews, 2006, 51, 125-135.	9.1	37
168	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.	0.4	30
169	Trends in Suicidology: Personality as an Endophenotype for Molecular Genetic Investigations. PLoS Medicine, 2006, 3, e107.	3.9	32
170	Novel variants in the hotspot region of RP1 in South African patients with retinitis pigmentosa. Molecular Vision, 2006, 12, 177-83.	1.1	7
171	Neurocognitive Function as an Endophenotype for Genetic Studies of Bipolar Affective Disorder. NeuroMolecular Medicine, 2005, 7, 275-286.	1.8	54
172	Neuropsychological dysfunction in bipolar affective disorder: a critical opinion. Bipolar Disorders, 2005, 7, 216-235.	1.1	157
173	Molecular genetics of pseudoxanthoma elasticum: type and frequency of mutations inABCC6. Human Mutation, 2005, 26, 235-248.	1.1	365
174	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.	2.6	137
175	Mutation Spectrum and Founder Chromosomes for theABCA4Gene in South African Patients with Stargardt Disease. , 2004, 45, 1705.		37
176	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.	3.7	42
177	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.	3.3	108
178	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
179	Analysis of RPGR in a South African family with X-linked retinitis pigmentosa: research and diagnostic implications. Clinical Genetics, 2003, 64, 137-141.	1.0	5
180	Arg120stop nonsense mutation in the RP2 gene: mutational hotspot and germ line mosaicism?. Clinical Genetics, 2003, 65, 7-10.	1.0	10

#	Article	IF	Citations
181	The hereditary adult-onset ataxias in South Africa. Journal of the Neurological Sciences, 2003, 216, 47-54.	0.3	77
182	A rare homozygous rhodopsin splice-site mutation: the issue of when and whether to offer presymptomatic testing. Ophthalmic Genetics, 2003, 24, 225-232.	0.5	11
183	Retinal Degenerative Disorders in Southern Africa: A Molecular Genetic Approach. Advances in Experimental Medicine and Biology, 2003, 533, 35-40.	0.8	3
184	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.	1.5	11
185	A computer-based register for inherited retinal dystrophies in Southern Africa. Ophthalmic Genetics, 2002, 23, 61-65.	0.5	6
186	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.	2.6	207
187	Autosomal Dominant Craniometaphyseal Dysplasia Is Caused by Mutations in the Transmembrane Protein ANK. American Journal of Human Genetics, 2001, 68, 1321-1326.	2.6	177
188	LDL Receptor-Related Protein 5 (LRP5) Affects Bone Accrual and Eye Development. Cell, 2001, 107, 513-523.	13.5	2,055
189	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.	1.7	48
190	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.	1.8	31
191	Mutations in the pre-mRNA splicing factor gene PRPC8 in autosomal dominant retinitis pigmentosa (RP13). Human Molecular Genetics, 2001, 10, 1555-1562.	1.4	251
192	Unusual Frequencies of Rhodopsin Mutations and Polymorphisms in Southern African Patients with Retinitis Pigmentosa. , 2001, , 329-333.		2
193	Low frequency of rhodopsin mutations in South African patients with autosomal dominant retinitis pigmentosa. Clinical Genetics, 2000, 58, 77-78.	1.0	7
194	Heritable disorders of connective tissue. Best Practice and Research in Clinical Rheumatology, 2000, 14, 345-361.	1.4	68
195	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.	1.7	63
196	Mutations of the gene encoding the transmembrane transporter protein ABC-C6 cause pseudoxanthoma elasticum. Journal of Molecular Medicine, 2000, 78, 282-286.	1.7	118
197	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.	2.6	168
198	Genetic Heterogeneity of Usher Syndrome: Analysis of 151 Families with Usher Type I. American Journal of Human Genetics, 2000, 67, 1569-1574.	2.6	63

#	Article	IF	Citations
199	Molecular genetics improves the management of hereditary non-polyposis colorectal cancer. South African Medical Journal, 2000, 90, 709-14.	0.2	14
200	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.	1.4	17
201	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.	2.6	49
202	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.	2.6	97
203	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.	1.3	16
204	Rhodopsin mutation G109R in a family with autosomal dominant retinitis pigmentosa. Human Mutation, 1998, 11, S40-S41.	1.1	6
205	Stickler-like syndrome due to a dominant negative mutation in the COL2A1 gene. , 1998, 80, 6-11.		45
206	Correlation between Waardenburg syndrome phenotype and genotype in a population of individuals with identified PAX3 mutations. Human Genetics, 1998, 102, 499-506.	1.8	69
207	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.	0.7	13
208	Expanded CAG repeats in spinocerebellar ataxia (SCA1) segregate with distinct haplotypes in South African families. Human Genetics, 1997, 100, 131-137.	1.8	23
209	Retinitis pigmentosa locus on 17q (RP17): fine localization to 17q22 and exclusion of the PDEG and TIMP2 genes. Human Genetics, 1997, 101, 13-17.	1.8	18
210	Growth Factors In The Retina. , 1997, , 291-294.		2
211	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.	1.5	25
212	An eighth locus for autosomal dominant retinitis pigmentosa is linked to chromosome 17q. Human Molecular Genetics, 1995, 4, 1459-1462.	1.4	44
213	A splice junction mutation in PAX3 causes Waardenburg syndrome in a South African family. Human Molecular Genetics, 1994, 3, 197-198.	1.4	18
214	A new locus for autosomal dominant retinitis pigmentosa on the short arm of chromosome 17. Human Molecular Genetics, 1994, 3, 915-918.	1.4	106
215	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
216	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.	9.4	141

#	Article	IF	Citations
217	Brachydactylous dwarfs of Mseleni. American Journal of Medical Genetics Part A, 1993, 46, 636-640.	2.4	13
218	X-Linked Ocular Albinism and Sensorineural Deafness: Linkage to Xp22.3. Genomics, 1993, 18, 444-445.	1.3	26
219	Retinitis pigmentosa in Southern Africa. Clinical Genetics, 1993, 44, 232-235.	1.0	30
220	Molecular Investigation of Familial Beckwith-Wiedemann Syndrome: A Model for Paternal Imprinting. European Journal of Human Genetics, 1993, 1, 109-113.	1.4	17
221	Evidence for paternal imprinting in familial Beckwith-Wiedemann syndrome.. Journal of Medical Genetics, 1992, 29, 221-225.	1.5	53
222	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.	13.9	257
223	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
224	Hearing Impairment and Pigmentary Disturbance. Annals of the New York Academy of Sciences, 1991, 630, 152-166.	1.8	26
225	Beals syndrome: clinical and molecular investigations in a kindred of Indian descent. Clinical Genetics, 1991, 39, 181-188.	1.0	11
226	Piebaldism: an autonomous autosomal dominant entity. Clinical Genetics, 1991, 39, 330-337.	1.0	20
227	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
228	Nucleotide sequence and expression of a cloned Thiobacillus ferrooxidans recA gene in Escherichia coli. Gene, 1989, 78, 1-8.	1.0	44
229	Cloning and Expression in Escherichia coli of a rec A-like Gene from the Acidophilic Autotroph Thiobacillus ferrooxidans. Microbiology (United Kingdom), 1988, 134, 1141-1146.	0.7	7
230	Whole Exome Sequencing in South Africa: Stakeholder Views on Return of Individual Research Results and Incidental Findings. Frontiers in Genetics, 0, 13, .	1.1	5