Johanna M Rommens

List of Publications by Year
in descending order

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63 papers	10,605 citations	117453 34 h-index	143772 57 g-index
70 all docs	70 docs citations	70 times ranked	10722 citing authors

#	Article	IF	Citations
1	The Wilson disease gene is a putative copper transporting Pâ€“type ATPase similar to the Menkes gene. Nature Genetics, 1993, 5, 327-337.	9.4	1,855
2	Bone progenitor dysfunction induces myelodysplasia and secondary leukaemia. Nature, 2010, 464, 852-857.	13.7	980
3	De novo truncating mutations in E6-AP ubiquitin-protein ligase gene (UBE3A) in Angelman syndrome. Nature Genetics, 1997, 15, 74-77.	9.4	801
4	Short GCG expansions in the PABP2 gene cause oculopharyngeal muscular dystrophy. Nature Genetics, 1998, 18, 164-167.	9.4	751
5	The Relation between Genotype and Phenotype in Cystic Fibrosis â€” Analysis of the Most Common Mutation (Î”F₅₀₈). New England Journal of Medicine, 1990, 323, 1517-1522.	13.9	717
6	Mutations in SBDS are associated with Shwachmanâ€“Diamond syndrome. Nature Genetics, 2003, 33, 97-101.	9.4	688
7	Defining the disease liability of variants in the cystic fibrosis transmembrane conductance regulator gene. Nature Genetics, 2013, 45, 1160-1167.	9.4	513
8	A candidate prostate cancer susceptibility gene at chromosome 17p. Nature Genetics, 2001, 27, 172-180.	9.4	504
9	Mutations in the homeodomain of the human SIX3 gene cause holoprosencephaly. Nature Genetics, 1999, 22, 196-198.	9.4	398
10	BRCA2 germline mutations in male breast cancer cases and breast cancer families. Nature Genetics, 1996, 13, 123-125.	9.4	315
11	Multi-ion pore behaviour in the CFTR chloride channel. Nature, 1993, 366, 79-82.	13.7	246
12	Genome-wide association meta-analysis identifies five modifier loci of lung disease severity in cystic fibrosis. Nature Communications, 2015, 6, 8382.	5.8	242
13	Shwachman syndrome: Phenotypic manifestations of sibling sets and isolated cases in a large patient cohort are similar. Journal of Pediatrics, 1999, 135, 81-88.	0.9	231
14	Genome-wide association and linkage identify modifier loci of lung disease severity in cystic fibrosis at 11p13 and 20q13.2. Nature Genetics, 2011, 43, 539-546.	9.4	209
15	The cystic fibrosis mutation (Î”F508) does not influence the chloride channel activity of CFTR. Nature Genetics, 1993, 3, 311-316.	9.4	183
16	Draft consensus guidelines for diagnosis and treatment of Shwachmanâ€Diamond syndrome. Annals of the New York Academy of Sciences, 2011, 1242, 40-55.	1.8	183
17	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
18	Genetic Modifiers of Cystic Fibrosisâ€“Related Diabetes. Diabetes, 2013, 62, 3627-3635.	0.3	148

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19	Biallelic mutations in DNAJC21 cause Shwachman-Diamond syndrome. Blood, 2017, 129, 1557-1562.	0.6	104
20	Unraveling the complex genetic model for cystic fibrosis: pleiotropic effects of modifier genes on early cystic fibrosis-related morbidities. Human Genetics, 2014, 133, 151-161.	1.8	100
21	Cystic fibrosis gene modifierSLC26A9modulates airway response to CFTR-directed therapeutics. Human Molecular Genetics, 2016, 25, ddw290.	1.4	81
22	Determinants of the Nuclear Localization of the Heterodimeric DNA Fragmentation Factor (Icad/Cad). Journal of Cell Biology, 2000, 150, 321-334.	2.3	80
23	Serum pancreatic enzymes define the pancreatic phenotype in patients with Shwachman-Diamond syndrome. Journal of Pediatrics, 2002, 141, 259-265.	0.9	79
24	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
25	Evidence for a Causal Relationship Between Early Exocrine Pancreatic Disease and Cystic Fibrosisâ€"Related Diabetes: A Mendelian Randomization Study. Diabetes, 2014, 63, 2114-2119.	0.3	70
26	Phenotypic profiling of CFTR modulators in patient-derived respiratory epithelia. Npj Genomic Medicine, 2017, 2, 12.	1.7	66
27	Shwachman-Bodian Diamond syndrome is a multi-functional protein implicated in cellular stress responses. Human Molecular Genetics, 2009, 18, 3684-3695.	1.4	65
28	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
29	Genetic association and transcriptome integration identify contributing genes and tissues at cystic fibrosis modifier loci. PLoS Genetics, 2019, 15, e1008007.	1.5	56
30	Identification of an Alu retrotransposition event in close proximity to a strong candidate gene for Huntington's disease. Nature, 1993, 362, 370-373.	13.7	50
31	Deficiency of the ribosome biogenesis gene Sbds in hematopoietic stem and progenitor cells causes neutropenia in mice by attenuating lineage progression in myelocytes. Haematologica, 2015, 100, 1285-1293.	1.7	49
32	A Joint Location-Scale Test Improves Power to Detect Associated SNPs, Gene Sets, and Pathways. American Journal of Human Genetics, 2015, 97, 125-138.	2.6	48
33	Variants in Solute Carrier SLC26A9 Modify Prenatal Exocrine Pancreatic Damage in Cystic Fibrosis. Journal of Pediatrics, 2015, 166, 1152-1157.e6.	0.9	45
34	Deficiency of Sbds in the Mouse Pancreas Leads to Features of Shwachmanâ€"Diamond Syndrome, With Loss of Zymogen Granules. Gastroenterology, 2012, 143, 481-492.	0.6	40
35	In Vivo Senescence in the Sbds-Deficient Murine Pancreas: Cell-Type Specific Consequences of Translation Insufficiency. PLoS Genetics, 2015, 11, e1005288.	1.5	37
36	Prevalence of meconium ileus marks the severity of mutations of the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene. Genetics in Medicine, 2016, 18, 333-340.	1.1	37

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37	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
38	SBDS-Deficient Cells Have an Altered Homeostatic Equilibrium due to Translational Inefficiency Which Explains their Reduced Fitness and Provides a Logical Framework for Intervention. PLoS Genetics, 2017, 13, e1006552.	1.5	31
39	Human Homologs of Ubc6p Ubiquitin-conjugating Enzyme and Phosphorylation of HsUbc6e in Response to Endoplasmic Reticulum Stress. Journal of Biological Chemistry, 2006, 281, 21480-21490.	1.6	30
40	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
41	LocusFocus: Web-based colocalization for the annotation and functional follow-up of GWAS. PLoS Computational Biology, 2020, 16, e1008336.	1.5	28
42	SLC6A14, an amino acid transporter, modifies the primary CF defect in fluid secretion. ELife, 2018, 7, .	2.8	25
43	Mammographic density defined by higher than conventional brightness thresholds better predicts breast cancer risk. International Journal of Epidemiology, 2017, 46, dyw212.	0.9	24
44	Positive epistasis between disease-causing missense mutations and silent polymorphism with effect on mRNA translation velocity. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, .	3.3	18
45	Cystic fibrosisâ€“related diabetes onset can be predicted using biomarkers measured at birth. Genetics in Medicine, 2021, 23, 927-933.	1.1	17
46	Search for the FSHD gene using cDNA selection in a region spanning 100 kb on chromosome 4q35. Muscle and Nerve, 1995, 18, S19-S26.	1.0	13
47	Exclusion of linkage of Shwachman-Diamond syndrome to chromosome regions 6q and 12q implicated by a de novo translocation. , 1999, 85, 171-174.		12
48	Improving imputation in disease-relevant regions: lessons from cystic fibrosis. Npj Genomic Medicine, 2018, 3, 8.	1.7	9
49	Transcription mapping and expression analysis of candidate genes in the vicinity of the mouse Loop-tail mutation. Mammalian Genome, 2000, 11, 633-638.	1.0	8
50	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
51	Screening for Regulatory Variants in 460 kb Encompassing the CFTR Locus in Cystic Fibrosis Patients. Journal of Molecular Diagnostics, 2019, 21, 70-80.	1.2	7
52	Genetic evidence supports the development of SLC26A9 targeting therapies for the treatment of lung disease. Npj Genomic Medicine, 2022, 7, 28.	1.7	7
53	Dinucleotide repeat polymorphism near the RP3 locus in Xp21 (DXS1110). Human Molecular Genetics, 1993, 2, 821-821.	1.4	5
54	Editorial overview: Molecular and genetic bases of disease: Enter the post-GWAS era. Current Opinion in Genetics and Development, 2015, 33, 77-79.	1.5	5

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55	Bias in CFTR screening panels. Genetics in Medicine, 2016, 18, 209-209.	1.1	3
56	Expression of cystic fibrosis lung disease modifier genes in human airway models. Journal of Cystic Fibrosis, 2022, 21, 616-622.	0.3	3
57	Shwachman-Diamond Syndrome: Development of a North American Registry to Assess Long-Term Outcomes, Risk of Leukemia and Other Complications.. Blood, 2009, 114, 1363-1363.	0.6	2
58	Mouse Model for Shwachman-Diamond Syndrome with the R126T Disease Mutation Leads to Severe Growth and Developmental Deficiencies with Impairment of Hematopoiesis.. Blood, 2006, 108, 1283-1283.	0.6	1
59	Reply to coates and bush. Pediatric Pulmonology, 2004, 37, 382-382.	1.0	0
60	Sbds Deficient Neutrophils Exhibit Normal Numbers, Chemotaxis and Phagocytic Functions, but Impaired NADPH Oxidase Activity.. Blood, 2006, 108, 1634-1634.	0.6	0
61	Quebec Platelet Disorder Is Caused by a Cis-Acting Mutation near the Plasminogen Activator Gene (PLAU) That Increases PLAU Transcription by Megakaryocytes.. Blood, 2008, 112, 3394-3394.	0.6	0
62	Lessons from the Mouse Model. Blood, 2013, 122, SCI-35-SCI-35.	0.6	0
63	Deletion of Sbds from Hematopoietic Progenitors Causes Neutropenia in a Mouse Model of Shwachman-Diamond Syndrome By Specifically Blocking Myeloid Lineage Progression at Late Differentiation Stages. Blood, 2014, 124, 355-355.	0.6	0