Peter H Byers

List of Publications by Year
in descending order

240 papers	20,457 citations	8172 76 h-index	11928 134 g-index
248 all docs	248 docs citations	248 times ranked	11690 citing authors

#	Article	IF	Citations
1	Aneurysm Syndromes Caused by Mutations in the TGF-Î² Receptor. New England Journal of Medicine, 2006, 355, 788-798.	13.9	1,490
2	Clinical and Genetic Features of Ehlersâ€“Danlos Syndrome Type IV, the Vascular Type. New England Journal of Medicine, 2000, 342, 673-680.	13.9	1,219
3	Consortium for osteogenesis imperfecta mutations in the helical domain of type I collagen: regions rich in lethal mutations align with collagen binding sites for integrins and proteoglycans. Human Mutation, 2007, 28, 209-221.	1.1	620
4	Osteogenesis imperfecta. Nature Reviews Disease Primers, 2017, 3, 17052.	18.1	481
5	CRTAP Is Required for Prolyl 3- Hydroxylation and Mutations Cause Recessive Osteogenesis Imperfecta. Cell, 2006, 127, 291-304.	13.5	465
6	Loss-of-function mutations in TGFB2 cause a syndromic presentation of thoracic aortic aneurysm. Nature Genetics, 2012, 44, 922-927.	9.4	391
7	The bicuspid aortic valve: an integrated phenotypic classification of leaflet morphology and aortic root shape. Heart, 2008, 94, 1634-1638.	1.2	364
8	Human Ehlers-Danlos Syndrome Type VII C and Bovine Dermatosparaxis Are Caused by Mutations in the Procollagen I N-Proteinase Gene. American Journal of Human Genetics, 1999, 65, 308-317.	2.6	348
9	Osteogenesis imperfecta: translation of mutation to phenotype.. Journal of Medical Genetics, 1991, 28, 433-442.	1.5	292
10	Homozygosity for a Missense Mutation in SERPINH1, which Encodes the Collagen Chaperone Protein HSP47, Results in Severe Recessive Osteogenesis Imperfecta. American Journal of Human Genetics, 2010, 86, 389-398.	2.6	291
11	Mutations in the Gene Encoding the RER Protein FKBP65 Cause Autosomal-Recessive Osteogenesis Imperfecta. American Journal of Human Genetics, 2010, 86, 551-559.	2.6	278
12	Gene Targeting in Stem Cells from Individuals with Osteogenesis Imperfecta. Science, 2004, 303, 1198-1201.	6.0	271
13	Defect in Conversion of Procollagen to Collagen in a Form of Ehlers-Danlos Syndrome. Science, 1973, 182, 298-300.	6.0	266
14	Osteogenesis Imperfecta. Annual Review of Medicine, 1992, 43, 269-282.	5.0	244
15	Diagnosis, natural history, and management in vascular Ehlersâ€“Danlos syndrome. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2017, 175, 40-47.	0.7	239
16	Marfan syndrome: defective synthesis, secretion, and extracellular matrix formation of fibrillin by cultured dermal fibroblasts.. Journal of Clinical Investigation, 1992, 89, 79-86.	3.9	235
17	Survival is affected by mutation type and molecular mechanism in vascular Ehlersâ€“Danlos syndrome (EDS type IV). Genetics in Medicine, 2014, 16, 881-888.	1.1	217
18	Brittle bones - fragile molecules: disorders of collagen gene structure and expression. Trends in Genetics, 1990, 6, 293-299.	2.9	215

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19	X-Linked Cutis Laxa. New England Journal of Medicine, 1980, 303, 61-65.	13.9	205
20	Subtle structural alterations in the chains of type I procollagen produce osteogenesis imperfecta type II. Nature, 1985, 316, 363-366.	13.7	204
21	CRTAPandLEPRE1mutations in recessive osteogenesis imperfecta. Human Mutation, 2008, 29, 1435-1442.	1.1	196
22	Altered triple helical structure of type I procollagen in lethal perinatal osteogenesis imperfecta.. Journal of Biological Chemistry, 1985, 260, 1734-1742.	1.6	196
23	Haploinsufficiency of SF3B4, a Component of the Pre-mRNA Spliceosomal Complex, Causes Nager Syndrome. American Journal of Human Genetics, 2012, 90, 925-933.	2.6	188
24	Perinatal lethal osteogenesis imperfecta (OI type II): a biochemically heterogeneous disorder usually due to new mutations in the genes for type I collagen. American Journal of Human Genetics, 1988, 42, 237-48.	2.6	184
25	Altered triple helical structure of type I procollagen in lethal perinatal osteogenesis imperfecta. Journal of Biological Chemistry, 1985, 260, 1734-42.	1.6	180
26	WNT1 Mutations in Families Affected by Moderately Severe and Progressive Recessive Osteogenesis Imperfecta. American Journal of Human Genetics, 2013, 92, 590-597.	2.6	179
27	Cysteine in the triple-helical domain of one allelic product of the alpha 1(I) gene of type I collagen produces a lethal form of osteogenesis imperfecta.. Journal of Biological Chemistry, 1984, 259, 11129-11138.	1.6	179
28	Cerebrovascular complications in Ehlers-Danlos syndrome type IV. Annals of Neurology, 1995, 38, 960-964.	2.8	176
29	Analysis of multigenerational families with thoracic aortic aneurysms and dissections due to TGFBR1 or TGFBR2 mutations. Journal of Medical Genetics, 2009, 46, 607-613.	1.5	172
30	Haploinsufficiency for One COL3A1 Allele of Type III Procollagen Results in a Phenotype Similar to the Vascular Form of Ehlers-Danlos Syndrome, Ehlers-Danlos Syndrome Type IV. American Journal of Human Genetics, 2001, 69, 989-1001.	2.6	168
31	Reduced secretion of structurally abnormal type I procollagen in a form of osteogenesis imperfecta.. Proceedings of the National Academy of Sciences of the United States of America, 1981, 78, 5142-5146.	3.3	167
32	Pre- and Postnatal Transplantation of Fetal Mesenchymal Stem Cells in Osteogenesis Imperfecta: A Two-Center Experience. Stem Cells Translational Medicine, 2014, 3, 255-264.	1.6	162
33	Cysteine in the triple-helical domain of one allelic product of the alpha 1(I) gene of type I collagen produces a lethal form of osteogenesis imperfecta. Journal of Biological Chemistry, 1984, 259, 11129-38.	1.6	148
34	Rare Autosomal Recessive Cardiac Valvular Form of Ehlers-Danlos Syndrome Results from Mutations in the COL1A2 Gene That Activate the Nonsense-Mediated RNA Decay Pathway. American Journal of Human Genetics, 2004, 74, 917-930.	2.6	147
35	Recurrence of lethal osteogenesis imperfecta due to parental mosaicism for a dominant mutation in a human type I collagen gene (COL1A1). American Journal of Human Genetics, 1990, 46, 591-601.	2.6	145
36	The Ehlersâ€“Danlos syndromes. Nature Reviews Disease Primers, 2020, 6, 64.	18.1	144

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37	Usefulness of Bicuspid Aortic Valve Phenotype to Predict Elastic Properties of the Ascending Aorta. American Journal of Cardiology, 2007, 99, 686-690.	0.7	138
38	Type I osteogenesis imperfecta: a nonfunctional allele for pro alpha 1 (I) chains of type I procollagen.. Proceedings of the National Academy of Sciences of the United States of America, 1982, 79, 3838-3842.	3.3	137
39	Mutations in FKBP10, which result in Bruck syndrome and recessive forms of osteogenesis imperfecta, inhibit the hydroxylation of telopeptide lysines in bone collagen. Human Molecular Genetics, 2013, 22, 1-17.	1.4	135
40	Clinical and ultrastructural heterogeneity of type IV Ehlers-Danlos syndrome. Human Genetics, 1979, 47, 141-150.	1.8	134
41	PREGNANCY COMPLICATIONS IN TYPE IV EHLERS-DANLOS SYNDROME. Lancet, The, 1983, 321, 50-53.	6.3	130
42	Endoplasmic Reticulum-mediated Quality Control of Type I Collagen Production by Cells from Osteogenesis Imperfecta Patients with Mutations in the proÎ±1(I) Chain Carboxyl-terminal Propeptide which Impair Subunit Assembly. Journal of Biological Chemistry, 1995, 270, 8642-8649.	1.6	129
43	Pregnancy-related deaths and complications in women with vascular Ehlersâ€“Danlos syndrome. Genetics in Medicine, 2014, 16, 874-880.	1.1	127
44	Structural Abnormalities in the Dermal Collagen and Elastic Matrix from the Skin of Patients with Inherited Connective Tissue Disorders.. Journal of Investigative Dermatology, 1982, 79, 7s-16s.	0.3	122
45	Null Alleles of the COL5A1 Gene of Type V Collagen Are a Cause of the Classical Forms of Ehlers-Danlos Syndrome (Types I and II). American Journal of Human Genetics, 2000, 66, 1757-1765.	2.6	122
46	BiP binds type I procollagen pro alpha chains with mutations in the carboxyl-terminal propeptide synthesized by cells from patients with osteogenesis imperfecta. Journal of Biological Chemistry, 1993, 268, 18226-33.	1.6	122
47	Intron-mediated recombination may cause a deletion in an alpha 1 type I collagen chain in a lethal form of osteogenesis imperfecta.. Proceedings of the National Academy of Sciences of the United States of America, 1985, 82, 2870-2874.	3.3	121
48	Delineation of the Marfan phenotype associated with mutations in exons 23â€“32 of theFBN1 gene. , 1996, 62, 233-242.		120
49	Lethal osteogenesis imperfecta resulting from a single nucleotide change in one human pro alpha 1(I) collagen allele.. Proceedings of the National Academy of Sciences of the United States of America, 1986, 83, 6045-6047.	3.3	118
50	Mutations in PPIB (cyclophilin B) delay type I procollagen chain association and result in perinatal lethal to moderate osteogenesis imperfecta phenotypes. Human Molecular Genetics, 2011, 20, 1595-1609.	1.4	118
51	Ehlers-Danlos syndrome type VIIA and VIIB result from splice-junction mutations or genomic deletions that involve exon 6 in theCOL1A1 andCOL1A2 genes of type I collagen. American Journal of Medical Genetics Part A, 1997, 72, 94-105.	2.4	117
52	A translocation interrupts the COL5A1 gene in a patient with Ehlersâ€“Danlos syndrome and hypomelanosis of Ito. Nature Genetics, 1996, 13, 361-365.	9.4	116
53	Killing the messenger: new insights into nonsense-mediated mRNA decay. Journal of Clinical Investigation, 2002, 109, 3-6.	3.9	114
54	Marfan syndrome: abnormal alpha 2 chain in type I collagen.. Proceedings of the National Academy of Sciences of the United States of America, 1981, 78, 7745-7749.	3.3	112

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55	Identification, characterization and expression analysis of a new fibrillar collagen gene, COL27A1. Matrix Biology, 2003, 22, 3-14.	1.5	112
56	Mutation and polymorphism spectrum in osteogenesis imperfecta type II: implications for genotypeâ€“phenotype relationships. Human Molecular Genetics, 2009, 18, 463-471.	1.4	107
57	EMQN best practice guidelines for the laboratory diagnosis of osteogenesis imperfecta. European Journal of Human Genetics, 2012, 20, 11-19.	1.4	107
58	Osteogenesis imperfecta type I: molecular heterogeneity for COL1A1 null alleles of type I collagen. American Journal of Human Genetics, 1994, 55, 638-47.	2.6	107
59	Order of Intron Removal Influences Multiple Splice Outcomes, Including a Two-Exon Skip, in a COL5A1 Acceptor-Site Mutation That Results in Abnormal Pro-Î±1(V) N-Propeptides and Ehlers-Danlos Syndrome Type I. American Journal of Human Genetics, 2002, 71, 451-465.	2.6	100
60	Periodontal Ehlers-Danlos Syndrome Is Caused by Mutations in C1R and C1S , which Encode Subcomponents C1r and C1s of Complement. American Journal of Human Genetics, 2016, 99, 1005-1014.	2.6	100
61	COL3A1haploinsufficiency results in a variety of Ehlers-Danlos syndrome type IV with delayed onset of complications and longer life expectancy. Genetics in Medicine, 2011, 13, 717-722.	1.1	98
62	Molecular defects of type III procollagen in Ehlers-Danlos syndrome type IV. Human Genetics, 1989, 82, 104-108.	1.8	97
63	Interchain disulfide bonds in procollagen are located in a large nontriple-helical COOH-terminal domain.. Proceedings of the National Academy of Sciences of the United States of America, 1975, 72, 3009-3013.	3.3	96
64	Stability related bias in residues replacing glycines within the collagen triple helix (Gly-Xaa-Yaa) in inherited connective tissue disorders. Human Mutation, 2004, 24, 330-337.	1.1	95
65	Recessively Inherited Forms of Osteogenesis Imperfecta. Annual Review of Genetics, 2012, 46, 475-497.	3.2	94
66	Spontaneous multivessel cervical artery dissection in a patient with a substitution of alanine for glycine (G13A) in the alpha 1(I) chain of type I collagen. Neurology, 1996, 47, 552-556.	1.5	93
67	Distinct biochemical phenotypes predict clinical severity in nonlethal variants of osteogenesis imperfecta. American Journal of Human Genetics, 1990, 46, 975-82.	2.6	92
68	Abnormal alpha 2-chain in type I collagen from a patient with a form of osteogenesis imperfecta.. Journal of Clinical Investigation, 1983, 71, 689-697.	3.9	88
69	Skin is a window on heritable disorders of connective tissue. American Journal of Medical Genetics Part A, 1989, 34, 105-121.	2.4	87
70	Frameshift mutation near the 3' end of the COL1A1 gene of type I collagen predicts an elongated Pro alpha 1(I) chain and results in osteogenesis imperfecta type I.. Journal of Clinical Investigation, 1990, 85, 282-290.	3.9	87
71	Variable expression of osteogenesis imperfecta in a nuclear family is explained by somatic mosaicism for a lethal point mutation in the alpha 1(I) gene (COL1A1) of type I collagen in a parent. American Journal of Human Genetics, 1990, 46, 1034-40.	2.6	86
72	Structural Abnormalities in the Dermal Collagen and Elastic Matrix from the Skin of Patients with Inherited Connective Tissue Disorders. Journal of Investigative Dermatology, 1982, 79, 7-16.	0.3	84

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73	Human dermatosparaxis: a form of Ehlers-Danlos syndrome that results from failure to remove the amino-terminal propeptide of type I procollagen. American Journal of Human Genetics, 1992, 51, 235-44.	2.6	82
74	Recurrence of lethal osteogenesis imperfecta due to parental mosaicism for a mutation in the COL1A2 gene of type I collagen. The mosiac parent exhibits phenotypic features of a mild form of the disease. Human Mutation, 1992, 1, 47-54.	1.1	80
75	Inherited disorders of collagen gene structure and expression. American Journal of Medical Genetics Part A, 1989, 34, 72-80.	2.4	79
76	Studies of collagen synthesis and structure in the differentiation of child abuse from osteogenesis imperfecta. Journal of Pediatrics, 1996, 128, 542-547.	0.9	78
77	Genetic evaluation of suspected osteogenesis imperfecta (OI). Genetics in Medicine, 2006, 8, 383-388.	1.1	78
78	Gene Targeting of Mutant COL1A2 Alleles in Mesenchymal Stem Cells From Individuals With Osteogenesis Imperfecta. Molecular Therapy, 2008, 16, 187-193.	3.7	78
79	Spontaneous Direct Carotid-Cavernous Fistula in Ehlers-Danlos Syndrome Type IV: Two Case Reports and a Review of the Literature. Journal of Neuro-Ophthalmology, 2002, 22, 75-81.	0.4	74
80	Heterozygosity for a large deletion in the alpha 2(I) collagen gene has a dramatic effect on type I collagen secretion and produces perinatal lethal osteogenesis imperfecta.. Journal of Biological Chemistry, 1988, 263, 8398-8404.	1.6	74
81	Osteogenesis imperfecta. The position of substitution for glycine by cysteine in the triple helical domain of the pro alpha 1(I) chains of type I collagen determines the clinical phenotype.. Journal of Clinical Investigation, 1989, 84, 1206-1214.	3.9	74
82	Splicing Defects in the COL3A1 Gene: Marked Preference for 5â€² (Donor) Splice-Site Mutations in Patients with Exon-Skipping Mutations and Ehlers-Danlos Syndrome Type IV. American Journal of Human Genetics, 1997, 61, 1276-1286.	2.6	73
83	STRATEGIES AND OUTCOMES OF PRENATAL DIAGNOSIS FOR OSTEOGENESIS IMPERFECTA: A REVIEW OF BIOCHEMICAL AND MOLECULAR STUDIES COMPLETED IN 129 PREGNANCIES. , 1997, 17, 559-570.		73
84	Killing the messenger: new insights into nonsense-mediated mRNA decay. Journal of Clinical Investigation, 2002, 109, 3-6.	3.9	73
85	Dermatosparaxis in a Himalayan Cat: II. Ultrastructural Studies of Dermal Collagen. Journal of Investigative Dermatology, 1980, 74, 100-104.	0.3	72
86	Ehlers-Danlos syndrome type IV: cosegregation of the phenotype to a COL3A1 allele of type III procollagen. Human Genetics, 1986, 74, 41-6.	1.8	72
87	Testing for osteogenesis imperfecta in cases of suspected non-accidental injury. Journal of Medical Genetics, 2002, 39, 382-386.	1.5	72
88	Mutations in the COL3A1 Gene Result in the Ehlersâ€“Danlos Syndrome Type IV and Alterations in the Size and Distribution of the Major Collagen Fibrils of the Dermis. Journal of Investigative Dermatology, 1997, 108, 241-247.	0.3	71
89	Molecular mechanisms of classical Ehlers-Danlos syndrome (EDS). Human Mutation, 2009, 30, 995-1002.	1.1	70
90	Arginine for glycine substitution in the triple-helical domain of the products of one alpha 2(I) collagen allele (COL1A2) produces the osteogenesis imperfecta type IV phenotype.. Journal of Biological Chemistry, 1988, 263, 7734-7740.	1.6	69

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91	Type XXVII collagen at the transition of cartilage to bone during skeletogenesis. Bone, 2007, 41, 535-542.	1.4	67
92	Osteogenesis imperfecta type I is commonly due to a COL1A1 null allele of type I collagen. American Journal of Human Genetics, 1992, 51, 508-15.	2.6	66
93	Constitutive skipping of alternatively spliced exon 10 in the ATP7A gene abolishes Golgi localization of the menkes protein and produces the occipital horn syndrome [published erratum appears in Hum Mol Genet 1998 Jun;7(6):1059]. Human Molecular Genetics, 1998, 7, 465-469.	1.4	65
94	Heterozygosity for a large deletion in the alpha 2(I) collagen gene has a dramatic effect on type I collagen secretion and produces perinatal lethal osteogenesis imperfecta. Journal of Biological Chemistry, 1988, 263, 8398-404.	1.6	65
95	Prenatal diagnosis of lethal perinatal osteogenesis imperfecta (OI Type II). Journal of Pediatrics, 1982, 100, 127-133.	0.9	64
96	Recurrence of perinatal lethal osteogenesis imperfecta in sibships: Parsing the risk between parental mosaicism for dominant mutations and autosomal recessive inheritance. Genetics in Medicine, 2011, 13, 125-130.	1.1	64
97	The challenge of comprehensive and consistent sequence variant interpretation between clinical laboratories. Genetics in Medicine, 2016, 18, 20-24.	1.1	63
98	Redefinition of Exon 7 in the COL1A1 Gene of Type I Collagen by an Intron 8 Splice-Donorâ€“Site Mutation in a Form of Osteogenesis Imperfecta: Influence of Intron Splice Order on Outcome of Splice-Site Mutation. American Journal of Human Genetics, 1999, 65, 336-344.	2.6	62
99	Peptide Mapping of Collagen Chains Using CNBr Cleavage of Proteins Within Polyacrylamide Gels. Collagen and Related Research, 1981, 1, 543-548.	2.2	60
100	Osteogenesis imperfecta type III: mutations in the type I collagen structural genes, COL1A1 and COL1A2, are not necessarily responsible.. Journal of Medical Genetics, 1993, 30, 492-496.	1.5	60
101	Invited editorial comment: Osteogenesis imperfecta: Update and perspective. American Journal of Medical Genetics Part A, 1984, 17, 429-435.	2.4	59
102	A crossâ€sectional multicenter study ofÂosteogenesis imperfecta in North AmericaÂâ€“Âresults from the linked clinical research centers. Clinical Genetics, 2015, 87, 133-140.	1.0	59
103	Defective folding and stable association with protein disulfide isomerase/prolyl hydroxylase of type I procollagen with a deletion in the pro alpha 2(I) chain that preserves the Gly-X-Y repeat pattern. Journal of Biological Chemistry, 1992, 267, 7751-7.	1.6	59
104	Mutations in the carboxyl-terminal propeptide of the pro alpha 1(I) chain of type I collagen result in defective chain association and produce lethal osteogenesis imperfecta. Journal of Biological Chemistry, 1993, 268, 18218-25.	1.6	59
105	Ehlers-Danlos Syndrome: Recent Advances and Current Understanding of the Clinical and Genetic Heterogeneity.. Journal of Investigative Dermatology, 1994, 103, 47S-52S.	0.3	57
106	Defective C-propeptides of the ProÎ±2(I) Chain of Type I Procollagen Impede Molecular Assembly and Result in Osteogenesis Imperfecta. Journal of Biological Chemistry, 2008, 283, 16061-16067.	1.6	57
107	Ehlersâ€“Danlos syndrome: A showcase of conditions that lead to understanding matrix biology. Matrix Biology, 2014, 33, 10-15.	1.5	57
108	Thrombospondin II: Partial cDNA sequence, chromosome location, and expression of a second member of the thrombospondin gene family in humans. Genomics, 1992, 12, 421-429.	1.3	53

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109	Arginine for glycine substitution in the triple-helical domain of the products of one alpha 2(I) collagen allele (COL1A2) produces the osteogenesis imperfecta type IV phenotype. Journal of Biological Chemistry, 1988, 263, 7734-40.	1.6	53
110	Novel missense mutations in the TRPS1 transcription factor define the nuclear localization signal. European Journal of Human Genetics, 2004, 12, 121-126.	1.4	52
111	Generalized Connective Tissue Disease in Crtap-/- Mouse. PLoS ONE, 2010, 5, e10560.	1.1	52
112	Disruption of one intra-chain disulphide bond in the carboxyl-terminal propeptide of the proalpha1(I) chain of type I procollagen permits slow assembly and secretion of overmodified, but stable procollagen trimers and results in mild osteogenesis imperfecta. Journal of Medical Genetics, 2001, 38, 443-449.	1.5	51
113	The effects of different cysteine for glycine substitutions within alpha 2(I) chains. Evidence of distinct structural domains within the type I collagen triple helix. Journal of Biological Chemistry, 1991, 266, 2590-2594.	1.6	51
114	Osteogenesis imperfecta: perspectives and opportunities. Current Opinion in Pediatrics, 2000, 12, 603-609.	1.0	50
115	Folding defects in fibrillar collagens. Philosophical Transactions of the Royal Society B: Biological Sciences, 2001, 356, 151-158.	1.8	49
116	Ehlers Danlos syndrome type VIIB. Incomplete cleavage of abnormal type I procollagen by N-proteinase in vitro results in the formation of copolymers of collagen and partially cleaved pNcollagen that are near circular in cross-section. Journal of Biological Chemistry, 1992, 267, 9093-100.	1.6	49
117	Abnormal collagen fibril structure in the gravis form (type I) of Ehlers-Danlos syndrome. Laboratory Investigation, 1979, 40, 201-6.	1.7	48
118	Natural variation in four human collagen genes across an ethnically diverse population. Genomics, 2008, 91, 307-314.	1.3	47
119	Monoallelic and biallelic CREB3L1 variant causes mild and severe osteogenesis imperfecta, respectively. Genetics in Medicine, 2018, 20, 411-419.	1.1	47
120	Genetic Disorders of Collagen Metabolism. , 1982, 12, 1-87.		46
121	An RT-PCR-SSCP screening strategy for detection of mutations in the gene encoding the Î±1 chain of type I collagen: application to four patients with osteogenesis imperfecta. Human Molecular Genetics, 1993, 2, 1155-1160.	1.4	45
122	Heritable Collagen Disorders: The Paradigm of the Ehlersâ€"Danlos Syndrome. Journal of Investigative Dermatology, 2012, 132, E6-E11.	0.3	45
123	Molecular heterogeneity in osteogenesis imperfecta type I. American Journal of Medical Genetics Part A, 1993, 45, 223-227.	2.4	44
124	Osteogenesis imperfecta: mode of delivery and neonatal outcome. Obstetrics and Gynecology, 2001, 97, 66-69.	1.2	44
125	A homozygous B3GAT3 mutation causes a severe syndrome with multiple fractures, expanding the phenotype of linkeropathy syndromes. American Journal of Medical Genetics, Part A, 2015, 167, 2691-2696.	0.7	44
126	Collagens: building blocks at the end of the development line. Clinical Genetics, 2000, 58, 270-279.	1.0	43

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127	A single amino acid substitution (D1441Y) in the carboxyl-terminal propeptide of the proalpha1(I) chain of type I collagen results in a lethal variant of osteogenesis imperfecta with features of dense bone diseases. Journal of Medical Genetics, 2002, 39, 23-29.	1.5	42
128	A novel mutation causes a perinatal lethal form of osteogenesis imperfecta. An insertion in one alpha 1(I) collagen allele (COL1A1).. Journal of Biological Chemistry, 1988, 263, 7855-7861.	1.6	42
129	First-trimester prenatal diagnosis of osteogenesis imperfecta type II by DNA analysis and sonography. Prenatal Diagnosis, 1993, 13, 589-596.	1.1	40
130	Cesarean delivery is not associated with decreased at-birth fracture rates in osteogenesis imperfecta. Genetics in Medicine, 2016, 18, 570-576.	1.1	39
131	A multi-institutional experience in the aortic and arterial pathology in individuals with genetically confirmed vascular Ehlers-Danlos syndrome. Journal of Vascular Surgery, 2019, 70, 1543-1554.	0.6	39
132	Parental somatic and germ-line mosaicism for a multiexon deletion with unusual endpoints in a type III collagen (COL3A1) allele produces Ehlers-Danlos syndrome type IV in the heterozygous offspring. American Journal of Human Genetics, 1993, 53, 62-70.	2.6	39
133	The effects of different cysteine for glycine substitutions within alpha 2(I) chains. Evidence of distinct structural domains within the type I collagen triple helix. Journal of Biological Chemistry, 1991, 266, 2590-4.	1.6	39
134	Deletions and duplications of Gly-Xaa-Yaa triplet repeats in the triple helical domains of type I collagen chains disrupt helix formation and result in several types of osteogenesis imperfecta. Human Mutation, 2001, 18, 319-326.	1.1	38
135	Osteogenesis imperfecta due to recurrent point mutations at CpG dinucleotides in the COL1A1 gene of type I collagen. Human Genetics, 1991, 87, 33-40.	1.8	37
136	MOLECULAR BASIS OF HEREDITARY DISORDERS OF CONNECTIVE TISSUE. Annual Review of Medicine, 1994, 45, 149-163.	5.0	37
137	Partial COL1A2 gene duplication produces features of osteogenesis imperfecta and Ehlers-Danlos syndrome type VII. Human Genetics, 2000, 106, 19-28.	1.8	37
138	Refining the structure and content of clinical genomic reports. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2014, 166, 85-92.	0.7	37
139	Substitution of cysteine for glycine within the carboxyl-terminal telopeptide of the alpha 1 chain of type I collagen produces mild osteogenesis imperfecta.. Journal of Biological Chemistry, 1988, 263, 14605-14607.	1.6	36
140	Determination of the molecular basis of Marfan syndrome: a growth industry. Journal of Clinical Investigation, 2004, 114, 161-163.	3.9	36
141	Bi-allelic variants inCOL3A1encoding the ligand to GPR56 are associated with cobblestone-like cortical malformation, white matter changes and cerebellar cysts. Journal of Medical Genetics, 2017, 54, 432-440.	1.5	34
142	Growth characteristics in individuals with osteogenesis imperfecta in North America: results from a multicenter study. Genetics in Medicine, 2019, 21, 275-283.	1.1	34
143	Molecular Basis of Clinical Heterogeneity in the Ehlers-Danlos Syndrome. Annals of the New York Academy of Sciences, 1985, 460, 298-310.	1.8	33
144	Osteogenesis Imperfecta: The Molecular Basis of Clinical Heterogeneity. Annals of the New York Academy of Sciences, 1988, 543, 117-128.	1.8	33

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145	Vascular Ehlersâ€“Danlos Syndrome in siblings with biallelic COL3A1 sequence variants and marked clinical variability in the extended family. European Journal of Human Genetics, 2015, 23, 796-802.	1.4	33
146	A multicenter study to evaluate pulmonary function in osteogenesis imperfecta. Clinical Genetics, 2018, 94, 502-511.	1.0	33
147	A call for direct sequencing of full-length RNAs to identify all modifications. Nature Genetics, 2021, 53, 1113-1116.	9.4	33
148	Sequence and Characterization of the Complete Human Thrombospondin 2 cDNA: Potential Regulatory Role for the 3â€² Untranslated Region. Genomics, 1993, 17, 225-229.	1.3	32
149	Osteogenesis imperfecta type IV. Biochemical confirmation of genetic linkage to the pro alpha 2(I) gene of type I collagen.. Journal of Clinical Investigation, 1986, 78, 1449-1455.	3.9	32
150	Molecular Mechanisms of Connective Tissue Abnormalities in the Ehlers-Danlos Syndrome. Collagen and Related Research, 1981, 1, 475-489.	2.2	31
151	What every clinical geneticist should know about testing for osteogenesis imperfecta in suspected child abuse cases. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2015, 169, 307-313.	0.7	31
152	A novel mutation causes a perinatal lethal form of osteogenesis imperfecta. An insertion in one alpha 1(I) collagen allele (COL1A1). Journal of Biological Chemistry, 1988, 263, 7855-61.	1.6	31
153	Large kindred with Ehlers-Danlos syndrome type IV due to a point mutation (G571S) in theCOL3A1 gene of type III procollagen: Low risk of pregnancy complications and unexpected longevity in some affected relatives. , 1999, 82, 305-311.		29
154	A new variety of spondyloepiphyseal dysplasia characterized by punctate corneal dystrophy and abnormal dermal collagen fibrils. Human Genetics, 1978, 40, 157-169.	1.8	28
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