Marco Ritelli

List of Publications by Year
in descending order

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84	1,899	236612	329751
papers	citations	25 h-index	37 g-index

84	84	84	2628
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Clinical and molecular characterization of 40 patients with classic Ehlersâ€“Danlos syndrome: identification of 18 COL5A1 and 2 COL5A2 novel mutations. Orphanet Journal of Rare Diseases, 2013, 8, 58.	1.2	101
2	Differential diagnosis and diagnostic flow chart of joint hypermobility syndrome/ehlersâ€danlos syndrome hypermobility type compared to other heritable connective tissue disorders. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2015, 169, 6-22.	0.7	91
3	Gynecologic and obstetric implications of the joint hypermobility syndrome (a.k.a. Ehlersâ€“Danlos) Tj ETQq1 1 0.784314 rgBT /Overlo 158A, 2176-2182.	0.7	78
4	Nosology and inheritance pattern(s) of joint hypermobility syndrome and Ehlersâ€Danlos syndrome, hypermobility type: A study of intrafamilial and interfamilial variability in 23 Italian pedigrees. American Journal of Medical Genetics, Part A, 2014, 164, 3010-3020.	0.7	70
5	Thoracic Aortic Aneurysm in Infancy in Aneurysmsâ€“<scp>O</scp>steoarthritis Syndrome Due to a Novel <scp>SMAD</scp>3 Mutation: Further Delineation of the Phenotype. American Journal of Medical Genetics, Part A, 2013, 161, 1028-1035.	0.7	58
6	Obesity and the Risk of Intracerebral Hemorrhage. Stroke, 2013, 44, 1584-1589.	1.0	46
7	Cellular and Molecular Mechanisms in the Pathogenesis of Classical, Vascular, and Hypermobile Ehlersâ€'Danlos Syndromes. Genes, 2019, 10, 609.	1.0	46
8	Complications of Acute Stroke and the Occurrence of Early Seizures. Cerebrovascular Diseases, 2013, 35, 444-450.	0.8	45
9	Chronic mucocutaneous candidiasis and connective tissue disorder in humans with impaired JNK1-dependent responses to IL-17A/F and TGF-Î². Science Immunology, 2019, 4, .	5.6	45
10	Connective tissue anomalies in patients with spontaneous cervical artery dissection. Neurology, 2014, 83, 2032-2037.	1.5	42
11	GLUT10 deficiency leads to oxidative stress and non-canonical Î±vÎ²3 integrin-mediated TGFÎ² signalling associated with extracellular matrix disarray in arterial tortuosity syndrome skin fibroblasts. Human Molecular Genetics, 2015, 24, 6769-6787.	1.4	42
12	Transcriptome-Wide Expression Profiling in Skin Fibroblasts of Patients with Joint Hypermobility Syndrome/Ehlers-Danlos Syndrome Hypermobility Type. PLoS ONE, 2016, 11, e0161347.	1.1	40
13	Arterial Tortuosity Syndrome: homozygosity for two novel and one recurrent SLC2A10missense mutations in three families with severe cardiopulmonary complications in infancy and a literature review. BMC Medical Genetics, 2014, 15, 122.	2.1	36
14	Delineation of Ehlersâ€“Danlos syndrome phenotype due to the c.934C>T, p.(Arg312Cys) mutation in COL1A1: Report on a threeâ€generation family without cardiovascular events, and literature review. American Journal of Medical Genetics, Part A, 2017, 173, 524-530.	0.7	35
15	Dermal fibroblast-to-myofibroblast transition sustained by Î±vÃŸ3 integrin-ILK-Snail1/Slug signaling is a common feature for hypermobile Ehlers-Danlos syndrome and hypermobility spectrum disorders. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2018, 1864, 1010-1023.	1.8	34
16	Serum cholesterol levels, HMG-CoA reductase inhibitors and the risk of intracerebral haemorrhage. The Multicenter Study on Cerebral Haemorrhage in Italy (MUCH-Italy). Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, 924-929.	0.9	33
17	A Novel Homozygous <i>SLC2A9</i> Mutation Associated with Renal-Induced Hypouricemia. American Journal of Nephrology, 2016, 43, 245-250.	1.4	33
18	Antithrombotic medications and the etiology of intracerebral hemorrhage. Neurology, 2014, 82, 529-535.	1.5	32

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19	Spontaneous coronary artery dissection in a young woman with Loeysâ€“Dietz syndrome. American Journal of Medical Genetics, Part A, 2012, 158A, 1216-1218.	0.7	31
20	Expanding the clinical and mutational spectrum of B4GALT7-spondylodysplastic Ehlers-Danlos syndrome. Orphanet Journal of Rare Diseases, 2017, 12, 153.	1.2	31
21	Transcriptome analysis of skin fibroblasts with dominant negative COL3A1 mutations provides molecular insights into the etiopathology of vascular Ehlers-Danlos syndrome. PLoS ONE, 2018, 13, e0191220.	1.1	31
22	Spectrum of mucocutaneous manifestations in 277 patients with joint hypermobility syndrome/Ehlersâ€Danlos syndrome, hypermobility type. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2015, 169, 43-53.	0.7	30
23	Further delineation of Loeys-Dietz syndrome type 4 in a family with mild vascular involvement and a TGFB2 splicing mutation. BMC Medical Genetics, 2014, 15, 91.	2.1	29
24	Molecular Genetics and Pathogenesis of Ehlersâ€“Danlos Syndrome and Related Connective Tissue Disorders. Genes, 2020, 11, 547.	1.0	29
25	Insights in the etiopathology of galactosyltransferase II (GalT-II) deficiency from transcriptome-wide expression profiling of skin fibroblasts of two sisters with compound heterozygosity for two novel B3GALT6 mutations. Molecular Genetics and Metabolism Reports, 2015, 2, 1-15.	0.4	27
26	COL1â€related overlap disorder: A novel connective tissue disorder incorporating the osteogenesis imperfecta/Ehlersâ€Danlos syndrome overlap. Clinical Genetics, 2020, 97, 396-406.	1.0	27
27	Loeys-Dietz syndrome type I and type II: clinical findings and novel mutations in two Italian patients. Orphanet Journal of Rare Diseases, 2009, 4, 24.	1.2	26
28	Arterial tortuosity in patients with spontaneous cervical artery dissection. Neuroradiology, 2017, 59, 571-575.	1.1	26
29	Adult presentation of arterial tortuosity syndrome in a 51â€yearâ€old woman with a novel homozygous c.1411+1G>A mutation in the SLC2A10 gene. American Journal of Medical Genetics, Part A, 2012, 158A, 1164-1169.	0.7	25
30	Glucose transporter type 10â€”lacking in arterial tortuosity syndromeâ€”facilitates dehydroascorbic acid transport. FEBS Letters, 2016, 590, 1630-1640.	1.3	25
31	COL6A5variants in familial neuropathic chronic itch. Brain, 2017, 140, aww343.	3.7	25
32	Recurrent exercise-induced acute renal failure in a young Pakistani man with severe renal hypouricemia and SLC2A9compound heterozygosity. BMC Medical Genetics, 2014, 15, 3.	2.1	24
33	Multifaced Roles of the Î±vÎ²3 Integrin in Ehlersâ€“Danlos and Arterial Tortuosity Syndromesâ€™ Dermal Fibroblasts. International Journal of Molecular Sciences, 2018, 19, 982.	1.8	24
34	Further Defining the Phenotypic Spectrum of B3GAT3 Mutations and Literature Review on Linkeropathy Syndromes. Genes, 2019, 10, 631.	1.0	23
35	Expanding the Clinical and Mutational Spectrum of Recessive AEBP1-Related Classical-Like Ehlers-Danlos Syndrome. Genes, 2019, 10, 135.	1.0	23
36	GARFIELD-NGS: Genomic vARiants FIltering by dEep Learning moDels in NGS. Bioinformatics, 2018, 34, 3038-3040.	1.8	22

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37	Molecular insights in the pathogenesis of classical Ehlers-Danlos syndrome from transcriptome-wide expression profiling of patientsâ€™ skin fibroblasts. PLoS ONE, 2019, 14, e0211647.	1.1	22
38	Arterial tortuosity syndrome in two Italian paediatric patients. Orphanet Journal of Rare Diseases, 2009, 4, 20.	1.2	21
39	Recurring and Generalized Visceroptosis in Ehlersâ€“<scp>D</scp>anlos Syndrome Hypermobility Type. American Journal of Medical Genetics, Part A, 2013, 161, 1143-1147.	0.7	21
40	Mutations in TGFBR2 gene cause spontaneous cervical artery dissection. Journal of Neurology, Neurosurgery and Psychiatry, 2011, 82, 1372-1374.	0.9	20
41	Anticoagulants Resumption after Warfarin-Related Intracerebral Haemorrhage: The Multicenter Study on Cerebral Hemorrhage in Italy (MUCH-Italy). Thrombosis and Haemostasis, 2018, 118, 572-580.	1.8	20
42	Alcohol intake and the risk of intracerebral hemorrhage in the elderly. Neurology, 2018, 91, e227-e235.	1.5	20
43	Genotypic Categorization of Loeys-Dietz Syndrome Based on 24 Novel Families and Literature Data. Genes, 2019, 10, 764.	1.0	20
44	Multisystemic manifestations in a cohort of 75 classical Ehlers-Danlos syndrome patients: natural history and nosological perspectives. Orphanet Journal of Rare Diseases, 2020, 15, 197.	1.2	20
45	Type III and V collagens modulate the expression and assembly of EDA+ fibronectin in the extracellular matrix of defective Ehlersâ€“Danlos syndrome fibroblasts. Biochimica Et Biophysica Acta - General Subjects, 2012, 1820, 1576-1587.	1.1	18
46	Palmoplantar keratoderma, pseudoâ€ainhum, and universal atrichia: A new patient and review of the palmoplantar keratodermaâ€“congenital alopecia syndrome. American Journal of Medical Genetics, Part A, 2010, 152A, 2043-2047.	0.7	17
47	Effects of IFN-Î± on the Inflammatory Response of Swine Leukocytes to Bacterial Endotoxin. Journal of Interferon and Cytokine Research, 2005, 25, 202-208.	0.5	16
48	Diagnosis of vascular Ehlers-Danlos syndrome in Italy: Clinical findings and novel COL3A1 mutations. Journal of Dermatological Science, 2011, 64, 237-240.	1.0	16
49	Late diagnosis of lateral meningocele syndrome in a 55â€yearâ€old woman with symptoms of joint instability and chronic musculoskeletal pain. American Journal of Medical Genetics, Part A, 2014, 164, 528-534.	0.7	16
50	Clinical and Molecular Characterization of Classical-Like Ehlers-Danlos Syndrome Due to a Novel TNXB Variant. Genes, 2019, 10, 843.	1.0	16
51	GLUT10â€”Lacking in Arterial Tortuosity Syndromeâ€”Is Localized to the Endoplasmic Reticulum of Human Fibroblasts. International Journal of Molecular Sciences, 2017, 18, 1820.	1.8	15
52	Further delineation of FKBP14â€related Ehlersâ€“Danlos syndrome: A patient with early vascular complications and nonâ€progressive kyphoscoliosis, and literature review. American Journal of Medical Genetics, Part A, 2016, 170, 2031-2038.	0.7	14
53	A novel MAP3K7 splice mutation causes cardiospondylocarpofacial syndrome with features of hereditary connective tissue disorder. European Journal of Human Genetics, 2018, 26, 582-586.	1.4	14
54	Ehlersâ€“Danlos syndrome with lethal cardiac valvular dystrophy in males carrying a novel splice mutation in FLNA. American Journal of Medical Genetics, Part A, 2017, 173, 169-176.	0.7	13

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55	A classical Ehlers-Danlos syndrome family with incomplete presentation diagnosed by molecular testing. European Journal of Medical Genetics, 2018, 61, 17-20.	0.7	13
56	Skeletal fragility: an emerging complication of Ehlersâ€“Danlos syndrome. Endocrine, 2019, 63, 225-230.	1.1	13
57	Risk Profile of Symptomatic Lacunar Stroke Versus Nonlobar Intracerebral Hemorrhage. Stroke, 2016, 47, 2141-2143.	1.0	12
58	Clinical and molecular characterization of an 18â€monthâ€old infant with autosomal recessive cutis laxa type 1C due to a novel LTBP4 pathogenic variant, and literature review. Molecular Genetics & Genomic Medicine, 2019, 7, e00735.	0.6	12
59	Biological insights in the pathogenesis of hypermobile Ehlers-Danlos syndrome from proteome profiling of patients' dermal myofibroblasts. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2021, 1867, 166051.	1.8	12
60	Clinical, neuroradiological and molecular features of a patient affected by pseudoxhantoma elasticum associated to carotid rete mirabile: Case report. Clinical Neurology and Neurosurgery, 2012, 114, 758-761.	0.6	11
61	Absence of Collagen Flowers on Electron Microscopy and Identification of (Likely) Pathogenic COL5A1 Variants in Two Patients. Genes, 2019, 10, 762.	1.0	11
62	Characterization and expression pattern analysis of the facilitative glucose transporter 10 gene (slc2a10) in Danio rerio. International Journal of Developmental Biology, 2011, 55, 229-236.	0.3	9
63	Marfan syndrome: Report of a complex phenotype due to a 15q21.1 contiguos gene deletion encompassing FBN1, and literature review. American Journal of Medical Genetics, Part A, 2017, 173, 200-206.	0.7	9
64	Atypical variants in COL1A1 and COL3A1 associated with classical and vascular Ehlers-Danlos syndrome overlap phenotypes: expanding the clinical phenotype based on additional case reports. Clinical and Experimental Rheumatology, 2022, 40, 46-62.	0.4	9
65	Koolen-de Vries Syndrome: Clinical Report of an Adult and Literature Review. Cytogenetic and Genome Research, 2016, 150, 40-45.	0.6	8
66	Novel pathogenic TGFBR1 and SMAD3 variants identified after cerebrovascular events in adult patients with Loeys-dietz syndrome. European Journal of Medical Genetics, 2019, 62, 103727.	0.7	8
67	Expanding the PURA syndrome phenotype: A child with the recurrent PURA p.(Phe233del) pathogenic variant showing similarities with cutis laxa. Molecular Genetics & Genomic Medicine, 2021, 9, e1562.	0.6	8
68	Application of the 2017 criteria for vascular Ehlersâ€Ðanlos syndrome in 50 patients ascertained according to the Villefranche nosology. Clinical Genetics, 2020, 97, 287-295.	1.0	7
69	Genome-first approach for the characterization of a complex phenotype with combined NBAS and CUL4B deficiency. Bone, 2020, 140, 115571.	1.4	7
70	Insights into the molecular pathogenesis of cardiospondylocarpofacial syndrome: MAP3K7 c.737-7AÂ>ÂG variant alters the TGFÎ²-mediated Î±-SMA cytoskeleton assembly and autophagy. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2020, 1866, 165742.	1.8	7
71	Clinical and molecular characterization of a 13-year-old Indian boy with cutis laxa type 2B: Identification of two novel PYCR1 mutations by amplicon-based semiconductor exome sequencing. Journal of Dermatological Science, 2017, 88, 141-143.	1.0	6
72	Identification of two novel ATP6V0A2 mutations in an infant with cutis laxa by exome sequencing. Journal of Dermatological Science, 2014, 75, 66-68.	1.0	5

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73	TAB2 c.1398dup variant leads to haploinsufficiency and impairs extracellular matrix homeostasis. Human Mutation, 2019, 40, 1886-1898.	1.1	5
74	Clinical and Genetic Heterogeneity in a Large Family with Pseudoxanthoma Elasticum: MTHFR and SERPINE1 Variants as Possible Disease Modifiers in Developing Ischemic Stroke. Journal of Stroke and Cerebrovascular Diseases, 2021, 30, 105744.	0.7	5
75	Matrix Metalloproteinases Inhibition by Doxycycline Rescues Extracellular Matrix Organization and Partly Reverts Myofibroblast Differentiation in Hypermobile Ehlers-Danlos Syndrome Dermal Fibroblasts: A Potential Therapeutic Target?. Cells, 2021, 10, 3236.	1.8	5
76	Characterization of a Pseudoxanthoma elasticum-like patient with coagulation deficiency, cutaneous calcinosis and GGCX compound heterozygosity. Journal of Dermatological Science, 2018, 89, 201-204.	1.0	4
77	The FN13 peptide inhibits human tumor cells invasion through the modulation of Î±vÎ²3 integrins organization and the inactivation of ILK pathway. Biochimica Et Biophysica Acta - Molecular Cell Research, 2007, 1773, 747-763.	1.9	3
78	Identification of the novel COL5A1 c.3369_3431dup, p.(Glu1124_Gly1144dup) variant in a patient with incomplete classical Ehlersâ€“Danlos syndrome: The importance of phenotypeâ€guided genetic testing. Molecular Genetics & Genomic Medicine, 2020, 8, e1422.	0.6	2
79	Macrophage Cell Cultures for Rapid Isolation of Intracellular Bacteria: The Mycobacterium bovis Model. , 2006, 345, 203-218.		1
80	Erratum to â€œLetter to the Editor â€“ Diagnosis of vascular Ehlers-Danlos syndrome in Italy: Clinical findings and novel COL3A1 mutationsâ€•[J. Dermatol. Sci. 64 (2011) 237â€“248]. Journal of Dermatological Science, 2012, 65, 77.	1.0	1
81	Aortic dissection and stroke in a 37-year-old woman: discovering an emerging heritable connective tissue disorder. Internal and Emergency Medicine, 2015, 10, 165-170.	1.0	1
82	Hereditary alpha-tryptasemia modifies clinical phenotypes among individuals with congenital hypermobility disorders. Human Genetics and Genomics Advances, 2022, 3, 100094.	1.0	1
83	Age-dependent effect of susceptibility factors on the risk of intracerebral haemorrhage: Multicenter Study on Cerebral Hemorrhage in Italy (MUCH-Italy). Journal of Neurology, Neurosurgery and Psychiatry, 2021, 92, 1068-1071.	0.9	0
84	Generation of the induced pluripotent stem cell line UNIBSi017-A from an individual with cardiospondylocarpofacial syndrome and the MAP3K7 c.737-7A>G variant. Stem Cell Research, 2022, , 102837.	0.3	0