Immacolata Andolfo

List of Publications by Year
in descending order

100 papers	3,414 citations	172207 29 h-index	155451 55 g-index
104 all docs	104 docs citations	104 times ranked	4906 citing authors

#	Article	IF	Citations
1	Nobel prize in physiology or medicine 2021, receptors for temperature and touch: Implications for hematology. American Journal of Hematology, 2022, 97, 168-170.	2.0	5
2	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
3	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
4	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
5	Novel Insights and Future Perspective in Iron Metabolism and Anemia. Metabolites, 2022, 12, 138.	1.3	1
6	Deferasirox-induced robust and dose-dependent reversal of anemia in a patient with variants in the TRIB2 and ABCB6 genes. Blood Advances, 2022, , .	2.5	1
7	Germline rare variants of lectin pathway genes predispose to asymptomatic SARS-CoV-2 infection in elderly individuals. Genetics in Medicine, 2022, , .	1.1	7
8	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
9	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
10	A Novel Îµ[3]Î²-Thalassemia Deletion Associated with Severe Anemia at Birth and a Î²-Thalassemia Intermedia Phenotype Later in Life in Three Generations of a Greek Family. Hemoglobin, 2021, 45, 351-354.	0.4	3
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	A role of PIEZO1 in iron metabolism in mice and humans. Cell, 2021, 184, 969-982.e13.	13.5	108
13	Dysregulation of lipid metabolism and pathological inflammation in patients with COVID-19. Scientific Reports, 2021, 11, 2941.	1.6	102
14	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
15	Recommendations for pregnancy in Fanconi anemia. Expert Opinion on Biological Therapy, 2021, 21, 1-7.	1.4	2
16	Therapeutic targeting of Lyn kinase to treat chorea-acanthocytosis. Acta Neuropathologica Communications, 2021, 9, 81.	2.4	19
17	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
18	Complex Modes of Inheritance in Hereditary Red Blood Cell Disorders: A Case Series Study of 155 Patients. Genes, 2021, 12, 958.	1.0	22

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19	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
20	The EHA Research Roadmap: Anemias. HemaSphere, 2021, 5, e607.	1.2	7
21	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
22	Recommendations for diagnosis and treatment of methemoglobinemia. American Journal of Hematology, 2021, 96, 1666-1678.	2.0	56
23	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
24	Evaluation of the Main Regulators of Systemic Iron Homeostasis in Pyruvate Kinase Deficiency. Blood, 2021, 138, 1993-1993.	0.6	1
25	Nrf2 Plays a Key Role in Iron-Overload Cardiomyopathy. Blood, 2021, 138, 3068-3068.	0.6	0
26	Summary of Joint European Hematology Association (EHA) and EuroBloodNet Recommendations on Diagnosis and Treatment of Methemoglobinemia. HemaSphere, 2021, 5, e660.	1.2	1
27	Editorial: Genetics and Genomics of Red Blood Cells. Frontiers in Physiology, 2021, 12, 822156.	1.3	0
28	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
29	M170. GENETIC CHARACTERIZATION OF A COHORT OF PATIENTS AFFECTED BY SCHIZOPHRENIA. THE ROLE FOR RARE STRUCTURAL VARIANTS IN MODULATING TREATMENT RESISTANT ENDOPHENOTYPES: PRELIMINARY DATA. Schizophrenia Bulletin, 2020, 46, S201-S201.	2.3	0
30	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
31	Inherited microcytic anemias. Hematology American Society of Hematology Education Program, 2020, 2020, 465-470.	0.9	12
32	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
33	Congenital dyserythropoietic anemias. Blood, 2020, 136, 1274-1283.	0.6	62
34	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
35	Recommendations for Pregnancy in Rare Inherited Anemias. HemaSphere, 2020, 4, e446.	1.2	8
36	Genetic Analysis of the Coronavirus SARS-CoV-2 Host Protease TMPRSS2 in Different Populations. Frontiers in Genetics, 2020, 11, 872.	1.1	40

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37	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
38	Genetics and Genomics Approaches for Diagnosis and Research Into Hereditary Anemias. Frontiers in Physiology, 2020, 11, 613559.	1.3	27
39	Kinome multigenic panel identified novel druggable EPHB4â€V871I somatic variant in highâ€risk neuroblastoma. Journal of Cellular and Molecular Medicine, 2020, 24, 6459-6471.	1.6	7
40	Apparent recessive inheritance of sideroblastic anemia type 2 due to uniparental isodisomy at the SLC25A38 locus. Haematologica, 2020, 105, 2883-2886.	1.7	4
41	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
42	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
43	Advances in understanding the pathogenesis of red cell membrane disorders. British Journal of Haematology, 2019, 187, 13-24.	1.2	64
44	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
45	CoDysAn: A Telemedicine Tool to Improve Awareness and Diagnosis for Patients With Congenital Dyserythropoietic Anemia. Frontiers in Physiology, 2019, 10, 1063.	1.3	4
46	Erythrocyte ion content and dehydration modulate maximal Gardos channel activity in KCNN4 V282M/+ hereditary xerocytosis red cells. American Journal of Physiology - Cell Physiology, 2019, 317, C287-C302.	2.1	11
47	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
48	Hereditary spherocytosis and allied disorders. HemaSphere, 2019, 3, 157-159.	1.2	2
49	Resolution of sickle cell diseaseâ€“associated inflammation and tissue damage with 17R-resolvin D1. Blood, 2019, 133, 252-265.	0.6	50
50	Bitopertin, a selective oral GLYT1 inhibitor, improves anemia in a mouse model of Î²-thalassemia. JCI Insight, 2019, 4, .	2.3	19
51	Anaemias diagnosis by label-free quantitative phase imaging. , 2019, , .		0
52	Diagnostic decision support tool for anemias based on label-free holographic imaging. , 2019, , .		0
53	The Novel Role That Nrf2 Plays in Erythropoiesis during Aging. Blood, 2019, 134, 3502-3502.	0.6	0
54	Multiâ€gene panel testing improves diagnosis and management of patients with hereditary anemias. American Journal of Hematology, 2018, 93, 672-682.	2.0	117

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55	PIEZO1-R1864H rare variant accounts for a genetic phenotype-modifier role in dehydrated hereditary stomatocytosis. Haematologica, 2018, 103, e94-e97.	1.7	18
56	Hereditary stomatocytosis: An underdiagnosed condition. American Journal of Hematology, 2018, 93, 107-121.	2.0	54
57	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
58	Label-Free Optical Marker for Red-Blood-Cell Phenotyping of Inherited Anemias. Analytical Chemistry, 2018, 90, 7495-7501.	3.2	49
59	Increased Red Cell KCNN4 Activity in Sporadic Hereditary Xerocytosis Associated With Enhanced Single Channel Pressure Sensitivity of PIEZO1ÂMutant V598M. HemaSphere, 2018, 2, e55.	1.2	10
60	Recommendations regarding splenectomy in hereditary hemolytic anemias. Haematologica, 2017, 102, 1304-1313.	1.7	138
61	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
62	Data demonstrating the role of peroxiredoxin 2 as important anti-oxidant system in lung homeostasis. Data in Brief, 2017, 15, 376-381.	0.5	0
63	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
64	Peroxiredoxin-2 plays a pivotal role as multimodal cytoprotector in the early phase of pulmonary hypertension. Free Radical Biology and Medicine, 2017, 112, 376-386.	1.3	28
65	Functional characterization of novel ABCB6 mutations and their clinical implications in familial pseudohyperkalemia. Haematologica, 2016, 101, 909-917.	1.7	30
66	Increased levels of ERFE-encoding FAM132B in patients with congenital dyserythropoietic anemia type II. Blood, 2016, 128, 1899-1902.	0.6	26
67	New insights on hereditary erythrocyte membrane defects. Haematologica, 2016, 101, 1284-1294.	1.7	156
68	Next generation research and therapy in red blood cell diseases. Haematologica, 2016, 101, 515-517.	1.7	6
69	The European Hematology Association Roadmap for European Hematology Research: a consensus document. Haematologica, 2016, 101, 115-208.	1.7	67
70	Diagnosis and management of congenital dyserythropoietic anemias. Expert Review of Hematology, 2016, 9, 283-296.	1.0	76
71	Dietary Â-3 fatty acids protect against vasculopathy in a transgenic mouse model of sickle cell disease. Haematologica, 2015, 100, 870-880.	1.7	51
72	Novel Gardos channel mutations linked to dehydrated hereditary stomatocytosis (xerocytosis). American Journal of Hematology, 2015, 90, 921-926.	2.0	81

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73	Red cells in post-genomic era: impact of personalized medicine in the treatment of anemias. Haematologica, 2015, 100, 3-6.	1.7	18
74	Inhibition of hypoxia inducible factors combined with all-trans retinoic acid treatment enhances glial transdifferentiation of neuroblastoma cells. Scientific Reports, 2015, 5, 11158.	1.6	26
75	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
76	Erfe-Encoding FAM132B in Congenital Dyserythropoietic Anemia Type II. Blood, 2015, 126, 535-535.	0.6	0
77	Hereditary xerocytosis revisited. American Journal of Hematology, 2014, 89, 1142-1146.	2.0	47
78	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
79	Dietary Ï‰-3 Fatty Acid Supplementation As a Potential New Therapy for Vasculopathy in Sickle Cell Disease: Proof of Concept in a Transgenic Mouse Model. Blood, 2014, 124, 220-220.	0.6	5
80	MicroRNA 199b-5p delivery through stable nucleic acid lipid particles (SNALPs) in tumorigenic cell lines. Naunyn-Schmiedeberg's Archives of Pharmacology, 2013, 386, 287-302.	1.4	30
81	Trasferrin receptor 2 gene regulation by microRNA 221 in SH-SY5Y cells treated with MPP+ as Parkinson's disease cellular model. Neuroscience Research, 2013, 77, 121-127.	1.0	24
82	Rapid Cl^{â^'}/HCO₃^{â^'}exchange kinetics of AE1 in HEK293 cells and hereditary stomatocytosis red blood cells. American Journal of Physiology - Cell Physiology, 2013, 305, C654-C662.	2.1	10
83	Missense mutations in the ABCB6 transporter cause dominant familialpseudohyperkalemia. American Journal of Hematology, 2013, 88, 66-72.	2.0	67
84	Multiple clinical forms of dehydrated hereditary stomatocytosis arise from mutations in PIEZO1. Blood, 2013, 121, 3925-3935.	0.6	266
85	The micro-RNA 199b-5p regulatory circuit involves Hes1, CD15, and epigenetic modifications in medulloblastoma. Neuro-Oncology, 2012, 14, 596-612.	0.6	48
86	The metallophosphodiesterase Mpped2 impairs tumorigenesis in neuroblastoma. Cell Cycle, 2012, 11, 569-581.	1.3	30
87	Identification and characterization of the first <scp>SLC</scp>11<scp>A</scp>2 isoform 1a mutation causing a defect in splicing process and an hypomorphic allele expression of the <scp>SLC</scp>11<scp>A</scp>2 gene. British Journal of Haematology, 2012, 159, 492-495.	1.2	6
88	Identification of ALK germline mutation (3605delG) in pediatric anaplastic medulloblastoma. Journal of Human Genetics, 2012, 57, 682-684.	1.1	19
89	Missense Mutations in the ABCB6 Transporter Cause Dominant Familial Pseudohyperkalemia. Blood, 2012, 120, 3184-3184.	0.6	0
90	Correlation of NM23-H1 cytoplasmic expression with metastatic stage in human prostate cancer tissue. Naunyn-Schmiedeberg's Archives of Pharmacology, 2011, 384, 489-498.	1.4	25

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91	Detection of erbB2 copy number variations in plasma of patients with esophageal carcinoma. BMC Cancer, 2011, 11, 126.	1.1	22
92	Chromosome 9q and 16q Loss Identified by Genome-Wide Pooled-Analysis Are Associated with Tumor Aggressiveness in Patients with Classic Medulloblastoma. OMICS A Journal of Integrative Biology, 2011, 15, 273-280.	1.0	7
93	MiR-34a Targeting of Notch Ligand Delta-Like 1 Impairs CD15+/CD133+ Tumor-Propagating Cells and Supports Neural Differentiation in Medulloblastoma. PLoS ONE, 2011, 6, e24584.	1.1	149
94	In Vitro Characterization of R14W Mutation in SEC23B, the CDAII Causative Gene. Blood, 2011, 118, 2098-2098.	0.6	0
95	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
96	MicroRNA-199b-5p Impairs Cancer Stem Cells through Negative Regulation of HES1 in Medulloblastoma. PLoS ONE, 2009, 4, e4998.	1.1	233
97	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
98	886 SOCS3 AND IRS-1 GENE EXPRESSION DIFFERS BETWEEN GENOTYPE 1 AND GENOTYPE 2 HCV- INFECTED HEPG2 CELLS. Journal of Hepatology, 2009, 50, S322.	1.8	0
99	COPII Complex Characterization During Erythroid Differentiation and Its Involvement in CDAII Disease.. Blood, 2009, 114, 3009-3009.	0.6	0
100	Regulation of DMT1 (non IRE isoform) by MicroRNA LET-7D. Blood, 2008, 112, 416-416.	0.6	0