Roberta Russo

List of Publications by Year
in descending order

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	Differential diagnosis of hereditary anemias from a fraction of blood drop by digital holography and hierarchical machine learning. Biosensors and Bioelectronics, 2022, 201, 113945.	5.3	19
2	SEC23B Loss-of-Function Suppresses Hepcidin Expression by Impairing Glycosylation Pathway in Human Hepatic Cells. International Journal of Molecular Sciences, 2022, 23, 1304.	1.8	6
3	Novel PKLR missense mutation (A300P) causing pyruvate kinase deficiency in an Omani Kindredâ€"PK deficiency masquerading as congenital dyserythropoietic anemia. Clinical Case Reports (discontinued), 2022, 10, e05315.	0.2	1
4	Novel Insights and Future Perspective in Iron Metabolism and Anemia. Metabolites, 2022, 12, 138.	1.3	1
5	Common, low-frequency, rare, and ultra-rare coding variants contribute to COVID-19 severity. Human Genetics, 2022, 141, 147-173.	1.8	22
6	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
7	Hereditary anemia caused by multilocus inheritance of <i>PIEZO1</i>, <i>SLC4A1</i> and <i>ABCB6</i> mutations: a diagnostic and therapeutic challenge. Haematologica, 2022, 107, 2280-2284.	1.7	2
8	Germline rare variants of lectin pathway genes predispose to asymptomatic SARS-CoV-2 infection in elderly individuals. Genetics in Medicine, 2022, , .	1.1	7
9	The use of <scp>nextâ€generation</scp> sequencing in the diagnosis of rare inherited anaemias: A Joint BSH/EHA Good Practice Paper*. British Journal of Haematology, 2022, 198, 459-477.	1.2	3
10	The Use of Next-generation Sequencing in the Diagnosis of Rare Inherited Anaemias: A Joint BSH/EHA Good Practice Paper. HemaSphere, 2022, 6, e739.	1.2	6
11	Employing a systematic approach to biobanking and analyzing clinical and genetic data for advancing COVID-19 research. European Journal of Human Genetics, 2021, 29, 745-759.	1.4	35
12	Dysregulation of lipid metabolism and pathological inflammation in patients with COVID-19. Scientific Reports, 2021, 11, 2941.	1.6	102
13	Shorter androgen receptor polyQ alleles protect against life-threatening COVID-19 disease in European males. EBioMedicine, 2021, 65, 103246.	2.7	52
14	Association of Toll-like receptor 7 variants with life-threatening COVID-19 disease in males: findings from a nested case-control study. ELife, 2021, 10, .	2.8	145
15	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
16	Recommendations for pregnancy in Fanconi anemia. Expert Opinion on Biological Therapy, 2021, 21, 1-7.	1.4	2
17	Protective Role of a TMPRSS2 Variant on Severe COVID-19 Outcome in Young Males and Elderly Women. Genes, 2021, 12, 596.	1.0	39
18	The pyruvate kinase activator mitapivat reduces hemolysis and improves anemia in a Î²-thalassemia mouse model. Journal of Clinical Investigation, 2021, 131, .	3.9	39

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19	Regulatory Noncoding and Predicted Pathogenic Coding Variants of CCR5 Predispose to Severe COVID-19. International Journal of Molecular Sciences, 2021, 22, 5372.	1.8	16
20	Complex Modes of Inheritance in Hereditary Red Blood Cell Disorders: A Case Series Study of 155 Patients. Genes, 2021, 12, 958.	1.0	22
21	The TNFRSF13C H159Y Variant Is Associated with Severe COVID-19: A Retrospective Study of 500 Patients from Southern Italy. Genes, 2021, 12, 881.	1.0	12
22	The EHA Research Roadmap: Anemias. HemaSphere, 2021, 5, e607.	1.2	7
23	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
24	Recommendations for diagnosis and treatment of methemoglobinemia. American Journal of Hematology, 2021, 96, 1666-1678.	2.0	56
25	The frameshift Leu220Phefs*2 variant in KRIT1 accounts for early acute bleeding in patients affected by cerebral cavernous malformation. Interdisciplinary Neurosurgery: Advanced Techniques and Case Management, 2021, 26, 101367.	0.2	0
26	Genetic mechanisms of critical illness in COVID-19. Nature, 2021, 591, 92-98.	13.7	1,014
27	Evaluation of the Main Regulators of Systemic Iron Homeostasis in Pyruvate Kinase Deficiency. Blood, 2021, 138, 1993-1993.	0.6	1
28	ERN-EuroBloodNet European Registry of Patients Affected by Red Blood Cell Disorders and COVID-19. Blood, 2021, 138, 4058-4058.	0.6	0
29	PIEZO1 Mutation May Determine Early Onset of Clinical Manifestation of Anemia of Myelodysplastic Syndromes. Blood, 2021, 138, 1528-1528.	0.6	2
30	Summary of Joint European Hematology Association (EHA) and EuroBloodNet Recommendations on Diagnosis and Treatment of Methemoglobinemia. HemaSphere, 2021, 5, e660.	1.2	1
31	Editorial: Genetics and Genomics of Red Blood Cells. Frontiers in Physiology, 2021, 12, 822156.	1.3	0
32	Gainâ€ofâ€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	ACE2 gene variants may underlie interindividual variability and susceptibility to COVID-19 in the Italian population. European Journal of Human Genetics, 2020, 28, 1602-1614.	1.4	208
34	Inherited microcytic anemias. Hematology American Society of Hematology Education Program, 2020, 2020, 465-470.	0.9	12
35	RAP-011 Rescues the Disease Phenotype in a Cellular Model of Congenital Dyserythropoietic Anemia Type II by Inhibiting the SMAD2-3 Pathway. International Journal of Molecular Sciences, 2020, 21, 5577.	1.8	9
36	Congenital dyserythropoietic anemias. Blood, 2020, 136, 1274-1283.	0.6	62

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37	A novel PIEZO1 mutation in a patient with dehydrated hereditary stomatocytosis: a case report and a brief review of literature. Italian Journal of Pediatrics, 2020, 46, 102.	1.0	8
38	Recommendations for Pregnancy in Rare Inherited Anemias. HemaSphere, 2020, 4, e446.	1.2	8
39	Genetic Analysis of the Coronavirus SARS-CoV-2 Host Protease TMPRSS2 in Different Populations. Frontiers in Genetics, 2020, 11, 872.	1.1	40
40	Uridine treatment normalizes the <scp>congenital dyserythropoietic anemia type</scp><scp>II</scp>â€like hematological phenotype in a patient with homozygous mutation in the <scp>CAD</scp> gene. American Journal of Hematology, 2020, 95, 1423-1426.	2.0	8
41	Genetics and Genomics Approaches for Diagnosis and Research Into Hereditary Anemias. Frontiers in Physiology, 2020, 11, 613559.	1.3	27
42	Bimodal strategy for excellent audiological rehabilitation in a subject with a novel nonsense mutation of the SLC26A4 gene: A case report. International Journal of Pediatric Otorhinolaryngology, 2020, 134, 110018.	0.4	1
43	Hematopoietic Stem Cell Transplantation in Congenital Dyserythropetic Anemia Type II: A Case Report and Review of the Literature. Journal of Pediatric Hematology/Oncology, 2020, 42, e507-e510.	0.3	7
44	Apparent recessive inheritance of sideroblastic anemia type 2 due to uniparental isodisomy at the SLC25A38 locus. Haematologica, 2020, 105, 2883-2886.	1.7	4
45	The BMPâ€SMAD pathway mediates the impaired hepatic iron metabolism associated with the ERFEâ€A260S variant. American Journal of Hematology, 2019, 94, 1227-1235.	2.0	21
46	Germline mutations and new copy number variants among 40 pediatric cancer patients suspected for genetic predisposition. Clinical Genetics, 2019, 96, 359-365.	1.0	14
47	Advances in understanding the pathogenesis of red cell membrane disorders. British Journal of Haematology, 2019, 187, 13-24.	1.2	64
48	Characterization of Two Cases of Congenital Dyserythropoietic Anemia Type I Shed Light on the Uncharacterized C15orf41 Protein. Frontiers in Physiology, 2019, 10, 621.	1.3	16
49	CoDysAn: A Telemedicine Tool to Improve Awareness and Diagnosis for Patients With Congenital Dyserythropoietic Anemia. Frontiers in Physiology, 2019, 10, 1063.	1.3	4
50	PIEZO1 Hypomorphic Variants in Congenital Lymphatic Dysplasia Cause Shape and Hydration Alterations of Red Blood Cells. Frontiers in Physiology, 2019, 10, 258.	1.3	26
51	Hereditary spherocytosis and allied disorders. HemaSphere, 2019, 3, 157-159.	1.2	2
52	Anaemias diagnosis by label-free quantitative phase imaging. , 2019, , .		0
53	S1629 ERFEâ€A260S ACCOUNTS FOR HEPATIC IRON METABOLISM IMPAIRMENT BY ACTING ON THE BMPâ€SMAD PATHWAY. HemaSphere, 2019, 3, 753.	1.2	0
54	Diagnostic decision support tool for anemias based on label-free holographic imaging. , 2019, , .		0

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55	Multiâ€gene panel testing improves diagnosis and management of patients with hereditary anemias. American Journal of Hematology, 2018, 93, 672-682.	2.0	117
56	PIEZO1-R1864H rare variant accounts for a genetic phenotype-modifier role in dehydrated hereditary stomatocytosis. Haematologica, 2018, 103, e94-e97.	1.7	18
57	Hereditary stomatocytosis: An underdiagnosed condition. American Journal of Hematology, 2018, 93, 107-121.	2.0	54
58	Genotypeâ€phenotype correlation and risk stratification in a cohort of 123 hereditary stomatocytosis patients. American Journal of Hematology, 2018, 93, 1509-1517.	2.0	48
59	Label-Free Optical Marker for Red-Blood-Cell Phenotyping of Inherited Anemias. Analytical Chemistry, 2018, 90, 7495-7501.	3.2	49
60	Towards an External Quality Assessment for Next Generation Sequencing in the Diagnosis of Rare Inherited Anaemias. Blood, 2018, 132, 4936-4936.	0.6	1
61	Imatinib Protects Against Hypoxia/Reoxygenation Induced Lung and Kidney Injury in a Humanized Mouse Model for SCD. Blood, 2018, 132, 725-725.	0.6	1
62	Recommendations regarding splenectomy in hereditary hemolytic anemias. Haematologica, 2017, 102, 1304-1313.	1.7	138
63	GATA1 erythroid-specific regulation of SEC23B expression and its implication in the pathogenesis of congenital dyserythropoietic anemia type II. Haematologica, 2017, 102, e371-e374.	1.7	16
64	Targeted next generation sequencing identifies a novel Î²â€spectrin gene mutation A2059P in two Omani children with hereditary pyropoikilocytosis. American Journal of Hematology, 2017, 92, E607-E609.	2.0	8
65	Kinome expression profiling of human neuroblastoma tumors identifies potential drug targets for ultra high-risk patients. Carcinogenesis, 2017, 38, 1011-1020.	1.3	17
66	Protease inhibitorsâ€based therapy induces acquired spherocyticâ€like anaemia and ineffective erythropoiesis in chronic hepatitis C virus patients. Liver International, 2016, 36, 49-58.	1.9	11
67	Functional characterization of novel ABCB6 mutations and their clinical implications in familial pseudohyperkalemia. Haematologica, 2016, 101, 909-917.	1.7	30
68	Increased levels of ERFE-encoding FAM132B in patients with congenital dyserythropoietic anemia type II. Blood, 2016, 128, 1899-1902.	0.6	26
69	New insights on hereditary erythrocyte membrane defects. Haematologica, 2016, 101, 1284-1294.	1.7	156
70	Next generation research and therapy in red blood cell diseases. Haematologica, 2016, 101, 515-517.	1.7	6
71	The European Hematology Association Roadmap for European Hematology Research: a consensus document. Haematologica, 2016, 101, 115-208.	1.7	67
72	Diagnosis and management of congenital dyserythropoietic anemias. Expert Review of Hematology, 2016, 9, 283-296.	1.0	76

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73	Acquired spherocytic like anemia combined with ineffective erythropoiesis sustains anemia in patients with chronic hepatitis C infection receiving telaprevir or boceprevir-based triple therapy. Digestive and Liver Disease, 2015, 47, e15.	0.4	1
74	Novel Gardos channel mutations linked to dehydrated hereditary stomatocytosis (xerocytosis). American Journal of Hematology, 2015, 90, 921-926.	2.0	81
75	Red cells in post-genomic era: impact of personalized medicine in the treatment of anemias. Haematologica, 2015, 100, 3-6.	1.7	18
76	Congenital erythropoietic porphyria linked to <scp>GATA</scp>1â€<scp>R</scp>216<scp>W</scp> mutation: challenges for diagnosis. European Journal of Haematology, 2015, 94, 491-497.	1.1	39
77	Detection of Familial Pseudohyperkalemia Among Italian Blood Donors By Genetic Screening for the R276W Mutation in ABCB6. Blood, 2015, 126, 2132-2132.	0.6	0
78	Erfe-Encoding FAM132B in Congenital Dyserythropoietic Anemia Type II. Blood, 2015, 126, 535-535.	0.6	0
79	Genetic predictors of response to treatment of chronic hepatitis C virus infection in patients from southern Italy. Italian Journal of Medicine, 2014, , .	0.2	0
80	Successful hematopoietic stem cell transplantation in a patient with congenital dyserythropoietic anemia type <scp>II</scp>. Pediatric Transplantation, 2014, 18, E130-3.	0.5	25
81	Retrospective cohort study of 205 cases with congenital dyserythropoietic anemia type II: Definition of clinical and molecular spectrum and identification of new diagnostic scores. American Journal of Hematology, 2014, 89, E169-75.	2.0	68
82	Inherited hematological disorders due to defects in coat protein (COP)II complex. American Journal of Hematology, 2013, 88, 135-140.	2.0	16
83	Hypomorphic mutations of SEC23B gene account for mild phenotypes of congenital dyserythropoietic anemia type II. Blood Cells, Molecules, and Diseases, 2013, 51, 17-21.	0.6	33
84	Missense mutations in the ABCB6 transporter cause dominant familialpseudohyperkalemia. American Journal of Hematology, 2013, 88, 66-72.	2.0	67
85	Replication of GWAS-identified neuroblastoma risk loci strengthens the role of BARD1 and affirms the cumulative effect of genetic variations on disease susceptibility. Carcinogenesis, 2013, 34, 605-611.	1.3	95
86	Multiple clinical forms of dehydrated hereditary stomatocytosis arise from mutations in PIEZO1. Blood, 2013, 121, 3925-3935.	0.6	266
87	Clinical aspects and pathogenesis of congenital dyserythropoietic anemias: from morphology to molecular approach. Haematologica, 2012, 97, 1786-1794.	1.7	68
88	Integration of Pharmacogenetics and Pharmacogenomics in Drug Development: Implications for Regulatory and Medical Decision Making in Pediatric Diseases. Journal of Clinical Pharmacology, 2012, 52, 704-716.	1.0	15
89	A novel GLA mutation in a Fabry family with glucose-6-phosphate dehydrogenase deficiency. Journal of Nephrology, 2012, 25, 582-585.	0.9	2
90	Missense Mutations in the ABCB6 Transporter Cause Dominant Familial Pseudohyperkalemia. Blood, 2012, 120, 3184-3184.	0.6	0

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91	Congenital dyserythropoietic anemias. Current Opinion in Hematology, 2011, 18, 146-151.	1.2	44
92	Pediatric pharmacogenetic and pharmacogenomic studies: the current state and future perspectives. European Journal of Clinical Pharmacology, 2011, 67, 17-27.	0.8	19
93	Two founder mutations in the SEC23B gene account for the relatively high frequency of CDA II in the Italian population. American Journal of Hematology, 2011, 86, 727-732.	2.0	27
94	In Vitro Characterization of R14W Mutation in SEC23B, the CDAII Causative Gene. Blood, 2011, 118, 2098-2098.	0.6	0
95	Congenital dyserythropoietic anaemias: new acquisitions. Blood Transfusion, 2011, 9, 278-80.	0.3	2
96	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
97	Mutational spectrum in congenital dyserythropoietic anemia type II: Identification of 19 novel variants in SEC23B gene. American Journal of Hematology, 2010, 85, 915-920.	2.0	40
98	Galectin-1 and Its Involvement in Hepatocellular Carcinoma Aggressiveness. Molecular Medicine, 2010, 16, 102-115.	1.9	69
99	A Dyserythropoietic Anemia Associated with Homozygous Hb Plasencia [Î±125(H8)Leuâ†'Arg (Î±2)] (HBA2:c.377T>G), A Variant with an Unstable Î± Chain. Hemoglobin, 2010, 34, 576-581.	0.4	3
100	Regulation of divalent metal transporter 1 (DMT1) non-IRE isoform by the microRNA Let-7d in erythroid cells. Haematologica, 2010, 95, 1244-1252.	1.7	82
101	MDM2 SNP309 and p53 Arg72Pro in cutaneous melanoma: association between SNP309 GG genotype and tumor Breslow thickness. Journal of Human Genetics, 2010, 55, 518-524.	1.1	14
102	Allelic Distribution Assessment of SEC23B Mutations In Cda II Patients From Italy: Towards the Identification of Possible Founder Mutations. Blood, 2010, 116, 1005-1005.	0.6	0
103	SOCS3 and IRS-1 gene expression differs between genotype 1 and genotype 2 hepatitis C virus-infected HepG2 cells. Clinical Chemistry and Laboratory Medicine, 2009, 47, 1217-25.	1.4	22
104	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. Journal of Cancer Research and Clinical Oncology, 2009, 135, 1799-1807.	1.2	36
105	Mutations affecting the secretory COPII coat component SEC23B cause congenital dyserythropoietic anemia type II. Nature Genetics, 2009, 41, 936-940.	9.4	250
106	Elevated expression and polymorphisms of SOCS3 influence patient response to antiviral therapy in chronic hepatitis C. Gut, 2007, 57, 507-515.	6.1	84
107	Comparative Proteomic Expression Profile in All-transRetinoic Acid Differentiated Neuroblastoma Cell Line. Journal of Proteome Research, 2007, 6, 2550-2564.	1.8	30
108	Suppressor of cytokine signaling 3 (SOCS3) expression and hepatitis C virusâ€"related chronic hepatitis: Insulin resistance and response to antiviral therapy. Hepatology, 2007, 46, 1009-1015.	3.6	150

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109	Common Variants at 21q22.3 Locus Influence MX1 Gene Expression and Susceptibility to Severe COVID-19. SSRN Electronic Journal, 0, , .	0.4	0