

Mary-Claire King

List of Publications by Year in descending order

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237
papers

32,485
citations

5876

81
h-index

4203

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. <i>Science</i> , 2007, 316, 445-449.	6.0	2,497
2	Breast and Ovarian Cancer Risks Due to Inherited Mutations in BRCA1 and BRCA2. <i>Science</i> , 2003, 302, 643-646.	6.0	2,032
3	Rare Structural Variants Disrupt Multiple Genes in Neurodevelopmental Pathways in Schizophrenia. <i>Science</i> , 2008, 320, 539-543.	6.0	1,654
4	Genetic Heterogeneity in Human Disease. <i>Cell</i> , 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. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 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. <i>Clinical Cancer Research</i> , 2014, 20, 764-775.	3.2	803
7	Breast-Cancer Risk in Families with Mutations in <i>PALB2</i> . <i>New England Journal of Medicine</i> , 2014, 371, 497-506.	13.9	745
8	Tamoxifen and Breast Cancer Incidence Among Women With Inherited Mutations in BRCA1 and BRCA2. <i>JAMA - Journal of the American Medical Association</i> , 2001, 286, 2251.	3.8	673
9	Recurrent Rearrangements of Chromosome 1q21.1 and Variable Pediatric Phenotypes. <i>New England Journal of Medicine</i> , 2008, 359, 1685-1699.	13.9	663
10	Microduplications of 16p11.2 are associated with schizophrenia. <i>Nature Genetics</i> , 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. <i>Nature Genetics</i> , 1994, 8, 399-404.	9.4	590
12	Mutant Adenosine Deaminase 2 in a Polyarteritis Nodosa Vasculopathy. <i>New England Journal of Medicine</i> , 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. <i>JAMA - Journal of the American Medical Association</i> , 2006, 295, 1379.	3.8	565
14	A recurrent 16p12.1 microdeletion supports a two-hit model for severe developmental delay. <i>Nature Genetics</i> , 2010, 42, 203-209.	9.4	539
15	Host Genotype-Specific Therapies Can Optimize the Inflammatory Response to Mycobacterial Infections. <i>Cell</i> , 2012, 148, 434-446.	13.5	523
16	BRCA1 and BRCA2 and the genetics of breast and ovarian cancer. <i>Human Molecular Genetics</i> , 2001, 10, 705-713.	1.4	517
17	Spatial and Temporal Mapping of De Novo Mutations in Schizophrenia to a Fetal Prefrontal Cortical Network. <i>Cell</i> , 2013, 154, 518-529.	13.5	507
18	The <i>Ita4h</i> Locus Modulates Susceptibility to Mycobacterial Infection in Zebrafish and Humans. <i>Cell</i> , 2010, 140, 717-730.	13.5	501

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19	Structure of a BRCA1-BARD1 heterodimeric RING-RING complex. <i>Nature Structural Biology</i> , 2001, 8, 833-837.	9.7	446
20	Detection of inherited mutations for breast and ovarian cancer using genomic capture and massively parallel sequencing. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2010, 107, 12629-12633.	3.3	426
21	Concordance for Type 2 (non-insulin-dependent) diabetes mellitus in male twins. <i>Diabetologia</i> , 1987, 30, 763-8.	2.9	402
22	Ten Genes for Inherited Breast Cancer. <i>Cancer Cell</i> , 2007, 11, 103-105.	7.7	395
23	Enabling the genomic revolution in Africa. <i>Science</i> , 2014, 344, 1346-1348.	6.0	361
24	Growth retardation and tumour inhibition by BRCA1. <i>Nature Genetics</i> , 1996, 12, 298-302.	9.4	359
25	Mutation in Transcription Factor POU4F3 Associated with Inherited Progressive Hearing Loss in Humans. <i>Science</i> , 1998, 279, 1950-1954.	6.0	322
26	Insights into the functions of BRCA1 and BRCA2. <i>Trends in Genetics</i> , 2000, 16, 69-74.	2.9	307
27	Germline ETV6 mutations in familial thrombocytopenia and hematologic malignancy. <i>Nature Genetics</i> , 2015, 47, 180-185.	9.4	299
28	Duplications of the neuropeptide receptor gene VIPR2 confer significant risk for schizophrenia. <i>Nature</i> , 2011, 471, 499-503.	13.7	296
29	Schizophrenia: a common disease caused by multiple rare alleles. <i>British Journal of Psychiatry</i> , 2007, 190, 194-199.	1.7	290
30	Population-based screening for breast and ovarian cancer risk due to <i>BRCA1</i> and <i>BRCA2</i> . <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2014, 111, 14205-14210.	3.3	286
31	Mutations in COL11A2 cause non-syndromic hearing loss (DFNA13). <i>Nature Genetics</i> , 1999, 23, 413-419.	9.4	285
32	A recurrent germline PAX5 mutation confers susceptibility to pre-B cell acute lymphoblastic leukemia. <i>Nature Genetics</i> , 2013, 45, 1226-1231.	9.4	270
33	Whole Exome Sequencing and Homozygosity Mapping Identify Mutation in the Cell Polarity Protein GPM2 as the Cause of Nonsyndromic Hearing Loss DFNB82. <i>American Journal of Human Genetics</i> , 2010, 87, 90-94.	2.6	261
34	Inherited Mutations in PTEN That Are Associated with Breast Cancer, Cowden Disease, and Juvenile Polyposis. <i>American Journal of Human Genetics</i> , 1997, 61, 1254-1260.	2.6	252
35	Population-Based Screening for <i>BRCA1</i> and <i>BRCA2</i> . <i>JAMA - Journal of the American Medical Association</i> , 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. <i>Journal of Molecular Diagnostics</i> , 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. <i>American Journal of Human Genetics</i> , 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. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2002, 99, 7518-7523.	3.3	230
39	Mutations in mitochondrial histidyl tRNA synthetase <i>HARS2</i> cause ovarian dysgenesis and sensorineural hearing loss of Perrault syndrome. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2011, 108, 6543-6548.	3.3	225
40	BRCA1 transcriptionally regulates genes involved in breast tumorigenesis. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 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. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2001, 98, 130-135.	3.3	222
42	RISK FACTORS FOR CORONARY HEART DISEASE IN ADULT FEMALE TWINS. <i>American Journal of Epidemiology</i> , 1987, 125, 308-318.	1.6	215
43	Contribution of Inherited Mutations in the BRCA2-Interacting Protein PALB2 to Familial Breast Cancer. <i>Cancer Research</i> , 2011, 71, 2222-2229.	0.4	215
44	Identifying individuals by sequencing mitochondrial DNA from teeth. <i>Nature Genetics</i> , 1992, 2, 135-138.	9.4	208
45	DBC2, a candidate for a tumor suppressor gene involved in breast cancer. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2002, 99, 13647-13652.	3.3	202
46	BRCA1 is secreted and exhibits properties of a granin. <i>Nature Genetics</i> , 1996, 12, 303-308.	9.4	198
47	Perrault Syndrome Is Caused by Recessive Mutations in CLPP, Encoding a Mitochondrial ATP-Dependent Chambered Protease. <i>American Journal of Human Genetics</i> , 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. <i>Genome Biology</i> , 2011, 12, R89.	13.9	183
49	ColoSeq Provides Comprehensive Lynch and Polyposis Syndrome Mutational Analysis Using Massively Parallel Sequencing. <i>Journal of Molecular Diagnostics</i> , 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. <i>Blood</i> , 2016, 127, 1017-1023.	0.6	179
51	Accurate and exact CNV identification from targeted high-throughput sequence data. <i>BMC Genomics</i> , 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. <i>American Journal of Human Genetics</i> , 2013, 92, 614-620.	2.6	176
53	The Future of Psychiatric Research: Genomes and Neural Circuits. <i>Science</i> , 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. <i>American Journal of Human Genetics</i> , 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. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2014, 111, 18285-18290.	3.3	147
56	Genetic Information and the Workplace: Legislative Approaches and Policy Challenges. <i>Science</i> , 1997, 275, 1755-1757.	6.0	142
57	MALE INFLUENCES ON CERVICAL CANCER RISK. <i>American Journal of Epidemiology</i> , 1986, 123, 302-307.	1.6	136
58	Inherited mutations in cancer susceptibility genes are common among survivors of breast cancer who develop therapy-related leukemia. <i>Cancer</i> , 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. <i>American Journal of Human Genetics</i> , 2010, 87, 679-686.	2.6	128
60	Genetics of congenital deafness in the Palestinian population: multiple connexin26 alleles with shared origins in the Middle East. <i>Human Genetics</i> , 2002, 110, 284-289.	1.8	127
61	Mapping the Functional Domains of BRCA1. <i>Journal of Biological Chemistry</i> , 1999, 274, 5659-5665.	1.6	124
62	Genetic features of myelodysplastic syndrome and aplastic anemia in pediatric and young adult patients. <i>Haematologica</i> , 2016, 101, 1343-1350.	1.7	124
63	FAMILIAL BREAST CANCER IN A POPULATION-BASED SERIES. <i>American Journal of Epidemiology</i> , 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. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2013, 110, 2228-2233.	3.3	119
65	BRCA1 RING Domain Cancer-predisposing Mutations. <i>Journal of Biological Chemistry</i> , 2001, 276, 41399-41406.	1.6	118
66	The Founder Mutation MSH2*1906G>C Is an Important Cause of Hereditary Nonpolyposis Colorectal Cancer in the Ashkenazi Jewish Population. <i>American Journal of Human Genetics</i> , 2002, 71, 1395-1412.	2.6	118
67	Inherited predisposition to breast cancer among African American women. <i>Breast Cancer Research and Treatment</i> , 2015, 149, 31-39.	1.1	116
68	The CEPH consortium linkage map of human chromosome 1. <i>Genomics</i> , 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. <i>American Journal of Human Genetics</i> , 2006, 78, 144-152.	2.6	113
70	Genomic Views of Human History. <i>Science</i> , 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. <i>Human Molecular Genetics</i> , 2005, 14, 555-563.	1.4	109
72	Reduced transcript expression of genes affected by inherited and de novo CNVs in autism. <i>European Journal of Human Genetics</i> , 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. <i>Haematologica</i> , 2015, 100, 42-48.	1.7	108
74	Inherited predisposition to malignant mesothelioma and overall survival following platinum chemotherapy. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2019, 116, 9008-9013.	3.3	108
75	Tumor-specific p53 sequences in blood and peritoneal fluid of women with epithelial ovarian cancer. <i>American Journal of Obstetrics and Gynecology</i> , 2005, 193, 662-667.	0.7	105
76	Functional Characterization of a Novel BRCA1-Null Ovarian Cancer Cell Line in Response to Ionizing Radiation. <i>Molecular Cancer Research</i> , 2007, 5, 35-45.	1.5	105
77	Targeted long-read sequencing identifies missing disease-causing variation. <i>American Journal of Human Genetics</i> , 2021, 108, 1436-1449.	2.6	105
78	<i>BRCA1</i> , <i>TP53</i> , and <i>CHEK2</i> germline mutations in uterine serous carcinoma. <i>Cancer</i> , 2013, 119, 332-338.	2.0	99
79	Genomic Duplication and Overexpression of <i>TJP2/ZO-2</i> Leads to Altered Expression of Apoptosis Genes in Progressive Nonsyndromic Hearing Loss DFNA51. <i>American Journal of Human Genetics</i> , 2010, 87, 101-109.	2.6	95
80	Comparative analysis of cancer genes in the human and chimpanzee genomes. <i>BMC Genomics</i> , 2006, 7, 15.	1.2	94
81	Genetics of schizophrenia in the South African Xhosa. <i>Science</i> , 2020, 367, 569-573.	6.0	93
82	High-Density Genetic Map of the BRCA1 Region of Chromosome 17q12-q21. <i>Genomics</i> , 1993, 17, 618-623.	1.3	87
83	Mutations in Twinkle primase-helicase cause Perrault syndrome with neurologic features. <i>Neurology</i> , 2014, 83, 2054-2061.	1.5	86
84	Prevalence of founder BRCA1 and BRCA2 mutations among breast and ovarian cancer patients in Hungary. <i>Journal of Clinical Oncology</i> , 2000, 18, 737-740.		85
85	Five novel loci for inherited hearing loss mapped by SNP-based homozygosity profiles in Palestinian families. <i>European Journal of Human Genetics</i> , 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. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2006, 103, 3770-3774.	3.3	81
87	Albumin Differences among Ranid Frogs: Taxonomic and Phylogenetic Implications. <i>Systematic Zoology</i> , 1973, 22, 1.	1.6	80
88	Improving performance of multigene panels for genomic analysis of cancer predisposition. <i>Genetics in Medicine</i> , 2016, 18, 974-981.	1.1	80
89	Inherited Breast Cancer in Nigerian Women. <i>Journal of Clinical Oncology</i> , 2018, 36, 2820-2825.	0.8	80
90	The Cancer-predisposing Mutation C61G Disrupts Homodimer Formation in the NH2-terminal BRCA1 RING Finger Domain. <i>Journal of Biological Chemistry</i> , 1998, 273, 7795-7799.	1.6	79

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

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109	Evidence for a BRCA1 Founder Mutation in Families of West African Ancestry. <i>American Journal of Human Genetics</i> , 1999, 65, 575-578.	2.6	55
110	Cellular Composition of the Nipple Aspirate Specimen of Breast Fluid: I. The Benign Cells. <i>American Journal of Clinical Pathology</i> , 1975, 64, 728-738.	0.4	53
111	Complex germline rearrangement of BRCA1 associated with breast and ovarian cancer. <i>Genes Chromosomes and Cancer</i> , 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. <i>Human Genomics</i> , 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. <i>Genomics</i> , 1995, 25, 256-263.	1.3	50
114	Characterization of EZH1, a Human Homolog of Drosophila Enhancer of zeste near BRCA1. <i>Genomics</i> , 1996, 37, 161-171.	1.3	49
115	Genetic Influences on Changes in Body Mass Index: A Longitudinal Analysis of Women Twins. <i>Obesity</i> , 1997, 5, 326-331.	4.0	49
116	Genetic Epidemiology of Breast and Ovarian Cancers. <i>Epidemiologic Reviews</i> , 1997, 19, 69-79.	1.3	46
117	Loss of function germline mutations in RAD51D in women with ovarian carcinoma. <i>Gynecologic Oncology</i> , 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. <i>Clinical Cancer Research</i> , 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. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2018, 115, 1316-1321.	3.3	43
120	Germline variants drive myelodysplastic syndrome in young adults. <i>Leukemia</i> , 2021, 35, 2439-2444.	3.3	43
121	A girl with West syndrome and autistic features harboring a de novo TBL1XR1 mutation. <i>Journal of Human Genetics</i> , 2014, 59, 581-583.	1.1	42
122	Identifying Inherited and Acquired Genetic Factors Involved in Poor Stem Cell Mobilization and Donor-Derived Malignancy. <i>Biology of Blood and Marrow Transplantation</i> , 2016, 22, 2100-2103.	2.0	42
123	ENVIRONMENTAL AND BEHAVIORAL DETERMINANTS OF FASTING PLASMA GLUCOSE IN WOMEN. <i>American Journal of Epidemiology</i> , 1987, 125, 979-988.	1.6	41
124	Breast cancer genes: how many, where and who are they?. <i>Nature Genetics</i> , 1992, 2, 89-90.	9.4	41
125	Single nucleotide polymorphisms (SNPs) in the estrogen receptor gene and breast cancer susceptibility. <i>Journal of Steroid Biochemistry and Molecular Biology</i> , 1999, 71, 21-27.	1.2	41
126	A mouse model for human hearing loss DFNB30 due to loss of function of myosin IIIA. <i>Mammalian Genome</i> , 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. <i>Journal of Medical Genetics</i> , 2010, 47, 643-645.	1.5	40
128	Formation of Chimeric Genes by Copy-Number Variation as a Mutational Mechanism in Schizophrenia. <i>American Journal of Human Genetics</i> , 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. <i>International Journal of Cancer</i> , 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. <i>Tissue Antigens</i> , 1999, 54, 533-542.	1.0	36
131	Using Somatic Mutations from Tumors to Classify Variants in Mismatch Repair Genes. <i>American Journal of Human Genetics</i> , 2018, 103, 19-29.	2.6	36
132	Human genetics and human rights. <i>American Journal of Forensic Medicine and Pathology</i> , 1984, 5, 339-347.	0.4	34
133	FAM111B Mutation Is Associated With Inherited Exocrine Pancreatic Dysfunction. <i>Pancreas</i> , 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. <i>Gastroenterology</i> , 2017, 152, 1876-1880.e1.	0.6	34
135	Characterization of splice-altering mutations in inherited predisposition to cancer. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 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 1F gene PCDH15. <i>European Journal of Human Genetics</i> , 2009, 17, 554-564.	1.4	33
137	Inherited breast cancer: an emerging picture. <i>Clinical Genetics</i> , 1998, 54, 447-458.	1.0	32
138	Systematic misclassification of missense variants in BRCA1 and BRCA2. <i>Genetics in Medicine</i> , 2020, 22, 825-830.	1.1	32
139	Evidence of a founder BRCA1 mutation in Scotland. <i>British Journal of Cancer</i> , 2000, 82, 705-711.	2.9	31
140	Genomic analysis of inherited hearing loss in the Palestinian population. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2020, 117, 20070-20076.	3.3	31
141	BRCA1 and BRCA2 Mutations in Ashkenazi Jewish Families with Breast and Ovarian Cancer. <i>Genetic Testing and Molecular Biomarkers</i> , 1997, 1, 41-46.	1.7	29
142	Homozygous Loss-of-function Mutations in SOHLH1 in Patients With Nonsyndromic Hypergonadotropic Hypogonadism. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2015, 100, E808-E814.	1.8	29
143	Mechanism for survival of homozygous nonsense mutations in the tumor suppressor gene BRCA1. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2018, 115, 5241-5246.	3.3	29
144	International distribution and age estimation of the Portuguese BRCA2 c.156_157insAlu founder mutation. <i>Breast Cancer Research and Treatment</i> , 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 HIV-1 infection. Paediatric and Perinatal Epidemiology, 1992, 6, 215-224.	0.8	26
149	SeqHelp: A Program to Analyze Molecular Sequences Utilizing Common Computational 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 <i>GPSM2</i> is associated with recessive nonsyndromic hearing loss. Clinical Genetics, 2012, 81, 289-293.	1.0	26
152	Mitochondrial <i>PITRM1</i> 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
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