Willem H Ouwehand

List of Publications by Year
in descending order

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118 papers	24,378 citations	17440 63 h-index	19190 118 g-index
133 all docs	133 docs citations	133 times ranked	40181 citing authors

#	Article	IF	Citations
1	G proteinâ€“coupled receptor kinase 5 regulates thrombin signaling in platelets via PAR-1. Blood Advances, 2022, 6, 2319-2330.	5.2	8
2	Machine learning optimized polygenic scores for blood cell traits identify sex-specific trajectories and genetic correlations with disease. Cell Genomics, 2022, 2, 100086.	6.5	9
3	Whole-exome sequencing identifies rare genetic variants associated with human plasma metabolites. American Journal of Human Genetics, 2022, 109, 1038-1054.	6.2	17
4	Long-Read Sequencing Identifies the First Retrotransposon Insertion and Resolves Structural Variants Causing Antithrombin Deficiency. Thrombosis and Haemostasis, 2022, 122, 1369-1378.	3.4	9
5	Cell type-specific novel long non-coding RNA and circular RNA in the BLUEPRINT hematopoietic transcriptomes atlas. Haematologica, 2021, 106, 2613-2623.	3.5	12
6	Neutrophil specific granule and NETosis defects in gray platelet syndrome. Blood Advances, 2021, 5, 549-564.	5.2	18
7	A genome-wide meta-analysis yields 46 new loci associating with biomarkers of iron homeostasis. Communications Biology, 2021, 4, 156.	4.4	72
8	Transcriptional characterization of human megakaryocyte polyploidization and lineage commitment. Journal of Thrombosis and Haemostasis, 2021, 19, 1236-1249.	3.8	15
9	Effects of adiposity on the human plasma proteome: observational and Mendelian randomisation estimates. International Journal of Obesity, 2021, 45, 2221-2229.	3.4	31
10	Mitochondrial DNA variants modulate N-formylmethionine, proteostasis and risk of late-onset human diseases. Nature Medicine, 2021, 27, 1564-1575.	30.7	40
11	Large-scale cis- and trans-eQTL analyses identify thousands of genetic loci and polygenic scores that regulate blood gene expression. Nature Genetics, 2021, 53, 1300-1310.	21.4	590
12	Elevated levels of tissue factor pathway inhibitor in patients with mild to moderate bleeding tendency. Blood Advances, 2021, 5, 391-398.	5.2	18
13	Investigating rare pathogenic/likely pathogenic exonic variation in bipolar disorder. Molecular Psychiatry, 2021, 26, 5239-5250.	7.9	15
14	Polygenic basis and biomedical consequences of telomere length variation. Nature Genetics, 2021, 53, 1425-1433.	21.4	145
15	Comparison of four methods to measure haemoglobin concentrations in whole blood donors (<scp>COMPARE</scp>): A diagnostic accuracy study. Transfusion Medicine, 2021, 31, 94-103.	1.1	13
16	Integrative analysis of the plasma proteome and polygenic risk of cardiometabolic diseases. Nature Metabolism, 2021, 3, 1476-1483.	11.9	43
17	The Polygenic and Monogenic Basis of Blood Traits and Diseases. Cell, 2020, 182, 1214-1231.e11.	28.9	388
18	Large genome-wide association study identifies three novel risk variants for restless legs syndrome. Communications Biology, 2020, 3, 703.	4.4	40

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19	Development and validation of a universal blood donor genotyping platform: a multinational prospective study. Blood Advances, 2020, 4, 3495-3506.	5.2	31
20	Novel manifestations of immune dysregulation and granule defects in gray platelet syndrome. Blood, 2020, 136, 1956-1967.	1.4	34
21	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
22	Treatment of COVID-19 with remdesivir in the absence of humoral immunity: a case report. Nature Communications, 2020, 11, 6385.	12.8	103
23	Paired rRNA-depleted and polyA-selected RNA sequencing data and supporting multi-omics data from human T cells. Scientific Data, 2020, 7, 376.	5.3	15
24	Whole-genome sequencing of a sporadic primary immunodeficiency cohort. Nature, 2020, 583, 90-95.	27.8	148
25	Whole-genome sequencing of patients with rare diseases in a national health system. Nature, 2020, 583, 96-102.	27.8	338
26	The influence of rare variants in circulating metabolic biomarkers. PLoS Genetics, 2020, 16, e1008605.	3.5	9
27	Mutational and phenotypic characterization of hereditary hemorrhagic telangiectasia. Blood, 2020, 136, 1907-1918.	1.4	40
28	A coagulation defect arising from heterozygous premature termination of tissue factor. Journal of Clinical Investigation, 2020, 130, 5302-5312.	8.2	17
29	Longer-term efficiency and safety of increasing the frequency of whole blood donation (INTERVAL): extension study of a randomised trial of 20â€ˆ757 blood donors. Lancet Haematology,the, 2019, 6, e510-e520.	4.6	17
30	Germline mutations in the transcription factor IKZF5 cause thrombocytopenia. Blood, 2019, 134, 2070-2081.	1.4	29
31	A catalog of genetic loci associated with kidney function from analyses of a million individuals. Nature Genetics, 2019, 51, 957-972.	21.4	549
32	Curated diseaseâ€causing genes for bleeding, thrombotic, and platelet disorders: Communication from the SSC of the ISTH. Journal of Thrombosis and Haemostasis, 2019, 17, 1253-1260.	3.8	56
33	Diagnostic high-throughput sequencing of 2396 patients with bleeding, thrombotic, and platelet disorders. Blood, 2019, 134, 2082-2091.	1.4	131
34	How common are single gene mutations as a cause for lacunar stroke?. Neurology, 2019, 93, e2007-e2020.	1.1	26
35	Sphingolipid dysregulation due to lack of functional KDSR impairs proplatelet formation causing thrombocytopenia. Haematologica, 2019, 104, 1036-1045.	3.5	28
36	Identification of rare sequence variation underlying heritable pulmonary arterial hypertension. Nature Communications, 2018, 9, 1416.	12.8	279

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37	Loss-of-function nuclear factor Î¹B subunit 1 (NFKB1) variants are the most common monogenic cause of common variable immunodeficiency in Europeans. Journal of Allergy and Clinical Immunology, 2018, 142, 1285-1296.	2.9	185
38	Nbeal2 interacts with Dock7, Sec16a, and Vac14. Blood, 2018, 131, 1000-1011.	1.4	29
39	Complex structural variants in Mendelian disorders: identification and breakpoint resolution using short- and long-read genome sequencing. Genome Medicine, 2018, 10, 95.	8.2	111
40	GNE variants causing autosomal recessive macrothrombocytopenia without associated muscle wasting. Blood, 2018, 132, 1851-1854.	1.4	48
41	Dynamics of Transcription Regulation in Human Bone Marrow Myeloid Differentiation to Mature Blood Neutrophils. Cell Reports, 2018, 24, 2784-2794.	6.4	104
42	Phenotype description and response to thrombopoietin receptor agonist in DIAPH1-related disorder. Blood Advances, 2018, 2, 2341-2346.	5.2	33
43	Automated typing of red blood cell and platelet antigens: a whole-genome sequencing study. Lancet Haematology,the, 2018, 5, e241-e251.	4.6	70
44	De Novo Truncating Mutations in WASF1 Cause Intellectual Disability with Seizures. American Journal of Human Genetics, 2018, 103, 144-153.	6.2	36
45	Genomic atlas of the human plasma proteome. Nature, 2018, 558, 73-79.	27.8	1,180
46	Congenital macrothrombocytopenia with focal myelofibrosis due to mutations in human G6b-B is rescued in humanized mice. Blood, 2018, 132, 1399-1412.	1.4	37
47	The Human Phenotype Ontology in 2017. Nucleic Acids Research, 2017, 45, D865-D876.	14.5	699
48	Rare variants in GP1BB are responsible for autosomal dominant macrothrombocytopenia. Blood, 2017, 129, 520-524.	1.4	42
49	Germline variants in ETV6 underlie reduced platelet formation, platelet dysfunction and increased levels of circulating CD34 ⁺ progenitors. Haematologica, 2017, 102, 282-294.	3.5	70
50	Megakaryocytes in Myeloproliferative Neoplasms Have Unique Somatic Mutations. American Journal of Pathology, 2017, 187, 1512-1522.	3.8	12
51	Common genetic variation drives molecular heterogeneity in human iPSCs. Nature, 2017, 546, 370-375.	27.8	491
52	Detection of Atherosclerotic Inflammation by 68 Ga-DOTATATE PET Compared to [18 F]FDG PET Imaging. Journal of the American College of Cardiology, 2017, 69, 1774-1791.	2.8	321
53	Comprehensive Rare Variant Analysis via Whole-Genome Sequencing to Determine the Molecular Pathology of Inherited Retinal Disease. American Journal of Human Genetics, 2017, 100, 75-90.	6.2	343
54	Dawning of the age of genomics for platelet granule disorders: improving insight, diagnosis and management. British Journal of Haematology, 2017, 176, 705-720.	2.5	22

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55	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
56	Identification of novel risk loci for restless legs syndrome in genome-wide association studies in individuals of European ancestry: a meta-analysis. Lancet Neurology, The, 2017, 16, 898-907.	10.2	191
57	Efficiency and safety of varying the frequency of whole blood donation (INTERVAL): a randomised trial of 45â€˙000 donors. Lancet, The, 2017, 390, 2360-2371.	13.7	222
58	Platelet function is modified by common sequence variation in megakaryocyte super enhancers. Nature Communications, 2017, 8, 16058.	12.8	50
59	Expanded repertoire of RASGRP2 variants responsible for platelet dysfunction and severe bleeding. Blood, 2017, 130, 1026-1030.	1.4	38
60	Chromosome contacts in activated T cells identify autoimmune disease candidate genes. Genome Biology, 2017, 18, 165.	8.8	68
61	Mutations in tropomyosin 4 underlie a rare form of human macrothrombocytopenia. Journal of Clinical Investigation, 2017, 127, 814-829.	8.2	57
62	NBEAL2 is required for neutrophil and NK cell function and pathogen defense. Journal of Clinical Investigation, 2017, 127, 3521-3526.	8.2	25
63	Increased DNA methylation variability in type 1 diabetes across three immune effector cell types. Nature Communications, 2016, 7, 13555.	12.8	142
64	A dominant gain-of-function mutation in universal tyrosine kinase SRC causes thrombocytopenia, myelofibrosis, bleeding, and bone pathologies. Science Translational Medicine, 2016, 8, 328ra30.	12.4	87
65	A high-throughput sequencing test for diagnosing inherited bleeding, thrombotic, and platelet disorders. Blood, 2016, 127, 2791-2803.	1.4	157
66	Inherited platelet disorders: toward DNA-based diagnosis. Blood, 2016, 127, 2814-2823.	1.4	119
67	eFORGE: A Tool for Identifying Cell Type-Specific Signal in Epigenomic Data. Cell Reports, 2016, 17, 2137-2150.	6.4	102
68	A gain-of-function variant in DIAPH1 causes dominant macrothrombocytopenia and hearing loss. Blood, 2016, 127, 2903-2914.	1.4	121
69	Lineage-Specific Genome Architecture Links Enhancers and Non-coding Disease Variants to Target Gene Promoters. Cell, 2016, 167, 1369-1384.e19.	28.9	863
70	The Allelic Landscape of Human Blood Cell Trait Variation and Links to Common Complex Disease. Cell, 2016, 167, 1415-1429.e19.	28.9	1,052
71	Distinct Trends of DNA Methylation Patterning in the Innate and Adaptive Immune Systems. Cell Reports, 2016, 17, 2101-2111.	6.4	54
72	DNA Methylation Dynamics of Human Hematopoietic Stem Cell Differentiation. Cell Stem Cell, 2016, 19, 808-822.	11.1	216

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73	Genetic Drivers of Epigenetic and Transcriptional Variation in Human Immune Cells. Cell, 2016, 167, 1398-1414.e24.	28.9	573
74	Large-scale production of megakaryocytes from human pluripotent stem cells by chemically defined forward programming. Nature Communications, 2016, 7, 11208.	12.8	199
75	Recruitment and representativeness of blood donors in the INTERVAL randomised trial assessing varying inter-donation intervals. Trials, 2016, 17, 458.	1.6	17
76	Rare loss-of-function variants in SETD1A are associated with schizophrenia and developmental disorders. Nature Neuroscience, 2016, 19, 571-577.	14.8	388
77	Leukemia-Associated Somatic Mutations Drive Distinct Patterns of Age-Related Clonal Hemopoiesis. Cell Reports, 2015, 10, 1239-1245.	6.4	443
78	Human phenotype ontology annotation and cluster analysis to unravel genetic defects in 707 cases with unexplained bleeding and platelet disorders. Genome Medicine, 2015, 7, 36.	8.2	119
79	Single Nucleotide Variants in the Protein C Pathway and Mortality in Dialysis Patients. PLoS ONE, 2014, 9, e97251.	2.5	6
80	The INTERVAL trial to determine whether intervals between blood donations can be safely and acceptably decreased to optimise blood supply: study protocol for a randomised controlled trial. Trials, 2014, 15, 363.	1.6	112
81	Common genetic variants do not associate with CAD in familial hypercholesterolemia. European Journal of Human Genetics, 2014, 22, 809-813.	2.8	2
82	DNA methylation and body-mass index: a genome-wide analysis. Lancet, The, 2014, 383, 1990-1998.	13.7	686
83	The Human Phenotype Ontology project: linking molecular biology and disease through phenotype data. Nucleic Acids Research, 2014, 42, D966-D974.	14.5	698
84	Novel loci affecting iron homeostasis and their effects in individuals at risk for hemochromatosis. Nature Communications, 2014, 5, 4926.	12.8	192
85	Transcriptional diversity during lineage commitment of human blood progenitors. Science, 2014, 345, 1251033.	12.6	253
86	Defining the role of common variation in the genomic and biological architecture of adult human height. Nature Genetics, 2014, 46, 1173-1186.	21.4	1,818
87	Gray platelet syndrome: proinflammatory megakaryocytes and Î±-granule loss cause myelofibrosis and confer metastasis resistance in mice. Blood, 2014, 124, 3624-3635.	1.4	79
88	Large-scale association analysis identifies new risk loci for coronary artery disease. Nature Genetics, 2013, 45, 25-33.	21.4	1,439
89	Platelet Genomics. , 2013, , 67-89.		2
90	Automatic Event Detection within Thrombus Formation Based on Integer Programming. Lecture Notes in Computer Science, 2013, , 215-224.	1.3	0

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91	Bayesian refinement of association signals for 14 loci in 3 common diseases. Nature Genetics, 2012, 44, 1294-1301.	21.4	469
92	Seventy-five genetic loci influencing the human red blood cell. Nature, 2012, 492, 369-375.	27.8	320
93	Large-Scale Gene-Centric Meta-analysis across 32 Studies Identifies Multiple Lipid Loci. American Journal of Human Genetics, 2012, 91, 823-838.	6.2	227
94	Compound inheritance of a low-frequency regulatory SNP and a rare null mutation in exon-junction complex subunit RBM8A causes TAR syndrome. Nature Genetics, 2012, 44, 435-439.	21.4	355
95	Image-based characterization of thrombus formation in time-lapse DIC microscopy. Medical Image Analysis, 2012, 16, 915-931.	11.6	6
96	Exome sequencing identifies NBEAL2 as the causative gene for gray platelet syndrome. Nature Genetics, 2011, 43, 735-737.	21.4	245
97	New gene functions in megakaryopoiesis and platelet formation. Nature, 2011, 480, 201-208.	27.8	401
98	Genome-wide Analysis of Simultaneous GATA1/2, RUNX1, FLI1, and SCL Binding in Megakaryocytes Identifies Hematopoietic Regulators. Developmental Cell, 2011, 20, 597-609.	7.0	255
99	Silencing of RhoA nucleotide exchange factor, ARHGEF3, reveals its unexpected role in iron uptake. Blood, 2011, 118, 4967-4976.	1.4	34
100	Joint Thrombus and Vessel Segmentation Using Dynamic Texture Likelihoods and Shape Prior. Lecture Notes in Computer Science, 2011, 14, 579-586.	1.3	2
101	Transcription profiling in human platelets reveals LRRFIP1 as a novel protein regulating platelet function. Blood, 2010, 116, 4646-4656.	1.4	90
102	Cell-specific protein phenotypes for the autoimmune locus IL2RA using a genotype-selectable human bioresource. Nature Genetics, 2009, 41, 1011-1015.	21.4	249
103	A novel variant on chromosome 7q22.3 associated with mean platelet volume, counts, and function. Blood, 2009, 113, 3831-3837.	1.4	117
104	Functional genomics in zebrafish permits rapid characterization of novel platelet membrane proteins. Blood, 2009, 113, 4754-4762.	1.4	69
105	Identification of variation in the platelet transcriptome associated withGlycoprotein 6haplotype. Platelets, 2008, 19, 258-267.	2.3	9
106	A nonsynonymous SNP in the ITGB3 gene disrupts the conserved membrane-proximal cytoplasmic salt bridge in the Î±IIbÎ²3 integrin and cosegregates dominantly with abnormal proplatelet formation and macrothrombocytopenia. Blood, 2008, 111, 3407-3414.	1.4	94
107	Genomewide Association Analysis of Coronary Artery Disease. New England Journal of Medicine, 2007, 357, 443-453.	27.0	1,865
108	Comparative gene expression profiling of in vitro differentiated megakaryocytes and erythroblasts identifies novel activatory and inhibitory platelet membrane proteins. Blood, 2007, 109, 3260-3269.	1.4	153

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109	Microarray-based genotyping for blood groups: comparison of gene array and 5â€²-nuclease assay techniques with human platelet antigen as a model. Transfusion, 2005, 45, 654-659.	1.6	64
110	Platelet genomics and proteomics in human health and disease. Journal of Clinical Investigation, 2005, 115, 3370-3377.	8.2	146
111	The low-frequency allele of the platelet collagen signaling receptor glycoprotein VI is associated with reduced functional responses and expression. Blood, 2003, 101, 4372-4379.	1.4	124
112	Introduction to Antibody Engineering and Phage Display. Vox Sanguinis, 2000, 78, 72-79.	1.5	45
113	Threonine-145/Methionine-145 variants of baculovirus produced recombinant ligand binding domain of GPIbÎ± express HPA-2 epitopes and show equal binding of von Willebrand factor. Blood, 2000, 95, 205-211.	1.4	34
114	Introduction to Antibody Engineering and Phage Display. Vox Sanguinis, 2000, 78, 72-79.	1.5	24
115	The Natural History of Fetomaternal Alloimmunization to the Platelet-Specific Antigen HPA-1a (PlA1,) Tj ETQq1 1 0.784314 rgBT /Ove	1.4	10
116	Activation during preparation of therapeutic platelets affects deterioration during storage: a comparative flow cytometric study of different production methods. British Journal of Haematology, 1997, 98, 86-95.	2.5	157
117	Characterization of human variable domain antibody fragments against the U1 RNA-associated A protein, selected from a synthetic and a patient-derived combinatorial V gene library. European Journal of Immunology, 1996, 26, 629-639.	2.9	46
118	Human Antibody Fragments Specific for Human Blood Group Antigens from a Phage Display Library. Nature Biotechnology, 1993, 11, 1145-1149.	17.5	172