

Mathew S Lebo

List of Publications by Citations

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Version: 2024-04-27

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

82
papers

3,641
citations

32
h-index

59
g-index

107
ext. papers

4,926
ext. citations

8.7
avg, IF

4.71
L-index

#	Paper	IF	Citations
82	Distribution and clinical impact of functional variants in 50,726 whole-exome sequences from the DiscovEHR study. <i>Science</i> , 2016 , 354,	33.3	320
81	Performance of ACMG-AMP Variant-Interpretation Guidelines among Nine Laboratories in the Clinical Sequencing Exploratory Research Consortium. <i>American Journal of Human Genetics</i> , 2016 , 98, 1067-1076	11	271
80	Polygenic Prediction of Weight and Obesity Trajectories from Birth to Adulthood. <i>Cell</i> , 2019 , 177, 587-596.e9	36.9	265
79	Results of clinical genetic testing of 2,912 probands with hypertrophic cardiomyopathy: expanded panels offer limited additional sensitivity. <i>Genetics in Medicine</i> , 2015 , 17, 880-8	8.1	236
78	The landscape of genetic variation in dilated cardiomyopathy as surveyed by clinical DNA sequencing. <i>Genetics in Medicine</i> , 2014 , 16, 601-8	8.1	215
77	Gain-of-function mutations in the mechanically activated ion channel PIEZO2 cause a subtype of Distal Arthrogryposis. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2013 , 110, 4667-72	11.5	151
76	A systematic approach to assessing the clinical significance of genetic variants. <i>Clinical Genetics</i> , 2013 , 84, 453-63	4	124
75	The Impact of Whole-Genome Sequencing on the Primary Care and Outcomes of Healthy Adult Patients: A Pilot Randomized Trial. <i>Annals of Internal Medicine</i> , 2017 , 167, 159-169	8	112
74	Clinical Sequencing Exploratory Research Consortium: Accelerating Evidence-Based Practice of Genomic Medicine. <i>American Journal of Human Genetics</i> , 2016 , 98, 1051-1066	11	107
73	Navigating highly homologous genes in a molecular diagnostic setting: a resource for clinical next-generation sequencing. <i>Genetics in Medicine</i> , 2016 , 18, 1282-1289	8.1	103
72	Exome Sequencing-Based Screening for BRCA1/2 Expected Pathogenic Variants Among Adult Biobank Participants. <i>JAMA Network Open</i> , 2018 , 1, e182140	10.4	98
71	Polygenic background modifies penetrance of monogenic variants for tier 1 genomic conditions. <i>Nature Communications</i> , 2020 , 11, 3635	17.4	88
70	An international effort towards developing standards for best practices in analysis, interpretation and reporting of clinical genome sequencing results in the CLARITY Challenge. <i>Genome Biology</i> , 2014 , 15, R53	18.3	86
69	Interpretation of Genomic Sequencing Results in Healthy and Ill Newborns: Results from the BabySeq Project. <i>American Journal of Human Genetics</i> , 2019 , 104, 76-93	11	86
68	A Model for Genome-First Care: Returning Secondary Genomic Findings to Participants and Their Healthcare Providers in a Large Research Cohort. <i>American Journal of Human Genetics</i> , 2018 , 103, 328-337	11	72
67	A systematic approach to the reporting of medically relevant findings from whole genome sequencing. <i>BMC Medical Genetics</i> , 2014 , 15, 134	2.1	66
66	Harmonizing Clinical Sequencing and Interpretation for the eMERGE III Network. <i>American Journal of Human Genetics</i> , 2019 , 105, 588-605	11	63

65	Development and validation of a computational method for assessment of missense variants in hypertrophic cardiomyopathy. <i>American Journal of Human Genetics</i> , 2011 , 88, 183-92	11	60
64	A survey of informatics approaches to whole-exome and whole-genome clinical reporting in the electronic health record. <i>Genetics in Medicine</i> , 2013 , 15, 824-32	8.1	56
63	Ecdysone receptor acts in fruitless- expressing neurons to mediate drosophila courtship behaviors. <i>Current Biology</i> , 2009 , 19, 1447-52	6.3	50
62	A curated gene list for reporting results of newborn genomic sequencing. <i>Genetics in Medicine</i> , 2017 , 19, 809-818	8.1	47
61	Dynamic, mating-induced gene expression changes in female head and brain tissues of <i>Drosophila melanogaster</i> . <i>BMC Genomics</i> , 2010 , 11, 541	4.5	47
60	VisCap: inference and visualization of germ-line copy-number variants from targeted clinical sequencing data. <i>Genetics in Medicine</i> , 2016 , 18, 712-9	8.1	46
59	A Comparison of Whole Genome Sequencing to Multigene Panel Testing in Hypertrophic Cardiomyopathy Patients. <i>Circulation: Cardiovascular Genetics</i> , 2017 , 10,		45
58	American College of Medical Genetics and Genomics technical standards and guidelines: microarray analysis for chromosome abnormalities in neoplastic disorders. <i>Genetics in Medicine</i> , 2013 , 15, 484-94	8.1	45
57	Somatic, germline and sex hierarchy regulated gene expression during <i>Drosophila</i> metamorphosis. <i>BMC Genomics</i> , 2009 , 10, 80	4.5	44
56	Automated typing of red blood cell and platelet antigens: a whole-genome sequencing study. <i>Lancet Haematology</i> , 2018 , 5, e241-e251	14.6	35
55	Electronic health record phenotype in subjects with genetic variants associated with arrhythmogenic right ventricular cardiomyopathy: a study of 30,716 subjects with exome sequencing. <i>Genetics in Medicine</i> , 2017 , 19, 1245-1252	8.1	33
54	Early cancer diagnoses through BRCA1/2 screening of unselected adult biobank participants. <i>Genetics in Medicine</i> , 2018 , 20, 554-558	8.1	33
53	Parental interest in genomic sequencing of newborns: enrollment experience from the BabySeq Project. <i>Genetics in Medicine</i> , 2019 , 21, 622-630	8.1	33
52	The eMERGE genotype set of 83,717 subjects imputed to ~40 million variants genome wide and association with the herpes zoster medical record phenotype. <i>Genetic Epidemiology</i> , 2019 , 43, 63-81	2.6	32
51	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. <i>JAMA Network Open</i> , 2020 , 3, e203959	10.4	31
50	A Rigorous Interlaboratory Examination of the Need to Confirm Next-Generation Sequencing-Detected Variants with an Orthogonal Method in Clinical Genetic Testing. <i>Journal of Molecular Diagnostics</i> , 2019 , 21, 318-329	5.1	28
49	Next generation sequencing-based copy number analysis reveals low prevalence of deletions and duplications in 46 genes associated with genetic cardiomyopathies. <i>Molecular Genetics & Genomic Medicine</i> , 2016 , 4, 143-51	2.3	27
48	Canadian Open Genetics Repository (COGR): a unified clinical genomics database as a community resource for standardising and sharing genetic interpretations. <i>Journal of Medical Genetics</i> , 2015 , 52, 438-45	5.8	23

47	Analyzing and Reanalyzing the Genome: Findings from the MedSeq Project. <i>American Journal of Human Genetics</i> , 2019 , 105, 177-188	11	22
46	Best practices for the analytical validation of clinical whole-genome sequencing intended for the diagnosis of germline disease. <i>Npj Genomic Medicine</i> , 2020 , 5, 47	6.2	22
45	Variant classification changes over time in BRCA1 and BRCA2. <i>Genetics in Medicine</i> , 2019 , 21, 2248-2254	8.1	20
44	Data sharing as a national quality improvement program: reporting on BRCA1 and BRCA2 variant-interpretation comparisons through the Canadian Open Genetics Repository (COGR). <i>Genetics in Medicine</i> , 2018 , 20, 294-302	8.1	20
43	Retrospective study of prenatal ultrasound findings in newborns with a Noonan spectrum disorder. <i>Prenatal Diagnosis</i> , 2016 , 36, 418-23	3.2	18
42	Rare Genetic Variants Associated With Sudden Cardiac Death in Adults. <i>Journal of the American College of Cardiology</i> , 2019 , 74, 2623-2634	15.1	17
41	Hematopoietic mosaic chromosomal alterations increase the risk for diverse types of infection. <i>Nature Medicine</i> , 2021 , 27, 1012-1024	50.5	16
40	Bioinformatics Workflow for Clinical Whole Genome Sequencing at Partners HealthCare Personalized Medicine. <i>Journal of Personalized Medicine</i> , 2016 , 6,	3.6	16
39	Characterizing reduced coverage regions through comparison of exome and genome sequencing data across 10 centers. <i>Genetics in Medicine</i> , 2018 , 20, 855-866	8.1	16
38	Automated typing of red blood cell and platelet antigens from whole exome sequences. <i>Transfusion</i> , 2019 , 59, 3253-3263	2.9	15
37	Detecting Copy Number Variation via Next Generation Technology. <i>Current Genetic Medicine Reports</i> , 2016 , 4, 74-85	2.2	14
36	Using large sequencing data sets to refine intragenic disease regions and prioritize clinical variant interpretation. <i>Genetics in Medicine</i> , 2017 , 19, 496-504	8.1	11
35	Discordant results between conventional newborn screening and genomic sequencing in the BabySeq Project. <i>Genetics in Medicine</i> , 2021 , 23, 1372-1375	8.1	11
34	Designing and Implementing NGS Tests for Inherited Disorders: A Practical Framework with Step-by-Step Guidance for Clinical Laboratories. <i>Journal of Molecular Diagnostics</i> , 2019 , 21, 369-374	5.1	11
33	Juvenile myelomonocytic leukemia-associated variants are associated with neo-natal lethal Noonan syndrome. <i>European Journal of Human Genetics</i> , 2017 , 25, 509-511	5.3	9
32	A whole genome approach for discovering the genetic basis of blood group antigens: independent confirmation for P1 and Xg. <i>Transfusion</i> , 2019 , 59, 908-915	2.9	9
31	Considerations for clinical curation, classification, and reporting of low-penetrance and low effect size variants associated with disease risk. <i>Genetics in Medicine</i> , 2019 , 21, 2765-2773	8.1	8
30	Inherited CHST11/MIR3922 deletion is associated with a novel recessive syndrome presenting with skeletal malformation and malignant lymphoproliferative disease. <i>Molecular Genetics & Genomic Medicine</i> , 2015 , 3, 413-23	2.3	8

29	Psychosocial Effect of Newborn Genomic Sequencing on Families in the BabySeq Project: A Randomized Clinical Trial. <i>JAMA Pediatrics</i> , 2021 , 175, 1132-1141	8.3	8
28	Genetic variant pathogenicity prediction trained using disease-specific clinical sequencing data sets. <i>Genome Research</i> , 2019 , 29, 1144-1151	9.7	7
27	Expanding the Noonan spectrum/RASopathy NGS panel: Benefits of adding NF1 and SPRED1. <i>Molecular Genetics & Genomic Medicine</i> , 2020 , 8, e1180	2.3	7
26	A GC-wave correction algorithm that improves the analytical performance of aCGH. <i>Journal of Molecular Diagnostics</i> , 2012 , 14, 550-9	5.1	7
25	Information Technology Support for Clinical Genetic Testing within an Academic Medical Center. <i>Journal of Personalized Medicine</i> , 2016 , 6,	3.6	7
24	Design and Reporting Considerations for Genetic Screening Tests. <i>Journal of Molecular Diagnostics</i> , 2020 , 22, 599-609	5.1	6
23	New molecular genetic tests in the diagnosis of heart disease. <i>Clinics in Laboratory Medicine</i> , 2014 , 34, 137-56, vii-viii	2.1	6
22	Pathology informatics fellowship training: Focus on molecular pathology. <i>Journal of Pathology Informatics</i> , 2014 , 5, 11	4.4	6
21	Polygenic background modifies penetrance of monogenic variants conferring risk for coronary artery disease, breast cancer, or colorectal cancer		5
20	Hematopoietic mosaic chromosomal alterations and risk for infection among 767,891 individuals without blood cancer 2020 ,		5
19	An assessment of the role of vinculin loss of function variants in inherited cardiomyopathy. <i>Human Mutation</i> , 2020 , 41, 1577-1587	4.7	4
18	Quantifying Downstream Healthcare Utilization in Studies of Genomic Testing. <i>Value in Health</i> , 2020 , 23, 559-565	3.3	4
17	Reconciling newborn screening and a novel splice variant in associated with partial biotinidase deficiency: a BabySeq Project case report. <i>Journal of Physical Education and Sports Management</i> , 2018 , 4,	2.8	4
16	"Big Data" Gets Personal. <i>Science Translational Medicine</i> , 2016 , 8, 322fs3-3fs3	17.5	3
15	Returning actionable genomic results in a research biobank: Analytic validity, clinical implementation, and resource utilization. <i>American Journal of Human Genetics</i> , 2021 , 108, 2224-2237	11	3
14	Data sharing to improve concordance in variant interpretation across laboratories: results from the Canadian Open Genetics Repository. <i>Journal of Medical Genetics</i> , 2021 ,	5.8	3
13	Randomized prospective evaluation of genome sequencing versus standard-of-care as a first molecular diagnostic test. <i>Genetics in Medicine</i> , 2021 , 23, 1689-1696	8.1	3
12	Overcoming the challenges of interpreting complex and uncommon RH alleles from whole genomes. <i>Vox Sanguinis</i> , 2020 , 115, 790-801	3.1	2

11	A Model for Genome-First Care: Returning Secondary Genomic Findings to Participants and Their Healthcare Providers in a Large Research Cohort		2
10	Multiple GYPB gene deletions associated with the U- phenotype in those of African ancestry. <i>Transfusion</i> , 2020 , 60, 1294-1307	2.9	1
9	Reanalysis of eMERGE phase III sequence variants in 10,500 participants and infrastructure to support the automated return of knowledge updates.. <i>Genetics in Medicine</i> , 2021 ,	8.1	1
8	Bioinformatics in Clinical Genomic Sequencing. <i>Clinics in Laboratory Medicine</i> , 2020 , 40, 163-187	2.1	1
7	Airmen and health-care providersTattitudes toward the use of genomic sequencing in the US Air Force: findings from the MilSeq Project. <i>Genetics in Medicine</i> , 2020 , 22, 2003-2010	8.1	1
6	Harmonizing the Collection of Clinical Data on Genetic Testing Requisition Forms to Enhance Variant Interpretation in Hypertrophic Cardiomyopathy (HCM): A Study from the ClinGen Cardiomyopathy Variant Curation Expert Panel. <i>Journal of Molecular Diagnostics</i> , 2021 , 23, 589-598	5.1	1
5	A framework for automated gene selection in genomic applications. <i>Genetics in Medicine</i> , 2021 , 23, 1993-1997	8.1	1
4	Neptune: an environment for the delivery of genomic medicine. <i>Genetics in Medicine</i> , 2021 , 23, 1838-1848	8.1	1
3	Bioinformatics in Clinical Genomic Sequencing. <i>Advances in Molecular Pathology</i> , 2018 , 1, 9-26	0.3	1
2	Validation of a Trans-Ancestry Polygenic Risk Score for Type 2 Diabetes in Diverse Populations		1
1	Designing algorithms for determining significance of DNA missense changes. <i>Methods in Molecular Biology</i> , 2014 , 1168, 251-62	1.4	