# Paul N Schofield

List of Publications by Year
in descending order



| | | | |
|---|---|---|---|
| 133 papers | 5,982 citations | 87723 38 h-index | 88477 70 g-index |
| 159 all docs | 159 docs citations | 159 times ranked | 9080 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. | 6.5 | 698 |
| 2 | IGF2 is parentally imprinted during human embryogenesis and in the Beckwithâ€“Wiedemann syndrome. Nature Genetics, 1993, 4, 94-97. | 9.4 | 292 |
| 3 | The mammalian gene function resource: the international knockout mouse consortium. Mammalian Genome, 2012, 23, 580-586. | 1.0 | 292 |
| 4 | Molecular subtypes and phenotypic expression of Beckwithâ€“Wiedemann syndrome. European Journal of Human Genetics, 2005, 13, 1025-1032. | 1.4 | 284 |
| 5 | Prepublication data sharing. Nature, 2009, 461, 168-170. | 13.7 | 243 |
| 6 | The Human Phenotype Ontology: Semantic Unification of Common and Rare Disease. American Journal of Human Genetics, 2015, 97, 111-124. | 2.6 | 203 |
| 7 | The role of ontologies in biological and biomedical research: a functional perspective. Briefings in Bioinformatics, 2015, 16, 1069-1080. | 3.2 | 199 |
| 8 | PhenomeNET: a whole-phenome approach to disease gene discovery. Nucleic Acids Research, 2011, 39, e119-e119. | 6.5 | 195 |
| 9 | Deletions of chromosomal regulatory boundaries are associated with congenital disease. Genome Biology, 2014, 15, 423. | 3.8 | 144 |
| 10 | Post-publication sharing of data and tools. Nature, 2009, 461, 171-173. | 13.7 | 142 |
| 11 | A mouse homoeo box gene is expressed during embryogenesis and in adult kidney. Nature, 1985, 317, 745-748. | 13.7 | 128 |
| 12 | A decadal view of biodiversity informatics: challenges and priorities. BMC Ecology, 2013, 13, 16. | 3.0 | 110 |
| 13 | Imprinting of IGF2 and H19: lack of reciprocity in sporadic Beckwith- Wiedemann syndrome. Human Molecular Genetics, 1997, 6, 1543-1548. | 1.4 | 100 |
| 14 | Analysis of the human diseasome using phenotype similarity between common, genetic and infectious diseases. Scientific Reports, 2015, 5, 10888. | 1.6 | 85 |
| 15 | Frequent RASSF1A tumour suppressor gene promoter methylation in Wilms' tumour and colorectal cancer. Oncogene, 2002, 21, 7277-7282. | 2.6 | 82 |
| 16 | Radiation-induced genomic instability is associated with DNA methylation changes in cultured human keratinocytes. Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis, 2006, 597, 87-97. | 0.4 | 78 |
| 17 | The mouse as a model for understanding chronic diseases of aging: the histopathologic basis of aging in inbred mice. Pathobiology of Aging & Age Related Diseases, 2011, 1, 7179. | 1.1 | 78 |
| 18 | The Units Ontology: a tool for integrating units of measurement in science. Database: the Journal of Biological Databases and Curation, 2012, 2012, bas033-bas033. | 1.4 | 78 |

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|---|---|---|---|
| 19 | Abnormal skeletal and cardiac development, cardiomyopathy, muscle atrophy and cataracts in mice with a targeted disruption of the Nov (Ccn3) gene. BMC Developmental Biology, 2008, 8, 18. | 2.1 | 76 |
| 20 | Construction and accessibility of a cross-species phenotype ontology along with gene annotations for biomedical research. F1000Research, 2013, 2, 30. | 0.8 | 72 |
| 21 | Aber-OWL: a framework for ontology-based data access in biology. BMC Bioinformatics, 2015, 16, 26. | 1.2 | 68 |
| 22 | Entity/quality-based logical definitions for the human skeletal phenome using PATO. , 2009, 2009, 7069-72. | | 67 |
| 23 | The anatomy of phenotype ontologies: principles, properties and applications. Briefings in Bioinformatics, 2018, 19, 1008-1021. | 3.2 | 66 |
| 24 | Construction and accessibility of a cross-species phenotype ontology along with gene annotations for biomedical research. F1000Research, 2013, 2, 30. | 0.8 | 64 |
| 25 | Mouse genetic and phenotypic resources for human genetics. Human Mutation, 2012, 33, 826-836. | 1.1 | 58 |
| 26 | How is the mouse segmented?. Trends in Genetics, 1985, 1, 67-74. | 2.9 | 56 |
| 27 | Glucose regulates preproinsulin messenger RNA levels in a clonal cell line of simian virus 40-transformed B cells. FEBS Letters, 1987, 213, 149-154. | 1.3 | 53 |
| 28 | Insulin-like growth factor (IGF)-I, -II and IGF binding protein-2 (IGFBP-2) in the plasma of children with Wilms' tumour. European Journal of Cancer, 1993, 29, 1973-1977. | 1.3 | 53 |
| 29 | MouseFinder: Candidate disease genes from mouse phenotype data. Human Mutation, 2012, 33, 858-866. | 1.1 | 53 |
| 30 | Sustaining the Data and Bioresource Commons. Science, 2010, 330, 592-593. | 6.0 | 52 |
| 31 | Pathbase: a database of mutant mouse pathology. Nucleic Acids Research, 2004, 32, 512D-515. | 6.5 | 49 |
| 32 | DeepPVP: phenotype-based prioritization of causative variants using deep learning. BMC Bioinformatics, 2019, 20, 65. | 1.2 | 49 |
| 33 | Integration of mouse phenome data resources. Mammalian Genome, 2007, 18, 157-163. | 1.0 | 44 |
| 34 | Phenotypic overlap in the contribution of individual genes to CNV pathogenicity revealed by cross-species computational analysis of single-gene mutations in humans, mice and zebrafish. DMM Disease Models and Mechanisms, 2013, 6, 358-72. | 1.2 | 43 |
| 35 | Integrating mouse anatomy and pathology ontologies into a phenotyping database: Tools for data capture and training. Mammalian Genome, 2008, 19, 413-419. | 1.0 | 42 |
| 36 | Genomic imprinting and cancer; new paradigms in the genetics of neoplasia. Toxicology Letters, 2001, 120, 151-160. | 0.4 | 41 |

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|---|---|---|---|
| 37 | Regulation and specificity of glucose-stimulated insulin gene expression in human islets of Langerhans. FEBS Letters, 1987, 223, 131-137. | 1.3 | 40 |
| 38 | Altered Expression of novH Is Associated with Human Adrenocortical Tumorigenesis. Journal of Clinical Endocrinology and Metabolism, 2001, 86, 3929-3940. | 1.8 | 40 |
| 39 | Phenotype ontologies for mouse and man: bridging the semantic gap. DMM Disease Models and Mechanisms, 2010, 3, 281-289. | 1.2 | 39 |
| 40 | Interoperability between Biomedical Ontologies through Relation Expansion, Upper-Level Ontologies and Automatic Reasoning. PLoS ONE, 2011, 6, e22006. | 1.1 | 38 |
| 41 | Mouse model phenotypes provide information about human drug targets. Bioinformatics, 2014, 30, 719-725. | 1.8 | 38 |
| 42 | The GA4GH Phenopacket schema defines a computable representation of clinical data. Nature Biotechnology, 2022, 40, 817-820. | 9.4 | 38 |
| 43 | Reproducibility of histopathological findings in experimental pathology of the mouse: a sorry tail. Lab Animal, 2017, 46, 146-151. | 0.2 | 36 |
| 44 | A common layer of interoperability for biomedical ontologies based on OWL EL. Bioinformatics, 2011, 27, 1001-1008. | 1.8 | 35 |
| 45 | Towards BioDBcore: a community-defined information specification for biological databases. Nucleic Acids Research, 2011, 39, D7-D10. | 6.5 | 32 |
| 46 | Exploring the elephant: histopathology in high-throughput phenotyping of mutant mice. DMM Disease Models and Mechanisms, 2012, 5, 19-25. | 1.2 | 32 |
| 47 | The mouse pathology ontology, MPATH; structure and applications. Journal of Biomedical Semantics, 2013, 4, 18. | 0.9 | 32 |
| 48 | The Neurobehavior Ontology. International Review of Neurobiology, 2012, 103, 69-87. | 0.9 | 31 |
| 49 | Towards BioDBcore: a community-defined information specification for biological databases. Database: the Journal of Biological Databases and Curation, 2011, 2011, baq027-baq027. | 1.4 | 30 |
| 50 | Improving ontologies by automatic reasoning and evaluation of logical definitions. BMC Bioinformatics, 2011, 12, 418. | 1.2 | 29 |
| 51 | The role of insulin-like growth factors and IGF-binding proteins in the physiological and pathological processes of the kidney. Vigiliae Christianae, 1992, 62, 207-220. | 0.1 | 28 |
| 52 | Semantic prioritization of novel causative genomic variants. PLoS Computational Biology, 2017, 13, e1005500. | 1.5 | 28 |
| 53 | Integrating phenotype ontologies with PhenomeNET. Journal of Biomedical Semantics, 2017, 8, 58. | 0.9 | 28 |
| 54 | Evolving paradigms for the biological response to low dose ionizing radiation; the role of epigenetics. International Journal of Radiation Biology, 2018, 94, 769-781. | 1.0 | 28 |

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|---|---|---|---|
| 55 | Models for financial sustainability of biological databases and resources. Database: the Journal of Biological Databases and Curation, 2009, 2009, bap017-bap017. | 1.4 | 27 |
| 56 | One Medicine, One Pathology, and the One Health concept. Journal of the American Veterinary Medical Association, 2009, 234, 1530-1531. | 0.2 | 26 |
| 57 | Patterns, puzzles and paradigms: the riddle of the homeobox. Trends in Neurosciences, 1987, 10, 3-6. | 4.2 | 25 |
| 58 | Living inside the box: environmental effects on mouse models of human disease. DMM Disease Models and Mechanisms, 2018, 11, . | 1.2 | 25 |
| 59 | Establishing the Japan-Store house of animal radiobiology experiments (J-SHARE), a large-scale necropsy and histopathology archive providing international access to important radiobiology data. International Journal of Radiation Biology, 2019, 95, 1372-1377. | 1.0 | 25 |
| 60 | Genomic Structure and Chromosomal Mapping of the MousenovGene. Genomics, 1996, 38, 425-428. | 1.3 | 23 |
| 61 | Solutions for data integration in functional genomics: a critical assessment and case study. Briefings in Bioinformatics, 2008, 9, 532-544. | 3.2 | 23 |
| 62 | Pathbase: a new reference resource and database for laboratory mouse pathology. Radiation Protection Dosimetry, 2004, 112, 525-528. | 0.4 | 21 |
| 63 | Mouse, man, and meaning: bridging the semantics of mouse phenotype and human disease. Mammalian Genome, 2009, 20, 457-461. | 1.0 | 21 |
| 64 | XGAP: a uniform and extensible data model and software platform for genotype and phenotype experiments. Genome Biology, 2010, 11, R27. | 13.9 | 20 |
| 65 | The Effects of Fibroblast Growth Factors in Long-Term Primary Culture of Dystrophic (MDX) Mouse Muscle Myoblasts. Experimental Cell Research, 1994, 210, 86-93. | 1.2 | 19 |
| 66 | Computational tools for comparative phenomics: the role and promise of ontologies. Mammalian Genome, 2012, 23, 669-679. | 1.0 | 19 |
| 67 | Systematic Analysis of Experimental Phenotype Data Reveals Gene Functions. PLoS ONE, 2013, 8, e60847. | 1.1 | 19 |
| 68 | Analyzing gene expression data in mice with the Neuro Behavior Ontology. Mammalian Genome, 2014, 25, 32-40. | 1.0 | 19 |
| 69 | New approaches to the representation and analysis of phenotype knowledge in human diseases and their animal models. Briefings in Functional Genomics, 2011, 10, 258-265. | 1.3 | 18 |
| 70 | Linking PharmGKB to phenotype studies and animal models of disease for drug repurposing. Pacific Symposium on Biocomputing Pacific Symposium on Biocomputing, 2012, , 388-99. | 0.7 | 18 |
| 71 | PRIME importance of pathology expertise. Nature Biotechnology, 2009, 27, 24-25. | 9.4 | 17 |
| 72 | Pathology of the Laboratory Mouse. Toxicologic Pathology, 2011, 39, 559-562. | 0.9 | 17 |

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|---|---|---|---|
| 73 | Inbred mouse strains reveal biomarkers that are proâ€longevity, antilongevity or role switching. Aging Cell, 2014, 13, 729-738. | 3.0 | 17 |
| 74 | From tangled banks to toxic bunnies; a reflection on the issues involved in developing an ecosystem approach for environmental radiation protection. International Journal of Radiation Biology, 2022, 98, 1185-1200. | 1.0 | 17 |
| 75 | STATE COMPLEXITY OF ADDITIVE WEIGHTED FINITE AUTOMATA. International Journal of Foundations of Computer Science, 2007, 18, 1407-1416. | 0.8 | 16 |
| 76 | An integrative, translational approach to understanding rare and orphan genetically based diseases. Interface Focus, 2013, 3, 20120055. | 1.5 | 16 |
| 77 | Big data in radiation biology and epidemiology; an overview of the historical and contemporary landscape of data and biomaterial archives. International Journal of Radiation Biology, 2019, 95, 861-878. | 1.0 | 16 |
| 78 | Ontology-based prediction of cancer driver genes. Scientific Reports, 2019, 9, 17405. | 1.6 | 16 |
| 79 | Concentration-dependent modulation of basic fibroblast growth factor action on multiplication and locomotion of human teratocarcinoma cells. FEBS Letters, 1992, 298, 154-158. | 1.3 | 15 |
| 80 | The Cinderella Effect: Searching for the Best Fit between Mouse Models and Human Diseases. Journal of Investigative Dermatology, 2013, 133, 2509-2513. | 0.3 | 15 |
| 81 | When a duck is not a duck; a new interdisciplinary synthesis for environmental radiation protection. Environmental Research, 2018, 162, 318-324. | 3.7 | 15 |
| 82 | A Review of Current Standards and the Evolution of Histopathology Nomenclature for Laboratory Animals. ILAR Journal, 2018, 59, 29-39. | 1.8 | 15 |
| 83 | Using AberOWL for fast and scalable reasoning over BioPortal ontologies. Journal of Biomedical Semantics, 2016, 7, 49. | 0.9 | 14 |
| 84 | Systematic screening for skin, hair, and nail abnormalities in a large-scale knockout mouse program. PLoS ONE, 2017, 12, e0180682. | 1.1 | 14 |
| 85 | Dsprul: A spontaneous mouse mutation in desmoplakin as a model of Carvajal-Huerta syndrome. Experimental and Molecular Pathology, 2015, 98, 164-172. | 0.9 | 13 |
| 86 | Mouse genome-wide association study identifies polymorphisms on chromosomes 4, 11, and 15 for age-related cardiac fibrosis. Mammalian Genome, 2016, 27, 179-190. | 1.0 | 13 |
| 87 | PathoPhenoDB, linking human pathogens to their phenotypes in support of infectious disease research. Scientific Data, 2019, 6, 79. | 2.4 | 13 |
| 88 | Quantitative evaluation of ontology design patterns for combining pathology and anatomy ontologies. Scientific Reports, 2019, 9, 4025. | 1.6 | 13 |
| 89 | A Mouse by Any Other Name â€¦. Journal of Investigative Dermatology, 2009, 129, 1599-1601. | 0.3 | 12 |
| 90 | Finding and sharing: new approaches to registries of databases and services for the biomedical sciences. Database: the Journal of Biological Databases and Curation, 2010, 2010, baq014-baq014. | 1.4 | 12 |

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|---|---|---|---|
| 91 | Differentiation associated modulation of K-FGF expression in a human teratocarcinoma cell line and in primary germ cell tumours. FEBS Letters, 1991, 280, 8-10. | 1.3 | 11 |
| 92 | Living Long and Well: Prospects for a Personalized Approach to the Medicine of Ageing. Gerontology, 2016, 62, 409-416. | 1.4 | 11 |
| 93 | Genetic determinants of fibro-osseous lesions in aged inbred mice. Experimental and Molecular Pathology, 2016, 100, 92-100. | 0.9 | 10 |
| 94 | Bio-acoustic signaling; exploring the potential of sound as a mediator of low-dose radiation and stress responses in the environment. International Journal of Radiation Biology, 2022, 98, 1083-1097. | 1.0 | 10 |
| 95 | LINKING PHARMGKB TO PHENOTYPE STUDIES AND ANIMAL MODELS OF DISEASE FOR DRUG REPURPOSING. , 2011, , . | | 9 |
| 96 | Integration of global resources for human genetic variation and disease. Human Mutation, 2012, 33, 813-816. | 1.1 | 9 |
| 97 | DDIEM: drug database for inborn errors of metabolism. Orphanet Journal of Rare Diseases, 2020, 15, 146. | 1.2 | 9 |
| 98 | Similarity-based search of model organism, disease and drug effect phenotypes. Journal of Biomedical Semantics, 2015, 6, 6. | 0.9 | 8 |
| 99 | Show and tell: disclosure and data sharing in experimental pathology. DMM Disease Models and Mechanisms, 2016, 9, 601-605. | 1.2 | 8 |
| 100 | DermO; an ontology for the description of dermatologic disease. Journal of Biomedical Semantics, 2016, 7, 38. | 0.9 | 8 |
| 101 | OligoPVP: Phenotype-driven analysis of individual genomic information to prioritize oligogenic disease variants. Scientific Reports, 2018, 8, 14681. | 1.6 | 8 |
| 102 | Nail abnormalities identified in an ageing study of 30 inbred mouse strains. Experimental Dermatology, 2019, 28, 383-390. | 1.4 | 8 |
| 103 | CASIMIR: Coordination and Sustainability of International Mouse Informatics Resources. , 2008, , . | | 7 |
| 104 | Archiving lessons from radiobiology. Nature, 2010, 468, 634-634. | 13.7 | 7 |
| 105 | Identifying mouse models for skin cancer using the Mouse Tumor Biology Database. Experimental Dermatology, 2014, 23, 761-763. | 1.4 | 7 |
| 106 | Anatomy ontologies and potential users: bridging the gap. Journal of Biomedical Semantics, 2011, 2, S3. | 0.9 | 6 |
| 107 | Diversity of Spontaneous Neoplasms in Commonly Used Inbred Strains of Laboratory Mice. , 2012, , 411-426. | | 6 |
| 108 | Linking common human diseases to their phenotypes; development of a resource for human phenomics. Journal of Biomedical Semantics, 2021, 12, 17. | 0.9 | 6 |

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| 109 | Developmental regulation of insulin like growth factor II expression in the horse.. Cell Biology International, 1993, 17, 603-608. | 1.4 | 5 |
| 110 | Ontology-based validation and identification of regulatory phenotypes. Bioinformatics, 2018, 34, i857-i865. | 1.8 | 5 |
| 111 | Radiation, Oxidative Stress and Senescence; The Vascular Endothelial Cell as a Common Target. NATO Science for Peace and Security Series C: Environmental Security, 2007, , 325-334. | 0.1 | 5 |
| 112 | Contribution of model organism phenotypes to the computational identification of human disease genes. DMM Disease Models and Mechanisms, 2022, 15, . | 1.2 | 5 |
| 113 | Best behaviour? Ontologies and the formal description of animal behaviour. Mammalian Genome, 2015, 26, 540-547. | 1.0 | 4 |
| 114 | Growth Factor Synthesis By a Human Teratocarcinoma Cell Line: Implications for Autocrine Growth in the Human Embryo?. , 1990, , 49-59. | | 4 |
| 115 | GROWH FACTORS IN EARLY EMBRYOGENESIS. Reproduction in Domestic Animals, 1993, 28, 176-181. | 0.6 | 3 |
| 116 | Towards a Disease Ontology. Computational Biology, 2008, , 119-130. | 0.1 | 3 |
| 117 | Towards the integration of mouse databases - definition and implementation of solutions to two use-cases in mouse functional genomics. BMC Research Notes, 2010, 3, 16. | 0.6 | 3 |
| 118 | Mouse Resource Browser–a database of mouse databases. Database: the Journal of Biological Databases and Curation, 2010, 2010, baq010-baq010. | 1.4 | 3 |
| 119 | The Informatics of Developmental Phenotypes. , 2016, , 307-318. | | 3 |
| 120 | Genome wide conditional mouse knockout resources. Drug Discovery Today: Disease Models, 2016, 20, 3-12. | 1.2 | 3 |
| 121 | Hyaline Arteriolosclerosis in 30 Strains of Aged Inbred Mice. Veterinary Pathology, 2019, 56, 799-806. | 0.8 | 3 |
| 122 | SURVEY ON DATA MANAGEMENT IN RADIATION PROTECTION RESEARCH. Radiation Protection Dosimetry, 2019, 183, 233-236. | 0.4 | 3 |
| 123 | Multi-faceted semantic clustering with text-derived phenotypes. Computers in Biology and Medicine, 2021, 138, 104904. | 3.9 | 3 |
| 124 | Excavating the Genome: Large-Scale Mutagenesis Screening for the Discovery of New Mouse Models. Journal of Investigative Dermatology Symposium Proceedings, 2015, 17, 27-29. | 0.8 | 2 |
| 125 | Datamining with Ontologies. Methods in Molecular Biology, 2016, 1415, 385-397. | 0.4 | 2 |
| 126 | PATHBIO: an international training program for precision mouse phenotyping. Mammalian Genome, 2020, 31, 49-53. | 1.0 | 2 |

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|---|---|---|---|
| 127 | The Mouse Resource Browser (MRB) - A near-complete registry of mouse resources. , 2008, , . | | 1 |
| 128 | Digital preservation - financial sustainability of biological data and material resources. , 2008, , . | | 1 |
| 129 | Towards dynamic database infrastructures for mouse genetics. , 2008, , . | | 1 |
| 130 | Molecular control of muscle development: specification, determination and differentiation in the amniote embryo. Seminars in Fetal and Neonatal Medicine, 1999, 4, 79-91. | 2.8 | 0 |
| 131 | Experiences with Aber-OWL, an Ontology Repository with OWL EL Reasoning. Lecture Notes in Computer Science, 2016, , 81-86. | 1.0 | 0 |
| 132 | FAIRing the radiation science commons. BIO Web of Conferences, 2019, 14, 08002. | 0.1 | 0 |
| 133 | Exploring Sentiment as a Potential Indicator of Bias in Disease Ontologies. , 2021, , . | | 0 |