Richard C Trembath

List of Publications by Year
in descending order

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	Autoimmunity Is a Significant Feature of Idiopathic Pulmonary Arterial Hypertension. American Journal of Respiratory and Critical Care Medicine, 2022, 206, 81-93.	5.6	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.	8.4	24
3	Bayesian Inference Associates Rare KDR Variants With Specific Phenotypes in Pulmonary Arterial Hypertension. Circulation Genomic and Precision Medicine, 2021, 14, .	3.6	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.	8.2	43
5	Genomeâ€Wide Association Study Identifies Risk Loci for Cluster Headache. Annals of Neurology, 2021, 90, 193-202.	5.3	31
6	The power of genetic diversity in genome-wide association studies of lipids. Nature, 2021, 600, 675-679.	27.8	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.	1.9	71
8	Characterization of GDF2 Mutations and Levels of BMP9 and BMP10 in Pulmonary Arterial Hypertension. American Journal of Respiratory and Critical Care Medicine, 2020, 201, 575-585.	5.6	80
9	Whole Exome Sequence Analysis Provides Novel Insights into the Genetic Framework of Childhood-Onset Pulmonary Arterial Hypertension. Genes, 2020, 11, 1328.	2.4	14
10	Trans-ethnic and Ancestry-Specific Blood-Cell Genetics in 746,667 Individuals from 5 Global Populations. Cell, 2020, 182, 1198-1213.e14.	28.9	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.	5.6	45
12	Evaluating drug targets through human loss-of-function genetic variation. Nature, 2020, 581, 459-464.	27.8	115
13	A restricted spectrum of missense KMT2D variants cause a multiple malformations disorder distinct fromKabuki syndrome. Genetics in Medicine, 2020, 22, 867-877.	2.4	41
14	Mendelian randomisation analysis of red cell distribution width in pulmonary arterial hypertension. European Respiratory Journal, 2020, 55, 1901486.	6.7	26
15	Characterising a healthy adult with a rare HAO1 knockout to support a therapeutic strategy for primary hyperoxaluria. ELife, 2020, 9, .	6.0	45
16	Cross-disorder analysis of schizophrenia and 19 immune-mediated diseases identifies shared genetic risk. Human Molecular Genetics, 2019, 28, 3498-3513.	2.9	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.	4.7	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.	10.7	122

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19	Genetics and genomics of pulmonary arterial hypertension. European Respiratory Journal, 2019, 53, 1801899.	6.7	306
20	Identification of rare sequence variation underlying heritable pulmonary arterial hypertension. Nature Communications, 2018, 9, 1416.	12.8	279
21	Long- and short-term outcomes in renal allografts with deceased donors: A large recipient and donor genome-wide association study. American Journal of Transplantation, 2018, 18, 1370-1379.	4.7	47
22	Genome-wide meta-analysis implicates mediators of hair follicle development and morphogenesis in risk for severe acne. Nature Communications, 2018, 9, 5075.	12.8	48
23	Loss-of-Function ABCC8 Mutations in Pulmonary Arterial Hypertension. Circulation Genomic and Precision Medicine, 2018, 11, e002087.	3.6	62
24	Genetic correlations among psychiatric and immuneâ€related phenotypes based on genomeâ€wide association data. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2018, 177, 641-657.	1.7	158
25	Elucidating the genetic architecture of Adams-Oliver syndrome in a large European cohort. Human Mutation, 2018, 39, 1246-1261.	2.5	31
26	Large scale meta-analysis characterizes genetic architecture for common psoriasis associated variants. Nature Communications, 2017, 8, 15382.	12.8	251
27	miR-146b Probably Assists miRNA-146a inÂthe Suppression of Keratinocyte Proliferation and Inflammatory ResponsesÂin Psoriasis. Journal of Investigative Dermatology, 2017, 137, 1945-1954.	0.7	68
28	Phenotypic Characterization of EIF2AK4 Mutation Carriers in a Large Cohort of Patients Diagnosed Clinically With Pulmonary Arterial Hypertension. Circulation, 2017, 136, 2022-2033.	1.6	111
29	An analysis of IL-36 signature genes and individuals with IL1RL2 knockout mutations validates IL-36 as a psoriasis therapeutic target. Science Translational Medicine, 2017, 9, .	12.4	124
30	Estimating the human mutation rate from autozygous segments reveals population differences in human mutational processes. Nature Communications, 2017, 8, 303.	12.8	81
31	Exome-wide association study reveals novel psoriasis susceptibility locus at TNFSF15 and rare protective alleles in genes contributing to type I IFN signalling. Human Molecular Genetics, 2017, 26, 4301-4313.	2.9	41
32	AP1S3 Mutations Cause Skin Autoinflammation by Disrupting Keratinocyte Autophagy and Up-Regulating IL-36 Production. Journal of Investigative Dermatology, 2016, 136, 2251-2259.	0.7	128
33	Polymorphism in a lincRNA Associates with a Doubled Risk of Pneumococcal Bacteremia in Kenyan Children. American Journal of Human Genetics, 2016, 98, 1092-1100.	6.2	39
34	Health and population effects of rare gene knockouts in adult humans with related parents. Science, 2016, 352, 474-477.	12.6	272
35	Analysis of five chronic inflammatory diseases identifies 27 new associations and highlights disease-specific patterns at shared loci. Nature Genetics, 2016, 48, 510-518.	21.4	617
36	Germline ESR2 mutation predisposes to medullary thyroid carcinoma and causes up-regulation of RET expression. Human Molecular Genetics, 2016, 25, 1836-1845.	2.9	28

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37	Pulmonary Arterial Hypertension: A Current Perspective on Established and Emerging Molecular Genetic Defects. Human Mutation, 2015, 36, 1113-1127.	2.5	185
38	Germline Mutations in the CDKN2B Tumor Suppressor Gene Predispose to Renal Cell Carcinoma. Cancer Discovery, 2015, 5, 723-729.	9.4	88
39	Enhanced meta-analysis and replication studies identify five new psoriasis susceptibility loci. Nature Communications, 2015, 6, 7001.	12.8	156
40	Haploinsufficiency of the NOTCH1 Receptor as a Cause of Adamsâ€"Oliver Syndrome With Variable Cardiac Anomalies. Circulation: Cardiovascular Genetics, 2015, 8, 572-581.	5.1	84
41	Genome-wide Comparative Analysis of Atopic Dermatitis and Psoriasis Gives Insight into Opposing Genetic Mechanisms. American Journal of Human Genetics, 2015, 96, 104-120.	6.2	163
42	Activating CARD14 Mutations Are Associated with Generalized Pustular Psoriasis but Rarely Account for Familial Recurrence in Psoriasis Vulgaris. Journal of Investigative Dermatology, 2015, 135, 2964-2970.	0.7	89
43	IL36RN mutations define a severe autoinflammatory phenotype of generalized pustular psoriasis. Journal of Allergy and Clinical Immunology, 2015, 135, 1067-1070.e9.	2.9	115
44	Heterozygous Loss-of-Function Mutations in DLL4 Cause Adams-Oliver Syndrome. American Journal of Human Genetics, 2015, 97, 475-482.	6.2	73
45	Loss of IL36RN Function Does Not Confer Susceptibility to Psoriasis Vulgaris. Journal of Investigative Dermatology, 2014, 134, 271-273.	0.7	25
46	Genome-wide association study identifies three novel susceptibility loci for severe Acne vulgaris. Nature Communications, 2014, 5, 4020.	12.8	68
47	The correlation between reading and mathematics ability at age twelve has a substantial genetic component. Nature Communications, 2014, 5, 4204.	12.8	72
48	AP1S3 Mutations Are Associated with Pustular Psoriasis and Impaired Toll-like Receptor 3 Trafficking. American Journal of Human Genetics, 2014, 94, 790-797.	6.2	153
49	Î³-Secretase Mutations in Hidradenitis Suppurativa: New Insights into Disease Pathogenesis. Journal of Investigative Dermatology, 2013, 133, 601-607.	0.7	133
50	Mutations in the Î³-Secretase Genes NCSTN , PSENEN , and PSEN1 Underlie Rare Forms of Hidradenitis Suppurativa (Acne Inversa). Journal of Investigative Dermatology, 2012, 132, 2459-2461.	0.7	126
51	Mutations in IL36RN/IL1F5 Are Associated with the Severe Episodic Inflammatory Skin Disease Known as Generalized Pustular Psoriasis. American Journal of Human Genetics, 2011, 89, 432-437.	6.2	468
52	Molecular genetic characterization of SMAD signaling molecules in pulmonary arterial hypertension. Human Mutation, 2011, 32, 1385-1389.	2.5	152
53	A genome-wide association study identifies new psoriasis susceptibility loci and an interaction between HLA-C and ERAP1. Nature Genetics, 2010, 42, 985-990.	21.4	918
54	A strategy for translation. Lancet, The, 2007, 369, 1771-1773.	13.7	6

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55	Transforming Growth Factor-Î² Receptor Mutations and Pulmonary Arterial Hypertension in Childhood. Circulation, 2005, 111, 435-441.	1.6	222
56	BMPR2 Haploinsufficiency as the Inherited Molecular Mechanism for Primary Pulmonary Hypertension. American Journal of Human Genetics, 2001, 68, 92-102.	6.2	521
57	Clinical and Molecular Genetic Features of Pulmonary Hypertension in Patients with Hereditary Hemorrhagic Telangiectasia. New England Journal of Medicine, 2001, 345, 325-334.	27.0	676
58	Heterozygous germline mutations in BMPR2, encoding a TGF-Î² receptor, cause familial primary pulmonary hypertension. Nature Genetics, 2000, 26, 81-84.	21.4	1,388