Caroline F Wright

List of Publications by Year
in descending order

77 papers	7,090 citations	101543 36 h-index	79698 73 g-index
101 all docs	101 docs citations	101 times ranked	12672 citing authors

#	Article	IF	Citations
1	The Human Phenotype Ontology project: linking molecular biology and disease through phenotype data. Nucleic Acids Research, 2014, 42, D966-D974.	14.5	698
2	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
3	Paediatric genomics: diagnosing rare disease in children. Nature Reviews Genetics, 2018, 19, 253-268.	16.3	369
4	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
5	Distinct genetic architectures for syndromic and nonsyndromic congenital heart defects identified by exome sequencing. Nature Genetics, 2016, 48, 1060-1065.	21.4	351
6	Evidence for 28 genetic disorders discovered by combining healthcare and research data. Nature, 2020, 586, 757-762.	27.8	343
7	The importance of sequence diversity in the aggregation and evolution of proteins. Nature, 2005, 438, 878-881.	27.8	291
8	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
9	Common genetic variants contribute to risk of rare severe neurodevelopmental disorders. Nature, 2018, 562, 268-271.	27.8	246
10	De novo mutations in regulatory elements in neurodevelopmental disorders. Nature, 2018, 555, 611-616.	27.8	232
11	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
12	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
13	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
14	Quantifying the contribution of recessive coding variation to developmental disorders. Science, 2018, 362, 1161-1164.	12.6	158
15	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
16	Parallel protein-unfolding pathways revealed and mapped. Nature Structural and Molecular Biology, 2003, 10, 658-662.	8.2	153
17	The Deciphering Developmental Disorders (DDD) study. Developmental Medicine and Child Neurology, 2011, 53, 702-703.	2.1	153
18	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

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19	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
20	Flexible and scalable diagnostic filtering of genomic variants using G2P with Ensembl VEP. Nature Communications, 2019, 10, 2373.	12.8	86
21	DECIPHER: web-based, community resource for clinical interpretation of rare variants in developmental disorders. Human Molecular Genetics, 2012, 21, R37-R44.	2.9	74
22	Pathogenicity and selective constraint on variation near splice sites. Genome Research, 2019, 29, 159-170.	5.5	70
23	Review of massively parallel DNA sequencing technologies. The HUGO Journal, 2011, 5, 1-12.	4.1	67
24	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
25	Recommendations for clinical interpretation of variants found in non-coding regions of the genome. Genome Medicine, 2022, 14, .	8.2	65
26	Clinically-relevant postzygotic mosaicism in parents and children with developmental disorders in trio exome sequencing data. Nature Communications, 2019, 10, 2985.	12.8	64
27	Direct-to-consumer genetic testing. BMJ: British Medical Journal, 2019, 367, l5688.	2.3	64
28	Regulating direct-to-consumer genetic tests: What is all the fuss about?. Genetics in Medicine, 2011, 13, 295-300.	2.4	57
29	Assessing performance of pathogenicity predictors using clinically relevant variant datasets. Journal of Medical Genetics, 2021, 58, 547-555.	3.2	57
30	The Gene Curation Coalition: A global effort to harmonize geneâ€“disease evidence resources. Genetics in Medicine, 2022, 24, 1732-1742.	2.4	56
31	Non-Invasive Prenatal Diagnosis Using Cell-Free Fetal DNA Technology: Applications and Implications. Public Health Genomics, 2010, 13, 246-255.	1.0	55
32	De Novo Mutations in EBF3 Cause a Neurodevelopmental Syndrome. American Journal of Human Genetics, 2017, 100, 138-150.	6.2	52
33	Policy challenges of clinical genome sequencing. BMJ, The, 2013, 347, f6845-f6845.	6.0	50
34	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
35	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
36	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

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37	Size of the direct-to-consumer genomic testing market. Genetics in Medicine, 2010, 12, 594.	2.4	38
38	Facilitating Collaboration in Rare Genetic Disorders Through Effective Matchmaking in DECIPHER. Human Mutation, 2015, 36, 941-949.	2.5	38
39	Potential research participants support the return of raw sequence data. Journal of Medical Genetics, 2015, 52, 571-574.	3.2	38
40	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
41	Exome-wide assessment of the functional impact and pathogenicity of multinucleotide mutations. Genome Research, 2019, 29, 1047-1056.	5.5	34
42	Informatics and clinical genome sequencing: opening the black box. Genetics in Medicine, 2013, 15, 165-171.	2.4	33
43	Structural analysis of pathogenic mutations in theDYRK1Agene in patients with developmental disorders. Human Molecular Genetics, 2017, 26, ddw409.	2.9	33
44	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
45	The contribution of X-linked coding variation to severe developmental disorders. Nature Communications, 2021, 12, 627.	12.8	33
46	Expanded universal carrier screening and its implementation within a publicly funded healthcare service. Journal of Community Genetics, 2020, 11, 21-38.	1.2	31
47	Genomic variant sharing: a position statement. Wellcome Open Research, 2019, 4, 22.	1.8	31
48	Thermodynamic Characterisation of Two Transition States Along Parallel Protein Folding Pathways. Journal of Molecular Biology, 2004, 338, 445-451.	4.2	30
49	Strengthening the reporting of genetic risk prediction studies (GRIPS): explanation and elaboration. European Journal of Clinical Investigation, 2011, 41, 1010-1035.	3.4	30
50	Protein structure and phenotypic analysis of pathogenic and population missense variants inSTXBP1. Molecular Genetics & Genomic Medicine, 2017, 5, 495-507.	1.2	29
51	Use of SNP chips to detect rare pathogenic variants: retrospective, population based diagnostic evaluation. BMJ, The, 2021, 372, n214.	6.0	27
52	Biomarkers, Dementia, and Public Health. Annals of the New York Academy of Sciences, 2009, 1180, 11-19.	3.8	26
53	Principle of proportionality in genomic data sharing. Nature Reviews Genetics, 2016, 17, 1-2.	16.3	26
54	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

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55	Returning genome sequences to research participants: Policy and practice. Wellcome Open Research, 2017, 2, 15.	1.8	24
56	No expectation to share incidental findings in genomic research. Lancet, The, 2015, 385, 1289-1290.	13.7	19
57	Empirical research on the ethics of genomic research. American Journal of Medical Genetics, Part A, 2013, 161, 2099-2101.	1.2	17
58	The importance of loop length in the folding of an immunoglobulin domain. Protein Engineering, Design and Selection, 2004, 17, 443-453.	2.1	16
59	Evaluating variants classified as pathogenic in ClinVar in the DDD Study. Genetics in Medicine, 2021, 23, 571-575.	2.4	16
60	Strengthening the reporting of genetic risk prediction studies (GRIPS): explanation and elaboration. European Journal of Epidemiology, 2011, 26, 313-337.	5.7	14
61	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
62	Strengthening the reporting of genetic risk prediction studies (GRIPS): explanation and elaboration. European Journal of Human Genetics, 2011, 19, 615-615.	2.8	12
63	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
64	Strengthening the reporting of Genetic RIsk Prediction Studies (GRIPS): explanation and elaboration. Journal of Clinical Epidemiology, 2011, 64, e1-e22.	5.0	9
65	Risk Prediction Models: A Framework for Assessment. Public Health Genomics, 2012, 15, 98-105.	1.0	8
66	Finding Diagnostically Useful Patterns in Quantitative Phenotypic Data. American Journal of Human Genetics, 2019, 105, 933-946.	6.2	8
67	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
68	When genomic medicine reveals misattributed genetic relationshipsâ€"the debate about disclosure revisited. Genetics in Medicine, 2019, 21, 97-101.	2.4	7
69	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
70	Genomic variant sharing: a position statement. Wellcome Open Research, 0, 4, 22.	1.8	7
71	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
72	A new strategic phase for genomic medicine in UK health services. Genome Medicine, 2009, 1, 93.	8.2	4

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73	Realising the benefits of genetics for health. Lancet, The, 2010, 376, 1370-1371.	13.7	3
74	Direct-to-Consumer Genetic Testing. , 2012, , 215-236.		3
75	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
76	Conceptual issues for screening in the genomic era - time for an update?. Epidemiology Biostatistics and Public Health, 2014, 11, .	0.0	1
77	Quality Issues in the Evaluation and Regulation of Genetic Testing Services: A Public Health Approach. , 2010, , 267-275.		0