

Johanna M Rommens

List of Publications by Citations

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

60
papers

8,944
citations

32
h-index

70
g-index

70
ext. papers

9,944
ext. citations

13.6
avg, IF

4.92
L-index

#	Paper	IF	Citations
60	The Wilson disease gene is a putative copper transporting P-type ATPase similar to the Menkes gene. <i>Nature Genetics</i> , 1993 , 5, 327-37	36.3	1638
59	Bone progenitor dysfunction induces myelodysplasia and secondary leukaemia. <i>Nature</i> , 2010 , 464, 852-750.4	50.4	815
58	De novo truncating mutations in E6-AP ubiquitin-protein ligase gene (UBE3A) in Angelman syndrome. <i>Nature Genetics</i> , 1997 , 15, 74-7	36.3	677
57	Short GCG expansions in the PABP2 gene cause oculopharyngeal muscular dystrophy. <i>Nature Genetics</i> , 1998 , 18, 164-7	36.3	656
56	The relation between genotype and phenotype in cystic fibrosis--analysis of the most common mutation (delta F508). <i>New England Journal of Medicine</i> , 1990 , 323, 1517-22	59.2	621
55	Mutations in SBDS are associated with Shwachman-Diamond syndrome. <i>Nature Genetics</i> , 2003 , 33, 97-103	36.3	573
54	A candidate prostate cancer susceptibility gene at chromosome 17p. <i>Nature Genetics</i> , 2001 , 27, 172-80	36.3	469
53	Defining the disease liability of variants in the cystic fibrosis transmembrane conductance regulator gene. <i>Nature Genetics</i> , 2013 , 45, 1160-7	36.3	413
52	Mutations in the homeodomain of the human SIX3 gene cause holoprosencephaly. <i>Nature Genetics</i> , 1999 , 22, 196-8	36.3	360
51	BRCA2 germline mutations in male breast cancer cases and breast cancer families. <i>Nature Genetics</i> , 1996 , 13, 123-5	36.3	289
50	Multi-ion pore behaviour in the CFTR chloride channel. <i>Nature</i> , 1993 , 366, 79-82	50.4	226
49	Shwachman syndrome: phenotypic manifestations of sibling sets and isolated cases in a large patient cohort are similar. <i>Journal of Pediatrics</i> , 1999 , 135, 81-8	3.6	200
48	Genome-wide association and linkage identify modifier loci of lung disease severity in cystic fibrosis at 11p13 and 20q13.2. <i>Nature Genetics</i> , 2011 , 43, 539-46	36.3	181
47	Genome-wide association meta-analysis identifies five modifier loci of lung disease severity in cystic fibrosis. <i>Nature Communications</i> , 2015 , 6, 8382	17.4	179
46	The cystic fibrosis mutation (delta F508) does not influence the chloride channel activity of CFTR. <i>Nature Genetics</i> , 1993 , 3, 311-6	36.3	170
45	Draft consensus guidelines for diagnosis and treatment of Shwachman-Diamond syndrome. <i>Annals of the New York Academy of Sciences</i> , 2011 , 1242, 40-55	6.5	146
44	Multiple apical plasma membrane constituents are associated with susceptibility to meconium ileus in individuals with cystic fibrosis. <i>Nature Genetics</i> , 2012 , 44, 562-9	36.3	139

43	Genetic modifiers of cystic fibrosis-related diabetes. <i>Diabetes</i> , 2013 , 62, 3627-35	0.9	116
42	Unraveling the complex genetic model for cystic fibrosis: pleiotropic effects of modifier genes on early cystic fibrosis-related morbidities. <i>Human Genetics</i> , 2014 , 133, 151-61	6.3	81
41	Biallelic mutations in cause Shwachman-Diamond syndrome. <i>Blood</i> , 2017 , 129, 1557-1562	2.2	80
40	Determinants of the nuclear localization of the heterodimeric DNA fragmentation factor (ICAD/CAD). <i>Journal of Cell Biology</i> , 2000 , 150, 321-34	7.3	75
39	Serum pancreatic enzymes define the pancreatic phenotype in patients with Shwachman-Diamond syndrome. <i>Journal of Pediatrics</i> , 2002 , 141, 259-65	3.6	71
38	Cystic fibrosis gene modifier SLC26A9 modulates airway response to CFTR-directed therapeutics. <i>Human Molecular Genetics</i> , 2016 , 25, 4590-4600	5.6	62
37	Shwachman-Bodian Diamond syndrome is a multi-functional protein implicated in cellular stress responses. <i>Human Molecular Genetics</i> , 2009 , 18, 3684-95	5.6	58
36	Evidence for a causal relationship between early exocrine pancreatic disease and cystic fibrosis-related diabetes: a Mendelian randomization study. <i>Diabetes</i> , 2014 , 63, 2114-9	0.9	56
35	Identification of an Alu retrotransposition event in close proximity to a strong candidate gene for Huntington's disease. <i>Nature</i> , 1993 , 362, 370-3	50.4	48
34	Sources of Variation in Sweat Chloride Measurements in Cystic Fibrosis. <i>American Journal of Respiratory and Critical Care Medicine</i> , 2016 , 194, 1375-1382	10.2	46
33	Phenotypic profiling of CFTR modulators in patient-derived respiratory epithelia. <i>Npj Genomic Medicine</i> , 2017 , 2, 12	6.2	46
32	Correlating Cystic Fibrosis Transmembrane Conductance Regulator Function with Clinical Features to Inform Precision Treatment of Cystic Fibrosis. <i>American Journal of Respiratory and Critical Care Medicine</i> , 2019 , 199, 1116-1126	10.2	41
31	Deficiency of the ribosome biogenesis gene Sbds in hematopoietic stem and progenitor cells causes neutropenia in mice by attenuating lineage progression in myelocytes. <i>Haematologica</i> , 2015 , 100, 1285-93	6.6	39
30	Variants in Solute Carrier SLC26A9 Modify Prenatal Exocrine Pancreatic Damage in Cystic Fibrosis. <i>Journal of Pediatrics</i> , 2015 , 166, 1152-1157.e6	3.6	36
29	Genetic association and transcriptome integration identify contributing genes and tissues at cystic fibrosis modifier loci. <i>PLoS Genetics</i> , 2019 , 15, e1008007	6	33
28	Deficiency of Sbds in the mouse pancreas leads to features of Shwachman-Diamond syndrome, with loss of zymogen granules. <i>Gastroenterology</i> , 2012 , 143, 481-92	13.3	30
27	Prevalence of meconium ileus marks the severity of mutations of the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene. <i>Genetics in Medicine</i> , 2016 , 18, 333-40	8.1	28
26	A Joint Location-Scale Test Improves Power to Detect Associated SNPs, Gene Sets, and Pathways. <i>American Journal of Human Genetics</i> , 2015 , 97, 125-38	11	28

25	In Vivo Senescence in the Sbds-Deficient Murine Pancreas: Cell-Type Specific Consequences of Translation Insufficiency. <i>PLoS Genetics</i> , 2015 , 11, e1005288	6	28
24	SBDS-Deficient Cells Have an Altered Homeostatic Equilibrium due to Translational Inefficiency Which Explains their Reduced Fitness and Provides a Logical Framework for Intervention. <i>PLoS Genetics</i> , 2017 , 13, e1006552	6	24
23	Human homologs of Ubc6p ubiquitin-conjugating enzyme and phosphorylation of HsUbc6e in response to endoplasmic reticulum stress. <i>Journal of Biological Chemistry</i> , 2006 , 281, 21480-21490	5.4	23
22	Gene expression in transformed lymphocytes reveals variation in endomembrane and HLA pathways modifying cystic fibrosis pulmonary phenotypes. <i>American Journal of Human Genetics</i> , 2015 , 96, 318-28	11	20
21	Mammographic density defined by higher than conventional brightness thresholds better predicts breast cancer risk. <i>International Journal of Epidemiology</i> , 2017 , 46, 652-661	7.8	18
20	SLC6A14, an amino acid transporter, modifies the primary CF defect in fluid secretion. <i>ELife</i> , 2018 , 7,	8.9	17
19	Genetic Modifiers of Cystic Fibrosis-Related Diabetes Have Extensive Overlap With Type 2 Diabetes and Related Traits. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2020 , 105,	5.6	16
18	Exclusion of linkage of Shwachman-Diamond syndrome to chromosome regions 6q and 12q implicated by a de novo translocation. <i>American Journal of Medical Genetics Part A</i> , 1999 , 85, 171-4		11
17	Search for the FSHD gene using cDNA selection in a region spanning 100 kb on chromosome 4q35. <i>Muscle and Nerve</i> , 1995 , 18, S19-S26	3.4	9
16	LocusFocus: Web-based colocalization for the annotation and functional follow-up of GWAS. <i>PLoS Computational Biology</i> , 2020 , 16, e1008336	5	9
15	Transcription mapping and expression analysis of candidate genes in the vicinity of the mouse Loop-tail mutation. <i>Mammalian Genome</i> , 2000 , 11, 633-8	3.2	8
14	Improving imputation in disease-relevant regions: lessons from cystic fibrosis. <i>Npj Genomic Medicine</i> , 2018 , 3, 8	6.2	6
13	Dinucleotide repeat polymorphism near the RP3 locus in Xp21 (DXS1110). <i>Human Molecular Genetics</i> , 1993 , 2, 821	5.6	5
12	Screening for Regulatory Variants in 460 kb Encompassing the CFTR Locus in Cystic Fibrosis Patients. <i>Journal of Molecular Diagnostics</i> , 2019 , 21, 70-80	5.1	5
11	Positive epistasis between disease-causing missense mutations and silent polymorphism with effect on mRNA translation velocity. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2021 , 118,	11.5	5
10	Bias in CFTR screening panels. <i>Genetics in Medicine</i> , 2016 , 18, 209	8.1	3
9	Cystic fibrosis-related diabetes onset can be predicted using biomarkers measured at birth. <i>Genetics in Medicine</i> , 2021 , 23, 927-933	8.1	3
8	Shwachman-Diamond Syndrome: Development of a North American Registry to Assess Long-Term Outcomes, Risk of Leukemia and Other Complications.. <i>Blood</i> , 2009 , 114, 1363-1363	2.2	2

7	Genetic evidence supports the development of SLC26A9 targeting therapies for the treatment of lung disease.. <i>Npj Genomic Medicine</i> , 2022 , 7, 28	6.2	o
6	Reply to coates and bush. <i>Pediatric Pulmonology</i> , 2004 , 37, 382-382	3.5	
5	Sbds Deficient Neutrophils Exhibit Normal Numbers, Chemotaxis and Phagocytic Functions, but Impaired NADPH Oxidase Activity.. <i>Blood</i> , 2006 , 108, 1634-1634	2.2	
4	Mouse Model for Shwachman-Diamond Syndrome with the R126T Disease Mutation Leads to Severe Growth and Developmental Deficiencies with Impairment of Hematopoiesis.. <i>Blood</i> , 2006 , 108, 1283-1283	2.2	
3	Cystic Fibrosis Gene: Identification1-4		
2	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. <i>Blood</i> , 2014 , 124, 355-355	2.2	
1	Quebec Platelet Disorder Is Caused by a Cis-Acting Mutation near the Plasminogen Activator Gene (PLAU) That Increases PLAU Transcription by Megakaryocytes.. <i>Blood</i> , 2008 , 112, 3394-3394	2.2	