

Matthew T Wheeler Iii

List of Publications by Year in descending order

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Version: 2024-02-01

101
papers

8,935
citations

81900

39
h-index

45317

90
g-index

109
all docs

109
docs citations

109
times ranked

14191
citing authors

#	ARTICLE	IF	CITATIONS
1	The ubiquitin-modifying enzyme A20 is required for termination of Toll-like receptor responses. <i>Nature Immunology</i> , 2004, 5, 1052-1060.	14.5	1,016
2	Clinical assessment incorporating a personal genome. <i>Lancet, The</i> , 2010, 375, 1525-1535.	13.7	637
3	Abnormal Calcium Handling Properties Underlie Familial Hypertrophic Cardiomyopathy Pathology in Patient-Specific Induced Pluripotent Stem Cells. <i>Cell Stem Cell</i> , 2013, 12, 101-113.	11.1	584
4	Expansion of the Human Phenotype Ontology (HPO) knowledge base and resources. <i>Nucleic Acids Research</i> , 2019, 47, D1018-D1027.	14.5	539
5	Mavacamten for treatment of symptomatic obstructive hypertrophic cardiomyopathy (EXPLORER-HCM): a randomised, double-blind, placebo-controlled, phase 3 trial. <i>Lancet, The</i> , 2020, 396, 759-769.	13.7	481
6	Accuracy in Wrist-Worn, Sensor-Based Measurements of Heart Rate and Energy Expenditure in a Diverse Cohort. <i>Journal of Personalized Medicine</i> , 2017, 7, 3.	2.5	420
7	Clinical Interpretation and Implications of Whole-Genome Sequencing. <i>JAMA - Journal of the American Medical Association</i> , 2014, 311, 1035.	7.4	398
8	Effect of Genetic Diagnosis on Patients with Previously Undiagnosed Disease. <i>New England Journal of Medicine</i> , 2018, 379, 2131-2139.	27.0	261
9	Identification of rare-disease genes using blood transcriptome sequencing and large control cohorts. <i>Nature Medicine</i> , 2019, 25, 911-919.	30.7	221
10	Cost-Effectiveness of Preparticipation Screening for Prevention of Sudden Cardiac Death in Young Athletes. <i>Annals of Internal Medicine</i> , 2010, 152, 276.	3.9	211
11	Challenges in the clinical application of whole-genome sequencing. <i>Lancet, The</i> , 2010, 375, 1749-1751.	13.7	207
12	Interpretation of the Electrocardiogram of Young Athletes. <i>Circulation</i> , 2011, 124, 746-757.	1.6	204
13	A public resource facilitating clinical use of genomes. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2012, 109, 11920-11927.	7.1	194
14	Long-read genome sequencing identifies causal structural variation in a Mendelian disease. <i>Genetics in Medicine</i> , 2018, 20, 159-163.	2.4	189
15	MARRVEL: Integration of Human and Model Organism Genetic Resources to Facilitate Functional Annotation of the Human Genome. <i>American Journal of Human Genetics</i> , 2017, 100, 843-853.	6.2	181
16	Episodic coronary artery vasospasm and hypertension develop in the absence of Sur2 KATP channels. <i>Journal of Clinical Investigation</i> , 2002, 110, 203-208.	8.2	173
17	Effect of Moderate-Intensity Exercise Training on Peak Oxygen Consumption in Patients With Hypertrophic Cardiomyopathy. <i>JAMA - Journal of the American Medical Association</i> , 2017, 317, 1349.	7.4	160
18	Molecular Transducers of Physical Activity Consortium (MoTrPAC): Mapping the Dynamic Responses to Exercise. <i>Cell</i> , 2020, 181, 1464-1474.	28.9	147

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19	The Undiagnosed Diseases Network: Accelerating Discovery about Health and Disease. <i>American Journal of Human Genetics</i> , 2017, 100, 185-192.	6.2	142
20	Phased Whole-Genome Genetic Risk in a Family Quartet Using a Major Allele Reference Sequence. <i>PLoS Genetics</i> , 2011, 7, e1002280.	3.5	137
21	Episodic coronary artery vasospasm and hypertension develop in the absence of Sur2 KATP channels. <i>Journal of Clinical Investigation</i> , 2002, 110, 203-208.	8.2	129
22	Medical implications of technical accuracy in genome sequencing. <i>Genome Medicine</i> , 2016, 8, 24.	8.2	123
23	Regional Variation in <i>RBM20</i> Causes a Highly Penetrant Arrhythmogenic Cardiomyopathy. <i>Circulation: Heart Failure</i> , 2019, 12, e005371.	3.9	96
24	Gene Coexpression Network Topology of Cardiac Development, Hypertrophy, and Failure. <i>Circulation: Cardiovascular Genetics</i> , 2011, 4, 26-35.	5.1	88
25	Physical Activity and Other Health Behaviors in Adults With Hypertrophic Cardiomyopathy. <i>American Journal of Cardiology</i> , 2013, 111, 1034-1039.	1.6	86
26	zeta-Sarcoglycan, a novel component of the sarcoglycan complex, is reduced in muscular dystrophy. <i>Human Molecular Genetics</i> , 2002, 11, 2147-2154.	2.9	82
27	Addition of the Electrocardiogram to the Preparticipation Examination of College Athletes. <i>Clinical Journal of Sport Medicine</i> , 2010, 20, 98-105.	1.8	79
28	DNA Sequencing. <i>Circulation</i> , 2012, 125, 931-944.	1.6	72
29	ClinPhen extracts and prioritizes patient phenotypes directly from medical records to expedite genetic disease diagnosis. <i>Genetics in Medicine</i> , 2019, 21, 1585-1593.	2.4	67
30	Smooth muscle cell "extrinsic" vascular spasm arises from cardiomyocyte degeneration in sarcoglycan-deficient cardiomyopathy. <i>Journal of Clinical Investigation</i> , 2004, 113, 668-675.	8.2	64
31	Accuracy of Smartphone Camera Applications for Detecting Atrial Fibrillation. <i>JAMA Network Open</i> , 2020, 3, e202064.	5.9	62
32	A comprehensive iterative approach is highly effective in diagnosing individuals who are exome negative. <i>Genetics in Medicine</i> , 2019, 21, 161-172.	2.4	60
33	Biallelic Mutations in <i>ATP5F1D</i> , which Encodes a Subunit of ATP Synthase, Cause a Metabolic Disorder. <i>American Journal of Human Genetics</i> , 2018, 102, 494-504.	6.2	59
34	Targeting ferroptosis: A novel therapeutic strategy for the treatment of mitochondrial disease-related epilepsy. <i>PLoS ONE</i> , 2019, 14, e0214250.	2.5	59
35	Personalized Preventive Medicine: Genetics and the Response to Regular Exercise in Preventive Interventions. <i>Progress in Cardiovascular Diseases</i> , 2015, 57, 337-346.	3.1	57
36	The effect of digital physical activity interventions on daily step count: a randomised controlled crossover substudy of the MyHeart Counts Cardiovascular Health Study. <i>The Lancet Digital Health</i> , 2019, 1, e344-e352.	12.3	52

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37	Overexpression of β -Sarcoglycan Induces Severe Muscular Dystrophy. <i>Journal of Biological Chemistry</i> , 2001, 276, 21785-21790.	3.4	51
38	Valsartan in early-stage hypertrophic cardiomyopathy: a randomized phase 2 trial. <i>Nature Medicine</i> , 2021, 27, 1818-1824.	30.7	51
39	Time trajectories in the transcriptomic response to exercise - a meta-analysis. <i>Nature Communications</i> , 2021, 12, 3471.	12.8	48
40	Outcomes in Patients With Cardiac Amyloidosis Undergoing Heart Transplantation. <i>JACC: Heart Failure</i> , 2020, 8, 461-468.	4.1	46
41	Yield of whole exome sequencing in undiagnosed patients facing insurance coverage barriers to genetic testing. <i>Journal of Genetic Counseling</i> , 2019, 28, 1107-1118.	1.6	42
42	Identification of rare and common regulatory variants in pluripotent cells using population-scale transcriptomics. <i>Nature Genetics</i> , 2021, 53, 313-321.	21.4	42
43	Characterization of HNRNPA1 mutations defines diversity in pathogenic mechanisms and clinical presentation. <i>JCI Insight</i> , 2021, 6, .	5.0	38
44	De novo EIF2AK1 and EIF2AK2 Variants Are Associated with Developmental Delay, Leukoencephalopathy, and Neurologic Decompensation. <i>American Journal of Human Genetics</i> , 2020, 106, 570-583.	6.2	37
45	Smooth muscle cell "extrinsic vascular spasm arises from cardiomyocyte degeneration in sarcoglycan-deficient cardiomyopathy. <i>Journal of Clinical Investigation</i> , 2004, 113, 668-675.	8.2	37
46	Sarcoglycans in Vascular Smooth and Striated Muscle. <i>Trends in Cardiovascular Medicine</i> , 2003, 13, 238-243.	4.9	34
47	Secondary Coronary Artery Vasospasm Promotes Cardiomyopathy Progression. <i>American Journal of Pathology</i> , 2004, 164, 1063-1071.	3.8	34
48	Mechanisms of exercise intolerance in patients with hypertrophic cardiomyopathy. <i>American Heart Journal</i> , 2009, 158, e27-e34.	2.7	32
49	Functional nitric oxide synthase mislocalization in cardiomyopathy. <i>Journal of Molecular and Cellular Cardiology</i> , 2004, 36, 213-223.	1.9	31
50	Systems Genomics Identifies a Key Role for Hypocretin/Orexin Receptor-2 in Human Heart Failure. <i>Journal of the American College of Cardiology</i> , 2015, 66, 2522-2533.	2.8	31
51	Exercise restrictions trigger psychological difficulty in active and athletic adults with hypertrophic cardiomyopathy. <i>Open Heart</i> , 2016, 3, e000488.	2.3	29
52	Silencing of MYH7 ameliorates disease phenotypes in human iPSC-cardiomyocytes. <i>Physiological Genomics</i> , 2020, 52, 293-303.	2.3	29
53	Prevalence and Cumulative Risk of Familial Idiopathic Dilated Cardiomyopathy. <i>JAMA - Journal of the American Medical Association</i> , 2022, 327, 454.	7.4	28
54	Cytoskeletal defects in cardiomyopathy. <i>Journal of Molecular and Cellular Cardiology</i> , 2003, 35, 231-241.	1.9	26

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55	Sports genetics moving forward: lessons learned from medical research. <i>Physiological Genomics</i> , 2016, 48, 175-182.	2.3	26
56	Allele-Specific Silencing Ameliorates Restrictive Cardiomyopathy Attributable to a Human Myosin Regulatory Light Chain Mutation. <i>Circulation</i> , 2019, 140, 765-778.	1.6	26
57	The genetics of human performance. <i>Nature Reviews Genetics</i> , 2022, 23, 40-54.	16.3	25
58	Pathologic gene network rewiring implicates PPP1R3A as a central regulator in pressure overload heart failure. <i>Nature Communications</i> , 2019, 10, 2760.	12.8	22
59	Cardiomyopathy is independent of skeletal muscle disease in muscular dystrophy. <i>FASEB Journal</i> , 2002, 16, 1096-1098.	0.5	21
60	Skeletal Muscle Structure and Function. , 2006, , 674-681.		20
61	Systematic Comparison of Digital Electrocardiograms From Healthy Athletes and Patients With Hypertrophic Cardiomyopathy. <i>Journal of the American College of Cardiology</i> , 2015, 65, 2462-2463.	2.8	20
62	Extracutaneous manifestations in phacomatosis cesioflammea and cesiomarmorata: Case series and literature review. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 966-977.	1.2	20
63	Functional and structural analysis of cytokine-selective IL6ST defects that cause recessive hyper-IgE syndrome. <i>Journal of Allergy and Clinical Immunology</i> , 2021, 148, 585-598.	2.9	20
64	Perceived utility and disutility of genomic sequencing for pediatric patients: Perspectives from parents with diverse sociodemographic characteristics. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 1088-1101.	1.2	20
65	Cardiomyopathy in animal models of muscular dystrophy. <i>Current Opinion in Cardiology</i> , 2001, 16, 211-217.	1.8	19
66	It seems like COVID-19 now is the only disease present on Earth, living with a rare or undiagnosed disease during the COVID-19 pandemic. <i>Genetics in Medicine</i> , 2021, 23, 837-844.	2.4	19
67	Clinical sites of the Undiagnosed Diseases Network: unique contributions to genomic medicine and science. <i>Genetics in Medicine</i> , 2021, 23, 259-271.	2.4	18
68	Computerized Q wave dimensions in athletes and hypertrophic cardiomyopathy patients. <i>Journal of Electrocardiology</i> , 2015, 48, 362-367.	0.9	16
69	A New Approach to Rare Diseases of Children: The Undiagnosed Diseases Network. <i>Journal of Pediatrics</i> , 2018, 196, 291-297.e2.	1.8	15
70	Systems biology of heart failure, challenges and hopes. <i>Current Opinion in Cardiology</i> , 2011, 26, 314-321.	1.8	14
71	Applying current normative data to prognosis in heart failure: The Fitness Registry and the Importance of Exercise National Database (FRIEND). <i>International Journal of Cardiology</i> , 2018, 263, 75-79.	1.7	14
72	Improving risk stratification in heart failure with preserved ejection fraction by combining two validated risk scores. <i>Open Heart</i> , 2019, 6, e000961.	2.3	13

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73	Functional Cardiac Recovery and Hematologic Response to Chemotherapy in Patients With Light-Chain Amyloidosis (from the Stanford University Amyloidosis Registry). <i>American Journal of Cardiology</i> , 2017, 120, 1381-1386.	1.6	12
74	Findings From Cardiovascular Evaluation of National Collegiate Athletic Association Division I Collegiate Student-Athletes After Asymptomatic or Mildly Symptomatic SARS-CoV-2 Infection. <i>Clinical Journal of Sport Medicine</i> , 2022, 32, 103-107.	1.8	12
75	A toolkit for genetics providers in follow-up of patients with non-diagnostic exome sequencing. <i>Journal of Genetic Counseling</i> , 2019, 28, 213-228.	1.6	11
76	Value of Strain Imaging and Maximal Oxygen Consumption in Patients With Hypertrophic Cardiomyopathy. <i>American Journal of Cardiology</i> , 2017, 120, 1203-1208.	1.6	10
77	Defining genotype-phenotype relationships in patients with hypertrophic cardiomyopathy using cardiovascular magnetic resonance imaging. <i>PLoS ONE</i> , 2019, 14, e0217612.	2.5	10
78	Baseline Characteristics of the VANISH Cohort. <i>Circulation: Heart Failure</i> , 2019, 12, e006231.	3.9	10
79	"Doctors can read about it, they can know about it, but they've never lived with it": How parents use social media throughout the diagnostic odyssey. <i>Journal of Genetic Counseling</i> , 2021, 30, 1707-1718.	1.6	10
80	Genomic Context Differs Between Human Dilated Cardiomyopathy and Hypertrophic Cardiomyopathy. <i>Journal of the American Heart Association</i> , 2021, 10, e019944.	3.7	9
81	TOWARDS TRANSCRIPTOMICS AS A PRIMARY TOOL FOR RARE DISEASE INVESTIGATION. <i>Journal of Physical Education and Sports Management</i> , 2022, , mcs.a006198.	1.2	9
82	Examining QRS amplitude criteria for electrocardiographic left ventricular hypertrophy in recommendations for screening criteria in athletes. <i>Journal of Electrocardiology</i> , 2015, 48, 368-372.	0.9	8
83	Exome sequencing identifies de novo pathogenic variants in <i>FBN1</i> and <i>TRPS1</i> in a patient with a complex connective tissue phenotype. <i>Journal of Physical Education and Sports Management</i> , 2017, 3, a001388.	1.2	8
84	Contractile reserve and cardiopulmonary exercise parameters in patients with dilated cardiomyopathy, the two dimensions of exercise testing. <i>Echocardiography</i> , 2017, 34, 1179-1186.	0.9	8
85	Mono- and Biallelic Protein-Truncating Variants in Alpha-Actinin 2 Cause Cardiomyopathy Through Distinct Mechanisms. <i>Circulation Genomic and Precision Medicine</i> , 2021, 14, CIRCGEN121003419.	3.6	8
86	The response to cardiac resynchronization therapy in <i>LMNA</i> cardiomyopathy. <i>European Journal of Heart Failure</i> , 2022, 24, 685-693.	7.1	7
87	The interaction of coronary tone and cardiac fibrosis. <i>Current Atherosclerosis Reports</i> , 2005, 7, 219-226.	4.8	6
88	Hypertrophic Cardiomyopathy. <i>Journal of the American College of Cardiology</i> , 2015, 65, 570-572.	2.8	5
89	Genome Sequencing in Hypertrophic Cardiomyopathy. <i>Journal of the American College of Cardiology</i> , 2018, 72, 430-433.	2.8	5
90	Compound heterozygous <i>KCTD7</i> variants in progressive myoclonus epilepsy. <i>Journal of Neurogenetics</i> , 2021, 35, 74-83.	1.4	4

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91	Pharmacogenetics of Heart Failure: Evidence, Opportunities, and Challenges for Cardiovascular Pharmacogenomics. <i>Journal of Cardiovascular Translational Research</i> , 2008, 1, 25-36.	2.4	3
92	Extremely elevated lipoprotein(a), combined hyperlipidemia, and premature atherosclerosis in a Chinese family. <i>Journal of Clinical Lipidology</i> , 2010, 4, 543-547.	1.5	2
93	Repeats and Survival in Myotonic Dystrophy Type 1. <i>Circulation: Cardiovascular Genetics</i> , 2017, 10, .	5.1	2
94	Biallelic UBE4A loss-of-function variants cause intellectual disability and global developmental delay. <i>Genetics in Medicine</i> , 2021, 23, 661-668.	2.4	2
95	Cardiopulmonary Exercise Testing With Echocardiography to Assess Recovery in Patients With Ventricular Assist Devices. <i>ASAIO Journal</i> , 2021, Publish Ahead of Print, 1134-1138.	1.6	2
96	Generation of three induced pluripotent stem cell lines from hypertrophic cardiomyopathy patients carrying MYH7 mutations. <i>Stem Cell Research</i> , 2021, 55, 102455.	0.7	2
97	Generation of three induced pluripotent stem cell lines from hypertrophic cardiomyopathy patients carrying TNNI3 mutations. <i>Stem Cell Research</i> , 2021, 57, 102597.	0.7	1
98	Letter by Wheeler et al Regarding Article, "Recognition and Significance of Pathological T-Wave Inversions in Athletes". <i>Circulation</i> , 2015, 132, e180.	1.6	0
99	Multimodality Imaging for Risk Assessment of Inherited Cardiomyopathies. <i>Current Cardiovascular Risk Reports</i> , 2020, 14, 1.	2.0	0
100	Abstract 21307: Beta-Blockers Inferior to Calcium Channel Blockers in Hypertrophic Cardiomyopathy. <i>Circulation</i> , 2017, 136, .	1.6	0
101	Wnt Signaling Interactor WTIP (Wilms Tumor Interacting Protein) Underlies Novel Mechanism for Cardiac Hypertrophy. <i>Circulation Genomic and Precision Medicine</i> , 0, , .	3.6	0