

# Richard C Trembath

## List of Publications by Year in descending order

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Version: 2024-02-01

58  
papers

10,118  
citations

76196

40  
h-index

123241

61  
g-index

72  
all docs

72  
docs citations

72  
times ranked

13764  
citing authors

#	ARTICLE	IF	CITATIONS
1	Autoimmunity Is a Significant Feature of Idiopathic Pulmonary Arterial Hypertension. American Journal of Respiratory and Critical Care Medicine, 2022, 206, 81-93.	2.5	9
2	Integrating polygenic risk scores in the prediction of type 2 diabetes risk and subtypes in British Pakistanis and Bangladeshis: A population-based cohort study. PLoS Medicine, 2022, 19, e1003981.	3.9	24
3	Bayesian Inference Associates Rare <i>KDR</i> Variants With Specific Phenotypes in Pulmonary Arterial Hypertension. Circulation Genomic and Precision Medicine, 2021, 14, .	1.6	29
4	Rare variant analysis of 4241 pulmonary arterial hypertension cases from an international consortium implicates FBLN2, PDGFD, and rare de novo variants in PAH. Genome Medicine, 2021, 13, 80.	3.6	43
5	Genome-Wide Association Study Identifies Risk Loci for Cluster Headache. Annals of Neurology, 2021, 90, 193-202.	2.8	31
6	The power of genetic diversity in genome-wide association studies of lipids. Nature, 2021, 600, 675-679.	13.7	353
7	Cohort Profile: East London Genes & Health (ELGH), a community-based population genomics and health study in British Bangladeshi and British Pakistani people. International Journal of Epidemiology, 2020, 49, 20-21i.	0.9	71
8	Characterization of <i>GDF2</i> Mutations and Levels of BMP9 and BMP10 in Pulmonary Arterial Hypertension. American Journal of Respiratory and Critical Care Medicine, 2020, 201, 575-585.	2.5	80
9	Whole Exome Sequence Analysis Provides Novel Insights into the Genetic Framework of Childhood-Onset Pulmonary Arterial Hypertension. Genes, 2020, 11, 1328.	1.0	14
10	Trans-ethnic and Ancestry-Specific Blood-Cell Genetics in 746,667 Individuals from 5 Global Populations. Cell, 2020, 182, 1198-1213.e14.	13.5	353
11	Whole-Blood RNA Profiles Associated with Pulmonary Arterial Hypertension and Clinical Outcome. American Journal of Respiratory and Critical Care Medicine, 2020, 202, 586-594.	2.5	45
12	Evaluating drug targets through human loss-of-function genetic variation. Nature, 2020, 581, 459-464.	13.7	115
13	A restricted spectrum of missense <i>KMT2D</i> variants cause a multiple malformations disorder distinct from Kabuki syndrome. Genetics in Medicine, 2020, 22, 867-877.	1.1	41
14	Mendelian randomisation analysis of red cell distribution width in pulmonary arterial hypertension. European Respiratory Journal, 2020, 55, 1901486.	3.1	26
15	Characterising a healthy adult with a rare <i>HAO1</i> knockout to support a therapeutic strategy for primary hyperoxaluria. ELife, 2020, 9, .	2.8	45
16	Cross-disorder analysis of schizophrenia and 19 immune-mediated diseases identifies shared genetic risk. Human Molecular Genetics, 2019, 28, 3498-3513.	1.4	65
17	The impact of donor and recipient common clinical and genetic variation on estimated glomerular filtration rate in a European renal transplant population. American Journal of Transplantation, 2019, 19, 2262-2273.	2.6	13
18	Genetic determinants of risk in pulmonary arterial hypertension: international genome-wide association studies and meta-analysis. Lancet Respiratory Medicine, the, 2019, 7, 227-238.	5.2	122

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19	Genetics and genomics of pulmonary arterial hypertension. <i>European Respiratory Journal</i> , 2019, 53, 1801899.	3.1	306
20	Identification of rare sequence variation underlying heritable pulmonary arterial hypertension. <i>Nature Communications</i> , 2018, 9, 1416.	5.8	279
21	Long- and short-term outcomes in renal allografts with deceased donors: A large recipient and donor genome-wide association study. <i>American Journal of Transplantation</i> , 2018, 18, 1370-1379.	2.6	47
22	Genome-wide meta-analysis implicates mediators of hair follicle development and morphogenesis in risk for severe acne. <i>Nature Communications</i> , 2018, 9, 5075.	5.8	48
23	Loss-of-Function <i>ABCC8</i> Mutations in Pulmonary Arterial Hypertension. <i>Circulation Genomic and Precision Medicine</i> , 2018, 11, e002087.	1.6	62
24	Genetic correlations among psychiatric and immune-related phenotypes based on genome-wide association data. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2018, 177, 641-657.	1.1	158
25	Elucidating the genetic architecture of Adams-Oliver syndrome in a large European cohort. <i>Human Mutation</i> , 2018, 39, 1246-1261.	1.1	31
26	Large scale meta-analysis characterizes genetic architecture for common psoriasis associated variants. <i>Nature Communications</i> , 2017, 8, 15382.	5.8	251
27	miR-146b Probably Assists miRNA-146a in the Suppression of Keratinocyte Proliferation and Inflammatory Responses in Psoriasis. <i>Journal of Investigative Dermatology</i> , 2017, 137, 1945-1954.	0.3	68
28	Phenotypic Characterization of <i>EIF2AK4</i> Mutation Carriers in a Large Cohort of Patients Diagnosed Clinically With Pulmonary Arterial Hypertension. <i>Circulation</i> , 2017, 136, 2022-2033.	1.6	111
29	An analysis of IL-36 signature genes and individuals with <i>IL1RL2</i> knockout mutations validates IL-36 as a psoriasis therapeutic target. <i>Science Translational Medicine</i> , 2017, 9, .	5.8	124
30	Estimating the human mutation rate from autozygous segments reveals population differences in human mutational processes. <i>Nature Communications</i> , 2017, 8, 303.	5.8	81
31	Exome-wide association study reveals novel psoriasis susceptibility locus at <i>TNFSF15</i> and rare protective alleles in genes contributing to type I IFN signalling. <i>Human Molecular Genetics</i> , 2017, 26, 4301-4313.	1.4	41
32	<i>AP1S3</i> Mutations Cause Skin Autoinflammation by Disrupting Keratinocyte Autophagy and Up-Regulating IL-36 Production. <i>Journal of Investigative Dermatology</i> , 2016, 136, 2251-2259.	0.3	128
33	Polymorphism in a lincRNA Associates with a Doubled Risk of Pneumococcal Bacteremia in Kenyan Children. <i>American Journal of Human Genetics</i> , 2016, 98, 1092-1100.	2.6	39
34	Health and population effects of rare gene knockouts in adult humans with related parents. <i>Science</i> , 2016, 352, 474-477.	6.0	272
35	Analysis of five chronic inflammatory diseases identifies 27 new associations and highlights disease-specific patterns at shared loci. <i>Nature Genetics</i> , 2016, 48, 510-518.	9.4	617
36	Germline <i>ESR2</i> mutation predisposes to medullary thyroid carcinoma and causes up-regulation of <i>RET</i> expression. <i>Human Molecular Genetics</i> , 2016, 25, 1836-1845.	1.4	28

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37	Pulmonary Arterial Hypertension: A Current Perspective on Established and Emerging Molecular Genetic Defects. <i>Human Mutation</i> , 2015, 36, 1113-1127.	1.1	185
38	Germline Mutations in the <i>CDKN2B</i> Tumor Suppressor Gene Predispose to Renal Cell Carcinoma. <i>Cancer Discovery</i> , 2015, 5, 723-729.	7.7	88
39	Enhanced meta-analysis and replication studies identify five new psoriasis susceptibility loci. <i>Nature Communications</i> , 2015, 6, 7001.	5.8	156
40	Haploinsufficiency of the NOTCH1 Receptor as a Cause of Adams-Oliver Syndrome With Variable Cardiac Anomalies. <i>Circulation: Cardiovascular Genetics</i> , 2015, 8, 572-581.	5.1	84
41	Genome-wide Comparative Analysis of Atopic Dermatitis and Psoriasis Gives Insight into Opposing Genetic Mechanisms. <i>American Journal of Human Genetics</i> , 2015, 96, 104-120.	2.6	163
42	Activating CARD14 Mutations Are Associated with Generalized Pustular Psoriasis but Rarely Account for Familial Recurrence in Psoriasis Vulgaris. <i>Journal of Investigative Dermatology</i> , 2015, 135, 2964-2970.	0.3	89
43	IL36RN mutations define a severe autoinflammatory phenotype of generalized pustular psoriasis. <i>Journal of Allergy and Clinical Immunology</i> , 2015, 135, 1067-1070.e9.	1.5	115
44	Heterozygous Loss-of-Function Mutations in DLL4 Cause Adams-Oliver Syndrome. <i>American Journal of Human Genetics</i> , 2015, 97, 475-482.	2.6	73
45	Loss of IL36RN Function Does Not Confer Susceptibility to Psoriasis Vulgaris. <i>Journal of Investigative Dermatology</i> , 2014, 134, 271-273.	0.3	25
46	Genome-wide association study identifies three novel susceptibility loci for severe Acne vulgaris. <i>Nature Communications</i> , 2014, 5, 4020.	5.8	68
47	The correlation between reading and mathematics ability at age twelve has a substantial genetic component. <i>Nature Communications</i> , 2014, 5, 4204.	5.8	72
48	AP1S3 Mutations Are Associated with Pustular Psoriasis and Impaired Toll-like Receptor 3 Trafficking. <i>American Journal of Human Genetics</i> , 2014, 94, 790-797.	2.6	153
49	$\hat{I}^3$ -Secretase Mutations in Hidradenitis Suppurativa: New Insights into Disease Pathogenesis. <i>Journal of Investigative Dermatology</i> , 2013, 133, 601-607.	0.3	133
50	Mutations in the $\hat{I}^3$ -Secretase Genes NCSTN , PSENEN , and PSEN1 Underlie Rare Forms of Hidradenitis Suppurativa (Acne Inversa). <i>Journal of Investigative Dermatology</i> , 2012, 132, 2459-2461.	0.3	126
51	Mutations in IL36RN/IL1F5 Are Associated with the Severe Episodic Inflammatory Skin Disease Known as Generalized Pustular Psoriasis. <i>American Journal of Human Genetics</i> , 2011, 89, 432-437.	2.6	468
52	Molecular genetic characterization of SMAD signaling molecules in pulmonary arterial hypertension. <i>Human Mutation</i> , 2011, 32, 1385-1389.	1.1	152
53	A genome-wide association study identifies new psoriasis susceptibility loci and an interaction between HLA-C and ERAP1. <i>Nature Genetics</i> , 2010, 42, 985-990.	9.4	918
54	A strategy for translation. <i>Lancet</i> , The, 2007, 369, 1771-1773.	6.3	6

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55	Transforming Growth Factor- $\beta$ 2 Receptor Mutations and Pulmonary Arterial Hypertension in Childhood. <i>Circulation</i> , 2005, 111, 435-441.	1.6	222
56	BMPR2 Haploinsufficiency as the Inherited Molecular Mechanism for Primary Pulmonary Hypertension. <i>American Journal of Human Genetics</i> , 2001, 68, 92-102.	2.6	521
57	Clinical and Molecular Genetic Features of Pulmonary Hypertension in Patients with Hereditary Hemorrhagic Telangiectasia. <i>New England Journal of Medicine</i> , 2001, 345, 325-334.	13.9	676
58	Heterozygous germline mutations in BMPR2, encoding a TGF- $\beta$ 2 receptor, cause familial primary pulmonary hypertension. <i>Nature Genetics</i> , 2000, 26, 81-84.	9.4	1,388