Michael Brudno

List of Publications by Year
in descending order

89 papers	8,990 citations	126907 33 h-index	58581 82 g-index
96 all docs	96 docs citations	96 times ranked	17561 citing authors

#	Article	IF	Citations
1	Similarity network fusion for aggregating data types on a genomic scale. Nature Methods, 2014, 11, 333-337.	19.0	1,392
2	The Human Phenotype Ontology in 2017. Nucleic Acids Research, 2017, 45, D865-D876.	14.5	699
3	The Human Phenotype Ontology project: linking molecular biology and disease through phenotype data. Nucleic Acids Research, 2014, 42, D966-D974.	14.5	698
4	Expansion of the Human Phenotype Ontology (HPO) knowledge base and resources. Nucleic Acids Research, 2019, 47, D1018-D1027.	14.5	539
5	Genomic analysis of diffuse intrinsic pontine gliomas identifies three molecular subgroups and recurrent activating ACVR1 mutations. Nature Genetics, 2014, 46, 451-456.	21.4	525
6	SHRiMP: Accurate Mapping of Short Color-space Reads. PLoS Computational Biology, 2009, 5, e1000386.	3.2	485
7	The Matchmaker Exchange: A Platform for Rare Disease Gene Discovery. Human Mutation, 2015, 36, 915-921.	2.5	390
8	Glocal alignment: finding rearrangements during alignment. Bioinformatics, 2003, 19, i54-i62.	4.1	333
9	International Cooperation to Enable the Diagnosis of All Rare Genetic Diseases. American Journal of Human Genetics, 2017, 100, 695-705.	6.2	305
10	Whole-genome sequencing expands diagnostic utility and improves clinical management in paediatric medicine. Npj Genomic Medicine, 2016, 1, .	3.8	295
11	FORGE Canada Consortium: Outcomes of a 2-Year National Rare-Disease Gene-Discovery Project. American Journal of Human Genetics, 2014, 94, 809-817.	6.2	219
12	PhenoTips: Patient Phenotyping Software for Clinical and Research Use. Human Mutation, 2013, 34, 1057-1065.	2.5	207
13	Alterations in ALK/ROS1/NTRK/MET drive a group of infantile hemispheric gliomas. Nature Communications, 2019, 10, 4343.	12.8	200
14	Integrated (epi)-Genomic Analyses Identify Subgroup-Specific Therapeutic Targets in CNS Rhabdoid Tumors. Cancer Cell, 2016, 30, 891-908.	16.8	191
15	Expanding the Boundaries of RNA Sequencing as a Diagnostic Tool for Rare Mendelian Disease. American Journal of Human Genetics, 2019, 104, 466-483.	6.2	176
16	CHARGE and Kabuki Syndromes: Gene-Specific DNA Methylation Signatures Identify Epigenetic Mechanisms Linking These Clinically Overlapping Conditions. American Journal of Human Genetics, 2017, 100, 773-788.	6.2	166
17	Spatial genomic heterogeneity in diffuse intrinsic pontine and midline high-grade glioma: implications for diagnostic biopsy and targeted therapeutics. Acta Neuropathologica Communications, 2016, 4, 1.	5.2	144
18	PRISM: Pair-read informed split-read mapping for base-pair level detection of insertion, deletion and structural variants. Bioinformatics, 2012, 28, 2576-2583.	4.1	109

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19	PhenomeCentral: A Portal for Phenotypic and Genotypic Matchmaking of Patients with Rare Genetic Diseases. Human Mutation, 2015, 36, 931-940.	2.5	107
20	Alignathon: a competitive assessment of whole-genome alignment methods. Genome Research, 2014, 24, 2077-2089.	5.5	102
21	GA4GH: International policies and standards for data sharing across genomic research and healthcare. Cell Genomics, 2021, 1, 100029.	6.5	94
22	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
23	Prevalence and Clinical Features of Inflammatory Bowel Diseases Associated With Monogenic Variants, Identified by Whole-Exome Sequencing in 1000 Children at a Single Center. Gastroenterology, 2020, 158, 2208-2220.	1.3	81
24	deBGA: read alignment with de Bruijn graph-based seed and extension. Bioinformatics, 2016, 32, 3224-3232.	4.1	74
25	Lactase nonpersistence is directed by DNA-variation-dependent epigenetic aging. Nature Structural and Molecular Biology, 2016, 23, 566-573.	8.2	72
26	Consent Codes: Upholding Standard Data Use Conditions. PLoS Genetics, 2016, 12, e1005772.	3.5	65
27	Impact of assisted reproduction, infertility, sex and paternal factors on the placental DNA methylome. Human Molecular Genetics, 2019, 28, 372-385.	2.9	61
28	DNA Methylation Signature for EZH2 Functionally Classifies Sequence Variants in Three PRC2 Complex Genes. American Journal of Human Genetics, 2020, 106, 596-610.	6.2	59
29	Centroid-based Deep Metric Learning for Speaker Recognition. , 2019, , .		55
30	Genome-wide placental DNA methylation analysis of severely growth-discordant monochorionic twins reveals novel epigenetic targets for intrauterine growth restriction. Clinical Epigenetics, 2016, 8, 70.	4.1	51
31	The Matchmaker Exchange API: Automating Patient Matching Through the Exchange of Structured Phenotypic and Genotypic Profiles. Human Mutation, 2015, 36, 922-927.	2.5	50
32	The missing indels: an estimate of indel variation in a human genome and analysis of factors that impede detection. Nucleic Acids Research, 2015, 43, 7217-7228.	14.5	47
33	Tamoxifen therapy in a murine model of myotubular myopathy. Nature Communications, 2018, 9, 4849.	12.8	41
34	Identifying Clinical Terms in Medical Text Using Ontology-Guided Machine Learning. JMIR Medical Informatics, 2019, 7, e12596.	2.6	38
35	Registered access: authorizing data access. European Journal of Human Genetics, 2018, 26, 1721-1731.	2.8	33
36	Care and cost consequences of pediatric whole genome sequencing compared to chromosome microarray. European Journal of Human Genetics, 2017, 25, 1303-1312.	2.8	32

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37	Control of Long-Term Synaptic Potentiation and Learning by Alternative Splicing of the NMDA Receptor Subunit GluN1. Cell Reports, 2019, 29, 4285-4294.e5.	6.4	32
38	Identification of complex genomic rearrangements in cancers using CouGaR. Genome Research, 2017, 27, 107-117.	5.5	31
39	Multiple whole genome alignments and novel biomedical applications at the VISTA portal. Nucleic Acids Research, 2007, 35, W669-W674.	14.5	30
40	Phenotyping: Targeting genotype's rich cousin for diagnosis. Journal of Paediatrics and Child Health, 2015, 51, 381-386.	0.8	29
41	Harmonising phenomics information for a better interoperability in the rare disease field. European Journal of Medical Genetics, 2018, 61, 706-714.	1.3	29
42	GenomeVISTAâ€"an integrated software package for whole-genome alignment and visualization. Bioinformatics, 2014, 30, 2654-2655.	4.1	27
43	Obsessive-compulsive disorder and attention-deficit/hyperactivity disorder: distinct associations with DNA methylation and genetic variation. Journal of Neurodevelopmental Disorders, 2020, 12, 23.	3.1	27
44	More Text Please! Understanding and Supporting the Use of Visualization for Clinical Text Overview. , 2018, , .		25
45	New insights into DNA methylation signatures: SMARCA2 variants in Nicolaides-Baraitser syndrome. BMC Medical Genomics, 2019, 12, 105.	1.5	25
46	International federation of genomic medicine databases using GA4GH standards. Cell Genomics, 2021, 1, 100032.	6.5	22
47	DNA methylation signature is prognostic of choroid plexus tumor aggressiveness. Clinical Epigenetics, 2019, 11, 117.	4.1	21
48	A Cross-Sectional Study of Nemaline Myopathy. Neurology, 2021, 96, e1425-e1436.	1.1	21
49	Detecting Alu insertions from high-throughput sequencing data. Nucleic Acids Research, 2013, 41, e169-e169.	14.5	20
50	Improved Diagnosis and Care for Rare Diseases through Implementation of Precision Public Health Framework. Advances in Experimental Medicine and Biology, 2017, 1031, 55-94.	1.6	20
51	PhenoLines: Phenotype Comparison Visualizations for Disease Subtyping via Topic Models. IEEE Transactions on Visualization and Computer Graphics, 2018, 24, 371-381.	4.4	20
52	Probabilistic method for detecting copy number variation in a fetal genome using maternal plasma sequencing. Bioinformatics, 2014, 30, i212-i218.	4.1	18
53	CReSCENT: CanceR Single Cell ExpressioN Toolkit. Nucleic Acids Research, 2020, 48, W372-W379.	14.5	18
54	Splicing is an alternate oncogenic pathway activation mechanism in glioma. Nature Communications, 2022, 13, 588.	12.8	17

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55	EpigenCentral: Portal for DNA methylation data analysis and classification in rare diseases. Human Mutation, 2020, 41, 1722-1733.	2.5	15
56	Outcome of over 1500 matches through the Matchmaker Exchange for rare disease gene discovery: The 2-year experience of Care4Rare Canada. Genetics in Medicine, 2022, 24, 100-108.	2.4	15
57	Enabling Global Clinical Collaborations on Identifiable Patient Data: The Minerva Initiative. Frontiers in Genetics, 2019, 10, 611.	2.3	14
58	The Genomic Birthday Paradox: How Much Is Enough?. Human Mutation, 2015, 36, 989-997.	2.5	13
59	â€œMatchingâ€•consent to purpose: The example of the Matchmaker Exchange. Human Mutation, 2017, 38, 1281-1285.	2.5	13
60	Speaker Diarization with Session-Level Speaker Embedding Refinement Using Graph Neural Networks. , 2020, , .		12
61	Assessment of Machine Learningâ€“Based Medical Directives to Expedite Care in Pediatric Emergency Medicine. JAMA Network Open, 2022, 5, e222599.	5.9	12
62	A novel strain of cynomolgus macaque cytomegalovirus: implications for host-virus co-evolution. BMC Genomics, 2016, 17, 277.	2.8	11
63	Automatically disambiguating medical acronyms with ontology-aware deep learning. Nature Communications, 2021, 12, 5319.	12.8	10
64	CanDIG: Federated network across Canada for multi-omic and health data discovery and analysis. Cell Genomics, 2021, 1, 100033.	6.5	10
65	Prioritizing Clinically Relevant Copy Number Variation from Genetic Interactions and Gene Function Data. PLoS ONE, 2015, 10, e0139656.	2.5	9
66	An Introduction to the Lagan Alignment Toolkit. Methods in Molecular Biology, 2007, 395, 205-219.	0.9	9
67	PhenomeCentral: 7 years of rare disease matchmaking. Human Mutation, 2022, , .	2.5	9
68	Donâ€™t brush off buccal data heterogeneity. Epigenetics, 2019, 14, 109-117.	2.7	8
69	Assembly and characterization of novelAluinserts detected from next-generation sequencing data. Mobile Genetic Elements, 2014, 4, 1-7.	1.8	6
70	Cell-free DNA fragment-size distribution analysis for non-invasive prenatal CNV prediction. Bioinformatics, 2016, 32, 1662-1669.	4.1	6
71	Extract and componentâ€specific sensitization patterns in Canadian moderateâ€toâ€severe preschool asthmatics. Allergy: European Journal of Allergy and Clinical Immunology, 2019, 74, 2519-2521.	5.7	6
72	MetaFusion: a high-confidence metacaller for filtering and prioritizing RNA-seq gene fusion candidates. Bioinformatics, 2021, 37, 3144-3151.	4.1	6

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73	PhenoPad: Building AI enabled note-taking interfaces for patient encounters. Npj Digital Medicine, 2022, 5, 12.	10.9	6
74	Genomics4RD: An integrated platform to share Canadian deep-phenotype and multiomic data for international rare disease gene discovery.. Human Mutation, 2022, , .	2.5	4
75	Distributed Cognition and Process Management Enabling Individualized Translational Research: The NIH Undiagnosed Diseases Program Experience. Frontiers in Medicine, 2016, 3, 39.	2.6	3
76	An MRspec database query and visualization engine with applications as a clinical diagnostic and research tool. Molecular Genetics and Metabolism, 2016, 119, 300-306.	1.1	3
77	Structural Variant in Mitochondrial-Associated Gene (MRPL3) Induces Adult-Onset Neurodegeneration with Memory Impairment in the Mouse. Journal of Neuroscience, 2020, 40, 4576-4585.	3.6	3
78	MIXTURE MODEL FOR SUB-PHENOTYPING IN GWAS. , 2011, , .		2
79	From Clinic to Computer and Back Again: Practical Considerations When Designing and Implementing Machine Learning Solutions for Pediatrics. Current Treatment Options in Pediatrics, 2020, 6, 336-349.	0.6	2
80	Phenotate: crowdsourcing phenotype annotations as exercises in undergraduate classes. Genetics in Medicine, 2020, 22, 1391-1400.	2.4	2
81	Child Neurology: RNA Sequencing for the Diagnosis of Lissencephaly. Neurology, 2021, 97, .	1.1	2
82	MG-132â€…Diagnostic utility of whole genome sequencing in paediatric medicine. Journal of Medical Genetics, 2015, 52, A12.1-A12.	3.2	1
83	Identifying Clinical Terms in Free-Text Notes Using Ontology-Guided Machine Learning. Lecture Notes in Computer Science, 2019, , 19-34.	1.3	1
84	A Canadian Study of Cisplatin Metabolomics and Nephrotoxicity (ACCENT): A Clinical Research Protocol. Canadian Journal of Kidney Health and Disease, 2021, 8, 205435812110577.	1.1	1
85	A report on the 2009 SIG on short read sequencing and algorithms (Short-SIG). Bioinformatics, 2009, 25, 2863-2864.	4.1	0
86	MG-108â€…Beyond the ACMG 56: Parental choices and initial results from a comprehensive whole genome sequencing-based search for predictive genomic variants in children. Journal of Medical Genetics, 2015, 52, A3.2-A4.	3.2	0
87	HGG-18. ALTERNATIVE SPLICING OF NEUROFIBROMIN 1 IS ASSOCIATED WITH ELEVATED MAPK ACTIVITY AND POOR PROGNOSIS IN HIGH-GRADE GLIOMA. Neuro-Oncology, 2019, 21, ii90-ii90.	1.2	0
88	Creation of an electronic patient-reported outcome measure platform Voxe: a mixed methods study protocol in paediatric solid organ transplantation. BMJ Open, 2021, 11, e053119.	1.9	0
89	Characterization of Key Sexually Dimorphic Regulators in Pain Processing. Canadian Journal of Pain, 0, , .	1.7	0