Peter H Byers

List of Publications by Citations

238 papers	16,888 citations	72 h-index	123 g-index
248 ext. papers	18,687 ext. citations	8.9 avg, IF	6.15 L-index

#	Paper	IF	Citations
238	Aneurysm syndromes caused by mutations in the TGF-beta receptor. *New England Journal of Medicine*, **2006**, 355, 788-98	59.2	1243
237	Clinical and genetic features of Ehlers-Danlos syndrome type IV, the vascular type. *New England Journal of Medicine*, **2000**, 342, 673-80	59.2	1011
236	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, 209-21	4.7	504
235	CRTAP is required for prolyl 3- hydroxylation and mutations cause recessive osteogenesis imperfecta. *Cell*, **2006**, 127, 291-304	56.2	394
234	Loss-of-function mutations in TGFB2 cause a syndromic presentation of thoracic aortic aneurysm. *Nature Genetics*, **2012**, 44, 922-7	36.3	323
233	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, 308-17	11	302
232	Osteogenesis imperfecta. *Nature Reviews Disease Primers*, **2017**, 3, 17052	51.1	292
231	The bicuspid aortic valve: an integrated phenotypic classification of leaflet morphology and aortic root shape. *Heart*, **2008**, 94, 1634-8	5.1	287
230	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, 389-98	11	253
229	Osteogenesis imperfecta: translation of mutation to phenotype. *Journal of Medical Genetics*, **1991**, 28, 433-42	5.8	249
228	Gene targeting in stem cells from individuals with osteogenesis imperfecta. *Science*, **2004**, 303, 1198-201	33.3	244
227	Defect in conversion of procollagen to collagen in a form of Ehlers-Danlos syndrome. *Science*, **1973**, 182, 298-300	33.3	242
226	Mutations in the gene encoding the RER protein FKBP65 cause autosomal-recessive osteogenesis imperfecta. *American Journal of Human Genetics*, **2010**, 86, 551-9	11	238
225	Osteogenesis imperfecta. *Annual Review of Medicine*, **1992**, 43, 269-82	17.4	223
224	Brittle bones--fragile molecules: disorders of collagen gene structure and expression. *Trends in Genetics*, **1990**, 6, 293-300	8.5	196
223	Marfan syndrome: defective synthesis, secretion, and extracellular matrix formation of fibrillin by cultured dermal fibroblasts. *Journal of Clinical Investigation*, **1992**, 89, 79-86	15.9	194
222	Subtle structural alterations in the chains of type I procollagen produce osteogenesis imperfecta type II. *Nature*, **1985**, 316, 363-6	50.4	187

221	X-linked cutis laxa: defective cross-link formation in collagen due to decreased lysyl oxidase activity. *New England Journal of Medicine*, **1980**, 303, 61-5	59.2	181
220	Altered triple helical structure of type I procollagen in lethal perinatal osteogenesis imperfecta. *Journal of Biological Chemistry*, **1985**, 260, 1734-42	5.4	177
219	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	11	173
218	CRTAP and LEPRE1 mutations in recessive osteogenesis imperfecta. *Human Mutation*, **2008**, 29, 1435-42	4.7	172
217	WNT1 mutations in families affected by moderately severe and progressive recessive osteogenesis imperfecta. *American Journal of Human Genetics*, **2013**, 92, 590-7	11	156
216	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, 5142-6	11.5	154
215	Cerebrovascular complications in Ehlers-Danlos syndrome type IV. *Annals of Neurology*, **1995**, 38, 960-4	9.4	153
214	Survival is affected by mutation type and molecular mechanism in vascular Ehlers-Danlos syndrome (EDS type IV). *Genetics in Medicine*, **2014**, 16, 881-8	8.1	151
213	Altered triple helical structure of type I procollagen in lethal perinatal osteogenesis imperfecta.. *Journal of Biological Chemistry*, **1985**, 260, 1734-1742	5.4	151
212	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	5.4	144
211	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	11	142
210	Diagnosis, natural history, and management in vascular Ehlers-Danlos syndrome. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2017**, 175, 40-47	3.1	140
209	Analysis of multigenerational families with thoracic aortic aneurysms and dissections due to TGFBR1 or TGFBR2 mutations. *Journal of Medical Genetics*, **2009**, 46, 607-13	5.8	140
208	Haploinsufficiency of SF3B4, a component of the pre-mRNA spliceosomal complex, causes Nager syndrome. *American Journal of Human Genetics*, **2012**, 90, 925-33	11	135
207	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	11	133
206	Pre- and postnatal transplantation of fetal mesenchymal stem cells in osteogenesis imperfecta: a two-center experience. *Stem Cells Translational Medicine*, **2014**, 3, 255-64	6.9	131
205	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, 917-30	11	127
204	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	5.4	125

203	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, 3838-42	11.5	120
202	Clinical and ultrastructural heterogeneity of type IV Ehlers-Danlos syndrome. *Human Genetics*, **1979**, 47, 141-50	6.3	119
201	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	5.6	117
200	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	5.4	117
199	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 Suppl 1, 7s-16s	4.3	116
198	Usefulness of bicuspid aortic valve phenotype to predict elastic properties of the ascending aorta. *American Journal of Cardiology*, **2007**, 99, 686-90	3	114
197	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, 2870-4	11.5	111
196	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, 1757-65	11	110
195	A translocation interrupts the COL5A1 gene in a patient with Ehlers-Danlos syndrome and hypomelanosis of Ito. *Nature Genetics*, **1996**, 13, 361-5	36.3	108
194	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, 6045-7	11.5	108
193	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, 7745-9	11.5	105
192	Endoplasmic reticulum-mediated quality control of type I collagen production by cells from osteogenesis imperfecta patients with mutations in the pro alpha 1 (I) chain carboxyl-terminal propeptide which impair subunit assembly. *Journal of Biological Chemistry*, **1995**, 270, 8642-9	5.4	104
191	Killing the messenger: new insights into nonsense-mediated mRNA decay. *Journal of Clinical Investigation*, **2002**, 109, 3-6	15.9	103
190	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, 1595-609	5.6	102
189	Delineation of the Marfan phenotype associated with mutations in exons 23-32 of the FBN1 gene. *American Journal of Medical Genetics Part A*, **1996**, 62, 233-42		101
188	EMQN best practice guidelines for the laboratory diagnosis of osteogenesis imperfecta. *European Journal of Human Genetics*, **2012**, 20, 11-9	5.3	99
187	Pregnancy complications in type IV Ehlers-Danlos Syndrome. *Lancet, The*, **1983**, 1, 50-3	40	99
186	Ehlers-Danlos syndrome type VIIA and VIIB result from splice-junction mutations or genomic deletions that involve exon 6 in the COL1A1 and COL1A2 genes of type I collagen. *American Journal of Medical Genetics Part A*, **1997**, 72, 94-105		96

185	Identification, characterization and expression analysis of a new fibrillar collagen gene, COL27A1. *Matrix Biology*, **2003**, 22, 3-14	11.4	96
184	Osteogenesis imperfecta type I: molecular heterogeneity for COL1A1 null alleles of type I collagen. *American Journal of Human Genetics*, **1994**, 55, 638-47	11	96
183	Mutation and polymorphism spectrum in osteogenesis imperfecta type II: implications for genotype-phenotype relationships. *Human Molecular Genetics*, **2009**, 18, 463-71	5.6	95
182	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, 3009-13	11.5	87
181	Pregnancy-related deaths and complications in women with vascular Ehlers-Danlos syndrome. *Genetics in Medicine*, **2014**, 16, 874-80	8.1	86
180	Molecular defects of type III procollagen in Ehlers-Danlos syndrome type IV. *Human Genetics*, **1989**, 82, 104-8	6.3	85
179	Distinct biochemical phenotypes predict clinical severity in nonlethal variants of osteogenesis imperfecta. *American Journal of Human Genetics*, **1990**, 46, 975-82	11	85
178	Order of intron removal influences multiple splice outcomes, including a two-exon skip, in a COL5A1 acceptor-site mutation that results in abnormal pro-alpha1(V) N-propeptides and Ehlers-Danlos syndrome type I. *American Journal of Human Genetics*, **2002**, 71, 451-65	11	83
177	Stability related bias in residues replacing glycines within the collagen triple helix (Gly-Xaa-Yaa) in inherited connective tissue disorders. *Human Mutation*, **2004**, 24, 330-7	4.7	82
176	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, 552-6	6.5	82
175	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	11	82
174	Frameshift mutation near the 3Pend 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, 282-90	15.9	80
173	COL3A1 haploinsufficiency results in a variety of Ehlers-Danlos syndrome type IV with delayed onset of complications and longer life expectancy. *Genetics in Medicine*, **2011**, 13, 717-22	8.1	79
172	Recessively inherited forms of osteogenesis imperfecta. *Annual Review of Genetics*, **2012**, 46, 475-97	14.5	78
171	Abnormal alpha 2-chain in type I collagen from a patient with a form of osteogenesis imperfecta. *Journal of Clinical Investigation*, **1983**, 71, 689-97	15.9	78
170	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	11	77
169	Inherited disorders of collagen gene structure and expression. *American Journal of Medical Genetics Part A*, **1989**, 34, 72-80		75
168	Skin is a window on heritable disorders of connective tissue. *American Journal of Medical Genetics Part A*, **1989**, 34, 105-21		74

167	Gene targeting of mutant COL1A2 alleles in mesenchymal stem cells from individuals with osteogenesis imperfecta. *Molecular Therapy*, **2008**, 16, 187-93	11.7	73
166	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	11	70
165	Recurrence of lethal osteogenesis imperfecta due to parental mosaicism for a mutation in the COL1A2 gene of type I collagen. The mosaic parent exhibits phenotypic features of a mild form of the disease. *Human Mutation*, **1992**, 1, 47-54	4.7	68
164	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, 1206-14	15.9	68
163	Molecular mechanisms of classical Ehlers-Danlos syndrome (EDS). *Human Mutation*, **2009**, 30, 995-1002	4.7	65
162	Testing for osteogenesis imperfecta in cases of suspected non-accidental injury. *Journal of Medical Genetics*, **2002**, 39, 382-6	5.8	64
161	Ehlers-Danlos syndrome type IV: cosegregation of the phenotype to a COL3A1 allele of type III procollagen. *Human Genetics*, **1986**, 74, 41-6	6.3	64
160	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	2.6	63
159	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	5.4	63
158	Splicing defects in the COL3A1 gene: marked preference for 5P(donor) spice-site mutations in patients with exon-skipping mutations and Ehlers-Danlos syndrome type IV. *American Journal of Human Genetics*, **1997**, 61, 1276-86	11	61
157	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	11	61
156	Studies of collagen synthesis and structure in the differentiation of child abuse from osteogenesis imperfecta. *Journal of Pediatrics*, **1996**, 128, 542-7	3.6	59
155	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	5.4	58
154	Peptide mapping of collagen chains using CNBr cleavage of proteins within polyacrylamide gels. *Collagen and Related Research*, **1981**, 1, 543-8		57
153	Genetic evaluation of suspected osteogenesis imperfecta (OI). *Genetics in Medicine*, **2006**, 8, 383-8	8.1	56
152	Type XXVII collagen at the transition of cartilage to bone during skeletogenesis. *Bone*, **2007**, 41, 535-42	4.7	56
151	Dermatosparaxis in a Himalayan cat: II. Ultrastructural studies of dermal collagen. *Journal of Investigative Dermatology*, **1980**, 74, 100-4	4.3	56
150	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	5.4	56

149	Redefinition of exon 7 in the COL1A1 gene of type I collagen by an intron 8 splice-donor-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, 336-44	11	54
148	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	5.4	54
147	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, 125-30	8.1	53
146	Mutations in the COL3A1 gene result in the Ehlers-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, 241-7	4.3	53
145	STRATEGIES AND OUTCOMES OF PRENATAL DIAGNOSIS FOR OSTEOGENESIS IMPERFECTA: A REVIEW OF BIOCHEMICAL AND MOLECULAR STUDIES COMPLETED IN 129 PREGNANCIES **1997**, 17, 559-570		52
144	Constitutive skipping of alternatively spliced exon 10 in the ATP7A gene abolishes Golgi localization of the menkes protein and produces the occipital horn syndrome. *Human Molecular Genetics*, **1998**, 7, 465-9	5.6	52
143	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	5.4	52
142	Osteogenesis imperfecta: update and perspective. *American Journal of Medical Genetics Part A*, **1984**, 17, 429-35		51
141	Prenatal diagnosis of lethal perinatal osteogenesis imperfecta (OI type II). *Journal of Pediatrics*, **1982**, 100, 127-33	3.6	50
140	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	5.4	50
139	Ehlers-Danlos syndrome: recent advances and current understanding of the clinical and genetic heterogeneity. *Journal of Investigative Dermatology*, **1994**, 103, 47S-52S	4.3	49
138	Thrombospondin II: partial cDNA sequence, chromosome location, and expression of a second member of the thrombospondin gene family in humans. *Genomics*, **1992**, 12, 421-9	4.3	49
137	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	5.4	49
136	The Ehlers-Danlos syndromes. *Nature Reviews Disease Primers*, **2020**, 6, 64	51.1	47
135	The challenge of comprehensive and consistent sequence variant interpretation between clinical laboratories. *Genetics in Medicine*, **2016**, 18, 20-4	8.1	46
134	Killing the messenger: new insights into nonsense-mediated mRNA decay. *Journal of Clinical Investigation*, **2002**, 109, 3-6	15.9	46
133	A cross-sectional multicenter study of osteogenesis imperfecta in North America - results from the linked clinical research centers. *Clinical Genetics*, **2015**, 87, 133-40	4	45
132	Ehlers-Danlos syndrome: a showcase of conditions that lead to understanding matrix biology. *Matrix Biology*, **2014**, 33, 10-5	11.4	45

131	Defective C-propeptides of the proalpha2(I) chain of type I procollagen impede molecular assembly and result in osteogenesis imperfecta. *Journal of Biological Chemistry*, **2008**, 283, 16061-7	5.4	45
130	Generalized connective tissue disease in Crtap-/- mouse. *PLoS ONE*, **2010**, 5, e10560	3.7	45
129	Novel missense mutations in the TRPS1 transcription factor define the nuclear localization signal. *European Journal of Human Genetics*, **2004**, 12, 121-6	5.3	44
128	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, 443-9	5.8	44
127	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, 492-6	5.8	44
126	Abnormal collagen fibril structure in the gravis form (type I) of Ehlers-Danlos syndrome. *Laboratory Investigation*, **1979**, 40, 201-6	5.9	44
125	Folding defects in fibrillar collagens. *Philosophical Transactions of the Royal Society B: Biological Sciences*, **2001**, 356, 151-7; discussion 157-8	5.8	42
124	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	5.4	41
123	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	4.3	40
122	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**, 167A, 2691-6	2.5	39
121	Genetic disorders of collagen metabolism. *Advances in Human Genetics*, **1982**, 12, 1-87		39
120	Heritable collagen disorders: the paradigm of the Ehlers-Danlos syndrome. *Journal of Investigative Dermatology*, **2012**, 132, E6-11	4.3	38
119	Osteogenesis imperfecta: perspectives and opportunities. *Current Opinion in Pediatrics*, **2000**, 12, 603-9	3.2	38
118	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	5.4	37
117	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	11	37
116	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, 23-9	5.8	36
115	An RT-PCR-SSCP screening strategy for detection of mutations in the gene encoding the alpha 1 chain of type I collagen: application to four patients with osteogenesis imperfecta. *Human Molecular Genetics*, **1993**, 2, 1155-60	5.6	36
114	Natural variation in four human collagen genes across an ethnically diverse population. *Genomics*, **2008**, 91, 307-14	4.3	35

113	Collagens: building blocks at the end of the development line. *Clinical Genetics*, **2000**, 58, 270-9	4	35
112	Partial COL1A2 gene duplication produces features of osteogenesis imperfecta and Ehlers-Danlos syndrome type VII. *Human Genetics*, **2000**, 106, 19-28	6.3	33
111	Molecular basis of hereditary disorders of connective tissue. *Annual Review of Medicine*, **1994**, 45, 149-63	17.4	33
110	Osteogenesis imperfecta: mode of delivery and neonatal outcome. *Obstetrics and Gynecology*, **2001**, 97, 66-9	4.9	32
109	Osteogenesis imperfecta: the molecular basis of clinical heterogeneity. *Annals of the New York Academy of Sciences*, **1988**, 543, 117-28	6.5	32
108	Determination of the molecular basis of Marfan syndrome: a growth industry. *Journal of Clinical Investigation*, **2004**, 114, 161-163	15.9	32
107	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	5.4	32
106	Osteogenesis imperfecta due to recurrent point mutations at CpG dinucleotides in the COL1A1 gene of type I collagen. *Human Genetics*, **1991**, 87, 33-40	6.3	31
105	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, 1449-55	15.9	31
104	Refining the structure and content of clinical genomic reports. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2014**, 166C, 85-92	3.1	30
103	First-trimester prenatal diagnosis of osteogenesis imperfecta type II by DNA analysis and sonography. *Prenatal Diagnosis*, **1993**, 13, 589-96	3.2	30
102	Molecular basis of clinical heterogeneity in the Ehlers-Danlos syndrome. *Annals of the New York Academy of Sciences*, **1985**, 460, 298-310	6.5	30
101	Molecular mechanisms of connective tissue abnormalities in the Ehlers-Danlos syndrome. *Collagen and Related Research*, **1981**, 1, 475-89		30
100	Sequence and characterization of the complete human thrombospondin 2 cDNA: potential regulatory role for the 3Puntranslated region. *Genomics*, **1993**, 17, 225-9	4.3	29
99	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	5.4	29
98	Monoallelic and biallelic CREB3L1 variant causes mild and severe osteogenesis imperfecta, respectively. *Genetics in Medicine*, **2018**, 20, 411-419	8.1	28
97	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, 319-26	4.7	28
96	A new variety of spondyloepiphyseal dysplasia characterized by punctate corneal dystrophy and abnormal dermal collagen fibrils. *Human Genetics*, **1978**, 40, 157-69	6.3	28

95	Cesarean delivery is not associated with decreased at-birth fracture rates in osteogenesis imperfecta. *Genetics in Medicine*, **2016**, 18, 570-6	8.1	27
94	Molecular heterogeneity in osteogenesis imperfecta type I. *American Journal of Medical Genetics Part A*, **1993**, 45, 223-7		27
93	Bi-allelic variants in encoding 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	5.8	26
92	Neurological presentation of Ehlers-Danlos syndrome type IV in a family with parental mosaicism. *Clinical Genetics*, **2003**, 63, 510-5	4	26
91	Vascular Ehlers-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	5.3	25
90	Sequence of the coding region of the bovine fibrillin cDNA and localization to bovine chromosome 10. *Genomics*, **1994**, 23, 480-5	4.3	25
89	Altered secretion of type III procollagen in a form of type IV Ehlers-Danlos syndrome. Biochemical studies in cultured fibroblasts. *Laboratory Investigation*, **1981**, 44, 336-41	5.9	25
88	Ehlers-Danlos syndrome type IV: a subset of patients distinguished by low serum levels of the amino-terminal propeptide of type III procollagen. *American Journal of Medical Genetics Part A*, **1989**, 34, 68-71		24
87	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	5.4	24
86	Multiexon deletion in the procollagen III gene is associated with mild Ehlers-Danlos syndrome type IV. *Journal of Biological Chemistry*, **1991**, 266, 5244-8	5.4	23
85	Successful endovascular repair of acute type B aortic dissection in undiagnosed Ehlers-Danlos syndrome type IV. *European Journal of Vascular and Endovascular Surgery*, **2009**, 38, 608-9	2.3	22
84	Multiple vascular and bowel ruptures in an adolescent male with sporadic Ehlers-Danlos syndrome type IV. *Pediatric and Developmental Pathology*, **1999**, 2, 86-93	2.2	22
83	Large kindred with Ehlers-Danlos syndrome type IV due to a point mutation (G571S) in the COL3A1 gene of type III procollagen: Low risk of pregnancy complications and unexpected longevity in some affected relatives **1999**, 82, 305-311		22
82	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, 307-13	3.1	21
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4	ABNORMALITIES IN THE PRODUCTION OF EXTRACELLULAR MATRIX BY CELLS FROM INDIVIDUALS WITH INHERITED DISORDERS OF COLLAGEN BIOSYNTHESIS**Supported in part by grants from the USPHS (AM 21557, AM 30426, GM 07266), Clinical Research Grants from the March of Dimes Birth Defects Foundation (6-298 and 6-312), a grant from the Osteogenesis Imperfecta Foundation, and	
3	Marfan syndrome resulting from a rare pathogenic FBN1 variant, ascertained through a proband with IgG4-related arteriopathy. *American Journal of Medical Genetics, Part A*, **2021**, 185, 2180-2189	2.5
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