Christopher Mark Watson

List of Publications by Year
in descending order

50 papers	1,298 citations	361413 20 h-index	377865 34 g-index
52 all docs	52 docs citations	52 times ranked	2607 citing authors

#	Article	IF	Citations
1	Molecular diagnoses in the congenital malformations caused by ciliopathies cohort of the 100,000 Genomes Project. Journal of Medical Genetics, 2022, 59, 737-747.	3.2	11
2	Longâ€read nanopore DNA sequencing can resolve complex intragenic duplication/deletion variants, providing information to enable preimplantation genetic diagnosis. Prenatal Diagnosis, 2022, 42, 226-232.	2.3	6
3	Unlocking the potential of the <scp>UK</scp> 100,000 Genomes Projectâ€"lessons learned from analysis of the â€œCongenital Malformations caused by Ciliopathiesâ€•cohort. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2022, 190, 5-8.	1.6	2
4	Identification of a novel MAGT1 mutation supports a diagnosis of XMEN disease. Genes and Immunity, 2022, 23, 66-72.	4.1	8
5	Long-read sequencing to resolve the parent of origin of a de novo pathogenicUBE3Avariant. Journal of Medical Genetics, 2022, 59, 1082-1086.	3.2	4
6	Assessing the utility of long-read nanopore sequencing for rapid and efficient characterization of mobile element insertions. Laboratory Investigation, 2021, 101, 442-449.	3.7	9
7	Long-read nanopore sequencing enables accurate confirmation of a recurrent PMS2 insertionâ€"deletion variant located in a region of complex genomic architecture. Cancer Genetics, 2021, 256-257, 122-126.	0.4	4
8	Cas9-based enrichment and single-molecule sequencing for precise characterization of genomic duplications. Laboratory Investigation, 2020, 100, 135-146.	3.7	33
9	Longâ€read nanopore sequencing resolves a TMEM231 gene conversion event causing Meckelâ€"Gruber syndrome. Human Mutation, 2020, 41, 525-531.	2.5	18
10	Clinical utility of NGS diagnosis and disease stratification in a multiethnic primary ciliary dyskinesia cohort. Journal of Medical Genetics, 2020, 57, 322-330.	3.2	50
11	A crowdsourced set of curated structural variants for the human genome. PLoS Computational Biology, 2020, 16, e1007933.	3.2	6
12	A crowdsourced set of curated structural variants for the human genome. , 2020, 16, e1007933.		0
13	A crowdsourced set of curated structural variants for the human genome. , 2020, 16, e1007933.		0
14	A crowdsourced set of curated structural variants for the human genome. , 2020, 16, e1007933.		0
15	A crowdsourced set of curated structural variants for the human genome. , 2020, 16, e1007933.		0
16	An alternative to array-based diagnostics: a prospectively recruited cohort, comparing arrayCGH to next-generation sequencing to evaluate foetal structural abnormalities. Journal of Obstetrics and Gynaecology, 2019, 39, 328-334.	0.9	4
17	RNAseq Supports the Molecular Genetic Diagnosis of Late-Onset ADA Deficiency. Journal of Clinical Immunology, 2019, 39, 270-273.	3.8	0
18	DNAAF1 links heart laterality with the AAA+ ATPase RUVBL1 and ciliary intraflagellar transport. Human Molecular Genetics, 2018, 27, 529-545.	2.9	45

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19	High prevalence of CCDC103 p.His154Pro mutation causing primary ciliary dyskinesia disrupts protein oligomerisation and is associated with normal diagnostic investigations. Thorax, 2018, 73, 157-166.	5.6	63
20	Biallelic Mutations in LRRC56, Encoding a Protein Associated with Intraflagellar Transport, Cause Mucociliary Clearance and Laterality Defects. American Journal of Human Genetics, 2018, 103, 727-739.	6.2	49
21	Increased Sensitivity of Diagnostic Mutation Detection by Re-analysis Incorporating Local Reassembly of Sequence Reads. Molecular Diagnosis and Therapy, 2017, 21, 685-692.	3.8	4
22	Characterization and Genomic Localization of a SMAD4 Processed Pseudogene. Journal of Molecular Diagnostics, 2017, 19, 933-940.	2.8	5
23	m6aViewer: software for the detection, analysis, and visualization of N⁶-methyladenosine peaks from m⁶A-seq/ME-RIP sequencing data. Rna, 2017, 23, 1493-1501.	3.5	34
24	A tubulin alpha 8 mouse knockout model indicates a likely role in spermatogenesis but not in brain development. PLoS ONE, 2017, 12, e0174264.	2.5	23
25	Deficiency of the myogenic factor MyoD causes a perinatally lethal fetal akinesia. Journal of Medical Genetics, 2016, 53, 264-269.	3.2	15
26	Biallelic Mutations in PDE10A Lead to Loss of Striatal PDE10A and a Hyperkinetic Movement Disorder with Onset in Infancy. American Journal of Human Genetics, 2016, 98, 735-743.	6.2	65
27	Enhanced diagnostic yield in Meckel-Gruber and Joubert syndrome through exome sequencing supplemented with split-read mapping. BMC Medical Genetics, 2016, 17, 1.	2.1	67
28	A Chromosome 7 Pericentric Inversion Defined at Single-Nucleotide Resolution Using Diagnostic Whole Genome Sequencing in a Patient with Hand-Foot-Genital Syndrome. PLoS ONE, 2016, 11, e0157075.	2.5	5
29	Gene discovery for motile cilia disorders: mutation spectrum in primary ciliary dyskinesia and discovery of mutations in CCDC151. Cilia, 2015, 4, P30.	1.8	0
30	Identification of a mutation in the ubiquitin-fold modifier 1-specific peptidase 2 gene, UFSP2, in an extended South African family with Beukes hip dysplasia. South African Medical Journal, 2015, 105, 558.	0.6	49
31	Towards a Next-Generation Sequencing Diagnostic Service for Tumour Genotyping: A Comparison of Panels and Platforms. BioMed Research International, 2015, 2015, 1-6.	1.9	12
32	Association Between Missense Mutations in theBBS2Gene and Nonsyndromic Retinitis Pigmentosa. JAMA Ophthalmology, 2015, 133, 312.	2.5	43
33	GeneTIER: prioritization of candidate disease genes using tissue-specific gene expression profiles. Bioinformatics, 2015, 31, 2728-2735.	4.1	25
34	Rapid Detection of Rare Deleterious Variants by Next Generation Sequencing with Optional Microarray SNP Genotype Data. Human Mutation, 2015, 36, 823-830.	2.5	15
35	A distinctive oral phenotype points to FAM 20A mutations not identified by S anger sequencing. Molecular Genetics & Genomic Medicine, 2015, 3, 543-549.	1.2	7
36	OVA: integrating molecular and physical phenotype data from multiple biomedical domain ontologies with variant filtering for enhanced variant prioritization. Bioinformatics, 2015, 31, 3822-3829.	4.1	24

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37	Novel C8orf37 mutations cause retinitis pigmentosa in consanguineous families of Pakistani origin. Molecular Vision, 2015, 21, 236-43.	1.1	10
38	Mutation Screening of Retinal Dystrophy Patients by Targeted Capture from Tagged Pooled DNAs and Next Generation Sequencing. PLoS ONE, 2014, 9, e104281.	2.5	20
39	CCDC151 Mutations Cause Primary Ciliary Dyskinesia by Disruption of the Outer Dynein Arm Docking Complex Formation. American Journal of Human Genetics, 2014, 95, 257-274.	6.2	149
40	Robust Diagnostic Genetic Testing Using Solution Capture Enrichment and a Novel Variantâ€Filtering Interface. Human Mutation, 2014, 35, 434-441.	2.5	38
41	Diagnostic whole genome sequencing and splitâ€read mapping for nucleotide resolution breakpoint identification in CNTNAP2 deficiency syndrome. American Journal of Medical Genetics, Part A, 2014, 164, 2649-2655.	1.2	29
42	Detection of somatic mutations in tumors using unaligned clonal sequencing data. Laboratory Investigation, 2014, 94, 1173-1183.	3.7	2
43	Diagnosis of copy number variation by Illumina next generation sequencing is comparable in performance to oligonucleotide array comparative genomic hybridisation. Genomics, 2013, 102, 174-181.	2.9	48
44	Simple and Efficient Identification of Rare Recessive Pathologically Important Sequence Variants from Next Generation Exome Sequence Data. Human Mutation, 2013, 34, 945-952.	2.5	4
45	Mutation Detection by Clonal Sequencing of PCR Amplicons and Grouped Read Typing is Applicable to Clinical Diagnostics. Human Mutation, 2013, 34, 248-254.	2.5	8
46	Structure of Ubiquitin-fold Modifier 1-specific Protease UfSP2. Journal of Biological Chemistry, 2011, 286, 10248-10257.	3.4	47
47	Concordant Association of Insulin Degrading Enzyme Gene (IDE) Variants with IDE mRNA, AÂŸ, and Alzheimer's Disease. PLoS ONE, 2010, 5, e8764.	2.5	48
48	Largeâ€scale analysis of association between GDF5 and FRZB variants and osteoarthritis of the hip, knee, and hand. Arthritis and Rheumatism, 2009, 60, 1710-1721.	6.7	181
49	An enhanced method for targeted next generation sequencing copy number variant detection using ExomeDepth. Wellcome Open Research, 0, 2, 49.	1.8	4
50	Uncovering the burden of hidden ciliopathies in the 100 000 Genomes Project: a reverse phenotyping approach. Journal of Medical Genetics, 0, , jmedgenet-2022-108476.	3.2	3