Anna Savoia

List of Publications by Year
in descending order

132 papers	7,826 citations	50276 46 h-index	53230 85 g-index
133 all docs	133 docs citations	133 times ranked	5984 citing authors

#	Article	IF	Citations
1	Dysregulation of oncogenic factors by GFI1B p32: investigation of a novel <I>GFI1B</I> germline mutation. Haematologica, 2022, 107, 260-267.	3.5	1
2	GNE-related thrombocytopenia: evidence for a mutational hotspot in the ADP/substrate domain of the GNE bifunctional enzyme. Haematologica, 2022, 107, 750-754.	3.5	11
3	Things come in threes: A new complex allele and a novel deletion within the CFTR gene complicate an accurate diagnosis of cystic fibrosis. Molecular Genetics & Genomic Medicine, 2022, 10, e1926.	1.2	2
4	ETV6-related thrombocytopenia: dominant negative effect of mutations as common pathogenic mechanism. Haematologica, 2022, 107, 2249-2254.	3.5	3
5	Genomic integrity and mitochondrial metabolism defects in Warsaw syndrome cells: a comparison with Fanconi anemia. Journal of Cellular Physiology, 2021, 236, 5664-5675.	4.1	1
6	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
7	Chronic Thrombocytopenia in Children: What Could It Hide?. Blood, 2020, 136, 33-34.	1.4	0
8	Two further patients with Warsaw breakage syndrome. Is a mild phenotype possible?. Molecular Genetics & Genomic Medicine, 2019, 7, e639.	1.2	10
9	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
10	Loss-of-function mutations in PTPRJ cause a new form of inherited thrombocytopenia. Blood, 2019, 133, 1346-1357.	1.4	40
11	MYH9: Structure, functions and role of non-muscle myosin IIA in human disease. Gene, 2018, 664, 152-167.	2.2	187
12	Hypomorphic FANCA mutations correlate with mild mitochondrial and clinical phenotype in Fanconi anemia. Haematologica, 2018, 103, 417-426.	3.5	26
13	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
14	Thrombopoietin mutation in congenital amegakaryocytic thrombocytopenia treatable withÂromiplostim. EMBO Molecular Medicine, 2018, 10, 63-75.	6.9	47
15	<scp>ACTN</scp>1 mutations lead to a benign form of platelet macrocytosis not always associated with thrombocytopenia. British Journal of Haematology, 2018, 183, 276-288.	2.5	16
16	Mutations of RUNX1 in families with inherited thrombocytopenia. American Journal of Hematology, 2017, 92, E86-E88.	4.1	15
17	MYH9 gene mutations associated with bleeding. Platelets, 2017, 28, 312-315.	2.3	29
18	Gray platelet syndrome: Novel mutations of the NBEAL2 gene. American Journal of Hematology, 2017, 92, E20-E22.	4.1	12

#	Article	IF	Citations
19	Molecular diagnosis of thrombocytopeniaâ€absent radius syndrome using nextâ€generation sequencing. International Journal of Laboratory Hematology, 2016, 38, 412-418.	1.3	10
20	Molecular basis of inherited thrombocytopenias. Clinical Genetics, 2016, 89, 154-162.	2.0	22
21	Nonmuscle Myosin Heavy Chain IIA Mutation Predicts Severity and Progression of Sensorineural Hearing Loss in Patients With MYH9-Related Disease. Ear and Hearing, 2016, 37, 112-120.	2.1	24
22	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 50 612 Td (Hematol	4.1	33
23	Clinical and pathogenic features of ETV6 -related thrombocytopenia with predisposition to acute lymphoblastic leukemia. Haematologica, 2016, 101, 1333-1342.	3.5	92
24	Molecular basis of inherited thrombocytopenias: an update. Current Opinion in Hematology, 2016, 23, 486-492.	2.5	17
25	Evaluation of energy metabolism and calcium homeostasis in cells affected by Shwachman-Diamond syndrome. Scientific Reports, 2016, 6, 25441.	3.3	39
26	ACTN1-related thrombocytopenia: identification of novel families for phenotypic characterization. Blood, 2015, 125, 869-872.	1.4	57
27	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
28	Abnormal cytoplasmic extensions associated with active Î±IIbÎ²3 are probably the cause for macrothrombocytopenia in Glanzmann thrombasthenia-like syndrome. Blood Coagulation and Fibrinolysis, 2015, 26, 302-308.	1.0	17
29	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
30	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
31	<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
32	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
33	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
34	Unusual splice site mutations disrupt FANCA exon 8 definition. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2014, 1842, 1052-1058.	3.8	16
35	Polyubiquitinated proteins, proteasome, and glycogen characterize the particle-rich cytoplasmic structure (PaCS) of neoplastic and fetal cells. Histochemistry and Cell Biology, 2014, 141, 483-497.	1.7	8
36	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

#	Article	IF	Citations
37	Spectrum of the Mutations in Bernard-Soulier Syndrome. Human Mutation, 2014, 35, 1033-1045.	2.5	124
38	Platelet diameters in inherited thrombocytopenias: analysis of 376 patients with all known disorders. Blood, 2014, 124, e4-e10.	1.4	112
39	Analysis of 339 pregnancies in 181 women with 13 different forms of inherited thrombocytopenia. Haematologica, 2014, 99, 1387-1394.	3.5	63
40	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
41	Inherited thrombocytopenias frequently diagnosed in adults. Journal of Thrombosis and Haemostasis, 2013, 11, 1006-1019.	3.8	87
42	Correlation between platelet phenotype and NBEAL2 genotype in patients with congenital thrombocytopenia and Â-granule deficiency. Haematologica, 2013, 98, 868-874.	3.5	49
43	ANKRD26-related thrombocytopenia and myeloid malignancies. Blood, 2013, 122, 1987-1989.	1.4	145
44	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
45	Fanconi Anemia Patients Are More Susceptible to Infection with Tumor Virus SV40. PLoS ONE, 2013, 8, e79683.	2.5	6
46	MYH9-related Disorders. Journal of Pediatric Hematology/Oncology, 2012, 34, 412-415.	0.6	4
47	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
48	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
49	Genetics of familial forms of thrombocytopenia. Human Genetics, 2012, 131, 1821-1832.	3.8	85
50	Alteration of Liver Enzymes Is a Feature of the Myh9-Related Disease Syndrome. PLoS ONE, 2012, 7, e35986.	2.5	38
51	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
52	Clinical and genetic aspects of Bernard-Soulier syndrome: searching for genotype/phenotype correlations. Haematologica, 2011, 96, 417-423.	3.5	90
53	Recent advances in the understanding and management of MYH9â€related inherited thrombocytopenias. British Journal of Haematology, 2011, 154, 161-174.	2.5	196
54	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

#	Article	IF	Citations
55	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
56	Eltrombopag for the treatment of the inherited thrombocytopenia deriving from MYH9 mutations. Blood, 2010, 116, 5832-5837.	1.4	141
57	MYH9-related disease: Report on five German families and description of a novel mutation. Annals of Hematology, 2010, 89, 1057-1059.	1.8	8
58	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
59	Expression and association data strongly support JARID2 involvement in nonsyndromic cleft lip with or without cleft palate. Human Mutation, 2010, 31, 794-800.	2.5	19
60	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
61	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
62	MYH9related disease: A novel missense Ala95Asp mutation of theMYH9gene. Platelets, 2009, 20, 598-602.	2.3	10
63	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
64	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
65	Dominant inheritance of a novel integrin Â3 mutation associated with a hereditary macrothrombocytopenia and platelet dysfunction in two Italian families. Haematologica, 2009, 94, 663-669.	3.5	64
66	Position of nonmuscle myosin heavy chain IIA (NMMHC-IIA) mutations predicts the natural history ofMYH9-related disease. Human Mutation, 2008, 29, 409-417.	2.5	172
67	Investigation of MYH14 as a candidate gene in cleft lip with or without cleft palate. European Journal of Oral Sciences, 2008, 116, 287-290.	1.5	4
68	Cleft lip with or without cleft palate: implication of the heavy chain of non-muscle myosin IIA. Journal of Medical Genetics, 2007, 44, 387-392.	3.2	23
69	Congenital amegakaryocytic thrombocytopenia: clinical and biological consequences of five novel mutations. Haematologica, 2007, 92, 1186-1193.	3.5	53
70	Why the disorder induced by GATA1 Arg216Gln mutation should be called â€œX-linked thrombocytopenia with thalassemiaâ€•rather than â€œX-linked gray platelet syndromeâ€• Blood, 2007, 110, 2770-2771.	1.4	13
71	A novel Leu153Ser mutation of the Fanconi anemia FANCD2 gene is associated with severe chemotherapy toxicity in a pediatric T-cell acute lymphoblastic leukemia. Leukemia, 2007, 21, 72-78.	7.2	21
72	Unexplained recurrent venous thrombosis in a patient withMYH9-related disease. Platelets, 2006, 17, 274-275.	2.3	17

#	Article	IF	Citations
73	Autosomal dominant thrombocytopenias with reduced expression of glycoprotein Ia. Thrombosis and Haemostasis, 2006, 95, 483-489.	3.4	26
74	Cord blood in vitro expanded CD41+ cells: identification of novel components of megakaryocytopoiesis. Journal of Thrombosis and Haemostasis, 2006, 4, 848-860.	3.8	23
75	Pathogenetic mechanisms of hematological abnormalities of patients with MYH9 mutations. Human Molecular Genetics, 2005, 14, 3169-3178.	2.9	52
76	Dissecting clinical findings: platelet defects segregate independently of deafness and cataract in a family affected by an apparent syndromic form of macrothrombocytopenia. International Journal of Molecular Medicine, 2005, 16, 437.	4.0	0
77	Effects of the R216Q mutation of GATA-1 on erythropoiesis and megakaryocytopoiesis. Thrombosis and Haemostasis, 2004, 91, 129-140.	3.4	105
78	Inherited Thrombocytopenias: Molecular Mechanisms. Seminars in Thrombosis and Hemostasis, 2004, 30, 513-523.	2.7	34
79	Spectrum ofPTCH mutations in Italian nevoid basal cell-carcinoma syndrome patients: Identification of thirteen novel alleles. Human Mutation, 2004, 24, 441-441.	2.5	20
80	Correlation between the clinical phenotype of MYH9 -related disease and tissue distribution of class II nonmuscle myosin heavy chains. Genomics, 2004, 83, 1125-1133.	2.9	69
81	Nonmuscle Myosin Heavy-Chain Gene MYH14 Is Expressed in Cochlea and Mutated in Patients Affected by Autosomal Dominant Hearing Impairment (DFNA4). American Journal of Human Genetics, 2004, 74, 770-776.	6.2	150
82	Application of a diagnostic algorithm for inherited thrombocytopenias to 46 consecutive patients. Haematologica, 2004, 89, 1219-25.	3.5	51
83	Genetics, clinical and pathological features of glomerulonephrites associated with mutations of nonmuscle myosin IIA (Fechtner syndrome). American Journal of Kidney Diseases, 2003, 41, 95-104.	1.9	94
84	Spectrum ofFANCA mutations in Italian Fanconi anemia patients: Identification of six novel alleles and phenotypic characterization of the S858R variant. Human Mutation, 2003, 22, 338-339.	2.5	35
85	Title is missing!. Medicine (United States), 2003, 82, 203-215.	1.0	30
86	MYH9-Related Disease. Medicine (United States), 2003, 82, 203-215.	1.0	255
87	Inherited thrombocytopenias: a proposed diagnostic algorithm from the Italian Gruppo di Studio delle Piastrine. Haematologica, 2003, 88, 582-92.	3.5	91
88	Cloning of the murine non-muscle myosin heavy chain IIA gene ortholog of human MYH9 responsible for May-Hegglin, Sebastian, Fechtner, and Epstein syndromes. Gene, 2002, 286, 215-222.	2.2	41
89	Epstein syndrome: another renal disorder with mutations in the nonmuscle myosin heavy chainÂ9 gene. Human Genetics, 2002, 110, 182-186.	3.8	45
90	Immunocytochemistry for the heavy chain of the non-muscle myosin IIA as a diagnostic tool forMYH9-related disorders. British Journal of Haematology, 2002, 117, 164-167.	2.5	47

#	Article	IF	Citations
91	Combined 17Â-Hydroxylase/17,20-Lyase Deficiency Caused by Phe93Cys Mutation in the CYP17 Gene. Journal of Clinical Endocrinology and Metabolism, 2002, 87, 898-905.	3.6	27
92	Inherited thrombocytopenias: from genes to therapy. Haematologica, 2002, 87, 860-80.	3.5	92
93	Defective expression of GPIb/IX/V complex in platelets from patients with May-Hegglin anomaly and Sebastian syndrome. Haematologica, 2002, 87, 943-7.	3.5	28
94	Molecular and Functional Analyses of the Human and Mouse Genes Encoding AFG3L1, a Mitochondrial Metalloprotease Homologous to the Human Spastic Paraplegia Protein. Genomics, 2001, 76, 58-65.	2.9	43
95	Autosomal dominant macrothrombocytopenia in Italy is most frequently a type of heterozygous Bernard-Soulier syndrome. Blood, 2001, 97, 1330-1335.	1.4	174
96	A new case of Acromegaloid Facial Appearance (AFA) syndrome with an expanded phenotype. Clinical Dysmorphology, 2000, 9, 221-222.	0.3	12
97	Mutations in MYH9 result in the May-Hegglin anomaly, and Fechtner and Sebastian syndromes. Nature Genetics, 2000, 26, 103-105.	21.4	397
98	The Gene for May-Hegglin Anomaly Localizes to a <1-Mb Region on Chromosome 22q12.3-13.1. American Journal of Human Genetics, 2000, 66, 1449-1454.	6.2	40
99	Nuclear Localization of the Fanconi Anemia Protein FANCC Is Required for Functional Activity. Blood, 1999, 93, 4025-4026.	1.4	20
100	Nevoid basal cell carcinoma syndrome. Clinical findings in 37 Italian affected individuals. Clinical Genetics, 1999, 55, 34-40.	2.0	143
101	Spontaneous functional correction of homozygous Fanconi anaemia alleles reveals novel mechanistic basis for reverse mosaicism. Nature Genetics, 1999, 22, 379-383.	21.4	190
102	Mutation analysis of the Fanconi anaemia A gene in breast tumours with loss of heterozygosity at 16q24.3. British Journal of Cancer, 1999, 79, 1049-1052.	6.4	15
103	An Autosomal Dominant Thrombocytopenia Gene Maps to Chromosomal Region 10p. American Journal of Human Genetics, 1999, 65, 1401-1405.	6.2	60
104	ThePISSLREGene: Structure, Exon Skipping, and Exclusion as Tumor Suppressor in Breast Cancer. Genomics, 1999, 56, 90-97.	2.9	39
105	Characterization of Copine VII, a New Member of the Copine Family, and Its Exclusion as a Candidate in Sporadic Breast Cancers with Loss of Heterozygosity at 16q24.3. Genomics, 1999, 61, 219-226.	2.9	20
106	Familial Dominant Thrombocytopenia: Clinical, Biologic, and Molecular Studies. Pediatric Research, 1999, 46, 548-548.	2.3	14
107	Mutational screening of thrombopoietin receptor gene (c-mpl) in patients with congenital thrombocytopenia and absent radii (TAR). British Journal of Haematology, 1998, 103, 311-314.	2.5	35
108	Construction of a High-Resolution Physical and Transcription Map of Chromosome 16q24.3: A Region of Frequent Loss of Heterozygosity in Sporadic Breast Cancer. Genomics, 1998, 50, 1-8.	2.9	28

#	Article	IF	Citations
109	Fine Exonâ€“Intron Structure of the Fanconi Anemia Group A (FAA) Gene and Characterization of Two Genomic Deletions. Genomics, 1998, 51, 463-467.	2.9	46
110	Characterization and Screening for Mutations of the Growth Arrest-Specific 11 (GAS11) andC16orf3Genes at 16q24.3 in Breast Cancer. Genomics, 1998, 52, 325-331.	2.9	47
111	Identification of the gene encoding the human mitochondrial RNA polymerase (h-mtRPOL) by cyberscreening of the Expressed Sequence Tags database. Human Molecular Genetics, 1997, 6, 615-625.	2.9	178
112	P53 Activates Fanconi Anemia Group C Gene Expression. Human Molecular Genetics, 1997, 6, 277-283.	2.9	23
113	The Genomic Organization of the Fanconi Anemia Group A (FAA) Gene. Genomics, 1997, 41, 309-314.	2.9	51
114	Mutations of the Fanconi Anemia Group A Gene (FAA) in Italian Patients. American Journal of Human Genetics, 1997, 61, 1246-1253.	6.2	55
115	Molecular characterization of Fanconi anaemia group C (FAC) gene polymorphisms. Molecular and Cellular Probes, 1996, 10, 213-218.	2.1	5
116	Fanconi anaemia in Italy: High prevalence of complementation group A in two geographic clusters. Human Genetics, 1996, 97, 599-603.	3.8	40
117	Identification of three novel mutations in the PIG-A gene in paroxysmal nocturnal haemoglobinuria (PNH) patients. Human Genetics, 1996, 97, 45-48.	3.8	21
118	Linkage analysis of Fanconi anaemia in Italy and mapping of the complementation group A gene. Human Genetics, 1996, 99, 93-97.	3.8	7
119	Expression cloning of a cDNA for the major Fanconi anaemia gene, FAA. Nature Genetics, 1996, 14, 320-323.	21.4	401
120	Positional cloning of the Fanconi anaemia group A gene. Nature Genetics, 1996, 14, 324-328.	21.4	294
121	Fanconi anaemia in Italy: high prevalence of complementation group A in two geographic clusters. Human Genetics, 1996, 97, 599-603.	3.8	4
122	Localisation of the Fanconi anaemia complementation group A gene to chromosome 16q24.3. Nature Genetics, 1995, 11, 338-340.	21.4	89
123	Characterization of the 5' region of the Fanconi anaemia group C (FACC) gene. Human Molecular Genetics, 1995, 4, 1321-1326.	2.9	13
124	Phosphatase inhibitors activate normal and defective CFTR chloride channels.. Proceedings of the National Academy of Sciences of the United States of America, 1994, 91, 9160-9164.	7.1	156
125	Haplotype analysis to determine the position of a mutation among closely linked DNA markers. Human Molecular Genetics, 1993, 2, 1007-1014.	2.9	19
126	EcoRI RFLP in the Fanconi anaemia complementation group C gene (FACC). Human Molecular Genetics, 1993, 2, 1509-1509.	2.9	2

#	Article	IF	Citations
127	The search for South European cystic fibrosis mutations: Identification of two new mutations, four variants, and intronic sequences. Genomics, 1991, 10, 193-200.	2.9	117
128	Polymorphic DNA haplotypes and Î”F508 deletion in 212 Italian CF families. Human Genetics, 1990, 85, 420-421.	3.8	12
129	Î” F508 gene deletion and prenatal diagnosis of cystic fibrosis in Italian and Spanish families. Prenatal Diagnosis, 1990, 10, 413-414.	2.3	8
130	Frequency Distribution of the Alleles of Several Variable Number of Tandem Repeat DNA Polymorphisms in the Italian Population. Human Heredity, 1990, 40, 61-68.	0.8	16
131	Linkage disequilibrium for DNA haplotypes near the cystic fibrosis locus in two South European populations. Human Genetics, 1989, 83, 175-178.	3.8	17
132	First-trimester prenatal diagnosis of cystic fibrosis using the polymerase chain reaction: Report of eight cases. Prenatal Diagnosis, 1989, 9, 349-355.	2.3	15