Fiona Cunningham

List of Publications by Year in Descending Order

48 papers	19,953 citations	33 h-index	56 g-index
56 ext. papers	28,001 ext. citations	16.2 avg, IF	6.09 L-index

#	Paper	IF	Citations
48	DECIPHER: Supporting the interpretation and sharing of rare disease phenotype-linked variant data to advance diagnosis and research.. *Human Mutation*, **2022**,	4.7	2
47	The Gene Curation Coalition: A global effort to harmonize gene-disease evidence resources.. *Genetics in Medicine*, **2022**,	8.1	4
46	GA4GH: International policies and standards for data sharing across genomic research and healthcare.. *Cell Genomics*, **2021**, 1, 100029-100029		20
45	The European Variation Archive: a FAIR resource of genomic variation for all species. *Nucleic Acids Research*, **2021**,	20.1	10
44	Ensembl 2022. *Nucleic Acids Research*, **2021**,	20.1	72
43	Ensembl 2021. *Nucleic Acids Research*, **2021**, 49, D884-D891	20.1	324
42	GENCODE 2021. *Nucleic Acids Research*, **2021**, 49, D916-D923	20.1	82
41	The value of primary transcripts to the clinical and non-clinical genomics community: Survey results and roadmap for improvements. *Molecular Genetics & Genomic Medicine*, **2021**, e1786	2.3	2
40	Whole-genome sequencing of patients with rare diseases in a national health system. *Nature*, **2020**, 583, 96-102	50.4	139
39	Ensembl 2020. *Nucleic Acids Research*, **2020**, 48, D682-D688	20.1	645
38	Flexible and scalable diagnostic filtering of genomic variants using G2P with Ensembl VEP. *Nature Communications*, **2019**, 10, 2373	17.4	22
37	IQSEC2-related encephalopathy in males and females: a comparative study including 37 novel patients. *Genetics in Medicine*, **2019**, 21, 837-849	8.1	32
36	The NHGRI-EBI GWAS Catalog of published genome-wide association studies, targeted arrays and summary statistics 2019. *Nucleic Acids Research*, **2019**, 47, D1005-D1012	20.1	1422
35	GENCODE reference annotation for the human and mouse genomes. *Nucleic Acids Research*, **2019**, 47, D766-D773	20.1	1140
34	Ensembl 2019. *Nucleic Acids Research*, **2019**, 47, D745-D751	20.1	554
33	Ensembl 2018. *Nucleic Acids Research*, **2018**, 46, D754-D761	20.1	1822
32	A standardized framework for representation of ancestry data in genomics studies, with application to the NHGRI-EBI GWAS Catalog. *Genome Biology*, **2018**, 19, 21	18.3	87

31	Scaling up data curation using deep learning: An application to literature triage in genomic variation resources. *PLoS Computational Biology*, **2018**, 14, e1006390	5	21
30	Ensembl variation resources. *Database: the Journal of Biological Databases and Curation*, **2018**, 2018,	5	230
29	Haplosaurus computes protein haplotypes for use in precision drug design. *Nature Communications*, **2018**, 9, 4128	17.4	8
28	Ensembl 2017. *Nucleic Acids Research*, **2017**, 45, D635-D642	20.1	404
27	The new NHGRI-EBI Catalog of published genome-wide association studies (GWAS Catalog). *Nucleic Acids Research*, **2017**, 45, D896-D901	20.1	1321
26	Ensembl 2016. *Nucleic Acids Research*, **2016**, 44, D710-6	20.1	1094
25	The Ensembl Variant Effect Predictor. *Genome Biology*, **2016**, 17, 122	18.3	2595
24	Integrated allelic, transcriptional, and phenomic dissection of the cardiac effects of titin truncations in health and disease. *Science Translational Medicine*, **2015**, 7, 270ra6	17.5	267
23	Ensembl 2015. *Nucleic Acids Research*, **2015**, 43, D662-9	20.1	1013
22	Improving the Sequence Ontology terminology for genomic variant annotation. *Journal of Biomedical Semantics*, **2015**, 6, 32	2.2	20
21	Ensembl 2014. *Nucleic Acids Research*, **2014**, 42, D749-55	20.1	1087
20	Locus Reference Genomic: reference sequences for the reporting of clinically relevant sequence variants. *Nucleic Acids Research*, **2014**, 42, D873-8	20.1	58
19	Integrative annotation of variants from 1092 humans: application to cancer genomics. *Science*, **2013** , 342, 1235587	33.3	281
18	Ensembl 2013. *Nucleic Acids Research*, **2013**, 41, D48-55	20.1	797
17	Ensembl 2012. *Nucleic Acids Research*, **2012**, 40, D84-90	20.1	798
16	Paralogous annotation of disease-causing variants in long QT syndrome genes. *Human Mutation*, **2012**, 33, 1188-1191	4.7	26
15	A combined functional annotation score for non-synonymous variants. *Human Heredity*, **2012**, 73, 47-51	1.1	79
14	Ensembl 2011. *Nucleic Acids Research*, **2011**, 39, D800-6	20.1	590

13	Modernizing reference genome assemblies. *PLoS Biology*, **2011**, 9, e1001091	9.7	294
12	Ensembl's 10th year. *Nucleic Acids Research*, **2010**, 38, D557-62	20.1	240
11	Deriving the consequences of genomic variants with the Ensembl API and SNP Effect Predictor. *Bioinformatics*, **2010**, 26, 2069-70	7.2	1240
10	A standard variation file format for human genome sequences. *Genome Biology*, **2010**, 11, R88	18.3	71
9	Locus Reference Genomic sequences: an improved basis for describing human DNA variants. *Genome Medicine*, **2010**, 2, 24	14.4	86
8	A database and API for variation, dense genotyping and resequencing data. *BMC Bioinformatics*, **2010**, 11, 238	3.6	26
7	Ensembl variation resources. *BMC Genomics*, **2010**, 11, 293	4.5	104
6	TranscriptSNPView: a genome-wide catalog of mouse coding variation. *Nature Genetics*, **2006**, 38, 853	36.3	14
5	WormBase: a comprehensive data resource for Caenorhabditis biology and genomics. *Nucleic Acids Research*, **2005**, 33, D383-9	20.1	140
4	WormBase: a multi-species resource for nematode biology and genomics. *Nucleic Acids Research*, **2004**, 32, D411-7	20.1	543
3	WormBase: a cross-species database for comparative genomics. *Nucleic Acids Research*, **2003**, 31, 133-7	20.1	90
2	Annotation and curation of human genomic variations: an ELIXIR Implementation Study. *F1000Research*,9, 1207	3.6	
1	A standardized framework for representation of ancestry data in genomics studies, with application to the NHGRI-EBI GWAS Catalog		2