Kelly A Frazer

List of Publications by Year
in descending order

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132 papers	25,289 citations	22099 59 h-index	11288 136 g-index
161 all docs	161 docs citations	161 times ranked	37964 citing authors

#	Article	IF	Citations
1	A second generation human haplotype map of over 3.1 million SNPs. Nature, 2007, 449, 851-861.	13.7	4,137
2	VISTA: computational tools for comparative genomics. Nucleic Acids Research, 2004, 32, W273-W279.	6.5	2,033
3	Genome-wide detection and characterization of positive selection in human populations. Nature, 2007, 449, 913-918.	13.7	1,788
4	Blocks of Limited Haplotype Diversity Revealed by High-Resolution Scanning of Human Chromosome 21. Science, 2001, 294, 1719-1723.	6.0	1,082
5	Whole-Genome Patterns of Common DNA Variation in Three Human Populations. Science, 2005, 307, 1072-1079.	6.0	1,074
6	PipMaker---A Web Server for Aligning Two Genomic DNA Sequences. Genome Research, 2000, 10, 577-586.	2.4	1,070
7	Human genetic variation and its contribution to complex traits. Nature Reviews Genetics, 2009, 10, 241-251.	7.7	942
8	PI3Kî³ is a molecular switch that controls immune suppression. Nature, 2016, 539, 437-442.	13.7	884
9	Common Sequence Polymorphisms Shaping Genetic Diversity in Arabidopsis thaliana. Science, 2007, 317, 338-342.	6.0	689
10	Genomewide SNP variation reveals relationships among landraces and modern varieties of rice. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 12273-12278.	3.3	581
11	Common vs. rare allele hypotheses for complex diseases. Current Opinion in Genetics and Development, 2009, 19, 212-219.	1.5	568
12	9p21 DNA variants associated with coronary artery disease impair interferon-î³ signalling response. Nature, 2011, 470, 264-268.	13.7	557
13	Evaluation of next generation sequencing platforms for population targeted sequencing studies. Genome Biology, 2009, 10, R32.	13.9	510
14	High-Resolution Whole-Genome Association Study of Parkinson Disease. American Journal of Human Genetics, 2005, 77, 685-693.	2.6	479
15	Implementing genomic medicine in the clinic: the future is here. Genetics in Medicine, 2013, 15, 258-267.	1.1	472
16	Microdroplet-based PCR enrichment for large-scale targeted sequencing. Nature Biotechnology, 2009, 27, 1025-1031.	9.4	425
17	A sequence-based variation map of 8.27 million SNPs in inbred mouse strains. Nature, 2007, 448, 1050-1053.	13.7	406
18	Identification of Liver Cancer Progenitors Whose Malignant Progression Depends on Autocrine IL-6 Signaling. Cell, 2013, 155, 384-396.	13.5	384

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19	Common deletions and SNPs are in linkage disequilibrium in the human genome. Nature Genetics, 2006, 38, 82-85.	9.4	338
20	Genome-wide scan identifies variation in MLXIPL associated with plasma triglycerides. Nature Genetics, 2008, 40, 149-151.	9.4	303
21	Active Conservation of Noncoding Sequences Revealed by Three-Way Species Comparisons. Genome Research, 2000, 10, 1304-1306.	2.4	279
22	Fine-scale recombination patterns differ between chimpanzees and humans. Nature Genetics, 2005, 37, 429-434.	9.4	263
23	Cross-Species Sequence Comparisons: A Review of Methods and Available Resources. Genome Research, 2003, 13, 1-12.	2.4	210
24	ADAR1 Activation Drives Leukemia Stem Cell Self-Renewal by Impairing Let-7 Biogenesis. Cell Stem Cell, 2016, 19, 177-191.	5.2	182
25	Transcriptome Sequencing Reveals Potential Mechanism of Cryptic 3â€™ Splice Site Selection in SF3B1-mutated Cancers. PLoS Computational Biology, 2015, 11, e1004105.	1.5	177
26	A Pan-BCL2 Inhibitor Renders Bone-Marrow-Resident Human Leukemia Stem Cells Sensitive to Tyrosine Kinase Inhibition. Cell Stem Cell, 2013, 12, 316-328.	5.2	167
27	Genomic and transcriptomic association studies identify 16 novel susceptibility loci for venous thromboembolism. Blood, 2019, 134, 1645-1657.	0.6	162
28	Large-Scale Profiling Reveals the Influence of Genetic Variation on Gene Expression in Human Induced Pluripotent Stem Cells. Cell Stem Cell, 2017, 20, 533-546.e7.	5.2	157
29	ADAR1 promotes malignant progenitor reprogramming in chronic myeloid leukemia. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, 1041-1046.	3.3	148
30	iPSCORE: A Resource of 222 iPSC Lines Enabling Functional Characterization of Genetic Variation across a Variety of Cell Types. Stem Cell Reports, 2017, 8, 1086-1100.	2.3	147
31	Analysis of allelic differential expression in human white blood cells. Genome Research, 2006, 16, 331-339.	2.4	134
32	miR-150 influences B-cell receptor signaling in chronic lymphocytic leukemia by regulating expression of GAB1 and FOXP1. Blood, 2014, 124, 84-95.	0.6	129
33	Computational and Biological Analysis of 680 kb of DNA Sequence from the Human 5q31 Cytokine Gene Cluster Region. Genome Research, 1997, 7, 495-512.	2.4	124
34	The apolipoprotein(a) gene is regulated by sex hormones and acuteâ€“phase inducers in YAC transgenic mice. Nature Genetics, 1995, 9, 424-431.	9.4	120
35	Whole-Genome Sequencing Uncovers the Genetic Basis of Chronic Mountain Sickness in Andean Highlanders. American Journal of Human Genetics, 2013, 93, 452-462.	2.6	115
36	Experimental selection of hypoxia-tolerant Drosophila melanogaster. Proceedings of the National Academy of Sciences of the United States of America, 2011, 108, 2349-2354.	3.3	105

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37	Enrichment of sequencing targets from the human genome by solution hybridization. Genome Biology, 2009, 10, R116.	13.9	104
38	High-level ROR1 associates with accelerated disease progression in chronic lymphocytic leukemia. Blood, 2016, 128, 2931-2940.	0.6	102
39	Effective filtering strategies to improve data quality from population-based whole exome sequencing studies. BMC Bioinformatics, 2014, 15, 125.	1.2	101
40	Accurate detection and genotyping of SNPs utilizing population sequencing data. Genome Research, 2010, 20, 537-545.	2.4	100
41	Subtle changes in chromatin loop contact propensity are associated with differential gene regulation and expression. Nature Communications, 2019, 10, 1054.	5.8	100
42	Cellular deconvolution of GTEx tissues powers discovery of disease and cell-type associated regulatory variants. Nature Communications, 2020, 11, 955.	5.8	96
43	Fine Mapping in 94 Inbred Mouse Strains Using a High-Density Haplotype Resource. Genetics, 2010, 185, 1081-1095.	1.2	95
44	Genome-wide mutational landscape of mucinous carcinomatosis peritonei of appendiceal origin. Genome Medicine, 2014, 6, 43.	3.6	94
45	PTEN regulates glioblastoma oncogenesis through chromatin-associated complexes of DAXX and histone H3.3. Nature Communications, 2017, 8, 15223.	5.8	94
46	Systematic analysis of binding of transcription factors to noncoding variants. Nature, 2021, 591, 147-151.	13.7	89
47	Insights into the Mutational Burden of Human Induced Pluripotent Stem Cells from an Integrative Multi-Omics Approach. Cell Reports, 2018, 24, 883-894.	2.9	85
48	Application of pooled genotyping to scan candidate regions for association with HDL cholesterol levels. Human Genomics, 2004, 1, 421.	1.4	83
49	Pancreatic islet chromatin accessibility and conformation reveals distal enhancer networks of type 2 diabetes risk. Nature Communications, 2019, 10, 2078.	5.8	82
50	Genomic DNA Insertions and Deletions Occur Frequently Between Humans and Nonhuman Primates. Genome Research, 2003, 13, 341-346.	2.4	81
51	Detection of low prevalence somatic mutations in solid tumors with ultra-deep targeted sequencing. Genome Biology, 2011, 12, R124.	13.9	81
52	Segmental Phylogenetic Relationships of Inbred Mouse Strains Revealed by Fine-Scale Analysis of Sequence Variation Across 4.6 Mb of Mouse Genome. Genome Research, 2004, 14, 1493-1500.	2.4	78
53	Identification of high-confidence somatic mutations in whole genome sequence of formalin-fixed breast cancer specimens. Nucleic Acids Research, 2012, 40, e107-e107.	6.5	78
54	Enhancer release and retargeting activates disease-susceptibility genes. Nature, 2021, 595, 735-740.	13.7	76

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55	Brief Report: Oxidative Stress Mediates Cardiomyocyte Apoptosis in a Human Model of Danon Disease and Heart Failure. Stem Cells, 2015, 33, 2343-2350.	1.4	74
56	Noncoding Sequences Conserved in a Limited Number of Mammals in the SIM2 Interval are Frequently Functional. Genome Research, 2004, 14, 367-372.	2.4	73
57	The Genomics Gold Rush. JAMA - Journal of the American Medical Association, 2007, 298, 218.	3.8	71
58	Whole genome sequencing of Ethiopian highlanders reveals conserved hypoxia tolerance genes. Genome Biology, 2014, 15, R36.	13.9	71
59	BAP1 mutation is a frequent somatic event in peritoneal malignant mesothelioma. Journal of Translational Medicine, 2015, 13, 122.	1.8	69
60	Properties of structural variants and short tandem repeats associated with gene expression and complex traits. Nature Communications, 2020, 11, 2927.	5.8	67
61	Functional screening of an asthma QTL in YAC transgenic mice. Nature Genetics, 1999, 23, 241-244.	9.4	64
62	Allele-Specific KRT1 Expression Is a Complex Trait. PLoS Genetics, 2006, 2, e93.	1.5	64
63	Systematic transcriptome analysis reveals tumor-specific isoforms for ovarian cancer diagnosis and therapy. Proceedings of the National Academy of Sciences of the United States of America, 2015, 112, E3050-7.	3.3	64
64	High-Throughput and Cost-Effective Characterization of Induced Pluripotent Stem Cells. Stem Cell Reports, 2017, 8, 1101-1111.	2.3	64
65	Sensitive gene fusion detection using ambiguously mapping RNA-Seq read pairs. Bioinformatics, 2011, 27, 1068-1075.	1.8	53
66	Glioblastoma cellular cross-talk converges on NF-Î¹B to attenuate EGFR inhibitor sensitivity. Genes and Development, 2017, 31, 1212-1227.	2.7	53
67	Method for improving sequence coverage uniformity of targeted genomic intervals amplified by LR-PCR using Illumina GA sequencing-by-synthesis technology. BioTechniques, 2009, 46, 229-231.	0.8	51
68	The resequencing imperative. Nature Genetics, 2007, 39, 439-440.	9.4	47
69	An RNA editing fingerprint of cancer stem cell reprogramming. Journal of Translational Medicine, 2015, 13, 52.	1.8	46
70	Longitudinal assessment of tumor development using cancer avatars derived from genetically engineered pluripotent stem cells. Nature Communications, 2020, 11, 550.	5.8	45
71	Photoreceptor Localization of the KIF3A and KIF3B Subunits of the Heterotrimeric Microtubule Motor Kinesin II in Vertebrate Retina. Experimental Eye Research, 1999, 69, 491-503.	1.2	44
72	Association of Human iPSC Gene Signatures and X Chromosome Dosage with Two Distinct Cardiac Differentiation Trajectories. Stem Cell Reports, 2019, 13, 924-938.	2.3	44

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73	Variation in conserved non-coding sequences on chromosome 5q and susceptibility to asthma and atopy. Respiratory Research, 2005, 6, 145.	1.4	43
74	Identification of rare and common regulatory variants in pluripotent cells using population-scale transcriptomics. Nature Genetics, 2021, 53, 313-321.	9.4	42
75	Decoding the human genome. Genome Research, 2012, 22, 1599-1601.	2.4	37
76	Cell-Surface Marker Signature for Enrichment of Ventricular Cardiomyocytes Derived from Human Embryonic Stem Cells. Stem Cell Reports, 2018, 11, 828-841.	2.3	37
77	Mutations in topoisomerase IIÎ² result in a B cell immunodeficiency. Nature Communications, 2019, 10, 3644.	5.8	37
78	Reversion to an embryonic alternative splicing program enhances leukemia stem cell self-renewal. Proceedings of the National Academy of Sciences of the United States of America, 2015, 112, 15444-15449.	3.3	36
79	Pgltools: a genomic arithmetic tool suite for manipulation of Hi-C peak and other chromatin interaction data. BMC Bioinformatics, 2017, 18, 207.	1.2	35
80	Allele-specific NKX2-5 binding underlies multiple genetic associations with human electrocardiographic traits. Nature Genetics, 2019, 51, 1506-1517.	9.4	35
81	Population sequencing of two endocannabinoid metabolic genes identifies rare and common regulatory variants associated with extreme obesity and metabolite level. Genome Biology, 2010, 11, R118.	13.9	34
82	Systematic genetic analysis of the MHC region reveals mechanistic underpinnings of HLA type associations with disease. ELife, 2019, 8, .	2.8	34
83	Kataegis Expression Signature in Breast Cancer Is Associated with Late Onset, Better Prognosis, and Higher HER2 Levels. Cell Reports, 2016, 16, 672-683.	2.9	33
84	Aberrant DNA Methylation in Human iPSCs Associates with MYC-Binding Motifs in a Clone-Specific Manner Independent of Genetics. Cell Stem Cell, 2017, 20, 505-517.e6.	5.2	33
85	Human iPSC-Derived Retinal Pigment Epithelium: A Model System for Prioritizing and Functionally Characterizing Causal Variants at AMD Risk Loci. Stem Cell Reports, 2019, 12, 1342-1353.	2.3	32
86	In Vitro Human Keratinocyte Migration Rates Are Associated with SNPs in the KRT1 Interval. PLoS ONE, 2007, 2, e697.	1.1	31
87	Genetic determinants of phenotypic diversity in humans. Genome Biology, 2008, 9, 215.	13.9	31
88	Establishing the involvement of the novel geneAGBL5in retinitis pigmentosa by whole genome sequencing. Physiological Genomics, 2016, 48, 922-927.	1.0	29
89	Biased estimates of clonal evolution and subclonal heterogeneity can arise from PCR duplicates in deep sequencing experiments. Genome Biology, 2014, 15, 420.	3.8	28
90	Joint effects of cancer and variants in the factor 5 gene on the risk of venous thromboembolism. Haematologica, 2016, 101, 1046-1053.	1.7	28

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91	Identification of Common and Rare Genetic Variation Associated With Plasma Protein Levels Using Whole-Exome Sequencing and Mass Spectrometry. Circulation Genomic and Precision Medicine, 2018, 11, e002170.	1.6	26
92	A radiation hybrid map of the region on human chromosome 22 containing the neurofibromatosis type 2 locus. Genomics, 1992, 14, 574-584.	1.3	25
93	High-Resolution Mutational Profiling Suggests the Genetic Validity of Glioblastoma Patient-Derived Pre-Clinical Models. PLoS ONE, 2013, 8, e56185.	1.1	25
94	SARS-CoV-2 susceptibility and COVID-19 disease severity are associated with genetic variants affecting gene expression in a variety of tissues. Cell Reports, 2021, 37, 110020.	2.9	25
95	Faithful Expression of the Human 5q31 Cytokine Cluster in Transgenic Mice. Journal of Immunology, 2000, 164, 4569-4574.	0.4	23
96	Mutascope: sensitive detection of somatic mutations from deep amplicon sequencing. Bioinformatics, 2013, 29, 1908-1909.	1.8	22
97	Identifying DNase I hypersensitive sites as driver distal regulatory elements in breast cancer. Nature Communications, 2017, 8, 436.	5.8	22
98	A largeâ€scale exome array analysis of venous thromboembolism. Genetic Epidemiology, 2019, 43, 449-457.	0.6	22
99	Discovery and quality analysis of a comprehensive set of structural variants and short tandem repeats. Nature Communications, 2020, 11, 2928.	5.8	22
100	Elucidating the role of 8q24 in colorectal cancer. Nature Genetics, 2009, 41, 868-869.	9.4	21
101	A Network of microRNAs Acts to Promote Cell Cycle Exit and Differentiation of Human Pancreatic Endocrine Cells. IScience, 2019, 21, 681-694.	1.9	21
102	Ultraâ€Sharp Nanowire Arrays Natively Permeate, Record, and Stimulate Intracellular Activity in Neuronal and Cardiac Networks. Advanced Functional Materials, 2022, 32, 2108378.	7.8	21
103	In heart failure reactivation of RNA-binding proteins is associated with the expression of 1,523 fetal-specific isoforms. PLoS Computational Biology, 2022, 18, e1009918.	1.5	19
104	Associations Between Common and Rare Exonic Genetic Variants and Serum Levels of 20 Cardiovascular-Related Proteins. Circulation: Cardiovascular Genetics, 2016, 9, 375-383.	5.1	18
105	Evaluation of ultra-deep targeted sequencing for personalized breast cancer care. Breast Cancer Research, 2013, 15, R115.	2.2	16
106	Network-based analysis identifies epigenetic biomarkers of esophageal squamous cell carcinoma progression. Bioinformatics, 2014, 30, 3054-3061.	1.8	15
107	Activation of hedgehog signaling associates with early disease progression in chronic lymphocytic leukemia. Blood, 2019, 133, 2651-2663.	0.6	15
108	Mutational Profiling Can Establish Clonal or Independent Origin in Synchronous Bilateral Breast and Other Tumors. PLoS ONE, 2015, 10, e0142487.	1.1	15

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109	Whole Genome Sequencing Revealed Mutations in Two Independent Genes as the Underlying Cause of Retinal Degeneration in an Ashkenazi Jewish Pedigree. Genes, 2017, 8, 210.	1.0	14
110	Deciphering the genetic architecture and ethnographic distribution of IRD in three ethnic populations by whole genome sequence analysis. PLoS Genetics, 2021, 17, e1009848.	1.5	13
111	Multiâ€phenotype analyses of hemostatic traits with cardiovascular events reveal novel genetic associations. Journal of Thrombosis and Haemostasis, 2022, 20, 1331-1349.	1.9	12
112	Genetic ancestry of participants in the National Childrenâ€™s Study. Genome Biology, 2014, 15, R22.	13.9	11
113	IFT88 mutations identified in individuals with non-syndromic recessive retinal degeneration result in abnormal ciliogenesis. Human Genetics, 2018, 137, 447-458.	1.8	11
114	Evaluation of the SNP tagging approach in an independent population sampleâ€”array-based SNP discovery in Sami. Human Genetics, 2007, 122, 141-150.	1.8	10
115	A mutation in IFT43 causes non-syndromic recessive retinal degeneration. Human Molecular Genetics, 2017, 26, 4741-4751.	1.4	10
116	Efficient Prioritization of Multiple Causal eQTL Variants via Sparse Polygenic Modeling. Genetics, 2017, 207, 1301-1312.	1.2	10
117	Genomic integrity of human induced pluripotent stem cells across nine studies in the NHLBI NextGen program. Stem Cell Research, 2020, 46, 101803.	0.3	10
118	Fibrinogen gamma gene rs2066865 and risk of cancer-related venous thromboembolism. Haematologica, 2020, 105, 1963-1968.	1.7	10
119	Whole Transcriptome Sequencing Enables Discovery and Analysis of Viruses in Archived Primary Central Nervous System Lymphomas. PLoS ONE, 2013, 8, e73956.	1.1	9
120	Consanguinity and rare mutations outside of MCCC genes underlie nonspecific phenotypes of MCCD. Genetics in Medicine, 2015, 17, 660-667.	1.1	9
121	Transcriptome Sequencing of Tumor Subpopulations Reveals a Spectrum of Therapeutic Options for Squamous Cell Lung Cancer. PLoS ONE, 2013, 8, e58714.	1.1	9
122	Homozygous GNAS 393C-Allele Carriers with Locally Advanced Esophageal Cancer Fail to Benefit from Platinum-Based Preoperative Chemoradiotherapy. Annals of Surgical Oncology, 2014, 21, 4375-4382.	0.7	8
123	Effect of prothrombotic genotypes on the risk of venous thromboembolism in patients with and without ischemic stroke. The TromsÃ¸ Study. Journal of Thrombosis and Haemostasis, 2019, 17, 749-758.	1.9	8
124	In vitro Differentiation of Human iPSC-derived Cardiovascular Progenitor Cells (iPSC-CVPCs). Bio-protocol, 2020, 10, e3755.	0.2	8
125	In vitro Differentiation of Human iPSC-derived Retinal Pigment Epithelium Cells (iPSC-RPE). Bio-protocol, 2019, 9, e3469.	0.2	6
126	Detection and validation of novel mutations in MERTK in a simplex case of retinal degeneration using WGS and hiPSCâ€“RPEs model. Human Mutation, 2021, 42, 189-199.	1.1	5

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127	Genomics Links Inflammation With Neurocognitive Impairment in Children Living With Human Immunodeficiency Virus Type-1. Journal of Infectious Diseases, 2021, 224, 870-880.	1.9	3
128	Implementing Genomic Medicine in the Clinic. Obstetrical and Gynecological Survey, 2013, 68, 621-623.	0.2	1
129	MiningABs: mining associated biomarkers across multi-connected gene expression datasets. BMC Bioinformatics, 2014, 15, 173.	1.2	1
130	Revealing Instability: Genetic Variation Underlies Variability in mESC Pluripotency. Cell Stem Cell, 2020, 27, 347-349.	5.2	0
131	Inhibition Of Inflammation Driven Leukemia Stem Cell Self-Renewal With a Selective JAK2 Antagonist. Blood, 2013, 122, 1481-1481.	0.6	0
132	The Role Of CD44 Isoform Expression In Niche Resident Chronic Myeloid Leukemia Stem Cell Evolution. Blood, 2013, 122, 4028-4028.	0.6	0