Garry R Cutting

List of Publications by Year
in descending order

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104 papers	7,696 citations	87723 38 h-index	53109 85 g-index
110 all docs	110 docs citations	110 times ranked	7808 citing authors

#	Article	IF	Citations
1	Cystic fibrosis genetics: from molecular understanding to clinical application. Nature Reviews Genetics, 2015, 16, 45-56.	7.7	730
2	Defining the disease liability of variants in the cystic fibrosis transmembrane conductance regulator gene. Nature Genetics, 2013, 45, 1160-1167.	9.4	513
3	Genetic basis of variable exon 9 skipping in cystic fibrosis transmembrane conductance regulator mRNA. Nature Genetics, 1993, 3, 151-156.	9.4	489
4	From CFTR biology toward combinatorial pharmacotherapy: expanded classification of cystic fibrosis mutations. Molecular Biology of the Cell, 2016, 27, 424-433.	0.9	446
5	Laboratory standards and guidelines for population-based cystic fibrosis carrier screening. Genetics in Medicine, 2001, 3, 149-154.	1.1	440
6	A cluster of cystic fibrosis mutations in the first nucleotide-binding fold of the cystic fibrosis conductance regulator protein. Nature, 1990, 346, 366-369.	13.7	416
7	Recommendations for application of the functional evidence PS3/BS3 criterion using the ACMG/AMP sequence variant interpretation framework. Genome Medicine, 2020, 12, 3.	3.6	312
8	ABCD1 mutations and the X-linked adrenoleukodystrophy mutation database: Role in diagnosis and clinical correlations. Human Mutation, 2001, 18, 499-515.	1.1	261
9	Genome-wide association meta-analysis identifies five modifier loci of lung disease severity in cystic fibrosis. Nature Communications, 2015, 6, 8382.	5.8	242
10	Modifier genes in Mendelian disorders: the example of cystic fibrosis. Annals of the New York Academy of Sciences, 2010, 1214, 57-69.	1.8	233
11	Detection of a cystic fibrosis modifier locus for meconium ileus on human chromosome 19q13. Nature Genetics, 1999, 22, 128-129.	9.4	216
12	CFTR modulator theratyping: Current status, gaps and future directions. Journal of Cystic Fibrosis, 2019, 18, 22-34.	0.3	208
13	Multiple apical plasma membrane constituents are associated with susceptibility to meconium ileus in individuals with cystic fibrosis. Nature Genetics, 2012, 44, 562-569.	9.4	177
14	Heritability of Lung Disease Severity in Cystic Fibrosis. American Journal of Respiratory and Critical Care Medicine, 2007, 175, 1036-1043.	2.5	171
15	Five Percent of Normal Cystic Fibrosis Transmembrane Conductance Regulator mRNA Ameliorates the Severity of Pulmonary Disease in Cystic Fibrosis. American Journal of Respiratory Cell and Molecular Biology, 2002, 27, 619-627.	1.4	158
16	Genetic Modifiers of Cystic Fibrosisâ€“Related Diabetes. Diabetes, 2013, 62, 3627-3635.	0.3	148
17	MODIFIER GENETICS: Cystic Fibrosis. Annual Review of Genomics and Human Genetics, 2005, 6, 237-260.	2.5	141
18	Clustering of fibrillin (FBN1) missense mutations in Marfan syndrome patients at cysteine residues in EGF-like domains. Human Mutation, 1992, 1, 366-374.	1.1	131

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19	Relative Contribution of Genetic and Nongenetic Modifiers to Intestinal Obstruction in Cystic Fibrosis. Gastroenterology, 2006, 131, 1030-1039.	0.6	128
20	Interactions Between Secondhand Smoke and Genes That Affect Cystic Fibrosis Lung Disease. JAMA - Journal of the American Medical Association, 2008, 299, 417-24.	3.8	125
21	Increased Prevalence of Chronic Rhinosinusitis in Carriers of a Cystic Fibrosis Mutation. JAMA Otolaryngology, 2005, 131, 237.	1.5	121
22	GABACreceptor Ï•subunits are heterogeneously expressed in the human CNS and form homo- and heterooligomers with distinct physical properties. European Journal of Neuroscience, 1999, 11, 41-50.	1.2	106
23	Residual function of cystic fibrosis mutants predicts response to small molecule CFTR modulators. JCI Insight, 2018, 3, .	2.3	86
24	Simplified Multiplex-PCR Diagnosis of Common Southeast Asian Deletional Determinants of Î±-Thalassemia. Clinical Chemistry, 2000, 46, 1692-1695.	1.5	83
25	Correlating Cystic Fibrosis Transmembrane Conductance Regulator Function with Clinical Features to Inform Precision Treatment of Cystic Fibrosis. American Journal of Respiratory and Critical Care Medicine, 2019, 199, 1116-1126.	2.5	76
26	Functional Assays Are Essential for Interpretation of Missense Variants Associated with Variable Expressivity. American Journal of Human Genetics, 2018, 102, 1062-1077.	2.6	69
27	Systematic Computational Identification of Variants That Activate Exonic and Intronic Cryptic Splice Sites. American Journal of Human Genetics, 2017, 100, 751-765.	2.6	68
28	Genetic Modifiers Play a Substantial Role in Diabetes Complicating Cystic Fibrosis. Journal of Clinical Endocrinology and Metabolism, 2009, 94, 1302-1309.	1.8	66
29	Sources of Variation in Sweat Chloride Measurements in Cystic Fibrosis. American Journal of Respiratory and Critical Care Medicine, 2016, 194, 1375-1382.	2.5	62
30	Sensitivity of the denaturing gradient gel electrophoresis technique in detection of known mutations and novel Asian mutations in theCFTR gene. , 1997, 9, 136-147.		58
31	Sweat test for cystic fibrosis: Wearable sweat sensor vs. standard laboratory test. Journal of Cystic Fibrosis, 2018, 17, e35-e38.	0.3	57
32	Experimental Assessment of Splicing Variants Using Expression Minigenes and Comparison with In Silico Predictions. Human Mutation, 2014, 35, 1249-1259.	1.1	56
33	Genetic association and transcriptome integration identify contributing genes and tissues at cystic fibrosis modifier loci. PLoS Genetics, 2019, 15, e1008007.	1.5	56
34	A novel lung disease phenotype adjusted for mortality attrition for cystic fibrosis Genetic modifier studies. Pediatric Pulmonology, 2011, 46, 857-869.	1.0	50
35	Cystic fibrosis transmembrane conductance regulator and the etiology and pathogenesis of cystic fibrosis. FASEB Journal, 1992, 6, 2775-2782.	0.2	49
36	The microbiome in pediatric cystic fibrosis patients: the role of shared environment suggests a window of intervention. Microbiome, 2014, 2, 14.	4.9	46

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37	Variants in Solute Carrier SLC26A9 Modify Prenatal Exocrine Pancreatic Damage in Cystic Fibrosis. Journal of Pediatrics, 2015, 166, 1152-1157.e6.	0.9	45
38	Capitalizing on the heterogeneous effects of CFTR nonsense and frameshift variants to inform therapeutic strategy for cystic fibrosis. PLoS Genetics, 2018, 14, e1007723.	1.5	44
39	CFTR variant testing: a technical standard of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2020, 22, 1288-1295.	1.1	39
40	A comparison of GABAC and Ï•subunit receptors from the white perch retina. Visual Neuroscience, 1997, 14, 843-851.	0.5	38
41	Deep resequencing of CFTR in 762 F508del homozygotes reveals clusters of non-coding variants associated with cystic fibrosis disease traits. Human Genome Variation, 2016, 3, 16038.	0.4	34
42	Genetic Modifiers of Cystic Fibrosis-Related Diabetes Have Extensive Overlap With Type 2 Diabetes and Related Traits. Journal of Clinical Endocrinology and Metabolism, 2020, 105, 1401-1415.	1.8	34
43	Cystic fibrosis transmembrane conductance regulator modulators reduce the risk of recurrent acute pancreatitis among adult patients with pancreas sufficient cystic fibrosis. Pancreatology, 2019, 19, 1023-1026.	0.5	33
44	Increased expression of anion transporter SLC26A9 delays diabetes onset in cystic fibrosis. Journal of Clinical Investigation, 2019, 130, 272-286.	3.9	33
45	Annotating DNA Variants Is the Next Major Goal for Human Genetics. American Journal of Human Genetics, 2014, 94, 5-10.	2.6	32
46	Codon bias and the folding dynamics of the cystic fibrosis transmembrane conductance regulator. Cellular and Molecular Biology Letters, 2016, 21, 23.	2.7	32
47	Loss of carbonic anhydrase XII function in individuals with elevated sweat chloride concentration and pulmonary airway disease. Human Molecular Genetics, 2016, 25, 1923-1933.	1.4	32
48	Gene Expression in Transformed Lymphocytes Reveals Variation in Endomembrane and HLA Pathways Modifying Cystic Fibrosis Pulmonary Phenotypes. American Journal of Human Genetics, 2015, 96, 318-328.	2.6	28
49	Creation and characterization of an airway epithelial cell line for stable expression of CFTR variants. Journal of Cystic Fibrosis, 2016, 15, 285-294.	0.3	28
50	Clinical and genetic studies of an autosomal dominant cone-rod dystrophy with features of Stargardt disease. Ophthalmic Genetics, 1999, 20, 71-81.	0.5	27
51	A deep learning approach to identify gene targets of a therapeutic for human splicing disorders. Nature Communications, 2021, 12, 3332.	5.8	26
52	Integrated Transcriptomic and Proteomic Analysis of Human Eccrine Sweat Glands Identifies Missing and Novel Proteins. Molecular and Cellular Proteomics, 2019, 18, 1382-1395.	2.5	25
53	Evaluation of both exonic and intronic variants for effects on RNA splicing allows for accurate assessment of the effectiveness of precision therapies. PLoS Genetics, 2020, 16, e1009100.	1.5	23
54	The relationship of lung function with ambient temperature. PLoS ONE, 2018, 13, e0191409.	1.1	21

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55	Development and Evaluation of a PCR-based, Line Probe Assay for the Detection of 58 Alleles in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) Gene. Clinical Chemistry, 2002, 48, 1121-1123.	1.5	19
56	Transformative therapies for rare CFTR missense alleles. Current Opinion in Pharmacology, 2017, 34, 76-82.	1.7	19
57	The genetics and genomics of cystic fibrosis. Journal of Cystic Fibrosis, 2020, 19, S5-S9.	0.3	18
58	Out-of-clinic measurement of sweat chloride using a wearable sensor during low-intensity exercise. Npj Digital Medicine, 2020, 3, 49.	5.7	17
59	Cystic fibrosisâ€"related diabetes onset can be predicted using biomarkers measured at birth. Genetics in Medicine, 2021, 23, 927-933.	1.1	17
60	Identification of a novel nonsense mutation (L88X) in exon 3 of the cystic fibrosis transmembrane conductance regulator gene in a native Korean cystic fibrosis chromosome. Human Mutation, 1992, 1, 501-502.	1.1	15
61	AGTR2 absence or antagonism prevents cystic fibrosis pulmonary manifestations. Journal of Cystic Fibrosis, 2019, 18, 127-134.	0.3	15
62	Respiratory pathogens mediate the association between lung function and temperature in cystic fibrosis. Journal of Cystic Fibrosis, 2016, 15, 794-801.	0.3	14
63	A sequence upstream of canonical PDZ-binding motif within CFTR COOH-terminus enhances NHERF1 interaction. American Journal of Physiology - Lung Cellular and Molecular Physiology, 2016, 311, L1170-L1182.	1.3	13
64	Complete CFTR gene sequencing in 5,058 individuals with cystic fibrosis informs variant-specific treatment. Journal of Cystic Fibrosis, 2022, 21, 463-470.	0.3	13
65	A new cover and new challenges forHuman Mutation. Human Mutation, 2008, 29, 1-1.	1.1	12
66	Common miR-590 Variant rs6971711 Present Only in African Americans Reduces miR-590 Biogenesis. PLoS ONE, 2016, 11, e0156065.	1.1	12
67	SLC26A9 SNP rs7512462 is not associated with lung disease severity or lung function response to ivacaftor in cystic fibrosis patients with G551D-CFTR. Journal of Cystic Fibrosis, 2021, 20, 851-856.	0.3	11
68	Verifying nomenclature of DNA variants in submitted manuscripts: Guidance for journals. Human Mutation, 2021, 42, 3-7.	1.1	10
69	Novel variation at chr11p13 associated with cystic fibrosis lung disease severity. Human Genome Variation, 2016, 3, 16020.	0.4	9
70	Treating Specific Variants Causing Cystic Fibrosis. JAMA - Journal of the American Medical Association, 2017, 318, 2130.	3.8	9
71	Improving imputation in disease-relevant regions: lessons from cystic fibrosis. Npj Genomic Medicine, 2018, 3, 8.	1.7	9
72	Caution advised in the use of CFTR modulator treatment for individuals harboring specific CFTR variants. Journal of Cystic Fibrosis, 2022, 21, 856-860.	0.3	8

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73	Cystic fibrosis transmembrane conductance regulator function, not TAS2R38 gene haplotypes, predict sinus surgery in children and young adults with cystic fibrosis. International Forum of Allergy and Rhinology, 2020, 10, 748-754.	1.5	7
74	Two steps closer to gene therapy for cystic fibrosis. Nature Genetics, 1992, 2, 4-5.	9.4	6
75	â€œMutation in briefâ€•online articles discontinued-introducing â€œbrief reportsâ€• Human Mutation, 2011, 32, 1-1.	1.1	6
76	Lessons from the CAGIâ€4 Hopkins clinical panel challenge. Human Mutation, 2017, 38, 1155-1168.	1.1	6
77	CFTRvariants are associated with chronic bronchitis in smokers. European Respiratory Journal, 2022, 60, 2101994.	3.1	6
78	Leveraging TOPMed imputation server and constructing a cohort-specific imputation reference panel to enhance genotype imputation among cystic fibrosis patients. Human Genetics and Genomics Advances, 2022, 3, 100090.	1.0	6
79	Genetic heterogeneity and cystic fibrosis. Human Mutation, 2009, 30, v-v.	1.1	5
80	Genetic Normalization of Differentiating Aneuploid Human Embryos. Nature Precedings, 2011, , .	0.1	4
81	Sensitivity of the denaturing gradient gel electrophoresis technique in detection of known mutations and novel Asian mutations in the CFTR gene. Human Mutation, 1997, 9, 136-147.	1.1	4
82	Accurate assignment of disease liability to genetic variants using only population data. Genetics in Medicine, 2022, 24, 87-99.	1.1	4
83	Bias in CFTR screening panels. Genetics in Medicine, 2016, 18, 209-209.	1.1	3
84	Potentially lethal cystic fibrosis gene variant in the orangutan. American Journal of Primatology, 2020, 83, e23097.	0.8	3
85	Comparing encounter-based and annualized chronic pseudomonas infection definitions in cystic fibrosis. Journal of Cystic Fibrosis, 2022, 21, 40-44.	0.3	3
86	Response to Biesecker etÂal.. American Journal of Human Genetics, 2021, 108, 1807-1808.	2.6	3
87	CFTR bearing variant p.Phe312del exhibits function inconsistent with phenotype and negligible response to ivacaftor. JCI Insight, 2022, 7, .	2.3	3
88	DNA sequencing analysis of cystic fibrosis transmembrane conductance regulator gene identifies cystic fibrosisâ€•associated variants in the Severe Asthma Research Program. Pediatric Pulmonology, 2022, 57, 1782-1788.	1.0	3
89	A novel ?-thalassemia intermedia phenotype containing Nt494+129T?C and NT494+132C?A mutationsin cis and a Nt168C?T (?o 39 point) mutationin trans. American Journal of Hematology, 2001, 67, 57-58.	2.0	2
90	Mandatory variant submission-Our experiences. Human Mutation, 2012, 33, 1-1.	1.1	2

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91	Predictive effects of low birth weight and small for gestational age status on respiratory and nutritional outcomes in cystic fibrosis. Journal of Cystic Fibrosis, 2020, 19, 888-895.	0.3	2
92	25 Years ofHuman Mutation. Human Mutation, 2016, 37, 503-504.	1.1	1
93	Characterizing mucociliary clearance in young children with cystic fibrosis. Pediatric Research, 2021, , .	1.1	1
94	Accounting for population structure in genetic studies of cystic fibrosis. Human Genetics and Genomics Advances, 2022, 3, 100117.	1.0	1
95	Cystic Fibrosis: Using genetic association to identify modifiers of disease variability in cystic fibrosis. European Journal of Human Genetics, 2006, 14, 890-891.	1.4	0
96	Future challenges for human mutation research. Human Mutation, 2007, 28, 929-930.	1.1	0
97	Title is missing!. , 2020, 16, e1009100.		0
98	Title is missing!. , 2020, 16, e1009100.		0
99	Title is missing!. , 2020, 16, e1009100.		0
100	Title is missing!. , 2020, 16, e1009100.		0
101	Mining GWAS and eQTL data for CF lung disease modifiers by gene expression imputation. , 2020, 15, e0239189.		0
102	Mining GWAS and eQTL data for CF lung disease modifiers by gene expression imputation. , 2020, 15, e0239189.		0
103	Mining GWAS and eQTL data for CF lung disease modifiers by gene expression imputation. , 2020, 15, e0239189.		0
104	Mining GWAS and eQTL data for CF lung disease modifiers by gene expression imputation. , 2020, 15, e0239189.		0