Anne C Goodeve

List of Publications by Citations

130 papers	4,796 citations	34 h-index	67 g-index
135 ext. papers	5,221 ext. citations	5.1 avg, IF	5.24 L-index

#	Paper	IF	Citations
130	Phenotype and genotype of a cohort of families historically diagnosed with type 1 von Willebrand disease in the European study, Molecular and Clinical Markers for the Diagnosis and Management of Type 1 von Willebrand Disease (MCMDM-1VWD). *Blood*, **2007**, 109, 112-21	2.2	309
129	Identification of novel FLT-3 Asp835 mutations in adult acute myeloid leukaemia. *British Journal of Haematology*, **2001**, 113, 983-8	4.5	303
128	Incidence and prognosis of c-KIT and FLT3 mutations in core binding factor (CBF) acute myeloid leukaemias. *British Journal of Haematology*, **2003**, 121, 775-7	4.5	253
127	FLT3 internal tandem duplication mutations in adult acute myeloid leukaemia define a high-risk group. *British Journal of Haematology*, **2000**, 111, 190-5	4.5	227
126	The diagnosis and management of von Willebrand disease: a United Kingdom Haemophilia Centre Doctors Organization guideline approved by the British Committee for Standards in Haematology. *British Journal of Haematology*, **2014**, 167, 453-65	4.5	222
125	c-kit proto-oncogene exon 8 in-frame deletion plus insertion mutations in acute myeloid leukaemia. *British Journal of Haematology*, **1999**, 105, 894-900	4.5	201
124	Response to desmopressin is influenced by the genotype and phenotype in type 1 von Willebrand disease (VWD): results from the European Study MCMDM-1VWD. *Blood*, **2008**, 111, 3531-9	2.2	162
123	Principles of care for the diagnosis and treatment of von Willebrand disease. *Haematologica*, **2013**, 98, 667-74	6.6	139
122	A high-throughput sequencing test for diagnosing inherited bleeding, thrombotic, and platelet disorders. *Blood*, **2016**, 127, 2791-803	2.2	135
121	HLA Class II Profile: A Weak Determinant of Factor VIII Inhibitor Development in Severe Haemophilia A. *Thrombosis and Haemostasis*, **1997**, 77, 234-237	7	129
120	Somatic mosaicism in hemophilia A: a fairly common event. *American Journal of Human Genetics*, **2001**, 69, 75-87	11	125
119	Identification of type 1 von Willebrand disease patients with reduced von Willebrand factor survival by assay of the VWF propeptide in the European study: molecular and clinical markers for the diagnosis and management of type 1 VWD (MCMDM-1VWD). *Blood*, **2008**, 111, 4979-85	2.2	123
118	The genetic basis of von Willebrand disease. *Blood Reviews*, **2010**, 24, 123-34	11.1	114
117	The molecular basis of hemophilia A: genotype-phenotype relationships and inhibitor development. *Seminars in Thrombosis and Hemostasis*, **2003**, 29, 23-30	5.3	100
116	Precise Carrier Diagnosis in Families with Haemophilia A: Use of Conformation Sensitive Gel Electrophoresis for Mutation Screening and Polymorphism Analysis. *Thrombosis and Haemostasis*, **1998**, 79, 723-726	7	98
115	Clinical and laboratory variability in a cohort of patients diagnosed with type 1 VWD in the United States. *Blood*, **2016**, 127, 2481-8	2.2	76
114	Aberrant methylation of the negative regulators RASSFIA, SHP-1 and SOCS-1 in myelodysplastic syndromes and acute myeloid leukaemia. *British Journal of Haematology*, **2005**, 129, 60-5	4.5	71

113	VWF propeptide and ratios between VWF, VWF propeptide, and FVIII in the characterization of type 1 von Willebrand disease. *Blood*, **2013**, 121, 2336-9	2.2	69
112	Methylation of the suppressor of cytokine signaling 3 gene (SOCS3) in myeloproliferative disorders. *Haematologica*, **2008**, 93, 1635-44	6.6	68
111	Analysis of intracellular storage and regulated secretion of 3 von Willebrand disease-causing variants of von Willebrand factor. *Blood*, **2003**, 102, 2452-8	2.2	67
110	The international society on thrombosis and haematosis von Willebrand disease database: an update. *Seminars in Thrombosis and Hemostasis*, **2011**, 37, 470-9	5.3	61
109	A study of Wilson disease mutations in Britain. *Human Mutation*, **1999**, 14, 304-11	4.7	61
108	von Willebrand disease. *Genetics in Medicine*, **2011**, 13, 365-76	8.1	60
107	Genomic structure of human FLT3: implications for mutational analysis. *British Journal of Haematology*, **2001**, 113, 1076-7	4.5	60
106	Identification and characterization of a novel P2Y 12 variant in a patient diagnosed with type 1 von Willebrand disease in the European MCMDM-1VWD study. *Blood*, **2009**, 113, 4110-3	2.2	59
105	Two novel type 2N von Willebrand disease-causing mutations that result in defective factor VIII binding, multimerization, and secretion of von Willebrand factor. *Blood*, **2000**, 95, 2000-2007	2.2	57
104	Hemophilia B: molecular pathogenesis and mutation analysis. *Journal of Thrombosis and Haemostasis*, **2015**, 13, 1184-95	15.4	56
103	Relationship between Factor VIII Mutation Type and Inhibitor Development in a Cohort of Previously Untreated Patients Treated with Recombinant Factor VIII (Recombinate). *Thrombosis and Haemostasis*, **2000**, 83, 844-848	7	51
102	Late Relapsing Childhood Lymphoblastic Leukemia. *Blood*, **1998**, 92, 2334-2337	2.2	51
101	Factor VIII gene rearrangements in patients with severe haemophilia A. *Lancet, The*, **1994**, 343, 329-30	40	51
100	A novel von Willebrand disease-causing mutation (Arg273Trp) in the von Willebrand factor propeptide that results in defective multimerization and secretion. *Blood*, **2000**, 96, 560-568	2.2	49
99	A Standard Nomenclature for von Willebrand Factor Gene Mutations and Polymorphisms. *Thrombosis and Haemostasis*, **2001**, 85, 929-931	7	43
98	Specific and global coagulation assays in the diagnosis of discrepant mild hemophilia A. *Haematologica*, **2013**, 98, 1980-7	6.6	35
97	A rapid method for haemophilia B mutation detection using conformation sensitive gel electrophoresis. *British Journal of Haematology*, **1999**, 104, 915-8	4.5	34
96	HLA class II profile: a weak determinant of factor VIII inhibitor development in severe haemophilia A. UKHCDO Inhibitor Working Party. *Thrombosis and Haemostasis*, **1997**, 77, 234-7	7	34

95	Second hit mutations in the RTK/RAS signaling pathway in acute myeloid leukemia with inv(16). *Haematologica*, **2004**, 89, 106	6.6	34
94	IIb variants defined by next-generation sequencing: predicting variants likely to cause Glanzmann thrombasthenia. *Proceedings of the National Academy of Sciences of the United States of America*, **2015**, 112, E1898-907	11.5	33
93	The impact of bleeding history, von Willebrand factor and PFA-100( ) on the diagnosis of type 1 von Willebrand disease: results from the European study MCMDM-1VWD. *British Journal of Haematology*, **2010**, 151, 245-51	4.5	32
92	Tribbles-1 and -2 are tumour suppressors, down-regulated in human acute myeloid leukaemia. *Immunology Letters*, **2010**, 130, 115-24	4.1	32
91	Homeologous recombination between AluSx-sequences as a cause of hemophilia. *Human Mutation*, **2004**, 24, 440	4.7	32
90	Curated disease-causing genes for bleeding, thrombotic, and platelet disorders: Communication from the SSC of the ISTH. *Journal of Thrombosis and Haemostasis*, **2019**, 17, 1253-1260	15.4	31
89	BCR-ABL Transcript With an e19a2 (c3a2) Junction in Classical Chronic Myeloid Leukemia. *Blood*, **1997**, 89, 3064-3064	2.2	31
88	Nomenclature of genetic variants in hemostasis. *Journal of Thrombosis and Haemostasis*, **2011**, 9, 852-5	15.4	29
87	Are aberrant BCR--ABL transcripts more common than previously thought?. *British Journal of Haematology*, **2000**, 111, 1109-11	4.5	28
86	The incidence of inhibitor development according to specific mutations--and treatment?. *Blood Coagulation and Fibrinolysis*, **2003**, 14 Suppl 1, S17-21	1	27
85	Mutational analysis of class III receptor tyrosine kinases (C-KIT, C-FMS, FLT3) in idiopathic myelofibrosis. *British Journal of Haematology*, **2003**, 120, 464-70	4.5	27
84	Assessment of the F9 genotype-specific FIX inhibitor risks and characterisation of 10 novel severe F9 defects in the first molecular series of Argentinian patients with haemophilia B. *Thrombosis and Haemostasis*, **2013**, 109, 24-33	7	26
83	Characterisation of Type 2N von Willebrand Disease Using Phenotypic and Molecular Techniques. *Thrombosis and Haemostasis*, **1996**, 75, 959-964	7	26
82	JAK2 V617F Mutation is uncommon in chronic myelomonocytic leukaemia. *British Journal of Haematology*, **2005**, 130, 968	4.5	25
81	Genetic testing for von Willebrand disease: the case for. *Journal of Thrombosis and Haemostasis*, **2010**, 8, 13-6	15.4	24
80	Gene structure, expression profiling and mutation analysis of the tumour suppressor SHIP1 in Caucasian acute myeloid leukaemia. *Leukemia*, **2007**, 21, 2390-3	10.7	24
79	Sixteen novel hemophilia A causative mutations in the first Argentinian series of severe molecular defects. *Haematologica*, **2007**, 92, 842-5	6.6	24
78	Endoplasmic reticulum retention and prolonged association of a von Willebrand's disease-causing von Willebrand factor variant with ERp57 and calnexin. *Biochemical and Biophysical Research Communications*, **2001**, 280, 448-53	3.4	24

77	Evaluation of a semi-automated von Willebrand factor multimer assay, the Hydragel 5 von Willebrand multimer, by two European Centers. *Research and Practice in Thrombosis and Haemostasis*, **2018**, 2, 790-799	5.1	23
76	Laboratory methods for the genetic diagnosis of bleeding disorders. *International Journal of Laboratory Hematology*, **1998**, 20, 3-19		21
75	Type 1 von Willebrand disease: application of emerging data to clinical practice. *Haemophilia*, **2008**, 14, 685-96	3.3	19
74	The UK National External Quality Assessment Scheme (UK NEQAS) for molecular genetic testing in haemophilia. *Thrombosis and Haemostasis*, **2006**, 96, 597-601	7	19
73	Mutations in PTPN11 are uncommon in adult myelodysplastic syndromes and acute myeloid leukaemia. *British Journal of Haematology*, **2004**, 124, 843-4	4.5	19
72	Molecular genetic testing of hemophilia A. *Seminars in Thrombosis and Hemostasis*, **2008**, 34, 491-501	5.3	18
71	A novel von Willebrand disease-causing mutation (Arg273Trp) in the von Willebrand factor propeptide that results in defective multimerization and secretion. *Blood*, **2000**, 96, 560-8	2.2	18
70	p.Tyr365Cys change in factor VIII: haemophilia A, but not as we know it. *British Journal of Haematology*, **2011**, 154, 618-25	4.5	16
69	Quality in molecular biology testing for inherited thrombophilia disorders. *Seminars in Thrombosis and Hemostasis*, **2012**, 38, 600-12	5.3	16
68	Clonal stability in late-relapsing childhood lymphoblastic leukaemia. *British Journal of Haematology*, **1997**, 98, 992-4	4.5	15
67	A novel von Willebrand disease□causing mutation (Arg273Trp) in the von Willebrand factor propeptide that results in defective multimerization and secretion. *Blood*, **2000**, 96, 560-568	2.2	15
66	Analysis of factor VIII gene intron 1 inversion in Argentinian families with severe haemophilia A and a review of the literature. *Blood Coagulation and Fibrinolysis*, **2004**, 15, 569-572	1	15
65	Diagnosing von Willebrand disease: genetic analysis. *Hematology American Society of Hematology Education Program*, **2016**, 2016, 678-682	3.1	15
64	c-FMS mutational analysis in acute myeloid leukaemia. *British Journal of Haematology*, **2003**, 123, 749-50	4.5	14
63	Next Generation Sequencing in Newborn Screening in the United Kingdom National Health Service. *International Journal of Neonatal Screening*, **2019**, 5, 40	2.6	14
62	Polymorphic variation within the VWF gene contributes to the failure to detect mutations in patients historically diagnosed with type 1 von Willebrand disease from the MCMDM-1VWD cohort. *Haematologica*, **2010**, 95, 2163-5	6.6	13
61	A rapid and cost effective method for analysis of dinucleotide repeat polymorphisms in the factor VIII gene. *Blood Coagulation and Fibrinolysis*, **1996**, 7, 672-7	1	13
60	Mutational analysis of the von Willebrand factor gene in type 1 von Willebrand disease using conformation sensitive gel electrophoresis: a comparison of fluorescent and manual techniques. *Haematologica*, **2007**, 92, 550-3	6.6	13

59	Precise carrier diagnosis in families with haemophilia A: use of conformation sensitive gel electrophoresis for mutation screening and polymorphism analysis. *Thrombosis and Haemostasis*, **1998**, 79, 723-6	7	13
58	Identification and characterisation of mutations associated with von Willebrand disease in a Turkish patient cohort. *Thrombosis and Haemostasis*, **2013**, 110, 264-74	7	12
57	Advances in carrier detection in haemophilia. *Haemophilia*, **1998**, 4, 358-64	3.3	12
56	The common single nucleotide variants c.2365A>G and c.2385T>C modify VWF biosynthesis and clearance. *Blood Advances*, **2018**, 2, 1585-1594	7.8	12
55	Diagnosis and management of von Willebrand disease in the United Kingdom. *Seminars in Thrombosis and Hemostasis*, **2011**, 37, 488-94	5.3	11
54	Haemophilia A and von Willebrand's disease. *Haemophilia*, **2010**, 16 Suppl 5, 79-84	3.3	10
53	Reassortants of influenza B viruses for use in vaccines: an evaluation. *Archives of Virology*, **1985**, 83, 169-79	2.6	10
52	Bleeding symptoms in patients diagnosed as type 3 von Willebrand disease: Results from 3WINTERS-IPS, an international and collaborative cross-sectional study. *Journal of Thrombosis and Haemostasis*, **2020**, 18, 2145-2154	15.4	9
51	Genomics of bleeding disorders. *Haemophilia*, **2014**, 20 Suppl 4, 50-3	3.3	9
50	Genetics of type 1 von Willebrand disease. *Current Opinion in Hematology*, **2007**, 14, 444-9	3.3	9
49	FLT3 internal tandem duplication mutations are rare in agnogenic myeloid metaplasia. *Blood*, **2002**, 100, 364	2.2	9
48	A comparison of the allelic frequencies of ten DNA polymorphisms associated with factor VIII and factor IX genes in Thai and Western European populations. *Blood Coagulation and Fibrinolysis*, **1994**, 5, 29-35	1	9
47	Relationship between factor VIII mutation type and inhibitor development in a cohort of previously untreated patients treated with recombinant factor VIII (Recombinate). Recombinate PUP Study Group. *Thrombosis and Haemostasis*, **2000**, 83, 844-8	7	9
46	The molecular basis of von Willebrand disease: the under investigated, the unexpected and the overlooked. *Haematologica*, **2011**, 96, 798-800	6.6	8
45	Two novel type 2N von Willebrand disease-causing mutations that result in defective factor VIII binding, multimerization, and secretion of von Willebrand factor. *Blood*, **2000**, 95, 2000-7	2.2	8
44	FLT3 internal tandem duplication mutations in adult acute myeloid leukaemia define a high-risk group. *British Journal of Haematology*, **2000**, 111, 190-195	4.5	7
43	A standard nomenclature for von Willebrand factor gene mutations and polymorphisms. On behalf of the ISTH SSC Subcommittee on von Willebrand factor. *Thrombosis and Haemostasis*, **2001**, 85, 929-31	7	7
42	The UK National External Quality Assessment Scheme for heritable bleeding disorders. *Seminars in Thrombosis and Hemostasis*, **2014**, 40, 261-8	5.3	6

41	Is routine molecular screening for common alpha-thalassaemia deletions necessary as part of an antenatal screening programme?. *Journal of Medical Screening*, **2007**, 14, 60-1	1.4	6
40	Null Alleles Are not a Common Cause of Type 1 von Willebrand Disease in the British Population. *Thrombosis and Haemostasis*, **1999**, 82, 1373-1375	7	6
39	Genetics of haemostasis. *Haemophilia*, **2012**, 18 Suppl 4, 73-80	3.3	5
38	Activating loop mutations in the PDGFR alpha and beta genes are rare in core binding factor acute myeloid leukaemia. *British Journal of Haematology*, **2004**, 127, 123-4	4.5	5
37	A standard nomenclature for von Willebrand factor gene mutations and polymorphisms. *Best Practice and Research in Clinical Haematology*, **2001**, 14, 235-40	4.2	5
36	The Complete Type I VWD Cohort Of The Zimmerman Program For The Molecular and Clinical Biology Of VWD - Phenotypic Assignment, Mutation Frequency, and Bleeding Assessment. *Blood*, **2013**, 122, 332-332	2.2	5
35	Identification and Characterization of Novel Variations in Platelet G-Protein Coupled Receptor (GPCR) Genes in Patients Historically Diagnosed with Type 1 von Willebrand Disease. *PLoS ONE*, **2015**, 10, e0143913	3.7	5
34	The UK National External Quality Assessment Scheme (UK NEQAS) for molecular genetic testing in haemophilia. *Thrombosis and Haemostasis*, **2006**, 96, 597-601	7	5
33	p.P2063S: a neutral VWF variant masquerading as a mutation. *Annals of Hematology*, **2014**, 93, 505-6	3	3
32	VWF sequence variants: a data goldmine. *Blood*, **2013**, 122, 471-3	2.2	3
31	A second case of Hb Renert [beta133(H11)Val --> Ala]. *Hemoglobin*, **2001**, 25, 337-40	0.6	3
30	Characterization of large in-frame von Willebrand factor deletions highlights differing pathogenic mechanisms. *Blood Advances*, **2020**, 4, 2979-2990	7.8	2
29	When 1 plus 1 equals 3 in VWD. *Blood*, **2009**, 114, 933-4	2.2	2
28	Vicenza deciphered: modeling the von Willebrand disease enigma: commentary on accelerated clearance alone explains ultralarge multimers in VWD Vicenza. *Journal of Thrombosis and Haemostasis*, **2010**, 8, 1271-2	15.4	2
27	Re-evaluation of three Israeli families initially diagnosed with type 1 von Willebrand disease in light of the ISTH update on von Willebrand factor pathophysiology and classification. *Haemophilia*, **2008**, 14, 621-4	3.3	2
26	Molecular Diagnosis of von Willebrand Disease: The Genotype114-126		2
25	A common splice site mutation is shared by two families with different type 2N von Willebrand disease mutations. *Thrombosis and Haemostasis*, **1999**, 82, 1061-4	7	2
24	Genetic analysis of bleeding disorders. *Haemophilia*, **2016**, 22 Suppl 5, 79-83	3.3	1

23	von Willebrand Disease: Molecular Aspects **2010**, 278-285		1
22	Intracellular Retention, Enhanced Clearance, and Defective FVIII Binding Are Common Features of Von Willebrand Factor D'-D3 Domain Mutations in Patients with Von Willebrand Disease Type 1 From the European Mcmdm-1VWD Study. *Blood*, **2012**, 120, 99-99	2.2	1
21	Characterisation of type 2N von Willebrand disease using phenotypic and molecular techniques. *Thrombosis and Haemostasis*, **1996**, 75, 959-64	7	1
20	Authors' response to Dong-Zhi Li's letter: Routine molecular screening for common -Thalassaemia deletions is necessary as part of an antenatal screening programme*Journal of Medical Screening*, **2008**, 15, 47-47	1.4	0
19	Are aberrant BCRABL transcripts more common than previously thought?. *British Journal of Haematology*, **2000**, 111, 1109-1111	4.5	0
18	Genotypes of European and Iranian patients with type 3 von Willebrand disease enrolled in 3WINTERS-IPS. *Blood Advances*, **2021**, 5, 2987-3001	7.8	0
17	von Willebrand Disease: Molecular Aspects **2014**, 353-361		
16	Another step towards understanding hemophilia A molecular pathogenesis. *Journal of Thrombosis and Haemostasis*, **2010**, 8, 2693-5	15.4	
15	Enhanced carrier and prenatal diagnosis in the Italian haemophilia B population. *Blood Transfusion*, **2007**, 5, 117-9	3.6	
14	Comparison of Phenotypic Assessment and Mutation Detection in the Diagnosis of Carrier State in Hemophilia: Identification of 10 Novel Mutations.. *Blood*, **2004**, 104, 4020-4020	2.2	
13	Promoter Methylation of RASSFIA, SHP1 and SOCS1 Genes in Acute Myeloid Leukaemia (AML) and Myelodysplastic Syndromes (MDS).. *Blood*, **2004**, 104, 2999-2999	2.2	
12	Investigation of Underlying Reasons of Factor VIII Deficiency in Haemophilia A Patients with Undetectable Mutations in the Factor VIII Gene.. *Blood*, **2006**, 108, 1042-1042	2.2	
11	Profile of Mutations Identified in the 3WINTERS-IPS Project on European & Iranian Patients with Previously Diagnosed Type 3 Von Willebrand Disease.. *Blood*, **2018**, 132, 1184-1184	2.2	
10	Clustering of Bleeding Symptoms in Patients Previously Diagnosed As Type 3 Von Willebrand Disease: Results from a Large Cohort of Type 3 Von Willebrand Disease (the 3Winters-Ips Project). *Blood*, **2018**, 132, 2465-2465	2.2	
9	von Willebrand Factor/Factor VIII Binding Is not Affected by the Arg89Gln Polymorphism in von Willebrand Factor. *Thrombosis and Haemostasis*, **1996**, 76, 820-821	7	
8	Late Relapsing Childhood Lymphoblastic Leukemia. *Blood*, **1998**, 92, 2334-2337	2.2	
7	Characterization and Expression of an In-Frame Exon 33-34 Deletion Causing Type 1 VWD. *Blood*, **2014**, 124, 1525-1525	2.2	
6	Identification of Novel Mutations with Molecular Modelling of Missense Mutations of Congenital Afibrinogenemia Patients in Pakistan. *Blood*, **2015**, 126, 4664-4664	2.2	

List of Publications

5	Investigation of the Role of Copy Number Variation In the Pathogenesis of Type 1 Von Willebrand Disease. *Blood*, **2010**, 116, 2218-2218	2.2
4	Laboratory Analysis of Von Willebrand Disease: Molecular Analysis204-215	
3	In silico analysis highlights the copy number variation mechanism responsible for the historically reported VWF exon 42 deletion. *Haemophilia*, **2016**, 22, e484-7	3.3
2	Genetic and Laboratory Diagnosis **2018**, 153-166	
1	Phenotypic and Genotypic Signatures of VWF Exon 18 in Eastern Saudi Patients Previously Diagnosed with Type 1 von Willebrand Disease. *International Journal of General Medicine*,Volume 15, 5385-5394	2.3