

Richard van Wijk

List of Publications by Year in descending order

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Version: 2024-02-01

78
papers

1,958
citations

304368

22
h-index

276539

41
g-index

78
all docs

78
docs citations

78
times ranked

2862
citing authors

#	ARTICLE	IF	CITATIONS
1	The energy-less red blood cell is lost: erythrocyte enzyme abnormalities of glycolysis. <i>Blood</i> , 2005, 106, 4034-4042.	0.6	256
2	Squeezing for Life – Properties of Red Blood Cell Deformability. <i>Frontiers in Physiology</i> , 2018, 9, 656.	1.3	213
3	A novel CBFA2 single-nucleotide mutation in familial platelet disorder with propensity to develop myeloid malignancies. <i>Blood</i> , 2001, 98, 2856-2858.	0.6	127
4	Red Blood Cells: Chasing Interactions. <i>Frontiers in Physiology</i> , 2019, 10, 945.	1.3	92
5	The fluid membrane determines mechanics of erythrocyte extracellular vesicles and is softened in hereditary spherocytosis. <i>Nature Communications</i> , 2018, 9, 4960.	5.8	79
6	Identification of a new VHL exon and complex splicing alterations in familial erythrocytosis or von Hippel-Lindau disease. <i>Blood</i> , 2018, 132, 469-483.	0.6	70
7	Ribosomal Protein Mutations Induce Autophagy through S6 Kinase Inhibition of the Insulin Pathway. <i>PLoS Genetics</i> , 2014, 10, e1004371.	1.5	58
8	Recommendations for diagnosis and treatment of methemoglobinemia. <i>American Journal of Hematology</i> , 2021, 96, 1666-1678.	2.0	56
9	Addressing the diagnostic gaps in pyruvate kinase deficiency: Consensus recommendations on the diagnosis of pyruvate kinase deficiency. <i>American Journal of Hematology</i> , 2019, 94, 149-161.	2.0	55
10	Disruption of a novel regulatory element in the erythroid-specific promoter of the human PKLR gene causes severe pyruvate kinase deficiency. <i>Blood</i> , 2003, 101, 1596-1602.	0.6	50
11	Rapid and reproducible characterization of sickling during automated deoxygenation in sickle cell disease patients. <i>American Journal of Hematology</i> , 2019, 94, 575-584.	2.0	47
12	Genotype–phenotype correlation and molecular heterogeneity in pyruvate kinase deficiency. <i>American Journal of Hematology</i> , 2020, 95, 472-482.	2.0	47
13	AG-348 (Mitapivat), an allosteric activator of red blood cell pyruvate kinase, increases enzymatic activity, protein stability, and ATP levels over a broad range of PKLR genotypes. <i>Haematologica</i> , 2020, 106, 238-249.	1.7	45
14	The Complexity of Genotype–Phenotype Correlations in Hereditary Spherocytosis: A Cohort of 95 Patients. <i>HemaSphere</i> , 2019, 3, e276.	1.2	43
15	Worldwide study of hematopoietic allogeneic stem cell transplantation in pyruvate kinase deficiency. <i>Haematologica</i> , 2018, 103, e82-e86.	1.7	42
16	Erythrocytosis associated with a novel missense mutation in the HIF2A gene. <i>Haematologica</i> , 2010, 95, 829-832.	1.7	35
17	Fifteen novel mutations in <i>PKLR</i> associated with pyruvate kinase (PK) deficiency: Structural implications of amino acid substitutions in PK. <i>Human Mutation</i> , 2009, 30, 446-453.	1.1	33
18	The EPO-FGF23 Signaling Pathway in Erythroid Progenitor Cells: Opening a New Area of Research. <i>Frontiers in Physiology</i> , 2019, 10, 304.	1.3	33

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19	Severe Ankyrin-R deficiency results in impaired surface retention and lysosomal degradation of RhAG in human erythroblasts. <i>Haematologica</i> , 2016, 101, 1018-1027.	1.7	31
20	HK Utrecht: missense mutation in the active site of human hexokinase associated with hexokinase deficiency and severe nonspherocytic hemolytic anemia. <i>Blood</i> , 2003, 101, 345-347.	0.6	30
21	The variable manifestations of disease in pyruvate kinase deficiency and their management. <i>Haematologica</i> , 2020, 105, 2229-2239.	1.7	30
22	Density, heterogeneity and deformability of red cells as markers of clinical severity in hereditary spherocytosis. <i>Haematologica</i> , 2020, 105, 338-347.	1.7	27
23	Red blood cell phenotyping from 3D confocal images using artificial neural networks. <i>PLoS Computational Biology</i> , 2021, 17, e1008934.	1.5	26
24	Analysis of a cohort of 101 CD41 patients: description of 24 new molecular variants and genotype-phenotype correlations. <i>British Journal of Haematology</i> , 2016, 175, 696-704.	1.2	25
25	PIEZO1 gain-of-function mutations delay reticulocyte maturation in hereditary xerocytosis. <i>Haematologica</i> , 2020, 105, e268-e271.	1.7	24
26	Hereditary Xerocytosis due to Mutations in PIEZO1 Gene Associated with Heterozygous Pyruvate Kinase Deficiency and Beta-Thalassemia Trait in Two Unrelated Families. <i>Case Reports in Hematology</i> , 2017, 2017, 1-8.	0.3	22
27	Decreased activity and stability of pyruvate kinase in sickle cell disease: a novel target for mitapivat therapy. <i>Blood</i> , 2021, 137, 2997-3001.	0.6	22
28	Ex vivo analysis of aberrant splicing induced by two donor site mutations in PKLR of a patient with severe pyruvate kinase deficiency. <i>British Journal of Haematology</i> , 2004, 125, 253-263.	1.2	21
29	Oxygen gradient ektacytometry-derived biomarkers are associated with vaso-occlusive crises and correlate with treatment response in sickle cell disease. <i>American Journal of Hematology</i> , 2021, 96, E29-E32.	2.0	21
30	Safety and efficacy of mitapivat, an oral pyruvate kinase activator, in sickle cell disease: A phase 2, open-label study. <i>American Journal of Hematology</i> , 2022, 97, .	2.0	21
31	De novo variants in HK1 associated with neurodevelopmental abnormalities and visual impairment. <i>European Journal of Human Genetics</i> , 2019, 27, 1081-1089.	1.4	19
32	Methodological aspects of the oxygen scan in sickle cell disease: A need for standardization. <i>American Journal of Hematology</i> , 2020, 95, E5-E8.	2.0	18
33	Oxidative stress in sickle cell disease; more than a DAMP squib. <i>Clinical Hemorheology and Microcirculation</i> , 2018, 68, 239-250.	0.9	17
34	Loss-of-function zinc finger mutation in the EGLN1 gene associated with erythrocytosis. <i>Blood</i> , 2018, 132, 1455-1458.	0.6	15
35	Untargeted metabolic profiling in dried blood spots identifies disease fingerprint for pyruvate kinase deficiency. <i>Haematologica</i> , 2021, 106, 2720-2725.	1.7	14
36	Interplay of erythropoietin, fibroblast growth factor 23, and erythroferrone in patients with hereditary hemolytic anemia. <i>Blood Advances</i> , 2020, 4, 1678-1682.	2.5	13

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37	Mechanical Stress Induces Ca ²⁺ -Dependent Signal Transduction in Erythroblasts and Modulates Erythropoiesis. <i>International Journal of Molecular Sciences</i> , 2021, 22, 955.	1.8	13
38	Distinct phenotypic expression of two de novo missense mutations affecting the dimer interface of glucose-6-phosphate dehydrogenase. <i>Blood Cells, Molecules, and Diseases</i> , 2004, 32, 112-117.	0.6	12
39	Effects of Genotypes and Treatment on Oxygenation Parameters in Sickle Cell Disease. <i>Cells</i> , 2021, 10, 811.	1.8	10
40	Methodological aspects of oxygen gradient ektacytometry in sickle cell disease: Effects of sample storage on outcome parameters in distinct patient subgroups. <i>Clinical Hemorheology and Microcirculation</i> , 2021, 77, 391-394.	0.9	10
41	Molecular characterization of six new cases of red blood cell hexokinase deficiency yields four novel mutations in HK1. <i>Blood Cells, Molecules, and Diseases</i> , 2016, 59, 71-76.	0.6	9
42	Characterization of Sickling During Controlled Automated Deoxygenation with Oxygen Gradient Ektacytometry. <i>Journal of Visualized Experiments</i> , 2019, , .	0.2	9
43	GATA-1 Defects in Diamond-Blackfan Anemia: Phenotypic Characterization Points to a Specific Subset of Disease. <i>Genes</i> , 2022, 13, 447.	1.0	9
44	Novel Homozygous Mutation of the Internal Translation Initiation Start Site of <i>VHL</i> is Exclusively Associated with Erythrocytosis: Indications for Distinct Functional Roles of von Hippel-Lindau Tumor Suppressor Isoforms. <i>Human Mutation</i> , 2015, 36, 1039-1042.	1.1	8
45	A Previously Unrecognized Ca ²⁺ -Inhibited Nonselective Cation Channel in Red Blood Cells. <i>HemaSphere</i> , 2018, 2, e146.	1.2	8
46	Iron overload in patients with rare hereditary hemolytic anemia: Evidence-based suggestion on whom and how to screen. <i>American Journal of Hematology</i> , 2018, 93, E374-E376.	2.0	8
47	Red Blood Cell Membrane Conductance in Hereditary Haemolytic Anaemias. <i>Frontiers in Physiology</i> , 2019, 10, 386.	1.3	8
48	A Proposed Concept for Defective Mitophagy Leading to Late Stage Ineffective Erythropoiesis in Pyruvate Kinase Deficiency. <i>Frontiers in Physiology</i> , 2020, 11, 609103.	1.3	7
49	A Novel Mutation of Glucose Phosphate Isomerase (GPI) Causing Severe Neonatal Anemia Due to GPI Deficiency. <i>Journal of Pediatric Hematology/Oncology</i> , 2019, 41, e186-e189.	0.3	6
50	Impaired Cytoskeletal and Membrane Biophysical Properties of Acanthocytes in Hypobetalipoproteinemia – A Case Study. <i>Frontiers in Physiology</i> , 2021, 12, 638027.	1.3	6
51	Pyruvate kinase deficiency and severe congenital hemolytic anemia in a double heterozygous patient with paternal transmission of an early germ-line <i>de novo</i> mutation. <i>American Journal of Hematology</i> , 2015, 90, E217-9.	2.0	5
52	The Interplay between Drivers of Erythropoiesis and Iron Homeostasis in Rare Hereditary Anemias: Tipping the Balance. <i>International Journal of Molecular Sciences</i> , 2021, 22, 2204.	1.8	5
53	Binding of Erythrocyte ICAM-4 to the Platelet Activated Integrin α IIb β 3 leads to a Direct Erythrocyte-Platelet Adhesion Under Venous Flow Shear Rate. <i>Blood</i> , 2012, 120, 105-105.	0.6	5
54	Proton pump inhibition for secondary hemochromatosis in hereditary anemia: a phase III placebo-controlled randomized cross-over clinical trial. <i>American Journal of Hematology</i> , 2022, 97, 924-932.	2.0	5

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55	Proteomics reveals reduced expression of transketolase in pyrimidine 5-aminonucleotidase deficient patients. <i>Proteomics - Clinical Applications</i> , 2016, 10, 859-869.	0.8	4
56	Dried blood spot metabolomics reveals a metabolic fingerprint with diagnostic potential for Diamond Blackfan Anaemia. <i>British Journal of Haematology</i> , 2021, 193, 1185-1193.	1.2	4
57	Safety and Efficacy of Mitapivat (AG-348), an Oral Activator of Pyruvate Kinase R, in Subjects with Sickle Cell Disease: A Phase 2, Open-Label Study (ESTIMATE). <i>Blood</i> , 2021, 138, 2047-2047.	0.6	4
58	Congenital Hemolytic Anemia Because of Glucose Phosphate Isomerase Deficiency: Identification of 2 Novel Missense Mutations in the GPI Gene. <i>Journal of Pediatric Hematology/Oncology</i> , 2020, 42, e696-e697.	0.3	3
59	Comparisons of oxygen gradient ektacytometry parameters between sickle cell patients with or without β -thalassaemia. <i>British Journal of Haematology</i> , 2021, 195, 629-633.	1.2	3
60	Organ involvement occurs in all forms of hereditary haemolytic anaemia. <i>British Journal of Haematology</i> , 2019, 185, 602-605.	1.2	2
61	Rapid diagnosis of hereditary haemolytic anaemias using automated rheoscopy and supervised machine learning. <i>British Journal of Haematology</i> , 2020, 190, e250-e255.	1.2	2
62	Liver Iron Retention Estimated from Utilization of Oral and Intravenous Radioiron in Various Anemias and Hemochromatosis in Humans. <i>International Journal of Molecular Sciences</i> , 2020, 21, 1077.	1.8	2
63	Metabolic Fingerprint in Hereditary Spherocytosis Correlates With Red Blood Cell Characteristics and Clinical Severity. <i>HemaSphere</i> , 2021, 5, e591.	1.2	2
64	Facilitating EMA binding test performance using fluorescent beads combined with next-generation sequencing. <i>EJHaem</i> , 2021, 2, 716-728.	0.4	2
65	Identification of Biomarkers That Are Associated with Clinical Complications of Hemoglobin SC Disease and Sickle Cell Anemia. <i>Blood</i> , 2021, 138, 962-962.	0.6	2
66	Editorial: Pathophysiology of Rare Hemolytic Anemias. <i>Frontiers in Physiology</i> , 2020, 11, 601746.	1.3	1
67	Rare Anemias: Are Their Names Just Smoke and Mirrors?. <i>Frontiers in Physiology</i> , 2021, 12, 690604.	1.3	1
68	The Oxygenscan: A Rapid and Reproducible Test to Determine Patient-Specific, Clinically Relevant Biomarkers of Disease Severity in Sickle Cell Anemia. <i>Blood</i> , 2018, 132, 2360-2360.	0.6	1
69	Tibetan Enriched PKLR Variant Is Beneficial to High Altitude Adaption By Improving Oxygen Delivery. <i>Blood</i> , 2018, 132, 1027-1027.	0.6	1
70	A Comprehensive Analysis of the Erythropoietin-erythroferrone-hepcidin Pathway in Hereditary Hemolytic Anemias. <i>HemaSphere</i> , 2021, 5, e627.	1.2	1
71	Summary of Joint European Hematology Association (EHA) and EuroBloodNet Recommendations on Diagnosis and Treatment of Methemoglobinemia. <i>HemaSphere</i> , 2021, 5, e660.	1.2	1
72	Infantile Pyknocytosis in a Premature Dichorionic Diamniotic Twin. <i>Journal of Pediatric Hematology/Oncology</i> , 2020, Publish Ahead of Print, e1037-e1039.	0.3	1

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73	Untargeted Metabolomic Fingerprinting As a Potential Tool in the Diagnostic Evaluation of Diamond Blackfan Anemia. <i>Blood</i> , 2020, 136, 7-8.	0.6	1
74	Editorial: New Methods for Red Blood Cell Research and Diagnosis. <i>Frontiers in Physiology</i> , 2021, 12, 755664.	1.3	0
75	GATA-1 Binding to Exon 1 Directs High Level Erythroid-Specific Expression of the Human Protoporphyrinogen Oxidase Gene.. <i>Blood</i> , 2005, 106, 1682-1682.	0.6	0
76	The Oxygenscan Provides Clinically Relevant Biomarkers for Treatment Efficacy That Are Associated with Frequency of Vaso-Occlusive Crisis in Sickle Cell Disease. <i>Blood</i> , 2019, 134, 2275-2275.	0.6	0
77	Red Cell Rheology Biomarkers to Assess Cure in Gene-Based Therapies. <i>Blood</i> , 2020, 136, 11-12.	0.6	0
78	Comment on: Oxygen gradient ektacytometry does not predict pain in children with sickle cell anaemia. <i>British Journal of Haematology</i> , 2022, , .	1.2	0