Caroline F Wright

List of Publications by Year
in descending order

77 papers	7,090 citations	101535 36 h-index	79691 73 g-index
101 all docs	101 docs citations	101 times ranked	12672 citing authors

#	Article	IF	Citations
1	The Gene Curation Coalition: A global effort to harmonize geneâ€“disease evidence resources. Genetics in Medicine, 2022, 24, 1732-1742.	2.4	56
2	Rare genetic variants in genes and loci linked to dominant monogenic developmental disorders cause milder related phenotypes in the general population. American Journal of Human Genetics, 2022, 109, 1308-1316.	6.2	35
3	Recommendations for clinical interpretation of variants found in non-coding regions of the genome. Genome Medicine, 2022, 14, .	8.2	65
4	Assessing performance of pathogenicity predictors using clinically relevant variant datasets. Journal of Medical Genetics, 2021, 58, 547-555.	3.2	57
5	Evaluating variants classified as pathogenic in ClinVar in the DDD Study. Genetics in Medicine, 2021, 23, 571-575.	2.4	16
6	Systematic assessment of outcomes following a genetic diagnosis identified through a large-scale research study into developmental disorders. Genetics in Medicine, 2021, 23, 1058-1064.	2.4	7
7	Use of SNP chips to detect rare pathogenic variants: retrospective, population based diagnostic evaluation. BMJ, The, 2021, 372, n214.	6.0	27
8	Common genetic variants with fetal effects on birth weight are enriched for proximity to genes implicated in rare developmental disorders. Human Molecular Genetics, 2021, 30, 1057-1066.	2.9	1
9	Unreliability of genotyping arrays for detecting very rare variants in human genetic studies: Example from a recent study of MC4R. Cell, 2021, 184, 1651.	28.9	8
10	Non-coding region variants upstream of MEF2C cause severe developmental disorder through three distinct loss-of-function mechanisms. American Journal of Human Genetics, 2021, 108, 1083-1094.	6.2	42
11	The contribution of X-linked coding variation to severe developmental disorders. Nature Communications, 2021, 12, 627.	12.8	33
12	Detecting cryptic clinically relevant structural variation in exome-sequencing data increases diagnostic yield for developmental disorders. American Journal of Human Genetics, 2021, 108, 2186-2194.	6.2	12
13	100,000 Genomes Pilot on Rare-Disease Diagnosis in Health Care â€" Preliminary Report. New England Journal of Medicine, 2021, 385, 1868-1880.	27.0	352
14	Expanded universal carrier screening and its implementation within a publicly funded healthcare service. Journal of Community Genetics, 2020, 11, 21-38.	1.2	31
15	Evidence for 28 genetic disorders discovered by combining healthcare and research data. Nature, 2020, 586, 757-762.	27.8	343
16	Large Copy-Number Variants in UK Biobank Caused by Clonal Hematopoiesis May Confound Penetrance Estimates. American Journal of Human Genetics, 2020, 107, 325-329.	6.2	6
17	Recurrent De Novo NAHR Reciprocal Duplications in the ATAD3 Gene Cluster Cause a Neurogenetic Trait with Perturbed Cholesterol and Mitochondrial Metabolism. American Journal of Human Genetics, 2020, 106, 272-279.	6.2	33
18	When genomic medicine reveals misattributed genetic relationshipsâ€"the debate about disclosure revisited. Genetics in Medicine, 2019, 21, 97-101.	2.4	7

#	Article	IF	Citations
19	Clinically-relevant postzygotic mosaicism in parents and children with developmental disorders in trio exome sequencing data. Nature Communications, 2019, 10, 2985.	12.8	64
20	Finding Diagnostically Useful Patterns in Quantitative Phenotypic Data. American Journal of Human Genetics, 2019, 105, 933-946.	6.2	8
21	Direct-to-consumer genetic testing. BMJ: British Medical Journal, 2019, 367, l5688.	2.3	64
22	Assessing the Pathogenicity, Penetrance, and Expressivity of Putative Disease-Causing Variants in a Population Setting. American Journal of Human Genetics, 2019, 104, 275-286.	6.2	158
23	Exome-wide assessment of the functional impact and pathogenicity of multinucleotide mutations. Genome Research, 2019, 29, 1047-1056.	5.5	34
24	Flexible and scalable diagnostic filtering of genomic variants using G2P with Ensembl VEP. Nature Communications, 2019, 10, 2373.	12.8	86
25	Using Structural Analysis In Silico to Assess the Impact of Missense Variants in MEN1. Journal of the Endocrine Society, 2019, 3, 2258-2275.	0.2	14
26	Pathogenicity and selective constraint on variation near splice sites. Genome Research, 2019, 29, 159-170.	5.5	70
27	Genomic variant sharing: a position statement. Wellcome Open Research, 2019, 4, 22.	1.8	31
28	Paediatric genomics: diagnosing rare disease in children. Nature Reviews Genetics, 2018, 19, 253-268.	16.3	369
29	Making new genetic diagnoses with old data: iterative reanalysis and reporting from genome-wide data in 1,133 families with developmental disorders. Genetics in Medicine, 2018, 20, 1216-1223.	2.4	255
30	KIAA1109 Variants Are Associated with a Severe Disorder of Brain Development and Arthrogryposis. American Journal of Human Genetics, 2018, 102, 116-132.	6.2	46
31	De novo mutations in regulatory elements in neurodevelopmental disorders. Nature, 2018, 555, 611-616.	27.8	232
32	Quantifying the contribution of recessive coding variation to developmental disorders. Science, 2018, 362, 1161-1164.	12.6	158
33	Common genetic variants contribute to risk of rare severe neurodevelopmental disorders. Nature, 2018, 562, 268-271.	27.8	246
34	Structural analysis of pathogenic mutations in theDYRK1Agene in patients with developmental disorders. Human Molecular Genetics, 2017, 26, ddw409.	2.9	33
35	De Novo Mutations in EBF3 Cause a Neurodevelopmental Syndrome. American Journal of Human Genetics, 2017, 100, 138-150.	6.2	52
36	Protein structure and phenotypic analysis of pathogenic and population missense variants inSTXBP1. Molecular Genetics & Genomic Medicine, 2017, 5, 495-507.	1.2	29

#	Article	IF	Citations
37	Returning genome sequences to research participants: Policy and practice. Wellcome Open Research, 2017, 2, 15.	1.8	24
38	Distinct genetic architectures for syndromic and nonsyndromic congenital heart defects identified by exome sequencing. Nature Genetics, 2016, 48, 1060-1065.	21.4	351
39	Principle of proportionality in genomic data sharing. Nature Reviews Genetics, 2016, 17, 1-2.	16.3	26
40	Integrating population variation and protein structural analysis to improve clinical interpretation of missense variation: application to the WD40 domain. Human Molecular Genetics, 2016, 25, 927-935.	2.9	26
41	Attitudes of nearly 7000 health professionals, genomic researchers and publics toward the return of incidental results from sequencing research. European Journal of Human Genetics, 2016, 24, 21-29.	2.8	161
42	Genetic diagnosis of developmental disorders in the DDD study: a scalable analysis of genome-wide research data. Lancet, The, 2015, 385, 1305-1314.	13.7	651
43	Facilitating Collaboration in Rare Genetic Disorders Through Effective Matchmaking in DECIPHER. Human Mutation, 2015, 36, 941-949.	2.5	38
44	Potential research participants support the return of raw sequence data. Journal of Medical Genetics, 2015, 52, 571-574.	3.2	38
45	No expectation to share incidental findings in genomic research. Lancet, The, 2015, 385, 1289-1290.	13.7	19
46	Discovery of four recessive developmental disorders using probabilistic genotype and phenotype matching among 4,125 families. Nature Genetics, 2015, 47, 1363-1369.	21.4	133
47	The Human Phenotype Ontology project: linking molecular biology and disease through phenotype data. Nucleic Acids Research, 2014, 42, D966-D974.	14.5	698
48	DECIPHER: database for the interpretation of phenotype-linked plausibly pathogenic sequence and copy-number variation. Nucleic Acids Research, 2014, 42, D993-D1000.	14.5	195
49	Conceptual issues for screening in the genomic era - time for an update?. Epidemiology Biostatistics and Public Health, 2014, 11, .	0.0	1
50	Informatics and clinical genome sequencing: opening the black box. Genetics in Medicine, 2013, 15, 165-171.	2.4	33
51	Policy challenges of clinical genome sequencing. BMJ, The, 2013, 347, f6845-f6845.	6.0	50
52	Empirical research on the ethics of genomic research. American Journal of Medical Genetics, Part A, 2013, 161, 2099-2101.	1.2	17
53	Risk Prediction Models: A Framework for Assessment. Public Health Genomics, 2012, 15, 98-105.	1.0	8
54	DECIPHER: web-based, community resource for clinical interpretation of rare variants in developmental disorders. Human Molecular Genetics, 2012, 21, R37-R44.	2.9	74

#	Article	IF	Citations
55	Non-invasive prenatal diagnostic test accuracy for fetal sex using cell-free DNA a review and meta-analysis. BMC Research Notes, 2012, 5, 476.	1.4	66
56	Direct-to-Consumer Genetic Testing. , 2012, , 215-236.		3
57	Strengthening the reporting of Genetic RIsk Prediction Studies (GRIPS): explanation and elaboration. Journal of Clinical Epidemiology, 2011, 64, e1-e22.	5.0	9
58	Strengthening the reporting of genetic risk prediction studies (GRIPS): explanation and elaboration. European Journal of Clinical Investigation, 2011, 41, 1010-1035.	3.4	30
59	The Deciphering Developmental Disorders (DDD) study. Developmental Medicine and Child Neurology, 2011, 53, 702-703.	2.1	153
60	Strengthening the reporting of genetic risk prediction studies (GRIPS): explanation and elaboration. European Journal of Human Genetics, 2011, 19, 615-615.	2.8	12
61	Review of massively parallel DNA sequencing technologies. The HUGO Journal, 2011, 5, 1-12.	4.1	67
62	Strengthening the reporting of genetic risk prediction studies (GRIPS): explanation and elaboration. European Journal of Epidemiology, 2011, 26, 313-337.	5.7	14
63	Regulating direct-to-consumer genetic tests: What is all the fuss about?. Genetics in Medicine, 2011, 13, 295-300.	2.4	57
64	Size of the direct-to-consumer genomic testing market. Genetics in Medicine, 2010, 12, 594.	2.4	38
65	Non-Invasive Prenatal Diagnosis Using Cell-Free Fetal DNA Technology: Applications and Implications. Public Health Genomics, 2010, 13, 246-255.	1.0	55
66	Realising the benefits of genetics for health. Lancet, The, 2010, 376, 1370-1371.	13.7	3
67	Extending the reach of public health genomics: What should be the agenda for public health in an era of genome-based and â€œpersonalizedâ€•medicine?. Genetics in Medicine, 2010, 12, 785-791.	2.4	95
68	Quality Issues in the Evaluation and Regulation of Genetic Testing Services: A Public Health Approach. , 2010, , 267-275.		0
69	Biomarkers, Dementia, and Public Health. Annals of the New York Academy of Sciences, 2009, 1180, 11-19.	3.8	26
70	Cell-free fetal DNA and RNA in maternal blood: implications for safer antenatal testing. BMJ: British Medical Journal, 2009, 339, b2451-b2451.	2.3	39
71	A new strategic phase for genomic medicine in UK health services. Genome Medicine, 2009, 1, 93.	8.2	4
72	The use of cell-free fetal nucleic acids in maternal blood for non-invasive prenatal diagnosis. Human Reproduction Update, 2008, 15, 139-151.	10.8	197

#	Article	IF	Citations
73	The importance of sequence diversity in the aggregation and evolution of proteins. Nature, 2005, 438, 878-881.	27.8	291
74	The importance of loop length in the folding of an immunoglobulin domain. Protein Engineering, Design and Selection, 2004, 17, 443-453.	2.1	16
75	Thermodynamic Characterisation of Two Transition States Along Parallel Protein Folding Pathways. Journal of Molecular Biology, 2004, 338, 445-451.	4.2	30
76	Parallel protein-unfolding pathways revealed and mapped. Nature Structural and Molecular Biology, 2003, 10, 658-662.	8.2	153
77	Genomic variant sharing: a position statement. Wellcome Open Research, 0, 4, 22.	1.8	7