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	Expression cloning of a cDNA for the major Fanconi anaemia gene, FAA. Nature Genetics, 1996, 14, 320-323.	21.4	401
2	Mutations in MYH9 result in the May-Hegglin anomaly, and Fechtner and Sebastian syndromes. Nature Genetics, 2000, 26, 103-105.	21.4	397
3	Positional cloning of the Fanconi anaemia group A gene. Nature Genetics, 1996, 14, 324-328.	21.4	294
4	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
5	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
6	MYH9-Related Disease. Medicine (United States), 2003, 82, 203-215.	1.0	255
7	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
8	Recent advances in the understanding and management of MYH9â€related inherited thrombocytopenias. British Journal of Haematology, 2011, 154, 161-174.	2.5	196
9	Spontaneous functional correction of homozygous Fanconi anaemia alleles reveals novel mechanistic basis for reverse mosaicism. Nature Genetics, 1999, 22, 379-383.	21.4	190
10	MYH9: Structure, functions and role of non-muscle myosin IIA in human disease. Gene, 2018, 664, 152-167.	2.2	187
11	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
12	Autosomal dominant macrothrombocytopenia in Italy is most frequently a type of heterozygous Bernard-Soulier syndrome. Blood, 2001, 97, 1330-1335.	1.4	174
13	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
14	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
15	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
16	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
17	ANKRD26-related thrombocytopenia and myeloid malignancies. Blood, 2013, 122, 1987-1989.	1.4	145
18	Nevoid basal cell carcinoma syndrome. Clinical findings in 37 Italian affected individuals. Clinical Genetics, 1999, 55, 34-40.	2.0	143

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19	Eltrombopag for the treatment of the inherited thrombocytopenia deriving from MYH9 mutations. Blood, 2010, 116, 5832-5837.	1.4	141
20	Spectrum of the Mutations in Bernard-Soulier Syndrome. Human Mutation, 2014, 35, 1033-1045.	2.5	124
21	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
22	Platelet diameters in inherited thrombocytopenias: analysis of 376 patients with all known disorders. Blood, 2014, 124, e4-e10.	1.4	112
23	Effects of the R216Q mutation of GATA-1 on erythropoiesis and megakaryocytopoiesis. Thrombosis and Haemostasis, 2004, 91, 129-140.	3.4	105
24	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
25	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
26	Clinical and pathogenic features of ETV6 -related thrombocytopenia with predisposition to acute lymphoblastic leukemia. Haematologica, 2016, 101, 1333-1342.	3.5	92
27	Inherited thrombocytopenias: from genes to therapy. Haematologica, 2002, 87, 860-80.	3.5	92
28	Inherited thrombocytopenias: a proposed diagnostic algorithm from the Italian Gruppo di Studio delle Piastrine. Haematologica, 2003, 88, 582-92.	3.5	91
29	Clinical and genetic aspects of Bernard-Soulier syndrome: searching for genotype/phenotype correlations. Haematologica, 2011, 96, 417-423.	3.5	90
30	Localisation of the Fanconi anaemia complementation group A gene to chromosome 16q24.3. Nature Genetics, 1995, 11, 338-340.	21.4	89
31	Inherited thrombocytopenias frequently diagnosed in adults. Journal of Thrombosis and Haemostasis, 2013, 11, 1006-1019.	3.8	87
32	Genetics of familial forms of thrombocytopenia. Human Genetics, 2012, 131, 1821-1832.	3.8	85
33	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
34	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
35	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
36	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

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37	Analysis of 339 pregnancies in 181 women with 13 different forms of inherited thrombocytopenia. Haematologica, 2014, 99, 1387-1394.	3.5	63
38	An Autosomal Dominant Thrombocytopenia Gene Maps to Chromosomal Region 10p. American Journal of Human Genetics, 1999, 65, 1401-1405.	6.2	60
39	ACTN1-related thrombocytopenia: identification of novel families for phenotypic characterization. Blood, 2015, 125, 869-872.	1.4	57
40	Mutations of the Fanconi Anemia Group A Gene (FAA) in Italian Patients. American Journal of Human Genetics, 1997, 61, 1246-1253.	6.2	55
41	Congenital amegakaryocytic thrombocytopenia: clinical and biological consequences of five novel mutations. Haematologica, 2007, 92, 1186-1193.	3.5	53
42	Pathogenetic mechanisms of hematological abnormalities of patients with MYH9 mutations. Human Molecular Genetics, 2005, 14, 3169-3178.	2.9	52
43	The Genomic Organization of the Fanconi Anemia Group A (FAA) Gene. Genomics, 1997, 41, 309-314.	2.9	51
44	Application of a diagnostic algorithm for inherited thrombocytopenias to 46 consecutive patients. Haematologica, 2004, 89, 1219-25.	3.5	51
45	Correlation between platelet phenotype and NBEAL2 genotype in patients with congenital thrombocytopenia and Â-granule deficiency. Haematologica, 2013, 98, 868-874.	3.5	49
46	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
47	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
48	Thrombopoietin mutation in congenital amegakaryocytic thrombocytopenia treatable withÂromiplostim. EMBO Molecular Medicine, 2018, 10, 63-75.	6.9	47
49	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
50	Epstein syndrome: another renal disorder with mutations in the nonmuscle myosin heavy chainÂ9 gene. Human Genetics, 2002, 110, 182-186.	3.8	45
51	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
52	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
53	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
54	Fanconi anaemia in Italy: High prevalence of complementation group A in two geographic clusters. Human Genetics, 1996, 97, 599-603.	3.8	40

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55	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
56	Loss-of-function mutations in PTPRJ cause a new form of inherited thrombocytopenia. Blood, 2019, 133, 1346-1357.	1.4	40
57	ThePISSLREGene: Structure, Exon Skipping, and Exclusion as Tumor Suppressor in Breast Cancer. Genomics, 1999, 56, 90-97.	2.9	39
58	Evaluation of energy metabolism and calcium homeostasis in cells affected by Shwachman-Diamond syndrome. Scientific Reports, 2016, 6, 25441.	3.3	39
59	Alteration of Liver Enzymes Is a Feature of the Myh9-Related Disease Syndrome. PLoS ONE, 2012, 7, e35986.	2.5	38
60	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
61	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
62	Inherited Thrombocytopenias: Molecular Mechanisms. Seminars in Thrombosis and Hemostasis, 2004, 30, 513-523.	2.7	34
63	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 ETQq1 1 0.784314 rgBT /Overlock 10 Tf 50 412 T	4.1	33
64	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
65	Title is missing!. Medicine (United States), 2003, 82, 203-215.	1.0	30
66	MYH9 gene mutations associated with bleeding. Platelets, 2017, 28, 312-315.	2.3	29
67	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
68	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
69	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
70	Autosomal dominant thrombocytopenias with reduced expression of glycoprotein Ia. Thrombosis and Haemostasis, 2006, 95, 483-489.	3.4	26
71	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
72	Hypomorphic FANCA mutations correlate with mild mitochondrial and clinical phenotype in Fanconi anemia. Haematologica, 2018, 103, 417-426.	3.5	26

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73	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
74	P53 Activates Fanconi Anemia Group C Gene Expression. Human Molecular Genetics, 1997, 6, 277-283.	2.9	23
75	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
76	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
77	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
78	Molecular basis of inherited thrombocytopenias. Clinical Genetics, 2016, 89, 154-162.	2.0	22
79	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
80	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
81	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
82	Nuclear Localization of the Fanconi Anemia Protein FANCC Is Required for Functional Activity. Blood, 1999, 93, 4025-4026.	1.4	20
83	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
84	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
85	Haplotype analysis to determine the position of a mutation among closely linked DNA markers. Human Molecular Genetics, 1993, 2, 1007-1014.	2.9	19
86	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
87	Linkage disequilibrium for DNA haplotypes near the cystic fibrosis locus in two South European populations. Human Genetics, 1989, 83, 175-178.	3.8	17
88	Unexplained recurrent venous thrombosis in a patient withMYH9-related disease. Platelets, 2006, 17, 274-275.	2.3	17
89	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
90	Molecular basis of inherited thrombocytopenias: an update. Current Opinion in Hematology, 2016, 23, 486-492.	2.5	17

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91	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
92	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
93	Unusual splice site mutations disrupt FANCA exon 8 definition. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2014, 1842, 1052-1058.	3.8	16
94	<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
95	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
96	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
97	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
98	Mutations of RUNX1 in families with inherited thrombocytopenia. American Journal of Hematology, 2017, 92, E86-E88.	4.1	15
99	<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
100	Familial Dominant Thrombocytopenia: Clinical, Biologic, and Molecular Studies. Pediatric Research, 1999, 46, 548-548.	2.3	14
101	Characterization of the 5' region of the Fanconi anaemia group C (FACC) gene. Human Molecular Genetics, 1995, 4, 1321-1326.	2.9	13
102	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
103	Polymorphic DNA haplotypes and Î”F508 deletion in 212 Italian CF families. Human Genetics, 1990, 85, 420-421.	3.8	12
104	A new case of Acromegaloid Facial Appearance (AFA) syndrome with an expanded phenotype. Clinical Dysmorphology, 2000, 9, 221-222.	0.3	12
105	Gray platelet syndrome: Novel mutations of the NBEAL2 gene. American Journal of Hematology, 2017, 92, E20-E22.	4.1	12
106	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
107	MYH9related disease: A novel missense Ala95Asp mutation of theMYH9gene. Platelets, 2009, 20, 598-602.	2.3	10
108	Molecular diagnosis of thrombocytopeniaâ€absent radius syndrome using nextâ€generation sequencing. International Journal of Laboratory Hematology, 2016, 38, 412-418.	1.3	10

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109	Two further patients with Warsaw breakage syndrome. Is a mild phenotype possible?. Molecular Genetics & Genomic Medicine, 2019, 7, e639.	1.2	10
110	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
111	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
112	Î” F508 gene deletion and prenatal diagnosis of cystic fibrosis in Italian and Spanish families. Prenatal Diagnosis, 1990, 10, 413-414.	2.3	8
113	MYH9-related disease: Report on five German families and description of a novel mutation. Annals of Hematology, 2010, 89, 1057-1059.	1.8	8
114	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
115	Linkage analysis of Fanconi anaemia in Italy and mapping of the complementation group A gene. Human Genetics, 1996, 99, 93-97.	3.8	7
116	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
117	Fanconi Anemia Patients Are More Susceptible to Infection with Tumor Virus SV40. PLoS ONE, 2013, 8, e79683.	2.5	6
118	Molecular characterization of Fanconi anaemia group C (FAC) gene polymorphisms. Molecular and Cellular Probes, 1996, 10, 213-218.	2.1	5
119	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
120	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
121	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
122	MYH9-related Disorders. Journal of Pediatric Hematology/Oncology, 2012, 34, 412-415.	0.6	4
123	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
124	Fanconi anaemia in Italy: high prevalence of complementation group A in two geographic clusters. Human Genetics, 1996, 97, 599-603.	3.8	4
125	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
126	ETV6-related thrombocytopenia: dominant negative effect of mutations as common pathogenic mechanism. Haematologica, 2022, 107, 2249-2254.	3.5	3

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127	EcoRI RFLP in the Fanconi anaemia complementation group C gene (FACC). Human Molecular Genetics, 1993, 2, 1509-1509.	2.9	2
128	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
129	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
130	Dysregulation of oncogenic factors by GFI1B p32: investigation of a novel <I>GFI1B</I> germline mutation. Haematologica, 2022, 107, 260-267.	3.5	1
131	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
132	Chronic Thrombocytopenia in Children: What Could It Hide?. Blood, 2020, 136, 33-34.	1.4	0