

# Mark Yandell

## List of Publications by Year in descending order

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Version: 2024-02-01

73  
papers

7,187  
citations

109321

35  
h-index

79698

73  
g-index

82  
all docs

82  
docs citations

82  
times ranked

14441  
citing authors

#	ARTICLE	IF	CITATIONS
1	Causal and Candidate Gene Variants in a Large Cohort of Women With Primary Ovarian Insufficiency. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2022, 107, 685-714.	3.6	13
2	An explainable artificial intelligence approach for predicting cardiovascular outcomes using electronic health records. , 2022, 1, e0000004.		13
3	Comprehensive variant calling from whole-genome sequencing identifies a complex inversion that disrupts <i>ZFPM2</i> in familial congenital diaphragmatic hernia. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2022, 10, e1888.	1.2	6
4	Common Variation in Cytoskeletal Genes Is Associated with Conotruncal Heart Defects. <i>Genes</i> , 2021, 12, 655.	2.4	2
5	The genomic basis of evolutionary differentiation among honey bees. <i>Genome Research</i> , 2021, 31, 1203-1215.	5.5	17
6	Identification of Somatic Gene Signatures in Circulating Cell-Free DNA Associated with Disease Progression in Metastatic Prostate Cancer by a Novel Machine Learning Platform. <i>Oncologist</i> , 2021, 26, 751-760.	3.7	9
7	Two Genomic Loci Control Three Eye Colors in the Domestic Pigeon ( <i>Columba livia</i> ). <i>Molecular Biology and Evolution</i> , 2021, 38, 5376-5390.	8.9	11
8	Addressing Ethical and Laboratory Challenges for Initiation of a Rapid Whole Genome Sequencing Program. <i>Journal of Clinical and Translational Science</i> , 2021, 5, 1-13.	0.6	2
9	Noncoding sequence variants define a novel regulatory element in the first intron of the <i>N-acetylglutamate synthase</i> gene. <i>Human Mutation</i> , 2021, 42, 1624-1636.	2.5	3
10	A ROR2 coding variant is associated with craniofacial variation in domestic pigeons. <i>Current Biology</i> , 2021, 31, 5069-5076.e5.	3.9	14
11	A Poisson binomial-based statistical testing framework for comorbidity discovery across electronic health record datasets. <i>Nature Computational Science</i> , 2021, 1, 694-702.	8.0	6
12	The history and geographic distribution of a KCNQ1 atrial fibrillation risk allele. <i>Nature Communications</i> , 2021, 12, 6442.	12.8	7
13	Development of a Portable Tool to Identify Patients With Atrial Fibrillation Using Clinical Notes From the Electronic Medical Record. <i>Circulation: Cardiovascular Quality and Outcomes</i> , 2020, 13, e006516.	2.2	2
14	Curses or Cures: A Review of the Numerous Benefits Versus the Biosecurity Concerns of Conotoxin Research. <i>Biomedicines</i> , 2020, 8, 235.	3.2	27
15	Deep whole-genome sequencing of multiple proband tissues and parental blood reveals the complex genetic etiology of congenital diaphragmatic hernias. <i>Human Genetics and Genomics Advances</i> , 2020, 1, 100008.	1.7	5
16	Clinical and molecular epidemiology of invasive <i>Staphylococcus aureus</i> infection in Utah children; continued dominance of MSSA over MRSA. <i>PLoS ONE</i> , 2020, 15, e0238991.	2.5	20
17	A copy number variant is associated with a spectrum of pigmentation patterns in the rock pigeon ( <i>Columba livia</i> ). <i>PLoS Genetics</i> , 2020, 16, e1008274.	3.5	34
18	Comprehensive Analysis of <i>AR</i> Alterations in Circulating Tumor DNA from Patients with Advanced Prostate Cancer. <i>Oncologist</i> , 2020, 25, 327-333.	3.7	27

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19	Pulmonary Eosinophilic Granulomatosis with Polyangiitis Has IgG4 Plasma Cells and Immunoregulatory Features. <i>American Journal of Pathology</i> , 2020, 190, 1438-1448.	3.8	7
20	Transcriptomic Profiling Reveals Extraordinary Diversity of Venom Peptides in Unexplored Predatory Gastropods of the Genus <i>Clavus</i> . <i>Genome Biology and Evolution</i> , 2020, 12, 684-700.	2.5	17
21	De novo and recessive forms of congenital heart disease have distinct genetic and phenotypic landscapes. <i>Nature Communications</i> , 2019, 10, 4722.	12.8	58
22	Identification of Polycystic Kidney Disease 1 Like 1 Gene Variants in Children With Biliary Atresia Splenic Malformation Syndrome. <i>Hepatology</i> , 2019, 70, 899-910.	7.3	58
23	Improved Genome Assembly and Annotation for the Rock Pigeon ( <i>Columba livia</i> ). <i>G3: Genes, Genomes, Genetics</i> , 2018, 8, 1391-1398.	1.8	62
24	Primary Ovarian Insufficiency and Azoospermia in Carriers of a Homozygous PSMC3IP Stop Gain Mutation. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2018, 103, 555-563.	3.6	45
25	The sea lamprey germline genome provides insights into programmed genome rearrangement and vertebrate evolution. <i>Nature Genetics</i> , 2018, 50, 270-277.	21.4	262
26	XPAT: a toolkit to conduct cross-platform association studies with heterogeneous sequencing datasets. <i>Nucleic Acids Research</i> , 2018, 46, e32-e32.	14.5	6
27	Discovery of Novel Conotoxin Candidates Using Machine Learning. <i>Toxins</i> , 2018, 10, 503.	3.4	20
28	Introgression of regulatory alleles and a missense coding mutation drive plumage pattern diversity in the rock pigeon. <i>ELife</i> , 2018, 7, .	6.0	66
29	The VAAST Variant Prioritizer (VVP): ultrafast, easy to use whole genome variant prioritization tool. <i>BMC Bioinformatics</i> , 2018, 19, 57.	2.6	29
30	Regulatory Divergence in Wound-Responsive Gene Expression between Domesticated and Wild Tomato. <i>Plant Cell</i> , 2018, 30, 1445-1460.	6.6	23
31	Hormone-like peptides in the venoms of marine cone snails. <i>General and Comparative Endocrinology</i> , 2017, 244, 11-18.	1.8	63
32	Viral Pathogen Detection by Metagenomics and Pan-Viral Group Polymerase Chain Reaction in Children With Pneumonia Lacking Identifiable Etiology. <i>Journal of Infectious Diseases</i> , 2017, 215, 1407-1415.	4.0	85
33	Emergence of a Viral RNA Polymerase Variant during Gene Copy Number Amplification Promotes Rapid Evolution of Vaccinia Virus. <i>Journal of Virology</i> , 2017, 91, .	3.4	36
34	Human Bocavirus Capsid Messenger RNA Detection in Children With Pneumonia. <i>Journal of Infectious Diseases</i> , 2017, 216, 688-696.	4.0	22
35	The Douglas-Fir Genome Sequence Reveals Specialization of the Photosynthetic Apparatus in Pinaceae. <i>G3: Genes, Genomes, Genetics</i> , 2017, 7, 3157-3167.	1.8	103
36	Settling the score: variant prioritization and Mendelian disease. <i>Nature Reviews Genetics</i> , 2017, 18, 599-612.	16.3	213

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37	The Venom Repertoire of <i>Conus gloriamaris</i> (Chemnitz, 1777), the Glory of the Sea. <i>Marine Drugs</i> , 2017, 15, 145.	4.6	27
38	Divergence of the Venom Exogene Repertoire in Two Sister Species of <i>Turriconus</i> . <i>Genome Biology and Evolution</i> , 2017, 9, 2211-2225.	2.5	30
39	Molecular shifts in limb identity underlie development of feathered feet in two domestic avian species. <i>ELife</i> , 2016, 5, e12115.	6.0	64
40	Antimicrobial Functions of Lactoferrin Promote Genetic Conflicts in Ancient Primates and Modern Humans. <i>PLoS Genetics</i> , 2016, 12, e1006063.	3.5	32
41	VARPRISM: incorporating variant prioritization in tests of de novo mutation association. <i>Genome Medicine</i> , 2016, 8, 91.	8.2	7
42	The draft genome sequence and annotation of the desert woodrat <i>Neotoma lepida</i> . <i>Genomics Data</i> , 2016, 9, 58-59.	1.3	9
43	Venom Insulins of Cone Snails Diversify Rapidly and Track Prey Taxa. <i>Molecular Biology and Evolution</i> , 2016, 33, 2924-2934.	8.9	44
44	Taxonomer: an interactive metagenomics analysis portal for universal pathogen detection and host mRNA expression profiling. <i>Genome Biology</i> , 2016, 17, 111.	8.8	152
45	Unbiased Detection of Respiratory Viruses by Use of RNA Sequencing-Based Metagenomics: a Systematic Comparison to a Commercial PCR Panel. <i>Journal of Clinical Microbiology</i> , 2016, 54, 1000-1007.	3.9	177
46	The spotted gar genome illuminates vertebrate evolution and facilitates human-teleost comparisons. <i>Nature Genetics</i> , 2016, 48, 427-437.	21.4	545
47	Rapid expansion of the protein disulfide isomerase gene family facilitates the folding of venom peptides. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2016, 113, 3227-3232.	7.1	39
48	Exome analysis of a family with Wolffâ€“Parkinsonâ€“White syndrome identifies a novel disease locus. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 2975-2984.	1.2	17
49	An Introduction to Genome Annotation. <i>Current Protocols in Bioinformatics</i> , 2015, 52, 4.1.1-4.1.17.	25.8	4
50	Wham: Identifying Structural Variants of Biological Consequence. <i>PLoS Computational Biology</i> , 2015, 11, e1004572.	3.2	105
51	Shared Segment Analysis and Next-Generation Sequencing Implicates the Retinoic Acid Signaling Pathway in Total Anomalous Pulmonary Venous Return (TAPVR). <i>PLoS ONE</i> , 2015, 10, e0131514.	2.5	16
52	Specialized insulin is used for chemical warfare by fish-hunting cone snails. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2015, 112, 1743-1748.	7.1	134
53	Muscle stem cells contribute to myofibres in sedentary adult mice. <i>Nature Communications</i> , 2015, 6, 7087.	12.8	222
54	Genomic signatures of evolutionary transitions from solitary to group living. <i>Science</i> , 2015, 348, 1139-1143.	12.6	357

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55	Evidence for extensive horizontal gene transfer from the draft genome of a tardigrade. Proceedings of the National Academy of Sciences of the United States of America, 2015, 112, 15976-15981.	7.1	145
56	Coordinated control of senescence by lncRNA and a novel T-box3 co-repressor complex. ELife, 2014, 3, .	6.0	81
57	Automated Update, Revision, and Quality Control of the Maize Genome Annotations Using MAKER-P Improves the B73 RefGen_v3 Gene Models and Identifies New Genes. Plant Physiology, 2014, 167, 25-39.	4.8	53
58	TBX3 Regulates Splicing In Vivo: A Novel Molecular Mechanism for Ulnar-Mammary Syndrome. PLoS Genetics, 2014, 10, e1004247.	3.5	31
59	Transiently Active Wnt/ $\beta$ -Catenin Signaling Is Not Required but Must Be Silenced for Stem Cell Function during Muscle Regeneration. Stem Cell Reports, 2014, 3, 475-488.	4.8	76
60	Transposable element islands facilitate adaptation to novel environments in an invasive species. Nature Communications, 2014, 5, 5495.	12.8	183
61	DisA-dependent striated fiber elongation is required to organize ciliary arrays. Journal of Cell Biology, 2014, 207, 705-715.	5.2	43
62	Pharmacogenomics of 17-alpha hydroxyprogesterone caproate for recurrent preterm birth prevention. American Journal of Obstetrics and Gynecology, 2014, 210, 321.e1-321.e21.	1.3	23
63	A unified test of linkage analysis and rare-variant association for analysis of pedigree sequence data. Nature Biotechnology, 2014, 32, 663-669.	17.5	93
64	Phevor Combines Multiple Biomedical Ontologies for Accurate Identification of Disease-Causing Alleles in Single Individuals and Small Nuclear Families. American Journal of Human Genetics, 2014, 94, 599-610.	6.2	175
65	Using VAAST to Identify Disease-Associated Variants in Next-Generation Sequencing Data. Current Protocols in Human Genetics, 2014, 81, 6.14.1-25.	3.5	21
66	Autoimmunity due to RAG deficiency and estimated disease incidence in RAG1/2 mutations. Journal of Allergy and Clinical Immunology, 2014, 133, 880-882.e10.	2.9	54
67	Epistatic and Combinatorial Effects of Pigmentary Gene Mutations in the Domestic Pigeon. Current Biology, 2014, 24, 459-464.	3.9	75
68	Next Generation Sequencing to Delineate the Mutational Landscape of Chronic Myelomonocytic Leukemia (CMML): Novel Disease Genes and Correlations with Survival. Blood, 2014, 124, 4637-4637.	1.4	0
69	VAAST 2.0: Improved Variant Classification and Disease-Associated Gene Identification Using a Conservation-Controlled Amino Acid Substitution Matrix. Genetic Epidemiology, 2013, 37, 622-634.	1.3	128
70	A beginner's guide to eukaryotic genome annotation. Nature Reviews Genetics, 2012, 13, 329-342.	16.3	553
71	A probabilistic disease-gene finder for personal genomes. Genome Research, 2011, 21, 1529-1542.	5.5	182
72	MAKER2: an annotation pipeline and genome-database management tool for second-generation genome projects. BMC Bioinformatics, 2011, 12, 491.	2.6	1,654

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73	Using VAAST to Identify an X-Linked Disorder Resulting in Lethality in Male Infants Due to N-Terminal Acetyltransferase Deficiency. American Journal of Human Genetics, 2011, 89, 28-43.	6.2	222