List of Publications by Year in descending order

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179 papers	37,713 citations	7069 78 h-index	5519 163 g-index
184 all docs	184 docs citations	184 times ranked	31941 citing authors

#	Article	IF	Citations
1	Tumour stem cells and drug resistance. Nature Reviews Cancer, 2005, 5, 275-284.	12.8	3,360
2	Identification of the von Hippel-Lindau disease tumor suppressor gene. Science, 1993, 260, 1317-1320.	6.0	2,723
3	Mutations of the Human Homolog of Drosophila patched in the Nevoid Basal Cell Carcinoma Syndrome. Cell, 1996, 85, 841-851.	13.5	2,150
4	From The Cover: A common haplotype in the complement regulatory gene factor H (HF1/CFH) predisposes individuals to age-related macular degeneration. Proceedings of the National Academy of Sciences of the United States of America, 2005, 102, 7227-7232.	3.3	1,867
5	Germline and somatic mutations in the tyrosine kinase domain of the MET proto-oncogene in papillary renal carcinomas. Nature Genetics, 1997, 16, 68-73.	9.4	1,461
6	A photoreceptor cell-specific ATP-binding transporter gene (ABCR) is mutated in recessive Starqardt macular dystrophy. Nature Genetics, 1997, 15, 236-246.	9.4	1,277
7	Variation in factor B (BF) and complement component 2 (C2) genes is associated with age-related macular degeneration. Nature Genetics, 2006, 38, 458-462.	9.4	1,001
8	The human ATP-binding cassette (ABC) transporter superfamily. Journal of Lipid Research, 2001, 42, 1007-1017.	2.0	965
9	The Human ATP-Binding Cassette (ABC) Transporter Superfamily. Genome Research, 2001, 11, 1156-1166.	2.4	932
10	Contrasting Genetic Influence of CCR2 and CCR5 Variants on HIV-1 Infection and Disease Progression. Science, 1997, 277, 959-965.	6.0	860
11	Mutation of the Stargardt Disease Gene (ABCR) in Age-Related Macular Degeneration. Science, 1997, 277, 1805-1807.	6.0	844
12	Cell-cycle control of c-myc but not c-ras expression is lost following chemical transformation. Cell, 1984, 36, 241-247.	13.5	769
13	The role of the human homologue of Drosophila patched in sporadic basal cell carcinomas. Nature Genetics, 1996, 14, 78-81.	9.4	713
14	Genetic Restriction of AIDS Pathogenesis by an SDF-1 Chemokine Gene Variant. Science, 1998, 279, 389-393.	6.0	674
15	Single-Cell Exome Sequencing Reveals Single-Nucleotide Mutation Characteristics of a Kidney Tumor. Cell, 2012, 148, 886-895.	13.5	622
16	ABCA3Gene Mutations in Newborns with Fatal Surfactant Deficiency. New England Journal of Medicine, 2004, 350, 1296-1303.	13.9	621
17	EVOLUTION OF THE ATP-BINDING CASSETTE (ABC) TRANSPORTER SUPERFAMILY IN VERTEBRATES. Annual Review of Genomics and Human Genetics, 2005, 6, 123-142.	2.5	540
18	Identification of a gene, ABCG5, important in the regulation of dietary cholesterol absorption. Nature Genetics, 2001, 27, 79-83.	9.4	539

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19	Germline mutations in the von Hippel-Lindau disease tumor suppressor gene: Correlations with phenotype. Human Mutation, 1995, 5, 66-75.	1.1	526
20	Mechanism of met oncogene activation. Cell, 1986, 45, 895-904.	13.5	523
21	Dating the Origin of the CCR5-Î”32 AIDS-Resistance Allele by the Coalescence of Haplotypes. American Journal of Human Genetics, 1998, 62, 1507-1515.	2.6	507
22	Novel mutations of the MET proto-oncogene in papillary renal carcinomas. Oncogene, 1999, 18, 2343-2350.	2.6	487
23	A closely linked genetic marker for cystic fibrosis. Nature, 1985, 318, 382-384.	13.7	470
24	ABCG2: A perspective. Advanced Drug Delivery Reviews, 2009, 61, 3-13.	6.6	409
25	Whole-genome and whole-exome sequencing of bladder cancer identifies frequent alterations in genes involved in sister chromatid cohesion and segregation. Nature Genetics, 2013, 45, 1459-1463.	9.4	400
26	Multiple mutations in highly conserved residues are found in mildly affected cystic fibrosis patients. Cell, 1990, 61, 863-870.	13.5	391
27	Retinitis pigmentosa caused by a homozygous mutation in the Stargardt disease gene ABCR. Nature Genetics, 1998, 18, 11-12.	9.4	382
28	ABC Transporters, Drug Resistance, and Cancer Stem Cells. Journal of Mammary Gland Biology and Neoplasia, 2009, 14, 3-9.	1.0	377
29	Detecting single base substitutions as heteroduplex polymorphisms. Genomics, 1992, 12, 301-306.	1.3	369
30	Two Genes That Map to the STSL Locus Cause Sitosterolemia: Genomic Structure and Spectrum of Mutations Involving Sterolin-1 and Sterolin-2, Encoded by ABCG5 and ABCG8, Respectively. American Journal of Human Genetics, 2001, 69, 278-290.	2.6	318
31	A novel germ line juxtamembrane Met mutation in human gastric cancer. Oncogene, 2000, 19, 4947-4953.	2.6	308
32	HIV-1 infection in a man homozygous for CCR5â—µ32. Lancet, The, 1997, 349, 1219.	6.3	305
33	The multidrug resistance transporter ABCG2 (breast cancer resistance protein 1) effluxes Hoechst 33342 and is overexpressed in hematopoietic stem cells. Clinical Cancer Research, 2002, 8, 22-8.	3.2	303
34	The human met oncogene is related to the tyrosine kinase oncogenes. Nature, 1985, 318, 385-388.	13.7	302
35	ABCA3Mutations Associated with Pediatric Interstitial Lung Disease. American Journal of Respiratory and Critical Care Medicine, 2005, 172, 1026-1031.	2.5	290
36	Genotype/Phenotype Analysis of a Photoreceptor-Specific ATP-Binding Cassette Transporter Gene, ABCR, in Stargardt Disease. American Journal of Human Genetics, 1999, 64, 422-434.	2.6	277

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37	Genome-wide association study provides evidence for a breast cancer risk locus at 6q22.33. Proceedings of the National Academy of Sciences of the United States of America, 2008, 105, 4340-4345.	3.3	274
38	Inherited GATA3 variants are associated with Ph-like childhood acute lymphoblastic leukemia and risk of relapse. Nature Genetics, 2013, 45, 1494-1498.	9.4	264
39	Complete characterization of the human ABC gene family. Journal of Bioenergetics and Biomembranes, 2001, 33, 475-479.	1.0	249
40	Optimization of the single-strand conformation polymorphism (SSCP) technique for detection of point mutations. Human Mutation, 1993, 2, 404-414.	1.1	246
41	Mapping of a Gene Causing Familial Mediterranean Fever to the Short Arm of Chromosome 16. New England Journal of Medicine, 1992, 326, 1509-1513.	13.9	229
42	Extended haplotypes in the complement factor H (CFH) and CFHâ€related (CFHR) family of genes protect against ageâ€related macular degeneration: Characterization, ethnic distribution and evolutionary implications. Annals of Medicine, 2006, 38, 592-604.	1.5	217
43	Novel Susceptibility Variants at 10p12.31-12.2 for Childhood Acute Lymphoblastic Leukemia in Ethnically Diverse Populations. Journal of the National Cancer Institute, 2013, 105, 733-742.	3.0	208
44	Sensitivity of single-strand conformation polymorphism and heteroduplex method for mutation detection in the cystic fibrosis gene. Human Molecular Genetics, 1994, 3, 801-807.	1.4	202
45	The B30.2(SPRY) Domain of the Retroviral Restriction Factor TRIM5Î± Exhibits Lineage-Specific Length and Sequence Variation in Primates. Journal of Virology, 2005, 79, 6111-6121.	1.5	181
46	A Mammalian patched Homolog Is Expressed in Target Tissues of sonic hedgehog and Maps to a Region Associated with Developmental Abnormalities. Journal of Biological Chemistry, 1996, 271, 12125-12128.	1.6	171
47	Structural and functional diversity calls for a new classification of ABC transporters. FEBS Letters, 2020, 594, 3767-3775.	1.3	169
48	ARID5B Genetic Polymorphisms Contribute to Racial Disparities in the Incidence and Treatment Outcome of Childhood Acute Lymphoblastic Leukemia. Journal of Clinical Oncology, 2012, 30, 751-757.	0.8	165
49	Novel Alleles of the Chemokine-Receptor Gene CCR5. American Journal of Human Genetics, 1997, 61, 1261-1267.	2.6	152
50	BALANCEDPOLYMORPHISMSELECTED BYGENETICVERSUSINFECTIOUSHUMANDISEASE. Annual Review of Genomics and Human Genetics, 2002, 3, 263-292.	2.5	150
51	Evolution of the vertebrate ABC gene family: Analysis of gene birth and death. Genomics, 2006, 88, 1-11.	1.3	150
52	Evaluation of the Best disease gene in patients with age-related macular degeneration and other maculopathies. Human Genetics, 1999, 104, 449-453.	1.8	145
53	Two new genes from the human ATP-binding cassette transporter superfamily, ABCC11 and ABCC12, tandemly duplicated on chromosome 16q12. Gene, 2001, 273, 89-96.	1.0	143
54	The Genetics of ATPâ€Binding Cassette Transporters. Methods in Enzymology, 2005, 400, 409-429.	0.4	142

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55	Comparison of variations detection between whole-genome amplification methods used in single-cell resequencing. GigaScience, 2015, 4, 37.	3.3	141
56	Influence of theCCR2-V64IPolymorphism on Human Immunodeficiency Virus Type 1 Coreceptor Activity and on Chemokine Receptor Function of CCR2b, CCR3, CCR5, and CXCR4. Journal of Virology, 1998, 72, 7450-7458.	1.5	138
57	Analysis of theABCA4Gene by Next-Generation Sequencing. , 2011, 52, 8479.		133
58	Evidence of association ofAPOEwith age-related macular degeneration - a pooled analysis of 15 studies. Human Mutation, 2011, 32, 1407-1416.	1.1	130
59	The Essential Vertebrate ABCE1 Protein Interacts with Eukaryotic Initiation Factors. Journal of Biological Chemistry, 2006, 281, 7452-7457.	1.6	129
60	A High-Throughput Cell-Based Assay for Inhibitors of ABCG2 Activity. Journal of Biomolecular Screening, 2006, 11, 176-183.	2.6	128
61	Effects of human TRIM5Î± polymorphisms on antiretroviral function and susceptibility to human immunodeficiency virus infection. Virology, 2006, 354, 15-27.	1.1	116
62	Surfactant Composition and Function in Patients with ABCA3 Mutations. Pediatric Research, 2006, 59, 801-805.	1.1	112
63	A genome-wide association study of susceptibility to acute lymphoblastic leukemia in adolescents and young adults. Blood, 2015, 125, 680-686.	0.6	110
64	Single-Nucleotide Polymorphism (SNP) Analysis in the ABC Half-Transporter ABCG2 (MXR/BCRP/ABCP1). Cancer Biology and Therapy, 2002, 1, 696-702.	1.5	109
65	A frame-shift mutation in the cystic fibrosis gene. Nature, 1990, 344, 665-667.	13.7	108
66	The rod photoreceptor ATP-binding cassette transporter gene, ABCR, and retinal disease: from monogenic to multifactorial. Vision Research, 1999, 39, 2537-2544.	0.7	108
67	Monocyte-derived neutrophil chemotactic factor (MDNCF/IL-8) resides in a gene cluster along with several other members of the platelet factor 4 gene superfamily. Human Genetics, 1990, 84, 185-7.	1.8	107
68	The ABC transporter gene family of Daphnia pulex. BMC Genomics, 2009, 10, 170.	1.2	107
69	Extended haplotypes in the complement factor H (CFH) and CFH-related (CFHR) family of genes protect against age-related macular degeneration: characterization, ethnic distribution and evolutionary implications. Annals of Medicine, 2006, 38, 592-604.	1.5	106
70	DRD2 Dopamine Receptor Genotype, Linkage Disequilibrium, and Alcoholism in American Indians and Other Populations. Alcoholism: Clinical and Experimental Research, 1993, 17, 199-204.	1.4	105
71	Concurrent Alterations in TERT, KDM6A, and the BRCA Pathway in Bladder Cancer. Clinical Cancer Research, 2014, 20, 4935-4948.	3.2	101
72	Single-cell sequencing analysis characterizes common and cell-lineage-specific mutations in a muscle-invasive bladder cancer. GigaScience, 2012, 1, 12.	3.3	99

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73	Estrogen Receptor Genotypes and Haplotypes Associated with Breast Cancer Risk. Cancer Research, 2004, 64, 8891-8900.	0.4	97
74	CCR2 chemokine receptor and AIDS progression. Nature Medicine, 1997, 3, 1052-1053.	15.2	96
75	Mutational Analysis of ABCG2:â€‰ Role of the GXXXG Motif. Biochemistry, 2004, 43, 9448-9456.	1.2	96
76	Mapping of the serotonin 5-HT1DÎ² autoreceptor gene on chromosome 6 and direct analysis for sequence variants. American Journal of Medical Genetics Part A, 1995, 60, 157-161.	2.4	91
77	Evolution of a cytoplasmic tripartite motif (TRIM) protein in cows that restricts retroviral infection. Proceedings of the National Academy of Sciences of the United States of America, 2006, 103, 7454-7459.	3.3	85
78	Evolution of ABC transporters by gene duplication and their role in human disease. Biological Chemistry, 2011, 392, 29-37.	1.2	84
79	Transcriptional activation of immunoglobulin Î± heavy-chain genes by translocation of the c-myc oncogene. Nature, 1983, 305, 443-446.	13.7	82
80	Multilocus analysis of age-related macular degeneration. European Journal of Human Genetics, 2009, 17, 1190-1199.	1.4	78
81	Heterogeneity in the severity of cystic fibrosis and the role of CFTR gene mutations. Human Genetics, 1994, 93, 364-8.	1.8	77
82	Amplification of 4q21-q22 and theMXR gene in independently derived mitoxantrone-resistant cell lines. , 2000, 27, 110-116.		73
83	Typing of HLA-DQA1 and DQB1 using DNA single-strand conformation polymorphism. Human Immunology, 1992, 33, 208-212.	1.2	72
84	XIIXVI. Yeast sequencing reports. Mapping and sequencing of two yeast genes belonging to the ATP-binding cassette superfamily. Yeast, 1994, 10, 377-383.	0.8	68
85	Genome Analysis of Latin American Cervical Cancer: Frequent Activation of the PIK3CA Pathway. Clinical Cancer Research, 2015, 21, 5360-5370.	3.2	68
86	Naturally Occurring CCR5 Extracellular and Transmembrane Domain Variants Affect HIV-1 Co-receptor and Ligand Binding Function. Journal of Biological Chemistry, 1999, 274, 16228-16234.	1.6	65
87	Unique features of TRIM5Î± among closely related human TRIM family members. Virology, 2007, 360, 419-433.	1.1	64
88	Mutations in the human ATP-binding cassette transportersABCG5 andABCG8 in sitosterolemia. Human Mutation, 2002, 20, 151-151.	1.1	61
89	Approaches to identify genes for complex human diseases: Lessons from Mendelian disorders. Human Mutation, 2003, 22, 261-274.	1.1	61
90	In Search of AIDS-Resistance Genes. Scientific American, 1997, 277, 44-51.	1.0	59

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91	New inhibitors of ABCG2 identified by high-throughput screening. Molecular Cancer Therapeutics, 2007, 6, 3271-3278.	1.9	57
92	DHPLC screening of cystic fibrosis gene mutations. Human Mutation, 2002, 19, 374-383.	1.1	53
93	Cloning and organization of the abc and mdl genes of Escherichia coli: relationship to eukaryotic multidrug resistance. Gene, 1993, 136, 231-236.	1.0	52
94	Mutational scanning of the ABCR gene with double-gradient denaturing-gradient gel electrophoresis (DG-DGGE) in Italian Stargardt disease patients. Human Genetics, 2001, 109, 326-338.	1.8	52
95	Comparative genome analysis of potential regulatory elements in the ABCG5â€“ABCG8 gene cluster. Biochemical and Biophysical Research Communications, 2002, 295, 276-282.	1.0	52
96	Physical mapping of the cystic fibrosis region by pulsed-field gel electrophoresis. Genomics, 1988, 2, 346-354.	1.3	51
97	The ABCR Gene in Recessive and Dominant Stargardt Diseases: A Genetic Pathway in Macular Degeneration. Genomics, 1999, 60, 234-237.	1.3	51
98	Applications of heteroduplex analysis for mutation detection in disease genes. Human Mutation, 1995, 6, 281-287.	1.1	48
99	Evolutionary analysis of a cluster of ATP-binding cassette (ABC) genes. Mammalian Genome, 2003, 14, 7-20.	1.0	44
100	Risk for HIV-1 Infection Associated With a Common CXCL12 (SDF1) Polymorphism and CXCR4 Variation in an African Population. Journal of Acquired Immune Deficiency Syndromes (1999), 2005, 40, 521-526.	0.9	44
101	The abcc6a Gene Expression Is Required for Normal Zebrafish Development. Journal of Investigative Dermatology, 2010, 130, 2561-2568.	0.3	43
102	Isolation and Chromosomal Mapping of a Novel ATP-Binding Cassette Transporter Conserved in Mouse and Human. Genomics, 1997, 41, 275-278.	1.3	42
103	Expression of 25 Human ABC Transporters in the Yeast Pichia pastoris and Characterization of the Purified ABCC3 ATPase Activity. Biochemistry, 2007, 46, 7992-8003.	1.2	42
104	A deletion of two nucleotides in exon 10 of the CFTR gene in a Soviet family with cystic fibrosis causing early infant death. Genomics, 1991, 10, 298-299.	1.3	41
105	Polymorphism and genetic mapping of the human oxytocin receptor gene on chromosome 3. American Journal of Medical Genetics Part A, 1995, 60, 183-187.	2.4	41
106	Phenotypic Expressions of CCR5-Î”32/Î”32 Homozygosity. Journal of Acquired Immune Deficiency Syndromes, 1999, 22, 75.	0.3	41
107	Detection of three rare frameshift mutations in the cystic fibrosis gene in an African-American (CF444delA), an Italian (CF2522insC), and a Soviet (CF3821delT)1. Genomics, 1991, 10, 266-269.	1.3	37
108	Variation and evolution of the ABC transporter genes ABCB1, ABCC1, ABCG2, ABCG5 and ABCG8: implication for pharmacogenetics and disease. Drug Metabolism and Drug Interactions, 2011, 26, 169-179.	0.3	37

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109	Conserved Intramolecular Disulfide Bond Is Critical to Trafficking and Fate of ATP-binding Cassette (ABC) Transporters ABCB6 and Sulfonylurea Receptor 1 (SUR1)/ABCC8. Journal of Biological Chemistry, 2011, 286, 8481-8492.	1.6	37
110	Degeneration of an ATP-binding cassette transporter gene, ABCC13, in different mammalian lineages. Genomics, 2004, 84, 34-46.	1.3	36
111	Mutational Studies of G553 in TM5 of ABCG2:Â A Residue Potentially Involved in Dimerizationâ€. Biochemistry, 2006, 45, 5251-5260.	1.2	36
112	Novel mutations in the gene encoding ATP binding cassette protein member A3 (ABCA3) resulting in fatal neonatal lung disease. Acta Paediatrica, International Journal of Paediatrics, 2007, 96, 185-190.	0.7	35
113	Purification and ATP Hydrolysis of the Putative Cholesterol Transporters ABCG5 and ABCG8â€. Biochemistry, 2006, 45, 9929-9939.	1.2	34
114	An ATP-binding cassette gene (ABCG3) closely related to the multidrug transporter ABCG2 (MXR/ABCP) has an unusual ATP-binding domain. Mammalian Genome, 2001, 12, 86-88.	1.0	32
115	Statistical estimation and pedigree analysis ofCCR2-CCR5 haplotypes. Human Genetics, 2001, 108, 484-493.	1.8	32
116	Three additional DNA polymorphisms in the met gene and D7S8 locus: Use in prenatal diagnosis of cystic fibrosis. Journal of Pediatrics, 1987, 111, 490-495.	0.9	31
117	A new mutation (1078delT) in exon 7 of the CFTR gene in a southern French adult with cystic fibrosis. Genomics, 1992, 13, 907-908.	1.3	28
118	Identification of 18 Mouse ABC Genes and Characterization of the ABC Superfamily in Mus musculus. Genomics, 2000, 64, 24-31.	1.3	28
119	A group of notI jumping and linking clones cover 2.5 Mb in the 3p21â€“p22 region suspected to contain a tumor suppressor gene. Cancer Genetics and Cytogenetics, 1995, 81, 144-150.	1.0	27
120	A de Novo cystic fibrosis mutation: CGA (Arg) to TGA (stop) at codon 851 of the CFTR gene. Genomics, 1991, 11, 778-779.	1.3	26
121	Polarity, proliferation and the hedgehog pathway. Nature Genetics, 1996, 14, 245-247.	9.4	26
122	Arginine 383 is a crucial residue in ABCG2 biogenesis. Biochimica Et Biophysica Acta - Biomembranes, 2009, 1788, 1434-1443.	1.4	26
123	The ERCC6 Gene and Age-Related Macular Degeneration. PLoS ONE, 2010, 5, e13786.	1.1	26
124	The gene causing familial mediterranean fever maps to the short arm of chromosome 16 in Druze and Moslem Arab families. Human Genetics, 1994, 94, 576-7.	1.8	25
125	The SERPING1 gene and age-related macular degeneration. Lancet, The, 2009, 374, 875-876.	6.3	25
126	Chromosomal localization of the met proto-oncogene in the mouse and cat genome. Genomics, 1987, 1, 167-173.	1.3	24

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127	Approaches to localizing disease genes as applied to cystic fibrosis. Nucleic Acids Research, 1990, 18, 345-350.	6.5	24
128	Molecular Evolutionary Analysis of ABCB5: The Ancestral Gene Is a Full Transporter with Potentially Deleterious Single Nucleotide Polymorphisms. PLoS ONE, 2011, 6, e16318.	1.1	24
129	Mapping of the serotonin 5-HT1DÎ± autoreceptor gene (HTR1D) on chromosome 1 using a silent polymorphism in the coding region. American Journal of Medical Genetics Part A, 1995, 60, 162-164.	2.4	23
130	Cystic fibrosis gene mutations detected in hereditary pancreatitis. Pflugers Archiv European Journal of Physiology, 1996, 431, R191-R192.	1.3	23
131	The use of DNA heteroduplex patterns to map recombination within the HLA class II region. Human Immunology, 1992, 33, 114-121.	1.2	22
132	Bringing age-related macular degeneration into focus. Nature Genetics, 2008, 40, 820-821.	9.4	22
133	The 6q22.33 Locus and Breast Cancer Susceptibility. Cancer Epidemiology Biomarkers and Prevention, 2009, 18, 2468-2475.	1.1	22
134	Single-stranded conformation polymorphism analysis of the CFTR gene in slovenian cystic fibrosis patients: Detection of mutations and sequence variations. Human Mutation, 1993, 2, 286-292.	1.1	21
135	Alternative splicing in the first nucleotide binding fold of CFTR. Human Molecular Genetics, 1993, 2, 231-235.	1.4	21
136	Resolving DNA mutations. Nature Genetics, 1995, 9, 103-104.	9.4	20
137	Identifying and characterizing a five-gene cluster of ATP-binding cassette transporters mapping to human chromosome 17q24: a new subgroup within the ABCA subfamily. GeneScreen, 2001, 1, 157-164.	0.7	20
138	Association Assessment of Copy Number Polymorphism and Risk of Age-Related Macular Degeneration. Ophthalmology, 2011, 118, 2442-2446.	2.5	20
139	CXCR4 Polymorphisms and HIV-1 Pathogenesis. Journal of Acquired Immune Deficiency Syndromes, 1998, 19, 430.	0.3	19
140	Haplotype structure and linkage disequilibrium in chemokine and chemokine receptor genes. Human Genomics, 2004, 1, 255.	1.4	18
141	Cystic fibrosis-related diabetes is associated with HLADQB1 alleles encoding Asp-57âˆ’ molecules. Journal of Clinical Immunology, 1994, 14, 353-358.	2.0	17
142	[9] Cloning of novel ABC transporter genes. Methods in Enzymology, 1998, 292, 116-130.	0.4	17
143	Three ATP-binding cassette transporter genes, Abca14 , Abca15 , and Abca16 , form a cluster on mouse Chromosome 7F3. Mammalian Genome, 2004, 15, 335-343.	1.0	17
144	Wholeâ€exome sequencing of nevoid basal cell carcinoma syndrome families and review of Human Gene Mutation Database PTCH1 mutation data. Molecular Genetics & Genomic Medicine, 2018, 6, 1168-1180.	0.6	16

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145	Polymorphisms in the 3â€² untranslated region of the IÎºB/MAD-3 (NFKBI) gene located on chromosome 14. Human Genetics, 1994, 93, 694-696.	1.8	15
146	Screening for CF mutations in adult cystic fibrosis patients with a directed and optimized SSCP strategy. Human Mutation, 1994, 3, 231-238.	1.1	15
147	Moving out: from sterol transport to drug resistance â€“ the ABCG subfamily of efflux pumps. Drug Metabolism and Drug Interactions, 2011, 26, 105-11.	0.3	15
148	Screening for cystic fibrosis mutations in Southern France: Identification of a frameshift mutation and two missense variations. Human Mutation, 1992, 1, 310-313.	1.1	13
149	Two new mutations detected by single-strand conformation polymorphism analysis in cystic fibrosis from Russia. Human Genetics, 1993, 91, 63-5.	1.8	13
150	Genetics of the Nevoid Basal Cell Carcinoma Syndrome. Advances in Cancer Research, 1996, 70, 49-61.	1.9	13
151	Variants in ARID5B gene are associated with the development of acute lymphoblastic leukemia in Mexican children. Annals of Hematology, 2019, 98, 2379-2388.	0.8	11
152	Targeted Deep Sequencing of Bladder Tumors Reveals Novel Associations between Cancer Gene Mutations and Mutational Signatures with Major Risk Factors. Clinical Cancer Research, 2021, 27, 3725-3733.	3.2	11
153	Towards a unified model of tumor suppression: lessons learned from the human patched gene. Biochimica Et Biophysica Acta: Reviews on Cancer, 1997, 1332, M43-M52.	3.3	10
154	Evidence that hereditary pancreatitis is genetically heterogeneous disorder. Pflugers Archiv European Journal of Physiology, 2000, 439, r050-r052.	1.3	10
155	A 22-bp deletion in the coding region of the cystic fibrosis gene. Genomics, 1992, 13, 235-236.	1.3	9
156	Identification of a rare cystic fibrosis mutation (S4X) in a Slovenian population. Human Molecular Genetics, 1993, 2, 315-316.	1.4	8
157	Evidence for linkage of the gene causing familial Mediterranean fever to chromosome 17q in non-Ashkenazi Jewish families: second locus or type I error?. Human Genetics, 1993, 91, 527-534.	1.8	7
158	Study of mutant and polyvariant mutant CFTR genes in patients with congenital absence of the vas deferens. Pflugers Archiv European Journal of Physiology, 2000, 439, r053-r055.	1.3	7
159	A rare null allele potentially encoding a dominant-negative TRIM5Î± protein in Baka pygmies. Virology, 2009, 391, 140-147.	1.1	6
160	Characterization of a YAC contig containing the NBCCS locus and a novel Kruppel-type zinc finger sequence on chromosome segment 9q22.3. , 1997, 18, 212-218.		5
161	Natural Animal Models of Human Genetic Diseases. , 2002, 70, 31-46.		5
162	HUMAN AND DROSOPHILA ABC PROTEINS. , 2003, , 47-61.		5

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163	Multicenter cohort association study of SLC2A1 single nucleotide polymorphisms and age-related macular degeneration. Molecular Vision, 2012, 18, 657-74.	1.1	5
164	Prenatal diagnosis and linkage disequilibrium with cystic fibrosis for markers surrounding D7S8. Human Genetics, 1990, 85, 275-8.	1.8	3
165	Screening methods for cystic fibrosis transmembrane conductance regulator (CFTR) gene mutations in non-human primates. Pflugers Archiv European Journal of Physiology, 2000, 439, r012-r013.	1.3	3
166	Characterisation of SNP haplotype structure in chemokine and chemokine receptor genes using CEPH pedigrees and statistical estimation. Human Genomics, 2004, 1, 195-207.	1.4	3
167	scDPN for High-throughput Single-cell CNV Detection to Uncover Clonal Evolution During HCC Recurrence. Genomics, Proteomics and Bioinformatics, 2021, 19, 346-357.	3.0	3
168	Identification of Cystic Fibrosis Mutations. Advances in Experimental Medicine and Biology, 1991, 290, 45-51.	0.8	2
169	Heteroduplex Analysis. , 1996, , 241-251.		2
170	Linkage Analysis for Monogenic Traits. , 2010, , 211-241.		1
171	D7S449 detects a HindIII polymorphism tightly linked to the MET gene on chromosome 7. Nucleic Acids Research, 1990, 18, 7199-7199.	6.5	0
172	[13] Comparison of the sensitivity of single-strand conformational polymorphism and heteroduplex methods. Methods in Neurosciences, 1995, 26, 194-209.	0.5	0
173	Cancer Stem Cells and New Therapeutic Approaches. , 0, , 217-232.		0
174	From Gene to Therapy: Understanding Human Disease through Genetics. Colloquium Series on the Genetic Basis of Human Disease, 2017, 5, i-89.	0.0	0
175	The Identification of Genes for Complex Genetic Diseases. Stadler Genetics Symposia Series, 2000, , 35-41.	0.0	0
176	Breast Cancer Stem Cells. , 2009, , 167-192.		0
177	Screening methods for cystic fibrosis transmembrane conductance regulator (CFTR) gene mutations in non-human primates. Pflugers Archiv European Journal of Physiology, 2000, 439, R12-R13.	1.3	0
178	Evidence that hereditary pancreatitis is genetically heterogeneous disorder. Pflugers Archiv European Journal of Physiology, 2000, 439, R50-R52.	1.3	0
179	Study of mutant and polyvariant mutant CFTR genes in patients with congenital absence of the vas deferens. Pflugers Archiv European Journal of Physiology, 2000, 439, R53-R55.	1.3	0