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	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
2	Splicing is an alternate oncogenic pathway activation mechanism in glioma. Nature Communications, 2022, 13, 588.	12.8	17
3	PhenoPad: Building AI enabled note-taking interfaces for patient encounters. Npj Digital Medicine, 2022, 5, 12.	10.9	6
4	PhenomeCentral: 7 years of rare disease matchmaking. Human Mutation, 2022, , .	2.5	9
5	Assessment of Machine Learningâ€“Based Medical Directives to Expedite Care in Pediatric Emergency Medicine. JAMA Network Open, 2022, 5, e222599.	5.9	12
6	Genomics4RD: An integrated platform to share Canadian deep-phenotype and multiomic data for international rare disease gene discovery.. Human Mutation, 2022, , .	2.5	4
7	A Cross-Sectional Study of Nemaline Myopathy. Neurology, 2021, 96, e1425-e1436.	1.1	21
8	MetaFusion: a high-confidence metacaller for filtering and prioritizing RNA-seq gene fusion candidates. Bioinformatics, 2021, 37, 3144-3151.	4.1	6
9	Child Neurology: RNA Sequencing for the Diagnosis of Lissencephaly. Neurology, 2021, 97, .	1.1	2
10	Automatically disambiguating medical acronyms with ontology-aware deep learning. Nature Communications, 2021, 12, 5319.	12.8	10
11	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
12	GA4GH: International policies and standards for data sharing across genomic research and healthcare. Cell Genomics, 2021, 1, 100029.	6.5	94
13	CanDIG: Federated network across Canada for multi-omic and health data discovery and analysis. Cell Genomics, 2021, 1, 100033.	6.5	10
14	International federation of genomic medicine databases using GA4GH standards. Cell Genomics, 2021, 1, 100032.	6.5	22
15	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
16	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
17	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
18	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

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19	Phenotate: crowdsourcing phenotype annotations as exercises in undergraduate classes. Genetics in Medicine, 2020, 22, 1391-1400.	2.4	2
20	CReSCENT: CanceR Single Cell ExpressioN Toolkit. Nucleic Acids Research, 2020, 48, W372-W379.	14.5	18
21	EpigenCentral: Portal for DNA methylation data analysis and classification in rare diseases. Human Mutation, 2020, 41, 1722-1733.	2.5	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. Gastroenterology, 2020, 158, 2208-2220.	1.3	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. American Journal of Human Genetics, 2020, 106, 596-610.	6.2	59
25	DNA methylation signature is prognostic of choroid plexus tumor aggressiveness. Clinical Epigenetics, 2019, 11, 117.	4.1	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. BMC Medical Genomics, 2019, 12, 105.	1.5	25
28	Alterations in ALK/ROS1/NTRK/MET drive a group of infantile hemispheric gliomas. Nature Communications, 2019, 10, 4343.	12.8	200
29	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
30	Enabling Global Clinical Collaborations on Identifiable Patient Data: The Minerva Initiative. Frontiers in Genetics, 2019, 10, 611.	2.3	14
31	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
32	Identifying Clinical Terms in Free-Text Notes Using Ontology-Guided Machine Learning. Lecture Notes in Computer Science, 2019, , 19-34.	1.3	1
33	Donâ€™t brush off buccal data heterogeneity. Epigenetics, 2019, 14, 109-117.	2.7	8
34	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
35	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
36	Expansion of the Human Phenotype Ontology (HPO) knowledge base and resources. Nucleic Acids Research, 2019, 47, D1018-D1027.	14.5	539

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37	Impact of assisted reproduction, infertility, sex and paternal factors on the placental DNA methylome. Human Molecular Genetics, 2019, 28, 372-385.	2.9	61
38	Identifying Clinical Terms in Medical Text Using Ontology-Guided Machine Learning. JMIR Medical Informatics, 2019, 7, e12596.	2.6	38
39	Harmonising phenomics information for a better interoperability in the rare disease field. European Journal of Medical Genetics, 2018, 61, 706-714.	1.3	29
40	Tamoxifen therapy in a murine model of myotubular myopathy. Nature Communications, 2018, 9, 4849.	12.8	41
41	More Text Please! Understanding and Supporting the Use of Visualization for Clinical Text Overview. , 2018, , .		25
42	Registered access: authorizing data access. European Journal of Human Genetics, 2018, 26, 1721-1731.	2.8	33
43	PhenoLines: Phenotype Comparison Visualizations for Disease Subtyping via Topic Models. IEEE Transactions on Visualization and Computer Graphics, 2018, 24, 371-381.	4.4	20
44	The Human Phenotype Ontology in 2017. Nucleic Acids Research, 2017, 45, D865-D876.	14.5	699
45	International Cooperation to Enable the Diagnosis of All Rare Genetic Diseases. American Journal of Human Genetics, 2017, 100, 695-705.	6.2	305
46	â€œMatchingâ€•consent to purpose: The example of the Matchmaker Exchange. Human Mutation, 2017, 38, 1281-1285.	2.5	13
47	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
48	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
49	Identification of complex genomic rearrangements in cancers using CouGaR. Genome Research, 2017, 27, 107-117.	5.5	31
50	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
51	Distributed Cognition and Process Management Enabling Individualized Translational Research: The NIH Undiagnosed Diseases Program Experience. Frontiers in Medicine, 2016, 3, 39.	2.6	3
52	Consent Codes: Upholding Standard Data Use Conditions. PLoS Genetics, 2016, 12, e1005772.	3.5	65
53	deBGA: read alignment with de Bruijn graph-based seed and extension. Bioinformatics, 2016, 32, 3224-3232.	4.1	74
54	Whole-genome sequencing expands diagnostic utility and improves clinical management in paediatric medicine. Npj Genomic Medicine, 2016, 1, .	3.8	295

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55	Integrated (epi)-Genomic Analyses Identify Subgroup-Specific Therapeutic Targets in CNS Rhabdoid Tumors. Cancer Cell, 2016, 30, 891-908.	16.8	191
56	Cell-free DNA fragment-size distribution analysis for non-invasive prenatal CNV prediction. Bioinformatics, 2016, 32, 1662-1669.	4.1	6
57	Lactase nonpersistence is directed by DNA-variation-dependent epigenetic aging. Nature Structural and Molecular Biology, 2016, 23, 566-573.	8.2	72
58	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
59	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
60	A novel strain of cynomolgus macaque cytomegalovirus: implications for host-virus co-evolution. BMC Genomics, 2016, 17, 277.	2.8	11
61	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
62	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
63	PhenomeCentral: A Portal for Phenotypic and Genotypic Matchmaking of Patients with Rare Genetic Diseases. Human Mutation, 2015, 36, 931-940.	2.5	107
64	The Genomic Birthday Paradox: How Much Is Enough?. Human Mutation, 2015, 36, 989-997.	2.5	13
65	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
66	MG-132â€...Diagnostic utility of whole genome sequencing in paediatric medicine. Journal of Medical Genetics, 2015, 52, A12.1-A12.	3.2	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. Journal of Medical Genetics, 2015, 52, A3.2-A4.	3.2	0
68	Prioritizing Clinically Relevant Copy Number Variation from Genetic Interactions and Gene Function Data. PLoS ONE, 2015, 10, e0139656.	2.5	9
69	Phenotyping: Targeting genotype's rich cousin for diagnosis. Journal of Paediatrics and Child Health, 2015, 51, 381-386.	0.8	29
70	The Matchmaker Exchange: A Platform for Rare Disease Gene Discovery. Human Mutation, 2015, 36, 915-921.	2.5	390
71	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
72	Probabilistic method for detecting copy number variation in a fetal genome using maternal plasma sequencing. Bioinformatics, 2014, 30, i212-i218.	4.1	18

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73	Assembly and characterization of novelAluinserts detected from next-generation sequencing data. Mobile Genetic Elements, 2014, 4, 1-7.	1.8	6
74	Alignathon: a competitive assessment of whole-genome alignment methods. Genome Research, 2014, 24, 2077-2089.	5.5	102
75	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
76	The Human Phenotype Ontology project: linking molecular biology and disease through phenotype data. Nucleic Acids Research, 2014, 42, D966-D974.	14.5	698
77	GenomeVISTAâ€"an integrated software package for whole-genome alignment and visualization. Bioinformatics, 2014, 30, 2654-2655.	4.1	27
78	Similarity network fusion for aggregating data types on a genomic scale. Nature Methods, 2014, 11, 333-337.	19.0	1,392
79	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
80	PhenoTips: Patient Phenotyping Software for Clinical and Research Use. Human Mutation, 2013, 34, 1057-1065.	2.5	207
81	Detecting Alu insertions from high-throughput sequencing data. Nucleic Acids Research, 2013, 41, e169-e169.	14.5	20
82	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
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). Bioinformatics, 2009, 25, 2863-2864.	4.1	0
85	SHRiMP: Accurate Mapping of Short Color-space Reads. PLoS Computational Biology, 2009, 5, e1000386.	3.2	485
86	Multiple whole genome alignments and novel biomedical applications at the VISTA portal. Nucleic Acids Research, 2007, 35, W669-W674.	14.5	30
87	An Introduction to the Lagan Alignment Toolkit. Methods in Molecular Biology, 2007, 395, 205-219.	0.9	9
88	Glocal alignment: finding rearrangements during alignment. Bioinformatics, 2003, 19, i54-i62.	4.1	333
89	Characterization of Key Sexually Dimorphic Regulators in Pain Processing. Canadian Journal of Pain, 0, , .	1.7	0