Maritha J Kotze

List of Publications by Year
in descending order

107 papers	3,295 citations	257450 24 h-index	161849 54 g-index
112 all docs	112 docs citations	112 times ranked	5711 citing authors

#	Article	IF	Citations
1	The Genetic Structure and History of Africans and African Americans. Science, 2009, 324, 1035-1044.	12.6	1,267
2	Genetic Origins of Lactase Persistence and the Spread of Pastoralism in Africa. American Journal of Human Genetics, 2014, 94, 496-510.	6.2	174
3	A Third Major Locus for Autosomal Dominant Hypercholesterolemia Maps to 1p34.1-p32. American Journal of Human Genetics, 1999, 64, 1378-1387.	6.2	154
4	The UMD-LDLR database: additions to the software and 490 new entries to the database. Human Mutation, 2002, 20, 81-87.	2.5	105
5	APRI: a simple bedside marker for advanced fibrosis that can avoid liver biopsy in patients with NAFLD/NASH. South African Medical Journal, 2011, 101, 477-80.	0.6	94
6	Inhibition of cholesterol synthesis by atorvastatin in homozygous familial hypercholesterolaemia. Atherosclerosis, 2000, 150, 421-428.	0.8	85
7	Fat Mass and Obesity-Associated (FTO) Gene Polymorphisms Are Associated with Physical Activity, Food Intake, Eating Behaviors, Psychological Health, and Modeled Change in Body Mass Index in Overweight/Obese Caucasian Adults. Nutrients, 2014, 6, 3130-3152.	4.1	70
8	Analysis of the NRAMP1 Gene Implicated in Iron Transport: Association with Multiple Sclerosis and Age Effects. Blood Cells, Molecules, and Diseases, 2001, 27, 44-53.	1.4	61
9	A Reverse-Hybridization Assay for the Rapid and Simultaneous Detection of NineHFEGene Mutations. Genetic Testing and Molecular Biomarkers, 2000, 4, 121-124.	1.7	50
10	Recent Origin and Spread of a Common Lithuanian Mutation, G197del LDLR, Causing Familial Hypercholesterolemia: Positive Selection Is Not Always Necessary to Account for Disease Incidence among Ashkenazi Jews. American Journal of Human Genetics, 2001, 68, 1172-1188.	6.2	46
11	Expression of the SLC11A1 (NRAMP1) 5â€²-(GT)n repeat: Opposite effect in the presence of â^'237Câ†'T. Blood Cells, Molecules, and Diseases, 2004, 33, 45-50.	1.4	45
12	Concentrations of the atherogenic Lp(a) are elevated in familial hypercholesterolaemia: a sib pair and family analysis. European Journal of Human Genetics, 1998, 6, 50-60.	2.8	41
13	Ethnic Variation and In Vivo Effects of the â^'93tâ†'g Promoter Variant in the Lipoprotein Lipase Gene. Arteriosclerosis, Thrombosis, and Vascular Biology, 1997, 17, 2672-2678.	2.4	40
14	Analysis of genes implicated in iron regulation in individuals presenting with primary iron overload. Human Genetics, 2004, 115, 409-417.	3.8	40
15	Iron and the folate-vitamin B12-methylation pathway in multiple sclerosis. Metabolic Brain Disease, 2006, 21, 117-133.	2.9	40
16	A framework for tiered informed consent for health genomic research in Africa. Nature Genetics, 2019, 51, 1566-1571.	21.4	40
17	Mutations in the Gene for Lipoprotein Lipase. Arteriosclerosis, Thrombosis, and Vascular Biology, 1995, 15, 1704-1712.	2.4	39
18	The conundrum of iron in multiple sclerosis â€“ time for an individualised approach. Metabolic Brain Disease, 2012, 27, 239-253.	2.9	34

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19	Pathology supported genetic testing and treatment of cardiovascular disease in middle age for prevention of Alzheimerâ€™s disease. Metabolic Brain Disease, 2012, 27, 255-266.	2.9	30
20	Analysis of the three common mutations in the CARD15 gene (R702W, G908R and 1007fs) in South African colored patients with inflammatory bowel disease. Molecular and Cellular Probes, 2005, 19, 278-281.	2.1	29
21	The fat mass and obesity-associated FTO rs9939609 polymorphism is associated with elevated homocysteine levels in patients with multiple sclerosis screened for vascular risk factors. Metabolic Brain Disease, 2014, 29, 409-19.	2.9	29
22	Genomic medicine and risk prediction across the disease spectrum. Critical Reviews in Clinical Laboratory Sciences, 2015, 52, 120-137.	6.1	29
23	Intrafamilial variability in the clinical expression of familial hypercholesterolemia: importance of risk factor determination for genetic counselling. Clinical Genetics, 2008, 43, 295-299.	2.0	27
24	Efficacy of vitamin E compared with either simvastatin or atorvastatin in preventing the progression of atherosclerosis in homozygous familial hypercholesterolemia. American Journal of Cardiology, 1999, 84, 1344-1346.	1.6	25
25	CpG hotspot mutations at the LDL receptor locus are a frequent cause of familial hypercholesterolemia among South African Indians. Clinical Genetics, 1997, 51, 394-398.	2.0	25
26	Founder mutations in the LDL receptor gene contribute significantly to the familial hypercholesterolemia phenotype in the indigenous South African population of mixed ancestry. Clinical Genetics, 1999, 55, 340-345.	2.0	24
27	Somatic mutations of theAPC,KRAS, andTP53 genes in nonpolypoid colorectal adenomas. Genes Chromosomes and Cancer, 2000, 27, 202-208.	2.8	24
28	Mutational and genetic origin of LDL receptor gene mutations detected in both Belgian and Dutch familial hypercholesterolemics. Human Genetics, 1997, 100, 266-270.	3.8	23
29	Novel mutations identified using a comprehensive CCR5-denaturing gradient gel electrophoresis assay. Aids, 2001, 15, 171-177.	2.2	23
30	The â^'237Câ†'T promoter polymorphism of the SLC11A1 gene is associated with a protective effect in relation to inflammatory bowel disease in the South African population. International Journal of Colorectal Disease, 2006, 21, 402-408.	2.2	23
31	Novel RET mutations in Hirschsprung's disease patients from the diverse South African population. European Journal of Human Genetics, 2001, 9, 419-423.	2.8	22
32	Analysis of viral and genetic factors in South African patients with multiple sclerosis. Metabolic Brain Disease, 2006, 21, 156-162.	2.9	22
33	Significance of dietary folate intake, homocysteine levels and MTHFR 677 C>T genotyping in South African patients diagnosed with depression: test development for clinical application. Metabolic Brain Disease, 2014, 29, 377-384.	2.9	19
34	Fractional anisotropy of white matter, disability and blood iron parameters in multiple sclerosis. Metabolic Brain Disease, 2018, 33, 545-557.	2.9	19
35	Association of functional polymorphisms of SLC11A1 with risk of esophageal cancer in the South African Colored population. Cancer Genetics and Cytogenetics, 2005, 159, 48-52.	1.0	18
36	The tumor genetics of acral melanoma: What should a dermatologist know?. JAAD International, 2020, 1, 135-147.	2.2	18

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37	Two novel point mutations causing receptor-negative familial hypercholesterolemia in a South African Indian homozygote. Atherosclerosis, 1996, 125, 111-119.	0.8	17
38	Multiple sclerosis, porphyria-like symptoms, and a history of iron deficiency anemia in a family of Scottish descent. , 1999, 86, 194-196.		15
39	Pathology-supported genetic testing directed at shared disease pathways for optimized health in later life. Personalized Medicine, 2013, 10, 497-507.	1.5	15
40	Simultaneous Detection of Multiple Familial Hypercholesterolemia Mutations Facilitates an Improved Diagnostic Service in South African Patients at High Risk of Cardiovascular Disease. Molecular Diagnosis and Therapy, 2003, 7, 169-174.	1.1	14
41	Exome Sequencing in a Family with Luminal-Type Breast Cancer Underpinned by Variation in the Methylation Pathway. International Journal of Molecular Sciences, 2017, 18, 467.	4.1	14
42	Lack of clinical manifestation of hereditary haemochromatosis in South African patients with multiple sclerosis. Metabolic Brain Disease, 2006, 21, 105-116.	2.9	13
43	Globally Rare BRCA2 Variants With Founder Haplotypes in the South African Population: Implications for Point-of-Care Testing Based on a Single-Institution BRCA1/2 Next-Generation Sequencing Study. Frontiers in Oncology, 2020, 10, 619469.	2.8	13
44	Detection of two point mutations causing familial defective apolipoprotein B-100 by heteroduplex analysis. Molecular and Cellular Probes, 1994, 8, 513-518.	2.1	12
45	DNA screening of hyperlipidemic Afrikaners for familial hypercholesterolemia. Clinical Genetics, 1992, 42, 43-46.	2.0	12
46	MammaPrint Pre-screen Algorithm (MPA) reduces chemotherapy in patients with early-stage breast cancer. South African Medical Journal, 2013, 103, 522.	0.6	12
47	Pioneering Informed Consent for Return of Research Results to Breast Cancer Patients Facing Barriers to Implementation of Genomic Medicine: The Kenyan BRCA1/2 Testing Experience Using Whole Exome Sequencing. Frontiers in Genetics, 2020, 11, 170.	2.3	12
48	A de novo duplication in the low density lipoprotein receptor gene. Human Mutation, 1995, 6, 181-183.	2.5	11
49	Significance of Linkage Disequilibrium between Mutation C282Y and a MseI Polymorphism in Population Screening and DNA Diagnosis of Hemochromatosis. Blood Cells, Molecules, and Diseases, 1999, 25, 250-252.	1.4	11
50	Impact of MammaPrint on Clinical Decision-Making in South African Patients with Early-Stage Breast Cancer. Breast Journal, 2016, 22, 442-446.	1.0	11
51	Towards AI-Enabled Multimodal Diagnostics and Management of COVID-19 and Comorbidities in Resource-Limited Settings. Informatics, 2021, 8, 63.	3.9	11
52	The relationship between measurement of in vivo brain glutamate and markers of iron metabolism: A proton magnetic resonance spectroscopy study in healthy adults. European Journal of Neuroscience, 2020, 51, 984-990.	2.6	10
53	Human whole genome sequencing in South Africa. Scientific Reports, 2021, 11, 606.	3.3	10
54	A 3-basepair deletion in repeat 1 of the LDL receptor promoter reduces transcriptional activity in a South African Pedi. Journal of Lipid Research, 1998, 39, 1021-1024.	4.2	10

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55	A View on Genomic Medicine Activities in Africa: Implications for Policy. Frontiers in Genetics, 2022, 13, 769919.	2.3	10
56	Mutation analysis in familial hypercholesterolemia patients of different ancestries: identification of three novel LDLR gene mutations. Molecular and Cellular Probes, 1998, 12, 149-152.	2.1	9
57	CYP2D6 genotyping and use of antidepressants in breast cancer patients: test development for clinical application. Metabolic Brain Disease, 2012, 27, 319-326.	2.9	9
58	Identification of an iron-responsive subtype in two children diagnosed with relapsing-remitting multiple sclerosis using whole exome sequencing. Molecular Genetics and Metabolism Reports, 2019, 19, 100465.	1.1	9
59	Detection of p53 gene mutations in oral squamous cell carcinomas of a black African population sample. Human Mutation, 1998, 11, 39-44.	2.5	8
60	Familial hypercholesterolemia: potential diagnostic value of mutation screening in a pediatric population of South Africa. Clinical Genetics, 1998, 54, 74-78.	2.0	8
61	Postmenopausal Breast Cancer, Aromatase Inhibitors, and Bone Health: What the Surgeon Should Know. World Journal of Surgery, 2016, 40, 2149-2156.	1.6	8
62	Composite prognostic models across the non-alcoholic fatty liver disease spectrum: Clinical application in developing countries. World Journal of Hepatology, 2015, 7, 1192.	2.0	8
63	Pathology-supported genetic testing as a method for disability prevention in multiple sclerosis (MS). Part I. Targeting a metabolic model rather than autoimmunity. Metabolic Brain Disease, 2021, 36, 1151-1167.	2.9	7
64	Clinical Relevance of Apolipoprotein E Genotyping Based on a Family History of Alzheimer's Disease. Current Alzheimer Research, 2015, 12, 210-217.	1.4	7
65	Effect of rapamycin on hepatic osteodystrophy in rats with portasystemic shunting. World Journal of Gastroenterology, 2006, 12, 4504.	3.3	7
66	Analysis of two mutations in the MTHFR gene associated with mild hyperhomocysteinaemia--heterogeneous distribution in the South African population. South African Medical Journal, 2002, 92, 464-7.	0.6	7
67	Two novel frameshift mutations in the low density lipoprotein receptor gene generated by endogenous sequence-directed mechanisms. Human Genetics, 1995, 96, 401-406.	3.8	6
68	Screening South African familial adenomatous polyposis families for the five-nucleotide deletion at codon 1309 of the APC gene. Molecular and Cellular Probes, 1995, 9, 49-51.	2.1	6
69	Application of advanced molecular technology in the diagnosis and management of genetic disorders in South Africa. South African Medical Journal, 2016, 106, 114.	0.6	6
70	Pioneering BRCA1/2 Point-Of-Care Testing for Integration of Germline and Tumor Genetics in Breast Cancer Risk Management: A Vision for the Future of Translational Pharmacogenomics. Frontiers in Oncology, 2021, 11, 619817.	2.8	6
71	Health Claims on the Benefits of Moderate Alcohol Consumption in Relation to Genetic Profiles. Journal of Wine Research, 2011, 22, 123-129.	1.5	5
72	Incorporating microarray assessment of HER2 status in clinical practice supports individualised therapy in early-stage breast cancer. Breast, 2015, 24, 137-142.	2.2	5

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73	Multiple sclerosis-like diagnosis as a complication of previously treated malaria in an iron and vitamin D deficient Nigerian patient. Metabolic Brain Disease, 2016, 31, 197-204.	2.9	5
74	Reclassification of early stage breast cancer into treatment groups by combining the use of immunohistochemistry and microarray analysis. South African Journal of Science, 2019, 115, .	0.7	5
75	CYP19A1 rs10046 Pharmacogenetics in Postmenopausal Breast Cancer Patients Treated with Aromatase Inhibitors: One-year Follow-up. Current Pharmaceutical Design, 2020, 26, 6007-6012.	1.9	5
76	Two novel and two known low-density lipoprotein receptor gene mutations in German patients with familial hypercholesterolemia. Human Mutation, 1998, 11, S232-S233.	2.5	4
77	Multiple APC mutations in sporadic flat colorectal adenomas. European Journal of Human Genetics, 1999, 7, 928-932.	2.8	4
78	Cardiovascular genetic assessment and treatment in middle age to reduce the risk of heart disease and dementia in old age. South African Family Practice: Official Journal of the South African Academy of Family Practice/Primary Care, 2006, 48, 53-54.	0.6	4
79	Apolipoprotein E ε-4 as a genetic determinant of Alzheimer’s disease heterogeneity. Degenerative Neurological and Neuromuscular Disease, 2015, 5, 9.	1.3	4
80	Apolipoprotein E genotyping and questionnaire-based assessment of lifestyle risk factors in dyslipidemic patients with a family history of Alzheimerâ€™s disease: test development for clinical application. Metabolic Brain Disease, 2016, 31, 213-224.	2.9	4
81	Baseline bone health status in multi-ethnic South African postmenopausal breast cancer patients at initiation of aromatase inhibitor therapy: A descriptive study. PLoS ONE, 2019, 14, e0214153.	2.5	4
82	Pathology-supported genetic testing as a method for disability prevention in multiple sclerosis (MS). Part II. Insights from two MS cases. Metabolic Brain Disease, 2021, 36, 1169-1181.	2.9	4
83	Pharmacogenetics of Aromatase Inhibitors in Endocrine Responsive Breast Cancer: Lessons Learnt from Tamoxifen and CYP2D6 Genotyping. Anti-Cancer Agents in Medicinal Chemistry, 2018, 17, 1805-1813.	1.7	4
84	Multiple mutations underlying familial hypercholesterolemia in the South African population. Human Genetics, 1989, 83, 67-70.	3.8	3
85	A nonsense mutation (Arg-196-Term) in exon 6 of the human TP53 gene identified in small cell lung carcinoma. Molecular and Cellular Probes, 1996, 10, 393-395.	2.1	3
86	Identification of a novel single base insertion in the adenomatous polyposis coli gene. Human Mutation, 1996, 7, 68-69.	2.5	3
87	Novel stop mutation causing familial hypercholesterolemia in a Costa Rican family. Molecular and Cellular Probes, 1997, 11, 457-458.	2.1	3
88	MedPed FH: a paradigm for other common monogenic diseases in South Africa. Atherosclerosis, 1999, 144, 467-468.	0.8	3
89	Overrepresentation of the founder PPOX gene mutation R59W in a South African patient with severe clinical manifestation of porphyria. Experimental Dermatology, 2005, 14, 50-55.	2.9	3
90	Clinical Overestimation of HER2 Positivity in Early Estrogen and Progesterone Receptorâ€“Positive Breast Cancer and the Value of Molecular Subtyping Using BluePrint. Journal of Global Oncology, 2017, 3, 314-322.	0.5	3

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91	Role of Iron Metabolism in Multiple Sclerosis. , 2003, , 399-414.		3
92	Detection of the Pvull RFLP in intron 15 of the LDL receptor gene by longâ€distance PCR. Clinical Genetics, 1996, 49, 277-278.	2.0	2
93	Clinicopathological features and associations in a series of South African acral melanomas. Pigment Cell and Melanoma Research, 2021, 34, 1120-1122.	3.3	2
94	The cost impact of unselective vs selective MammaPrint testing in early-stage breast cancer in Southern Africa. Breast, 2021, 59, 87-93.	2.2	2
95	Influence of Genetic Factors on the Development of Breast Cancer in the Older Woman. Current Aging Science, 2012, 5, 140-147.	1.2	2
96	Application of Molecular Genetics in Clinical Practice. South African Family Practice: Official Journal of the South African Academy of Family Practice/Primary Care, 2005, 47, 3-3.	0.6	1
97	A rare silent G to T mutation in exon 4 of the human low density lipoprotein receptor gene. Clinical Genetics, 1995, 47, 101-102.	2.0	1
98	Exome sequencing combined with semantic discovery identifies strong disease-associated candidates in a single case of relapsing remitting multiple sclerosis. BMC Proceedings, 2012, 6, .	1.6	1
99	Rooibos: Effect on Iron Status in South African Adults at Risk for Coronary Heart Disease. ACS Symposium Series, 2013, , 103-114.	0.5	1
100	Mo1051 TNF-Î± Rs1800629 (â^'308 G>a) Genotyping May Identify Obese Individuals At High Risk for Development of Non-Alcoholic Fatty Liver Disease (NAFLD). Gastroenterology, 2013, 144, S-1022.	1.3	1
101	Haplotype analysis excludes the functional protoporphyrinogen oxidase promoter polymorphism -1081G>A as a modifying factor in the clinical expression of variegate porphyria. Cellular and Molecular Biology, 2002, 48, 57-60.	0.9	1
102	Health implications and counselling for paternity testing. South African Family Practice: Official Journal of the South African Academy of Family Practice/Primary Care, 2006, 48, 34-34.	0.6	0
103	Rooibos: Effect on Iron Status of South African Adults at Risk for Coronary Heart Disease. Free Radical Biology and Medicine, 2012, 53, S85.	2.9	0
104	Tu1811 Correlation Between Mutations in the HFE Gene and Alanine Transaminase Levels May Increase the Risk of Cardiovascular Disease Events in Patients With NASH. Gastroenterology, 2012, 142, S-1023.	1.3	0
105	Mo1541 A Non-Invasive Pre-Screen Diagnostic Algorithm to Differentiate Between Genetic Hereditary Hemochromatosis and the Dysmetabolic Iron Overload Syndrome. Gastroenterology, 2016, 150, S1140.	1.3	0
106	Obesity and the metabolic syndrome: Impact of gene-diet interaction. South African Gastroenterology Review, 2010, 8, .	0.0	0
107	Abstract PO-056: Using MammaPrint to reduce the need for chemotherapy in early breast cancer during the COVID-19 pandemic. , 2020, , .		0