Daniela De Rocco

List of Publications by Year
in descending order

39 papers	2,124 citations	331670 21 h-index	315739 38 g-index
39 all docs	39 docs citations	39 times ranked	2460 citing authors

#	Article	IF	Citations
1	A Novel Mutation in GP1BB Reveals the Role of the Cytoplasmic Domain of GPIbÎ² in the Pathophysiology of Bernard-Soulier Syndrome and GPIb-IX Complex Assembly. International Journal of Molecular Sciences, 2021, 22, 10190.	4.1	4
2	MYH9-Related Thrombocytopenia: Four Novel Variants Affecting the Tail Domain of the Non-Muscle Myosin Heavy Chain IIA Associated with a Mild Clinical Evolution of the Disorder. Hamostaseologie, 2019, 39, 087-094.	1.9	3
3	Hypomorphic FANCA mutations correlate with mild mitochondrial and clinical phenotype in Fanconi anemia. Haematologica, 2018, 103, 417-426.	3.5	26
4	A new form of inherited thrombocytopenia due to monoallelic loss of function mutation in the thrombopoietin gene. British Journal of Haematology, 2018, 181, 698-701.	2.5	21
5	Thrombopoietin mutation in congenital amegakaryocytic thrombocytopenia treatable withÂromiplostim. EMBO Molecular Medicine, 2018, 10, 63-75.	6.9	47
6	Mutations of RUNX1 in families with inherited thrombocytopenia. American Journal of Hematology, 2017, 92, E86-E88.	4.1	15
7	MYH9 gene mutations associated with bleeding. Platelets, 2017, 28, 312-315.	2.3	29
8	Somatic, hematologic phenotype, longâ€term outcome, and effect of hematopoietic stem cell transplantation. An analysis of 97 Fanconi anemia patients from the Italian national database on behalf of the Marrow Failure Study Group of the AIEOP (Italian Association of Pediatric) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 452 Td (Hematol	4.1	33
9	Clinical and pathogenic features of ETV6 -related thrombocytopenia with predisposition to acute lymphoblastic leukemia. Haematologica, 2016, 101, 1333-1342.	3.5	92
10	Clinical aspects of Fanconi anemia individuals with the same mutation of FANCF identified by next generation sequencing. Birth Defects Research Part A: Clinical and Molecular Teratology, 2015, 103, 1003-1010.	1.6	5
11	Impaired immune response to Candida albicans in cells from Fanconi anemia patients. Cytokine, 2015, 73, 203-207.	3.2	5
12	Germline mutations in ETV6 are associated with thrombocytopenia, red cell macrocytosis and predisposition to lymphoblastic leukemia. Nature Genetics, 2015, 47, 535-538.	21.4	274
13	Identification of point mutations and large intragenic deletions in Fanconi anemia using nextâ€generation sequencing technology. Molecular Genetics & Genomic Medicine, 2015, 3, 500-512.	1.2	9
14	<scp>R705H</scp> mutation of <scp>MYH9</scp> is associated with <scp>MYH9</scp>â€related disease and not only with nonâ€syndromic deafness <scp>DFNA17</scp>. Clinical Genetics, 2015, 88, 85-89.	2.0	14
15	Molecular analysis of Fanconi anemia: the experience of the Bone Marrow Failure Study Group of the Italian Association of Pediatric Onco-Hematology. Haematologica, 2014, 99, 1022-1031.	3.5	44
16	MYH9-Related Disease: A Novel Prognostic Model to Predict the Clinical Evolution of the Disease Based on Genotype-Phenotype Correlations. Human Mutation, 2014, 35, 236-247.	2.5	154
17	Unusual splice site mutations disrupt FANCA exon 8 definition. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2014, 1842, 1052-1058.	3.8	16
18	Mutations of cytochrome c identified in patients with thrombocytopenia THC4 affect both apoptosis and cellular bioenergetics. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2014, 1842, 269-274.	3.8	65

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19	Spectrum of the Mutations in Bernard-Soulier Syndrome. Human Mutation, 2014, 35, 1033-1045.	2.5	124
20	Analysis of 339 pregnancies in 181 women with 13 different forms of inherited thrombocytopenia. Haematologica, 2014, 99, 1387-1394.	3.5	63
21	MYH9-related disease: Five novel mutations expanding the spectrum of causative mutations and confirming genotype/phenotype correlations. European Journal of Medical Genetics, 2013, 56, 7-12.	1.3	26
22	Correlation between platelet phenotype and NBEAL2 genotype in patients with congenital thrombocytopenia and Â-granule deficiency. Haematologica, 2013, 98, 868-874.	3.5	49
23	ANKRD26-related thrombocytopenia and myeloid malignancies. Blood, 2013, 122, 1987-1989.	1.4	145
24	Apparent genotypeâ€“phenotype mismatch in a patient with MYH9-related disease: When the exception proves the rule. Thrombosis and Haemostasis, 2013, 110, 618-620.	3.4	6
25	Fanconi Anemia Patients Are More Susceptible to Infection with Tumor Virus SV40. PLoS ONE, 2013, 8, e79683.	2.5	6
26	Clinical and laboratory features of 103 patients from 42 Italian families with inherited thrombocytopenia derived from the monoallelic Ala156Val mutation of GPIbÂ (Bolzano mutation). Haematologica, 2012, 97, 82-88.	3.5	99
27	International collaboration as a tool for diagnosis of patients with inherited thrombocytopenia in the setting of a developing country. Journal of Thrombosis and Haemostasis, 2012, 10, 1653-1661.	3.8	22
28	Mutations in ANKRD26 are responsible for a frequent form of inherited thrombocytopenia: analysis of 78 patients from 21 families. Blood, 2011, 117, 6673-6680.	1.4	263
29	Clinical and genetic aspects of Bernard-Soulier syndrome: searching for genotype/phenotype correlations. Haematologica, 2011, 96, 417-423.	3.5	90
30	Mutations in the 5â€² UTR of ANKRD26, the Ankirin Repeat Domain 26 Gene, Cause an Autosomal-Dominant Form of Inherited Thrombocytopenia, THC2. American Journal of Human Genetics, 2011, 88, 115-120.	6.2	200
31	Clinical and Laboratory Features of 103 Patients From 42 Italian Families with Inherited Thrombocytopenia Derived From the Monoallelic Ala156Val Mutation of GPIb Alpha (Bolzano) Tj ETQq1 1 0.784314 rgBT /Overlock 10		
32	Heavy chain myosin 9-related disease (MYH9-RD): Neutrophil inclusions of myosin-9 as a pathognomonic sign of the disorder. Thrombosis and Haemostasis, 2010, 103, 826-832.	3.4	81
33	MYH9-related disease: Report on five German families and description of a novel mutation. Annals of Hematology, 2010, 89, 1057-1059.	1.8	8
34	MYH9 related disease: four novel mutations of the tail domain of myosinâ€9 correlating with a mild clinical phenotype. European Journal of Haematology, 2010, 84, 291-297.	2.2	32
35	A G to C transversion at the last nucleotide of exon 25 of the MYH9 gene results in a missense mutation rather than in a splicing defect. European Journal of Medical Genetics, 2010, 53, 256-260.	1.3	9
36	Megakaryocyte and platelet abnormalities in a patient with a W33C mutation in the conserved SH3-like domain of myosin heavy chain IIA. Thrombosis and Haemostasis, 2009, 102, 1241-1250.	3.4	15

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37	MYH9related disease: A novel missense Ala95Asp mutation of theMYH9gene. Platelets, 2009, 20, 598-602.	2.3	10
38	Identification of the first duplication in MYH9-related disease: A hot spot for unequal crossing-over within exon 24 of the MYH9 gene. European Journal of Medical Genetics, 2009, 52, 191-194.	1.3	16
39	Absence of CYCS mutations in a large Italian cohort of patients with inherited thrombocytopenias of unknown origin. Platelets, 2009, 20, 72-73.	2.3	4