Mary-Claire King

List of Publications by Citations

228 papers	26,749 citations	77 h-index	162 g-index
241 ext. papers	30,046 ext. citations	14.2 avg, IF	6.49 L-index

#	Paper	IF	Citations
228	Strong association of de novo copy number mutations with autism. *Science*, **2007**, 316, 445-9	33.3	2126
227	Breast and ovarian cancer risks due to inherited mutations in BRCA1 and BRCA2. *Science*, **2003**, 302, 643-6	33.3	1746
226	Rare structural variants disrupt multiple genes in neurodevelopmental pathways in schizophrenia. *Science*, **2008**, 320, 539-43	33.3	1443
225	Genetic heterogeneity in human disease. *Cell*, **2010**, 141, 210-7	56.2	737
224	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, 18032-7	11.5	676
223	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, 764-75	12.9	590
222	Recurrent rearrangements of chromosome 1q21.1 and variable pediatric phenotypes. *New England Journal of Medicine*, **2008**, 359, 1685-99	59.2	587
221	Breast-cancer risk in families with mutations in PALB2. *New England Journal of Medicine*, **2014**, 371, 497-506	59.2	576
220	Tamoxifen and breast cancer incidence among women with inherited mutations in BRCA1 and BRCA2: National Surgical Adjuvant Breast and Bowel Project (NSABP-P1) Breast Cancer Prevention Trial. *JAMA - Journal of the American Medical Association*, **2001**, 286, 2251-6	27.4	564
219	Microduplications of 16p11.2 are associated with schizophrenia. *Nature Genetics*, **2009**, 41, 1223-7	36.3	550
218	Confirmation of BRCA1 by analysis of germline mutations linked to breast and ovarian cancer in ten families. *Nature Genetics*, **1994**, 8, 399-404	36.3	538
217	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, 1379-88	27.4	502
216	A recurrent 16p12.1 microdeletion supports a two-hit model for severe developmental delay. *Nature Genetics*, **2010**, 42, 203-9	36.3	461
215	BRCA1 and BRCA2 and the genetics of breast and ovarian cancer. *Human Molecular Genetics*, **2001**, 10, 705-13	5.6	427
214	Host genotype-specific therapies can optimize the inflammatory response to mycobacterial infections. *Cell*, **2012**, 148, 434-46	56.2	417
213	Mutant adenosine deaminase 2 in a polyarteritis nodosa vasculopathy. *New England Journal of Medicine*, **2014**, 370, 921-31	59.2	409
212	Spatial and temporal mapping of de novo mutations in schizophrenia to a fetal prefrontal cortical network. *Cell*, **2013**, 154, 518-29	56.2	406

211	The lta4h locus modulates susceptibility to mycobacterial infection in zebrafish and humans. *Cell*, **2010**, 140, 717-30	56.2	405
210	Structure of a BRCA1-BARD1 heterodimeric RING-RING complex. *Nature Structural Biology*, **2001**, 8, 833-7		381
209	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, 12629-33	11.5	372
208	Growth retardation and tumour inhibition by BRCA1. *Nature Genetics*, **1996**, 12, 298-302	36.3	338
207	Concordance for type 2 (non-insulin-dependent) diabetes mellitus in male twins. *Diabetologia*, **1987** , 30, 763-8	10.3	314
206	Mutation in transcription factor POU4F3 associated with inherited progressive hearing loss in humans. *Science*, **1998**, 279, 1950-4	33.3	269
205	Insights into the functions of BRCA1 and BRCA2. *Trends in Genetics*, **2000**, 16, 69-74	8.5	260
204	Duplications of the neuropeptide receptor gene VIPR2 confer significant risk for schizophrenia. *Nature*, **2011**, 471, 499-503	50.4	257
203	Schizophrenia: a common disease caused by multiple rare alleles. *British Journal of Psychiatry*, **2007**, 190, 194-9	5.4	257
202	Research capacity. Enabling the genomic revolution in Africa. *Science*, **2014**, 344, 1346-8	33.3	256
201	Germline ETV6 mutations in familial thrombocytopenia and hematologic malignancy. *Nature Genetics*, **2015**, 47, 180-5	36.3	239
200	Inherited mutations in PTEN that are associated with breast cancer, cowden disease, and juvenile polyposis. *American Journal of Human Genetics*, **1997**, 61, 1254-60	11	226
199	Mutations in COL11A2 cause non-syndromic hearing loss (DFNA13). *Nature Genetics*, **1999**, 23, 413-9	36.3	225
198	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, 90-4	11	223
197	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, 14205-10	11.5	216
196	Mutations in the DBP-deficiency protein HSD17B4 cause ovarian dysgenesis, hearing loss, and ataxia of Perrault Syndrome. *American Journal of Human Genetics*, **2010**, 87, 282-8	11	209
195	A recurrent germline PAX5 mutation confers susceptibility to pre-B cell acute lymphoblastic leukemia. *Nature Genetics*, **2013**, 45, 1226-1231	36.3	205
194	Risk factors for coronary heart disease in adult female twins. Genetic heritability and shared environmental influences. *American Journal of Epidemiology*, **1987**, 125, 308-18	3.8	204

193	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	5.1	203
192	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, 7518-23	11.5	202
191	BRCA1 transcriptionally regulates genes involved in breast tumorigenesis. *Proceedings of the National Academy of Sciences of the United States of America*, **2002**, 99, 7560-5	11.5	201
190	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, 6543-8	11.5	200
189	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, 130-5	11.5	197
188	Identifying individuals by sequencing mitochondrial DNA from teeth. *Nature Genetics*, **1992**, 2, 135-8	36.3	186
187	Population-based screening for BRCA1 and BRCA2: 2014 Lasker Award. *JAMA - Journal of the American Medical Association*, **2014**, 312, 1091-2	27.4	181
186	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, 13647-52	11.5	180
185	Contribution of inherited mutations in the BRCA2-interacting protein PALB2 to familial breast cancer. *Cancer Research*, **2011**, 71, 2222-9	10.1	178
184	BRCA1 is secreted and exhibits properties of a granin. *Nature Genetics*, **1996**, 12, 303-8	36.3	176
183	ColoSeq provides comprehensive lynch and polyposis syndrome mutational analysis using massively parallel sequencing. *Journal of Molecular Diagnostics*, **2012**, 14, 357-66	5.1	163
182	Targeted genomic capture and massively parallel sequencing to identify genes for hereditary hearing loss in Middle Eastern families. *Genome Biology*, **2011**, 12, R89	18.3	163
181	Accurate and exact CNV identification from targeted high-throughput sequence data. *BMC Genomics*, **2011**, 12, 184	4.5	156
180	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, 614-20	11	153
179	Perrault syndrome is caused by recessive mutations in CLPP, encoding a mitochondrial ATP-dependent chambered protease. *American Journal of Human Genetics*, **2013**, 92, 605-13	11	152
178	Haplotype and phenotype analysis of nine recurrent BRCA2 mutations in 111 families: results of an international study. *American Journal of Human Genetics*, **1998**, 62, 1381-8	11	138
177	Medicine. The future of psychiatric research: genomes and neural circuits. *Science*, **2010**, 327, 1580-1	33.3	137
176	Male influences on cervical cancer risk. *American Journal of Epidemiology*, **1986**, 123, 302-7	3.8	121

175	Genetic information and the workplace: legislative approaches and policy changes. *Science*, **1997**, 275, 1755-7	33.3	118
174	Novel germ line DDX41 mutations define families with a lower age of MDS/AML onset and lymphoid malignancies. *Blood*, **2016**, 127, 1017-23	2.2	117
173	Familial breast cancer in a population-based series. *American Journal of Epidemiology*, **1986**, 123, 15-21	3.8	113
172	The founder mutation MSH2*1906G-->C is an important cause of hereditary nonpolyposis colorectal cancer in the Ashkenazi Jewish population. *American Journal of Human Genetics*, **2002**, 71, 1395-412	11	109
171	Mapping the functional domains of BRCA1. Interaction of the ring finger domains of BRCA1 and BARD1. *Journal of Biological Chemistry*, **1999**, 274, 5659-65	5.4	109
170	The CEPH consortium linkage map of human chromosome 1. *Genomics*, **1991**, 9, 686-700	4.3	108
169	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, 679-86	11	107
168	Genetics of congenital deafness in the Palestinian population: multiple connexin 26 alleles with shared origins in the Middle East. *Human Genetics*, **2002**, 110, 284-9	6.3	107
167	BRCA1 RING domain cancer-predisposing mutations. Structural consequences and effects on protein-protein interactions. *Journal of Biological Chemistry*, **2001**, 276, 41399-406	5.4	103
166	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, 2228-33	11.5	97
165	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, 18285-90	11.5	96
164	Inherited predisposition to breast cancer among African American women. *Breast Cancer Research and Treatment*, **2015**, 149, 31-9	4.4	95
163	Functional and genomic approaches reveal an ancient CHEK2 allele associated with breast cancer in the Ashkenazi Jewish population. *Human Molecular Genetics*, **2005**, 14, 555-63	5.6	94
162	Inherited mutations in cancer susceptibility genes are common among survivors of breast cancer who develop therapy-related leukemia. *Cancer*, **2016**, 122, 304-11	6.4	89
161	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, 144-52	11	88
160	Reduced transcript expression of genes affected by inherited and de novo CNVs in autism. *European Journal of Human Genetics*, **2011**, 19, 727-31	5.3	87
159	Tumor-specific p53 sequences in blood and peritoneal fluid of women with epithelial ovarian cancer. *American Journal of Obstetrics and Gynecology*, **2005**, 193, 662-7	6.4	85
158	Functional characterization of a novel BRCA1-null ovarian cancer cell line in response to ionizing radiation. *Molecular Cancer Research*, **2007**, 5, 35-45	6.6	84

157	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, 101-9	11	82
156	Genomic analysis of bone marrow failure and myelodysplastic syndromes reveals phenotypic and diagnostic complexity. *Haematologica*, **2015**, 100, 42-8	6.6	81
155	BRCA1, TP53, and CHEK2 germline mutations in uterine serous carcinoma. *Cancer*, **2013**, 119, 332-8	6.4	80
154	Genomic views of human history. *Science*, **1999**, 286, 451-3	33.3	80
153	High-density genetic map of the BRCA1 region of chromosome 17q12-q21. *Genomics*, **1993**, 17, 618-23	4.3	79
152	Genetic features of myelodysplastic syndrome and aplastic anemia in pediatric and young adult patients. *Haematologica*, **2016**, 101, 1343-1350	6.6	74
151	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, 3770-4	11.5	74
150	Prevalence of founder BRCA1 and BRCA2 mutations among breast and ovarian cancer patients in Hungary. *International Journal of Cancer*, **2000**, 86, 737-40	7.5	74
149	Mutations in Twinkle primase-helicase cause Perrault syndrome with neurologic features. *Neurology*, **2014**, 83, 2054-61	6.5	73
148	Five novel loci for inherited hearing loss mapped by SNP-based homozygosity profiles in Palestinian families. *European Journal of Human Genetics*, **2010**, 18, 407-13	5.3	68
147	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	11.5	67
146	Comparative analysis of cancer genes in the human and chimpanzee genomes. *BMC Genomics*, **2006** , 7, 15	4.5	67
145	The cancer-predisposing mutation C61G disrupts homodimer formation in the NH2-terminal BRCA1 RING finger domain. *Journal of Biological Chemistry*, **1998**, 273, 7795-9	5.4	67
144	Genetic epidemiology. *Annual Review of Public Health*, **1984**, 5, 1-52	20.6	65
143	Targeted capture and sequencing for detection of mutations causing early onset epileptic encephalopathy. *Epilepsia*, **2013**, 54, 1262-9	6.4	64
142	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, 438-42		61
141	Improving performance of multigene panels for genomic analysis of cancer predisposition. *Genetics in Medicine*, **2016**, 18, 974-81	8.1	60
140	Inherited mutation of the luteinizing hormone/choriogonadotropin receptor (LHCGR) in empty follicle syndrome. *Fertility and Sterility*, **2011**, 96, e125-30	4.8	60

139	The APCI1307K allele and breast cancer risk. *Nature Genetics*, **1998**, 20, 13-4	36.3	59
138	Essential Role of BRCA2 in Ovarian Development and Function. *New England Journal of Medicine*, **2018**, 379, 1042-1049	59.2	57
137	Maternal famine, de novo mutations, and schizophrenia. *JAMA - Journal of the American Medical Association*, **2006**, 296, 582-4	27.4	56
136	Response to DNA damage of CHEK2 missense mutations in familial breast cancer. *Human Molecular Genetics*, **2012**, 21, 2738-44	5.6	52
135	Albumin Differences among Ranid Frogs: Taxonomic and Phylogenetic Implications. *Systematic Zoology*, **1973**, 22, 1		52
134	Inherited Breast Cancer in Nigerian Women. *Journal of Clinical Oncology*, **2018**, 36, 2820-2825	2.2	51
133	"The race" to clone BRCA1. *Science*, **2014**, 343, 1462-5	33.3	48
132	Genomic analysis of mental illness: a changing landscape. *JAMA - Journal of the American Medical Association*, **2010**, 303, 2523-4	27.4	48
131	A truncating mutation in SERPINB6 is associated with autosomal-recessive nonsyndromic sensorineural hearing loss. *American Journal of Human Genetics*, **2010**, 86, 797-804	11	48
130	Cellular composition of the nipple aspirate specimen of breast fluid. I. The benign cells. *American Journal of Clinical Pathology*, **1975**, 64, 728-38	1.9	48
129	A genomewide screen for suppressors of Alu-mediated rearrangements reveals a role for PIF1. *PLoS ONE*, **2012**, 7, e30748	3.7	47
128	Complex germline rearrangement of BRCA1 associated with breast and ovarian cancer. *Genes Chromosomes and Cancer*, **2000**, 29, 58-62	5	47
127	Race: a genetic melting-pot. *Nature*, **2003**, 424, 374	50.4	46
126	Evidence for a BRCA1 founder mutation in families of West African ancestry. *American Journal of Human Genetics*, **1999**, 65, 575-8	11	45
125	22 genes from chromosome 17q21: cloning, sequencing, and characterization of mutations in breast cancer families and tumors. *Genomics*, **1995**, 25, 256-63	4.3	45
124	Genetics of schizophrenia in the South African Xhosa. *Science*, **2020**, 367, 569-573	33.3	44
123	Human genetics. Mapping human history. *Science*, **2002**, 298, 2342-3	33.3	43
122	Characterization of EZH1, a human homolog of Drosophila Enhancer of zeste near BRCA1. *Genomics* , **1996**, 37, 161-71	4.3	43

121	Genetic Predisposition to Breast Cancer Due to Mutations Other Than BRCA1 and BRCA2 Founder Alleles Among Ashkenazi Jewish Women. *JAMA Oncology*, **2017**, 3, 1647-1653	13.4	42
120	Genetic epidemiology of breast and ovarian cancers. *Epidemiologic Reviews*, **1997**, 19, 69-79	4.1	41
119	Single nucleotide polymorphisms (SNPs) in the estrogen receptor gene and breast cancer susceptibility. *Journal of Steroid Biochemistry and Molecular Biology*, **1999**, 71, 21-7	5.1	41
118	Genomic analysis of a heterogeneous Mendelian phenotype: multiple novel alleles for inherited hearing loss in the Palestinian population. *Human Genomics*, **2006**, 2, 203-11	6.8	39
117	Loss of function germline mutations in RAD51D in women with ovarian carcinoma. *Gynecologic Oncology*, **2012**, 127, 552-5	4.9	38
116	Heritability of longitudinal changes in coronary-heart-disease risk factors in women twins. *American Journal of Human Genetics*, **1997**, 60, 1502-12	11	38
115	Environmental and behavioral determinants of fasting plasma glucose in women. A matched co-twin analysis. *American Journal of Epidemiology*, **1987**, 125, 979-88	3.8	36
114	Genetic influences on changes in body mass index: a longitudinal analysis of women twins. *Obesity*, **1997**, 5, 326-31		35
113	Formation of chimeric genes by copy-number variation as a mutational mechanism in schizophrenia. *American Journal of Human Genetics*, **2013**, 93, 697-710	11	34
112	A girl with West syndrome and autistic features harboring a de novo TBL1XR1 mutation. *Journal of Human Genetics*, **2014**, 59, 581-3	4.3	34
111	A mouse model for human hearing loss DFNB30 due to loss of function of myosin IIIA. *Mammalian Genome*, **2011**, 22, 170-7	3.2	34
110	Nonsense mutation of the stereociliar membrane protein gene PTPRQ in human hearing loss DFNB84. *Journal of Medical Genetics*, **2010**, 47, 643-5	5.8	32
109	Hardy-Weinberg testing for HLA class II (DRB1, DQA1, DQB1, and DPB1) loci in 26 human ethnic groups. *Tissue Antigens*, **1999**, 54, 533-42		32
108	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	4.7	31
107	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	11.5	27
106	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, 554-64	5.3	27
105	BRCA1 and BRCA2 mutations in Ashkenazi Jewish families with breast and ovarian cancer. *Genetic Testing and Molecular Biomarkers*, **1997**, 1, 41-6		27
104	Inherited breast cancer: an emerging picture. *Clinical Genetics*, **1998**, 54, 447-58	4	27

103	Human genetics and human rights. Identifying the families of kidnapped children. *American Journal of Forensic Medicine and Pathology*, **1984**, 5, 339-47	1	26
102	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	13.3	25
101	Homozygous loss-of-function mutations in SOHLH1 in patients with nonsyndromic hypergonadotropic hypogonadism. *Journal of Clinical Endocrinology and Metabolism*, **2015**, 100, E808-14	5.6	25
100	Evidence of a founder BRCA1 mutation in Scotland. *British Journal of Cancer*, **2000**, 82, 705-11	8.7	25
99	Genetic risk factors for perinatally acquired HIV-1 infection. *Paediatric and Perinatal Epidemiology*, **1992**, 6, 215-24	2.7	24
98	Inherited thrombocytopenia associated with mutation of UDP-galactose-4-epimerase (GALE). *Human Molecular Genetics*, **2019**, 28, 133-142	5.6	24
97	Intensive Surveillance with Biannual Dynamic Contrast-Enhanced Magnetic Resonance Imaging Downstages Breast Cancer in Mutation Carriers. *Clinical Cancer Research*, **2019**, 25, 1786-1794	12.9	23
96	SeqHelp: a program to analyze molecular sequences utilizing common computational resources. *Genome Research*, **1998**, 8, 306-12	9.7	22
95	The inheritance of immunoglobulin E: genetic linkage analysis. *American Journal of Medical Genetics Part A*, **1983**, 16, 575-81		22
94	Mitochondrial peptidase loss-of-function in childhood cerebellar atrophy. *Journal of Medical Genetics*, **2018**, 55, 599-606	5.8	22
93	Vesicular acetylcholine transporter defect underlies devastating congenital myasthenia syndrome. *Neurology*, **2017**, 88, 1021-1028	6.5	21
92	A truncating mutation in GPSM2 is associated with recessive non-syndromic hearing loss. *Clinical Genetics*, **2012**, 81, 289-93	4	21
91	International distribution and age estimation of the Portuguese BRCA2 c.156_157insAlu founder mutation. *Breast Cancer Research and Treatment*, **2011**, 127, 671-9	4.4	21
90	MYO3A Causes Human Dominant Deafness and Interacts with Protocadherin 15-CD2 Isoform. *Human Mutation*, **2016**, 37, 481-7	4.7	21
89	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	7.5	20
88	Mechanism for survival of homozygous nonsense mutations in the tumor suppressor gene. *Proceedings of the National Academy of Sciences of the United States of America*, **2018**, 115, 5241-5246	11.5	20
87	Collaborative genomics for human health and cooperation in the Mediterranean region. *Nature Genetics*, **2010**, 42, 641-5	36.3	20
86	Using Somatic Mutations from Tumors to Classify Variants in Mismatch Repair Genes. *American Journal of Human Genetics*, **2018**, 103, 19-29	11	19

85	Reply to Bnd secreted tumour suppressors. *Nature Genetics*, **1996**, 13, 269-272	36.3	19
84	Rare HRAS alleles and susceptibility to human breast cancer. *Genomics*, **1990**, 6, 188-91	4.3	19
83	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, 203-13	3.8	18
82	African-American HLA class II allele and haplotype diversity. *Tissue Antigens*, **1996**, 48, 636-44		18
81	Long terminal repeat and nef gene variants of human immunodeficiency virus type 1 in perinatally infected long-term survivors and rapid progressors. *AIDS Research and Human Retroviruses*, **1997**, 13, 1611-23	1.6	17
80	Diagnosis of twin zygosity by self-assessment and by genetic analysis. *Acta Geneticae Medicae Et Gemellologiae*, **1980**, 29, 121-6		17
79	Characterization of splice-altering mutations in inherited predisposition to cancer. *Proceedings of the National Academy of Sciences of the United States of America*, **2019**,	11.5	17
78	Gene Discovery for Complex Traits: Lessons from Africa. *Cell*, **2017**, 171, 261-264	56.2	16
77	Whole Exome Sequencing Identifies Candidate Genes Associated with Hereditary Predisposition to Uveal Melanoma. *Ophthalmology*, **2020**, 127, 668-678	7.3	16
76	Targeted long-read sequencing identifies missing disease-causing variation. *American Journal of Human Genetics*, **2021**, 108, 1436-1449	11	16
75	De novo mutation in with epigenetic effects on neurodevelopment. *Proceedings of the National Academy of Sciences of the United States of America*, **2018**, 115, 1558-1563	11.5	15
74	Genetics of hearing loss in the Arab population of Northern Israel. *European Journal of Human Genetics*, **2018**, 26, 1840-1847	5.3	15
73	A YAC-, P1-, and cosmid-based physical map of the BRCA1 region on chromosome 17q21. *Genomics*, **1995**, 25, 264-73	4.3	15
72	Genetic analysis of human breast cancer: Literature review and description of family data in workshop. *Genetic Epidemiology*, **1986**, 3, 1-13	2.6	15
71	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, 1430-5	5.3	15
70	Systematic misclassification of missense variants in BRCA1 and BRCA2 "coldspots". *Genetics in Medicine*, **2020**, 22, 825-830	8.1	14
69	A novel BRCA2-binding protein and breast and ovarian tumorigenesis. *New England Journal of Medicine*, **2004**, 350, 1252-3	59.2	14
68	An application of DNA sequencing to a human rights problem. *Molecular Genetic Medicine*, **1991**, 1, 117-31		14

67	FAM111B Mutation Is Associated With Inherited Exocrine Pancreatic Dysfunction. *Pancreas*, **2016**, 45, 858-62	2.6	14
66	Identification of a new BRCA2 large genomic deletion associated with high risk male breast cancer. *Hereditary Cancer in Clinical Practice*, **2015**, 13, 2	2.3	13
65	Diamond Blackfan Anemia: A Nonclassical Patient With Diagnosis Assisted by Genomic Analysis. *Journal of Pediatric Hematology/Oncology*, **2016**, 38, e260-2	1.2	13
64	Inheritance of the shared epitope and long-term outcomes of rheumatoid arthritis among community-based Caucasian females. *Genetic Epidemiology*, **1998**, 15, 61-72	2.6	13
63	Genetic analysis of human breast cancer: a synthesis of contributions to GAW IV. *Genetic Epidemiology*, **1986**, 1, 15-35	2.6	12
62	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	11.5	12
61	Clinical usefulness of genetic information for predicting radiographic damage in rheumatoid arthritis. *Journal of Rheumatology*, **2002**, 29, 2068-73	4.1	12
60	Deep sequencing with intronic capture enables identification of an APC exon 10 inversion in a patient with polyposis. *Genetics in Medicine*, **2014**, 16, 783-6	8.1	11
59	Germline mutations of inhibins in early-onset ovarian epithelial tumors. *Human Mutation*, **2014**, 35, 294-7	4.7	11
58	Garrod's fourth inborn error of metabolism solved by the identification of mutations causing pentosuria. *Proceedings of the National Academy of Sciences of the United States of America*, **2011**, 108, 18313-7	11.5	11
57	Genomic sequencing in the service of human rights. *International Journal of Epidemiology*, **2002**, 31, 53-8	7.8	11
56	Germline variants drive myelodysplastic syndrome in young adults. *Leukemia*, **2021**, 35, 2439-2444	10.7	11
55	Relative predispositional effects and mode of inheritance of HLA-DRB1 alleles among community-based Caucasian females with rheumatoid arthritis. *Genetic Epidemiology*, **1998**, 15, 123-34	2.6	10
54	Infantile onset spinocerebellar ataxia caused by compound heterozygosity for Twinkle mutations and modeling of Twinkle mutations causing recessive disease. *Journal of Physical Education and Sports Management*, **2016**, 2, a001107	2.8	9
53	Telomere biology disorder prevalence and phenotypes in adults with familial hematologic and/or pulmonary presentations. *Blood Advances*, **2020**, 4, 4873-4886	7.8	9
52	Alpha-thalassemia intellectual disability: variable phenotypic expression among males with a recurrent nonsense mutation□□t.109C>T (p.R37X). *Clinical Genetics*, **2015**, 87, 461-6	4	8
51	Abundant novel transcriptional units and unconventional gene pairs on human chromosome 22. *Genome Research*, **2006**, 16, 45-54	9.7	8
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