Caroline F Wright

List of Publications by Year
in descending order

132 papers	9,229 citations	69737 41 h-index	52210 86 g-index
178 all docs	178 docs citations	178 times ranked	25077 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.	14.0	708
2	Genetic diagnosis of developmental disorders in the DDD study: a scalable analysis of genome-wide research data. Lancet, The, 2015, 385, 1305-1314.	12.1	670
3	Living risk prediction algorithm (QCOVID) for risk of hospital admission and mortality from coronavirus 19 in adults: national derivation and validation cohort study. BMJ, The, 2020, 371, m3731.	7.8	482
4	100,000 Genomes Pilot on Rare-Disease Diagnosis in Health Care â€" Preliminary Report. New England Journal of Medicine, 2021, 385, 1868-1880.	30.1	413
5	Paediatric genomics: diagnosing rare disease in children. Nature Reviews Genetics, 2018, 19, 253-268.	16.7	400
6	Evidence for 28 genetic disorders discovered by combining healthcare and research data. Nature, 2020, 586, 757-762.	36.2	393
7	Distinct genetic architectures for syndromic and nonsyndromic congenital heart defects identified by exome sequencing. Nature Genetics, 2016, 48, 1060-1065.	20.4	368
8	The importance of sequence diversity in the aggregation and evolution of proteins. Nature, 2005, 438, 878-881.	36.2	294
9	Engineering bacteria for diagnostic and therapeutic applications. Nature Reviews Microbiology, 2018, 16, 214-225.	29.2	289
10	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	273
11	Common genetic variants contribute to risk of rare severe neurodevelopmental disorders. Nature, 2018, 562, 268-271.	36.2	258
12	De novo mutations in regulatory elements in neurodevelopmental disorders. Nature, 2018, 555, 611-616.	36.2	242
13	DECIPHER: database for the interpretation of phenotype-linked plausibly pathogenic sequence and copy-number variation. Nucleic Acids Research, 2014, 42, D993-D1000.	14.0	205
14	The use of cell-free fetal nucleic acids in maternal blood for non-invasive prenatal diagnosis. Human Reproduction Update, 2008, 15, 139-151.	12.0	199
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.	6.1	168
16	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.	2.9	165
17	The Deciphering Developmental Disorders (DDD) study. Developmental Medicine and Child Neurology, 2011, 53, 702-703.	2.7	157
18	Parallel protein-unfolding pathways revealed and mapped. Nature Structural and Molecular Biology, 2003, 10, 658-662.	8.1	153

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19	New strategies in sport nutrition to increase exercise performance. Free Radical Biology and Medicine, 2016, 98, 144-158.	4.5	144
20	Discovery of four recessive developmental disorders using probabilistic genotype and phenotype matching among 4,125 families. Nature Genetics, 2015, 47, 1363-1369.	20.4	135
21	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	100
22	Flexible and scalable diagnostic filtering of genomic variants using G2P with Ensembl VEP. Nature Communications, 2019, 10, 2373.	13.2	97
23	Recommendations for clinical interpretation of variants found in non-coding regions of the genome. Genome Medicine, 2022, 14, .	8.5	92
24	Validation of the prognostic value of <scp>MMP</scp>â€7 in idiopathic pulmonary fibrosis. Respirology, 2017, 22, 486-493.	2.9	91
25	Genomic Diagnosis of Rare Pediatric Disease in the United Kingdom and Ireland. New England Journal of Medicine, 2023, 388, 1559-1571.	30.1	76
26	DECIPHER: web-based, community resource for clinical interpretation of rare variants in developmental disorders. Human Molecular Genetics, 2012, 21, R37-R44.	3.0	74
27	Pathogenicity and selective constraint on variation near splice sites. Genome Research, 2019, 29, 159-170.	5.6	71
28	Review of massively parallel DNA sequencing technologies. The HUGO Journal, 2011, 5, 1-12.	0.8	69
29	The Gene Curation Coalition: A global effort to harmonize geneâ€“disease evidence resources. Genetics in Medicine, 2022, 24, 1732-1742.	2.4	69
30	Assessing performance of pathogenicity predictors using clinically relevant variant datasets. Journal of Medical Genetics, 2021, 58, 547-555.	3.6	68
31	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	67
32	Clinically-relevant postzygotic mosaicism in parents and children with developmental disorders in trio exome sequencing data. Nature Communications, 2019, 10, 2985.	13.2	67
33	Regulating direct-to-consumer genetic tests: What is all the fuss about?. Genetics in Medicine, 2011, 13, 295-300.	2.4	58
34	De Novo Mutations in EBF3 Cause a Neurodevelopmental Syndrome. American Journal of Human Genetics, 2017, 100, 138-150.	6.1	56
35	Methodological Encounters with the Phenomenal Kind. Philosophy and Phenomenological Research, 2012, 84, 307-344.	1.1	53
36	KIAA1109 Variants Are Associated with a Severe Disorder of Brain Development and Arthrogryposis. American Journal of Human Genetics, 2018, 102, 116-132.	6.1	49

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37	Social Innovation for Sustainability Transformation and its Diverging Development Paths in Marginalised Rural Areas. Sociologia Ruralis, 2021, 61, 344-371.	3.3	48
38	A sensitive electrochemical sensor for lead based on gold nanoparticles/nitrogen-doped graphene composites functionalized with l-cysteine-modified electrode. Journal of Solid State Electrochemistry, 2016, 20, 327-335.	2.6	44
39	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.	6.1	44
40	Cost analysis and cost-effectiveness of NT-proBNP-guided heart failure specialist care in addition to home-based nurse care. European Journal of Clinical Investigation, 2011, 41, 315-322.	3.4	43
41	Facilitating Collaboration in Rare Genetic Disorders Through Effective Matchmaking in DECIPHER. Human Mutation, 2015, 36, 941-949.	2.8	41
42	Cell-free fetal DNA and RNA in maternal blood: implications for safer antenatal testing. BMJ: British Medical Journal, 2009, 339, b2451-b2451.	5.6	40
43	Potential research participants support the return of raw sequence data. Journal of Medical Genetics, 2015, 52, 571-574.	3.6	40
44	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.	6.1	40
45	Size of the direct-to-consumer genomic testing market. Genetics in Medicine, 2010, 12, 594.	2.4	38
46	Exome-wide assessment of the functional impact and pathogenicity of multinucleotide mutations. Genome Research, 2019, 29, 1047-1056.	5.6	38
47	Expanded universal carrier screening and its implementation within a publicly funded healthcare service. Journal of Community Genetics, 2020, 11, 21-38.	1.3	38
48	The contribution of X-linked coding variation to severe developmental disorders. Nature Communications, 2021, 12, 627.	13.2	36
49	Protein structure and phenotypic analysis of pathogenic and population missense variants inSTXBP1. Molecular Genetics & Genomic Medicine, 2017, 5, 495-507.	1.3	35
50	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.	6.1	35
51	Genomic variant sharing: a position statement. Wellcome Open Research, 2019, 4, 22.	1.9	35
52	ALMA-IMF. Astronomy and Astrophysics, 2022, 662, A8.	5.3	35
53	Informatics and clinical genome sequencing: opening the black box. Genetics in Medicine, 2013, 15, 165-171.	2.4	33
54	The Impact of Tidal Phase on Hurricane Sandy's Flooding Around New York City and Long Island Sound. Journal of Extreme Events, 2014, 01, 1450006.	0.8	33

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55	Structural analysis of pathogenic mutations in theDYRK1Agene in patients with developmental disorders. Human Molecular Genetics, 2017, 26, ddw409.	3.0	33
56	A plant oilâ€containing <scp>pH</scp> 4 emulsion improves epidermal barrier structure and enhances ceramide levels in aged skin. International Journal of Cosmetic Science, 2017, 39, 284-291.	2.7	32
57	Strengthening the reporting of genetic risk prediction studies (GRIPS): explanation and elaboration. European Journal of Clinical Investigation, 2011, 41, 1010-1035.	3.4	31
58	Use of SNP chips to detect rare pathogenic variants: retrospective, population based diagnostic evaluation. BMJ, The, 2021, 372, n214.	7.8	31
59	Thermodynamic Characterisation of Two Transition States Along Parallel Protein Folding Pathways. Journal of Molecular Biology, 2004, 338, 445-451.	4.3	30
60	Principle of proportionality in genomic data sharing. Nature Reviews Genetics, 2016, 17, 1-2.	16.7	27
61	Systematic development and characterization of curcumin-loaded nanogel for topical application. Drug Development and Industrial Pharmacy, 2020, 46, 1443-1457.	2.1	27
62	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.	3.0	26
63	Returning genome sequences to research participants: Policy and practice. Wellcome Open Research, 2017, 2, 15.	1.9	24
64	An enclosed in-gel PCR amplification cassette with multi-target, multi-sample detection for platform molecular diagnostics. Lab on A Chip, 2013, 13, 2576.	6.1	23
65	No expectation to share incidental findings in genomic research. Lancet, The, 2015, 385, 1289-1290.	12.1	20
66	Microwave Radiation as a Pre-Treatment for Standard and Innovative Fragmentation Techniques in Concrete Recycling. Materials, 2019, 12, 488.	3.0	20
67	Exploring Consumersâ€™ Purchase Intention of rPET Bottle-Based Apparel in an Emerging Economy. Journal of Open Innovation: Technology, Market, and Complexity, 2021, 7, 22.	5.4	19
68	Proteinuria and Haematuria are Frequently Present in Patients with Lung Cancer. Nephrology Dialysis Transplantation, 1989, 4, 947-950.	0.8	18
69	Empirical research on the ethics of genomic research. American Journal of Medical Genetics, Part A, 2013, 161, 2099-2101.	1.5	18
70	Evaluating variants classified as pathogenic in ClinVar in the DDD Study. Genetics in Medicine, 2021, 23, 571-575.	2.4	18
71	Search for new physics in top quark production with additional leptons in proton-proton collisions at $$ sqrt{s} $$ = 13 TeV using effective field theory. Journal of High Energy Physics, 2021, 2021, 1.	4.8	17
72	The importance of loop length in the folding of an immunoglobulin domain. Protein Engineering, Design and Selection, 2004, 17, 443-453.	2.4	16

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73	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	16
74	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.	6.1	16
75	Gain-of-function mutations in KCNK3 cause a developmental disorder with sleep apnea. Nature Genetics, 2022, 54, 1534-1543.	20.4	16
76	Experimental beta limit in an average minimum-B tandem mirror. Nuclear Fusion, 1990, 30, 1061-1078.	3.4	14
77	Strengthening the reporting of genetic risk prediction studies (GRIPS): explanation and elaboration. European Journal of Epidemiology, 2011, 26, 313-337.	5.9	14
78	An insight into the protospacer adjacent motif of Streptococcus pyogenes Cas9 with artificially stimulated RNA-guided-Cas9 DNA cleavage flexibility. RSC Advances, 2016, 6, 33514-33522.	3.7	14
79	Evaluating value mediation in patients with chronic low-back pain using virtual reality: contributions for empirical research in Value Sensitive Design. Health and Technology, 2022, 12, 765-778.	3.7	13
80	Strengthening the reporting of genetic risk prediction studies (GRIPS): explanation and elaboration. European Journal of Human Genetics, 2011, 19, 615-615.	2.9	12
81	Penetrance of pathogenic genetic variants associated with premature ovarian insufficiency. Nature Medicine, 2023, 29, 1692-1699.	30.1	12
82	Search for resonances decaying to a pair of Higgs bosons in the b$$ overline{mathrm{b}} $$q$$ overline{mathrm{q}} $$â€™â„“Î½ final state in proton-proton collisions at $$ sqrt{s} $$ = 13 TeV. Journal of High Energy Physics, 2019, 2019, 1.	4.8	11
83	Ectopic activation of GABAB receptors inhibits neurogenesis and metamorphosis in the cnidarian Nematostella vectensis. Nature Ecology and Evolution, 2021, 5, 111-121.	8.0	11
84	Strengthening the reporting of Genetic RIsk Prediction Studies (GRIPS): explanation and elaboration. Journal of Clinical Epidemiology, 2011, 64, e1-e22.	5.0	9
85	When genomic medicine reveals misattributed genetic relationshipsâ€”the debate about disclosure revisited. Genetics in Medicine, 2019, 21, 97-101.	2.4	9
86	Finding Diagnostically Useful Patterns in Quantitative Phenotypic Data. American Journal of Human Genetics, 2019, 105, 933-946.	6.1	9
87	Unreliability of genotyping arrays for detecting very rare variants in human genetic studies: Example from a recent study of MC4R. Cell, 2021, 184, 1651.	27.8	9
88	Delayed Laparoscopic Cholecystectomy for Acute Calculous Cholecystitis: Is it Time for a Change?. World Journal of Surgery, 2017, 41, 1762-1768.	1.4	8
89	Large Copy-Number Variants in UK Biobank Caused by Clonal Hematopoiesis May Confound Penetrance Estimates. American Journal of Human Genetics, 2020, 107, 325-329.	6.1	7
90	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

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91	Genomic variant sharing: a position statement. Wellcome Open Research, 0, 4, 22.	1.9	7
92	ELK1 has a dual activating and repressive role in human embryonic stem cells. Wellcome Open Research, 2019, 4, 41.	1.9	6
93	The effects of ulinastatin on cardiac and hepatic energy metabolism in rats subjected to hypovolemic shock. Journal of Anesthesia, 1990, 4, 40-44.	1.8	5
94	Improving the Assessment of COVID-19â€“Associated Posttraumatic Stress Disorder. JAMA Psychiatry, 2021, 78, 795.	11.4	5
95	Primate-specific ZNF808 is essential for pancreatic development in humans. Nature Genetics, 2023, 55, 2075-2081.	20.4	5
96	van der Waals interactions between the air-bearing surface and a lubricated glass disk: A comparative study. Applied Physics Letters, 2006, 88, 022509.	3.2	4
97	A new strategic phase for genomic medicine in UK health services. Genome Medicine, 2009, 1, 93.	8.5	4
98	Seed germination of the wild banana Musa ornata (Musaceae). Seed Science and Technology, 2014, 42, 16-27.	1.4	4
99	A Parametric study of radial tool actuation in orbital EDM. Advances in Materials and Processing Technologies, 2015, 1, 394-403.	1.4	4
100	Importance of adopting standardized MANE transcripts in clinical reporting. Genetics in Medicine, 2023, 25, 100331.	2.4	4
101	Realising the benefits of genetics for health. Lancet, The, 2010, 376, 1370-1371.	12.1	3
102	Application of Hysteretic Trends in the Preconsolidation Stress of Unsaturated Soils. Geotechnical and Geological Engineering, 2018, 36, 193-207.	1.7	3
103	A multiple mechanism model for measuring extraâ€•and intraparticle mass transport in ion exchange resins. AICHE Journal, 2020, 66, e16825.	3.6	3
104	Direct-to-Consumer Genetic Testing. , 2012, , 215-236.		3
105	Calculating the gross coke yield from satellite batch coking. Coke and Chemistry, 2008, 51, 14-19.	0.4	2
106	Chen Jitong, Les Parisiens peints par un Chinois, and the Literary Self-Fashioning of a Chinese Boulevardier in Fin-de-siÃ¨cle Paris. Esprit Createur, 2016, 56, 90-103.	0.0	2
107	Gait biomechanics: A clinically relevant outcome measure for preclinical research of musculoskeletal trauma. Journal of Orthopaedic Research, 2021, 39, 1139-1151.	2.4	2
108	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.	3.0	2

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109	Evaluation of in silico pathogenicity prediction tools for the classification of small in-frame indels. BMC Medical Genomics, 2023, 16, .	1.5	2
110	Evaluating the use of paralogous protein domains to increase data availability for missense variant classification. Genome Medicine, 2023, 15, .	8.5	2
111	Synthetic Degradable Polyvinyl Alcohol Polymer and Its Blends with Starch and Celluloseâ€"A Comprehensive Overview. Polymers, 2024, 16, 1356.	4.6	2
112	De novo variants in the RNU4-2 snRNA cause a frequent neurodevelopmental syndrome. Nature, 0, , .	36.2	2
113	Flexural testing of carbon FLP composite bars with annular and square cross section. Production Engineering Archives, 2020, 26, 138-143.	2.5	1
114	Genetic modifiers of rare variants in monogenic developmental disorder loci. Nature Genetics, 2024, 56, 861-868.	20.4	1
115	Cuba's International Relations, The Anatomy of a Nationalistic Foreign Policy. By H. Michael Erisman. (Boulder, CO: Westview Press, 1985. Pp. xv + 203. $34.00, cloth; $13.95, paper.). American Political Science Review, 1986, 80, 1396-1397.	4.2	0
116	Quality Measures in Surgical Palliative Care: Adapting Existing Palliative Care Measures to Improve Care for Seriously Ill Surgical Patients. Journal of the American College of Surgeons, 2018, 227, S160.	0.5	0
117	Macular function in severe disbinocular amblyopia. Acta Ophthalmologica, 0, 85, 0-0.	0.3	0
118	Quality Issues in the Evaluation and Regulation of Genetic Testing Services: A Public Health Approach. , 2010, , 267-275.		0
119	An Econometric Analysis of Advertising, Retail Availability, and Sales of a New Brand. Lecture Notes in Economics and Mathematical Systems, 1976, , 371-373.	0.0	0
120	Ein facettenreiches Fach prÃsentiert sich. Der Deutsche Dermatologe, 2020, 68, 170-199.	0.0	0
121	Pentecostal theologyâ€™s problem (Pt 1 2:2): MaÅ¡i ke phepa ke le nosi, selabe se tla le motsaya kgamelo â€“ a Setswana proverb. HTS Teologiese Studies / Theological Studies, 2022, 78, .	0.4	0
122	Remediating sub-Saharan Airline challenges and limitations. Journal of Transport and Supply Chain Management, 0, 16, .	0.8	0
123	Historical origin and meaning of the term â€žglial tumorâ€œ. International Bulletin of Otorhinolaryngology, 2023, 18, 12.	0.0	0
124	Vibration-Based Nondestructive Evaluation of Bio-Printed Constructs Using Phase-Based Motion Estimation. , 2022, , .		0
125	Cyclic Magnetic Activity in KIC7542091 Eclipsing Binary System. Turkish Journal of Astronomy and Astrophysics, 0, , .	0.0	0
126	Prevalence, Risk Factors, and Perinatal Outcomes of Velamentous Umbilical Cord Insertion in Twin Pregnancies: A Single-Center Retrospective Study. Journal of Clinical Medicine, 2024, 13, 1396.	2.5	0

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127	Guidance for estimating penetrance of monogenic disease-causing variants in population cohorts. Nature Genetics, 0, , .	20.4	0
128	FAKTOR YANG MEMPENGARUHI DAN MENINGKATKAN KINERJA PEGAWAI UKM DONAT DI PURI GADING. Journal Science Innovation and Technology (SINTECH), 2024, 4, 13-16.	0.1	0
129	Genetic links between ovarian ageing, cancer risk and de novo mutation rates. Nature, 0, , .	36.2	0
130	Federated analysis of autosomal recessive coding variants in 29,745 developmental disorder patients from diverse populations. Nature Genetics, 0, , .	20.4	0
131	Phenotypic spectrum of dual diagnoses in developmental disorders. American Journal of Human Genetics, 2024, , .	6.1	0
132	Whole-genome sequencing in 333,100 individuals reveals rare non-coding single variant and aggregate associations with height. Nature Communications, 2024, 15, .	13.2	0