

Raj Ramesar

List of Publications by Year in descending order

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Version: 2025-02-01

178
papers

8,498
citations

54494

42
h-index

39432

88
g-index

204
all docs

204
docs citations

204
times ranked

12625
citing authors

#	ARTICLE	IF	CITATIONS
1	Towards Uncovering the Role of Incomplete Penetrance in Maculopathies through Sequencing of 105 Disease-Associated Genes. <i>Biomolecules</i> , 2024, 14, 367.	4.4	1
2	Genetic Factors Contributing to the Pathogenesis of Essential Hypertension in Two African Populations. <i>Journal of Personalized Medicine</i> , 2024, 14, 323.	2.7	0
3	Human Leukocyte Antigen-Allelic Variations May Influence the Age at Cancer Diagnosis in Lynch Syndrome. <i>Journal of Personalized Medicine</i> , 2024, 14, 575.	2.7	0
4	Influence of Genetic Polymorphisms on the Age at Cancer Diagnosis in a Homogenous Lynch Syndrome Cohort of Individuals Carrying the MLH1:c.1528C>T South African Founder Variant. <i>Biomedicines</i> , 2024, 12, 2201.	3.6	0
5	Genomic landscape of colorectal carcinoma in sub-Saharan Africa. <i>Journal of Clinical Pathology</i> , 2023, 76, 5-10.	1.7	3
6	Rationale, Design, and the Baseline Characteristics of the RHDGen (The Genetics of Rheumatic Heart) Tj ETQq0 0 0.rgBT /Overlock 10 Tf	3.2	1
7	Massively Parallel Sequencing of 43 Arrhythmia Genes in a Selected SUDI Cohort from Cape Town. <i>Journal of Pediatric Genetics</i> , 2022, 11, 292-297.	0.6	1
8	Correspondence on Cancer risks by gene, age, and gender in 6350 carriers of pathogenic mismatch repair variants: findings from the Prospective Lynch Syndrome Database by Dominguez-Valentin et al. <i>Genetics in Medicine</i> , 2022, 24, 1148-1150.	2.3	3
9	Global Globin Network Consensus Paper: Classification and Stratified Roadmaps for Improved Thalassaemia Care and Prevention in 32 Countries. <i>Journal of Personalized Medicine</i> , 2022, 12, 552.	2.7	11
10	Organophosphate pesticide exposure as a risk factor for attempted suicide in Cape Town, South Africa: A case-control study. <i>Archives of Environmental and Occupational Health</i> , 2022, 77, 789-799.	1.7	0
11	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. <i>Cancers</i> , 2022, 14, 2901.	4.0	1
12	Whole Exome Sequencing in South Africa: Stakeholder Views on Return of Individual Research Results and Incidental Findings. <i>Frontiers in Genetics</i> , 2022, 13, .	2.4	9
13	Cancer Prevention with Resistant Starch in Lynch Syndrome Patients in the CAPP2-Randomized Placebo Controlled Trial: Planned 10-Year Follow-up. <i>Cancer Prevention Research</i> , 2022, 15, 623-634.	1.1	33
14	Investigation of Copy Number Variation in South African Patients With Congenital Heart Defects. <i>Circulation Genomic and Precision Medicine</i> , 2022, 15, .	3.2	2
15	A comparative cost analysis of two screening strategies for colorectal cancer in Lynch Syndrome in a South African tertiary hospital. <i>Cancer Causes and Control</i> , 2022, 34, 161-169.	1.8	2
16	Overlap in genetic risk for cross-disorder vulnerability to mental disorders and genetic risk for altered subcortical brain volumes. <i>Journal of Affective Disorders</i> , 2021, 282, 740-756.	4.7	5
17	Low-coverage sequencing cost-effectively detects known and novel variation in underrepresented populations. <i>American Journal of Human Genetics</i> , 2021, 108, 656-668.	6.8	47
18	Variation in the risk of colorectal cancer in families with Lynch syndrome: a retrospective cohort study. <i>Lancet Oncology</i> , The, 2021, 22, 1014-1022.	21.9	74

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19	A review of systems biology research of anxiety disorders. <i>Revista Brasileira De Psiquiatria</i> , 2021, 43, 414-423.	1.1	9
20	Association of Novel Locus With Rheumatic Heart Disease in Black African Individuals. <i>JAMA Cardiology</i> , 2021, 6, 1000.	9.7	23
21	Variations in seasonal solar insolation are associated with a history of suicide attempts in bipolar I disorder. <i>International Journal of Bipolar Disorders</i> , 2021, 9, .	2.6	11
22	PROTEA, A Southern African Multicenter Congenital Heart Disease Registry and Biorepository: Rationale, Design, and Initial Results. <i>Frontiers in Pediatrics</i> , 2021, 9, .	1.9	12
23	Mseleni joint disease: an endemic arthritis of unknown cause. <i>Lancet Rheumatology</i> , The, 2020, 2, e8-e9.	9.2	4
24	Structural Variants Create New Topological-Associated Domains and Ectopic Retinal Enhancer-Gene Contact in Dominant Retinitis Pigmentosa. <i>American Journal of Human Genetics</i> , 2020, 107, 802-814.	6.8	80
25	Whole-exome sequencing in an Afrikaner family with bipolar disorder. <i>Journal of Affective Disorders</i> , 2020, 276, 69-75.	4.7	4
26	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. <i>Genes</i> , 2020, 11, 800.	2.7	11
27	Association of Sex With Frequent and Mild <i>ABCA4</i> Alleles in Stargardt Disease. <i>JAMA Ophthalmology</i> , 2020, 138, 1035.	4.2	27
28	Resolving the dark matter of ABCA4 for 1054 Stargardt disease probands through integrated genomics and transcriptomics. <i>Genetics in Medicine</i> , 2020, 22, 1235-1246.	2.3	95
29	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. <i>Lancet</i> , The, 2020, 395, 1855-1863.	35.3	257
30	Trends in Suicide Mortality in South Africa, 1997 to 2016. <i>International Journal of Environmental Research and Public Health</i> , 2020, 17, 1850.	3.1	28
31	Assessment of candidate variants causative of inborn metabolic diseases in SUDI cases in South Africa, and a case report. <i>International Journal of Legal Medicine</i> , 2020, 134, 1639-1645.	2.0	4
32	Age, absolute CD4 count, and CD4 percentage in relation to HPV infection and the stage of cervical disease in HIV-1-positive women. <i>Medicine (United States)</i> , 2020, 99, e19273.	1.3	14
33	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. <i>Journal of Cancer</i> , 2019, 10, 2145-2152.	2.7	16
34	Investigation on the hereditary basis of colorectal cancers in an African population with frequent early onset cases. <i>PLoS ONE</i> , 2019, 14, e0224023.	2.5	9
35	Association between solar insolation and a history of suicide attempts in bipolar I disorder. <i>Journal of Psychiatric Research</i> , 2019, 113, 1-9.	3.1	28
36	Concordance of genetic variation that increases risk for Tourette Syndrome and that influences its underlying neurocircuitry. <i>Translational Psychiatry</i> , 2019, 9, .	5.7	19

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37	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. <i>Frontiers in Oncology</i> , 2019, 9, .	2.7	7
38	SU1111GENOME-WIDE ANALYSIS OF COPY NUMBER VARIATION IN A SOUTH AFRICAN XHOSA POPULATION AFFECTED BY SCHIZOPHRENIA. <i>European Neuropsychopharmacology</i> , 2019, 29, S1325-S1326.	0.9	0
39	Massively parallel sequencing in sudden unexpected death in infants: A case report in South Africa. <i>Forensic Science International: Genetics Supplement Series</i> , 2019, 7, 459-461.	0.5	3
40	Concordance of genetic variation that increases risk for anxiety disorders and posttraumatic stress disorders and that influences their underlying neurocircuitry. <i>Journal of Affective Disorders</i> , 2019, 245, 885-896.	4.7	20
41	Detecting genetic modifiers of spondyloepimetaphyseal dysplasia with joint laxity in the Caucasian Afrikaner community. <i>Human Molecular Genetics</i> , 2019, 28, 1053-1063.	3.1	4
42	A caseâ€“control study of risk factors for colorectal cancer in an African population. <i>European Journal of Cancer Prevention</i> , 2019, 28, 145-150.	2.1	25
43	Update on Inherited Retinal Disease in South Africa: Encouraging Diversity in Molecular Genetics. <i>Advances in Experimental Medicine and Biology</i> , 2019, , 257-261.	0.0	3
44	ASSAf consensus study on the ethical, legal and social implications of genetics and genomics in South Africa. <i>South African Journal of Science</i> , 2018, 114, .	0.8	6
45	Impact of Host Molecular Genetic Variations and HIV/HPV Co-infection on Cervical Cancer Progression: A Systematic review. <i>Oncomedicine</i> , 2018, 3, 82-93.	0.6	7
46	Dietary patterns and colorectal cancer risk in Zimbabwe: A population based case-control study. <i>Cancer Epidemiology</i> , 2018, 57, 33-38.	2.1	11
47	Dravet syndrome in South African infants: Tools for an early diagnosis. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2018, 62, 99-105.	2.3	11
48	Significant concordance of genetic variation that increases both the risk for obsessiveâ€“compulsive disorder and the volumes of the nucleus accumbens and putamen. <i>British Journal of Psychiatry</i> , 2018, 213, 430-436.	2.3	29
49	Clinical Application of Epilepsy Genetics in Africa: Is Now the Time?. <i>Frontiers in Neurology</i> , 2018, 9, .	2.5	27
50	A Systematic Review of Molecular Autopsy Studies in Sudden Infant Death Cases. <i>Journal of Pediatric Genetics</i> , 2018, 07, 143-149.	0.6	13
51	Whole-Exome Sequencing Identifies Biallelic IDH3A Variants as a Cause of Retinitis Pigmentosa Accompanied by Pseudocoloboma. <i>Ophthalmology</i> , 2017, 124, 992-1003.	4.9	31
52	The shifting epidemiology of colorectal cancer in sub-Saharan Africa. <i>The Lancet Gastroenterology and Hepatology</i> , 2017, 2, 377-383.	22.9	52
53	Genetic variation within <i>GRIN2B</i> in adolescents with alcohol use disorder may be associated with larger left posterior cingulate cortex volume. <i>Acta Neuropsychiatrica</i> , 2017, 29, 252-258.	2.3	7
54	A review of the optimisation of the use of formalin fixed paraffin embedded tissue for molecular analysis in a forensic post-mortem setting. <i>Forensic Science International</i> , 2017, 280, 181-187.	2.2	20

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55	Reconstructing Prehistoric African Population Structure. <i>Cell</i> , 2017, 171, 59-71.e21.	35.1	252
56	Whole-genome sequencing for an enhanced understanding of genetic variation among South Africans. <i>Nature Communications</i> , 2017, 8, .	14.1	84
57	New-onset diabetes after transplant: Incidence, risk factors and outcome. <i>South African Medical Journal</i> , 2017, 107, 791.	0.7	9
58	Ethical considerations in forensic genetics research on tissue samples collected post-mortem in Cape Town, South Africa. <i>BMC Medical Ethics</i> , 2017, 18, .	2.8	13
59	Osteogenesis imperfecta type 3 in South Africa: Causative mutations in FKBP10. <i>South African Medical Journal</i> , 2017, 107, 457.	0.7	11
60	Digitotalar dysmorphism: Molecular elucidation. <i>South African Medical Journal</i> , 2016, 106, 253.	0.7	2
61	Molecular Diagnosis of Inherited Retinal Diseases in Indigenous African Populations by Whole-Exome Sequencing. , 2016, 57, 6374.		19
62	Inherited retinal disorders in South Africa and the clinical impact of evolving technologies. <i>South African Medical Journal</i> , 2016, 106, 33.	0.7	5
63	The incidence and histo-pathological characteristics of colorectal cancer in a population based cancer registry in Zimbabwe. <i>Cancer Epidemiology</i> , 2016, 44, 96-100.	2.1	17
64	Minimum Information Required for a DMET Experiment Reporting. <i>Pharmacogenomics</i> , 2016, 17, 1533-1545.	1.8	6
65	Toward a Global Roadmap for Precision Medicine in Psychiatry: Challenges and Opportunities. <i>OMICS A Journal of Integrative Biology</i> , 2016, 20, 557-564.	1.9	15
66	Pharmacokinetics of rosuvastatin in 30 healthy Zimbabwean individuals of African ancestry. <i>British Journal of Clinical Pharmacology</i> , 2016, 82, 326-328.	2.8	13
67	Promoter region variation in NFE2L2 influences susceptibility to ototoxicity in patients exposed to high cumulative doses of cisplatin. <i>Pharmacogenomics Journal</i> , 2016, 17, 515-520.	3.1	21
68	Large Scale Genetic Research on Neuropsychiatric Disorders in African Populations is Needed. <i>EBioMedicine</i> , 2015, 2, 1259-1261.	10.0	30
69	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. <i>South African Medical Journal</i> , 2015, 105, 558.	0.7	48
70	A Founder Mutation in <i>MYO7A</i> Underlies a Significant Proportion of Usher Syndrome in Indigenous South Africans: Implications for the African Diaspora. , 2015, 56, 6671.		10
71	A Genomic Portrait of Haplotype Diversity and Signatures of Selection in Indigenous Southern African Populations. <i>PLoS Genetics</i> , 2015, 11, e1005052.	3.3	36
72	The Importance of G Protein-Coupled Receptor Kinase 4 (GRK4) in Pathogenesis of Salt Sensitivity, Salt Sensitive Hypertension and Response to Antihypertensive Treatment. <i>International Journal of Molecular Sciences</i> , 2015, 16, 5741-5749.	4.5	25

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73	Influence of light exposure during early life on the age of onset of bipolar disorder. <i>Journal of Psychiatric Research</i> , 2015, 64, 1-8.	3.1	40
74	Obesity, Aspirin, and Risk of Colorectal Cancer in Carriers of Hereditary Colorectal Cancer: A Prospective Investigation in the CAPP2 Study. <i>Journal of Clinical Oncology</i> , 2015, 33, 3591-3597.	17.1	98
75	Exploring researchers'™ experiences of working with a researcher-driven, population-specific community advisory board in a South African schizophrenia genomics study. <i>BMC Medical Ethics</i> , 2015, 16, .	2.8	26
76	Possible involvement of the circadian pathway in alcohol use disorder in a South African adolescent cohort. <i>Metabolic Brain Disease</i> , 2015, 31, 75-80.	3.0	3
77	Glutamatergic and HPA-axis pathway genes in bipolar disorder comorbid with alcohol- and substance use disorders. <i>Metabolic Brain Disease</i> , 2015, 31, 183-189.	3.0	7
78	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. <i>PLoS ONE</i> , 2014, 9, e100516.	2.5	83
79	High prevalence of cisplatin-induced ototoxicity in Cape Town, South Africa. <i>South African Medical Journal</i> , 2014, 104, 288.	0.7	32
80	Genetic Variation in Otos is Associated with Cisplatin-Induced Ototoxicity. <i>Pharmacogenomics</i> , 2014, 15, 1667-1676.	1.8	22
81	The BDNFp.Val66Met polymorphism, childhood trauma, and brain volumes in adolescents with alcohol abuse. <i>BMC Psychiatry</i> , 2014, 14, .	3.4	11
82	Ready to Put Metadata on the Post-2015 Development Agenda? Linking Data Publications to Responsible Innovation and Science Diplomacy. <i>OMICS A Journal of Integrative Biology</i> , 2014, 18, 1-9.	1.9	29
83	Would you terminate a pregnancy affected by sickle cell disease? Analysis of views of patients in Cameroon. <i>Journal of Medical Ethics</i> , 2014, 40, 615-620.	2.0	23
84	Psychosocial Burden of Sickle Cell Disease on Parents with an Affected Child in Cameroon. <i>Journal of Genetic Counseling</i> , 2014, 23, 192-201.	1.9	41
85	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. <i>Metabolic Brain Disease</i> , 2014, 29, 333-340.	3.0	5
86	Psychosocial Stressors of Sickle Cell Disease on Adult Patients in Cameroon. <i>Journal of Genetic Counseling</i> , 2014, 23, 948-956.	1.9	9
87	Coinheritance of sickle cell anemia and ð±thalassemia delays disease onset and could improve survival in cameroonian's patients (Sub-Saharan Africa). <i>American Journal of Hematology</i> , 2014, 89, 664-665.	6.3	14
88	A mutation in a splicing factor that causes retinitis pigmentosa has a transcriptome-wide effect on mRNA splicing. <i>BMC Research Notes</i> , 2014, 7, .	1.6	5
89	Fertility and apparent genetic anticipation in Lynch syndrome. <i>Familial Cancer</i> , 2014, 13, 369-374.	1.7	3
90	Relationship between sunlight and the age of onset of bipolar disorder: An international multisite study. <i>Journal of Affective Disorders</i> , 2014, 167, 104-111.	4.7	42

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91	African dawn. Nature, 2014, 517, 276-277.	40.1	4
92	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
93	Stargardt Disease: towards developing a model to predict phenotype. European Journal of Human Genetics, 2013, 21, 1173-1176.	3.1	11
94	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.	1.9	9
95	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.	2.0	16
96	Lynch Syndrome: Genetic Counselling of At-Risk Individuals and Families. , 2013, , 171-190.		0
97	Direct-to-consumer genetic testing: To test or not to test, that is the question. South African Medical Journal, 2013, 103, 510.	0.7	11
98	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.	0.7	0
99	Determining Ancestry Proportions in Complex Admixture Scenarios in South Africa Using a Novel Proxy Ancestry Selection Method. PLoS ONE, 2013, 8, e73971.	2.5	30
100	PXR and CAR single nucleotide polymorphisms influence plasma efavirenz levels in South African HIV/AIDS patients. BMC Medical Genetics, 2012, 13, .	2.1	44
101	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.	21.9	82
102	UCT's contribution to medical genetics in Africa - from the past into the future. South African Medical Journal, 2012, 102, 446.	0.7	11
103	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.	35.3	801
104	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.	0.5	5
105	UCT and CPGR join forces with international Pharmacogenomics initiative focussing on African diseases. EMBnet Journal, 2011, 17, 3.	0.6	0
106	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.	1.9	40
107	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.	1.7	15
108	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.	1.8	2

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109	Psychosis and relapse in bipolar disorder are related to GRM3, DAOA, and GRIN2B genotype. African Journal of Psychiatry, 2010, 13, .	0.0	16
110	Advancing public health genomics in Africa through prospective cohort studies. Journal of Epidemiology and Community Health, 2010, 64, 585-586.	3.4	8
111	Report on the 6th African Society of Human Genetics (AfSHG) Meeting, March 12-15, 2009, Yaounde, Cameroon. American Journal of Tropical Medicine and Hygiene, 2010, 83, 226-229.	2.2	5
112	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
113	Computational Analysis of Candidate Disease Genes and Variants for Salt-Sensitive Hypertension in Indigenous Southern Africans. PLoS ONE, 2010, 5, e12989.	2.5	12
114	Across Culture and Health Systems: Africa. , 2010, , 471-486.		0
115	Clinical Utility of the ABCR400 Microarray. JAMA Ophthalmology, 2009, 127, 549.	2.4	3
116	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.	2.3	38
117	6000 Aspirin prevents cancer in Lynch syndrome. European Journal of Cancer, Supplement, 2009, 7, 320-321.	0.5	6
118	Bipolar Disorder: Emotional Dysregulation and Neuronal Vulnerability. CNS Spectrums, 2009, 14, 122-126.	0.7	9
119	Neuropsychological status of bipolar I disorder: impact of psychosis. British Journal of Psychiatry, 2009, 194, 243-251.	2.3	53
120	Catechol-o-Methyltransferase Genotype and Childhood Trauma May Interact to Impact Schizotypal Personality Traits. Behavior Genetics, 2009, 40, 415-423.	1.8	31
121	Management of a South African family with retinitis pigmentosa—should potential therapy influence translational research protocols?. Journal of Ocular Biology, Diseases, and Informatics, 2008, 1, .	0.5	1
122	Genetic studies of African populations: an overview on disease susceptibility and response to vaccines and therapeutics. Human Genetics, 2008, 123, 557-598.	3.1	72
123	South Africa: from species cradle to genomic applications. Nature Reviews Genetics, 2008, 9, S19-S23.	19.1	36
124	Dysthymic and anxiety-related personality traits in bipolar spectrum illness. Journal of Affective Disorders, 2008, 109, 305-311.	4.7	35
125	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
126	S25. Results of the CAPP-2-trial (Aspirin and resistant starch) in HNPCC gene carriers. European Journal of Cancer, Supplement, 2008, 6, 25.	0.5	3

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127	The Human Variome Project. <i>Science</i> , 2008, 322, 861-862.	38.2	58
128	Effect of Aspirin or Resistant Starch on Colorectal Neoplasia in the Lynch Syndrome. <i>New England Journal of Medicine</i> , 2008, 359, 2567-2578.	25.5	251
129	The relationship between childhood abuse and dissociation. Is it influenced by catechol-O-methyltransferase (COMT) activity?. <i>International Journal of Neuropsychopharmacology</i> , 2008, 11, .	2.0	51
130	Prioritization of candidate disease genes for metabolic syndrome by computational analysis of its defining phenotypes. <i>Physiological Genomics</i> , 2008, 35, 55-64.	2.5	18
131	Genotype and Childhood Sexual Trauma Moderate Neurocognitive Performance: A Possible Role for Brain-Derived Neurotrophic Factor and Apolipoprotein E Variants. <i>Biological Psychiatry</i> , 2007, 62, 391-399.	1.7	58
132	Recommendations of the 2006 Human Variome Project meeting. <i>Nature Genetics</i> , 2007, 39, 433-436.	16.3	53
133	A linkage and family-based association analysis of a potential neurocognitive endophenotype of bipolar disorder. <i>NeuroMolecular Medicine</i> , 2007, 9, 101-116.	3.7	7
134	The extracolonic cancer spectrum in females with the common "South African" hMLH1 c.C1528T mutation. <i>Familial Cancer</i> , 2007, 7, 191-198.	1.7	13
135	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
136	Warriors Versus Worriers: The Role of COMT Gene Variants. <i>CNS Spectrums</i> , 2006, 11, 745-748.	0.7	129
137	Arrhythmogenic right ventricular cardiomyopathy type 6 (ARVC6): support for the locus assignment, narrowing of the critical region and mutation screening of three candidate genes. <i>BMC Medical Genetics</i> , 2006, 7, .	2.1	16
138	A brain-behaviour initiative for South Africa: the time is right. <i>Metabolic Brain Disease</i> , 2006, 21, 266-271.	3.0	10
139	Apolipoprotein E variants and cognition in healthy individuals: A critical opinion. <i>Brain Research Reviews</i> , 2006, 51, 125-135.	6.9	34
140	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. <i>Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis</i> , 2006, 602, 175-181.	1.8	31
141	Trends in Suicidology: Personality as an Endophenotype for Molecular Genetic Investigations. <i>PLoS Medicine</i> , 2006, 3, e107.	8.1	33
142	Neurocognitive Function as an Endophenotype for Genetic Studies of Bipolar Affective Disorder. <i>NeuroMolecular Medicine</i> , 2005, 7, 275-286.	3.7	48
143	A Mutation in the Variable Repeat Region of the Aggrecan Gene (AGC1) Causes a Form of Spondyloepiphyseal Dysplasia Associated with Severe, Premature Osteoarthritis. <i>American Journal of Human Genetics</i> , 2005, 77, 484-490.	6.8	130
144	Mutation Spectrum and Founder Chromosomes for the ABCA4 Gene in South African Patients with Stargardt Disease. , 2004, 45, 1705.		34

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145	Novel presenilin 1 mutation with profound neurofibrillary pathology in an indigenous Southern African family with early-onset Alzheimer's disease. <i>Brain</i> , 2004, 127, 133-142.	8.9	42
146	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.	7.7	97
147	The hereditary adult-onset ataxias in South Africa. <i>Journal of the Neurological Sciences</i> , 2003, 216, 47-54.	1.4	72
148	A rare homozygous rhodopsin splice-site mutation: the issue of when and whether to offer presymptomatic testing. <i>Ophthalmic Genetics</i> , 2003, 24, 225-232.	1.3	11
149	Retinal Degenerative Disorders in Southern Africa: A Molecular Genetic Approach. <i>Advances in Experimental Medicine and Biology</i> , 2003, , 35-40.	0.0	3
150	A computer-based register for inherited retinal dystrophies in Southern Africa. <i>Ophthalmic Genetics</i> , 2002, 23, 61-65.	1.3	6
151	Autosomal Dominant Craniometaphyseal Dysplasia Is Caused by Mutations in the Transmembrane Protein ANK. <i>American Journal of Human Genetics</i> , 2001, 68, 1321-1326.	6.8	160
152	LDL Receptor-Related Protein 5 (LRP5) Affects Bone Accrual and Eye Development. <i>Cell</i> , 2001, 107, 513-523.	35.1	1,929
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165	The authors reply. <i>Diseases of the Colon and Rectum</i> , 1998, 41, 1254-1255.	1.8	0
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