Fransiska Malfait

List of Publications by Year
in descending order

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129 papers	7,347 citations	53794 45 h-index	60623 81 g-index

131 all docs	131 docs citations	131 times ranked	6420 citing authors

#	Article	IF	Citations
1	The 2017 international classification of the Ehlersâ€“Danlos syndromes. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2017, 175, 8-26.	1.6	1,163
2	A framework for the classification of joint hypermobility and related conditions. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2017, 175, 148-157.	1.6	356
3	Clinical and genetic aspects of Ehlers-Danlos syndrome, classic type. Genetics in Medicine, 2010, 12, 597-605.	2.4	220
4	Ehlers-Danlos syndromes and Marfan syndrome. Best Practice and Research in Clinical Rheumatology, 2008, 22, 165-189.	3.3	205
5	Bleeding and bruising in patients with Ehlersâ€“Danlos syndrome and other collagen vascular disorders. British Journal of Haematology, 2004, 127, 491-500.	2.5	190
6	Musculoskeletal complaints, physical activity and health-related quality of life among patients with the Ehlersâ€“Danlos syndrome hypermobility type. Disability and Rehabilitation, 2010, 32, 1339-1345.	1.8	186
7	The Ehlersâ€“Danlos syndromes, rare types. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2017, 175, 70-115.	1.6	168
8	Mutations in FKBP10 cause recessive osteogenesis imperfecta and bruck syndrome. Journal of Bone and Mineral Research, 2011, 26, 666-672.	2.8	149
9	The Ehlersâ€“Danlos syndromes. Nature Reviews Disease Primers, 2020, 6, 64.	30.5	144
10	Three arginine to cysteine substitutions in the pro-alpha (I)-collagen chain cause Ehlers-Danlos syndrome with a propensity to arterial rupture in early adulthood. Human Mutation, 2007, 28, 387-395.	2.5	139
11	Comprehensive molecular analysis demonstrates type V collagen mutations in over 90% of patients with classic EDS and allows to refine diagnostic criteria. Human Mutation, 2012, 33, 1485-1493.	2.5	133
12	The molecular basis of classic Ehlers-Danlos syndrome: A comprehensive study of biochemical and molecular findings in 48 unrelated patients. Human Mutation, 2005, 25, 28-37.	2.5	117
13	Novel Types of Mutation Responsible for the Dermatosparactic Type of Ehlersâ€“Danlos Syndrome (Type) Tj ETQq1 1 0.784314 rgBT /O 123, 656-663.	0.7	116
14	Defective Initiation of Glycosaminoglycan Synthesis due to B3GALT6 Mutations Causes a Pleiotropic Ehlers-Danlos-Syndrome-like Connective Tissue Disorder. American Journal of Human Genetics, 2013, 92, 935-945.	6.2	116
15	Dysautonomia and its underlying mechanisms in the hypermobility type of Ehlersâ€“Danlos syndrome. Seminars in Arthritis and Rheumatism, 2014, 44, 93-100.	3.4	116
16	Ehlersâ€“Danlos syndrome, classical type. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2017, 175, 27-39.	1.6	116
17	EMQN best practice guidelines for the laboratory diagnosis of osteogenesis imperfecta. European Journal of Human Genetics, 2012, 20, 11-19.	2.8	107
18	Musculocontractural Ehlers-Danlos Syndrome (former EDS type VIB) and adducted thumb clubfoot syndrome (ATCS) represent a single clinical entity caused by mutations in the dermatan-4-sulfotransferase 1 encoding CHST14 gene. Human Mutation, 2010, 31, 1233-1239.	2.5	105

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19	Deficiency for the ER-stress transducer OASIS causes severe recessive osteogenesis imperfecta in humans. Orphanet Journal of Rare Diseases, 2013, 8, 154.	2.7	98
20	Medication, Surgery, and Physiotherapy Among Patients With the Hypermobility Type of Ehlers-Danlos Syndrome. Archives of Physical Medicine and Rehabilitation, 2011, 92, 1106-1112.	0.9	94
21	The Ehlers-Danlos Syndrome. Advances in Experimental Medicine and Biology, 2014, 802, 129-143.	1.6	93
22	Molecular genetics inclassic Ehlers-Danlos syndrome. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2005, 139C, 17-23.	1.6	86
23	Vascular aspects of the Ehlers-Danlos Syndromes. Matrix Biology, 2018, 71-72, 380-395.	3.6	85
24	Joint position sense and vibratory perception sense in patients with Ehlersâ€“Danlos syndrome type III (hypermobility type). Clinical Rheumatology, 2010, 29, 289-295.	2.2	84
25	Balance, gait, falls, and fear of falling in women with the hypermobility type of Ehlersâ€Danlos syndrome. Arthritis Care and Research, 2011, 63, 1432-1439.	3.4	84
26	Muscle mass, muscle strength, functional performance, and physical impairment in women with the hypermobility type of Ehlersâ€Danlos syndrome. Arthritis Care and Research, 2012, 64, 1584-1592.	3.4	84
27	Recommendations for anesthesia and perioperative management in patients with Ehlers-Danlos syndrome(s). Orphanet Journal of Rare Diseases, 2014, 9, 109.	2.7	83
28	Mutations in ATP6V1E1 or ATP6V1A Cause Autosomal-Recessive Cutis Laxa. American Journal of Human Genetics, 2017, 100, 216-227.	6.2	82
29	Autonomic symptom burden in the hypermobility type of Ehlersâ€“Danlos syndrome: A comparative study with two other EDS types, fibromyalgia, and healthy controls. Seminars in Arthritis and Rheumatism, 2014, 44, 353-361.	3.4	81
30	Bleeding in the heritable connective tissue disorders: Mechanisms, diagnosis and treatment. Blood Reviews, 2009, 23, 191-197.	5.7	77
31	Zebrafish type I collagen mutants faithfully recapitulate human type I collagenopathies. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, E8037-E8046.	7.1	77
32	The natural history, including orofacial features of three patients with Ehlers-Danlos syndrome, dermatosparaxis type (EDS type VIIC). American Journal of Medical Genetics Part A, 2004, 131A, 18-28.	2.4	75
33	Impairment and impact of pain in female patients with Ehlers-Danlos syndrome: A comparative study with fibromyalgia and rheumatoid arthritis. Arthritis and Rheumatism, 2011, 63, 1979-1987.	6.7	73
34	Helical mutations in type I collagen that affect the processing of the amino-propeptide result in an Osteogenesis Imperfecta/Ehlers-Danlos Syndrome overlap syndrome. Orphanet Journal of Rare Diseases, 2013, 8, 78.	2.7	73
35	Chronic pain in patients with the hypermobility type of Ehlersâ€“Danlos syndrome: evidence for generalized hyperalgesia. Clinical Rheumatology, 2015, 34, 1121-1129.	2.2	72
36	Clinical utility gene card for: osteogenesis imperfecta. European Journal of Human Genetics, 2013, 21, 1-4.	2.8	68

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37	Comprehensive Clinical and Molecular Analysis of 12 Families with Type 1 Recessive Cutis Laxa. Human Mutation, 2013, 34, 111-121.	2.5	67
38	Clinical heterogeneity in patients with the hypermobility type of Ehlers-Danlos Syndrome. Research in Developmental Disabilities, 2013, 34, 873-881.	2.2	66
39	Genetic Heterogeneity and Clinical Variability in Musculocontractural Ehlers-Danlos Syndrome Caused by Impaired Dermatan Sulfate Biosynthesis. Human Mutation, 2015, 36, 535-547.	2.5	65
40	Loss of Type I Collagen Telopeptide Lysyl Hydroxylation Causes Musculoskeletal Abnormalities in a Zebrafish Model of Bruck Syndrome. Journal of Bone and Mineral Research, 2016, 31, 1930-1942.	2.8	65
41	Vascular phenotypes in nonvascular subtypes of the Ehlers-Danlos syndrome: a systematic review. Genetics in Medicine, 2018, 20, 562-573.	2.4	62
42	COL5A1 signal peptide mutations interfere with protein secretion and cause classic Ehlers-Danlos syndrome. Human Mutation, 2009, 30, E395-E403.	2.5	57
43	The RIN2 syndrome: a new autosomal recessive connective tissue disorder caused by deficiency of Ras and Rab interactor 2 (RIN2). Human Genetics, 2010, 128, 79-88.	3.8	51
44	The Genetics of Soft Connective Tissue Disorders. Annual Review of Genomics and Human Genetics, 2015, 16, 229-255.	6.2	50
45	Identification of binding partners interacting with the Î±1-N-propeptide of typeÂV collagen. Biochemical Journal, 2011, 433, 371-381.	3.7	49
46	Osteogenesis imperfecta: the audiological phenotype lacks correlation with the genotype. Orphanet Journal of Rare Diseases, 2011, 6, 88.	2.7	48
47	Muscleâ€“tendon tissue properties in the hypermobility type of Ehlersâ€Danlos syndrome. Arthritis Care and Research, 2012, 64, 766-772.	3.4	48
48	Novel pathogenic COL11A1/COL11A2 variants in Stickler syndrome detected by targeted NGS and exome sequencing. Molecular Genetics and Metabolism, 2014, 113, 230-235.	1.1	48
49	Type III collagen affects dermal and vascular collagen fibrillogenesis and tissue integrity in a mutant Col3a1 transgenic mouse model. Matrix Biology, 2018, 70, 72-83.	3.6	48
50	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, .	11.9	45
51	Mutations in PLOD3, encoding lysyl hydroxylase 3, cause a complex connective tissue disorder including recessive dystrophic epidermolysis bullosa-like blistering phenotype with abnormal anchoring fibrils and type VII collagen deficiency. Matrix Biology, 2019, 81, 91-106.	3.6	45
52	Low tendon stiffness and abnormal ultrastructure distinguish classic Ehlersâ€Danlos syndrome from benign joint hypermobility syndrome in patients. FASEB Journal, 2014, 28, 4668-4676.	0.5	44
53	Defective Proteolytic Processing of Fibrillar Procollagens and Prodecorin Due to Biallelic BMP1 Mutations Results in a Severe, Progressive Form of Osteogenesis Imperfecta. Journal of Bone and Mineral Research, 2015, 30, 1445-1456.	2.8	42
54	Genetic Defects in TAPT1 Disrupt Ciliogenesis and Cause a Complex Lethal Osteochondrodysplasia. American Journal of Human Genetics, 2015, 97, 521-534.	6.2	39

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55	Type I Procollagen C-Propeptide Defects: Study of Genotype-Phenotype Correlation and Predictive Role of Crystal Structure. Human Mutation, 2014, 35, n/a-n/a.	2.5	38
56	Expanding the clinical and mutational spectrum of the Ehlersâ€“Danlos syndrome, dermatosparaxis type. Genetics in Medicine, 2016, 18, 882-891.	2.4	37
57	P4HA1 mutations cause a unique congenital disorder of connective tissue involving tendon, bone, muscle and the eye. Human Molecular Genetics, 2017, 26, 2207-2217.	2.9	37
58	A Novel Splice Variant in the N-propeptide of COL5A1 Causes an EDS Phenotype with Severe Kyphoscoliosis and Eye Involvement. PLoS ONE, 2011, 6, e20121.	2.5	36
59	Orthostatic intolerance and fatigue in the hypermobility type of Ehlers-Danlos Syndrome. Rheumatology, 2016, 55, 1412-1420.	1.9	35
60	The impact of COVID-19 on rare and complex connective tissue diseases: the experience of ERN ReCONNET. Nature Reviews Rheumatology, 2021, 17, 177-184.	8.0	35
61	Copper Deficiency in Patients with Cystinosis with Cysteamine Toxicity. Journal of Pediatrics, 2013, 163, 754-760.	1.8	34
62	Biallelic B3GALT6 mutations cause spondylodysplastic Ehlersâ€“Danlos syndrome. Human Molecular Genetics, 2018, 27, 3475-3487.	2.9	34
63	Twenty patients including 7 probands with autosomal dominant cutis laxa confirm clinical and molecular homogeneity. Orphanet Journal of Rare Diseases, 2013, 8, 36.	2.7	33
64	Novel defects in collagen XII and VI expand the mixed myopathy/Ehlersâ€“Danlos syndrome spectrum and lead to variant-specific alterations in the extracellular matrix. Genetics in Medicine, 2020, 22, 112-123.	2.4	33
65	Compound heterozygous mutations of the TNXB gene cause primary myopathy. Neuromuscular Disorders, 2013, 23, 664-669.	0.6	32
66	Bi-allelic AEBP1 mutations in two patients with Ehlersâ€“Danlos syndrome. Human Molecular Genetics, 2019, 28, 1853-1864.	2.9	29
67	Ehlers-Danlos Syndrome, Hypermobility Type, Is Linked to Chromosome 8p22-8p21.1 in an Extended Belgian Family. Disease Markers, 2015, 2015, 1-9.	1.3	28
68	COL1â€related overlap disorder: A novel connective tissue disorder incorporating the osteogenesis imperfecta/Ehlersâ€Danlos syndrome overlap. Clinical Genetics, 2020, 97, 396-406.	2.0	27
69	Analysis of chromosomal radiosensitivity of healthy BRCA2 mutation carriers and non-carriers in BRCA families with the G2 micronucleus assay. Oncology Reports, 2017, 37, 1379-1386.	2.6	26
70	Genetic analysis of osteogenesis imperfecta in the <scp>P</scp>alestinian population: molecular screening of 49 affected families. Molecular Genetics & Genomic Medicine, 2018, 6, 15-26.	1.2	26
71	Increased chromosomal radiosensitivity in asymptomatic carriers of a heterozygous BRCA1 mutation. Breast Cancer Research, 2016, 18, 52.	5.0	25
72	Updating the Evidence on Functional Capacity Evaluation Methods: A Systematic Review. Journal of Occupational Rehabilitation, 2018, 28, 418-428.	2.2	23

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73	Ehlers-Danlos syndromes: state of the art on clinical practice guidelines. RMD Open, 2018, 4, e000790.	3.8	23
74	A homozygous pathogenic missense variant broadens the phenotypic and mutational spectrum of CREB3L1-related osteogenesis imperfecta. Human Molecular Genetics, 2019, 28, 1801-1809.	2.9	21
75	Loss of <scp>TANGO1</scp> Leads to Absence of Bone Mineralization. JBMR Plus, 2021, 5, e10451.	2.7	21
76	Pain in the Ehlersâ€“Danlos syndromes: Mechanisms, models, and challenges. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2021, 187, 429-445.	1.6	21
77	Retinitis Pigmentosa, Cutis Laxa, and Pseudoxanthoma Elasticumâ€“Like Skin Manifestations Associated with GGCX Mutations. Journal of Investigative Dermatology, 2014, 134, 2331-2338.	0.7	20
78	Knowledge, assessment, and management of adults with joint hypermobility syndrome/Ehlersâ€“Danlos syndrome hypermobility type among flemish physiotherapists. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2015, 169, 76-83.	1.6	19
79	Hypomorphic zebrafish models mimic the musculoskeletal phenotype of Î²4GalT7-deficient Ehlers-Danlos syndrome. Matrix Biology, 2020, 89, 59-75.	3.6	19
80	More than meets the eye: Expanding and reviewing the clinical and mutational spectrum of brittle cornea syndrome. Human Mutation, 2021, 42, 711-730.	2.5	19
81	Eight years experience from a skeletal dysplasia referral center in a tertiary hospital in Southern India: A model for the diagnosis and treatment of rare diseases in a developing country. American Journal of Medical Genetics, Part A, 2014, 164, 2317-2323.	1.2	18
82	Discriminative Features in Three Autosomal Recessive Cutis Laxa Syndromes: Cutis Laxa IIA, Cutis Laxa IIB, and Geroderma Osteoplastica. International Journal of Molecular Sciences, 2017, 18, 635.	4.1	18
83	Delineation of musculocontractural Ehlersâ€“Danlos Syndrome caused by dermatan sulfate epimerase deficiency. Molecular Genetics & Genomic Medicine, 2020, 8, e1197.	1.2	18
84	Clinical and molecular features of 66 patients with musculocontractural Ehlersâ€'Danlos syndrome caused by pathogenic variants in CHST14 (mcEDS-CHST14). Journal of Medical Genetics, 2021, , jmedgenet-2020-107623.	3.2	18
85	Hypermobility, the Ehlers-Danlos syndromes and chronic pain. Clinical and Experimental Rheumatology, 2017, 35 Suppl 107, 116-122.	0.8	17
86	A Novel Mutation in the Lysyl Hydroxylase 1 Gene Causes Decreased Lysyl Hydroxylase Activity in an Ehlersâ€“Danlos VIA Patient. Journal of Investigative Dermatology, 2005, 124, 914-918.	0.7	16
87	Ptosis as a unique hallmark for autosomal recessive WNT1â€associated osteogenesis imperfecta. American Journal of Medical Genetics, Part A, 2019, 179, 908-914.	1.2	16
88	The effect of five isometric exercises on glenohumeral translations in healthy subjects and patients with the hypermobility type of the ehlers-danlos syndrome (heds) or hypermobility spectrum disorder (hsd) with multidirectional shoulder instability: an observational study. Physiotherapy, 2020, 107, 11-18.	0.4	15
89	Four decades in the making: Collagen III and mechanisms of vascular Ehlers Danlos Syndrome. Matrix Biology Plus, 2021, 12, 100090.	3.5	15
90	Tissueâ€specific mosaicism for a lethal osteogenesis imperfecta COL1A1 mutation causes mild OI/EDS overlap syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 1047-1050.	1.2	13

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91	The influence of Ehlers-Danlos syndrome â€“ hypermobility type, on motherhood: A phenomenological, hermeneutical study. Research in Developmental Disabilities, 2017, 60, 135-144.	2.2	13
92	Pain-related behaviors and abnormal cutaneous innervation in a murine model of classical Ehlersâ€“Danlos syndrome. Pain, 2020, 161, 2274-2283.	4.2	13
93	Auditory phenotype in Stickler syndrome: results of audiometric analysis in 20 patients. European Archives of Oto-Rhino-Laryngology, 2016, 273, 3025-3034.	1.6	12
94	Clinical and molecular characteristics of 168 probands and 65 relatives with a clinical presentation of classical Ehlersâ€“Danlos syndrome. Human Mutation, 2021, 42, 1294-1306.	2.5	12
95	The added value of a European Reference Network on rare and complex connective tissue and musculoskeletal diseases: insights after the first 5 years of the ERN ReCONNET. Clinical and Experimental Rheumatology, 2022, 40, 3-11.	0.8	12
96	b3galt6 Knock-Out Zebrafish Recapitulate Î²3GalT6-Deficiency Disorders in Human and Reveal a Trisaccharide Proteoglycan Linkage Region. Frontiers in Cell and Developmental Biology, 2020, 8, 597857.	3.7	11
97	Animal Models of Ehlersâ€“Danlos Syndromes: Phenotype, Pathogenesis, and Translational Potential. Frontiers in Genetics, 2021, 12, 726474.	2.3	11
98	The clinical and mutational spectrum of B3GAT3 linkeropathy: two case reports and literature review. Orphanet Journal of Rare Diseases, 2019, 14, 138.	2.7	9
99	Muscle activity and scapular kinematics in individuals with multidirectional shoulder instability: A systematic review. Annals of Physical and Rehabilitation Medicine, 2021, 64, 101457.	2.3	9
100	Aberrant binding of mutant HSP47 affects posttranslational modification of type I collagen and leads to osteogenesis imperfecta. PLoS Genetics, 2021, 17, e1009339.	3.5	9
101	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.	0.8	9
102	RIN2 syndrome: Expanding the clinical phenotype. American Journal of Medical Genetics, Part A, 2016, 170, 2408-2415.	1.2	8
103	Does muscle strength change over time in patients with hypermobile Ehlers-Danlos syndrome/ Hypermobility Spectrum Disorder? An 8-year follow-up study. Arthritis Care and Research, 2020, 73, 1041-1048.	3.4	8
104	Home-based exercise therapy for treating shoulder instability in patients with hypermobile Ehlers-Danlos syndrome/hypermobility spectrum disorders. A randomized trial. Disability and Rehabilitation, 2023, 45, 1811-1821.	1.8	7
105	Patient perspectives on employment participation in the â€œhypermobile Ehlersâ€“Danlos syndromeâ€• Disability and Rehabilitation, 2021, 43, 668-677.	1.8	6
106	Cephalometrics in Stickler syndrome: Objectification of the typical facial appearance. Journal of Cranio-Maxillo-Facial Surgery, 2016, 44, 848-853.	1.7	5
107	Electromyographic Muscle Activity and <scp>Threeâ€Dimensional</scp> Scapular Kinematics in Patients With Multidirectional Shoulder Instability: A Study in the Hypermobile Type of the <scp>Ehlersâ€Danlos</scp> Syndrome and the Hypermobility Spectrum Disorders. Arthritis Care and Research, 2022, 74, 833-840.	3.4	5
108	Altered Multisegment Ankle and Foot Kinematics During Gait in Patients With Hypermobile <scp>Ehlersâ€Danlos</scp> Syndrome/Hypermobility Spectrum Disorder: A Caseâ€“Control Study. Arthritis Care and Research, 2022, 74, 841-848.	3.4	5

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109	DUP25 remains unconfirmed. American Journal of Medical Genetics Part A, 2004, 131A, 320-321.	2.4	4
110	The Ehlersâ€“Danlos Syndromes against the Backdrop of Inborn Errors of Metabolism. Genes, 2022, 13, 265.	2.4	4
111	Glycosaminoglycan linkage region of urinary bikunin as a potentially useful biomarker for <scp>Î²3GalT6</scp>â€deficient spondylodysplastic <scp>Ehlersâ€“Danlos</scp> syndrome. JIMD Reports, 2022, 63, 462-467.	1.5	4
112	Genetics of the Ehlersâ€“Danlos syndrome: more than collagen disorders. Expert Opinion on Orphan Drugs, 2015, 3, 379-392.	0.8	3
113	Biallelic variants in MESD, which encodes a WNT-signaling-related protein, in four new families with recessively inherited osteogenesis imperfecta. Human Genetics and Genomics Advances, 2021, 2, 100051.	1.7	3
114	Exploring pain mechanisms in hypermobile <scp>Ehlersâ€Danlos</scp> syndrome: A caseâ€“control study. European Journal of Pain, 2022, 26, 1355-1367.	2.8	3
115	New insights on the clinical variability of FKBP10 mutations. European Journal of Medical Genetics, 2020, 63, 103980.	1.3	2
116	Societal participation in ehlers-danlos syndromes and hypermobility spectrum disorder, compared to fibromyalgia and healthy controls. PLoS ONE, 2022, 17, e0269608.	2.5	2
117	Foot kinematics in the hypermobility type of Ehlersâ€“Danlos syndrome using the Ghent Foot Model. Footwear Science, 2017, 9, S118-S120.	2.1	1
118	Collagens in the Physiopathology of the Ehlersâ€“Danlos Syndromes. Biology of Extracellular Matrix, 2021, , 55-119.	0.3	1
119	Caffey disease is associated with distinct arginine to cysteine substitutions in the proÎ±1(I) chain of type I procollagen. Genetics in Medicine, 2021, 23, 2378-2385.	2.4	1
120	Physical activity and sleep in patients with hypermobile Ehlersâ€“Danlos syndrome and patients with generalized hypermobility spectrum disorder. Edorium Journal of Disability and Rehabilitation, 2020, 6, 1.	0.3	1
121	The Impact of Hypermobile â€œEhlers-Danlos Syndromeâ€•and Hypermobile Spectrum Disorder on Interpersonal Interactions and Relationships. Frontiers in Rehabilitation Sciences, 2022, 3, .	1.2	1
122	Muscle Strength, Muscle Mass and Physical Impairment in Women with hypermobile Ehlers-Danlos syndrome and Hypermobility Spectrum Disorder.. Journal of Musculoskeletal Neuronal Interactions, 2022, 22, 5-14.	0.1	1
123	Short stature, severe aortic root dilation, skin hyperextensibility, extreme joint laxity and craniofacial dysmorphic features: a probable new syndrome. Clinical Dysmorphology, 2010, 19, 119-122.	0.3	0
124	Reply to the letter to the editor by Marc Williams. Genetics in Medicine, 2011, 13, 77-78.	2.4	0
125	Compound heterozygous mutations of the TNXB gene cause primary myopathy. Neuromuscular Disorders, 2014, 24, 89.	0.6	0
126	FRI0607â€…IDENTIFICATION OF UNMET NEEDS RELATED TO RARE AND COMPLEX CONNECTIVE TISSUE AND MUSCULOSKELETAL DISEASES (RCTDS) ACROSS EU: THE EXPERIENCE OF THE ERN RECONNET. , 2019, , .		0

#	Article	IF	Citations
127	The Ehlersâ€”Danlos Syndrome. , 2009, , 467-475.		0
128	The added value of a European Reference Network on rare and complex connective tissue and musculoskeletal diseases: insights after the first 5 years of the ERN ReCONNET.. Clinical and Experimental Rheumatology, 2022, , .	0.8	0
129	Editorial: Molecular Mechanisms of Heritable Connective Tissue Disorders. Frontiers in Genetics, 2022, 13, 866665.	2.3	0