Kelly A Frazer

List of Publications by Citations

138 papers	20,378 citations	54 h-index	142 g-index
161 ext. papers	23,341 ext. citations	14.3 avg, IF	6 L-index

#	Paper	IF	Citations
138	A second generation human haplotype map of over 3.1 million SNPs. *Nature*, **2007**, 449, 851-61	50.4	3647
137	VISTA: computational tools for comparative genomics. *Nucleic Acids Research*, **2004**, 32, W273-9	20.1	1441
136	Genome-wide detection and characterization of positive selection in human populations. *Nature*, **2007**, 449, 913-8	50.4	1367
135	Whole-genome patterns of common DNA variation in three human populations. *Science*, **2005**, 307, 1072-9	33.3	972
134	Blocks of limited haplotype diversity revealed by high-resolution scanning of human chromosome 21. *Science*, **2001**, 294, 1719-23	33.3	948
133	PipMaker--a web server for aligning two genomic DNA sequences. *Genome Research*, **2000**, 10, 577-86	9.7	906
132	Human genetic variation and its contribution to complex traits. *Nature Reviews Genetics*, **2009**, 10, 241-51	30.1	778
131	PI3K is a molecular switch that controls immune suppression. *Nature*, **2016**, 539, 437-442	50.4	609
130	Common sequence polymorphisms shaping genetic diversity in Arabidopsis thaliana. *Science*, **2007**, 317, 338-42	33.3	596
129	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, 12273-8	11.5	499
128	9p21 DNA variants associated with coronary artery disease impair interferon- signalling response. *Nature*, **2011**, 470, 264-8	50.4	490
127	Common vs. rare allele hypotheses for complex diseases. *Current Opinion in Genetics and Development*, **2009**, 19, 212-9	4.9	490
126	Evaluation of next generation sequencing platforms for population targeted sequencing studies. *Genome Biology*, **2009**, 10, R32	18.3	451
125	High-resolution whole-genome association study of Parkinson disease. *American Journal of Human Genetics*, **2005**, 77, 685-93	11	433
124	Implementing genomic medicine in the clinic: the future is here. *Genetics in Medicine*, **2013**, 15, 258-67	8.1	385
123	Microdroplet-based PCR enrichment for large-scale targeted sequencing. *Nature Biotechnology*, **2009**, 27, 1025-31	44.5	373
122	A sequence-based variation map of 8.27 million SNPs in inbred mouse strains. *Nature*, **2007**, 448, 1050-3	50.4	352

121	Identification of liver cancer progenitors whose malignant progression depends on autocrine IL-6 signaling. *Cell*, **2013**, 155, 384-96	56.2	323
120	Common deletions and SNPs are in linkage disequilibrium in the human genome. *Nature Genetics*, **2006**, 38, 82-5	36.3	303
119	Genome-wide scan identifies variation in MLXIPL associated with plasma triglycerides. *Nature Genetics*, **2008**, 40, 149-51	36.3	262
118	Active conservation of noncoding sequences revealed by three-way species comparisons. *Genome Research*, **2000**, 10, 1304-6	9.7	240
117	Fine-scale recombination patterns differ between chimpanzees and humans. *Nature Genetics*, **2005**, 37, 429-34	36.3	226
116	Cross-species sequence comparisons: a review of methods and available resources. *Genome Research*, **2003**, 13, 1-12	9.7	170
115	Transcriptome sequencing reveals potential mechanism of cryptic 3Usplice site selection in SF3B1-mutated cancers. *PLoS Computational Biology*, **2015**, 11, e1004105	5	145
114	A Pan-BCL2 inhibitor renders bone-marrow-resident human leukemia stem cells sensitive to tyrosine kinase inhibition. *Cell Stem Cell*, **2013**, 12, 316-28	18	140
113	ADAR1 Activation Drives Leukemia Stem Cell Self-Renewal by Impairing Let-7 Biogenesis. *Cell Stem Cell*, **2016**, 19, 177-191	18	120
112	Analysis of allelic differential expression in human white blood cells. *Genome Research*, **2006**, 16, 331-9	9.7	119
111	ADAR1 promotes malignant progenitor reprogramming in chronic myeloid leukemia. *Proceedings of the National Academy of Sciences of the United States of America*, **2013**, 110, 1041-6	11.5	118
110	The apolipoprotein(a) gene is regulated by sex hormones and acute-phase inducers in YAC transgenic mice. *Nature Genetics*, **1995**, 9, 424-31	36.3	112
109	Computational and biological analysis of 680 kb of DNA sequence from the human 5q31 cytokine gene cluster region. *Genome Research*, **1997**, 7, 495-512	9.7	111
108	miR-150 influences B-cell receptor signaling in chronic lymphocytic leukemia by regulating expression of GAB1 and FOXP1. *Blood*, **2014**, 124, 84-95	2.2	106
107	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	18	105
106	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	8	93
105	Enrichment of sequencing targets from the human genome by solution hybridization. *Genome Biology*, **2009**, 10, R116	18.3	92
104	Whole-genome sequencing uncovers the genetic basis of chronic mountain sickness in Andean highlanders. *American Journal of Human Genetics*, **2013**, 93, 452-62	11	90

103	Experimental selection of hypoxia-tolerant Drosophila melanogaster. *Proceedings of the National Academy of Sciences of the United States of America*, **2011**, 108, 2349-54	11.5	85
102	Accurate detection and genotyping of SNPs utilizing population sequencing data. *Genome Research*, **2010**, 20, 537-45	9.7	84
101	Fine mapping in 94 inbred mouse strains using a high-density haplotype resource. *Genetics*, **2010**, 185, 1081-95	4	82
100	High-level ROR1 associates with accelerated disease progression in chronic lymphocytic leukemia. *Blood*, **2016**, 128, 2931-2940	2.2	75
99	Genome-wide mutational landscape of mucinous carcinomatosis peritonei of appendiceal origin. *Genome Medicine*, **2014**, 6, 43	14.4	73
98	Detection of low prevalence somatic mutations in solid tumors with ultra-deep targeted sequencing. *Genome Biology*, **2011**, 12, R124	18.3	73
97	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, 1493-500	9.7	71
96	Identification of high-confidence somatic mutations in whole genome sequence of formalin-fixed breast cancer specimens. *Nucleic Acids Research*, **2012**, 40, e107	20.1	69
95	Genomic DNA insertions and deletions occur frequently between humans and nonhuman primates. *Genome Research*, **2003**, 13, 341-6	9.7	68
94	Application of pooled genotyping to scan candidate regions for association with HDL cholesterol levels. *Human Genomics*, **2004**, 1, 421-34	6.8	68
93	Effective filtering strategies to improve data quality from population-based whole exome sequencing studies. *BMC Bioinformatics*, **2014**, 15, 125	3.6	67
92	PTEN regulates glioblastoma oncogenesis through chromatin-associated complexes of DAXX and histone H3.3. *Nature Communications*, **2017**, 8, 15223	17.4	66
91	Genomic and transcriptomic association studies identify 16 novel susceptibility loci for venous thromboembolism. *Blood*, **2019**, 134, 1645-1657	2.2	63
90	Brief Report: Oxidative Stress Mediates Cardiomyocyte Apoptosis in a Human Model of Danon Disease and Heart Failure. *Stem Cells*, **2015**, 33, 2343-50	5.8	61
89	Noncoding sequences conserved in a limited number of mammals in the SIM2 interval are frequently functional. *Genome Research*, **2004**, 14, 367-72	9.7	61
88	Whole genome sequencing of Ethiopian highlanders reveals conserved hypoxia tolerance genes. *Genome Biology*, **2014**, 15, R36	18.3	59
87	Allele-specific KRT1 expression is a complex trait. *PLoS Genetics*, **2006**, 2, e93	6	59
86	The genomics gold rush. *JAMA - Journal of the American Medical Association*, **2007**, 298, 218-21	27.4	59

85	BAP1 mutation is a frequent somatic event in peritoneal malignant mesothelioma. *Journal of Translational Medicine*, **2015**, 13, 122	8.5	55
84	Functional screening of an asthma QTL in YAC transgenic mice. *Nature Genetics*, **1999**, 23, 241-4	36.3	53
83	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	11.5	51
82	Method for improving sequence coverage uniformity of targeted genomic intervals amplified by LR-PCR using Illumina GA sequencing-by-synthesis technology. *BioTechniques*, **2009**, 46, 229-31	2.5	51
81	Insights into the Mutational Burden of Human Induced Pluripotent Stem Cells from an Integrative Multi-Omics Approach. *Cell Reports*, **2018**, 24, 883-894	10.6	49
80	High-Throughput and Cost-Effective Characterization of Induced Pluripotent Stem Cells. *Stem Cell Reports*, **2017**, 8, 1101-1111	8	48
79	Sensitive gene fusion detection using ambiguously mapping RNA-Seq read pairs. *Bioinformatics*, **2011**, 27, 1068-75	7.2	48
78	Subtle changes in chromatin loop contact propensity are associated with differential gene regulation and expression. *Nature Communications*, **2019**, 10, 1054	17.4	42
77	Pancreatic islet chromatin accessibility and conformation reveals distal enhancer networks of type 2 diabetes risk. *Nature Communications*, **2019**, 10, 2078	17.4	41
76	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	3.7	39
75	Glioblastoma cellular cross-talk converges on NF-B to attenuate EGFR inhibitor sensitivity. *Genes and Development*, **2017**, 31, 1212-1227	12.6	38
74	Cellular deconvolution of GTEx tissues powers discovery of disease and cell-type associated regulatory variants. *Nature Communications*, **2020**, 11, 955	17.4	37
73	An RNA editing fingerprint of cancer stem cell reprogramming. *Journal of Translational Medicine*, **2015**, 13, 52	8.5	36
72	Variation in conserved non-coding sequences on chromosome 5q and susceptibility to asthma and atopy. *Respiratory Research*, **2005**, 6, 145	7.3	36
71	Decoding the human genome. *Genome Research*, **2012**, 22, 1599-601	9.7	32
70	Cell-Surface Marker Signature for Enrichment of Ventricular Cardiomyocytes Derived from Human Embryonic Stem Cells. *Stem Cell Reports*, **2018**, 11, 828-841	8	29
69	In vitro human keratinocyte migration rates are associated with SNPs in the KRT1 interval. *PLoS ONE*, **2007**, 2, e697	3.7	28
68	Association of Human iPSC Gene Signatures and X Chromosome Dosage with Two Distinct Cardiac Differentiation Trajectories. *Stem Cell Reports*, **2019**, 13, 924-938	8	26

67	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	18	25
66	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	8	24
65	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, 15444-9	11.5	24
64	Mutations in topoisomerase II□esult in a B cell immunodeficiency. *Nature Communications*, **2019**, 10, 3644	17.4	24
63	Genetic determinants of phenotypic diversity in humans. *Genome Biology*, **2008**, 9, 215	18.3	24
62	Longitudinal assessment of tumor development using cancer avatars derived from genetically engineered pluripotent stem cells. *Nature Communications*, **2020**, 11, 550	17.4	23
61	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	18.3	23
60	Systematic analysis of binding of transcription factors to noncoding variants. *Nature*, **2021**, 591, 147-151	50.4	23
59	Properties of structural variants and short tandem repeats associated with gene expression and complex traits. *Nature Communications*, **2020**, 11, 2927	17.4	22
58	Biased estimates of clonal evolution and subclonal heterogeneity can arise from PCR duplicates in deep sequencing experiments. *Genome Biology*, **2014**, 15, 420	18.3	22
57	High-resolution mutational profiling suggests the genetic validity of glioblastoma patient-derived pre-clinical models. *PLoS ONE*, **2013**, 8, e56185	3.7	21
56	Mutascope: sensitive detection of somatic mutations from deep amplicon sequencing. *Bioinformatics*, **2013**, 29, 1908-9	7.2	20
55	A radiation hybrid map of the region on human chromosome 22 containing the neurofibromatosis type 2 locus. *Genomics*, **1992**, 14, 574-84	4.3	20
54	Joint effects of cancer and variants in the factor 5 gene on the risk of venous thromboembolism. *Haematologica*, **2016**, 101, 1046-53	6.6	20
53	Kataegis Expression Signature in Breast Cancer Is Associated with Late Onset, Better Prognosis, and Higher HER2 Levels. *Cell Reports*, **2016**, 16, 672-83	10.6	19
52	Pgltools: a genomic arithmetic tool suite for manipulation of Hi-C peak and other chromatin interaction data. *BMC Bioinformatics*, **2017**, 18, 207	3.6	18
51	Establishing the involvement of the novel gene AGBL5 in retinitis pigmentosa by whole genome sequencing. *Physiological Genomics*, **2016**, 48, 922-927	3.6	18
50	Faithful expression of the human 5q31 cytokine cluster in transgenic mice. *Journal of Immunology*, **2000**, 164, 4569-74	5.3	16

49	Identification of rare and common regulatory variants in pluripotent cells using population-scale transcriptomics. *Nature Genetics*, **2021**, 53, 313-321	36.3	16
48	A Network of microRNAs Acts to Promote Cell Cycle Exit and Differentiation of Human Pancreatic Endocrine Cells. *IScience*, **2019**, 21, 681-694	6.1	15
47	Evaluation of ultra-deep targeted sequencing for personalized breast cancer care. *Breast Cancer Research*, **2013**, 15, R115	8.3	14
46	Identifying DNase I hypersensitive sites as driver distal regulatory elements in breast cancer. *Nature Communications*, **2017**, 8, 436	17.4	14
45	Mutational Profiling Can Establish Clonal or Independent Origin in Synchronous Bilateral Breast and Other Tumors. *PLoS ONE*, **2015**, 10, e0142487	3.7	14
44	Allele-specific NKX2-5 binding underlies multiple genetic associations with human electrocardiographic traits. *Nature Genetics*, **2019**, 51, 1506-1517	36.3	14
43	Associations Between Common and Rare Exonic Genetic Variants and Serum Levels of 20 Cardiovascular-Related Proteins: The Troms Study. *Circulation: Cardiovascular Genetics*, **2016**, 9, 375-83		13
42	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	5.2	13
41	Systematic genetic analysis of the MHC region reveals mechanistic underpinnings of HLA type associations with disease. *ELife*, **2019**, 8,	8.9	12
40	Enhancer release and retargeting activates disease-susceptibility genes. *Nature*, **2021**, 595, 735-740	50.4	12
39	A large-scale exome array analysis of venous thromboembolism. *Genetic Epidemiology*, **2019**, 43, 449-457	2.6	11
38	Discovery and quality analysis of a comprehensive set of structural variants and short tandem repeats. *Nature Communications*, **2020**, 11, 2928	17.4	11
37	Activation of hedgehog signaling associates with early disease progression in chronic lymphocytic leukemia. *Blood*, **2019**, 133, 2651-2663	2.2	10
36	Whole Genome Sequencing Revealed Mutations in Two Independent Genes as the Underlying Cause of Retinal Degeneration in an Ashkenazi Jewish Pedigree. *Genes*, **2017**, 8,	4.2	10
35	Evaluation of the SNP tagging approach in an independent population sample--array-based SNP discovery in Sami. *Human Genetics*, **2007**, 122, 141-50	6.3	10
34	Network-based analysis identifies epigenetic biomarkers of esophageal squamous cell carcinoma progression. *Bioinformatics*, **2014**, 30, 3054-61	7.2	9
33	Whole transcriptome sequencing enables discovery and analysis of viruses in archived primary central nervous system lymphomas. *PLoS ONE*, **2013**, 8, e73956	3.7	9
32	Systematic assessment of regulatory effects of human disease variants in pluripotent cells		9

31	A mutation in IFT43 causes non-syndromic recessive retinal degeneration. *Human Molecular Genetics*, **2017**, 26, 4741-4751	5.6	8
30	IFT88 mutations identified in individuals with non-syndromic recessive retinal degeneration result in abnormal ciliogenesis. *Human Genetics*, **2018**, 137, 447-458	6.3	8
29	Consanguinity and rare mutations outside of MCCC genes underlie nonspecific phenotypes of MCCD. *Genetics in Medicine*, **2015**, 17, 660-7	8.1	8
28	Transcriptome sequencing of tumor subpopulations reveals a spectrum of therapeutic options for squamous cell lung cancer. *PLoS ONE*, **2013**, 8, e58714	3.7	8
27	Genetic ancestry of participants in the National Children's Study. *Genome Biology*, **2014**, 15, R22	18.3	7
26	Homozygous GNAS 393C-allele carriers with locally advanced esophageal cancer fail to benefit from platinum-based preoperative chemoradiotherapy. *Annals of Surgical Oncology*, **2014**, 21, 4375-82	3.1	6
25	Efficient Prioritization of Multiple Causal eQTL Variants via Sparse Polygenic Modeling. *Genetics*, **2017**, 207, 1301-1312	4	5
24	Fibrinogen gamma gene and risk of cancer-related venous thromboembolism. *Haematologica*, **2020**, 105, 1963-1968	6.6	5
23	Ultra-Sharp Nanowire Arrays Natively Permeate, Record, and Stimulate Intracellular Activity in Neuronal and Cardiac Networks. *Advanced Functional Materials*,2108378	15.6	4
22	Cellular deconvolution of GTEx tissues powers eQTL studies to discover thousands of novel disease and cell-type associated regulatory variants		4
21	Differentiation of Human iPSC-derived Retinal Pigment Epithelium Cells (iPSC-RPE). *Bio-protocol*, **2019**, 9, e3469	0.9	3
20	Genomic properties of structural variants and short tandem repeats that impact gene expression and complex traits in humans		3
19	Effect of prothrombotic genotypes on the risk of venous thromboembolism in patients with and without ischemic stroke. The Troms Study. *Journal of Thrombosis and Haemostasis*, **2019**, 17, 749-758	15.4	3
18	Correction: Genome-wide mutational landscape of mucinous carcinomatosis peritonei of appendiceal origin. *Genome Medicine*, **2014**, 6, 53	14.4	2
17	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	10.6	2
16	Integration of phased Hi-C and molecular phenotype data to study genetic and epigenetic effects on chromatin looping		2
15	Discovery and Quality Analysis of a Comprehensive Set of Structural Variants and Short Tandem Repeats		2
14	Genomic integrity of human induced pluripotent stem cells across nine studies in the NHLBI NextGen program. *Stem Cell Research*, **2020**, 46, 101803	1.6	2

13	Allele-specific NKX2-5 binding underlies multiple genetic associations with human EKG traits		2
12	Detection and validation of novel mutations in MERTK in a simplex case of retinal degeneration using WGS and hiPSC-RPEs model. *Human Mutation*, **2021**, 42, 189-199	4.7	2
11	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	5	2
10	Differentiation of Human iPSC-derived Cardiovascular Progenitor Cells (iPSC-CVPCs). *Bio-protocol*, **2020**, 10, e3755	0.9	1
9	Human iPSC gene signatures and X chromosome dosage impact response to WNT inhibition and cardiac differentiation fate		1
8	Genomics Links Inflammation With Neurocognitive Impairment in Children Living With Human Immunodeficiency Virus Type-1. *Journal of Infectious Diseases*, **2021**, 224, 870-880	7	1
7	Human iPSC-derived retinal pigment epithelium: a model system for identifying and functionally characterizing causal variants at AMD risk loci		1
6	Chromatin co-accessibility is highly structured, spans entire chromosomes, and mediates long range regulatory genetic effects		1
5	Deciphering the genetic architecture and ethnographic distribution of IRD in three ethnic populations by whole genome sequence analysis. *PLoS Genetics*, **2021**, 17, e1009848	6	0
4	MiningABs: mining associated biomarkers across multi-connected gene expression datasets. *BMC Bioinformatics*, **2014**, 15, 173	3.6	
3	Inhibition Of Inflammation Driven Leukemia Stem Cell Self-Renewal With a Selective JAK2 Antagonist. *Blood*, **2013**, 122, 1481-1481	2.2	
2	The Role Of CD44 Isoform Expression In Niche Resident Chronic Myeloid Leukemia Stem Cell Evolution. *Blood*, **2013**, 122, 4028-4028	2.2	
1	Revealing Instability: Genetic Variation Underlies Variability in mESC Pluripotency. *Cell Stem Cell*, **2020**, 27, 347-349	18	