Mathew S Lebo

List of Publications by Year in Descending Order

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	Returning actionable genomic results in a research biobank: Analytic validity, clinical implementation, and resource utilization. *American Journal of Human Genetics*, **2021**, 108, 2224-2237	11	3
81	Reanalysis of eMERGE phase III sequence variants in 10,500 participants and infrastructure to support the automated return of knowledge updates.. *Genetics in Medicine*, **2021**,	8.1	1
80	Discordant results between conventional newborn screening and genomic sequencing in the BabySeq Project. *Genetics in Medicine*, **2021**, 23, 1372-1375	8.1	11
79	Data sharing to improve concordance in variant interpretation across laboratories: results from the Canadian Open Genetics Repository. *Journal of Medical Genetics*, **2021**,	5.8	3
78	Randomized prospective evaluation of genome sequencing versus standard-of-care as a first molecular diagnostic test. *Genetics in Medicine*, **2021**, 23, 1689-1696	8.1	3
77	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. *Journal of Molecular Diagnostics*, **2021**, 23, 589-598	5.1	1
76	A framework for automated gene selection in genomic applications. *Genetics in Medicine*, **2021**, 23, 1993-1997	8.1	1
75	Hematopoietic mosaic chromosomal alterations increase the risk for diverse types of infection. *Nature Medicine*, **2021**, 27, 1012-1024	50.5	16
74	Neptune: an environment for the delivery of genomic medicine. *Genetics in Medicine*, **2021**, 23, 1838-1848	8.1	1
73	Psychosocial Effect of Newborn Genomic Sequencing on Families in the BabySeq Project: A Randomized Clinical Trial. *JAMA Pediatrics*, **2021**, 175, 1132-1141	8.3	8
72	Multiple GYPB gene deletions associated with the U- phenotype in those of African ancestry. *Transfusion*, **2020**, 60, 1294-1307	2.9	1
71	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	10.4	31
70	Overcoming the challenges of interpreting complex and uncommon RH alleles from whole genomes. *Vox Sanguinis*, **2020**, 115, 790-801	3.1	2
69	An assessment of the role of vinculin loss of function variants in inherited cardiomyopathy. *Human Mutation*, **2020**, 41, 1577-1587	4.7	4
68	Expanding the Noonan spectrum/RASopathy NGS panel: Benefits of adding NF1 and SPRED1. *Molecular Genetics & Genomic Medicine*, **2020**, 8, e1180	2.3	7
67	Design and Reporting Considerations for Genetic Screening Tests. *Journal of Molecular Diagnostics*, **2020**, 22, 599-609	5.1	6
66	Quantifying Downstream Healthcare Utilization in Studies of Genomic Testing. *Value in Health*, **2020**, 23, 559-565	3.3	4

65	Bioinformatics in Clinical Genomic Sequencing. *Clinics in Laboratory Medicine*, **2020**, 40, 163-187	2.1	1
64	Hematopoietic mosaic chromosomal alterations and risk for infection among 767,891 individuals without blood cancer **2020**,		5
63	Best practices for the analytical validation of clinical whole-genome sequencing intended for the diagnosis of germline disease. *Npj Genomic Medicine*, **2020**, 5, 47	6.2	22
62	Airmen and health-care providersTattitudes toward the use of genomic sequencing in the US Air Force: findings from the MilSeq Project. *Genetics in Medicine*, **2020**, 22, 2003-2010	8.1	1
61	Polygenic background modifies penetrance of monogenic variants for tier 1 genomic conditions. *Nature Communications*, **2020**, 11, 3635	17.4	88
60	Harmonizing Clinical Sequencing and Interpretation for the eMERGE III Network. *American Journal of Human Genetics*, **2019**, 105, 588-605	11	63
59	A Rigorous Interlaboratory Examination of the Need to Confirm Next-Generation Sequencing-Detected Variants with an Orthogonal Method□n Clinical Genetic Testing. *Journal of Molecular Diagnostics*, **2019**, 21, 318-329	5.1	28
58	Genetic variant pathogenicity prediction trained using disease-specific clinical sequencing data sets. *Genome Research*, **2019**, 29, 1144-1151	9.7	7
57	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	8.1	8
56	Polygenic Prediction of Weight and Obesity Trajectories from Birth to Adulthood. *Cell*, **2019**, 177, 587-596.e9	36.2	265
55	Variant classification changes over time in BRCA1 and BRCA2. *Genetics in Medicine*, **2019**, 21, 2248-2254	8.1	20
54	Automated typing of red blood cell and platelet antigens from whole exome sequences. *Transfusion*, **2019**, 59, 3253-3263	2.9	15
53	Analyzing and Reanalyzing the Genome: Findings from the MedSeq Project. *American Journal of Human Genetics*, **2019**, 105, 177-188	11	22
52	Rare Genetic Variants Associated With Sudden Cardiac Death in Adults. *Journal of the American College of Cardiology*, **2019**, 74, 2623-2634	15.1	17
51	Parental interest in genomic sequencing of newborns: enrollment experience from the BabySeq Project. *Genetics in Medicine*, **2019**, 21, 622-630	8.1	33
50	Interpretation of Genomic Sequencing Results in Healthy and Ill Newborns: Results from the BabySeq Project. *American Journal of Human Genetics*, **2019**, 104, 76-93	11	86
49	Designing and Implementing NGS Tests for Inherited Disorders: A Practical Framework with Step-by-Step Guidance for Clinical Laboratories. *Journal of Molecular Diagnostics*, **2019**, 21, 369-374	5.1	11
48	A whole genome approach for discovering the genetic basis of blood group antigens: independent confirmation for P1 and Xg. *Transfusion*, **2019**, 59, 908-915	2.9	9

47	The eMERGE genotype set of 83,717 subjects imputed to ~40 million variants genome wide and association with the herpes zoster medical record phenotype. *Genetic Epidemiology*, **2019**, 43, 63-81	2.6	32
46	Early cancer diagnoses through BRCA1/2 screening of unselected adult biobank participants. *Genetics in Medicine*, **2018**, 20, 554-558	8.1	33
45	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	8.1	20
44	Reconciling newborn screening and a novel splice variant in associated with partial biotinidase deficiency: a BabySeq Project case report. *Journal of Physical Education and Sports Management*, **2018**, 4,	2.8	4
43	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	11	72
42	Characterizing reduced coverage regions through comparison of exome and genome sequencing data across 10 centers. *Genetics in Medicine*, **2018**, 20, 855-866	8.1	16
41	Exome Sequencing-Based Screening for BRCA1/2 Expected Pathogenic Variants Among Adult Biobank Participants. *JAMA Network Open*, **2018**, 1, e182140	10.4	98
40	Bioinformatics in Clinical Genomic Sequencing. *Advances in Molecular Pathology*, **2018**, 1, 9-26	0.3	1
39	Automated typing of red blood cell and platelet antigens: a whole-genome sequencing study. *Lancet Haematology,the*, **2018**, 5, e241-e251	14.6	35
38	Juvenile myelomonocytic leukemia-associated variants are associated with neo-natal lethal Noonan syndrome. *European Journal of Human Genetics*, **2017**, 25, 509-511	5.3	9
37	A curated gene list for reporting results of newborn genomic sequencing. *Genetics in Medicine*, **2017**, 19, 809-818	8.1	47
36	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	8.1	33
35	A Comparison of Whole Genome Sequencing to Multigene Panel Testing in Hypertrophic Cardiomyopathy Patients. *Circulation: Cardiovascular Genetics*, **2017**, 10,		45
34	The Impact of Whole-Genome Sequencing on the Primary Care and Outcomes of Healthy Adult Patients: A Pilot Randomized Trial. *Annals of Internal Medicine*, **2017**, 167, 159-169	8	112
33	Using large sequencing data sets to refine intragenic disease regions and prioritize clinical variant interpretation. *Genetics in Medicine*, **2017**, 19, 496-504	8.1	11
32	"Big Data" Gets Personal. *Science Translational Medicine*, **2016**, 8, 322fs3-3fs3	17.5	3
31	Next generation sequencing-based copy number analysis reveals low prevalence of deletions and duplications in 46 genes associated with genetic cardiomyopathies. *Molecular Genetics & Genomic Medicine*, **2016**, 4, 143-51	2.3	27
30	VisCap: inference and visualization of germ-line copy-number variants from targeted clinical sequencing data. *Genetics in Medicine*, **2016**, 18, 712-9	8.1	46

29	Bioinformatics Workflow for Clinical Whole Genome Sequencing at Partners HealthCare Personalized Medicine. *Journal of Personalized Medicine*, **2016**, 6,	3.6	16
28	Information Technology Support for Clinical Genetic Testing within an Academic Medical Center. *Journal of Personalized Medicine*, **2016**, 6,	3.6	7
27	Detecting Copy Number Variation via Next Generation Technology. *Current Genetic Medicine Reports*, **2016**, 4, 74-85	2.2	14
26	Retrospective study of prenatal ultrasound findings in newborns with a Noonan spectrum disorder. *Prenatal Diagnosis*, **2016**, 36, 418-23	3.2	18
25	Distribution and clinical impact of functional variants in 50,726 whole-exome sequences from the DiscovEHR study. *Science*, **2016**, 354,	33.3	320
24	Navigating highly homologous genes in a molecular diagnostic setting: a resource for clinical next-generation sequencing. *Genetics in Medicine*, **2016**, 18, 1282-1289	8.1	103
23	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	11	271
22	Clinical Sequencing Exploratory Research Consortium: Accelerating Evidence-Based Practice of Genomic Medicine. *American Journal of Human Genetics*, **2016**, 98, 1051-1066	11	107
21	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, 413-23	2.3	8
20	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, 438-45	5.8	23
19	Results of clinical genetic testing of 2,912 probands with hypertrophic cardiomyopathy: expanded panels offer limited additional sensitivity. *Genetics in Medicine*, **2015**, 17, 880-8	8.1	236
18	The landscape of genetic variation in dilated cardiomyopathy as surveyed by clinical DNA sequencing. *Genetics in Medicine*, **2014**, 16, 601-8	8.1	215
17	New molecular genetic tests in the diagnosis of heart disease. *Clinics in Laboratory Medicine*, **2014**, 34, 137-56, vii-viii	2.1	6
16	A systematic approach to the reporting of medically relevant findings from whole genome sequencing. *BMC Medical Genetics*, **2014**, 15, 134	2.1	66
15	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	18.3	86
14	Pathology informatics fellowship training: Focus on molecular pathology. *Journal of Pathology Informatics*, **2014**, 5, 11	4.4	6
13	Designing algorithms for determining significance of DNA missense changes. *Methods in Molecular Biology*, **2014**, 1168, 251-62	1.4	
12	A systematic approach to assessing the clinical significance of genetic variants. *Clinical Genetics*, **2013**, 84, 453-63	4	124

11	A survey of informatics approaches to whole-exome and whole-genome clinical reporting in the electronic health record. *Genetics in Medicine*, **2013**, 15, 824-32	8.1	56
10	American College of Medical Genetics and Genomics technical standards and guidelines: microarray analysis for chromosome abnormalities in neoplastic disorders. *Genetics in Medicine*, **2013**, 15, 484-94	8.1	45
9	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, 4667-72	11.5	151
8	A GC-wave correction algorithm that improves the analytical performance of aCGH. *Journal of Molecular Diagnostics*, **2012**, 14, 550-9	5.1	7
7	Development and validation of a computational method for assessment of missense variants in hypertrophic cardiomyopathy. *American Journal of Human Genetics*, **2011**, 88, 183-92	11	60
6	Dynamic, mating-induced gene expression changes in female head and brain tissues of Drosophila melanogaster. *BMC Genomics*, **2010**, 11, 541	4.5	47
5	Somatic, germline and sex hierarchy regulated gene expression during Drosophila metamorphosis. *BMC Genomics*, **2009**, 10, 80	4.5	44
4	Ecdysone receptor acts in fruitless- expressing neurons to mediate drosophila courtship behaviors. *Current Biology*, **2009**, 19, 1447-52	6.3	50
3	A Model for Genome-First Care: Returning Secondary Genomic Findings to Participants and Their Healthcare Providers in a Large Research Cohort		2
2	Polygenic background modifies penetrance of monogenic variants conferring risk for coronary artery disease, breast cancer, or colorectal cancer		5
1	Validation of a Trans-Ancestry Polygenic Risk Score for Type 2 Diabetes in Diverse Populations		1