

# Richard C Trembath

## List of Publications by Year in descending order

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Version: 2024-02-01

58  
papers

10,118  
citations

76326

40  
h-index

123424

61  
g-index

72  
all docs

72  
docs citations

72  
times ranked

13764  
citing authors

#	ARTICLE	IF	CITATIONS
1	Heterozygous germline mutations in BMPR2, encoding a TGF- $\beta$ 2 receptor, cause familial primary pulmonary hypertension. <i>Nature Genetics</i> , 2000, 26, 81-84.	21.4	1,388
2	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.	21.4	918
3	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.	27.0	676
4	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.	21.4	617
5	BMPR2 Haploinsufficiency as the Inherited Molecular Mechanism for Primary Pulmonary Hypertension. <i>American Journal of Human Genetics</i> , 2001, 68, 92-102.	6.2	521
6	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.	6.2	468
7	Trans-ethnic and Ancestry-Specific Blood-Cell Genetics in 746,667 Individuals from 5 Global Populations. <i>Cell</i> , 2020, 182, 1198-1213.e14.	28.9	353
8	The power of genetic diversity in genome-wide association studies of lipids. <i>Nature</i> , 2021, 600, 675-679.	27.8	353
9	Genetics and genomics of pulmonary arterial hypertension. <i>European Respiratory Journal</i> , 2019, 53, 1801899.	6.7	306
10	Identification of rare sequence variation underlying heritable pulmonary arterial hypertension. <i>Nature Communications</i> , 2018, 9, 1416.	12.8	279
11	Health and population effects of rare gene knockouts in adult humans with related parents. <i>Science</i> , 2016, 352, 474-477.	12.6	272
12	Large scale meta-analysis characterizes genetic architecture for common psoriasis associated variants. <i>Nature Communications</i> , 2017, 8, 15382.	12.8	251
13	Transforming Growth Factor- $\beta$ 2 Receptor Mutations and Pulmonary Arterial Hypertension in Childhood. <i>Circulation</i> , 2005, 111, 435-441.	1.6	222
14	Pulmonary Arterial Hypertension: A Current Perspective on Established and Emerging Molecular Genetic Defects. <i>Human Mutation</i> , 2015, 36, 1113-1127.	2.5	185
15	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.	6.2	163
16	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.	1.7	158
17	Enhanced meta-analysis and replication studies identify five new psoriasis susceptibility loci. <i>Nature Communications</i> , 2015, 6, 7001.	12.8	156
18	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.	6.2	153

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19	Molecular genetic characterization of SMAD signaling molecules in pulmonary arterial hypertension. <i>Human Mutation</i> , 2011, 32, 1385-1389.	2.5	152
20	Î³-Secretase Mutations in Hidradenitis Suppurativa: New Insights into Disease Pathogenesis. <i>Journal of Investigative Dermatology</i> , 2013, 133, 601-607.	0.7	133
21	AP1S3 Mutations Cause Skin Autoinflammation by Disrupting Keratinocyte Autophagy and Up-Regulating IL-36 Production. <i>Journal of Investigative Dermatology</i> , 2016, 136, 2251-2259.	0.7	128
22	Mutations in the Î³-Secretase Genes NCSTN , PSENEN , and PSEN1 Underlie Rare Forms of Hidradenitis Suppurativa (Acne Inversa). <i>Journal of Investigative Dermatology</i> , 2012, 132, 2459-2461.	0.7	126
23	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, .	12.4	124
24	Genetic determinants of risk in pulmonary arterial hypertension: international genome-wide association studies and meta-analysis. <i>Lancet Respiratory Medicine</i> , 2019, 7, 227-238.	10.7	122
25	IL36RN mutations define a severe autoinflammatory phenotype of generalized pustular psoriasis. <i>Journal of Allergy and Clinical Immunology</i> , 2015, 135, 1067-1070.e9.	2.9	115
26	Evaluating drug targets through human loss-of-function genetic variation. <i>Nature</i> , 2020, 581, 459-464.	27.8	115
27	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
28	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.	0.7	89
29	Germline Mutations in the <i>CDKN2B</i> Tumor Suppressor Gene Predispose to Renal Cell Carcinoma. <i>Cancer Discovery</i> , 2015, 5, 723-729.	9.4	88
30	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
31	Estimating the human mutation rate from autozygous segments reveals population differences in human mutational processes. <i>Nature Communications</i> , 2017, 8, 303.	12.8	81
32	Characterization of <i>GDF2</i> Mutations and Levels of BMP9 and BMP10 in Pulmonary Arterial Hypertension. <i>American Journal of Respiratory and Critical Care Medicine</i> , 2020, 201, 575-585.	5.6	80
33	Heterozygous Loss-of-Function Mutations in DLL4 Cause Adams-Oliver Syndrome. <i>American Journal of Human Genetics</i> , 2015, 97, 475-482.	6.2	73
34	The correlation between reading and mathematics ability at age twelve has a substantial genetic component. <i>Nature Communications</i> , 2014, 5, 4204.	12.8	72
35	Cohort Profile: East London Genes & Health (ELGH), a community-based population genomics and health study in British Bangladeshi and British Pakistani people. <i>International Journal of Epidemiology</i> , 2020, 49, 20-21i.	1.9	71
36	Genome-wide association study identifies three novel susceptibility loci for severe Acne vulgaris. <i>Nature Communications</i> , 2014, 5, 4020.	12.8	68

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37	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.	0.7	68
38	Cross-disorder analysis of schizophrenia and 19 immune-mediated diseases identifies shared genetic risk. <i>Human Molecular Genetics</i> , 2019, 28, 3498-3513.	2.9	65
39	Loss-of-Function <i>ABCC8</i> Mutations in Pulmonary Arterial Hypertension. <i>Circulation Genomic and Precision Medicine</i> , 2018, 11, e002087.	3.6	62
40	Genome-wide meta-analysis implicates mediators of hair follicle development and morphogenesis in risk for severe acne. <i>Nature Communications</i> , 2018, 9, 5075.	12.8	48
41	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.	4.7	47
42	Whole-Blood RNA Profiles Associated with Pulmonary Arterial Hypertension and Clinical Outcome. <i>American Journal of Respiratory and Critical Care Medicine</i> , 2020, 202, 586-594.	5.6	45
43	Characterising a healthy adult with a rare <i>HAO1</i> knockout to support a therapeutic strategy for primary hyperoxaluria. <i>ELife</i> , 2020, 9, .	6.0	45
44	Rare variant analysis of 4241 pulmonary arterial hypertension cases from an international consortium implicates <i>FBLN2</i> , <i>PDGFD</i> , and rare de novo variants in PAH. <i>Genome Medicine</i> , 2021, 13, 80.	8.2	43
45	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.	2.9	41
46	A restricted spectrum of missense <i>KMT2D</i> variants cause a multiple malformations disorder distinct from Kabuki syndrome. <i>Genetics in Medicine</i> , 2020, 22, 867-877.	2.4	41
47	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.	6.2	39
48	Genome-Wide Association Study Identifies Risk Loci for Cluster Headache. <i>Annals of Neurology</i> , 2021, 90, 193-202.	5.3	31
49	Elucidating the genetic architecture of Adams-Oliver syndrome in a large European cohort. <i>Human Mutation</i> , 2018, 39, 1246-1261.	2.5	31
50	Bayesian Inference Associates Rare <i>KDR</i> Variants With Specific Phenotypes in Pulmonary Arterial Hypertension. <i>Circulation Genomic and Precision Medicine</i> , 2021, 14, .	3.6	29
51	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.	2.9	28
52	Mendelian randomisation analysis of red cell distribution width in pulmonary arterial hypertension. <i>European Respiratory Journal</i> , 2020, 55, 1901486.	6.7	26
53	Loss of <i>IL36RN</i> Function Does Not Confer Susceptibility to Psoriasis Vulgaris. <i>Journal of Investigative Dermatology</i> , 2014, 134, 271-273.	0.7	25
54	Integrating polygenic risk scores in the prediction of type 2 diabetes risk and subtypes in British Pakistanis and Bangladeshis: A population-based cohort study. <i>PLoS Medicine</i> , 2022, 19, e1003981.	8.4	24

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55	Whole Exome Sequence Analysis Provides Novel Insights into the Genetic Framework of Childhood-Onset Pulmonary Arterial Hypertension. <i>Genes</i> , 2020, 11, 1328.	2.4	14
56	The impact of donor and recipient common clinical and genetic variation on estimated glomerular filtration rate in a European renal transplant population. <i>American Journal of Transplantation</i> , 2019, 19, 2262-2273.	4.7	13
57	Autoimmunity Is a Significant Feature of Idiopathic Pulmonary Arterial Hypertension. <i>American Journal of Respiratory and Critical Care Medicine</i> , 2022, 206, 81-93.	5.6	9
58	A strategy for translation. <i>Lancet, The</i> , 2007, 369, 1771-1773.	13.7	6