# Christopher Mark Watson

List of Publications by Year
in descending order



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|---|---|---|---|
| 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 | 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 |
| 2 | 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 |
| 3 | 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 |
| 4 | 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 |
| 5 | 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 |
| 6 | 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 |
| 7 | 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 |
| 8 | 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 |
| 9 | Concordant Association of Insulin Degrading Enzyme Gene (IDE) Variants with IDE mRNA, AÃŸ, and Alzheimer's Disease. PLoS ONE, 2010, 5, e8764. | 2.5 | 48 |
| 10 | 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 |
| 11 | Structure of Ubiquitin-fold Modifier 1-specific Protease UfSP2. Journal of Biological Chemistry, 2011, 286, 10248-10257. | 3.4 | 47 |
| 12 | DNAAF1 links heart laterality with the AAA+ ATPase RUVBL1 and ciliary intraflagellar transport. Human Molecular Genetics, 2018, 27, 529-545. | 2.9 | 45 |
| 13 | Association Between Missense Mutations in theBBS2Gene and Nonsyndromic Retinitis Pigmentosa. JAMA Ophthalmology, 2015, 133, 312. | 2.5 | 43 |
| 14 | Robust Diagnostic Genetic Testing Using Solution Capture Enrichment and a Novel Variantâ€Filtering Interface. Human Mutation, 2014, 35, 434-441. | 2.5 | 38 |
| 15 | m6aViewer: software for the detection, analysis, and visualization of N<sup>6</sup>-methyladenosine peaks from m<sup>6</sup>A-seq/ME-RIP sequencing data. Rna, 2017, 23, 1493-1501. | 3.5 | 34 |
| 16 | Cas9-based enrichment and single-molecule sequencing for precise characterization of genomic duplications. Laboratory Investigation, 2020, 100, 135-146. | 3.7 | 33 |
| 17 | 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 |
| 18 | GeneTIER: prioritization of candidate disease genes using tissue-specific gene expression profiles. Bioinformatics, 2015, 31, 2728-2735. | 4.1 | 25 |

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|---|---|---|---|
| 19 | 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 |
| 20 | 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 |
| 21 | 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 |
| 22 | Longâ€read nanopore sequencing resolves a TMEM231 gene conversion event causing Meckelâ€“Gruber syndrome. Human Mutation, 2020, 41, 525-531. | 2.5 | 18 |
| 23 | 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 |
| 24 | Deficiency of the myogenic factor MyoD causes a perinatally lethal fetal akinesia. Journal of Medical Genetics, 2016, 53, 264-269. | 3.2 | 15 |
| 25 | 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 |
| 26 | 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 |
| 27 | Novel C8orf37 mutations cause retinitis pigmentosa in consanguineous families of Pakistani origin. Molecular Vision, 2015, 21, 236-43. | 1.1 | 10 |
| 28 | 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 |
| 29 | 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 |
| 30 | Identification of a novel MAGT1 mutation supports a diagnosis of XMEN disease. Genes and Immunity, 2022, 23, 66-72. | 4.1 | 8 |
| 31 | 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 |
| 32 | A crowdsourced set of curated structural variants for the human genome. PLoS Computational Biology, 2020, 16, e1007933. | 3.2 | 6 |
| 33 | 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 |
| 34 | Characterization and Genomic Localization of a SMAD4 Processed Pseudogene. Journal of Molecular Diagnostics, 2017, 19, 933-940. | 2.8 | 5 |
| 35 | 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 |
| 36 | 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 |

| # | Article | IF | Citations |
|---|---|---|---|
| 37 | 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 |
| 38 | 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 |
| 39 | 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 |
| 40 | An enhanced method for targeted next generation sequencing copy number variant detection using ExomeDepth. Wellcome Open Research, 0, 2, 49. | 1.8 | 4 |
| 41 | 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 |
| 42 | 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 |
| 43 | Detection of somatic mutations in tumors using unaligned clonal sequencing data. Laboratory Investigation, 2014, 94, 1173-1183. | 3.7 | 2 |
| 44 | 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 |
| 45 | 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 |
| 46 | RNAseq Supports the Molecular Genetic Diagnosis of Late-Onset ADA Deficiency. Journal of Clinical Immunology, 2019, 39, 270-273. | 3.8 | 0 |
| 47 | A crowdsourced set of curated structural variants for the human genome. , 2020, 16, e1007933. | | 0 |
| 48 | A crowdsourced set of curated structural variants for the human genome. , 2020, 16, e1007933. | | 0 |
| 49 | A crowdsourced set of curated structural variants for the human genome. , 2020, 16, e1007933. | | 0 |
| 50 | A crowdsourced set of curated structural variants for the human genome. , 2020, 16, e1007933. | | 0 |