Mathew S Lebo

List of Publications by Year
in descending order

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83	6,015	94433	82547
papers	citations	37 h-index	72 g-index

107	107	107	10963
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Data sharing to improve concordance in variant interpretation across laboratories: results from the Canadian Open Genetics Repository. Journal of Medical Genetics, 2022, 59, 571-578.	3.2	14
2	Reanalysis of eMERGE phase III sequence variants in 10,500 participants and infrastructure to support the automated return of knowledge updates. Genetics in Medicine, 2022, 24, 454-462.	2.4	6
3	Automated Pharmacogenomic Reports for Clinical Genome Sequencing. Journal of Molecular Diagnostics, 2022, 24, 205-218.	2.8	5
4	Harmonizing variant classification for return of results in the All of Us Research Program. Human Mutation, 2022, 43, 1114-1121.	2.5	7
5	Development of a clinical polygenic risk score assay and reporting workflow. Nature Medicine, 2022, 28, 1006-1013.	30.7	74
6	Association of Pathogenic DNA Variants Predisposing to Cardiomyopathy With Cardiovascular Disease Outcomes and All-Cause Mortality. JAMA Cardiology, 2022, 7, 723.	6.1	15
7	Development and validation of a trans-ancestry polygenic risk score for type 2 diabetes in diverse populations. Genome Medicine, 2022, 14, .	8.2	48
8	Discordant results between conventional newborn screening and genomic sequencing in the BabySeq Project. Genetics in Medicine, 2021, 23, 1372-1375.	2.4	47
9	Randomized prospective evaluation of genome sequencing versus standard-of-care as a first molecular diagnostic test. Genetics in Medicine, 2021, 23, 1689-1696.	2.4	17
10	Harmonizing the Collection of Clinical Data on Genetic Testing Requisition Forms to Enhance Variant Interpretation in Hypertrophic Cardiomyopathy (HCM). Journal of Molecular Diagnostics, 2021, 23, 589-598.	2.8	5
11	A framework for automated gene selection in genomic applications. Genetics in Medicine, 2021, 23, 1993-1997.	2.4	5
12	Hematopoietic mosaic chromosomal alterations increase the risk for diverse types of infection. Nature Medicine, 2021, 27, 1012-1024.	30.7	109
13	Neptune: an environment for the delivery of genomic medicine. Genetics in Medicine, 2021, 23, 1838-1846.	2.4	3
14	Psychosocial Effect of Newborn Genomic Sequencing on Families in the BabySeq Project. JAMA Pediatrics, 2021, 175, 1132.	6.2	35
15	Returning actionable genomic results in a research biobank: Analytic validity, clinical implementation, and resource utilization. American Journal of Human Genetics, 2021, 108, 2224-2237.	6.2	34
16	Best practices for the analytical validation of clinical whole-genome sequencing intended for the diagnosis of germline disease. Npj Genomic Medicine, 2020, 5, 47.	3.8	67
17	Airmen and health-care providersâ€™ attitudes toward the use of genomic sequencing in the US Air Force: findings from the MilSeq Project. Genetics in Medicine, 2020, 22, 2003-2010.	2.4	2
18	Polygenic background modifies penetrance of monogenic variants for tier 1 genomic conditions. Nature Communications, 2020, 11, 3635.	12.8	277

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19	Multiple GYPB gene deletions associated with the Uâ˜ phenotype in those of African ancestry. Transfusion, 2020, 60, 1294-1307.	1.6	12
20	Association of Rare Pathogenic DNA Variants for Familial Hypercholesterolemia, Hereditary Breast and Ovarian Cancer Syndrome, and Lynch Syndrome With Disease Risk in Adults According to Family History. JAMA Network Open, 2020, 3, e203959.	5.9	75
21	Overcoming the challenges of interpreting complex and uncommon RH alleles from whole genomes. Vox Sanguinis, 2020, 115, 790-801.	1.5	5
22	An assessment of the role of vinculin loss of function variants in inherited cardiomyopathy. Human Mutation, 2020, 41, 1577-1587.	2.5	10
23	Expanding the Noonan spectrum/RASopathy NGS panel: Benefits of adding NF1 and SPRED1. Molecular Genetics & Genomic Medicine, 2020, 8, e1180.	1.2	9
24	Design and Reporting Considerations for Genetic Screening Tests. Journal of Molecular Diagnostics, 2020, 22, 599-609.	2.8	15
25	Quantifying Downstream Healthcare Utilization in Studies of Genomic Testing. Value in Health, 2020, 23, 559-565.	0.3	6
26	Bioinformatics in Clinical Genomic Sequencing. Clinics in Laboratory Medicine, 2020, 40, 163-187.	1.4	5
27	Automated typing of red blood cell and platelet antigens from whole exome sequences. Transfusion, 2019, 59, 3253-3263.	1.6	32
28	Analyzing and Reanalyzing the Genome: Findings from the MedSeq Project. American Journal of Human Genetics, 2019, 105, 177-188.	6.2	38
29	Rare Genetic Variants Associated With Sudden Cardiac Death in Adults. Journal of the American College of Cardiology, 2019, 74, 2623-2634.	2.8	27
30	Harmonizing Clinical Sequencing and Interpretation for the eMERGE III Network. American Journal of Human Genetics, 2019, 105, 588-605.	6.2	99
31	A Rigorous Interlaboratory Examination of the Need to Confirm Next-Generation Sequencingâ€“Detected Variants with an Orthogonal MethodÂin Clinical Genetic Testing. Journal of Molecular Diagnostics, 2019, 21, 318-329.	2.8	49
32	Genetic variant pathogenicity prediction trained using disease-specific clinical sequencing data sets. Genome Research, 2019, 29, 1144-1151.	5.5	19
33	Considerations for clinical curation, classification, and reporting of low-penetrance and low effect size variants associated with disease risk. Genetics in Medicine, 2019, 21, 2765-2773.	2.4	20
34	Polygenic Prediction of Weight and Obesity Trajectories from Birth to Adulthood. Cell, 2019, 177, 587-596.e9.	28.9	516
35	Variant classification changes over time in BRCA1 and BRCA2. Genetics in Medicine, 2019, 21, 2248-2254.	2.4	37
36	Parental interest in genomic sequencing of newborns: enrollment experience from the BabySeq Project. Genetics in Medicine, 2019, 21, 622-630.	2.4	61

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37	Interpretation of Genomic Sequencing Results in Healthy and Ill Newborns: Results from the BabySeq Project. American Journal of Human Genetics, 2019, 104, 76-93.	6.2	176
38	Designing and Implementing NGS Tests for Inherited Disorders. Journal of Molecular Diagnostics, 2019, 21, 369-374.	2.8	23
39	A whole genome approach for discovering the genetic basis of blood group antigens: independent confirmation for P1 and Xg^a. Transfusion, 2019, 59, 908-915.	1.6	13
40	The eMERGE genotype set of 83,717 subjects imputed to ~40â€‰million variants genome wide and association with the herpes zoster medical record phenotype. Genetic Epidemiology, 2019, 43, 63-81.	1.3	63
41	Early cancer diagnoses through BRCA1/2 screening of unselected adult biobank participants. Genetics in Medicine, 2018, 20, 554-558.	2.4	46
42	Data sharing as a national quality improvement program: reporting on BRCA1 and BRCA2 variant-interpretation comparisons through the Canadian Open Genetics Repository (COGR). Genetics in Medicine, 2018, 20, 294-302.	2.4	27
43	Characterizing reduced coverage regions through comparison of exome and genome sequencing data across 10 centers. Genetics in Medicine, 2018, 20, 855-866.	2.4	22
44	Exome Sequencingâ€“Based Screening for BRCA1/2 Expected Pathogenic Variants Among Adult Biobank Participants. JAMA Network Open, 2018, 1, e182140.	5.9	163
45	Bioinformatics in Clinical Genomic Sequencing. Advances in Molecular Pathology, 2018, 1, 9-26.	0.4	1
46	Automated typing of red blood cell and platelet antigens: a whole-genome sequencing study. Lancet Haematology,the, 2018, 5, e241-e251.	4.6	70
47	Reconciling newborn screening and a novel splice variant in BTD associated with partial biotinidase deficiency: a BabySeq Project case report. Journal of Physical Education and Sports Management, 2018, 4, a002873.	1.2	7
48	A Model for Genome-First Care: Returning Secondary Genomic Findings to Participants and Their Healthcare Providers in a Large Research Cohort. American Journal of Human Genetics, 2018, 103, 328-337.	6.2	130
49	Juvenile myelomonocytic leukemia-associated variants are associated with neo-natal lethal Noonan syndrome. European Journal of Human Genetics, 2017, 25, 509-511.	2.8	18
50	A curated gene list for reporting results of newborn genomic sequencing. Genetics in Medicine, 2017, 19, 809-818.	2.4	79
51	Electronic health record phenotype in subjects with genetic variants associated with arrhythmogenic right ventricular cardiomyopathy: a study of 30,716 subjects with exome sequencing. Genetics in Medicine, 2017, 19, 1245-1252.	2.4	43
52	A Comparison of Whole Genome Sequencing to Multigene Panel Testing in Hypertrophic Cardiomyopathy Patients. Circulation: Cardiovascular Genetics, 2017, 10, .	5.1	62
53	The Impact of Whole-Genome Sequencing on the Primary Care and Outcomes of Healthy Adult Patients. Annals of Internal Medicine, 2017, 167, 159.	3.9	145
54	Using large sequencing data sets to refine intragenic disease regions and prioritize clinical variant interpretation. Genetics in Medicine, 2017, 19, 496-504.	2.4	15

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55	Bioinformatics Workflow for Clinical Whole Genome Sequencing at Partners HealthCare Personalized Medicine. Journal of Personalized Medicine, 2016, 6, 12.	2.5	22
56	Information Technology Support for Clinical Genetic Testing within an Academic Medical Center. Journal of Personalized Medicine, 2016, 6, 4.	2.5	7
57	Detecting Copy Number Variation via Next Generation Technology. Current Genetic Medicine Reports, 2016, 4, 74-85.	1.9	20
58	Retrospective study of prenatal ultrasound findings in newborns with a Noonan spectrum disorder. Prenatal Diagnosis, 2016, 36, 418-423.	2.3	25
59	Distribution and clinical impact of functional variants in 50,726 whole-exome sequences from the DiscovEHR study. Science, 2016, 354, .	12.6	464
60	Navigating highly homologous genes in a molecular diagnostic setting: a resource for clinical next-generation sequencing. Genetics in Medicine, 2016, 18, 1282-1289.	2.4	170
61	Performance of ACMG-AMP Variant-Interpretation Guidelines among Nine Laboratories in the Clinical Sequencing Exploratory Research Consortium. American Journal of Human Genetics, 2016, 98, 1067-1076.	6.2	432
62	Clinical Sequencing Exploratory Research Consortium: Accelerating Evidence-Based Practice of Genomic Medicine. American Journal of Human Genetics, 2016, 98, 1051-1066.	6.2	137
63	â€œBig dataâ€•gets personal. Science Translational Medicine, 2016, 8, 322fs3-3fs3.	12.4	4
64	Next generation sequencingâ€based copy number analysis reveals low prevalence of deletions and duplications in 46 genes associated with genetic cardiomyopathies. Molecular Genetics & Genomic Medicine, 2016, 4, 143-151.	1.2	29
65	VisCap: inference and visualization of germ-line copy-number variants from targeted clinical sequencing data. Genetics in Medicine, 2016, 18, 712-719.	2.4	61
66	Inherited CHST11/MIR3922 deletion is associated with a novel recessive syndrome presenting with skeletal malformation and malignant lymphoproliferative disease. Molecular Genetics & Genomic Medicine, 2015, 3, 413-423.	1.2	11
67	Canadian Open Genetics Repository (COGR): a unified clinical genomics database as a community resource for standardising and sharing genetic interpretations. Journal of Medical Genetics, 2015, 52, 438-445.	3.2	27
68	Results of clinical genetic testing of 2,912 probands with hypertrophic cardiomyopathy: expanded panels offer limited additional sensitivity. Genetics in Medicine, 2015, 17, 880-888.	2.4	344
69	A systematic approach to the reporting of medically relevant findings from whole genome sequencing. BMC Medical Genetics, 2014, 15, 134.	2.1	84
70	An international effort towards developing standards for best practices in analysis, interpretation and reporting of clinical genome sequencing results in the CLARITY Challenge. Genome Biology, 2014, 15, R53.	9.6	101
71	Pathology informatics fellowship training: Focus on molecular pathology. Journal of Pathology Informatics, 2014, 5, 11.	1.7	6
72	The landscape of genetic variation in dilated cardiomyopathy as surveyed by clinical DNA sequencing. Genetics in Medicine, 2014, 16, 601-608.	2.4	284

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73	New Molecular Genetic Tests in the Diagnosis of Heart Disease. Clinics in Laboratory Medicine, 2014, 34, 137-156.	1.4	9
74	Designing Algorithms for Determining Significance of DNA Missense Changes. Methods in Molecular Biology, 2014, 1168, 251-262.	0.9	0
75	A systematic approach to assessing the clinical significance of genetic variants. Clinical Genetics, 2013, 84, 453-463.	2.0	153
76	A survey of informatics approaches to whole-exome and whole-genome clinical reporting in the electronic health record. Genetics in Medicine, 2013, 15, 824-832.	2.4	62
77	American College of Medical Genetics and Genomics technical standards and guidelines: microarray analysis for chromosome abnormalities in neoplastic disorders. Genetics in Medicine, 2013, 15, 484-494.	2.4	54
78	Gain-of-function mutations in the mechanically activated ion channel PIEZO2 cause a subtype of Distal Arthrogryposis. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, 4667-4672.	7.1	193
79	A GC-Wave Correction Algorithm that Improves the Analytical Performance of aCGH. Journal of Molecular Diagnostics, 2012, 14, 550-559.	2.8	9
80	Development and Validation of a Computational Method for Assessment of Missense Variants in Hypertrophic Cardiomyopathy. American Journal of Human Genetics, 2011, 88, 183-192.	6.2	73
81	Dynamic, mating-induced gene expression changes in female head and brain tissues of Drosophila melanogaster. BMC Genomics, 2010, 11, 541.	2.8	60
82	Somatic, germline and sex hierarchy regulated gene expression during Drosophila metamorphosis. BMC Genomics, 2009, 10, 80.	2.8	49
83	Ecdysone Receptor Acts in fruitless- Expressing Neurons to Mediate Drosophila Courtship Behaviors. Current Biology, 2009, 19, 1447-1452.	3.9	57