

Michael Brudno

List of Publications by Year in descending order

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Version: 2024-02-01

89
papers

8,990
citations

145106

33
h-index

66518

82
g-index

96
all docs

96
docs citations

96
times ranked

19253
citing authors

#	ARTICLE	IF	CITATIONS
1	Outcome of over 1500 matches through the Matchmaker Exchange for rare disease gene discovery: The 2-year experience of Care4Rare Canada. <i>Genetics in Medicine</i> , 2022, 24, 100-108.	1.1	15
2	Splicing is an alternate oncogenic pathway activation mechanism in glioma. <i>Nature Communications</i> , 2022, 13, 588.	5.8	17
3	PhenoPad: Building AI enabled note-taking interfaces for patient encounters. <i>Npj Digital Medicine</i> , 2022, 5, 12.	5.7	6
4	PhenomeCentral: 7 years of rare disease matchmaking. <i>Human Mutation</i> , 2022, , .	1.1	9
5	Assessment of Machine Learning–Based Medical Directives to Expedite Care in Pediatric Emergency Medicine. <i>JAMA Network Open</i> , 2022, 5, e222599.	2.8	12
6	Genomics4RD: An integrated platform to share Canadian deep-phenotype and multiomic data for international rare disease gene discovery.. <i>Human Mutation</i> , 2022, , .	1.1	4
7	A Cross-Sectional Study of Nemaline Myopathy. <i>Neurology</i> , 2021, 96, e1425-e1436.	1.5	21
8	MetaFusion: a high-confidence metacaller for filtering and prioritizing RNA-seq gene fusion candidates. <i>Bioinformatics</i> , 2021, 37, 3144-3151.	1.8	6
9	Child Neurology: RNA Sequencing for the Diagnosis of Lissencephaly. <i>Neurology</i> , 2021, 97, .	1.5	2
10	Automatically disambiguating medical acronyms with ontology-aware deep learning. <i>Nature Communications</i> , 2021, 12, 5319.	5.8	10
11	Creation of an electronic patient-reported outcome measure platform Voxe: a mixed methods study protocol in paediatric solid organ transplantation. <i>BMJ Open</i> , 2021, 11, e053119.	0.8	0
12	GA4GH: International policies and standards for data sharing across genomic research and healthcare. <i>Cell Genomics</i> , 2021, 1, 100029.	3.0	94
13	CanDIG: Federated network across Canada for multi-omic and health data discovery and analysis. <i>Cell Genomics</i> , 2021, 1, 100033.	3.0	10
14	International federation of genomic medicine databases using GA4GH standards. <i>Cell Genomics</i> , 2021, 1, 100032.	3.0	22
15	A Canadian Study of Cisplatin Metabolomics and Nephrotoxicity (ACCENT): A Clinical Research Protocol. <i>Canadian Journal of Kidney Health and Disease</i> , 2021, 8, 205435812110577.	0.6	1
16	Obsessive-compulsive disorder and attention-deficit/hyperactivity disorder: distinct associations with DNA methylation and genetic variation. <i>Journal of Neurodevelopmental Disorders</i> , 2020, 12, 23.	1.5	27
17	From Clinic to Computer and Back Again: Practical Considerations When Designing and Implementing Machine Learning Solutions for Pediatrics. <i>Current Treatment Options in Pediatrics</i> , 2020, 6, 336-349.	0.2	2
18	Structural Variant in Mitochondrial-Associated Gene (MRPL3) Induces Adult-Onset Neurodegeneration with Memory Impairment in the Mouse. <i>Journal of Neuroscience</i> , 2020, 40, 4576-4585.	1.7	3

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19	Phenotate: crowdsourcing phenotype annotations as exercises in undergraduate classes. <i>Genetics in Medicine</i> , 2020, 22, 1391-1400.	1.1	2
20	CReSCENT: CanceR Single Cell ExpressioN Toolkit. <i>Nucleic Acids Research</i> , 2020, 48, W372-W379.	6.5	18
21	EpigenCentral: Portal for DNA methylation data analysis and classification in rare diseases. <i>Human Mutation</i> , 2020, 41, 1722-1733.	1.1	15
22	Prevalence and Clinical Features of Inflammatory Bowel Diseases Associated With Monogenic Variants, Identified by Whole-Exome Sequencing in 1000 Children at a Single Center. <i>Gastroenterology</i> , 2020, 158, 2208-2220.	0.6	81
23	Speaker Diarization with Session-Level Speaker Embedding Refinement Using Graph Neural Networks. , 2020, , .		12
24	DNA Methylation Signature for EZH2 Functionally Classifies Sequence Variants in Three PRC2 Complex Genes. <i>American Journal of Human Genetics</i> , 2020, 106, 596-610.	2.6	59
25	DNA methylation signature is prognostic of choroid plexus tumor aggressiveness. <i>Clinical Epigenetics</i> , 2019, 11, 117.	1.8	21
26	Centroid-based Deep Metric Learning for Speaker Recognition. , 2019, , .		55
27	New insights into DNA methylation signatures: SMARCA2 variants in Nicolaides-Baraitser syndrome. <i>BMC Medical Genomics</i> , 2019, 12, 105.	0.7	25
28	Alterations in ALK/ROS1/NTRK/MET drive a group of infantile hemispheric gliomas. <i>Nature Communications</i> , 2019, 10, 4343.	5.8	200
29	Extract and component-specific sensitization patterns in Canadian moderate-to-severe preschool asthmatics. <i>Allergy: European Journal of Allergy and Clinical Immunology</i> , 2019, 74, 2519-2521.	2.7	6
30	Enabling Global Clinical Collaborations on Identifiable Patient Data: The Minerva Initiative. <i>Frontiers in Genetics</i> , 2019, 10, 611.	1.1	14
31	HGG-18. ALTERNATIVE SPLICING OF NEUROFIBROMIN 1 IS ASSOCIATED WITH ELEVATED MAPK ACTIVITY AND POOR PROGNOSIS IN HIGH-GRADE GLIOMA. <i>Neuro-Oncology</i> , 2019, 21, ii90-ii90.	0.6	0
32	Identifying Clinical Terms in Free-Text Notes Using Ontology-Guided Machine Learning. <i>Lecture Notes in Computer Science</i> , 2019, , 19-34.	1.0	1
33	Don't brush off buccal data heterogeneity. <i>Epigenetics</i> , 2019, 14, 109-117.	1.3	8
34	Expanding the Boundaries of RNA Sequencing as a Diagnostic Tool for Rare Mendelian Disease. <i>American Journal of Human Genetics</i> , 2019, 104, 466-483.	2.6	176
35	Control of Long-Term Synaptic Potentiation and Learning by Alternative Splicing of the NMDA Receptor Subunit GluN1. <i>Cell Reports</i> , 2019, 29, 4285-4294.e5.	2.9	32
36	Expansion of the Human Phenotype Ontology (HPO) knowledge base and resources. <i>Nucleic Acids Research</i> , 2019, 47, D1018-D1027.	6.5	539

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37	Impact of assisted reproduction, infertility, sex and paternal factors on the placental DNA methylome. <i>Human Molecular Genetics</i> , 2019, 28, 372-385.	1.4	61
38	Identifying Clinical Terms in Medical Text Using Ontology-Guided Machine Learning. <i>JMIR Medical Informatics</i> , 2019, 7, e12596.	1.3	38
39	Harmonising phenomics information for a better interoperability in the rare disease field. <i>European Journal of Medical Genetics</i> , 2018, 61, 706-714.	0.7	29
40	Tamoxifen therapy in a murine model of myotubular myopathy. <i>Nature Communications</i> , 2018, 9, 4849.	5.8	41
41	More Text Please! Understanding and Supporting the Use of Visualization for Clinical Text Overview. , 2018, , .		25
42	Registered access: authorizing data access. <i>European Journal of Human Genetics</i> , 2018, 26, 1721-1731.	1.4	33
43	PhenoLines: Phenotype Comparison Visualizations for Disease Subtyping via Topic Models. <i>IEEE Transactions on Visualization and Computer Graphics</i> , 2018, 24, 371-381.	2.9	20
44	The Human Phenotype Ontology in 2017. <i>Nucleic Acids Research</i> , 2017, 45, D865-D876.	6.5	699
45	International Cooperation to Enable the Diagnosis of All Rare Genetic Diseases. <i>American Journal of Human Genetics</i> , 2017, 100, 695-705.	2.6	305
46	â€œMatchingâ€•consent to purpose: The example of the Matchmaker Exchange. <i>Human Mutation</i> , 2017, 38, 1281-1285.	1.1	13
47	Improved Diagnosis and Care for Rare Diseases through Implementation of Precision Public Health Framework. <i>Advances in Experimental Medicine and Biology</i> , 2017, 1031, 55-94.	0.8	20
48	Care and cost consequences of pediatric whole genome sequencing compared to chromosome microarray. <i>European Journal of Human Genetics</i> , 2017, 25, 1303-1312.	1.4	32
49	Identification of complex genomic rearrangements in cancers using CouGaR. <i>Genome Research</i> , 2017, 27, 107-117.	2.4	31
50	CHARGE and Kabuki Syndromes: Gene-Specific DNA Methylation Signatures Identify Epigenetic Mechanisms Linking These Clinically Overlapping Conditions. <i>American Journal of Human Genetics</i> , 2017, 100, 773-788.	2.6	166
51	Distributed Cognition and Process Management Enabling Individualized Translational Research: The NIH Undiagnosed Diseases Program Experience. <i>Frontiers in Medicine</i> , 2016, 3, 39.	1.2	3
52	Consent Codes: Upholding Standard Data Use Conditions. <i>PLoS Genetics</i> , 2016, 12, e1005772.	1.5	65
53	deBGA: read alignment with de Bruijn graph-based seed and extension. <i>Bioinformatics</i> , 2016, 32, 3224-3232.	1.8	74
54	Whole-genome sequencing expands diagnostic utility and improves clinical management in paediatric medicine. <i>Npj Genomic Medicine</i> , 2016, 1, .	1.7	295

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55	Integrated (epi)-Genomic Analyses Identify Subgroup-Specific Therapeutic Targets in CNS Rhabdoid Tumors. <i>Cancer Cell</i> , 2016, 30, 891-908.	7.7	191
56	Cell-free DNA fragment-size distribution analysis for non-invasive prenatal CNV prediction. <i>Bioinformatics</i> , 2016, 32, 1662-1669.	1.8	6
57	Lactase nonpersistence is directed by DNA-variation-dependent epigenetic aging. <i>Nature Structural and Molecular Biology</i> , 2016, 23, 566-573.	3.6	72
58	Genome-wide placental DNA methylation analysis of severely growth-discordant monozygotic twins reveals novel epigenetic targets for intrauterine growth restriction. <i>Clinical Epigenetics</i> , 2016, 8, 70.	1.8	51
59	An MRspec database query and visualization engine with applications as a clinical diagnostic and research tool. <i>Molecular Genetics and Metabolism</i> , 2016, 119, 300-306.	0.5	3
60	A novel strain of cynomolgus macaque cytomegalovirus: implications for host-virus co-evolution. <i>BMC Genomics</i> , 2016, 17, 277.	1.2	11
61	Spatial genomic heterogeneity in diffuse intrinsic pontine and midline high-grade glioma: implications for diagnostic biopsy and targeted therapeutics. <i>Acta Neuropathologica Communications</i> , 2016, 4, 1.	2.4	144
62	Computational evaluation of exome sequence data using human and model organism phenotypes improves diagnostic efficiency. <i>Genetics in Medicine</i> , 2016, 18, 608-617.	1.1	85
63	PhenomeCentral: A Portal for Phenotypic and Genotypic Matchmaking of Patients with Rare Genetic Diseases. <i>Human Mutation</i> , 2015, 36, 931-940.	1.1	107
64	The Genomic Birthday Paradox: How Much Is Enough?. <i>Human Mutation</i> , 2015, 36, 989-997.	1.1	13
65	The Matchmaker Exchange API: Automating Patient Matching Through the Exchange of Structured Phenotypic and Genotypic Profiles. <i>Human Mutation</i> , 2015, 36, 922-927.	1.1	50
66	MG-132â€¦Diagnostic utility of whole genome sequencing in paediatric medicine. <i>Journal of Medical Genetics</i> , 2015, 52, A12.1-A12.	1.5	1
67	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. <i>Journal of Medical Genetics</i> , 2015, 52, A3.2-A4.	1.5	0
68	Prioritizing Clinically Relevant Copy Number Variation from Genetic Interactions and Gene Function Data. <i>PLoS ONE</i> , 2015, 10, e0139656.	1.1	9
69	Phenotyping: Targeting genotype's rich cousin for diagnosis. <i>Journal of Paediatrics and Child Health</i> , 2015, 51, 381-386.	0.4	29
70	The Matchmaker Exchange: A Platform for Rare Disease Gene Discovery. <i>Human Mutation</i> , 2015, 36, 915-921.	1.1	390
71	The missing indels: an estimate of indel variation in a human genome and analysis of factors that impede detection. <i>Nucleic Acids Research</i> , 2015, 43, 7217-7228.	6.5	47
72	Probabilistic method for detecting copy number variation in a fetal genome using maternal plasma sequencing. <i>Bioinformatics</i> , 2014, 30, i212-i218.	1.8	18

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73	Assembly and characterization of novel <i>Alu</i> inserts detected from next-generation sequencing data. <i>Mobile Genetic Elements</i> , 2014, 4, 1-7.	1.8	6
74	Alignathon: a competitive assessment of whole-genome alignment methods. <i>Genome Research</i> , 2014, 24, 2077-2089.	2.4	102
75	Genomic analysis of diffuse intrinsic pontine gliomas identifies three molecular subgroups and recurrent activating ACVR1 mutations. <i>Nature Genetics</i> , 2014, 46, 451-456.	9.4	525
76	The Human Phenotype Ontology project: linking molecular biology and disease through phenotype data. <i>Nucleic Acids Research</i> , 2014, 42, D966-D974.	6.5	698
77	GenomeVISTA—an integrated software package for whole-genome alignment and visualization. <i>Bioinformatics</i> , 2014, 30, 2654-2655.	1.8	27
78	Similarity network fusion for aggregating data types on a genomic scale. <i>Nature Methods</i> , 2014, 11, 333-337.	9.0	1,392
79	FORGE Canada Consortium: Outcomes of a 2-Year National Rare-Disease Gene-Discovery Project. <i>American Journal of Human Genetics</i> , 2014, 94, 809-817.	2.6	219
80	PhenoTips: Patient Phenotyping Software for Clinical and Research Use. <i>Human Mutation</i> , 2013, 34, 1057-1065.	1.1	207
81	Detecting <i>Alu</i> insertions from high-throughput sequencing data. <i>Nucleic Acids Research</i> , 2013, 41, e169-e169.	6.5	20
82	PRISM: Pair-read informed split-read mapping for base-pair level detection of insertion, deletion and structural variants. <i>Bioinformatics</i> , 2012, 28, 2576-2583.	1.8	109
83	MIXTURE MODEL FOR SUB-PHENOTYPING IN GWAS. , 2011, , .		2
84	A report on the 2009 SIG on short read sequencing and algorithms (Short-SIG). <i>Bioinformatics</i> , 2009, 25, 2863-2864.	1.8	0
85	SHRiMP: Accurate Mapping of Short Color-space Reads. <i>PLoS Computational Biology</i> , 2009, 5, e1000386.	1.5	485
86	Multiple whole genome alignments and novel biomedical applications at the VISTA portal. <i>Nucleic Acids Research</i> , 2007, 35, W669-W674.	6.5	30
87	An Introduction to the Lagan Alignment Toolkit. <i>Methods in Molecular Biology</i> , 2007, 395, 205-219.	0.4	9
88	Glocal alignment: finding rearrangements during alignment. <i>Bioinformatics</i> , 2003, 19, i54-i62.	1.8	333
89	Characterization of Key Sexually Dimorphic Regulators in Pain Processing. <i>Canadian Journal of Pain</i> , 0, , .	0.6	0