

Roberta Russo

List of Publications by Year in descending order

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Version: 2024-02-01

109
papers

4,937
citations

101384

36
h-index

102304

66
g-index

117
all docs

117
docs citations

117
times ranked

8192
citing authors

#	ARTICLE	IF	CITATIONS
1	Genetic mechanisms of critical illness in COVID-19. <i>Nature</i> , 2021, 591, 92-98.	13.7	1,014
2	Multiple clinical forms of dehydrated hereditary stomatocytosis arise from mutations in PIEZO1. <i>Blood</i> , 2013, 121, 3925-3935.	0.6	266
3	Mutations affecting the secretory COPII coat component SEC23B cause congenital dyserythropoietic anemia type II. <i>Nature Genetics</i> , 2009, 41, 936-940.	9.4	250
4	ACE2 gene variants may underlie interindividual variability and susceptibility to COVID-19 in the Italian population. <i>European Journal of Human Genetics</i> , 2020, 28, 1602-1614.	1.4	208
5	New insights on hereditary erythrocyte membrane defects. <i>Haematologica</i> , 2016, 101, 1284-1294.	1.7	156
6	Suppressor of cytokine signaling 3 (SOCS3) expression and hepatitis C virus-related chronic hepatitis: Insulin resistance and response to antiviral therapy. <i>Hepatology</i> , 2007, 46, 1009-1015.	3.6	150
7	Association of Toll-like receptor 7 variants with life-threatening COVID-19 disease in males: findings from a nested case-control study. <i>ELife</i> , 2021, 10, .	2.8	145
8	Recommendations regarding splenectomy in hereditary hemolytic anemias. <i>Haematologica</i> , 2017, 102, 1304-1313.	1.7	138
9	Multi-gene panel testing improves diagnosis and management of patients with hereditary anemias. <i>American Journal of Hematology</i> , 2018, 93, 672-682.	2.0	117
10	Dysregulation of lipid metabolism and pathological inflammation in patients with COVID-19. <i>Scientific Reports</i> , 2021, 11, 2941.	1.6	102
11	Replication of GWAS-identified neuroblastoma risk loci strengthens the role of BARD1 and affirms the cumulative effect of genetic variations on disease susceptibility. <i>Carcinogenesis</i> , 2013, 34, 605-611.	1.3	95
12	Elevated expression and polymorphisms of SOCS3 influence patient response to antiviral therapy in chronic hepatitis C. <i>Gut</i> , 2007, 57, 507-515.	6.1	84
13	Regulation of divalent metal transporter 1 (DMT1) non-IRE isoform by the microRNA Let-7d in erythroid cells. <i>Haematologica</i> , 2010, 95, 1244-1252.	1.7	82
14	Novel Gardos channel mutations linked to dehydrated hereditary stomatocytosis (xerocytosis). <i>American Journal of Hematology</i> , 2015, 90, 921-926.	2.0	81
15	Diagnosis and management of congenital dyserythropoietic anemias. <i>Expert Review of Hematology</i> , 2016, 9, 283-296.	1.0	76
16	Galectin-1 and Its Involvement in Hepatocellular Carcinoma Aggressiveness. <i>Molecular Medicine</i> , 2010, 16, 102-115.	1.9	69
17	Clinical aspects and pathogenesis of congenital dyserythropoietic anemias: from morphology to molecular approach. <i>Haematologica</i> , 2012, 97, 1786-1794.	1.7	68
18	Retrospective cohort study of 205 cases with congenital dyserythropoietic anemia type II: Definition of clinical and molecular spectrum and identification of new diagnostic scores. <i>American Journal of Hematology</i> , 2014, 89, E169-75.	2.0	68

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19	Missense mutations in the ABCB6 transporter cause dominant familialpseudohyperkalemia. American Journal of Hematology, 2013, 88, 66-72.	2.0	67
20	The European Hematology Association Roadmap for European Hematology Research: a consensus document. Haematologica, 2016, 101, 115-208.	1.7	67
21	Advances in understanding the pathogenesis of red cell membrane disorders. British Journal of Haematology, 2019, 187, 13-24.	1.2	64
22	Congenital dyserythropoietic anemias. Blood, 2020, 136, 1274-1283.	0.6	62
23	Common variants at 21q22.3 locus influence MX1 and TMPRSS2 gene expression and susceptibility to severe COVID-19. IScience, 2021, 24, 102322.	1.9	60
24	Molecular analysis of 42 patients with congenital dyserythropoietic anemia type II: new mutations in the SEC23B gene and a search for a genotype-phenotype relationship. Haematologica, 2010, 95, 708-715.	1.7	56
25	The Serum Metabolome of Moderate and Severe COVID-19 Patients Reflects Possible Liver Alterations Involving Carbon and Nitrogen Metabolism. International Journal of Molecular Sciences, 2021, 22, 9548.	1.8	56
26	Recommendations for diagnosis and treatment of methemoglobinemia. American Journal of Hematology, 2021, 96, 1666-1678.	2.0	56
27	Hereditary stomatocytosis: An underdiagnosed condition. American Journal of Hematology, 2018, 93, 107-121.	2.0	54
28	Shorter androgen receptor polyQ alleles protect against life-threatening COVID-19 disease in European males. EBioMedicine, 2021, 65, 103246.	2.7	52
29	Label-Free Optical Marker for Red-Blood-Cell Phenotyping of Inherited Anemias. Analytical Chemistry, 2018, 90, 7495-7501.	3.2	49
30	Genotype-phenotype correlation and risk stratification in a cohort of 123 hereditary stomatocytosis patients. American Journal of Hematology, 2018, 93, 1509-1517.	2.0	48
31	Congenital dyserythropoietic anemias. Current Opinion in Hematology, 2011, 18, 146-151.	1.2	44
32	Gain-of-function mutations in PIEZO1 directly impair hepatic iron metabolism via the inhibition of the BMP/SMADs pathway. American Journal of Hematology, 2020, 95, 188-197.	2.0	44
33	Rare variants in Toll-like receptor 7 results in functional impairment and downregulation of cytokine-mediated signaling in COVID-19 patients. Genes and Immunity, 2022, 23, 51-56.	2.2	41
34	Mutational spectrum in congenital dyserythropoietic anemia type II: Identification of 19 novel variants in <i>SEC23B</i> gene. American Journal of Hematology, 2010, 85, 915-920.	2.0	40
35	Genetic Analysis of the Coronavirus SARS-CoV-2 Host Protease TMPRSS2 in Different Populations. Frontiers in Genetics, 2020, 11, 872.	1.1	40
36	Congenital erythropoietic porphyria linked to <i>GATA1</i> <i>R216W</i> mutation: challenges for diagnosis. European Journal of Haematology, 2015, 94, 491-497.	1.1	39

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37	Protective Role of a Tmprss2 Variant on Severe COVID-19 Outcome in Young Males and Elderly Women. <i>Genes</i> , 2021, 12, 596.	1.0	39
38	The pyruvate kinase activator mitapivat reduces hemolysis and improves anemia in a β^2 -thalassemia mouse model. <i>Journal of Clinical Investigation</i> , 2021, 131, .	3.9	39
39	A predicted functional single-nucleotide polymorphism of bone morphogenetic protein-4 gene affects mRNA expression and shows a significant association with cutaneous melanoma in Southern Italian population. <i>Journal of Cancer Research and Clinical Oncology</i> , 2009, 135, 1799-1807.	1.2	36
40	Employing a systematic approach to biobanking and analyzing clinical and genetic data for advancing COVID-19 research. <i>European Journal of Human Genetics</i> , 2021, 29, 745-759.	1.4	35
41	Hypomorphic mutations of SEC23B gene account for mild phenotypes of congenital dyserythropoietic anemia type II. <i>Blood Cells, Molecules, and Diseases</i> , 2013, 51, 17-21.	0.6	33
42	Comparative Proteomic Expression Profile in All-transRetinoic Acid Differentiated Neuroblastoma Cell Line. <i>Journal of Proteome Research</i> , 2007, 6, 2550-2564.	1.8	30
43	Functional characterization of novel ABCB6 mutations and their clinical implications in familial pseudohyperkalemia. <i>Haematologica</i> , 2016, 101, 909-917.	1.7	30
44	Two founder mutations in the <i>SEC23B</i> gene account for the relatively high frequency of CDA II in the Italian population. <i>American Journal of Hematology</i> , 2011, 86, 727-732.	2.0	27
45	Genetics and Genomics Approaches for Diagnosis and Research Into Hereditary Anemias. <i>Frontiers in Physiology</i> , 2020, 11, 613559.	1.3	27
46	Increased levels of ERFE-encoding FAM132B in patients with congenital dyserythropoietic anemia type II. <i>Blood</i> , 2016, 128, 1899-1902.	0.6	26
47	PIEZO1 Hypomorphic Variants in Congenital Lymphatic Dysplasia Cause Shape and Hydration Alterations of Red Blood Cells. <i>Frontiers in Physiology</i> , 2019, 10, 258.	1.3	26
48	Successful hematopoietic stem cell transplantation in a patient with congenital dyserythropoietic anemia type II. <i>Pediatric Transplantation</i> , 2014, 18, E130-3.	0.5	25
49	SOCS3 and IRS-1 gene expression differs between genotype 1 and genotype 2 hepatitis C virus-infected HepG2 cells. <i>Clinical Chemistry and Laboratory Medicine</i> , 2009, 47, 1217-25.	1.4	22
50	Complex Modes of Inheritance in Hereditary Red Blood Cell Disorders: A Case Series Study of 155 Patients. <i>Genes</i> , 2021, 12, 958.	1.0	22
51	Common, low-frequency, rare, and ultra-rare coding variants contribute to COVID-19 severity. <i>Human Genetics</i> , 2022, 141, 147-173.	1.8	22
52	The BMP-SMAD pathway mediates the impaired hepatic iron metabolism associated with the ERFE A260S variant. <i>American Journal of Hematology</i> , 2019, 94, 1227-1235.	2.0	21
53	Pediatric pharmacogenetic and pharmacogenomic studies: the current state and future perspectives. <i>European Journal of Clinical Pharmacology</i> , 2011, 67, 17-27.	0.8	19
54	Differential diagnosis of hereditary anemias from a fraction of blood drop by digital holography and hierarchical machine learning. <i>Biosensors and Bioelectronics</i> , 2022, 201, 113945.	5.3	19

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55	Red cells in post-genomic era: impact of personalized medicine in the treatment of anemias. <i>Haematologica</i> , 2015, 100, 3-6.	1.7	18
56	PIEZO1-R1864H rare variant accounts for a genetic phenotype-modifier role in dehydrated hereditary stomatocytosis. <i>Haematologica</i> , 2018, 103, e94-e97.	1.7	18
57	Kinome expression profiling of human neuroblastoma tumors identifies potential drug targets for ultra high-risk patients. <i>Carcinogenesis</i> , 2017, 38, 1011-1020.	1.3	17
58	Inherited hematological disorders due to defects in coat protein (COP)II complex. <i>American Journal of Hematology</i> , 2013, 88, 135-140.	2.0	16
59	GATA1 erythroid-specific regulation of SEC23B expression and its implication in the pathogenesis of congenital dyserythropoietic anemia type II. <i>Haematologica</i> , 2017, 102, e371-e374.	1.7	16
60	Characterization of Two Cases of Congenital Dyserythropoietic Anemia Type I Shed Light on the Uncharacterized C15orf41 Protein. <i>Frontiers in Physiology</i> , 2019, 10, 621.	1.3	16
61	Regulatory Noncoding and Predicted Pathogenic Coding Variants of CCR5 Predispose to Severe COVID-19. <i>International Journal of Molecular Sciences</i> , 2021, 22, 5372.	1.8	16
62	Integration of Pharmacogenetics and Pharmacogenomics in Drug Development: Implications for Regulatory and Medical Decision Making in Pediatric Diseases. <i>Journal of Clinical Pharmacology</i> , 2012, 52, 704-716.	1.0	15
63	MDM2 SNP309 and p53 Arg72Pro in cutaneous melanoma: association between SNP309 GG genotype and tumor Breslow thickness. <i>Journal of Human Genetics</i> , 2010, 55, 518-524.	1.1	14
64	Germline mutations and new copy number variants among 40 pediatric cancer patients suspected for genetic predisposition. <i>Clinical Genetics</i> , 2019, 96, 359-365.	1.0	14
65	Inherited microcytic anemias. <i>Hematology American Society of Hematology Education Program</i> , 2020, 2020, 465-470.	0.9	12
66	The TNFRSF13C H159Y Variant Is Associated with Severe COVID-19: A Retrospective Study of 500 Patients from Southern Italy. <i>Genes</i> , 2021, 12, 881.	1.0	12
67	Protease inhibitors-based therapy induces acquired spherocytic-like anaemia and ineffective erythropoiesis in chronic hepatitis C virus patients. <i>Liver International</i> , 2016, 36, 49-58.	1.9	11
68	RAP-011 Rescues the Disease Phenotype in a Cellular Model of Congenital Dyserythropoietic Anemia Type II by Inhibiting the SMAD2-3 Pathway. <i>International Journal of Molecular Sciences</i> , 2020, 21, 5577.	1.8	9
69	Targeted next generation sequencing identifies a novel β -spectrin gene mutation A2059P in two Omani children with hereditary pyropoikilocytosis. <i>American Journal of Hematology</i> , 2017, 92, E607-E609.	2.0	8
70	A novel PIEZO1 mutation in a patient with dehydrated hereditary stomatocytosis: a case report and a brief review of literature. <i>Italian Journal of Pediatrics</i> , 2020, 46, 102.	1.0	8
71	Recommendations for Pregnancy in Rare Inherited Anemias. <i>HemaSphere</i> , 2020, 4, e446.	1.2	8
72	Uridine treatment normalizes the congenital dyserythropoietic anemia type II-like hematological phenotype in a patient with homozygous mutation in the <i>CAD</i> gene. <i>American Journal of Hematology</i> , 2020, 95, 1423-1426.	2.0	8

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73	The EHA Research Roadmap: Anemias. <i>HemaSphere</i> , 2021, 5, e607.	1.2	7
74	Hematopoietic Stem Cell Transplantation in Congenital Dyserythropetic Anemia Type II: A Case Report and Review of the Literature. <i>Journal of Pediatric Hematology/Oncology</i> , 2020, 42, e507-e510.	0.3	7
75	Germline rare variants of lectin pathway genes predispose to asymptomatic SARS-CoV-2 infection in elderly individuals. <i>Genetics in Medicine</i> , 2022, , .	1.1	7
76	Next generation research and therapy in red blood cell diseases. <i>Haematologica</i> , 2016, 101, 515-517.	1.7	6
77	SEC23B Loss-of-Function Suppresses Hepcidin Expression by Impairing Glycosylation Pathway in Human Hepatic Cells. <i>International Journal of Molecular Sciences</i> , 2022, 23, 1304.	1.8	6
78	The Use of Next-generation Sequencing in the Diagnosis of Rare Inherited Anaemias: A Joint BSH/EHA Good Practice Paper. <i>HemaSphere</i> , 2022, 6, e739.	1.2	6
79	CoDysAn: A Telemedicine Tool to Improve Awareness and Diagnosis for Patients With Congenital Dyserythropoietic Anemia. <i>Frontiers in Physiology</i> , 2019, 10, 1063.	1.3	4
80	Apparent recessive inheritance of sideroblastic anemia type 2 due to uniparental isodisomy at the SLC25A38 locus. <i>Haematologica</i> , 2020, 105, 2883-2886.	1.7	4
81	A Dyserythropoietic Anemia Associated with Homozygous Hb Plasencia [$\pm 125(\text{H8})\text{Leu}\hat{+}\text{Arg} (\hat{\pm}2)$] (HBA2:c.377T>G), A Variant with an Unstable $\hat{\pm}$ Chain. <i>Hemoglobin</i> , 2010, 34, 576-581.	0.4	3
82	The use of <sc>nextâ€ generation</sc> sequencing in the diagnosis of rare inherited anaemias: A Joint BSH/EHA Good Practice Paper*. <i>British Journal of Haematology</i> , 2022, 198, 459-477.	1.2	3
83	Hereditary spherocytosis and allied disorders. <i>HemaSphere</i> , 2019, 3, 157-159.	1.2	2
84	Recommendations for pregnancy in Fanconi anemia. <i>Expert Opinion on Biological Therapy</i> , 2021, 21, 1-7.	1.4	2
85	A novel GLA mutation in a Fabry family with glucose-6-phosphate dehydrogenase deficiency. <i>Journal of Nephrology</i> , 2012, 25, 582-585.	0.9	2
86	Congenital dyserythropoietic anaemias: new acquisitions. <i>Blood Transfusion</i> , 2011, 9, 278-80.	0.3	2
87	<i>PIEZO1</i> Mutation May Determine Early Onset of Clinical Manifestation of Anemia of Myelodysplastic Syndromes. <i>Blood</i> , 2021, 138, 1528-1528.	0.6	2
88	Hereditary anemia caused by multilocus inheritance of <i>PIEZO1</i>, <i>SLC4A1</i> and <i>ABCB6</i> mutations: a diagnostic and therapeutic challenge. <i>Haematologica</i> , 2022, 107, 2280-2284.	1.7	2
89	Acquired spherocytic like anemia combined with ineffective erythropoiesis sustains anemia in patients with chronic hepatitis C infection receiving telaprevir or boceprevir-based triple therapy. <i>Digestive and Liver Disease</i> , 2015, 47, e15.	0.4	1
90	Bimodal strategy for excellent audiological rehabilitation in a subject with a novel nonsense mutation of the SLC26A4 gene: A case report. <i>International Journal of Pediatric Otorhinolaryngology</i> , 2020, 134, 110018.	0.4	1

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91	Towards an External Quality Assessment for Next Generation Sequencing in the Diagnosis of Rare Inherited Anaemias. <i>Blood</i> , 2018, 132, 4936-4936.	0.6	1
92	Imatinib Protects Against Hypoxia/Reoxygenation Induced Lung and Kidney Injury in a Humanized Mouse Model for SCD. <i>Blood</i> , 2018, 132, 725-725.	0.6	1
93	Evaluation of the Main Regulators of Systemic Iron Homeostasis in Pyruvate Kinase Deficiency. <i>Blood</i> , 2021, 138, 1993-1993.	0.6	1
94	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
95	Novel PKLR missense mutation (A300P) causing pyruvate kinase deficiency in an Omani Kindredâ€™PK deficiency masquerading as congenital dyserythropoietic anemia. <i>Clinical Case Reports (discontinued)</i> , 2022, 10, e05315.	0.2	1
96	Novel Insights and Future Perspective in Iron Metabolism and Anemia. <i>Metabolites</i> , 2022, 12, 138.	1.3	1
97	Genetic predictors of response to treatment of chronic hepatitis C virus infection in patients from southern Italy. <i>Italian Journal of Medicine</i> , 2014, , .	0.2	0
98	The frameshift Leu220Phefs*2 variant in KRIT1 accounts for early acute bleeding in patients affected by cerebral cavernous malformation. <i>Interdisciplinary Neurosurgery: Advanced Techniques and Case Management</i> , 2021, 26, 101367.	0.2	0
99	Common Variants at 21q22.3 Locus Influence <i>MX1</i> Gene Expression and Susceptibility to Severe COVID-19. <i>SSRN Electronic Journal</i> , 0, , .	0.4	0
100	Allelic Distribution Assessment of SEC23B Mutations In Cda II Patients From Italy: Towards the Identification of Possible Founder Mutations. <i>Blood</i> , 2010, 116, 1005-1005.	0.6	0
101	In Vitro Characterization of R14W Mutation in SEC23B, the CDAlI Causative Gene. <i>Blood</i> , 2011, 118, 2098-2098.	0.6	0
102	Missense Mutations in the ABCB6 Transporter Cause Dominant Familial Pseudohyperkalemia. <i>Blood</i> , 2012, 120, 3184-3184.	0.6	0
103	Detection of Familial Pseudohyperkalemia Among Italian Blood Donors By Genetic Screening for the R276W Mutation in ABCB6. <i>Blood</i> , 2015, 126, 2132-2132.	0.6	0
104	Erfe-Encoding FAM132B in Congenital Dyserythropoietic Anemia Type II. <i>Blood</i> , 2015, 126, 535-535.	0.6	0
105	Anaemias diagnosis by label-free quantitative phase imaging. , 2019, , .		0
106	S1629 ERFEâ€™A260S ACCOUNTS FOR HEPATIC IRON METABOLISM IMPAIRMENT BY ACTING ON THE BMPâ€™SMAD PATHWAY. <i>HemaSphere</i> , 2019, 3, 753.	1.2	0
107	Diagnostic decision support tool for anemias based on label-free holographic imaging. , 2019, , .		0
108	ERN-EuroBloodNet European Registry of Patients Affected by Red Blood Cell Disorders and COVID-19. <i>Blood</i> , 2021, 138, 4058-4058.	0.6	0

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109	Editorial: Genetics and Genomics of Red Blood Cells. <i>Frontiers in Physiology</i> , 2021, 12, 822156.	1.3	0