Damian Smedley

List of Publications by Year
in descending order

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93 papers	11,208 citations	57758 44 h-index	49909 87 g-index
110 all docs	110 docs citations	110 times ranked	17716 citing authors

#	Article	IF	Citations
1	Identifying genetic determinants of inflammatory pain in mice using a large-scale gene-targeted screen. Pain, 2022, 163, 1139-1157.	4.2	4
2	Extensive identification of genes involved in congenital and structural heart disorders and cardiomyopathy. , 2022, 1, 157-173.		22
3	The RDâ€Connect Genomeâ€Phenome Analysis Platform: Accelerating diagnosis, research, and gene discovery for rare diseases. Human Mutation, 2022, , .	2.5	18
4	Phenotypeâ€driven approaches to enhance variant prioritization and diagnosis of rare disease. Human Mutation, 2022, 43, 1071-1081.	2.5	17
5	The Clinical Variant Analysis Tool: Analyzing the evidence supporting reported genomic variation in clinical practice. Genetics in Medicine, 2022, 24, 1512-1522.	2.4	4
6	Evaluation of phenotype-driven gene prioritization methods for Mendelian diseases. Briefings in Bioinformatics, 2022, 23, .	6.5	6
7	SvAnna: efficient and accurate pathogenicity prediction of coding and regulatory structural variants in long-read genome sequencing. Genome Medicine, 2022, 14, 44.	8.2	7
8	A gene-to-patient approach uplifts novel disease gene discovery and identifies 18 putative novel disease genes. Genetics in Medicine, 2022, 24, 1697-1707.	2.4	14
9	A basement membrane discovery pipeline uncovers network complexity, regulators, and human disease associations. Science Advances, 2022, 8, eabn2265.	10.3	76
10	The Human Phenotype Ontology in 2021. Nucleic Acids Research, 2021, 49, D1207-D1217.	14.5	652
11	Heterozygous ANKRD17 loss-of-function variants cause a syndrome with intellectual disability, speech delay, and dysmorphism. American Journal of Human Genetics, 2021, 108, 1138-1150.	6.2	17
12	Pathogenic SPTBN1 variants cause an autosomal dominant neurodevelopmental syndrome. Nature Genetics, 2021, 53, 1006-1021.	21.4	44
13	Diffusion enables integration of heterogeneous data and user-driven learning in a desktop knowledge-base. PLoS Computational Biology, 2021, 17, e1009283.	3.2	4
14	Interpretable prioritization of splice variants in diagnostic next-generation sequencing. American Journal of Human Genetics, 2021, 108, 1564-1577.	6.2	36
15	Dimensional reduction of phenotypes from 53Â000 mouse models reveals a diverse landscape of gene function. Bioinformatics Advances, 2021, 1, vbab026.	2.4	0
16	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
17	Soft windowing application to improve analysis of high-throughput phenotyping data. Bioinformatics, 2020, 36, 1492-1500.	4.1	9
18	The Monarch Initiative in 2019: an integrative data and analytic platform connecting phenotypes to genotypes across species. Nucleic Acids Research, 2020, 48, D704-D715.	14.5	178

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19	Identification of UBAP1 mutations in juvenile hereditary spastic paraplegia in the 100,000 Genomes Project. European Journal of Human Genetics, 2020, 28, 1763-1768.	2.8	9
20	Interpretable Clinical Genomics with a Likelihood Ratio Paradigm. American Journal of Human Genetics, 2020, 107, 403-417.	6.2	56
21	Incremental data integration for tracking genotype-disease associations. PLoS Computational Biology, 2020, 16, e1007586.	3.2	7
22	High-throughput discovery of genetic determinants of circadian misalignment. PLoS Genetics, 2020, 16, e1008577.	3.5	10
23	The Deep Genome Project. Genome Biology, 2020, 21, 18.	8.8	30
24	An Improved Phenotype-Driven Tool for Rare Mendelian Variant Prioritization: Benchmarking Exomiser on Real Patient Whole-Exome Data. Genes, 2020, 11, 460.	2.4	42
25	Human and mouse essentiality screens as a resource for disease gene discovery. Nature Communications, 2020, 11, 655.	12.8	64
26	OpenStats: A robust and scalable software package for reproducible analysis of high-throughput phenotypic data. PLoS ONE, 2020, 15, e0242933.	2.5	12
27	High-throughput discovery of genetic determinants of circadian misalignment. , 2020, 16, e1008577.		0
28	High-throughput discovery of genetic determinants of circadian misalignment. , 2020, 16, e1008577.		0
29	High-throughput discovery of genetic determinants of circadian misalignment. , 2020, 16, e1008577.		0
30	High-throughput discovery of genetic determinants of circadian misalignment. , 2020, 16, e1008577.		0
31	Encoding Clinical Data with the Human Phenotype Ontology for Computational Differential Diagnostics. Current Protocols in Human Genetics, 2019, 103, e92.	3.5	29
32	PanelApp crowdsources expert knowledge to establish consensus diagnostic gene panels. Nature Genetics, 2019, 51, 1560-1565.	21.4	294
33	New models for human disease from the International Mouse Phenotyping Consortium. Mammalian Genome, 2019, 30, 143-150.	2.2	57
34	Expansion of the Human Phenotype Ontology (HPO) knowledge base and resources. Nucleic Acids Research, 2019, 47, D1018-D1027.	14.5	539
35	The 100â€‰000 Genomes Project: bringing whole genome sequencing to the NHS. BMJ: British Medical Journal, 2018, 361, k1687.	2.3	312
36	High-throughput mouse phenomics for characterizing mammalian gene function. Nature Reviews Genetics, 2018, 19, 357-370.	16.3	78

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37	Identification of genes required for eye development by high-throughput screening of mouse knockouts. Communications Biology, 2018, 1, 236.	4.4	37
38	matchbox: An open-source tool for patient matching via the Matchmaker Exchange. Human Mutation, 2018, 39, 1827-1834.	2.5	20
39	The International Mouse Phenotyping Consortium (IMPC): a functional catalogue of the mammalian genome that informs conservation. Conservation Genetics, 2018, 19, 995-1005.	1.5	82
40	The Human Phenotype Ontology in 2017. Nucleic Acids Research, 2017, 45, D865-D876.	14.5	699
41	The Monarch Initiative: an integrative data and analytic platform connecting phenotypes to genotypes across species. Nucleic Acids Research, 2017, 45, D712-D722.	14.5	306
42	A large scale hearing loss screen reveals an extensive unexplored genetic landscape for auditory dysfunction. Nature Communications, 2017, 8, 886.	12.8	116
43	Prevalence of sexual dimorphism in mammalian phenotypic traits. Nature Communications, 2017, 8, 15475.	12.8	200
44	Disease model discovery from 3,328 gene knockouts by The International Mouse Phenotyping Consortium. Nature Genetics, 2017, 49, 1231-1238.	21.4	216
45	Defining Disease, Diagnosis, and Translational Medicine within a Homeostatic Perturbation Paradigm: The National Institutes of Health Undiagnosed Diseases Program Experience. Frontiers in Medicine, 2017, 4, 62.	2.6	23
46	Distributed Cognition and Process Management Enabling Individualized Translational Research: The NIH Undiagnosed Diseases Program Experience. Frontiers in Medicine, 2016, 3, 39.	2.6	3
47	A Whole-Genome Analysis Framework for Effective Identification of Pathogenic Regulatory Variants in Mendelian Disease. American Journal of Human Genetics, 2016, 99, 595-606.	6.2	223
48	Navigating the Phenotype Frontier: The Monarch Initiative. Genetics, 2016, 203, 1491-1495.	2.9	65
49	Tools for exploring mouse models of human disease. Drug Discovery Today: Disease Models, 2016, 20, 21-26.	1.2	0
50	Computational evaluation of exome sequence data using human and model organism phenotypes improves diagnostic efficiency. Genetics in Medicine, 2016, 18, 608-617.	2.4	85
51	Use of Model Organism and Disease Databases to Support Matchmaking for Human Disease Gene Discovery. Human Mutation, 2015, 36, 979-984.	2.5	36
52	Phenotype-driven strategies for exome prioritization of human Mendelian disease genes. Genome Medicine, 2015, 7, 81.	8.2	97
53	PhenoMiner: from text to a database of phenotypes associated with OMIM diseases. Database: the Journal of Biological Databases and Curation, 2015, 2015, bav104.	3.0	29
54	Automatic concept recognition using the Human Phenotype Ontology reference and test suite corpora. Database: the Journal of Biological Databases and Curation, 2015, 2015, bav005-bav005.	3.0	55

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55	Generation of Silver Standard Concept Annotations from Biomedical Texts with Special Relevance to Phenotypes. PLoS ONE, 2015, 10, e0116040.	2.5	17
56	Linking gene expression to phenotypes via pathway information. Journal of Biomedical Semantics, 2015, 6, 17.	1.6	26
57	The BioMart community portal: an innovative alternative to large, centralized data repositories. Nucleic Acids Research, 2015, 43, W589-W598.	14.5	682
58	Next-generation diagnostics and disease-gene discovery with the Exomiser. Nature Protocols, 2015, 10, 2004-2015.	12.0	296
59	Applying the ARRIVE Guidelines to an In Vivo Database. PLoS Biology, 2015, 13, e1002151.	5.6	75
60	A mouse informatics platform for phenotypic and translational discovery. Mammalian Genome, 2015, 26, 413-421.	2.2	27
61	Disease insights through cross-species phenotype comparisons. Mammalian Genome, 2015, 26, 548-555.	2.2	19
62	Clinical interpretation of CNVs with cross-species phenotype data. Journal of Medical Genetics, 2014, 51, 766-772.	3.2	23
63	Walking the interactome for candidate prioritization in exome sequencing studies of Mendelian diseases. Bioinformatics, 2014, 30, 3215-3222.	4.1	91
64	Improved exome prioritization of disease genes through cross-species phenotype comparison. Genome Research, 2014, 24, 340-348.	5.5	300
65	The International Mouse Phenotyping Consortium Web Portal, a unified point of access for knockout mice and related phenotyping data. Nucleic Acids Research, 2014, 42, D802-D809.	14.5	252
66	Effective diagnosis of genetic disease by computational phenotype analysis of the disease-associated genome. Science Translational Medicine, 2014, 6, 252ra123.	12.4	223
67	Linking tissues to phenotypes using gene expression profiles. Database: the Journal of Biological Databases and Curation, 2014, 2014, bau017-bau017.	3.0	15
68	The Human Phenotype Ontology project: linking molecular biology and disease through phenotype data. Nucleic Acids Research, 2014, 42, D966-D974.	14.5	698
69	Jannovar: A Java Library for Exome Annotation. Human Mutation, 2014, 35, 548-555.	2.5	63
70	Using association rule mining to determine promising secondary phenotyping hypotheses. Bioinformatics, 2014, 30, i52-i59.	4.1	9
71	The influence of disease categories on gene candidate predictions from model organism phenotypes. Journal of Biomedical Semantics, 2014, 5, S4.	1.6	9
72	Genome-wide Generation and Systematic Phenotyping of Knockout Mice Reveals New Roles for Many Genes. Cell, 2013, 154, 452-464.	28.9	449

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73	PhenoDigm: analyzing curated annotations to associate animal models with human diseases. Database: the Journal of Biological Databases and Curation, 2013, 2013, bat025-bat025.	3.0	115
74	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.	2.4	43
75	Construction and accessibility of a cross-species phenotype ontology along with gene annotations for biomedical research. F1000Research, 2013, 2, 30.	1.6	72
76	Construction and accessibility of a cross-species phenotype ontology along with gene annotations for biomedical research. F1000Research, 2013, 2, 30.	1.6	64
77	The mammalian gene function resource: the international knockout mouse consortium. Mammalian Genome, 2012, 23, 580-586.	2.2	292
78	Beyond knockouts: cre resources for conditional mutagenesis. Mammalian Genome, 2012, 23, 587-599.	2.2	57
79	MouseFinder: Candidate disease genes from mouse phenotype data. Human Mutation, 2012, 33, 858-866.	2.5	53
80	The IKMC web portal: a central point of entry to data and resources from the International Knockout Mouse Consortium. Nucleic Acids Research, 2011, 39, D849-D855.	14.5	83
81	EMMA--mouse mutant resources for the international scientific community. Nucleic Acids Research, 2010, 38, D570-D576.	14.5	39
82	Sustaining the Data and Bioresource Commons. Science, 2010, 330, 592-593.	12.6	52
83	Solutions for data integration in functional genomics: a critical assessment and case study. Briefings in Bioinformatics, 2008, 9, 532-544.	6.5	23
84	EnsMart: A Generic System for Fast and Flexible Access to Biological Data. Genome Research, 2004, 14, 160-169.	5.5	348
85	An Overview of Ensembl. Genome Research, 2004, 14, 925-928.	5.5	391
86	Association and Haplotype Analysis of the Insulin-Degrading Enzyme (IDE) Gene, a Strong Positional and Biological Candidate for Type 2 Diabetes Susceptibility. Diabetes, 2003, 52, 1300-1305.	0.6	52
87	eVOC: A Controlled Vocabulary for Unifying Gene Expression Data. Genome Research, 2003, 13, 1222-1230.	5.5	144
88	A Genomewide Scan for Loci Predisposing to Type 2 Diabetes in a U.K. Population (The Diabetes UK) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 5 Locus on Chromosome 1q. American Journal of Human Genetics, 2001, 69, 553-569.	6.2	300
89	Characterization of chromosome 1 abnormalities in malignant melanomas. , 2000, 28, 121-125.		69
90	ZNF198-FGFR1 Transforms Ba/F3 Cells to Growth Factor Independence and Results in High Level Tyrosine Phosphorylation of STATS 1 and 5. Neoplasia, 1999, 1, 349-355.	5.3	49

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91	The Genomic Structure of ZNF198 and Location of Breakpoints in the t(8;13) Myeloproliferative Syndrome. Genomics, 1999, 55, 118-121.	2.9	24
92	Cloning and Mapping of Members of the MYM Family. Genomics, 1999, 60, 244-247.	2.9	28
93	Fusion of splicing factor genes PSF and NonO (p54nrb) to the TFE3 gene in papillary renal cell carcinoma. Oncogene, 1997, 15, 2233-2239.	5.9	298