Raj Ramesar

List of Publications by Citations

224 papers	9,915 citations	45 h-index	94 g-index
248 ext. papers	11,299 ext. citations	6.8 avg, IF	5.45 L-index

#	Paper	IF	Citations
224	LDL receptor-related protein 5 (LRP5) affects bone accrual and eye development. *Cell*, **2001**, 107, 513-23	56.2	1827
223	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, 2081-7	40	715
222	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	36.3	332
221	Molecular genetics of pseudoxanthoma elasticum: type and frequency of mutations in ABCC6. *Human Mutation*, **2005**, 26, 235-48	4.7	304
220	Research capacity. Enabling the genomic revolution in Africa. *Science*, **2014**, 344, 1346-8	33.3	256
219	The molecular genetics of cognition: dopamine, COMT and BDNF. *Genes, Brain and Behavior*, **2006**, 5, 311-28	3.6	235
218	Genetic linkage of the Marfan syndrome, ectopia lentis, and congenital contractural arachnodactyly to the fibrillin genes on chromosomes 15 and 5. The International Marfan Syndrome Collaborative Study. *New England Journal of Medicine*, **1992**, 326, 905-9	59.2	232
217	Effect of aspirin or resistant starch on colorectal neoplasia in the Lynch syndrome. *New England Journal of Medicine*, **2008**, 359, 2567-78	59.2	228
216	Reconstructing Prehistoric African Population Structure. *Cell*, **2017**, 171, 59-71.e21	56.2	201
215	Mutations in the pre-mRNA splicing factor gene PRPC8 in autosomal dominant retinitis pigmentosa (RP13). *Human Molecular Genetics*, **2001**, 10, 1555-62	5.6	198
214	CDH23 mutation and phenotype heterogeneity: a profile of 107 diverse families with Usher syndrome and nonsyndromic deafness. *American Journal of Human Genetics*, **2002**, 71, 262-75	11	183
213	Autosomal dominant craniometaphyseal dysplasia is caused by mutations in the transmembrane protein ANK. *American Journal of Human Genetics*, **2001**, 68, 1321-6	11	158
212	Genomic structure and identification of novel mutations in usherin, the gene responsible for Usher syndrome type IIa. *American Journal of Human Genetics*, **2000**, 66, 1199-210	11	146
211	Neuropsychological dysfunction in bipolar affective disorder: a critical opinion. *Bipolar Disorders*, **2005**, 7, 216-35	3.8	142
210	Mutational bias provides a model for the evolution of Huntingtonâ disease and predicts a general increase in disease prevalence. *Nature Genetics*, **1994**, 7, 525-30	36.3	127
209	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, 484-90	11	111
208	Genetic variants implicated in personality: a review of the more promising candidates. *American Journal of Medical Genetics Part A*, **2004**, 131B, 20-32		110

207	Warriors versus worriers: the role of COMT gene variants. *CNS Spectrums*, **2006**, 11, 745-8	1.8	108
206	A new locus for autosomal dominant retinitis pigmentosa on the short arm of chromosome 17. *Human Molecular Genetics*, **1994**, 3, 915-8	5.6	100
205	Mutations of the gene encoding the transmembrane transporter protein ABC-C6 cause pseudoxanthoma elasticum. *Journal of Molecular Medicine*, **2000**, 78, 282-6	5.5	98
204	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, 6617-22	11.5	96
203	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, 1604-16	11	91
202	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	40	88
201	Neuropsychological task performance in bipolar spectrum illness: genetics, alcohol abuse, medication and childhood trauma. *Bipolar Disorders*, **2008**, 10, 479-94	3.8	79
200	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, 3591-7	2.2	71
199	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, 1242-9	21.7	70
198	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	3.7	69
197	The hereditary adult-onset ataxias in South Africa. *Journal of the Neurological Sciences*, **2003**, 216, 47-54	3.2	68
196	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	3.7	67
195	Surveillance colonoscopy improves survival in a cohort of subjects with a single mismatch repair gene mutation. *Colorectal Disease*, **2009**, 11, 126-30	2.1	67
194	Genetic studies of African populations: an overview on disease susceptibility and response to vaccines and therapeutics. *Human Genetics*, **2008**, 123, 557-98	6.3	66
193	Correlation between Waardenburg syndrome phenotype and genotype in a population of individuals with identified PAX3 mutations. *Human Genetics*, **1998**, 102, 499-506	6.3	60
192	Whole-genome sequencing for an enhanced understanding of genetic variation among South Africans. *Nature Communications*, **2017**, 8, 2062	17.4	53
191	Genetic heterogeneity of Usher syndrome: analysis of 151 families with Usher type I. *American Journal of Human Genetics*, **2000**, 67, 1569-74	11	53
190	Genotype and childhood sexual trauma moderate neurocognitive performance: a possible role for brain-derived neurotrophic factor and apolipoprotein E variants. *Biological Psychiatry*, **2007**, 62, 391-9	7.9	52

189	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	5.5	51
188	GENETICS. The Human Variome Project. *Science*, **2008**, 322, 861-2	33.3	50
187	Neuropsychological status of bipolar I disorder: impact of psychosis. *British Journal of Psychiatry*, **2009**, 194, 243-51	5.4	48
186	Heritable disorders of connective tissue. *Best Practice and Research in Clinical Rheumatology*, **2000**, 14, 345-61	5.3	48
185	Hypomanic, cyclothymic and hostile personality traits in bipolar spectrum illness: a family-based study. *Journal of Psychiatric Research*, **2008**, 42, 920-9	5.2	47
184	Personality: is it a viable endophenotype for genetic studies of bipolar affective disorder?. *Bipolar Disorders*, **2006**, 8, 322-37	3.8	47
183	Neurocognitive function as an endophenotype for genetic studies of bipolar affective disorder. *NeuroMolecular Medicine*, **2005**, 7, 275-86	4.6	47
182	Personality endophenotypes for bipolar affective disorder: a family-based genetic association analysis. *Genes, Brain and Behavior*, **2008**, 7, 869-76	3.6	45
181	Evidence for paternal imprinting in familial Beckwith-Wiedemann syndrome. *Journal of Medical Genetics*, **1992**, 29, 221-5	5.8	45
180	Resolving the dark matter of ABCA4 for 1054 Stargardt disease probands through integrated genomics and transcriptomics. *Genetics in Medicine*, **2020**, 22, 1235-1246	8.1	44
179	Genetics of schizophrenia in the South African Xhosa. *Science*, **2020**, 367, 569-573	33.3	44
178	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, 904-8	11	44
177	An eighth locus for autosomal dominant retinitis pigmentosa is linked to chromosome 17q. *Human Molecular Genetics*, **1995**, 4, 1459-62	5.6	43
176	PXR and CAR single nucleotide polymorphisms influence plasma efavirenz levels in South African HIV/AIDS patients. *BMC Medical Genetics*, **2012**, 13, 112	2.1	40
175	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, 536-46	5.5	40
174	Nucleotide sequence and expression of a cloned Thiobacillus ferrooxidans recA gene in Escherichia coli. *Gene*, **1989**, 78, 1-8	3.8	39
173	Relationship between sunlight and the age of onset of bipolar disorder: an international multisite study. *Journal of Affective Disorders*, **2014**, 167, 104-11	6.6	37
172	Capturing all disease-causing mutations for clinical and research use: toward an effortless system for the Human Variome Project. *Genetics in Medicine*, **2009**, 11, 843-9	8.1	37

171	Stickler-like syndrome due to a dominant negative mutation in the COL2A1 gene. *American Journal of Medical Genetics Part A*, **1998**, 80, 6-11		37
170	Lateralization of hand skill in bipolar affective disorder. *Genes, Brain and Behavior*, **2007**, 6, 698-705	3.6	36
169	Mutation spectrum and founder chromosomes for the ABCA4 gene in South African patients with Stargardt disease. *Investigative Ophthalmology and Visual Science*, **2004**, 45, 1705-11		36
168	Influence of light exposure during early life on the age of onset of bipolar disorder. *Journal of Psychiatric Research*, **2015**, 64, 1-8	5.2	34
167	The relationship between childhood abuse and dissociation. Is it influenced by catechol-O-methyltransferase (COMT) activity?. *International Journal of Neuropsychopharmacology*, **2008**, 11, 149-61	5.8	34
166	Apolipoprotein E variants and cognition in healthy individuals: a critical opinion. *Brain Research Reviews*, **2006**, 51, 125-35		34
165	Trends in suicidology: personality as an endophenotype for molecular genetic investigations. *PLoS Medicine*, **2006**, 3, e107	11.6	32
164	A genomic portrait of haplotype diversity and signatures of selection in indigenous southern African populations. *PLoS Genetics*, **2015**, 11, e1005052	6	31
163	Emergency medical services in India: the present and future. *Prehospital and Disaster Medicine*, **2014**, 29, 307-10	0.8	31
162	Psychosocial burden of sickle cell disease on parents with an affected child in Cameroon. *Journal of Genetic Counseling*, **2014**, 23, 192-201	2.5	31
161	Novel presenilin 1 mutation with profound neurofibrillary pathology in an indigenous Southern African family with early-onset Alzheimer's disease. *Brain*, **2004**, 127, 133-42	11.2	31
160	The shifting epidemiology of colorectal cancer in sub-Saharan Africa. *The Lancet Gastroenterology and Hepatology*, **2017**, 2, 377-383	18.8	30
159	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, 558-63	1.5	30
158	Dysthymic and anxiety-related personality traits in bipolar spectrum illness. *Journal of Affective Disorders*, **2008**, 109, 305-11	6.6	30
157	Determining ancestry proportions in complex admixture scenarios in South Africa using a novel proxy ancestry selection method. *PLoS ONE*, **2013**, 8, e73971	3.7	29
156	Catechol-o-methyltransferase genotype and childhood trauma may interact to impact schizotypal personality traits. *Behavior Genetics*, **2010**, 40, 415-23	3.2	29
155	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, 175-81	3.3	28
154	Prognosis for splicing factor PRPF8 retinitis pigmentosa, novel mutations and correlation between human and yeast phenotypes. *Human Mutation*, **2010**, 31, E1361-76	4.7	27

153	Diverse prevalence of large deletions within the OA1 gene in ocular albinism type 1 patients from Europe and North America. *Human Genetics*, **2001**, 108, 51-4	6.3	27
152	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	3.8	26
151	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, 272-6		26
150	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	11	26
149	Whole-Exome Sequencing Identifies Biallelic IDH3A Variants as a Cause of Retinitis Pigmentosa Accompanied by Pseudocoloboma. *Ophthalmology*, **2017**, 124, 992-1003	7.3	25
148	Large Scale Genetic Research on Neuropsychiatric Disorders in African Populations is Needed. *EBioMedicine*, **2015**, 2, 1259-61	8.8	25
147	X-linked ocular albinism and sensorineural deafness: linkage to Xp22.3. *Genomics*, **1993**, 18, 444-5	4.3	25
146	Exploring researchersPexperiences of working with a researcher-driven, population-specific community advisory board in a South African schizophrenia genomics study. *BMC Medical Ethics*, **2015**, 16, 45	2.9	24
145	Influence of birth cohort on age of onset cluster analysis in bipolar I disorder. *European Psychiatry*, **2015**, 30, 99-105	6	24
144	Surgery for colonic cancer in HNPCC: total vs segmental colectomy. *Colorectal Disease*, **2011**, 13, 1395-9	2.1	24
143	Significant concordance of genetic variation that increases both the risk for obsessive-compulsive disorder and the volumes of the nucleus accumbens and putamen. *British Journal of Psychiatry*, **2018**, 213, 430-436	5.4	24
142	Capacity-building in human genetics for developing countries: initiatives and perspectives in sub-Saharan Africa. *Public Health Genomics*, **2010**, 13, 492-4	1.9	23
141	Identification of new cases of early-onset colorectal cancer with an MLH1 epimutation in an ethnically diverse South African cohort. *Clinical Genetics*, **2011**, 80, 428-34	4	22
140	Hearing impairment and pigmentary disturbance. *Annals of the New York Academy of Sciences*, **1991**, 630, 152-66	6.5	22
139	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, 334-9	2.6	21
138	Recommendations for genetic variation data capture in developing countries to ensure a comprehensive worldwide data collection. *Human Mutation*, **2011**, 32, 2-9	4.7	21
137	Expanded CAG repeats in spinocerebellar ataxia (SCA1) segregate with distinct haplotypes in South african families. *Human Genetics*, **1997**, 100, 131-7	6.3	21
136	Retinitis pigmentosa in southern Africa. *Clinical Genetics*, **1993**, 44, 232-5	4	21

135	Prioritization of candidate disease genes for metabolic syndrome by computational analysis of its defining phenotypes. *Physiological Genomics*, **2008**, 35, 55-64	3.6	21
134	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, 511-4	5.8	21
133	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, 5741-9	6.3	20
132	Spondyloepimetaphyseal dysplasia with joint laxity (Beighton type); mutation analysis in eight affected South African families. *Clinical Genetics*, **2015**, 87, 492-5	4	20
131	Would you terminate a pregnancy affected by sickle cell disease? Analysis of views of patients in Cameroon. *Journal of Medical Ethics*, **2014**, 40, 615-20	2.5	20
130	Toward a Global Roadmap for Precision Medicine in Psychiatry: Challenges and Opportunities. *OMICS A Journal of Integrative Biology*, **2016**, 20, 557-564	3.8	18
129	The burden of sickle cell disease in Cape Town. *South African Medical Journal*, **2012**, 102, 752-4	1.5	18
128	Retinitis pigmentosa locus on 17q (RP17): fine localization to 17q22 and exclusion of the PDEG and TIMP2 genes. *Human Genetics*, **1997**, 101, 13-7	6.3	18
127	High prevalence of cisplatin-induced ototoxicity in Cape Town, South Africa. *South African Medical Journal*, **2014**, 104, 288-91	1.5	17
126	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	17
125	Genetic variation in Otos is associated with cisplatin-induced ototoxicity. *Pharmacogenomics*, **2014**, 15, 1667-76	2.6	16
124	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	4.3	16
123	A splice junction mutation in PAX3 causes Waardenburg syndrome in a South African family. *Human Molecular Genetics*, **1994**, 3, 197-8	5.6	16
122	Association between solar insolation and a history of suicide attempts in bipolar I disorder. *Journal of Psychiatric Research*, **2019**, 113, 1-9	5.2	15
121	Coinheritance of sickle cell anemia and Ethalassemia delays disease onset and could improve survival in CameroonianS patients (Sub-Saharan Africa). *American Journal of Hematology*, **2014**, 89, 664-5	7.1	15
120	Promoter region variation in NFE2L2 influences susceptibility to ototoxicity in patients exposed to high cumulative doses of cisplatin. *Pharmacogenomics Journal*, **2017**, 17, 515-520	3.5	15
119	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, 332-8	5.3	15
118	Molecular investigation of familial Beckwith-Wiedemann syndrome: a model for paternal imprinting. *European Journal of Human Genetics*, **1993**, 1, 109-13	5.3	15

117	Association of Sex With Frequent and Mild ABCA4 Alleles in Stargardt Disease. *JAMA Ophthalmology*, **2020**, 138, 1035-1042	3.9	15
116	A case-control study of risk factors for colorectal cancer in an African population. *European Journal of Cancer Prevention*, **2019**, 28, 145-150	2	15
115	Solar insolation in springtime influences age of onset of bipolar I disorder. *Acta Psychiatrica Scandinavica*, **2017**, 136, 571-582	6.5	14
114	Beyond the Caster Semenya controversy: the case of the use of genetics for gender testing in sport. *Journal of Genetic Counseling*, **2010**, 19, 545-8	2.5	14
113	The extracolonic cancer spectrum in females with the common South African hMLH1 c.C1528T mutation. *Familial Cancer*, **2008**, 7, 191-8	3	14
112	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	6.6	14
111	Concordance of genetic variation that increases risk for tourette syndrome and that influences its underlying neurocircuitry. *Translational Psychiatry*, **2019**, 9, 120	8.6	13
110	Mobile colonoscopic surveillance provides quality care for hereditary nonpolyposis colorectal carcinoma families in South Africa. *Colorectal Disease*, **2007**, 9, 509-14	2.1	13
109	Molecular genetics improves the management of hereditary non-polyposis colorectal cancer. *South African Medical Journal*, **2000**, 90, 709-14	1.5	13
108	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	3.1	12
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, 357-63	3	12
106	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, 1250-3; discussion 1253-5	3.1	12
105	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	4.5	11
104	Piebaldism: an autonomous autosomal dominant entity. *Clinical Genetics*, **1991**, 39, 330-7	4	11
103	A rare homozygous rhodopsin splice-site mutation: the issue of when and whether to offer presymptomatic testing. *Ophthalmic Genetics*, **2003**, 24, 225-32	1.2	11
102	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	3.7	11
101	Molecular Diagnosis of Inherited Retinal Diseases in Indigenous African Populations by Whole-Exome Sequencing **2016**, 57, 6374-6381		11
100	Toward colorectal cancer control in Africa. *International Journal of Cancer*, **2016**, 138, 1033-4	7.5	11

99	Psychosocial stressors of sickle cell disease on adult patients in Cameroon. *Journal of Genetic Counseling*, **2014**, 23, 948-56	2.5	10
98	Predictive genetic testing in children: constitutional mismatch repair deficiency cancer predisposing syndrome. *Journal of Genetic Counseling*, **2014**, 23, 147-55	2.5	10
97	Stargardt disease: towards developing a model to predict phenotype. *European Journal of Human Genetics*, **2013**, 21, 1173-6	5.3	10
96	Human Variome Project country nodes: documenting genetic information within a country. *Human Mutation*, **2012**, 33, 1513-9	4.7	10
95	Advancing public health genomics in Africa through prospective cohort studies. *Journal of Epidemiology and Community Health*, **2010**, 64, 585-6	5.1	10
94	Cancer risk in a cohort of subjects carrying a single mismatch repair gene mutation. *Familial Cancer*, **2009**, 8, 519-23	3	10
93	Autosomal dominant (Beukes) premature degenerative osteoarthropathy of the hip joint unlinked to COL2A1. *American Journal of Medical Genetics Part A*, **1994**, 53, 348-51		10
92	Brachydactylous dwarfs of Mseleni. *American Journal of Medical Genetics Part A*, **1993**, 46, 636-40		10
91	Low-coverage sequencing cost-effectively detects known and novel variation in underrepresented populations. *American Journal of Human Genetics*, **2021**, 108, 656-668	11	10
90	The incidence and histo-pathological characteristics of colorectal cancer in a population based cancer registry in Zimbabwe. *Cancer Epidemiology*, **2016**, 44, 96-100	2.8	10
89	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	2.6	9
88	Clinical Application of Epilepsy Genetics in Africa: Is Now the Time?. *Frontiers in Neurology*, **2018**, 9, 276	4.1	9
87	The BDNF p.Val66Met polymorphism, childhood trauma, and brain volumes in adolescents with alcohol abuse. *BMC Psychiatry*, **2014**, 14, 328	4.2	9
86	UCTB contribution to medical genetics in Africa - from the past into the future. *South African Medical Journal*, **2012**, 102, 446-8	1.5	9
85	Psychosis and relapse in bipolar disorder are related to GRM3, DAOA, and GRIN2B genotype. *African Journal of Psychiatry*, **2010**, 13, 297-301		9
84	Bipolar disorder: emotional dysregulation and neuronal vulnerability. *CNS Spectrums*, **2009**, 14, 122-6	1.8	9
83	Beals syndrome: clinical and molecular investigations in a kindred of Indian descent. *Clinical Genetics*, **1991**, 39, 181-8	4	9
82	Immunohistochemistry detects mismatch repair gene defects in colorectal cancer. *Colorectal Disease*, **2006**, 8, 411-7	2.1	9

81	Arg120stop nonsense mutation in the RP2 gene: mutational hotspot and germ line mosaicism?. *Clinical Genetics*, **2004**, 65, 7-10	4	9
80	Polygenic risk for schizophrenia and associated brain structural changes: A systematic review. *Comprehensive Psychiatry*, **2019**, 88, 77-82	7.3	9
79	Pharmacokinetics of rosuvastatin in 30 healthy Zimbabwean individuals of African ancestry. *British Journal of Clinical Pharmacology*, **2016**, 82, 326-8	3.8	8
78	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, 383-7	3.5	8
77	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, 634-8	5.8	8
76	Dietary patterns and colorectal cancer risk in Zimbabwe: A population based case-control study. *Cancer Epidemiology*, **2018**, 57, 33-38	2.8	8
75	Mismatch repair deficiency in colorectal cancer patients in a low-incidence area. *South African Journal of Surgery*, **2013**, 51, 16-21	0.4	8
74	Osteogenesis imperfecta type 3 in South Africa: Causative mutations in FKBP10. *South African Medical Journal*, **2017**, 107, 457-462	1.5	7
73	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, 125-37	2.5	7
72	Direct-to-consumer genetic testing: to test or not to test, that is the question. *South African Medical Journal*, **2013**, 103, 510-2	1.5	7
71	Computational analysis of candidate disease genes and variants for salt-sensitive hypertension in indigenous Southern Africans. *PLoS ONE*, **2010**, 5, e12989	3.7	7
70	Association of Novel Locus With Rheumatic Heart Disease in Black African Individuals: Findings From the RHDGen Study. *JAMA Cardiology*, **2021**, 6, 1000-1011	16.2	7
69	Novel variants in the hotspot region of RP1 in South African patients with retinitis pigmentosa. *Molecular Vision*, **2006**, 12, 177-83	2.3	7
68	Trends in Suicide Mortality in South Africa, 1997 to 2016. *International Journal of Environmental Research and Public Health*, **2020**, 17,	4.6	6
67	Ethical considerations in forensic genetics research on tissue samples collected post-mortem in Cape Town, South Africa. *BMC Medical Ethics*, **2017**, 18, 66	2.9	6
66	New-onset diabetes after transplant: Incidence, risk factors and outcome. *South African Medical Journal*, **2017**, 107, 791-796	1.5	6
65	A Founder Mutation in MYO7A Underlies a Significant Proportion of Usher Syndrome in Indigenous South Africans: Implications for the African Diaspora **2015**, 56, 6671-8		6
64	Clinical utility of the ABCR400 microarray: basing a genetic service on a commercial gene chip. *JAMA Ophthalmology*, **2009**, 127, 549-54		6

63	A linkage and family-based association analysis of a potential neurocognitive endophenotype of bipolar disorder. *NeuroMolecular Medicine*, **2007**, 9, 101-16	4.6	6
62	Infrequent and low expression of cancer-testis antigens located on the X chromosome in colorectal cancer: implications for immunotherapy in South African populations. *Biotechnology Journal*, **2008**, 3, 1417-23	5.6	6
61	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, 833-8		6
60	Inherited retinal disorders in South Africa and the clinical impact of evolving technologies. *South African Medical Journal*, **2016**, 106, S33-7	1.5	6
59	Impact of Host Molecular Genetic Variations and HIV/HPV Co-infection on Cervical Cancer Progression: A Systematic review. *Oncomedicine*, **2018**, 3, 82-93	1	6
58	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	1.8	5
57	Glutamatergic and HPA-axis pathway genes in bipolar disorder comorbid with alcohol- and substance use disorders. *Metabolic Brain Disease*, **2016**, 31, 183-9	3.9	5
56	A Systematic Review of Molecular Autopsy Studies in Sudden Infant Death Cases. *Journal of Pediatric Genetics*, **2018**, 7, 143-149	0.7	5
55	A mutation in a splicing factor that causes retinitis pigmentosa has a transcriptome-wide effect on mRNA splicing. *BMC Research Notes*, **2014**, 7, 401	2.3	5
54	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	2.9	5
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