Johanna M Rommens

List of Publications by Year in Descending Order

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	Genetic evidence supports the development of SLC26A9 targeting therapies for the treatment of lung disease.. *Npj Genomic Medicine*, **2022**, 7, 28	6.2	0
59	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,	11.5	5
58	Cystic fibrosis-related diabetes onset can be predicted using biomarkers measured at birth. *Genetics in Medicine*, **2021**, 23, 927-933	8.1	3
57	LocusFocus: Web-based colocalization for the annotation and functional follow-up of GWAS. *PLoS Computational Biology*, **2020**, 16, e1008336	5	9
56	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,	5.6	16
55	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	10.2	41
54	Genetic association and transcriptome integration identify contributing genes and tissues at cystic fibrosis modifier loci. *PLoS Genetics*, **2019**, 15, e1008007	6	33
53	Screening for Regulatory Variants in 460 kb Encompassing the CFTR Locus in Cystic Fibrosis Patients. *Journal of Molecular Diagnostics*, **2019**, 21, 70-80	5.1	5
52	Improving imputation in disease-relevant regions: lessons from cystic fibrosis. *Npj Genomic Medicine* , **2018**, 3, 8	6.2	6
51	SLC6A14, an amino acid transporter, modifies the primary CF defect in fluid secretion. *ELife*, **2018**, 7,	8.9	17
50	Biallelic mutations in cause Shwachman-Diamond syndrome. *Blood*, **2017**, 129, 1557-1562	2.2	80
49	Mammographic density defined by higher than conventional brightness thresholds better predicts breast cancer risk. *International Journal of Epidemiology*, **2017**, 46, 652-661	7.8	18
48	Phenotypic profiling of CFTR modulators in patient-derived respiratory epithelia. *Npj Genomic Medicine*, **2017**, 2, 12	6.2	46
47	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	6	24
46	Prevalence of meconium ileus marks the severity of mutations of the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene. *Genetics in Medicine*, **2016**, 18, 333-40	8.1	28
45	Sources of Variation in Sweat Chloride Measurements in Cystic Fibrosis. *American Journal of Respiratory and Critical Care Medicine*, **2016**, 194, 1375-1382	10.2	46
44	Bias in CFTR screening panels. *Genetics in Medicine*, **2016**, 18, 209	8.1	3

43	Cystic fibrosis gene modifier SLC26A9 modulates airway response to CFTR-directed therapeutics. *Human Molecular Genetics*, **2016**, 25, 4590-4600	5.6	62
42	Variants in Solute Carrier SLC26A9 Modify Prenatal Exocrine Pancreatic Damage in Cystic Fibrosis. *Journal of Pediatrics*, **2015**, 166, 1152-1157.e6	3.6	36
41	Genome-wide association meta-analysis identifies five modifier loci of lung disease severity in cystic fibrosis. *Nature Communications*, **2015**, 6, 8382	17.4	179
40	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, 1285-93	6.6	39
39	A Joint Location-Scale Test Improves Power to Detect Associated SNPs, Gene Sets, and Pathways. *American Journal of Human Genetics*, **2015**, 97, 125-38	11	28
38	In Vivo Senescence in the Sbds-Deficient Murine Pancreas: Cell-Type Specific Consequences of Translation Insufficiency. *PLoS Genetics*, **2015**, 11, e1005288	6	28
37	Gene expression in transformed lymphocytes reveals variation in endomembrane and HLA pathways modifying cystic fibrosis pulmonary phenotypes. *American Journal of Human Genetics*, **2015**, 96, 318-28	11	20
36	Evidence for a causal relationship between early exocrine pancreatic disease and cystic fibrosis-related diabetes: a Mendelian randomization study. *Diabetes*, **2014**, 63, 2114-9	0.9	56
35	Unraveling the complex genetic model for cystic fibrosis: pleiotropic effects of modifier genes on early cystic fibrosis-related morbidities. *Human Genetics*, **2014**, 133, 151-61	6.3	81
34	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	2.2	
33	Genetic modifiers of cystic fibrosis-related diabetes. *Diabetes*, **2013**, 62, 3627-35	0.9	116
32	Defining the disease liability of variants in the cystic fibrosis transmembrane conductance regulator gene. *Nature Genetics*, **2013**, 45, 1160-7	36.3	413
31	Deficiency of Sbds in the mouse pancreas leads to features of Shwachman-Diamond syndrome, with loss of zymogen granules. *Gastroenterology*, **2012**, 143, 481-92	13.3	30
30	Multiple apical plasma membrane constituents are associated with susceptibility to meconium ileus in individuals with cystic fibrosis. *Nature Genetics*, **2012**, 44, 562-9	36.3	139
29	Genome-wide association and linkage identify modifier loci of lung disease severity in cystic fibrosis at 11p13 and 20q13.2. *Nature Genetics*, **2011**, 43, 539-46	36.3	181
28	Draft consensus guidelines for diagnosis and treatment of Shwachman-Diamond syndrome. *Annals of the New York Academy of Sciences*, **2011**, 1242, 40-55	6.5	146
27	Bone progenitor dysfunction induces myelodysplasia and secondary leukaemia. *Nature*, **2010**, 464, 852-7	50.4	815
26	Shwachman-Bodian Diamond syndrome is a multi-functional protein implicated in cellular stress responses. *Human Molecular Genetics*, **2009**, 18, 3684-95	5.6	58

25	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	2.2	2
24	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	2.2	
23	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	5.4	23
22	Sbds Deficient Neutrophils Exhibit Normal Numbers, Chemotaxis and Phagocytic Functions, but Impaired NADPH Oxidase Activity.. *Blood*, **2006**, 108, 1634-1634	2.2	
21	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	2.2	
20	Reply to coates and bush. *Pediatric Pulmonology*, **2004**, 37, 382-382	3.5	
19	Mutations in SBDS are associated with Shwachman-Diamond syndrome. *Nature Genetics*, **2003**, 33, 97-101	36.3	573
18	Serum pancreatic enzymes define the pancreatic phenotype in patients with Shwachman-Diamond syndrome. *Journal of Pediatrics*, **2002**, 141, 259-65	3.6	71
17	A candidate prostate cancer susceptibility gene at chromosome 17p. *Nature Genetics*, **2001**, 27, 172-80	36.3	469
16	Transcription mapping and expression analysis of candidate genes in the vicinity of the mouse Loop-tail mutation. *Mammalian Genome*, **2000**, 11, 633-8	3.2	8
15	Determinants of the nuclear localization of the heterodimeric DNA fragmentation factor (ICAD/CAD). *Journal of Cell Biology*, **2000**, 150, 321-34	7.3	75
14	Mutations in the homeodomain of the human SIX3 gene cause holoprosencephaly. *Nature Genetics*, **1999**, 22, 196-8	36.3	360
13	Exclusion of linkage of Shwachman-Diamond syndrome to chromosome regions 6q and 12q implicated by a de novo translocation. *American Journal of Medical Genetics Part A*, **1999**, 85, 171-4		11
12	Shwachman syndrome: phenotypic manifestations of sibling sets and isolated cases in a large patient cohort are similar. *Journal of Pediatrics*, **1999**, 135, 81-8	3.6	200
11	Short GCG expansions in the PABP2 gene cause oculopharyngeal muscular dystrophy. *Nature Genetics*, **1998**, 18, 164-7	36.3	656
10	De novo truncating mutations in E6-AP ubiquitin-protein ligase gene (UBE3A) in Angelman syndrome. *Nature Genetics*, **1997**, 15, 74-7	36.3	677
9	BRCA2 germline mutations in male breast cancer cases and breast cancer families. *Nature Genetics*, **1996**, 13, 123-5	36.3	289
8	Search for the FSHD gene using cDNA selection in a region spanning 100 kb on chromosome 4q35. *Muscle and Nerve*, **1995**, 18, S19-S26	3.4	9

7	Dinucleotide repeat polymorphism near the RP3 locus in Xp21 (DXS1110). *Human Molecular Genetics*, **1993**, 2, 821	5.6	5
6	Multi-ion pore behaviour in the CFTR chloride channel. *Nature*, **1993**, 366, 79-82	50.4	226
5	The cystic fibrosis mutation (delta F508) does not influence the chloride channel activity of CFTR. *Nature Genetics*, **1993**, 3, 311-6	36.3	170
4	The Wilson disease gene is a putative copper transporting P-type ATPase similar to the Menkes gene. *Nature Genetics*, **1993**, 5, 327-37	36.3	1638
3	Identification of an Alu retrotransposition event in close proximity to a strong candidate gene for Huntington's disease. *Nature*, **1993**, 362, 370-3	50.4	48
2	The relation between genotype and phenotype in cystic fibrosis--analysis of the most common mutation (delta F508). *New England Journal of Medicine*, **1990**, 323, 1517-22	59.2	621
1	Cystic Fibrosis Gene: Identification1-4		