

# Marina Colombi

## List of Publications by Year in descending order

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113  
papers

4,229  
citations

186265

28  
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128289

60  
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113  
all docs

113  
docs citations

113  
times ranked

4133  
citing authors

#	ARTICLE	IF	CITATIONS
1	The 2017 international classification of the Ehlers-Danlos syndromes. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2017, 175, 8-26.	1.6	1,163
2	Mutations in the facilitative glucose transporter GLUT10 alter angiogenesis and cause arterial tortuosity syndrome. Nature Genetics, 2006, 38, 452-457.	21.4	354
3	Mutations in FKBP14 Cause a Variant of Ehlers-Danlos Syndrome with Progressive Kyphoscoliosis, Myopathy, and Hearing Loss. American Journal of Human Genetics, 2012, 90, 201-216.	6.2	136
4	Ehlers-Danlos syndrome, classical type. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2017, 175, 27-39.	1.6	116
5	Mutations in PRDM5 in Brittle Cornea Syndrome Identify a Pathway Regulating Extracellular Matrix Development and Maintenance. American Journal of Human Genetics, 2011, 88, 767-777.	6.2	106
6	Clinical and molecular characterization of 40 patients with classic Ehlers-Danlos syndrome: identification of 18 COL5A1 and 2 COL5A2 novel mutations. Orphanet Journal of Rare Diseases, 2013, 8, 58.	2.7	101
7	Differential diagnosis and diagnostic flow chart of joint hypermobility syndrome/ehlers-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.	1.6	91
8	Human Fibroblasts with Mutations in COL5A1 and COL3A1 Genes Do Not Organize Collagens and Fibronectin in the Extracellular Matrix, Down-regulate $\alpha 2 \beta 1$ Integrin, and Recruit $\alpha 5 \beta 1$ Instead of $\alpha 5 \beta 2$ Integrin. Journal of Biological Chemistry, 2004, 279, 18157-18168.	3.4	90
9	Gynecologic and obstetric implications of the joint hypermobility syndrome (a.k.a. Ehlers-Danlos) Tj ETQq1 1 0.784314 rgBT /Overl 158A, 2176-2182.	1.2	78
10	Nosology and inheritance pattern(s) of joint hypermobility syndrome and Ehlers-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.	1.2	70
11	Genotype-Phenotype Correlation in Italian Patients with Dystrophic Epidermolysis Bullosa. Journal of Investigative Dermatology, 2002, 119, 1456-1462.	0.7	58
12	Thoracic Aortic Aneurysm in Infancy in Aneurysms-Osteoarthritis Syndrome Due to a Novel SMAD3 Mutation: Further Delineation of the Phenotype. American Journal of Medical Genetics, Part A, 2013, 161, 1028-1035.	1.2	58
13	RT-PCR detection of fibronectin EDA <sup>+</sup> and EDB <sup>+</sup> mRNA isoforms: Molecular markers for hepatocellular carcinoma. International Journal of Cancer, 1994, 56, 820-825.	5.1	46
14	Cellular and Molecular Mechanisms in the Pathogenesis of Classical, Vascular, and Hypermobile Ehlers-Danlos Syndromes. Genes, 2019, 10, 609.	2.4	46
15	Chronic mucocutaneous candidiasis and connective tissue disorder in humans with impaired JNK1-dependent responses to IL-17A/F and TGF- $\beta 2$ . Science Immunology, 2019, 4, .	11.9	45
16	Homozygosity mapping of a gene for arterial tortuosity syndrome to chromosome 20q13. Journal of Medical Genetics, 2003, 40, 747-751.	3.2	44
17	Exclusion of candidate genes in a family with arterial tortuosity syndrome. American Journal of Medical Genetics Part A, 2004, 126A, 221-228.	2.4	42
18	GLUT10 deficiency leads to oxidative stress and non-canonical $\alpha 5 \beta 3$ integrin-mediated TGF- $\beta 2$ signalling associated with extracellular matrix disarray in arterial tortuosity syndrome skin fibroblasts. Human Molecular Genetics, 2015, 24, 6769-6787.	2.9	42

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19	High prevalence of radiological vertebral fractures in adult patients with Ehlers-Danlos syndrome. <i>Bone</i> , 2016, 84, 88-92.	2.9	41
20	Transcriptome-Wide Expression Profiling in Skin Fibroblasts of Patients with Joint Hypermobility Syndrome/Ehlers-Danlos Syndrome Hypermobility Type. <i>PLoS ONE</i> , 2016, 11, e0161347.	2.5	40
21	Cavin-1 and Caveolin-1 are both required to support cell proliferation, migration and anchorage-independent cell growth in rhabdomyosarcoma. <i>Laboratory Investigation</i> , 2015, 95, 585-602.	3.7	37
22	Arterial Tortuosity Syndrome: homozygosity for two novel and one recurrent SLC2A10 missense mutations in three families with severe cardiopulmonary complications in infancy and a literature review. <i>BMC Medical Genetics</i> , 2014, 15, 122.	2.1	36
23	Delineation of Ehlers-Danlos syndrome phenotype due to the c.934C>T, p.(Arg312Cys) mutation in <i>COL1A1</i> : Report on a three-generation family without cardiovascular events, and literature review. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 524-530.	1.2	35
24	Dermal fibroblast-to-myofibroblast transition sustained by $\beta_3$ integrin-ILK-Snail1/Slug signaling is a common feature for hypermobile Ehlers-Danlos syndrome and hypermobility spectrum disorders. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2018, 1864, 1010-1023.	3.8	34
25	Dystrophic epidermolysis bullosa pruriginosa in Italy: clinical and molecular characterization. <i>Clinical Genetics</i> , 2006, 70, 339-347.	2.0	33
26	Generalized joint hypermobility, joint hypermobility syndrome and Ehlers-Danlos syndrome, hypermobility type. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2015, 169, 1-5.	1.6	33
27	Expanding the clinical and mutational spectrum of B4GALT7-spondylodysplastic Ehlers-Danlos syndrome. <i>Orphanet Journal of Rare Diseases</i> , 2017, 12, 153.	2.7	31
28	Transcriptome analysis of skin fibroblasts with dominant negative COL3A1 mutations provides molecular insights into the etiopathology of vascular Ehlers-Danlos syndrome. <i>PLoS ONE</i> , 2018, 13, e0191220.	2.5	31
29	Spectrum of mucocutaneous manifestations in 277 patients with joint hypermobility syndrome/Ehlers-Danlos syndrome, hypermobility type. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2015, 169, 43-53.	1.6	30
30	Further delineation of Loeys-Dietz syndrome type 4 in a family with mild vascular involvement and a TGFB2 splicing mutation. <i>BMC Medical Genetics</i> , 2014, 15, 91.	2.1	29
31	Molecular Genetics and Pathogenesis of Ehlers-Danlos Syndrome and Related Connective Tissue Disorders. <i>Genes</i> , 2020, 11, 547.	2.4	29
32	Two novel SLC2A10/GLUT10 mutations in a patient with arterial tortuosity syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2007, 143A, 216-218.	1.2	27
33	FAK-independent $\beta_3$ integrin-EGFR complexes rescue from anoikis matrix-defective fibroblasts. <i>Biochimica Et Biophysica Acta - Molecular Cell Research</i> , 2008, 1783, 1177-1188.	4.1	27
34	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. <i>Molecular Genetics and Metabolism Reports</i> , 2015, 2, 1-15.	1.1	27
35	<i>COL1</i> -related overlap disorder: A novel connective tissue disorder incorporating the osteogenesis imperfecta/Ehlers-Danlos syndrome overlap. <i>Clinical Genetics</i> , 2020, 97, 396-406.	2.0	27
36	Loeys-Dietz syndrome type I and type II: clinical findings and novel mutations in two Italian patients. <i>Orphanet Journal of Rare Diseases</i> , 2009, 4, 24.	2.7	26

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37	Spectrum of mucocutaneous, ocular and facial features and delineation of novel presentations in 62 classical Ehlers-Danlos syndrome patients. <i>Clinical Genetics</i> , 2017, 92, 624-631.	2.0	26
38	Adult presentation of arterial tortuosity syndrome in a 51-year-old woman with a novel homozygous c.1411+1G>A mutation in the <i>SLC2A10</i> gene. <i>American Journal of Medical Genetics, Part A</i> , 2012, 158A, 1164-1169.	1.2	25
39	Glucose transporter type 10-lacking in arterial tortuosity syndrome-facilitates dehydroascorbic acid transport. <i>FEBS Letters</i> , 2016, 590, 1630-1640.	2.8	25
40	<i>COL6A5</i> variants in familial neuropathic chronic itch. <i>Brain</i> , 2017, 140, aww343.	7.6	25
41	A family of fibronectin mRNAs in human normal and transformed cells. <i>Biochimica Et Biophysica Acta Gene Regulatory Mechanisms</i> , 1986, 868, 207-214.	2.4	24
42	Recurrent exercise-induced acute renal failure in a young Pakistani man with severe renal hypouricemia and <i>SLC2A9</i> compound heterozygosity. <i>BMC Medical Genetics</i> , 2014, 15, 3.	2.1	24
43	Multifaced Roles of the $\alpha 3$ Integrin in Ehlers-Danlos and Arterial Tortuosity Syndromes™ Dermal Fibroblasts. <i>International Journal of Molecular Sciences</i> , 2018, 19, 982.	4.1	24
44	Matrix Assembly Induction and Cell Migration and Invasion Inhibition by a 13-Amino Acid Fibronectin Peptide. <i>Journal of Biological Chemistry</i> , 2003, 278, 14346-14355.	3.4	23
45	Towards the genetic basis of cerebral venous thrombosis-the BEAST Consortium: a study protocol: Table 1. <i>BMJ Open</i> , 2016, 6, e012351.	1.9	23
46	Further Defining the Phenotypic Spectrum of <i>B3GAT3</i> Mutations and Literature Review on Linkeropathy Syndromes. <i>Genes</i> , 2019, 10, 631.	2.4	23
47	Expanding the Clinical and Mutational Spectrum of Recessive <i>AEBP1</i> -Related Classical-Like Ehlers-Danlos Syndrome. <i>Genes</i> , 2019, 10, 135.	2.4	23
48	Study of fibronectin mRNA in human laryngeal and ectocervical carcinomas by IN SITU hybridization and image analysis. <i>International Journal of Cancer</i> , 1992, 51, 692-697.	5.1	22
49	Rescue of Migratory Defects of Ehlers-Danlos Syndrome Fibroblasts In Vitro by Type V Collagen but not Insulin-Like Binding Protein-1. <i>Journal of Investigative Dermatology</i> , 2008, 128, 1915-1919.	0.7	22
50	Molecular insights in the pathogenesis of classical Ehlers-Danlos syndrome from transcriptome-wide expression profiling of patients' skin fibroblasts. <i>PLoS ONE</i> , 2019, 14, e0211647.	2.5	22
51	Severity classes in adults with hypermobile Ehlers-Danlos syndrome/hypermobility spectrum disorders: a pilot study of 105 Italian patients. <i>Rheumatology</i> , 2019, 58, 1722-1730.	1.9	22
52	Identification of a novel <i>TGFBR1</i> mutation in a Loey's-Dietz syndrome type II patient with vascular Ehlers-Danlos syndrome phenotype. <i>Clinical Genetics</i> , 2008, 73, 290-293.	2.0	21
53	Arterial tortuosity syndrome in two Italian paediatric patients. <i>Orphanet Journal of Rare Diseases</i> , 2009, 4, 20.	2.7	21
54	Recurring and Generalized Visceroptosis in Ehlers-Danlos Syndrome Hypermobility Type. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 1143-1147.	1.2	21

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55	Refining patterns of joint hypermobility, <i>habitus</i> , and orthopedic traits in joint hypermobility syndrome and Ehlers-Danlos syndrome, hypermobility type. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 914-929.	1.2	20
56	Genotypic Categorization of Loeys-Dietz Syndrome Based on 24 Novel Families and Literature Data. <i>Genes</i> , 2019, 10, 764.	2.4	20
57	Multisystemic manifestations in a cohort of 75 classical Ehlers-Danlos syndrome patients: natural history and nosological perspectives. <i>Orphanet Journal of Rare Diseases</i> , 2020, 15, 197.	2.7	20
58	Bruch's membrane abnormalities in PRDM5-related brittle cornea syndrome. <i>Orphanet Journal of Rare Diseases</i> , 2015, 10, 145.	2.7	19
59	Melatonin decreases cell proliferation, impairs myogenic differentiation and triggers apoptotic cell death in rhabdomyosarcoma cell lines. <i>Oncology Reports</i> , 2015, 34, 279-287.	2.6	19
60	Caveolin-1, Caveolin-2 and Cavin-1 are strong predictors of adipogenic differentiation in human tumors and cell lines of liposarcoma. <i>European Journal of Cell Biology</i> , 2016, 95, 252-264.	3.6	19
61	A recognizable systemic connective tissue disorder with polyvalvular heart dystrophy and dysmorphism associated with <i>TAB2</i> mutations. <i>Clinical Genetics</i> , 2018, 93, 126-133.	2.0	19
62	Exploring relationships between joint hypermobility and neurodevelopment in children (4-13 years) with hereditary connective tissue disorders and developmental coordination disorder. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2018, 177, 546-556.	1.7	19
63	Type III and V collagens modulate the expression and assembly of EDA+ fibronectin in the extracellular matrix of defective Ehlers-Danlos syndrome fibroblasts. <i>Biochimica Et Biophysica Acta - General Subjects</i> , 2012, 1820, 1576-1587.	2.4	18
64	Phosphocaveolin-1 Enforces Tumor Growth and Chemoresistance in Rhabdomyosarcoma. <i>PLoS ONE</i> , 2014, 9, e84618.	2.5	17
65	Three homozygous PTC mutations in the collagen type VII gene of patients affected by recessive dystrophic epidermolysis bullosa: Analysis of transcript levels in dermal fibroblasts. , 1999, 13, 439-452.		16
66	Diagnosis of vascular Ehlers-Danlos syndrome in Italy: Clinical findings and novel COL3A1 mutations. <i>Journal of Dermatological Science</i> , 2011, 64, 237-240.	1.9	16
67	Clinical and Molecular Characterization of Classical-Like Ehlers-Danlos Syndrome Due to a Novel TNXB Variant. <i>Genes</i> , 2019, 10, 843.	2.4	16
68	GLUT10 "Lacking in Arterial Tortuosity Syndrome" Is Localized to the Endoplasmic Reticulum of Human Fibroblasts. <i>International Journal of Molecular Sciences</i> , 2017, 18, 1820.	4.1	15
69	Further delineation of <i>FKBP14</i> -related Ehlers-Danlos syndrome: A patient with early vascular complications and non-progressive kyphoscoliosis, and literature review. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 2031-2038.	1.2	14
70	A novel MAP3K7 splice mutation causes cardiospondylocarpofacial syndrome with features of hereditary connective tissue disorder. <i>European Journal of Human Genetics</i> , 2018, 26, 582-586.	2.8	14
71	Ehlers-Danlos syndrome with lethal cardiac valvular dystrophy in males carrying a novel splice mutation in <i>FLNA</i> . <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 169-176.	1.2	13
72	A classical Ehlers-Danlos syndrome family with incomplete presentation diagnosed by molecular testing. <i>European Journal of Medical Genetics</i> , 2018, 61, 17-20.	1.3	13

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73	Skeletal fragility: an emerging complication of Ehlers-Danlos syndrome. <i>Endocrine</i> , 2019, 63, 225-230.	2.3	13
74	A -96C>T mutation in the promoter of the collagen type VII gene (COL7A1) abolishing transcription in a patient affected by recessive dystrophic epidermolysis bullosa. <i>Human Mutation</i> , 2000, 16, 275-275.	2.5	12
75	Clinical and molecular characterization of an 18-month-old infant with autosomal recessive cutis laxa type 1C due to a novel <i>LTBP4</i> pathogenic variant, and literature review. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2019, 7, e00735.	1.2	12
76	Biological insights in the pathogenesis of hypermobile Ehlers-Danlos syndrome from proteome profiling of patients' dermal myofibroblasts. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2021, 1867, 166051.	3.8	12
77	EFFECT OF DEXAMETHASONE ON THE ASSEMBLY OF THE MATRIX OF FIBRONECTIN AND ON ITS RECEPTORS ORGANIZATION IN EHLERS-DANLOS SYNDROME SKIN FIBROBLASTS. <i>Cell Biology International</i> , 1998, 22, 499-508.	3.0	11
78	Absence of Collagen Flowers on Electron Microscopy and Identification of (Likely) Pathogenic COL5A1 Variants in Two Patients. <i>Genes</i> , 2019, 10, 762.	2.4	11
79	Italian validation of the functional difficulties questionnaire (FDQ) and its correlation with major determinants of quality of life in adults with hypermobile Ehlers-Danlos syndrome/hypermobility spectrum disorder. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2019, 180, 25-34.	1.7	11
80	Altered fibronectin mRNA splicing in skin fibroblasts from Ehlers-Danlos syndrome patients: hybridization analysis. <i>Cell Biology International Reports</i> , 1991, 15, 1195-1206.	0.6	10
81	Genome-Wide Association Study Identifies First Locus Associated with Susceptibility to Cerebral Venous Thrombosis. <i>Annals of Neurology</i> , 2021, 90, 777-788.	5.3	10
82	Characterization and expression pattern analysis of the facilitative glucose transporter 10 gene ( <i>slc2a10</i> ) in <i>Danio rerio</i> . <i>International Journal of Developmental Biology</i> , 2011, 55, 229-236.	0.6	9
83	Marfan syndrome: Report of a complex phenotype due to a 15q21.1 contiguous gene deletion encompassing <i>FBN1</i> , and literature review. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 200-206.	1.2	9
84	Differential Enzymatic Activity of Rat ADAR2 Splicing Variants Is Due to Altered Capability to Interact with RNA in the Deaminase Domain. <i>Genes</i> , 2018, 9, 79.	2.4	9
85	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. <i>Clinical and Experimental Rheumatology</i> , 2022, 40, 46-62.	0.8	9
86	A frequent HindIII RFLP of the human fibronectin gene (FN1). <i>Nucleic Acids Research</i> , 1988, 16, 9074-9074.	14.5	8
87	Novel pathogenic TGFBR1 and SMAD3 variants identified after cerebrovascular events in adult patients with Loey's dietz syndrome. <i>European Journal of Medical Genetics</i> , 2019, 62, 103727.	1.3	8
88	Expanding the PURA syndrome phenotype: A child with the recurrent <i>PURA</i> p.(Phe233del) pathogenic variant showing similarities with cutis laxa. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2021, 9, e1562.	1.2	8
89	Correction of the defective extracellular matrix of Ehlers-Danlos syndrome skin fibroblasts by dexamethasone.. <i>Cell Biology International</i> , 1994, 18, 29-38.	3.0	7
90	Application of the 2017 criteria for vascular Ehlers-Danlos syndrome in 50 patients ascertained according to the Villefranche nosology. <i>Clinical Genetics</i> , 2020, 97, 287-295.	2.0	7

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91	Genome-first approach for the characterization of a complex phenotype with combined NBAS and CUL4B deficiency. <i>Bone</i> , 2020, 140, 115571.	2.9	7
92	Insights into the molecular pathogenesis of cardiospondylocarpofacial syndrome: MAP3K7 c.737-7AA&gt;AG variant alters the TGF $\beta$ 2-mediated $\beta$ 1-SMA cytoskeleton assembly and autophagy. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2020, 1866, 165742.	3.8	7
93	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. <i>Journal of Dermatological Science</i> , 2017, 88, 141-143.	1.9	6
94	Identification of two novel ATP6V0A2 mutations in an infant with cutis laxa by exome sequencing. <i>Journal of Dermatological Science</i> , 2014, 75, 66-68.	1.9	5
95	TAB2 c.1398dup variant leads to haploinsufficiency and impairs extracellular matrix homeostasis. <i>Human Mutation</i> , 2019, 40, 1886-1898.	2.5	5
96	Clinical and Genetic Heterogeneity in a Large Family with Pseudoxanthoma Elasticum: MTHFR and SERPINE1 Variants as Possible Disease Modifiers in Developing Ischemic Stroke. <i>Journal of Stroke and Cerebrovascular Diseases</i> , 2021, 30, 105744.	1.6	5
97	Clinical variability in two Macedonian families with Arterial tortuosity syndrome. <i>Balkan Journal of Medical Genetics</i> , 2018, 21, 47-52.	0.5	5
98	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?. <i>Cells</i> , 2021, 10, 3236.	4.1	5
99	The novel p.G150R missense mutation in the cartilage matrix protein subdomain of type VII collagen in compound heterozygosity with the c.682+1G>A COL7A1 splicing mutation leads to mild dystrophic epidermolysis bullosa. <i>Journal of Dermatological Science</i> , 2009, 53, 222-225.	1.9	4
100	Characterization of a Pseudoxanthoma elasticum-like patient with coagulation deficiency, cutaneous calcinosis and GGCX compound heterozygosity. <i>Journal of Dermatological Science</i> , 2018, 89, 201-204.	1.9	4
101	Decreased Nuclear Ascorbate Accumulation Accompanied with Altered Genomic Methylation Pattern in Fibroblasts from Arterial Tortuosity Syndrome Patients. <i>Oxidative Medicine and Cellular Longevity</i> , 2019, 2019, 1-11.	4.0	4
102	Plasminogen activators in nude mice xenotransplanted with human tumorigenic cells. <i>Invasion &amp; Metastasis</i> , 1995, 15, 22-33.	0.5	4
103	The type III-9 repeat of human fibronectin is encoded by a single exon which is not alternatively spliced.. <i>Cell Biology International</i> , 1993, 17, 989-992.	3.0	2
104	MURC/cavin-4 Is Co-Expressed with Caveolin-3 in Rhabdomyosarcoma Tumors and Its Silencing Prevents Myogenic Differentiation in the Human Embryonal RD Cell Line. <i>PLoS ONE</i> , 2015, 10, e0130287.	2.5	2
105	Identification of the novel <i>COL5A1</i> c.3369_3431dup, p.(Glu1124_Gly1144dup) variant in a patient with incomplete classical Ehlersâ€“Danlos syndrome: The importance of phenotypeâ€“guided genetic testing. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2020, 8, e1422.	1.2	2
106	Erratum to â€œLetter to the Editor â€“ Diagnosis of vascular Ehlers-Danlos syndrome in Italy: Clinical findings and novel COL3A1 mutationsâ€“[J. <i>Dermatol. Sci.</i> 64 (2011) 237â€“248]. <i>Journal of Dermatological Science</i> , 2012, 65, 77.	1.9	1
107	Compound heterozygosity of the novel âˆ’186C>T mutation in the COL7A1 promoter and the recurrent c.497insA mutation leads to generalized dystrophic epidermolysis bullosa. <i>British Journal of Dermatology</i> , 2013, 168, 904-906.	1.5	1
108	A Child With Self-Improving Hypotonia: Look at the Skin!. <i>Journal of Pediatrics</i> , 2020, 225, 269-270.	1.8	1

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109	Hereditary alpha-tryptasemia modifies clinical phenotypes among individuals with congenital hypermobility disorders. <i>Human Genetics and Genomics Advances</i> , 2022, 3, 100094.	1.7	1
110	Compound heterozygosity for a novel and a recurrent ABCC6 gene mutation in an Italian family with Pseudoxanthoma elasticum. <i>Journal of Dermatological Science</i> , 2008, 49, 252-255.	1.9	0
111	From the bedside to the bench and backwards: diagnostic approach and management of Ehlers-Danlos syndrome(s) in Italy. <i>Journal of Medical Rehabilitation</i> , 2016, 36, 9-27.	0.0	0
112	Arterial tortuosity syndrome causing recurrent transient ischemic attacks in young adult: a case report. <i>BMC Neurology</i> , 2021, 21, 464.	1.8	0
113	Angiokeratoma corporis diffusum and arteriovenous fistulas with dominant transmission in the absence of metabolic disorders. <i>Archives of Dermatology</i> , 1995, 131, 57-62.	1.4	0