Maria E De La Morena-Barrio

List of Publications by Year
in descending order

73	917	567144 15	580701 25
papers	citations	h-index	g-index

81	81	81	1063
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	International clinical guidelines for the management of phosphomannomutase 2â€congenital disorders of glycosylation: Diagnosis, treatment and follow up. Journal of Inherited Metabolic Disease, 2019, 42, 5-28.	1.7	91
2	The genetics of antithrombin. Thrombosis Research, 2018, 169, 23-29.	0.8	77
3	Multirefractory primary immune thrombocytopenia; targeting the decreased sialic acid content. Platelets, 2019, 30, 743-751.	1.1	45
4	Hypoglycosylation is a common finding in antithrombin deficiency in the absence of a SERPINC1 gene defect. Journal of Thrombosis and Haemostasis, 2016, 14, 1549-1560.	1.9	38
5	Amelioration of the severity of heparin-binding antithrombin mutations by posttranslational mosaicism. Blood, 2012, 120, 900-904.	0.6	37
6	Prognostic value of thrombin generation parameters in hospitalized COVID-19 patients. Scientific Reports, 2021, 11, 7792.	1.6	28
7	Antithrombin Dublin (p.Val30Glu): a relatively common variant with moderate thrombosis risk of causing transient antithrombin deficiency. Thrombosis and Haemostasis, 2016, 116, 146-154.	1.8	27
8	Regulatory regions of SERPINC1 gene: Identification of the first mutation associated with antithrombin deficiency. Thrombosis and Haemostasis, 2012, 107, 430-437.	1.8	26
9	High incidence of <scp>FXI</scp> deficiency in a Spanish town caused by 11 different mutations and the first duplication of F11: Results from the Yecla study. Haemophilia, 2017, 23, e488-e496.	1.0	22
10	Subcellular localization of antizyme inhibitor 2 in mammalian cells: Influence of intrinsic sequences and interaction with antizymes. Journal of Cellular Biochemistry, 2009, 107, 732-740.	1.2	21
11	Incidence and features of thrombosis in children with inherited antithrombin deficiency. Haematologica, 2019, 104, 2512-2518.	1.7	21
12	High levels of latent antithrombin in plasma from patients with antithrombin deficiency. Thrombosis and Haemostasis, 2017, 117, 880-888.	1.8	20
13	Elevated thrombin generation in patients with congenital disorder of glycosylation and combined coagulation factor deficiencies. Journal of Thrombosis and Haemostasis, 2019, 17, 1798-1807.	1.9	18
14	Genetic predisposition to fetal alcohol syndrome: association with congenital disorders of N-glycosylation. Pediatric Research, 2018, 83, 119-127.	1.1	17
15	A human antithrombin isoform dampens inflammatory responses and protects from organ damage during bacterial infection. Nature Microbiology, 2019, 4, 2442-2455.	5.9	17
16	A pilot study on the impact of congenital thrombophilia in COVIDâ€19. European Journal of Clinical Investigation, 2021, 51, e13546.	1.7	16
17	Proteomic analysis of platelet N-glycoproteins in PMM2-CDG patients. Thrombosis Research, 2014, 133, 412-417.	0.8	15
18	Neutrophil extracellular traps and von Willebrand factor are allies that negatively influence COVIDâ€19 outcomes. Clinical and Translational Medicine, 2021, 11, e268.	1.7	15

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19	Protective role of antithrombin in mouse models of liver injury. Journal of Hepatology, 2012, 57, 980-986.	1.8	14
20	The Infective Polymerization of Conformationally Unstable Antithrombin Mutants May Play a Role in the Clinical Severity of Antithrombin Deficiency. Molecular Medicine, 2012, 18, 762-770.	1.9	14
21	Congenital disorder of glycosylation (PMM2-CDG) in a patient with antithrombin deficiency and severe thrombophilia. Journal of Thrombosis and Haemostasis, 2012, 10, 2625-2627.	1.9	13
22	GPI-anchor and GPI-anchored protein expression in PMM2-CDG patients. Orphanet Journal of Rare Diseases, 2013, 8, 170.	1.2	13
23	Identification of Regulatory Mutations in SERPINC1 Affecting Vitamin D Response Elements Associated with Antithrombin Deficiency. PLoS ONE, 2016, 11, e0152159.	1.1	12
24	Assessment of two contact activation reagents for the diagnosis of congenital factor XI deficiency. Thrombosis Research, 2018, 163, 64-70.	0.8	12
25	MPI-CDG with transient hypoglycosylation and antithrombin deficiency. Haematologica, 2019, 104, e79-e82.	1.7	12
26	ALG12â€CDG: An unusual patient without intellectual disability and facial dysmorphism, and with a novel variant. Molecular Genetics & Genomic Medicine, 2020, 8, e1304.	0.6	12
27	Expanding the genetic spectrum of TUBB1-related thrombocytopenia. Blood Advances, 2021, 5, 5453-5467.	2.5	12
28	Heparin affinity of factor VIIa: Implications on the physiological inhibition by antithrombin and clearance of recombinant factor VIIa. Thrombosis Research, 2011, 127, 154-160.	0.8	11
29	Two SERPINC1 variants affecting N-glycosylation of Asn224 cause severe thrombophilia not detected by functional assays. Blood, 2022, 140, 140-151.	0.6	11
30	Control of post-translational modifications in antithrombin during murine post-natal development by miR-200a. Journal of Biomedical Science, 2013, 20, 29.	2.6	10
31	First case with antithrombin deficiency, mesenteric vein thrombosis and pregnancy: Multidisciplinary diagnosis and successful management. Thrombosis Research, 2016, 144, 72-75.	0.8	10
32	Defects of splicing in antithrombin deficiency. Research and Practice in Thrombosis and Haemostasis, 2017, 1, 216-222.	1.0	10
33	Congenital antithrombin deficiency in patients with splanchnic vein thrombosis. Liver International, 2020, 40, 1168-1177.	1.9	10
34	N-Glycosylation as a Tool to Study Antithrombin Secretion, Conformation, and Function. International Journal of Molecular Sciences, 2021, 22, 516.	1.8	10
35	Molecular and clinical characterization of transient antithrombin deficiency: A new concept in congenital thrombophilia. American Journal of Hematology, 2022, 97, 216-225.	2.0	10
36	The FXII c.-4T>C Polymorphism as a Disease Modifier in Patients With Hereditary Angioedema Due to the FXII p.Thr328Lys Variant. Frontiers in Genetics, 2020, 11, 1033.	1.1	9

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37	Factor XII in PMM2-CDG patients: role of N-glycosylation in the secretion and function of the first element of the contact pathway. Orphanet Journal of Rare Diseases, 2020, 15, 280.	1.2	9
38	Identification of Antithrombin-Modulating Genes. Role of LARGE, a Gene Encoding a Bifunctional Glycosyltransferase, in the Secretion of Proteins?. PLoS ONE, 2013, 8, e64998.	1.1	9
39	Natural Killer Cell Receptors and Cytotoxic Activity in Phosphomannomutase 2 Deficiency (PMM2-CDG). PLoS ONE, 2016, 11, e0158863.	1.1	9
40	Long-Read Sequencing Identifies the First Retrotransposon Insertion and Resolves Structural Variants Causing Antithrombin Deficiency. Thrombosis and Haemostasis, 2022, 122, 1369-1378.	1.8	9
41	Role of the Câ€sheet in the maturation of Nâ€glycans on antithrombin: functional relevance of pleiotropic mutations. Journal of Thrombosis and Haemostasis, 2014, 12, 1131-1140.	1.9	8
42	Genotypeâ€“phenotype gradient of SERPINC1 variants in a single family reveals a severe compound antithrombin deficiency in a dead embryo. British Journal of Haematology, 2020, 191, e32-e35.	1.2	8
43	A new method to quantify Î²-antithrombin glycoform in plasma reveals increased levels during the acute stroke event. Thrombosis Research, 2015, 136, 634-641.	0.8	7
44	High penetrance of inferior vena cava system atresia in severe thrombophilia caused by homozygous antithrombin Budapest 3 variant: Description of a new syndrome. American Journal of Hematology, 2021, 96, 1363-1373.	2.0	7
45	Molecular Dissection of Structural Variations Involved in Antithrombin Deficiency. Journal of Molecular Diagnostics, 2022, 24, 462-475.	1.2	7
46	The induction of cardiac ornithine decarboxylase by Î²₂â€adrenergic agents is associated with calcium channels and phosphorylation of ERK1/2. Journal of Cellular Biochemistry, 2013, 114, 1978-1986.	1.2	6
47	Uniparental disomy causes deficiencies of vitamin Kâ€dependent proteins. Journal of Thrombosis and Haemostasis, 2016, 14, 2410-2418.	1.9	6
48	Lost in translation: bioinformatic analysis of variations affecting the translation initiation codon in the human genome. Bioinformatics, 2018, 34, 3788-3794.	1.8	6
49	Identification of the first large intronic deletion responsible of type I antithrombin deficiency not detected by routine molecular diagnostic methods. British Journal of Haematology, 2019, 186, e82-e86.	1.2	6
50	Archeogenetics of F11 p.Cys38Arg: a 5400-year-old mutation identified in different southwestern European countries. Blood, 2019, 133, 2618-2622.	0.6	6
51	Antithrombin p.Thr147Ala: The First Founder Mutation in People of African Origin Responsible for Inherited Antithrombin Deficiency. Thrombosis and Haemostasis, 2021, 121, 182-191.	1.8	6
52	Gynaecological and obstetrical bleeding in Caucasian women with congenital factor XI deficiency: Results from a twenty-year, retrospective, observational study. Medicina ClÃnica, 2019, 153, 373-379.	0.3	6
53	Identification of a New Mechanism of Antithrombin Deficiency Hardly Detected by Current Methods: Duplication of SERPINC1 Exon 6. Thrombosis and Haemostasis, 2018, 118, 939-941.	1.8	5
54	A series of 10 Polish patients with thromboembolic events and antithrombin deficiency. Blood Coagulation and Fibrinolysis, 2019, 30, 193-198.	0.5	5

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55	Transient desialylation in combination with a novel antithrombin deficiency causing a severe and recurrent thrombosis despite anticoagulation therapy. Scientific Reports, 2017, 7, 44556.	1.6	4
56	Recurrent mutations in aSERPINC1hotspot associate with venous thrombosis without apparent antithrombin deficiency. Oncotarget, 2017, 8, 84417-84425.	0.8	4
57	Dissecting the transcriptional program of phosphomannomutase 2-deficient cells: Lymphoblastoide B cell lines as a valuable model for congenital disorders of glycosylation studies. Glycobiology, 2022, 32, 84-100.	1.3	3
58	Antithrombin deficiency as a still underdiagnosed thrombophilia: a primer for internists. Polish Archives of Internal Medicine, 2020, 130, 868-877.	0.3	3
59	Biochemical and cellular consequences of the antithrombin p.Met1? mutation identified in a severe thrombophilic family. Oncotarget, 2018, 9, 33202-33214.	0.8	3
60	Compound heterozygosity involving Antithrombin Cambridge II (p.Ala416Ser) in antithrombin deficiency. Thrombosis and Haemostasis, 2013, 109, 556-558.	1.8	2
61	Thrombin in the Activation of the Fluid Contact Phase in Patients with Hereditary Angioedema Carrying the F12 P.Thr309Lys Variant. Clinical Reviews in Allergy and Immunology, 2021, 60, 357-368.	2.9	2
62	A novel genetic variant in <scp>PTGS1</scp> affects Nâ€glycosylation of cyclooxygenaseâ€1 causing a dominantâ€negative effect on platelet function and bleeding diathesis. American Journal of Hematology, 2021, 96, E83-E88.	2.0	2
63	C0229 Regulatory regions of SERPINC1 gene: Identification of the first mutation associated with antithrombin deficiency. Thrombosis Research, 2012, 130, S118.	0.8	1
64	Anticoagulant therapy in patients with congenital FXI deficiency. Blood Advances, 2021, 5, 4083-4086.	2.5	1
65	C0094 High incidence of compound heterozygosity in antithrombin deficiency: Diagnostic and prognostic relevance. Thrombosis Research, 2012, 130, S117.	0.8	0
66	C0260: Identification of RXRA/VDR Elements Involved in the Regulation of Antithrombin Levels. Relevance in Antithrombin Deficiency. Thrombosis Research, 2014, 133, S10.	0.8	0
67	C0229: Mutations Creating New Glycosylation Sites Cause Different Types of Antithrombin Deficiency. Role of an N-Glycan at the Reactive Centre Loop. Thrombosis Research, 2014, 133, S15.	0.8	0
68	Angel: Towards a Multi-level Method for the Analysis of Variants in Individual Genomes. Lecture Notes in Computer Science, 2016, , 47-58.	1.0	0
69	Predominant mutations in a hotspot of SERPINC1 associated with venous thromboembolism in the Chinese population: a case-control study. Lancet, The, 2016, 388, S39.	6.3	0
70	Gynaecological and obstetrical bleeding in Caucasian women with congenital factor XI deficiency: Results from a twenty-year, retrospective, observational study. Medicina ClÃnica (English Edition), 2019, 153, 373-379.	0.1	0
71	Commentary on Acquired Factor XI Deficiency during SARS-CoV-2 Infection: Not Only Thrombosis. TH Open, 2020, 04, e231-e232.	0.7	0
72	When genetic and surname analyses meet historical sources: The C56R mutation associated with factor XI deficiency as a marker of human migration during the Spanish Reconquista. Medical Hypotheses, 2020, 141, 109709.	0.8	0

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73	Characterizing Coagulation FVII from iPSC-Hepatocytes-like Cells: Setting the Basis for Cell Therapy Development. Blood, 2020, 136, 4-4.	0.6	0