Clair A Francomano

List of Publications by Year
in descending order

130 papers	11,590 citations	41258 49 h-index	29081 104 g-index
134 all docs	134 docs citations	134 times ranked	9145 citing authors

#	Article	IF	Citations
1	Longitudinal analysis of symptoms in the <scp>Ehlersâ€Danlos</scp> syndromes. American Journal of Medical Genetics, Part A, 2022, 188, 1204-1213.	0.7	4
2	Hereditary alpha-tryptasemia modifies clinical phenotypes among individuals with congenital hypermobility disorders. Human Genetics and Genomics Advances, 2022, 3, 100094.	1.0	1
3	Atlanto-axial rotary instability (Fielding type 1): characteristic clinical and radiological findings, and treatment outcomes following alignment, fusion, and stabilization. Neurosurgical Review, 2021, 44, 1553-1568.	1.2	13
4	Letter to the editor regarding â€œAtlantoaxial dislocation due to os odontoideum in patients with Downâ€™s syndrome: literature review and case reportsâ€. Child's Nervous System, 2021, 37, 1041-1043.	0.6	0
5	The long road to rare disease diagnosis: a heroâ€™s journey. Molecular Genetics and Metabolism, 2021, 132, S297.	0.5	0
6	Prescription claims for immunomodulator and anti-inflammatory medications among persons with Ehlers-Danlos syndromes. Molecular Genetics and Metabolism, 2021, 132, S104-S105.	0.5	0
7	P084â€ƒEDS ECHO: virtual case based learning in the assessment and management of Ehlers-Danlos syndrome and hypermobility spectrum disorders. Rheumatology, 2021, 60, .	0.9	2
8	Refractory Syncope and Presyncope Associated with Atlantoaxial Instability: Preliminary Evidence of Improvement Following Surgical Stabilization. World Neurosurgery, 2021, 149, e854-e865.	0.7	7
9	Victor Almon McKusick : In the footsteps of Mendel and Osler. American Journal of Medical Genetics, Part A, 2021, 185, 3193-3201.	0.7	2
10	Patients with <scp>Ehlersâ€“Danlos</scp> syndrome on the diagnostic odyssey: Rethinking complexity and difficulty as a hero's journey. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2021, 187, 416-424.	0.7	23
11	Gastrointestinal medication burden among persons with the Ehlersâ€Danlos syndromes. Neurogastroenterology and Motility, 2021, 33, e14077.	1.6	6
12	The power of patientâ€led global collaboration. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2021, 187, 425-428.	0.7	2
13	Initial description and evaluation of EDS ECHO: An international effort to improve care for people with the Ehlersâ€Danlos syndromes and hypermobility spectrum disorders. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2021, 187, 609-615.	0.7	10
14	A caseâ€“control study of respiratory medication and coâ€occurring gastrointestinal prescription burden among persons with Ehlersâ€“Danlos syndromes. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2021, 187, 549-560.	0.7	0
15	Respiratory manifestations in the <scp>Ehlersâ€“Danlos</scp> syndromes. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2021, 187, 533-548.	0.7	8
16	<scp>Ehlersâ€“Danlos</scp> syndromes, hypermobility spectrum disorders, and associated <scp>coâ€morbidities</scp>: Reports from <scp>EDS ECHO</scp>. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2021, 187, 413-415.	0.7	17
17	Prescription Claims for Immunomodulator and <scp>Antiâ€Inflammatory</scp> Drugs among Persons with <scp>Ehlersâ€Danlos</scp> Syndromes. Arthritis Care and Research, 2021, , .	1.5	1
18	Defining renal phenotype in AlstrÃ¶m syndrome. Nephrology Dialysis Transplantation, 2020, 35, 994-1001.	0.4	20

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19	Arterial Elasticity in Ehlers-Danlos Syndromes. Genes, 2020, 11, 55.	1.0	13
20	The Ehlersâ€“Danlos syndromes. Nature Reviews Disease Primers, 2020, 6, 64.	18.1	144
21	Consensus clinical management guidelines for AlstrÃ¶m syndrome. Orphanet Journal of Rare Diseases, 2020, 15, 253.	1.2	49
22	Prevalence of hypermobile Ehlers-Danlos syndrome in postural orthostatic tachycardia syndrome. Autonomic Neuroscience: Basic and Clinical, 2020, 224, 102637.	1.4	32
23	Survey of Ehlersâ€'Danlos Patientsâ€™ ophthalmic surgery experiences. Molecular Genetics & Genomic Medicine, 2020, 8, e1155.	0.6	13
24	Factors affecting quality of life in children and adolescents with hypermobile Ehlersâ€Danlos syndrome/hypermobility spectrum disorders. American Journal of Medical Genetics, Part A, 2019, 179, 561-569.	0.7	39
25	Heritable disorders of connective tissue: Description of a data repository and initial cohort characterization. American Journal of Medical Genetics, Part A, 2019, 179, 552-560.	0.7	19
26	Use of Cluster Analysis to Delineate Symptom Profiles in an Ehlers-Danlos Syndrome Patient Population. Journal of Pain and Symptom Management, 2019, 58, 427-436.	0.6	12
27	Resistance to local anesthesia in people with the Ehlers-Danlos Syndromes presenting for dental surgery. Journal of Dental Anesthesia and Pain Medicine, 2019, 19, 261.	0.4	25
28	Use of prescription opioid and other drugs among a cohort of persons with Ehlersâ€“Danlos syndrome: A retrospective study. American Journal of Medical Genetics, Part A, 2019, 179, 397-403.	0.7	16
29	Postural tachycardia syndrome and other forms of orthostatic intolerance in Ehlers-Danlos syndrome. Autonomic Neuroscience: Basic and Clinical, 2018, 215, 89-96.	1.4	73
30	Bi-allelic Alterations in AEBP1 Lead to Defective Collagen Assembly and Connective Tissue Structure Resulting in a Variant of Ehlers-Danlos Syndrome. American Journal of Human Genetics, 2018, 102, 696-705.	2.6	105
31	Characteristics, Diagnosis, and Management of Ehlers-Danlos Syndromes. JAMA Facial Plastic Surgery, 2018, 20, 70-75.	2.2	34
32	Pain and sleep quality in children with nonâ€vascular Ehlersâ€“Danlos syndromes. American Journal of Medical Genetics, Part A, 2018, 176, 1858-1864.	0.7	12
33	Neurological and spinal manifestations of the Ehlersâ€“Danlos syndromes. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2017, 175, 195-211.	0.7	157
34	Pain management in the Ehlersâ€“Danlos syndromes. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2017, 175, 212-219.	0.7	116
35	The 2017 international classification of the Ehlersâ€“Danlos syndromes. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2017, 175, 8-26.	0.7	1,163
36	Clinical and biochemical profiles suggest fibromuscular dysplasia is a systemic disease with altered TGFâ€Î²2 expression and connective tissue features. FASEB Journal, 2014, 28, 3313-3324.	0.2	68

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37	Victor A. McKusick and Medical Genetics Among the Amish. , 2012, , 119-130.		2
38	Occipito-atlanto-axial Hypermobility : Clinical Features and Dynamic Analysis of Cranial Settling and Posterior Gliding of Occipital Condyle. Part 2 : Findings in Patients with Post-traumatic Condition. Spinal Surgery, 2009, 23, 176-182.	0.0	0
39	Occipito-atlanto-axial Hypermobility : Clinical Features and Dynamic Analysis of Cranial Settling and Posterior Gliding of Occipital Condyle. Part 1 : Findings in Patients with Hereditary Disorders of Connective Tissue and Ehlers-Danlos Syndrome. Spinal Surgery, 2009, 23, 168-175.	0.0	0
40	Association of Chiari malformation type I and tethered cord syndrome: preliminary results of sectioning filum terminale. World Neurosurgery, 2009, 72, 20-35.	1.3	117
41	Tethered Cord Syndrome : Preliminary Report of Clinical Features and Morphometric Analysis on Association of Chiari Malformation Type â.... Spinal Surgery, 2009, 23, 195-203.	0.0	1
42	Syndrome of occipitoatlantoaxial hypermobility, cranial settling, and Chiari malformation Type I in patients with hereditary disorders of connective tissue. Journal of Neurosurgery: Spine, 2007, 7, 601-609.	0.9	192
43	Growth and integration of neocartilage with native cartilage in vitro. Journal of Orthopaedic Research, 2005, 23, 433-439.	1.2	29
44	Complementary Medicine and Genetic Medicine: Polar Disciplines or Dynamic Partners?. Journal of Alternative and Complementary Medicine, 2005, 11, 343-347.	2.1	4
45	Hyaline cartilage engineered by chondrocytes in pellet culture: histological, immunohistochemical and ultrastructural analysis in comparison with cartilage explants. Journal of Anatomy, 2004, 205, 229-237.	0.9	117
46	Klotho gene variation and expression in 20 inbred mouse strains. Mammalian Genome, 2004, 15, 759-767.	1.0	11
47	Medical genetic studies in the Amish: Historical perspective. American Journal of Medical Genetics Part A, 2003, 121C, 1-4.	2.4	27
48	The major mutation in theRMRPgene causing CHH among the Amish is the same as that found in most Finnish cases. , 2003, 121C, 81-83.		40
49	The effects of pulsed low-intensity ultrasound on chondrocyte viability, proliferation, gene expression and matrix production. Ultrasound in Medicine and Biology, 2003, 29, 1645-1651.	0.7	125
50	A central nervous system specific mouse model for thanatophoric dysplasia type II. Human Molecular Genetics, 2003, 12, 2863-2871.	1.4	30
51	The Stickler syndrome: Genotype/phenotype correlation in 10 families with Stickler syndrome resulting from seven mutations in the type II collagen gene locus COL2A1. Genetics in Medicine, 2003, 5, 21-27.	1.1	83
52	Case 47: Dural Ectasia Associated with Marfan Syndrome. Radiology, 2002, 223, 767-771.	3.6	25
53	Gene Expression Profile of Human Bone Marrow Stromal Cells: High-Throughput Expressed Sequence Tag Sequencing Analysis. Genomics, 2002, 79, 7-17.	1.3	51
54	Structural and Functional Mutations of the Perlecan Gene Cause Schwartz-Jampel Syndrome, with Myotonic Myopathy and Chondrodysplasia. American Journal of Human Genetics, 2002, 70, 1368-1375.	2.6	168

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55	The influence of pulsed low-intensity ultrasound on matrix production of chondrocytes at different stages of differentiation: an explant study. Ultrasound in Medicine and Biology, 2002, 28, 1547-1553.	0.7	72
56	Issues surrounding prenatal genetic testing for achondroplasia. Prenatal Diagnosis, 2002, 22, 933-940.	1.1	29
57	Case 47. Radiology, 2002, 222, 513-514.	3.6	0
58	Characterization of a Human Gene Encoding Nucleosomal Binding Protein NSBP1. Genomics, 2001, 71, 163-173.	1.3	40
59	Thoracolumbar Spinal Abnormalities in Stickler Syndrome. Spine, 2001, 26, 403-409.	1.0	53
60	Comprehensive resource: Skeletal gene database. American Journal of Medical Genetics Part A, 2001, 106, 275-281.	2.4	7
61	A comparison of the Berlin and Ghent nosologies and the influence of dural ectasia in the diagnosis of Marfan syndrome. Genetics in Medicine, 2000, 2, 278-282.	1.1	45
62	Jeune asphyxiating thoracic dystrophy and short-rib polydactyly type III (Verma-Naumoff) are variants of the same disorder. , 2000, 90, 310-314.		48
63	Severe hypodontia and oral xanthomas in Alagille syndrome. American Journal of Medical Genetics Part A, 2000, 93, 250-252.	2.4	18
64	Rapid determination of COL2A1 mutations in individuals with Stickler syndrome: Analysis of potential premature termination codons. American Journal of Medical Genetics Part A, 2000, 94, 141-148.	2.4	28
65	Mutations in a new gene in Ellis-van Creveld syndrome and Weyers acrodental dysostosis. Nature Genetics, 2000, 24, 283-286.	9.4	323
66	A Skeletal Gene Database. Journal of Bone and Mineral Research, 2000, 15, 2095-2122.	3.1	50
67	Minocycline-induced generalized postinflammatory elastolysis. American Journal of Medicine, 2000, 109, 340.	0.6	8
68	Recommendations for national and local regulatory authorities concerning research in genetic markers of disease. American Heart Journal, 2000, 140, S3-S5.	1.2	0
69	A Novel Nemaline Myopathy in the Amish Caused by a Mutation in Troponin T1. American Journal of Human Genetics, 2000, 67, 814-821.	2.6	300
70	Distinct Missense Mutations of the FGFR3 Lys650 Codon Modulate Receptor Kinase Activation and the Severity of the Skeletal Dysplasia Phenotype. American Journal of Human Genetics, 2000, 67, 1411-1421.	2.6	154
71	The Molecular and Genetic Basis of Fibroblast Growth Factor Receptor 3 Disorders: The Achondroplasia Family of Skeletal Dysplasias, Muenke Craniosynostosis, and Crouzon Syndrome with Acanthosis Nigricans*. Endocrine Reviews, 2000, 21, 23-39.	8.9	262
72	Heterozygous mutations in the gene encoding noggin affect human joint morphogenesis. Nature Genetics, 1999, 21, 302-304.	9.4	329

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73	Mutations of CTSK Result in Pycnodysostosis via a Reduction in Cathepsin K Protein. Journal of Bone and Mineral Research, 1999, 14, 1649-1653.	3.1	49
74	Conservation of the Caenorhabditis elegans timing gene clk-1 from yeast to human: a gene required for ubiquinone biosynthesis with potential implications for aging. Mammalian Genome, 1999, 10, 1000-1004.	1.0	76
75	Severe achondroplasia with developmental delay and acanthosis nigricans (SADDAN): Phenotypic analysis of a new skeletal dysplasia caused by a Lys650Met mutation in fibroblast growth factor receptor 3. American Journal of Medical Genetics Part A, 1999, 85, 53-65.	2.4	106
76	Small deletions in the type II collagen triple helix produce Kniest dysplasia. , 1999, 85, 105-112.		59
77	A Novel Skeletal Dysplasia with Developmental Delay and Acanthosis Nigricans Is Caused by a Lys650Met Mutation in the Fibroblast Growth Factor Receptor 3 Gene. American Journal of Human Genetics, 1999, 64, 722-731.	2.6	151
78	Characterization of the Human Talin (TLN) Gene: Genomic Structure, Chromosomal Localization, and Expression Pattern. Genomics, 1999, 62, 316-319.	1.3	15
79	Small deletions in the type II collagen triple helix produce Kniest dysplasia. , 1999, 85, 105.		1
80	Physical mapping of the nail patella syndrome interval at 9q34: ordering of STSs and ESTs. Human Genetics, 1998, 103, 525-526.	1.8	8
81	Sleep-disordered breathing in children with achondroplasia. Journal of Pediatrics, 1998, 132, 667-671.	0.9	90
82	Mutations in Fibroblast Growth-Factor Receptor 3 in Sporadic Cases of Achondroplasia Occur Exclusively on the Paternally Derived Chromosome. American Journal of Human Genetics, 1998, 63, 711-716.	2.6	206
83	Multiple Molecular Mechanisms Underlying Subdiagnostic Variants of Marfan Syndrome. American Journal of Human Genetics, 1998, 63, 1703-1711.	2.6	85
84	Mutation Analysis of LMX1B Gene in Nail-Patella Syndrome Patients. American Journal of Human Genetics, 1998, 63, 1651-1658.	2.6	166
85	Software for Constructing and Verifying Pedigrees within Large Genealogies and an Application to the Old Order Amish of Lancasterâ€‰County. Genome Research, 1998, 8, 211-221.	2.4	102
86	Genetic and physical mapping of the McKusick-Kaufman syndrome. Human Molecular Genetics, 1998, 7, 475-481.	1.4	46
87	Hypochondroplasia: Molecular Analysis of the Fibroblast Growth Factor Receptor 3 Gene. Annals of the New York Academy of Sciences, 1996, 785, 182-187.	1.8	27
88	The Gene for the Ellisâ€“van Creveld Syndrome Is Located on Chromosome 4p16. Genomics, 1996, 35, 1-5.	1.3	117
89	Brachydactyly Type C Gene Maps to Human Chromosome 12q24. Genomics, 1996, 38, 45-50.	1.3	16
90	Bone dysplasias in man: molecular insights. Current Opinion in Genetics and Development, 1996, 6, 301-308.	1.5	41

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91	Exclusion of the MSX1 homeobox gene as the gene for the Ellis van Creveld syndrome in the Amish. Human Genetics, 1996, 98, 572-575.	1.8	14
92	Progress in medical genetics: Map-based gene discovery and the molecular pathology of skeletal dysplasias. , 1996, 63, 98-105.		16
93	Identical mutations in three different fibroblast growth factor receptor genes in autosomal dominant craniosynostosis syndromes. Nature Genetics, 1996, 14, 174-176.	9.4	306
94	Association Study of Transforming Growth Factor Alpha (TGFÎ±) TaqI Polymorphismand Oral Clefts: Indication of Gene-Environment Interaction in a Population-based Sample of Infants with Birth Defects. American Journal of Epidemiology, 1995, 141, 629-636.	1.6	226
95	Confirmatory linkage of hypochondroplasia to chromosome arm 4p. American Journal of Medical Genetics Part A, 1995, 57, 505-506.	2.4	7
96	Concentration of mutations causing schmid metaphyseal chondrodysplasia in the C-terminal noncollagenous domain of type X collagen. Human Mutation, 1995, 5, 121-125.	1.1	48
97	Key role for a minor collagen. Nature Genetics, 1995, 9, 6-8.	9.4	27
98	The question of heterogeneity in Marfan syndrome. Nature Genetics, 1995, 9, 228-229.	9.4	51
99	The gene for pycnodysostosis maps to human chromosome 1cenâ€“q21. Nature Genetics, 1995, 10, 238-239.	9.4	51
100	Mutations in exon 17B of cartilage oligomeric matrix protein (COMP) cause pseudoachondroplasia. Nature Genetics, 1995, 10, 325-329.	9.4	368
101	A recurrent mutation in the tyrosine kinase domain of fibroblast growth factor receptor 3 causes hypochondroplasia. Nature Genetics, 1995, 10, 357-359.	9.4	443
102	The Genetic Basis of Dwarfism. New England Journal of Medicine, 1995, 332, 58-59.	13.9	55
103	COL5a1: fine genetic mapping and exclusion as candidate gene in families with nail-patella syndrome, tuberous sclerosis 1, hereditary hemorrhagic telangiectasia, and Ehlersâ€”Danlos syndrome type II. Genomics, 1995, 25, 737-739.	1.3	36
104	Localization of the Gene (SYM1) for Proximal Symphalangism to Human Chromosome 17q21-q22. Genomics, 1995, 27, 225-229.	1.3	42
105	The status of online Mendelian inheritance in man (OMIM) medio 1994. Nucleic Acids Research, 1994, 22, 3470-3473.	6.5	66
106	Substitution of a cysteine residue in a non-calcium binding, EGF-like domain of fibrillin segregates with the Marfan syndrome in a large kindred. Human Molecular Genetics, 1994, 3, 1013-1014.	1.4	12
107	Additional mutations of type X collagen confirm COL10A1 as the Schmid metaphyseal chondrodysplasia locus. Human Molecular Genetics, 1994, 3, 303-307.	1.4	54
108	High-Resolution Genetic Mapping of the Cartilage-Hair Hypoplasia (CHH) Gene in Amish and Finnish Families. Genomics, 1994, 20, 347-353.	1.3	37

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109	Presymptomatic direct detection of adenomatous polyposis coli (APC) gene mutations in familial adenomatous polyposis. Human Genetics, 1993, 91, 307-11.	1.8	38
110	A type X collagen mutation causes Schmid metaphyseal chondrodysplasia. Nature Genetics, 1993, 5, 79-82.	9.4	253
111	Four Novel FBN1 Mutations: Significance for Mutant Transcript Level and EGF-like Domain Calcium Binding in the Pathogenesis of Marfan Syndrome. Genomics, 1993, 17, 468-475.	1.3	293
112	Linkage of typical pseudoachondroplasia to chromosome 19. Genomics, 1993, 18, 661-666.	1.3	57
113	Molecular Genetic Analysis in Autosomal Dominant Keratoconus. Cornea, 1992, 11, 302-308.	0.9	73
114	Dextromethorphan and high-dose benzoate therapy for nonketotic hyperglycinemia in an infant. Journal of Pediatrics, 1992, 121, 131-135.	0.9	84
115	Mutations in the fibrillin gene and variability of the Marfan syndrome. Journal of Molecular and Cellular Cardiology, 1992, 24, S76.	0.9	0
116	Diagnostic approaches to renal genetic disorders using DNA analysis. Pediatric Nephrology, 1992, 6, 113-118.	0.9	0
117	Clustering of fibrillin (FBN1) missense mutations in Marfan syndrome patients at cysteine residues in EGF-like domains. Human Mutation, 1992, 1, 366-374.	1.1	131
118	Exclusion of human proteoglycan link protein (CRTL1) and type II collagen (COL2A1) genes in pseudoachondroplasia. American Journal of Medical Genetics Part A, 1992, 44, 420-424.	2.4	12
119	The Marfan syndrome locus: Confirmation of assignment to chromosome 15 and identification of tightly linked markers at 15q15-q21.3. Genomics, 1991, 9, 355-361.	1.3	185
120	Marfan syndrome caused by a recurrent de novo missense mutation in the fibrillin gene. Nature, 1991, 352, 337-339.	13.7	1,901
121	The COL6A1 and COL6A2 genes exist as a gene cluster and detect highly informative DNA polymorphisms in the telomeric region of human chromosome 21q. Human Genetics, 1991, 87, 162-166.	1.8	32
122	Craniocervical decompression for cervicomedullary compression in pediatric patients with achondroplasia. Journal of Neurosurgery, 1990, 73, 375-382.	0.9	70
123	Use of denaturing gradient gel electrophoresis for detection of mutation and prospective diagnosis in late onset ornithine transcarbamylase deficiency. Genomics, 1990, 7, 167-172.	1.3	38
124	Marfan Syndrome: Exclusion of genetic linkage to three major collagen genes. American Journal of Medical Genetics Part A, 1988, 29, 457-462.	2.4	30
125	Achondropiasia is not caused by mutation in the gene for type II collagen. American Journal of Medical Genetics Part A, 1988, 29, 955-961.	2.4	16
126	Therapeutic approaches to cobalamin-C methylmalonic acidemia and homocystinuria. Journal of Pediatrics, 1988, 112, 32-39.	0.9	83

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127	Molecular Genetic Studies in Achondroplasia. , 1988, 48, 53-58.		3
128	Partial structure of the human ?2(IV) collagen chain and chromosomal localization of the gene (COL4A2). Human Genetics, 1987, 77, 318-324.	1.8	36
129	DNA Analysis in Genetic Disorders. Annual Review of Medicine, 1986, 37, 377-395.	5.0	19
130	Isolation and partial characterization of genomic clones coding for a human pro-.alpha.1(II) collagen chain and demonstration of restriction fragment length polymorphism at the 3' end of the gene. Biochemistry, 1985, 24, 6343-6348.	1.2	20