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	Marfan syndrome caused by a recurrent de novo missense mutation in the fibrillin gene. Nature, 1991, 352, 337-339.	13.7	1,901
2	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
3	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
4	Mutations in exon 17B of cartilage oligomeric matrix protein (COMP) cause pseudoachondroplasia. Nature Genetics, 1995, 10, 325-329.	9.4	368
5	Heterozygous mutations in the gene encoding noggin affect human joint morphogenesis. Nature Genetics, 1999, 21, 302-304.	9.4	329
6	Mutations in a new gene in Ellis-van Creveld syndrome and Weyers acrodental dysostosis. Nature Genetics, 2000, 24, 283-286.	9.4	323
7	Identical mutations in three different fibroblast growth factor receptor genes in autosomal dominant craniosynostosis syndromes. Nature Genetics, 1996, 14, 174-176.	9.4	306
8	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
9	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
10	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
11	A type X collagen mutation causes Schmid metaphyseal chondrodysplasia. Nature Genetics, 1993, 5, 79-82.	9.4	253
12	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
13	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
14	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
15	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
16	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
17	Mutation Analysis of LMX1B Gene in Nail-Patella Syndrome Patients. American Journal of Human Genetics, 1998, 63, 1651-1658.	2.6	166
18	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

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19	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
20	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
21	The Ehlersâ€“Danlos syndromes. Nature Reviews Disease Primers, 2020, 6, 64.	18.1	144
22	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
23	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
24	The Gene for the Ellisâ€“van Creveld Syndrome Is Located on Chromosome 4p16. Genomics, 1996, 35, 1-5.	1.3	117
25	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
26	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
27	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
28	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
29	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
30	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
31	Sleep-disordered breathing in children with achondroplasia. Journal of Pediatrics, 1998, 132, 667-671.	0.9	90
32	Multiple Molecular Mechanisms Underlying Subdiagnostic Variants of Marfan Syndrome. American Journal of Human Genetics, 1998, 63, 1703-1711.	2.6	85
33	Dextromethorphan and high-dose benzoate therapy for nonketotic hyperglycinemia in an infant. Journal of Pediatrics, 1992, 121, 131-135.	0.9	84
34	Therapeutic approaches to cobalamin-C methylmalonic acidemia and homocystinuria. Journal of Pediatrics, 1988, 112, 32-39.	0.9	83
35	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
36	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

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37	Molecular Genetic Analysis in Autosomal Dominant Keratoconus. Cornea, 1992, 11, 302-308.	0.9	73
38	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
39	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
40	Craniocervical decompression for cervicomedullary compression in pediatric patients with achondroplasia. Journal of Neurosurgery, 1990, 73, 375-382.	0.9	70
41	Clinical and biochemical profiles suggest fibromuscular dysplasia is a systemic disease with altered TGFâ€Î² expression and connective tissue features. FASEB Journal, 2014, 28, 3313-3324.	0.2	68
42	The status of online Mendelian inheritance in man (OMIM) medio 1994. Nucleic Acids Research, 1994, 22, 3470-3473.	6.5	66
43	Small deletions in the type II collagen triple helix produce Kniest dysplasia. , 1999, 85, 105-112.		59
44	Linkage of typical pseudoachondroplasia to chromosome 19. Genomics, 1993, 18, 661-666.	1.3	57
45	The Genetic Basis of Dwarfism. New England Journal of Medicine, 1995, 332, 58-59.	13.9	55
46	Additional mutations of type X collagen confirm COL10A1 as the Schmid metaphyseal chondrodysplasia locus. Human Molecular Genetics, 1994, 3, 303-307.	1.4	54
47	Thoracolumbar Spinal Abnormalities in Stickler Syndrome. Spine, 2001, 26, 403-409.	1.0	53
48	The question of heterogeneity in Marfan syndrome. Nature Genetics, 1995, 9, 228-229.	9.4	51
49	The gene for pycnodysostosis maps to human chromosome 1cenâ€“q21. Nature Genetics, 1995, 10, 238-239.	9.4	51
50	Gene Expression Profile of Human Bone Marrow Stromal Cells: High-Throughput Expressed Sequence Tag Sequencing Analysis. Genomics, 2002, 79, 7-17.	1.3	51
51	A Skeletal Gene Database. Journal of Bone and Mineral Research, 2000, 15, 2095-2122.	3.1	50
52	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
53	Consensus clinical management guidelines for AlstrÃ¶m syndrome. Orphanet Journal of Rare Diseases, 2020, 15, 253.	1.2	49
54	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

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55	Jeune asphyxiating thoracic dystrophy and short-rib polydactyly type III (Verma-Naumoff) are variants of the same disorder. , 2000, 90, 310-314.		48
56	Genetic and physical mapping of the McKusick-Kaufman syndrome. Human Molecular Genetics, 1998, 7, 475-481.	1.4	46
57	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
58	Localization of the Gene (SYM1) for Proximal Symphalangism to Human Chromosome 17q21-q22. Genomics, 1995, 27, 225-229.	1.3	42
59	Bone dysplasias in man: molecular insights. Current Opinion in Genetics and Development, 1996, 6, 301-308.	1.5	41
60	Characterization of a Human Gene Encoding Nucleosomal Binding Protein NSBP1. Genomics, 2001, 71, 163-173.	1.3	40
61	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
62	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
63	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
64	Presymptomatic direct detection of adenomatous polyposis coli (APC) gene mutations in familial adenomatous polyposis. Human Genetics, 1993, 91, 307-11.	1.8	38
65	High-Resolution Genetic Mapping of the Cartilage-Hair Hypoplasia (CHH) Gene in Amish and Finnish Families. Genomics, 1994, 20, 347-353.	1.3	37
66	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
67	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
68	Characteristics, Diagnosis, and Management of Ehlers-Danlos Syndromes. JAMA Facial Plastic Surgery, 2018, 20, 70-75.	2.2	34
69	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
70	Prevalence of hypermobile Ehlers-Danlos syndrome in postural orthostatic tachycardia syndrome. Autonomic Neuroscience: Basic and Clinical, 2020, 224, 102637.	1.4	32
71	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
72	A central nervous system specific mouse model for thanatophoric dysplasia type II. Human Molecular Genetics, 2003, 12, 2863-2871.	1.4	30

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73	Issues surrounding prenatal genetic testing for achondroplasia. Prenatal Diagnosis, 2002, 22, 933-940.	1.1	29
74	Growth and integration of neocartilage with native cartilage in vitro. Journal of Orthopaedic Research, 2005, 23, 433-439.	1.2	29
75	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
76	Key role for a minor collagen. Nature Genetics, 1995, 9, 6-8.	9.4	27
77	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
78	Medical genetic studies in the Amish: Historical perspective. American Journal of Medical Genetics Part A, 2003, 121C, 1-4.	2.4	27
79	Case 47: Dural Ectasia Associated with Marfan Syndrome. Radiology, 2002, 223, 767-771.	3.6	25
80	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
81	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
82	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
83	Defining renal phenotype in AlstrÃ¶m syndrome. Nephrology Dialysis Transplantation, 2020, 35, 994-1001.	0.4	20
84	DNA Analysis in Genetic Disorders. Annual Review of Medicine, 1986, 37, 377-395.	5.0	19
85	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
86	Severe hypodontia and oral xanthomas in Alagille syndrome. American Journal of Medical Genetics Part A, 2000, 93, 250-252.	2.4	18
87	<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
88	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
89	Brachydactyly Type C Gene Maps to Human Chromosome 12q24. Genomics, 1996, 38, 45-50.	1.3	16
90	Progress in medical genetics: Map-based gene discovery and the molecular pathology of skeletal dysplasias. , 1996, 63, 98-105.		16

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91	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
92	Characterization of the Human Talin (TLN) Gene: Genomic Structure, Chromosomal Localization, and Expression Pattern. Genomics, 1999, 62, 316-319.	1.3	15
93	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
94	Arterial Elasticity in Ehlers-Danlos Syndromes. Genes, 2020, 11, 55.	1.0	13
95	Survey of Ehlersâ€'Danlos Patientsâ€™ ophthalmic surgery experiences. Molecular Genetics & Genomic Medicine, 2020, 8, e1155.	0.6	13
96	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
97	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
98	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
99	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
100	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
101	Klotho gene variation and expression in 20 inbred mouse strains. Mammalian Genome, 2004, 15, 759-767.	1.0	11
102	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
103	Physical mapping of the nail patella syndrome interval at 9q34: ordering of STSs and ESTs. Human Genetics, 1998, 103, 525-526.	1.8	8
104	Minocycline-induced generalized postinflammatory elastolysis. American Journal of Medicine, 2000, 109, 340.	0.6	8
105	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
106	Confirmatory linkage of hypochondroplasia to chromosome arm 4p. American Journal of Medical Genetics Part A, 1995, 57, 505-506.	2.4	7
107	Comprehensive resource: Skeletal gene database. American Journal of Medical Genetics Part A, 2001, 106, 275-281.	2.4	7
108	Refractory Syncope and Presyncope Associated with Atlantoaxial Instability: Preliminary Evidence of Improvement Following Surgical Stabilization. World Neurosurgery, 2021, 149, e854-e865.	0.7	7

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109	Gastrointestinal medication burden among persons with the Ehlersâ€Danlos syndromes. Neurogastroenterology and Motility, 2021, 33, e14077.	1.6	6
110	Complementary Medicine and Genetic Medicine: Polar Disciplines or Dynamic Partners?. Journal of Alternative and Complementary Medicine, 2005, 11, 343-347.	2.1	4
111	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
112	Molecular Genetic Studies in Achondroplasia. , 1988, 48, 53-58.		3
113	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
114	Victor Almon McKusick : In the footsteps of Mendel and Osler. American Journal of Medical Genetics, Part A, 2021, 185, 3193-3201.	0.7	2
115	Victor A. McKusick and Medical Genetics Among the Amish. , 2012, , 119-130.		2
116	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
117	Small deletions in the type II collagen triple helix produce Kniest dysplasia. , 1999, 85, 105.		1
118	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
119	Hereditary alpha-tryptasemia modifies clinical phenotypes among individuals with congenital hypermobility disorders. Human Genetics and Genomics Advances, 2022, 3, 100094.	1.0	1
120	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
121	Mutations in the fibrillin gene and variability of the Marfan syndrome. Journal of Molecular and Cellular Cardiology, 1992, 24, S76.	0.9	0
122	Diagnostic approaches to renal genetic disorders using DNA analysis. Pediatric Nephrology, 1992, 6, 113-118.	0.9	0
123	Recommendations for national and local regulatory authorities concerning research in genetic markers of disease. American Heart Journal, 2000, 140, S3-S5.	1.2	0
124	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
125	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
126	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

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127	The long road to rare disease diagnosis: a heroâ€™s journey. Molecular Genetics and Metabolism, 2021, 132, S297.	0.5	0
128	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
129	Case 47. Radiology, 2002, 222, 513-514.	3.6	0
130	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