Christopher G Mathew

List of Publications by Year
in descending order

212 papers	45,588 citations	4831 87 h-index	2239 207 g-index
221 all docs	221 docs citations	221 times ranked	49574 citing authors

#	Article	IF	Citations
1	Processing and Analysis of Tissue Samples from Esophageal Cancer Patients in an African Setting. Biopreservation and Biobanking, 2022, 20, 185-194.	0.5	1
2	Genetic associations with carotid intima-media thickness link to atherosclerosis with sex-specific effects in sub-Saharan Africans. Nature Communications, 2022, 13, 855.	5.8	10
3	HPV types 16/18 L1 E6 and E7 proteins seropositivity and cervical cancer risk in HIV-positive and HIV-negative black South African women. Infectious Agents and Cancer, 2022, 17, 14.	1.2	3
4	Lifestyle factors associated with sex differences in Kaposi sarcoma incidence among adult black South Africans: A case-control study. Cancer Epidemiology, 2022, 78, 102158.	0.8	1
5	Esophageal Cancer Genomics in Africa: Recommendations for Future Research. Frontiers in Genetics, 2022, 13, 864575.	1.1	3
6	Epidemiology of Kaposiâ€™s sarcoma in sub-Saharan Africa. Cancer Epidemiology, 2022, 78, 102167.	0.8	14
7	Genetic Susceptibility to Breast Cancer in Sub-Saharan African Populations. JCO Global Oncology, 2021, 7, 1462-1471.	0.8	3
8	Ranking lifestyle risk factors for cervical cancer among Black women: A case-control study from Johannesburg, South Africa. PLoS ONE, 2021, 16, e0260319.	1.1	5
9	Genetic and Inflammatory Biomarkers Classify Small Intestine Inflammation in Asymptomatic First-degree Relatives of Patients With Crohnâ€™s Disease. Clinical Gastroenterology and Hepatology, 2020, 18, 908-916.e13.	2.4	18
10	Johannesburg Cancer Study (JCS): contribution to knowledge and opportunities arising from 20 years of data collection in an African setting. Cancer Epidemiology, 2020, 65, 101701.	0.8	11
11	The Relationship Between Environmental Exposure and Genetic Architecture of the 2q33 Locus With Esophageal Cancer in South Africa. Frontiers in Genetics, 2019, 10, 406.	1.1	3
12	Association of genetic variants in CHEK2 with oesophageal squamous cell carcinoma in the South African Black population. Carcinogenesis, 2019, 40, 513-520.	1.3	13
13	Novel and Known Gene-Smoking Interactions With cIMT Identified as Potential Drivers for Atherosclerosis Risk in West-African Populations of the AWI-Gen Study. Frontiers in Genetics, 2019, 10, 1354.	1.1	10
14	Exome Sequencing and Genotyping Identify a Rare Variant inNLRP7Gene Associated With Ulcerative Colitis. Journal of Crohn's and Colitis, 2018, 12, 321-326.	0.6	14
15	Exome array analysis of adverse reactions to fluoropyrimidine-based therapy for gastrointestinal cancer. PLoS ONE, 2018, 13, e0188911.	1.1	3
16	Genome-wide association study implicates immune activation of multiple integrin genes in inflammatory bowel disease. Nature Genetics, 2017, 49, 256-261.	9.4	943
17	Exploring the genetic architecture of inflammatory bowel disease by whole-genome sequencing identifies association at ADCY7. Nature Genetics, 2017, 49, 186-192.	9.4	153
18	Mutations in DONSON disrupt replication fork stability and cause microcephalic dwarfism. Nature Genetics, 2017, 49, 537-549.	9.4	81

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19	Fine-mapping inflammatory bowel disease loci to single-variant resolution. Nature, 2017, 547, 173-178.	13.7	473
20	Copy number variation of scavenger-receptor cysteine-rich domains within DMBT1 and Crohnâ€™s disease. European Journal of Human Genetics, 2016, 24, 1294-1300.	1.4	10
21	Genetic Association Analysis Reveals Differences in the Contribution of NOD2 Variants to the Clinical Phenotypes of Orofacial Granulomatosis. Inflammatory Bowel Diseases, 2016, 22, 1552-1558.	0.9	13
22	Genome-wide rare copy number variation screening in ulcerative colitis identifies potential susceptibility loci. BMC Medical Genetics, 2016, 17, 26.	2.1	14
23	Polymorphism in a lincRNA Associates with a Doubled Risk of Pneumococcal Bacteremia in Kenyan Children. American Journal of Human Genetics, 2016, 98, 1092-1100.	2.6	39
24	Pooled Sequencing of 531 Genes in Inflammatory Bowel Disease Identifies an Associated Rare Variant in BTNL2 and Implicates Other Immune Related Genes. PLoS Genetics, 2015, 11, e1004955.	1.5	59
25	Lamina propria macrophage phenotypes in relation to Escherichia coli in Crohnâ€™s disease. BMC Gastroenterology, 2015, 15, 75.	0.8	11
26	Meta-analysis of genome-wide association studies identifies novel loci that influence cupping and the glaucomatous process. Nature Communications, 2014, 5, 4883.	5.8	89
27	The correlation between reading and mathematics ability at age twelve has a substantial genetic component. Nature Communications, 2014, 5, 4204.	5.8	72
28	A Genome-wide Association Analysis of a Broad Psychosis Phenotype Identifies Three Loci for Further Investigation. Biological Psychiatry, 2014, 75, 386-397.	0.7	44
29	Genome-wide association analysis identifies 13 new risk loci for schizophrenia. Nature Genetics, 2013, 45, 1150-1159.	9.4	1,395
30	Association Between Variants of PRDM1 and NDP52 and Crohn's Disease, Based on Exome Sequencing and Functional Studies. Gastroenterology, 2013, 145, 339-347.	0.6	149
31	Common variants in the HLA-DRB1â€“HLA-DQA1 HLA class II region are associated with susceptibility to visceral leishmaniasis. Nature Genetics, 2013, 45, 208-213.	9.4	86
32	Genome-wide association analysis in Primary sclerosing cholangitis and ulcerative colitis identifies risk loci atGPR35andTCF4. Hepatology, 2013, 58, 1074-1083.	3.6	150
33	Negligible impact of rare autoimmune-locus coding-region variants on missing heritability. Nature, 2013, 498, 232-235.	13.7	184
34	Deep Resequencing of GWAS Loci Identifies Rare Variants in CARD9, IL23R and RNF186 That Are Associated with Ulcerative Colitis. PLoS Genetics, 2013, 9, e1003723.	1.5	185
35	Genome-wide association study of intraocular pressure identifies the GLCCI1/ICA1 region as a glaucoma susceptibility locus. Human Molecular Genetics, 2013, 22, 4653-4660.	1.4	29
36	Distinct genetic association at the PLCE1 locus with oesophageal squamous cell carcinoma in the South African population. Carcinogenesis, 2012, 33, 2155-2161.	1.3	44

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37	Using genome-wide complex trait analysis to quantify 'missing heritability' in Parkinson's disease. Human Molecular Genetics, 2012, 21, 4996-5009.	1.4	176
38	Effect of communicating DNA based risk assessments for Crohn's disease on smoking cessation: randomised controlled trial. BMJ, The, 2012, 345, e4708-e4708.	3.0	27
39	Rare and functional SIAE variants are not associated with autoimmune disease risk in up to 66,924 individuals of European ancestry. Nature Genetics, 2012, 44, 3-5.	9.4	44
40	Bayesian refinement of association signals for 14 loci in 3 common diseases. Nature Genetics, 2012, 44, 1294-1301.	9.4	469
41	Common variants at the MHC locus and at chromosome 16q24.1 predispose to Barrett's esophagus. Nature Genetics, 2012, 44, 1131-1136.	9.4	162
42	Genome-wide association study identifies a variant in HDAC9 associated with large vessel ischemic stroke. Nature Genetics, 2012, 44, 328-333.	9.4	375
43	Hostâ€“microbe interactions have shaped the genetic architecture of inflammatory bowel disease. Nature, 2012, 491, 119-124.	13.7	4,038
44	Identification of 15 new psoriasis susceptibility loci highlights the role of innate immunity. Nature Genetics, 2012, 44, 1341-1348.	9.4	848
45	Smokers with active CrohnÊ¼s disease have a clinically relevant dysbiosis of the gastrointestinal microbiota*. Inflammatory Bowel Diseases, 2012, 18, 1092-1100.	0.9	174
46	Combined Analysis of Genome-wide Association Studies for Crohn Disease and Psoriasis Identifies Seven Shared Susceptibility Loci. American Journal of Human Genetics, 2012, 90, 636-647.	2.6	290
47	Genetic risk and a primary role for cell-mediated immune mechanisms in multiple sclerosis. Nature, 2011, 476, 214-219.	13.7	2,400
48	Association of a Deletion of GSTT2B with an Altered Risk of Oesophageal Squamous Cell Carcinoma in a South African Population: A Case-Control Study. PLoS ONE, 2011, 6, e29366.	1.1	35
49	Common variants near ATM are associated with glycemic response to metformin in type 2 diabetes. Nature Genetics, 2011, 43, 117-120.	9.4	390
50	Meta-analysis identifies 29 additional ulcerative colitis risk loci, increasing the number of confirmed associations to 47. Nature Genetics, 2011, 43, 246-252.	9.4	1,201
51	Interaction between ERAP1 and HLA-B27 in ankylosing spondylitis implicates peptide handling in the mechanism for HLA-B27 in disease susceptibility. Nature Genetics, 2011, 43, 761-767.	9.4	778
52	Trial Protocol: Communicating DNA-based risk assessments for Crohn's disease: a randomised controlled trial assessing impact upon stopping smoking. BMC Public Health, 2011, 11, 44.	1.2	4
53	Population-specific genetic associations with oesophageal squamous cell carcinoma in South Africa. Carcinogenesis, 2011, 32, 1855-1861.	1.3	47
54	Dissection of the genetics of Parkinson's disease identifies an additional association 5' of SNCA and multiple associated haplotypes at 17q21. Human Molecular Genetics, 2011, 20, 345-353.	1.4	202

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55	Genome-wide association study of CNVs in 16,000 cases of eight common diseases and 3,000 shared controls. Nature, 2010, 464, 713-720.	13.7	737
56	Genome-wide association study for ulcerative colitis identifies risk loci at 7q22 and 22q13 (IL17REL). Nature Genetics, 2010, 42, 292-294.	9.4	177
57	Germline mutations in breast and ovarian cancer pedigrees establish RAD51C as a human cancer susceptibility gene. Nature Genetics, 2010, 42, 410-414.	9.4	638
58	Mutation of the RAD51C gene in a Fanconi anemiaâ€“like disorder. Nature Genetics, 2010, 42, 406-409.	9.4	360
59	Meta-analysis and imputation refines the association of 15q25 with smoking quantity. Nature Genetics, 2010, 42, 436-440.	9.4	581
60	A genome-wide association study identifies new psoriasis susceptibility loci and an interaction between HLA-C and ERAP1. Nature Genetics, 2010, 42, 985-990.	9.4	918
61	Genome-wide meta-analysis increases to 71 the number of confirmed Crohn's disease susceptibility loci. Nature Genetics, 2010, 42, 1118-1125.	9.4	2,284
62	Mutational Characterization of the Bile Acid Receptor TGR5 in Primary Sclerosing Cholangitis. PLoS ONE, 2010, 5, e12403.	1.1	106
63	Independent and population-specific association of risk variants at the IRGM locus with Crohn's disease. Human Molecular Genetics, 2010, 19, 1828-1839.	1.4	93
64	Measurement methods and accuracy in copy number variation: failure to replicate associations of beta-defensin copy number with Crohn's disease. Human Molecular Genetics, 2010, 19, 4930-4938.	1.4	81
65	Genome-wide association study identifies eight loci associated with blood pressure. Nature Genetics, 2009, 41, 666-676.	9.4	1,104
66	Genome-wide association study of ulcerative colitis identifies three new susceptibility loci, including the HNF4A region. Nature Genetics, 2009, 41, 1330-1334.	9.4	483
67	Common variants at five new loci associated with early-onset inflammatory bowel disease. Nature Genetics, 2009, 41, 1335-1340.	9.4	459
68	Searching for Genotype-Phenotype Structure: Using Hierarchical Log-Linear Models in Crohn Disease. American Journal of Human Genetics, 2009, 84, 178-187.	2.6	7
69	Investigation of Crohn's Disease Risk Loci in Ulcerative Colitis Further Defines Their Molecular Relationship. Gastroenterology, 2009, 136, 523-529.e3.	0.6	198
70	FANCG promotes formation of a newly identified protein complex containing BRCA2, FANCD2 and XRCC3. Oncogene, 2008, 27, 3641-3652.	2.6	82
71	Genetic determinants of ulcerative colitis include the ECM1 locus and five loci implicated in Crohn's disease. Nature Genetics, 2008, 40, 710-712.	9.4	403
72	Genome-wide association defines more than 30 distinct susceptibility loci for Crohn's disease. Nature Genetics, 2008, 40, 955-962.	9.4	2,422

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73	Sequence variants in IL10, ARPC2 and multiple other loci contribute to ulcerative colitis susceptibility. Nature Genetics, 2008, 40, 1319-1323.	9.4	534
74	New links to the pathogenesis of Crohn disease provided by genome-wide association scans. Nature Reviews Genetics, 2008, 9, 9-14.	7.7	186
75	Novel isoforms of the CARD8 (TUCAN) gene evade a nonsense mutation. European Journal of Human Genetics, 2008, 16, 619-625.	1.4	42
76	Cancer incidence in relatives of British Fanconi Anaemia patients. BMC Cancer, 2008, 8, 257.	1.1	23
77	Diverse effects of the CARD15 and IBD5 loci on clinical phenotype in 630 patients with CrohnÊ¼s disease. European Journal of Gastroenterology and Hepatology, 2008, 20, 37-45.	0.8	30
78	Gender-stratified analysis of DLG5 R30Q in 4707 patients with Crohn disease and 4973 controls from 12 Caucasian cohorts. Journal of Medical Genetics, 2007, 45, 36-42.	1.5	47
79	Estimating risks of common complex diseases across genetic and environmental factors: the example of Crohn disease. Journal of Medical Genetics, 2007, 44, 689-694.	1.5	43
80	Psoriasis is associated with pleiotropic susceptibility loci identified in type II diabetes and Crohn disease. Journal of Medical Genetics, 2007, 45, 114-116.	1.5	139
81	Identification, evolution, and association study of a novel promoter and first exon of the human NOD2 (CARD15) gene. Genomics, 2007, 90, 493-501.	1.3	5
82	IL23R Variation Determines Susceptibility But Not Disease Phenotype in Inflammatory Bowel Disease. Gastroenterology, 2007, 132, 1657-1664.	0.6	170
83	A Nonsynonymous SNP in ATG16L1 Predisposes to Ileal Crohnâ€™s Disease and Is Independent of CARD15 and IBD5. Gastroenterology, 2007, 132, 1665-1671.	0.6	268
84	Combined Evidence From Three Large British Association Studies Rejects TUCAN/CARD8 as an IBD Susceptibility Gene. Gastroenterology, 2007, 132, 2078-2080.	0.6	27
85	Systematic Association Mapping Identifies NELL1 as a Novel IBD Disease Gene. PLoS ONE, 2007, 2, e691.	1.1	123
86	Identification of the Fanconi Anemia Complementation Group I Gene, FANCI. Analytical Cellular Pathology, 2007, 29, 211-218.	0.7	89
87	Biallelic mutations in PALB2 cause Fanconi anemia subtype FA-N and predispose to childhood cancer. Nature Genetics, 2007, 39, 162-164.	9.4	556
88	A genome-wide association scan of nonsynonymous SNPs identifies a susceptibility variant for Crohn disease in ATG16L1. Nature Genetics, 2007, 39, 207-211.	9.4	1,712
89	Sequence variants in the autophagy gene IRGM and multiple other replicating loci contribute to Crohn's disease susceptibility. Nature Genetics, 2007, 39, 830-832.	9.4	1,063
90	Localization of type 1 diabetes susceptibility to the MHC class I genes HLA-B and HLA-A. Nature, 2007, 450, 887-892.	13.7	493

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91	Functional characterization of two novel 5' untranslated exons reveals a complex regulation of NOD2 protein expression. BMC Genomics, 2007, 8, 472.	1.2	28
92	Investigation of association of the DLG5 gene with phenotypes of inflammatory bowel disease in the British population. International Journal of Colorectal Disease, 2007, 22, 419-424.	1.0	9
93	Genetic Variation in Myosin IXB Is Associated With Ulcerative Colitis. Gastroenterology, 2006, 131, 1768-1774.	0.6	95
94	Tetratricopeptide-motif-mediated interaction of FANCG with recombination proteins XRCC3 and BRCA2. DNA Repair, 2006, 5, 629-640.	1.3	45
95	Recruiting first-degree relatives for prevention research: a comparison of clinician and proband-led methods of contact in Crohn's disease. European Journal of Human Genetics, 2006, 14, 1263-1268.	1.4	4
96	Evaluation of AGR2 and AGR3 as candidate genes for inflammatory bowel disease. Genes and Immunity, 2006, 7, 11-18.	2.2	113
97	Sequence variation, linkage disequilibrium and association with Crohn's disease on chromosome 5q31. Genes and Immunity, 2006, 7, 359-365.	2.2	26
98	Fanconi anaemia genes and susceptibility to cancer. Oncogene, 2006, 25, 5875-5884.	2.6	183
99	Detection of muramyl dipeptide-sensing pathway defects in patients with CrohnÊ¼s disease. Inflammatory Bowel Diseases, 2006, 12, 598-605.	0.9	21
100	Associations of allelic variants of the multidrug resistance gene (ABCB1 or MDR1) and Inflammatory Bowel Disease and their effects on disease behavior: A case-control and meta-analysis study. Inflammatory Bowel Diseases, 2006, 12, 263-271.	0.9	87
101	Mutation, selection, and evolution of the Crohn disease susceptibility geneCARD15. Human Mutation, 2006, 27, 44-54.	1.1	33
102	Direct or indirect association in a complex disease: the role ofSLC22A4 andSLC22A5 functional variants in Crohn disease. Human Mutation, 2006, 27, 778-785.	1.1	47
103	A common founder mutation in FANCA underlies the world's highest prevalence of Fanconi anemia in Gypsy families from Spain. Blood, 2005, 105, 1946-1949.	0.6	89
104	A common Fanconi anemia mutation in black populations of sub-Saharan Africa. Blood, 2005, 105, 3542-3544.	0.6	53
105	A general autoimmunity gene (PTPN22) is not associated with inflammatory bowel disease in a British population. Tissue Antigens, 2005, 66, 318-320.	1.0	28
106	The DNA helicase BRIP1 is defective in Fanconi anemia complementation group J. Nature Genetics, 2005, 37, 934-935.	9.4	399
107	A human ortholog of archaeal DNA repair protein Hef is defective in Fanconi anemia complementation group M. Nature Genetics, 2005, 37, 958-963.	9.4	395
108	Association of DLG5 R30Q variant with inflammatory bowel disease. European Journal of Human Genetics, 2005, 13, 835-839.	1.4	70

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109	Should chromosome breakage studies be performed in patients with VACTERL association?. American Journal of Medical Genetics, Part A, 2005, 137A, 55-58.	0.7	69
110	Nijmegen breakage syndrome diagnosed as Fanconi anaemia. Pediatric Blood and Cancer, 2005, 44, 494-499.	0.8	29
111	Synergy between TLR9 and NOD2 innate immune responses is lost in genetic Crohn's disease. Gut, 2005, 54, 1553-1557.	6.1	111
112	No association of the NFKB1 promoter polymorphism with ulcerative colitis in a British case control cohort. Gut, 2005, 54, 1205-1206.	6.1	34
113	Muramyl dipeptide and toll-like receptor sensitivity in NOD2-associated Crohn's disease. Lancet, The, 2005, 365, 1794-1796.	6.3	305
114	Genetics of inflammatory bowel disease: progress and prospects. Human Molecular Genetics, 2004, 13, 161R-168.	1.4	106
115	Inflammatory bowel disease susceptibility loci defined by genome scan meta-analysis of 1952 affected relative pairs. Human Molecular Genetics, 2004, 13, 763-770.	1.4	219
116	Quantitative PCR analysis reveals a high incidence of large intragenic deletions in the FANCA gene in Spanish Fanconi anemia patients. Cytogenetic and Genome Research, 2004, 104, 341-345.	0.6	21
117	Direct interaction of FANCD2 with BRCA2 in DNA damage response pathways. Human Molecular Genetics, 2004, 13, 1241-1248.	1.4	190
118	Genetic variation in DLG5 is associated with inflammatory bowel disease. Nature Genetics, 2004, 36, 476-480.	9.4	443
119	Deletion and reduced expression of the Fanconi anemia FANCA gene in sporadic acute myeloid leukemia. Leukemia, 2004, 18, 420-425.	3.3	78
120	Heterogeneity in Fanconi anemia: evidence for 2 new genetic subtypes. Blood, 2004, 103, 2498-2503.	0.6	212
121	Stratification by CARD15 variant genotype in a genome-wide search for inflammatory bowel disease susceptibility loci. Human Genetics, 2003, 113, 514-521.	1.8	15
122	Genetic variation in the IGSF6 gene and lack of association with inflammatory bowel disease. International Journal of Immunogenetics, 2003, 30, 187-190.	1.2	13
123	A Crohn's disease-associated insertion polymorphism (3020insC) in the NOD2 gene is not associated with psoriasis vulgaris, palmo-plantar pustular psoriasis or guttate psoriasis. Experimental Dermatology, 2003, 12, 506-509.	1.4	26
124	SNP Subset Selection for Genetic Association Studies. Annals of Human Genetics, 2003, 67, 543-556.	0.3	30
125	Genetic Variation at the Chromosome 16 Chemokine Gene Cluster: Development of a Strategy for Association Studies in Complex Disease. Annals of Human Genetics, 2003, 67, 377-390.	0.3	6
126	Bi-allelic silencing of the Fanconi anaemia gene FANCF in acute myeloid leukaemia. British Journal of Haematology, 2003, 123, 469-471.	1.2	65

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127	Disruption of the Fanconi anemiaâ€“BRCA pathway in cisplatin-sensitive ovarian tumors. Nature Medicine, 2003, 9, 568-574.	15.2	508
128	Lack of association between the C3435T MDR1 gene polymorphism and inflammatory bowel disease in two independent Northern European populations. Gastroenterology, 2003, 125, 1919-1920.	0.6	64
129	Prediction of pathogenic mutations in patients with early-onset breast cancer by family history. Lancet, The, 2003, 361, 1101-1102.	6.3	200
130	Yeast two-hybrid screens imply involvement of fanconi anemia proteins in transcription regulation, cell signaling, oxidative metabolism, and cellular transport. Experimental Cell Research, 2003, 289, 211-221.	1.2	55
131	Genetic Evidence for Interaction of the 5q31 Cytokine Locus and the CARD15 Gene in Crohn Disease. American Journal of Human Genetics, 2003, 72, 1018-1022.	2.6	111
132	Development of rheumatoid arthritis is not associated with two polymorphisms in the Crohn's disease gene CARD15. British Journal of Rheumatology, 2003, 42, 304-307.	2.5	22
133	Direct interaction of the Fanconi anaemia protein FANCG with BRCA2/FANCD1. Human Molecular Genetics, 2003, 12, 2503-2510.	1.4	106
134	Acquired FANCA dysfunction and cytogenetic instability in adult acute myelogenous leukemia. Blood, 2003, 102, 7-16.	0.6	56
135	Evidence for a NOD2-independent susceptibility locus for inflammatory bowel disease on chromosome 16p. Proceedings of the National Academy of Sciences of the United States of America, 2002, 99, 321-326.	3.3	106
136	Sex stratification of an inflammatory bowel disease genome search shows male-specific linkage to the HLA region of chromosome 6. European Journal of Human Genetics, 2002, 10, 259-265.	1.4	49
137	The contribution of NOD2 gene mutations to the risk and site of disease in inflammatory bowel disease. Gastroenterology, 2002, 122, 867-874.	0.6	670
138	Interaction of FANCD2 and NBS1 in the DNA damage response. Nature Cell Biology, 2002, 4, 913-920.	4.6	261
139	Association between insertion mutation in NOD2 gene and Crohn's disease in German and British populations. Lancet, The, 2001, 357, 1925-1928.	6.3	1,071
140	Molecular and genealogical evidence for a founder effect in Fanconi anemia families of the Afrikaner population of South Africa. Proceedings of the National Academy of Sciences of the United States of America, 2001, 98, 5734-5739.	3.3	100
141	Direct interactions of the five known Fanconi anaemia proteins suggest a common functional pathway. Human Molecular Genetics, 2001, 10, 423-429.	1.4	147
142	Fine mapping of the chromosome 3p susceptibility locus in inflammatory bowel disease. Gut, 2001, 48, 191-197.	6.1	72
143	The Fanconi anaemia gene FANCF encodes a novel protein with homology to ROM. Nature Genetics, 2000, 24, 15-16.	9.4	252
144	Spectrum of mutations in the Fanconi anaemia group G gene, FANCG/XRCC9. European Journal of Human Genetics, 2000, 8, 861-868.	1.4	61

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145	Investigation of Fanconi Anemia Protein Interactions by Yeast Two-Hybrid Analysis. Biochemical and Biophysical Research Communications, 2000, 268, 73-77.	1.0	27
146	Isolation of a cDNA Representing the Fanconi Anemia Complementation Group E Gene. American Journal of Human Genetics, 2000, 67, 1306-1308.	2.6	201
147	Complementation Analysis in Fanconi Anemia: Assignment of the Reference FA-H Patient to Group A. American Journal of Human Genetics, 2000, 67, 759-762.	2.6	115
148	Comparative Mutation Detection Screening of the Type VII Collagen Gene (COL7A1) Using the Protein Truncation Test, Fluorescent Chemical Cleavage of Mismatch, and Conformation Sensitive Gel Electrophoresis. Journal of Investigative Dermatology, 1999, 113, 673-686.	0.3	76
149	Spontaneous functional correction of homozygous Fanconi anaemia alleles reveals novel mechanistic basis for reverse mosaicism. Nature Genetics, 1999, 22, 379-383.	9.4	190
150	Identification of a C/G polymorphism in the promoter region of the BRCA1 gene and its use as a marker for rapid detection of promoter deletions. British Journal of Cancer, 1999, 79, 759-763.	2.9	8
151	Mutation analysis of the Fanconi anaemia A gene in breast tumours with loss of heterozygosity at 16q24.3. British Journal of Cancer, 1999, 79, 1049-1052.	2.9	15
152	A patient-derived mutant form of the Fanconi anemia protein, FANCA, is defective in nuclear accumulation. Experimental Hematology, 1999, 27, 587-593.	0.2	35
153	Prenatal onset spinal muscular atrophy. European Journal of Paediatric Neurology, 1999, 3, 65-72.	0.7	91
154	Identification of germline missense mutations and rare allelic variants in theATM gene in early-onset breast cancer. , 1999, 26, 286-294.		69
155	A Genomewide Analysis Provides Evidence for Novel Linkages in Inflammatory Bowel Disease in a Large European Cohort. American Journal of Human Genetics, 1999, 64, 808-816.	2.6	349
156	The Fanconi Anemia Group E Gene, FANCE, Maps to Chromosome 6p. American Journal of Human Genetics, 1999, 64, 1400-1405.	2.6	48
157	High Frequency of Large Intragenic Deletions in the Fanconi Anemia Group A Gene. American Journal of Human Genetics, 1999, 65, 1330-1341.	2.6	121
158	Linkage of Inflammatory Bowel Disease to Human Chromosome 6p. American Journal of Human Genetics, 1999, 65, 1647-1655.	2.6	215
159	ThePISSLREGene: Structure, Exon Skipping, and Exclusion as Tumor Suppressor in Breast Cancer. Genomics, 1999, 56, 90-97.	1.3	39
160	DNA diagnostics: goals and Challenges. British Medical Bulletin, 1999, 55, 325-339.	2.7	9
161	Identification of germline missense mutations and rare allelic variants in the ATM gene in early-onset breast cancer. , 1999, 26, 286.		4
162	Correlation of SMNt and SMNc gene copy number with age of onset and survival in spinal muscular atrophy. European Journal of Human Genetics, 1998, 6, 467-474.	1.4	108

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163	Exon 6 skipping in the fanconi anemia C gene associated with a nonsense/missense mutation (775Câ†'T) in exon 5: The first example of a nonsense mutation in one exon causing skipping of another downstream. Human Mutation, 1998, 11, S25-S27.	1.1	6
164	Identification of missense and truncating mutations in theBRCA1 gene in sporadic and familial breast and ovarian cancer. Genes Chromosomes and Cancer, 1998, 21, 244-249.	1.5	29
165	The interferon- Ã•gene as a positional and functional candidate gene for inflammatory bowel disease. International Journal of Colorectal Disease, 1998, 13, 260-263.	1.0	36
166	Genetic analysis of inflammatory bowel disease in a large European cohort supports linkage to chromosomes 12 and 16. Gastroenterology, 1998, 115, 1066-1071.	0.6	169
167	Construction of a High-Resolution Physical and Transcription Map of Chromosome 16q24.3: A Region of Frequent Loss of Heterozygosity in Sporadic Breast Cancer. Genomics, 1998, 50, 1-8.	1.3	28
168	Characterization and Screening for Mutations of the Growth Arrest-Specific 11 (GAS11) andC16orf3Genes at 16q24.3 in Breast Cancer. Genomics, 1998, 52, 325-331.	1.3	47
169	Identification of missense and truncating mutations in the BRCA1 gene in sporadic and familial breast and ovarian cancer. Genes Chromosomes and Cancer, 1998, 21, 244-249.	1.5	2
170	A pedigree-based linkage study of coeliac disease: failure to replicate previous positive findings. Annals of Human Genetics, 1998, 62, 25-32.	0.3	15
171	Dystrophin Point Mutation Screening Using a Multiplexed Protein Truncation Test. Genetic Testing and Molecular Biomarkers, 1997, 1, 115-123.	1.7	29
172	The Genomic Organization of the Fanconi Anemia Group A (FAA) Gene. Genomics, 1997, 41, 309-314.	1.3	51
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