Mary-Claire King

List of Publications by Year
in descending order

237 papers	32,485 citations	5891 81 h-index	4223 174 g-index
241 all docs	241 docs citations	241 times ranked	36170 citing authors

#	Article	IF	Citations
1	Strong Association of De Novo Copy Number Mutations with Autism. Science, 2007, 316, 445-449.	6.0	2,497
2	Breast and Ovarian Cancer Risks Due to Inherited Mutations in BRCA1 and BRCA2. Science, 2003, 302, 643-646.	6.0	2,032
3	Rare Structural Variants Disrupt Multiple Genes in Neurodevelopmental Pathways in Schizophrenia. Science, 2008, 320, 539-543.	6.0	1,654
4	Genetic Heterogeneity in Human Disease. Cell, 2010, 141, 210-217.	13.5	905
5	Mutations in 12 genes for inherited ovarian, fallopian tube, and peritoneal carcinoma identified by massively parallel sequencing. Proceedings of the National Academy of Sciences of the United States of America, 2011, 108, 18032-18037.	3.3	814
6	Germline and Somatic Mutations in Homologous Recombination Genes Predict Platinum Response and Survival in Ovarian, Fallopian Tube, and Peritoneal Carcinomas. Clinical Cancer Research, 2014, 20, 764-775.	3.2	803
7	Breast-Cancer Risk in Families with Mutations in PALB2. New England Journal of Medicine, 2014, 371, 497-506.	13.9	745
8	Tamoxifen and Breast Cancer Incidence Among Women With Inherited Mutations in BRCA1 and BRCA2. JAMA - Journal of the American Medical Association, 2001, 286, 2251.	3.8	673
9	Recurrent Rearrangements of Chromosome 1q21.1 and Variable Pediatric Phenotypes. New England Journal of Medicine, 2008, 359, 1685-1699.	13.9	663
10	Microduplications of 16p11.2 are associated with schizophrenia. Nature Genetics, 2009, 41, 1223-1227.	9.4	646
11	Confirmation of BRCA1 by analysis of germline mutations linked to breast and ovarian cancer in ten families. Nature Genetics, 1994, 8, 399-404.	9.4	590
12	Mutant Adenosine Deaminase 2 in a Polyarteritis Nodosa Vasculopathy. New England Journal of Medicine, 2014, 370, 921-931.	13.9	566
13	Spectrum of Mutations in BRCA1, BRCA2, CHEK2, and TP53 in Families at High Risk of Breast Cancer. JAMA - Journal of the American Medical Association, 2006, 295, 1379.	3.8	565
14	A recurrent 16p12.1 microdeletion supports a two-hit model for severe developmental delay. Nature Genetics, 2010, 42, 203-209.	9.4	539
15	Host Genotype-Specific Therapies Can Optimize the Inflammatory Response to Mycobacterial Infections. Cell, 2012, 148, 434-446.	13.5	523
16	BRCA1 and BRCA2 and the genetics of breast and ovarian cancer. Human Molecular Genetics, 2001, 10, 705-713.	1.4	517
17	Spatial and Temporal Mapping of De Novo Mutations in Schizophrenia to a Fetal Prefrontal Cortical Network. Cell, 2013, 154, 518-529.	13.5	507
18	The lta4h Locus Modulates Susceptibility to Mycobacterial Infection in Zebrafish and Humans. Cell, 2010, 140, 717-730.	13.5	501

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19	Structure of a BRCA1-BARD1 heterodimeric RING-RING complex. Nature Structural Biology, 2001, 8, 833-837.	9.7	446
20	Detection of inherited mutations for breast and ovarian cancer using genomic capture and massively parallel sequencing. Proceedings of the National Academy of Sciences of the United States of America, 2010, 107, 12629-12633.	3.3	426
21	Concordance for Type 2 (non-insulin-dependent) diabetes mellitus in male twins. Diabetologia, 1987, 30, 763-8.	2.9	402
22	Ten Genes for Inherited Breast Cancer. Cancer Cell, 2007, 11, 103-105.	7.7	395
23	Enabling the genomic revolution in Africa. Science, 2014, 344, 1346-1348.	6.0	361
24	Growth retardation and tumour inhibition by BRCA1. Nature Genetics, 1996, 12, 298-302.	9.4	359
25	Mutation in Transcription Factor POU4F3 Associated with Inherited Progressive Hearing Loss in Humans. Science, 1998, 279, 1950-1954.	6.0	322
26	Insights into the functions of BRCA1 and BRCA2. Trends in Genetics, 2000, 16, 69-74.	2.9	307
27	Germline ETV6 mutations in familial thrombocytopenia and hematologic malignancy. Nature Genetics, 2015, 47, 180-185.	9.4	299
28	Duplications of the neuropeptide receptor gene VIPR2 confer significant risk for schizophrenia. Nature, 2011, 471, 499-503.	13.7	296
29	Schizophrenia: a common disease caused by multiple rare alleles. British Journal of Psychiatry, 2007, 190, 194-199.	1.7	290
30	Population-based screening for breast and ovarian cancer risk due to BRCA1 and BRCA2. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, 14205-14210.	3.3	286
31	Mutations in COL11A2 cause non-syndromic hearing loss (DFNA13). Nature Genetics, 1999, 23, 413-419.	9.4	285
32	A recurrent germline PAX5 mutation confers susceptibility to pre-B cell acute lymphoblastic leukemia. Nature Genetics, 2013, 45, 1226-1231.	9.4	270
33	Whole Exome Sequencing and Homozygosity Mapping Identify Mutation in the Cell Polarity Protein GPSM2 as the Cause of Nonsyndromic Hearing Loss DFNB82. American Journal of Human Genetics, 2010, 87, 90-94.	2.6	261
34	Inherited Mutations in PTEN That Are Associated with Breast Cancer, Cowden Disease, and Juvenile Polyposis. American Journal of Human Genetics, 1997, 61, 1254-1260.	2.6	252
35	Population-Based Screening forBRCA1andBRCA2. JAMA - Journal of the American Medical Association, 2014, 312, 1091.	3.8	236
36	Validation and Implementation of Targeted Capture and Sequencing for the Detection of Actionable Mutation, Copy Number Variation, and Gene Rearrangement in Clinical Cancer Specimens. Journal of Molecular Diagnostics, 2014, 16, 56-67.	1.2	234

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37	Mutations in the DBP-Deficiency Protein HSD17B4 Cause Ovarian Dysgenesis, Hearing Loss, and Ataxia of Perrault Syndrome. American Journal of Human Genetics, 2010, 87, 282-288.	2.6	231
38	From flies' eyes to our ears: Mutations in a human class III myosin cause progressive nonsyndromic hearing loss DFNB30. Proceedings of the National Academy of Sciences of the United States of America, 2002, 99, 7518-7523.	3.3	230
39	Mutations in mitochondrial histidyl tRNA synthetase HARS2 cause ovarian dysgenesis and sensorineural hearing loss of Perrault syndrome. Proceedings of the National Academy of Sciences of the United States of America, 2011, 108, 6543-6548.	3.3	225
40	BRCA1 transcriptionally regulates genes involved in breast tumorigenesis. Proceedings of the National Academy of Sciences of the United States of America, 2002, 99, 7560-7565.	3.3	224
41	Identification of Id4 as a regulator of BRCA1 expression by using a ribozyme-library-based inverse genomics approach. Proceedings of the National Academy of Sciences of the United States of America, 2001, 98, 130-135.	3.3	222
42	RISK FACTORS FOR CORONARY HEART DISEASE IN ADULT FEMALE TWINS. American Journal of Epidemiology, 1987, 125, 308-318.	1.6	215
43	Contribution of Inherited Mutations in the BRCA2-Interacting Protein PALB2 to Familial Breast Cancer. Cancer Research, 2011, 71, 2222-2229.	0.4	215
44	Identifying individuals by sequencing mitochondrial DNA from teeth. Nature Genetics, 1992, 2, 135-138.	9.4	208
45	DBC2, a candidate for a tumor suppressor gene involved in breast cancer. Proceedings of the National Academy of Sciences of the United States of America, 2002, 99, 13647-13652.	3.3	202
46	BRCA1 is secreted and exhibits properties of a granin. Nature Genetics, 1996, 12, 303-308.	9.4	198
47	Perrault Syndrome Is Caused by Recessive Mutations in CLPP, Encoding a Mitochondrial ATP-Dependent Chambered Protease. American Journal of Human Genetics, 2013, 92, 605-613.	2.6	186
48	Targeted genomic capture and massively parallel sequencing to identify genes for hereditary hearing loss in middle eastern families. Genome Biology, 2011, 12, R89.	13.9	183
49	ColoSeq Provides Comprehensive Lynch and Polyposis Syndrome Mutational Analysis Using Massively Parallel Sequencing. Journal of Molecular Diagnostics, 2012, 14, 357-366.	1.2	179
50	Novel germ line DDX41 mutations define families with a lower age of MDS/AML onset and lymphoid malignancies. Blood, 2016, 127, 1017-1023.	0.6	179
51	Accurate and exact CNV identification from targeted high-throughput sequence data. BMC Genomics, 2011, 12, 184.	1.2	177
52	Mutations in LARS2, Encoding Mitochondrial Leucyl-tRNA Synthetase, Lead to Premature Ovarian Failure and Hearing Loss in Perrault Syndrome. American Journal of Human Genetics, 2013, 92, 614-620.	2.6	176
53	The Future of Psychiatric Research: Genomes and Neural Circuits. Science, 2010, 327, 1580-1581.	6.0	164
54	Haplotype and Phenotype Analysis of Nine Recurrent BRCA2 Mutations in 111 Families: Results of an International Study. American Journal of Human Genetics, 1998, 62, 1381-1388.	2.6	150

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55	Mitochondrial serine protease HTRA2 p.G399S in a kindred with essential tremor and Parkinson disease. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, 18285-18290.	3.3	147
56	Genetic Information and the Workplace: Legislative Approaches and Policy Challenges. Science, 1997, 275, 1755-1757.	6.0	142
57	MALE INFLUENCES ON CERVICAL CANCER RISK. American Journal of Epidemiology, 1986, 123, 302-307.	1.6	136
58	Inherited mutations in cancer susceptibility genes are common among survivors of breast cancer who develop therapyâ€related leukemia. Cancer, 2016, 122, 304-311.	2.0	129
59	MASP1 Mutations in Patients with Facial, Umbilical, Coccygeal, and Auditory Findings of Carnevale, Malpuech, OSA, and Michels Syndromes. American Journal of Human Genetics, 2010, 87, 679-686.	2.6	128
60	Genetics of congenital deafness in the Palestinian population: multiple connexinÂ26 alleles with shared origins in the Middle East. Human Genetics, 2002, 110, 284-289.	1.8	127
61	Mapping the Functional Domains of BRCA1. Journal of Biological Chemistry, 1999, 274, 5659-5665.	1.6	124
62	Genetic features of myelodysplastic syndrome and aplastic anemia in pediatric and young adult patients. Haematologica, 2016, 101, 1343-1350.	1.7	124
63	FAMILIAL BREAST CANCER IN A POPULATION-BASED SERIES. American Journal of Epidemiology, 1986, 123, 15-21.	1.6	120
64	Mutation of the ATP-gated P2X ₂ receptor leads to progressive hearing loss and increased susceptibility to noise. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, 2228-2233.	3.3	119
65	BRCA1 RING Domain Cancer-predisposing Mutations. Journal of Biological Chemistry, 2001, 276, 41399-41406.	1.6	118
66	The Founder Mutation MSH2*1906Gât'C Is an Important Cause of Hereditary Nonpolyposis Colorectal Cancer in the Ashkenazi Jewish Population. American Journal of Human Genetics, 2002, 71, 1395-1412.	2.6	118
67	Inherited predisposition to breast cancer among African American women. Breast Cancer Research and Treatment, 2015, 149, 31-39.	1.1	116
68	The CEPH consortium linkage map of human chromosome 1. Genomics, 1991, 9, 686-700.	1.3	113
69	Mutations in a Novel Isoform of TRIOBP That Encodes a Filamentous-Actin Binding Protein Are Responsible for DFNB28 Recessive Nonsyndromic Hearing Loss. American Journal of Human Genetics, 2006, 78, 144-152.	2.6	113
70	Genomic Views of Human History. Science, 1999, 286, 451-453.	6.0	109
71	Functional and genomic approaches reveal an ancient CHEK2 allele associated with breast cancer in the Ashkenazi Jewish population. Human Molecular Genetics, 2005, 14, 555-563.	1.4	109
72	Reduced transcript expression of genes affected by inherited and de novo CNVs in autism. European Journal of Human Genetics, 2011, 19, 727-731.	1.4	109

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73	Genomic analysis of bone marrow failure and myelodysplastic syndromes reveals phenotypic and diagnostic complexity. Haematologica, 2015, 100, 42-48.	1.7	108
74	Inherited predisposition to malignant mesothelioma and overall survival following platinum chemotherapy. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 9008-9013.	3.3	108
75	Tumor-specific p53 sequences in blood and peritoneal fluid of women with epithelial ovarian cancer. American Journal of Obstetrics and Gynecology, 2005, 193, 662-667.	0.7	105
76	Functional Characterization of a Novel BRCA1-Null Ovarian Cancer Cell Line in Response to Ionizing Radiation. Molecular Cancer Research, 2007, 5, 35-45.	1.5	105
77	Targeted long-read sequencing identifies missing disease-causing variation. American Journal of Human Genetics, 2021, 108, 1436-1449.	2.6	105
78	BRCA1, TP53, and CHEK2 germline mutations in uterine serous carcinoma. Cancer, 2013, 119, 332-338.	2.0	99
79	Genomic Duplication and Overexpression of TJP2/ZO-2 Leads to Altered Expression of Apoptosis Genes in Progressive Nonsyndromic Hearing Loss DFNA51. American Journal of Human Genetics, 2010, 87, 101-109.	2.6	95
80	Comparative analysis of cancer genes in the human and chimpanzee genomes. BMC Genomics, 2006, 7, 15.	1.2	94
81	Genetics of schizophrenia in the South African Xhosa. Science, 2020, 367, 569-573.	6.0	93
82	High-Density Genetic Map of the BRCA1 Region of Chromosome 17q12-q21. Genomics, 1993, 17, 618-623.	1.3	87
83	Mutations in Twinkle primase-helicase cause Perrault syndrome with neurologic features. Neurology, 2014, 83, 2054-2061.	1.5	86
84	Prevalence of founderBRCA1 andBRCA2 mutations among breast and ovarian cancer patients in hungary. , 2000, 86, 737-740.		85
85	Five novel loci for inherited hearing loss mapped by SNP-based homozygosity profiles in Palestinian families. European Journal of Human Genetics, 2010, 18, 407-413.	1.4	83
86	Familial clustering of site-specific cancer risks associated with BRCA1 and BRCA2 mutations in the Ashkenazi Jewish population. Proceedings of the National Academy of Sciences of the United States of America, 2006, 103, 3770-3774.	3.3	81
87	Albumin Differences among Ranid Frogs: Taxonomic and Phylogenetic Implications. Systematic Zoology, 1973, 22, 1.	1.6	80
88	Improving performance of multigene panels for genomic analysis of cancer predisposition. Genetics in Medicine, 2016, 18, 974-981.	1.1	80
89	Inherited Breast Cancer in Nigerian Women. Journal of Clinical Oncology, 2018, 36, 2820-2825.	0.8	80
90	The Cancer-predisposing Mutation C61G Disrupts Homodimer Formation in the NH2-terminal BRCA1 RING Finger Domain. Journal of Biological Chemistry, 1998, 273, 7795-7799.	1.6	79

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91	Essential Role ofBRCA2in Ovarian Development and Function. New England Journal of Medicine, 2018, 379, 1042-1049.	13.9	79
92	Targeted capture and sequencing for detection of mutations causing early onset epileptic encephalopathy. Epilepsia, 2013, 54, 1262-1269.	2.6	76
93	â€œThe Raceâ€•to Clone BRCA1. Science, 2014, 343, 1462-1465.	6.0	75
94	Genetic Epidemiology. Annual Review of Public Health, 1984, 5, 1-52.	7.6	73
95	Inherited thrombocytopenia associated with mutation of UDP-galactose-4-epimerase (GALE). Human Molecular Genetics, 2019, 28, 133-142.	1.4	73
96	Inherited mutation of the luteinizing hormone/choriogonadotropin receptor (LHCGR) in empty follicle syndrome. Fertility and Sterility, 2011, 96, e125-e130.	0.5	70
97	Heritability of Longitudinal Changes in Coronary-Heart-Disease Risk Factors in Women Twins. American Journal of Human Genetics, 1997, 60, 1502-1512.	2.6	69
98	Response to DNA damage of CHEK2 missense mutations in familial breast cancer. Human Molecular Genetics, 2012, 21, 2738-2744.	1.4	69
99	Maternal Famine, De Novo Mutations, and Schizophrenia. JAMA - Journal of the American Medical Association, 2006, 296, 582.	3.8	68
100	The APC I1307K allele and breast cancer risk. Nature Genetics, 1998, 20, 13-14.	9.4	65
101	Tumor necrosis factor a microsatellite polymorphism is associated with rheumatoid arthritis severity through an interaction with the HLA-DRB1 shared epitope. Arthritis and Rheumatism, 1999, 42, 438-442.	6.7	65
102	Race: A genetic melting-pot. Nature, 2003, 424, 374-374.	13.7	60
103	A Genomewide Screen for Suppressors of Alu-Mediated Rearrangements Reveals a Role for PIF1. PLoS ONE, 2012, 7, e30748.	1.1	59
104	Genomic Analysis of Mental Illness. JAMA - Journal of the American Medical Association, 2010, 303, 2523.	3.8	58
105	HUMAN GENETICS: Mapping Human History. Science, 2002, 298, 2342-2343.	6.0	57
106	Precision Medicine Meets Public Health: Population Screening for BRCA1 and BRCA2. Journal of the National Cancer Institute, 2014, 107, dju420-dju420.	3.0	57
107	Genetic Predisposition to Breast Cancer Due to Mutations Other Than BRCA1 and BRCA2 Founder Alleles Among Ashkenazi Jewish Women. JAMA Oncology, 2017, 3, 1647.	3.4	57
108	A Truncating Mutation in SERPINB6 Is Associated with Autosomal-Recessive Nonsyndromic Sensorineural Hearing Loss. American Journal of Human Genetics, 2010, 86, 797-804.	2.6	56

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109	Evidence for a BRCA1 Founder Mutation in Families of West African Ancestry. American Journal of Human Genetics, 1999, 65, 575-578.	2.6	55
110	Cellular Composition of the Nipple Aspirate Specimen of Breast Fluid: I. The Benign Cells. American Journal of Clinical Pathology, 1975, 64, 728-738.	0.4	53
111	Complex germline rearrangement of BRCA1 associated with breast and ovarian cancer. Genes Chromosomes and Cancer, 2000, 29, 58-62.	1.5	52
112	Genomic analysis of a heterogeneous Mendelian phenotype: multiple novel alleles for inherited hearing loss in the Palestinian population. Human Genomics, 2006, 2, 203-11.	1.4	51
113	22 genes from chromosome 17q21: cloning, sequencing, and characterization of mutations in breast cancer families and tumors. Genomics, 1995, 25, 256-263.	1.3	50
114	Characterization ofEZH1,a Human Homolog ofDrosophila Enhancer of zestenearBRCA1. Genomics, 1996, 37, 161-171.	1.3	49
115	Genetic Influences on Changes in Body Mass Index: A Longitudinal Analysis of Women Twins. Obesity, 1997, 5, 326-331.	4.0	49
116	Genetic Epidemiology of Breast and Ovarian Cancers. Epidemiologic Reviews, 1997, 19, 69-79.	1.3	46
117	Loss of function germline mutations in RAD51D in women with ovarian carcinoma. Gynecologic Oncology, 2012, 127, 552-555.	0.6	44
118	Intensive Surveillance with Biannual Dynamic Contrast-Enhanced Magnetic Resonance Imaging Downstages Breast Cancer in BRCA1 Mutation Carriers. Clinical Cancer Research, 2019, 25, 1786-1794.	3.2	44
119	BARD1 is necessary for ubiquitylation of nucleosomal histone H2A and for transcriptional regulation of estrogen metabolism genes. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, 1316-1321.	3.3	43
120	Germline variants drive myelodysplastic syndrome in young adults. Leukemia, 2021, 35, 2439-2444.	3.3	43
121	A girl with West syndrome and autistic features harboring a de novo TBL1XR1 mutation. Journal of Human Genetics, 2014, 59, 581-583.	1.1	42
122	Identifying Inherited and Acquired Genetic Factors Involved in Poor Stem Cell Mobilization and Donor-Derived Malignancy. Biology of Blood and Marrow Transplantation, 2016, 22, 2100-2103.	2.0	42
123	ENVIRONMENTAL AND BEHAVIORAL DETERMINANTS OF FASTING PLASMA GLUCOSE IN WOMEN. American Journal of Epidemiology, 1987, 125, 979-988.	1.6	41
124	Breast cancer genes: how many, where and who are they?. Nature Genetics, 1992, 2, 89-90.	9.4	41
125	Single nucleotide polymorphisms (SNPs) in the estrogen receptor gene and breast cancer susceptibility. Journal of Steroid Biochemistry and Molecular Biology, 1999, 71, 21-27.	1.2	41
126	A mouse model for human hearing loss DFNB30 due to loss of function of myosin IIIA. Mammalian Genome, 2011, 22, 170-177.	1.0	41

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127	Nonsense mutation of the stereociliar membrane protein gene PTPRQ in human hearing loss DFNB84. Journal of Medical Genetics, 2010, 47, 643-645.	1.5	40
128	Formation of Chimeric Genes by Copy-Number Variation as a Mutational Mechanism in Schizophrenia. American Journal of Human Genetics, 2013, 93, 697-710.	2.6	40
129	Genomic analysis of inherited breast cancer among Palestinian women: Genetic heterogeneity and a founder mutation in TP53. International Journal of Cancer, 2017, 141, 750-756.	2.3	40
130	Hardy-Weinberg testing for HLA class II (DRB1, DQA1, DQB1, AND DPB1) loci in 26 human ethnic groups. Tissue Antigens, 1999, 54, 533-542.	1.0	36
131	Using Somatic Mutations from Tumors to Classify Variants in Mismatch Repair Genes. American Journal of Human Genetics, 2018, 103, 19-29.	2.6	36
132	Human genetics and human rights. American Journal of Forensic Medicine and Pathology, 1984, 5, 339-347.	0.4	34
133	FAM111B Mutation Is Associated With Inherited Exocrine Pancreatic Dysfunction. Pancreas, 2016, 45, 858-862.	0.5	34
134	Features of Patients With Hereditary Mixed Polyposis Syndrome Caused by Duplication of GREM1 and Implications for Screening and Surveillance. Gastroenterology, 2017, 152, 1876-1880.e1.	0.6	34
135	Characterization of splice-altering mutations in inherited predisposition to cancer. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 26798-26807.	3.3	34
136	Profound, prelingual nonsyndromic deafness maps to chromosome 10q21 and is caused by a novel missense mutation in the Usher syndrome type IF gene PCDH15. European Journal of Human Genetics, 2009, 17, 554-564.	1.4	33
137	Inherited breast cancer: an emerging picture. Clinical Genetics, 1998, 54, 447-458.	1.0	32
138	Systematic misclassification of missense variants in BRCA1 and BRCA2 â€œcoldspotsâ€• Genetics in Medicine, 2020, 22, 825-830.	1.1	32
139	Evidence of a founder BRCA1 mutation in Scotland. British Journal of Cancer, 2000, 82, 705-711.	2.9	31
140	Genomic analysis of inherited hearing loss in the Palestinian population. Proceedings of the National Academy of Sciences of the United States of America, 2020, 117, 20070-20076.	3.3	31
141	BRCA1 and BRCA2 Mutations in Ashkenazi Jewish Families with Breast and Ovarian Cancer. Genetic Testing and Molecular Biomarkers, 1997, 1, 41-46.	1.7	29
142	Homozygous Loss-of-function Mutations inSOHLH1in Patients With Nonsyndromic Hypergonadotropic Hypogonadism. Journal of Clinical Endocrinology and Metabolism, 2015, 100, E808-E814.	1.8	29
143	Mechanism for survival of homozygous nonsense mutations in the tumor suppressor gene BRCA1. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, 5241-5246.	3.3	29
144	International distribution and age estimation of the Portuguese BRCA2 c.156_157insAlu founder mutation. Breast Cancer Research and Treatment, 2011, 127, 671-679.	1.1	27

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145	MYO3A Causes Human Dominant Deafness and Interacts with Protocadherin 15-CD2 Isoform. Human Mutation, 2016, 37, 481-487.	1.1	27
146	Gene Discovery for Complex Traits: Lessons from Africa. Cell, 2017, 171, 261-264.	13.5	27
147	Whole Exome Sequencing Identifies Candidate Genes Associated with Hereditary Predisposition to Uveal Melanoma. Ophthalmology, 2020, 127, 668-678.	2.5	27
148	Genetic risk factors for perinatally acquired HIVâ€1 infection. Paediatric and Perinatal Epidemiology, 1992, 6, 215-224.	0.8	26
149	SeqHelp: A Program to Analyze Molecular Sequences Utilizing Common Computationalâ€‰Resources. Genome Research, 1998, 8, 306-312.	2.4	26
150	Collaborative genomics for human health and cooperation in the Mediterranean region. Nature Genetics, 2010, 42, 641-645.	9.4	26
151	A truncating mutation in GPSM2 is associated with recessive nonâ€syndromic hearing loss. Clinical Genetics, 2012, 81, 289-293.	1.0	26
152	Mitochondrial PITRM1 peptidase loss-of-function in childhood cerebellar atrophy. Journal of Medical Genetics, 2018, 55, 599-606.	1.5	26
153	Vesicular acetylcholine transporter defect underlies devastating congenital myasthenia syndrome. Neurology, 2017, 88, 1021-1028.	1.5	25
154	De novo mutation in RING1 with epigenetic effects on neurodevelopment. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, 1558-1563.	3.3	24
155	Telomere biology disorder prevalence and phenotypes in adults with familial hematologic and/or pulmonary presentations. Blood Advances, 2020, 4, 4873-4886.	2.5	23
156	The inheritance of immunoglobulin E: Genetic linkage analysis. American Journal of Medical Genetics Part A, 1983, 16, 575-581.	2.4	22
157	Molecular diagnosis of childhood immune dysregulation, polyendocrinopathy, and enteropathy, and implications for clinical management. Journal of Allergy and Clinical Immunology, 2022, 149, 327-339.	1.5	22
158	Genetics of hearing loss in the Arab population of Northern Israel. European Journal of Human Genetics, 2018, 26, 1840-1847.	1.4	21
159	Reply to â€œâ€¦and secreted tumour suppressorsâ€• Nature Genetics, 1996, 13, 269-272.	9.4	20
160	Mutation of KREMEN1, a modulator of Wnt signaling, is responsible for ectodermal dysplasia including oligodontia in Palestinian families. European Journal of Human Genetics, 2016, 24, 1430-1435.	1.4	20
161	Diagnosis of Twin Zygosity by Self-Assessment and by Genetic Analysis. Acta Geneticae Medicae Et Gemellologiae, 1980, 29, 121-126.	0.2	19
162	Rare HRAS alleles and susceptibility to human breast cancer. Genomics, 1990, 6, 188-191.	1.3	19

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163	Africanâ€American HLA class II allele and haplotype diversity. Tissue Antigens, 1996, 48, 636-644.	1.0	19
164	Long Terminal Repeat andnefGene Variants of Human Immunodeficiency Virus Type 1 in Perinatally Infected Long-Term Survivors and Rapid Progressors. AIDS Research and Human Retroviruses, 1997, 13, 1611-1623.	0.5	19
165	CRISPRâ€“Cas9/long-read sequencing approach to identify cryptic mutations in BRCA1 and other tumour suppressor genes. Journal of Medical Genetics, 2021, 58, 850-852.	1.5	19
166	An Application of DNA Sequencing to a Human Rights Problem. , 1991, 1, 117-131.		19
167	Sexual orientation and the X. Nature, 1993, 364, 288-289.	13.7	18
168	Genomic structure and evolutionary context of the human feline leukemia virus subgroup C receptor (hFLVCR) gene: evidence for block duplications and de novo gene formation within duplicons of the hFLVCR locus. Gene, 2002, 286, 203-213.	1.0	18
169	Genetic analysis of human breast cancer: Literature review and description of family data in workshop. Genetic Epidemiology, 1986, 3, 1-13.	0.6	16
170	Hiding in Plain Sight â€" Somatic Mutation in Human Disease. New England Journal of Medicine, 2020, 383, 2680-2682.	13.9	16
171	A YAC-, P1-, and cosmid-based physical Map of the BRCA1 region on chromosome 17q21. Genomics, 1995, 25, 264-273.	1.3	15
172	Leaving Kansas â€¦ finding genes in 1997. Nature Genetics, 1997, 15, 8-10.	9.4	15
173	Spectrum of genes for inherited hearing loss in the Israeli Jewish population, including the novel human deafness gene <scp>ATOH1</scp>. Clinical Genetics, 2020, 98, 353-364.	1.0	15
174	Cisplatin +/â^' rucaparib after preoperative chemotherapy in patients with triple-negative or BRCA mutated breast cancer. Npj Breast Cancer, 2021, 7, 29.	2.3	15
175	A Novel BRCA2-Binding Protein and Breast and Ovarian Tumorigenesis. New England Journal of Medicine, 2004, 350, 1252-1253.	13.9	14
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