

CITATION REPORT

List of articles citing

Somatic mosaicism in hemophilia A: a fairly common event

DOI: 10.1086/321285

American Journal of Human Genetics, 2001, 69, 75-87.

Source: <https://exaly.com/paper-pdf/32393214/citation-report.pdf>

Version: 2024-04-27

This report has been generated based on the citations recorded by exaly.com for the above article. For the latest version of this publication list, visit the link given above.

The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

#	Paper	IF	Citations
135	Human germline mutation in the factor IX gene. 2001 , 487, 1-17		39
134	Post-zygotic mutations and somatic mosaicism in androgen insensitivity syndrome. 2001 , 17, 628		1
133	Possibility of somatic mosaicism of ELA2 mutation overlooked in an asymptomatic father transmitting severe congenital neutropenia to two offspring. 2002 , 118, 923; author reply 923-4		6
132	Somatic mosaicism for a MECP2 mutation associated with classic Rett syndrome in a boy. 2002 , 10, 77-81		81
131	Paternal mosaicism proves the pathogenic nature of mutations in neutrophil elastase in severe congenital neutropenia. 2002 , 100, 707-9		54
130	11 Hemophilia A Patients without Mutations in the Factor VIII Encoding Gene. <i>Thrombosis and Haemostasis</i> , 2002 , 88, 357-360	7	40
129	High throughput mutation screening of the factor VIII gene (F8C) in hemophilia A: 37 novel mutations and genotype-phenotype correlation. 2002 , 20, 267-74		33
128	Time for a redefinition of type 2m von willebrand disease. 2002 , 118, 922-922		
127	Polyclonal hypergammaglobulinaemia with hyperviscosity syndrome. 2002 , 118, 922-922		
126	Possibility of somatic mosaicism of ELA2 mutation overlooked in an asymptomatic father transmitting severe congenital neutropenia to two offspring. 2002 , 118, 923-923		1
125	Reply to benson and horwitz. 2002 , 118, 923-924		6
124	The use of vitamin k for reversal of over-warfarinization in children. 2002 , 118, 924-925		7
123	Reversal of coumarin-induced over-anticoagulation. 2002 , 118, 925-926		5
122	Reply to escobar. 2002 , 118, 926-926		1
121	Regarding the loss of cd20 after rituximab therapy. 2002 , 118, 927-927		
120	Consequences at mrna level of the pklr gene splicing mutations IVS10(+1)G->C and IVS8(+2)T->G causing pyruvate kinase deficiency. 2002 , 118, 927-928		
119	Somatic and germinal mosaicism for the steroid sulfatase gene deletion in a steroid sulfatase deficiency carrier. 2002 , 119, 972-5		10

118	Origin of amnion and implications for evaluation of the fetal genotype in cases of mosaicism. 2002 , 22, 1076-85		45
117	Mechanisms and consequences of somatic mosaicism in humans. 2002 , 3, 748-58		280
116	Genotype and phenotype of haemophilia A in Thai patients. <i>Haemophilia</i> , 2003 , 9, 179-86	3-3	7
115	Exclusion of mosaicism in Spanish haemophilia A families with inversion of intron 22. <i>Haemophilia</i> , 2003 , 9, 584-7	3-3	5
114	Thirty-four novel mutations detected in factor VIII gene by multiplex CSGE: modeling of 13 novel amino acid substitutions. 2003 , 1, 773-81		37
113	Somatic mosaicism and cancer: inference based on a conditional Luria-Delbrück distribution. 2003 , 223, 405-12		20
112	Somatic gene mutation and human disease other than cancer. 2003 , 543, 125-36		82
111	Genetic counseling of hemophilia carriers. 2003 , 29, 31-6		12
110	Recurrent familial hypocalcemia due to germline mosaicism for an activating mutation of the calcium-sensing receptor gene. 2003 , 88, 3674-81		39
109	Neutrophil elastase mutations in congenital neutropenia. 2003 , 8, 165-71		13
108	2% Hämophilie-A-Patienten ohne Mutation im FVIII-Gen. 2003 , 23, 1-5		8
107	Recurrent third-trimester fetal loss and maternal mosaicism for long-QT syndrome. 2004 , 109, 3029-34		73
106	Somatic mosaicism is rare in unaffected parents of patients with sporadic tuberous sclerosis. 2004 , 41, e69		21
105	Molecular basis of haemophilia A. <i>Haemophilia</i> , 2004 , 10 Suppl 4, 133-9	3-3	80
104	Germline mosaicism complicates molecular diagnosis of Lesch-Nyhan syndrome. 2004 , 24, 737-40		5
103	Genetic analysis of haemophilia A in Bulgaria. 2004 , 4, 2		3
102	Germline mosaicism resulting in the transmission of severe hemophilia B from a grandfather with a mild deficiency. 2004 , 129A, 13-5		16
101	Autoimmune lymphoproliferative syndrome with somatic Fas mutations. 2004 , 351, 1409-18		224

100	The molecular analysis of haemophilia A: a guideline from the UK haemophilia centre doctors organization haemophilia genetics laboratory network. <i>Haemophilia</i> , 2005 , 11, 387-97	3-3	51
99	First description of somatic mosaicism in MYH9 disorders. 2005 , 128, 360-5		21
98	Spectrum of molecular defects and mutation detection rate in patients with severe hemophilia A. 2005 , 26, 249-54		36
97	The fundamental theorem of neutral evolution: rates of substitution and mutation should factor in premeiotic clusters. 2005 , 125, 333-9		4
96	Heterozygous mutations of OTX2 cause severe ocular malformations. <i>American Journal of Human Genetics</i> , 2005 , 76, 1008-22	11	237
95	The origin of EFN1 mutations in craniofrontonasal syndrome: frequent somatic mosaicism and explanation of the paucity of carrier males. <i>American Journal of Human Genetics</i> , 2006 , 78, 999-1010	11	82
94	Genetic diagnosis of haemophilia and other inherited bleeding disorders. <i>Haemophilia</i> , 2006 , 12 Suppl 3, 82-9	3-3	102
93	Somatic APC mosaicism: an underestimated cause of polyposis coli. 2008 , 57, 71-6		111
92	Somatic APC mosaicism: a frequent cause of familial adenomatous polyposis (FAP). 2007 , 28, 985-92		108
91	Recurrent inversion with concomitant deletion and insertion events in the coagulation factor VIII gene suggests a new mechanism for X-chromosomal rearrangements causing hemophilia A. 2007 , 28, 1045		18
90	The molecular aetiology of haemophilia A in a New Zealand patient group. <i>Haemophilia</i> , 2007 , 13, 420-7	3-3	13
89	New efficient extragenic microsatellite markers for hemophilia a carrier state diagnostics. 2007 , 43, 689-696		
88	Genetik und Klinik der Hämophilie A und B. 2008 , 20, 190-196		
87	Somatic mosaicism for a PDHA1 mutation in a female with pyruvate dehydrogenase deficiency. 2008 , 124, 187-93		18
86	Familial CHARGE syndrome and the CHD7 gene: a recurrent missense mutation, intrafamilial recurrence and variability. <i>American Journal of Medical Genetics, Part A</i> , 2008 , 146A, 43-50	2-5	68
85	Heterozygous large deletions of Factor 8 gene in females identified by multiplex PCR-LC. <i>Haemophilia</i> , 2008 , 14, 599-606	3-3	8
84	Importance of investigating somatic and germline mutations in hemophilia A: a preliminary study from All India Institute of Medical Sciences, India. 2008 , 389, 103-8		7
83	Identification of 31 novel mutations in the F8 gene in Spanish hemophilia A patients: structural analysis of 20 missense mutations suggests new intermolecular binding sites. 2008 , 111, 3468-78		13

82	INHERITED BLEEDING DISORDERS IN PREGNANCY. 2009 , 20, 205-227		0
81	Detection of mosaic RB1 mutations in families with retinoblastoma. 2009 , 30, 842-51		122
80	Mosaics and haemophilia. <i>Haemophilia</i> , 2009 , 15, 1181-6	3.3	25
79	Improved criterion-referenced assessment in indirect tracking of haemophilia A using a 0.23 cM-resolution dense polymorphic marker set. <i>Haemophilia</i> , 2009 , 15, 1135-42	3.3	6
78	Understanding what determines the frequency and pattern of human germline mutations. 2009 , 10, 478-88		90
77	Recurrence risk due to germ line mosaicism: Duchenne and Becker muscular dystrophy. 2009 , 75, 465-72		60
76	Mosaicism in men in hemophilia: is it exceptional? Impact on genetic counselling. 2009 , 7, 367-9		11
75	Molecular mechanisms underlying hemophilia A phenotype in seven females. 2009 , 7, 976-82		44
74	Molecular diagnostics in hemostatic disorders. 2009 , 29, 367-90		4
73	Carrier analysis for hemophilia A: ideal versus acceptable. 2009 , 9, 203-7		4
72	Ultra deep sequencing detects a low rate of mosaic mutations in tuberous sclerosis complex. 2010 , 127, 573-82		61
71	Somatic gene mutation and human disease other than cancer: an update. 2010 , 705, 96-106		132
70	Somatic mosaicism in Menkes disease suggests choroid plexus-mediated copper transport to the developing brain. <i>American Journal of Medical Genetics, Part A</i> , 2010 , 152A, 2529-34	2.5	33
69	Somatic mosaicism in a case of apparently sporadic Creutzfeldt-Jakob disease carrying a de novo D178N mutation in the PRNP gene. 2010 , 153B, 1283-91		27
68	Preimplantation genetic diagnosis for hemophilia A using indirect linkage analysis and direct genotyping approaches. 2010 , 8, 783-9		28
67	Retraction. Mosaicism and haemophilia. by C.K. Kasper and C.H. Buzin. <i>Haemophilia</i> , 2010 , 16, 972	3.3	
66	Genetic counseling and pre-natal diagnosis in hemophilia. 194-200		
65	Occurrence of haemophilia A and B in a Chinese family with mosaicism of the F9 gene mutation in the HB index maternal grandfather. <i>Thrombosis and Haemostasis</i> , 2010 , 103, 1106-8	7	11

64	Mendelian Inheritance. 2010 , 22-65		
63	Hemophilia A and B. 2010 , 61-72		
62	Genotyping the factor VIII intron 22 inversion locus using fluorescent in situ hybridization. 2011 , 46, 151-8		0
61	X-linked adrenoleukodystrophy: ABCD1 de novo mutations and mosaicism. 2011 , 104, 160-6		36
60	Maternal de novo triple mosaicism for two single OCRL nucleotide substitutions (c.1736A>T, c.1736A>G) in a Lowe syndrome family. 2011 , 129, 513-9		17
59	Genotyping of intron 22-related rearrangements of F8 by inverse-shifting PCR in Egyptian hemophilia A patients. 2011 , 90, 579-84		18
58	Ready, Steady, Go! The Current State of Carriership Status Determination and Prenatal Diagnosis of Haemophilia a in Bulgaria. 2011 , 25, 2566-2571		1
57	Role of molecular genetics in hemophilia: from diagnosis to therapy. 2012 , 38, 64-78		21
56	On a Break with the X: The Role of Repair of Double-Stranded DNA Breaks in X-Linked Disease. 2012 , 26, 2829-2837		
55	In silico profiling of deleterious amino acid substitutions of potential pathological importance in haemophilia A and haemophilia B. 2012 , 19, 30		15
54	Hemophilia A. 2012 , 1005-1012		
53	Genotype-Phenotype Interaction Analyses in Hemophilia. 2012 ,		0
52	A novel de novo missense mutation in TP63 underlying germline mosaicism in AEC syndrome: implications for recurrence risk and prenatal diagnosis. <i>American Journal of Medical Genetics, Part A</i> , 2012 , 158A, 1957-61	2.5	15
51	The status of carrier and prenatal diagnosis of haemophilia in China. <i>Haemophilia</i> , 2012 , 18, 235-40	3.3	13
50	F8 gene dosage defects in atypical patients with severe haemophilia A. <i>Haemophilia</i> , 2012 , 18, 708-13	3.3	2
49	Hemophilias and Other Disorders of Hemostasis. 2013 , 1-33		9
48	X-Chromosome Inactivation. 2013 , 63-88		1
47	F8 genetic analysis strategies when standard approaches fail. 2014 , 34, 167-73		14

46	Historical review on genetic analysis in hemophilia A. 2014 , 40, 895-902		20
45	Factor VIII therapy for hemophilia A: current and future issues. 2014 , 7, 373-85		24
44	Parent of origin, mosaicism, and recurrence risk: probabilistic modeling explains the broken symmetry of transmission genetics. <i>American Journal of Human Genetics</i> , 2014 , 95, 345-59	11	76
43	Paternal Somatic Mosaicism of a Novel Frameshift Mutation in ELANE Causing Severe Congenital Neutropenia. 2015 , 62, 2229-31		7
42	Maternal low-level somatic mosaicism of Cys155Tyr of F9 in severe hemophilia B. 2015 , 26, 866-8		7
41	Combined coagulation factor VIII and factor IX deficiency (CDF8F9) in a patient from Lithuania. 2016 , 36, S29-S33		3
40	Identification of mutations in the F8 and F9 gene in families with haemophilia using targeted high-throughput sequencing. <i>Haemophilia</i> , 2016 , 22, e427-34	3.3	6
39	Origin of mutation in sporadic cases of severe haemophilia A in Sweden. 2016 , 90, 63-8		18
38	Hemophilia A and B. 2016 , 79-93		1
37	AccuCopy quantification combined with pre-amplification of long-distance PCR for fast analysis of intron 22 inversion in haemophilia A. 2016 , 458, 78-83		5
36	Novel calcium-sensing receptor cytoplasmic tail deletion mutation causing autosomal dominant hypocalcemia: molecular and clinical study. 2016 , 174, K1-K11		5
35	The second Team Haemophilia Education Meeting, 2016, Frankfurt, Germany. <i>European Journal of Haematology</i> , 2017 , 98 Suppl 85, 1-15	3.8	2
34	Somatic mosaicism in a severe haemophilia B family detected by allele specific PCR: An alert to the genetic diagnostic laboratories. <i>Thrombosis Research</i> , 2017 , 158, 138-139	8.2	1
33	Panel-based whole exome sequencing identifies novel mutations in microphthalmia and anophthalmia patients showing complex Mendelian inheritance patterns. <i>Molecular Genetics & Genomic Medicine</i> , 2017 , 5, 709-719	2.3	21
32	Type 2 Gaucher disease in an infant despite a normal maternal glucocerebrosidase gene. <i>American Journal of Medical Genetics, Part A</i> , 2017 , 173, 3211-3215	2.5	4
31	Hemophilia A. 2017 , 1319-1329		
30	Complex recombination with deletion in the F8 and duplication in the TMLHE mediated by int22h copies during early embryogenesis. <i>Thrombosis and Haemostasis</i> , 2017 , 117, 1478-1485	7	4
29	Molecular Diagnostics for Coagulopathies. 2017 , 221-233		

28	Genetic Counseling and Prenatal Diagnosis of Congenital Bleeding Disorders. 255-264		
27	Somatic/Germinal Mosaicism of a F8 Promoter Deletion Confounds Clinical Predictions in a Family with Haemophilia A: Key Role of Genotype Quantitation. <i>Thrombosis and Haemostasis</i> , 2018 , 118, 617-620	3.7	1
26	Spectrum and origin of mutations in sporadic cases of haemophilia A in China. <i>Haemophilia</i> , 2018 , 24, 291-298	3.3	12
25	RET somatic mutations are underrecognized in Hirschsprung disease. <i>Genetics in Medicine</i> , 2018 , 20, 770-777	4.7	20
24	Carriers of Hemophilia A and Hemophilia B. 2018 , 65-82		
23	The molecular basis of hemophilia. 2019 , 221-234		
22	The characteristics and spectrum of F9 mutations in Chinese sporadic haemophilia B pedigrees. <i>Haemophilia</i> , 2019 , 25, 316-323	3.3	2
21	Genomic mosaicism: A neglected factor that promotes variability in asthma diagnosis. <i>Medical Hypotheses</i> , 2019 , 127, 112-115	3.8	
20	Unexpected relevant role of gene mosaicism in patients with primary immunodeficiency diseases. <i>Journal of Allergy and Clinical Immunology</i> , 2019 , 143, 359-368	11.5	29
19	Genetic analysis for carrier diagnosis in hemophilia A and B in the Mexican population: 25 years of experience. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2020 , 184, 939-954	3.1	1
18	Detection of mosaics in hemophilia A by deep Ion Torrent sequencing and droplet digital PCR. <i>Research and Practice in Thrombosis and Haemostasis</i> , 2020 , 4, 1121-1130	5.1	0
17	Identification of the Intron 22 and Intron 1 Inversions of the Factor VIII Gene in Iraqi Kurdish Patients With Hemophilia A. <i>Clinical and Applied Thrombosis/Hemostasis</i> , 2020 , 26, 1076029619888293	3.3	2
16	Genetic mosaicism in haemophilia: A practical review to help evaluate the risk of transmitting the disease. <i>Haemophilia</i> , 2020 , 26, 375-383	3.3	7
15	Women with Hemophilia: Case Series of Reproductive Choices and Review of Literature. <i>TH Open</i> , 2021 , 5, e183-e187	2.7	2
14	Haemophilia. <i>Nature Reviews Disease Primers</i> , 2021 , 7, 45	51.1	10
13	related Congenital Stationary Night Blindness in Two Siblings due to Probable Maternal Germline Mosaicism. <i>Ophthalmic Genetics</i> , 2021 , 42, 588-592	1.2	0
12	Identification of F8 rearrangements in carrier and non-carrier mothers of haemophilia A patients. <i>Haemophilia</i> , 2021 , 27, e654-e658	3.3	
11	Genomic and Epigenetic Complexity of the FOXF1 Locus in 16q24.1: Implications for Development and Disease. <i>Current Genomics</i> , 2015 , 16, 107-16	2.6	41

10 Genetic Inheritance. **2004**, 31-56

9 Hemophilia.

8 Mosaicism and haemophilia. *Haemophilia*, **2009**,

3-3

7 Other Issues in Hemophilia Care. 97-104

6 Hemophilia A. **2016**, 1-11

5 Hemophilia A. **2006**, 476-481

4 Spectrum and tissue distribution of RB1 pathogenic alleles in mosaic retinoblastoma patients. 1-11

3 Disorders of Hemostasis and Thrombosis. **2023**, 173-211

2 Case Report: Novel pathogenic variant in NF1X in two sisters with Malan syndrome due to germline mosaicism. 13,

○

1 Insights into the Molecular Genetic of Hemophilia A and Hemophilia B: The Relevance of Genetic Testing in Routine Clinical Practice. **2022**, 42, 390-399

○