Peter H Byers

List of Publications by Year in Descending Order

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	Aortic dissection in pregnancy and the postpartum period.. *Seminars in Vascular Surgery*, **2022**, 35, 60-68	1.2	2
237	The Aortic Dissection Collaborative: Methods for building capacity for patient-centered outcomes research in the aortic dissection community.. *Seminars in Vascular Surgery*, **2022**, 35, 9-15	1.2	5
236	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	
235	Subtle differences in autonomic symptoms in people diagnosed with hypermobile Ehlers-Danlos syndrome and hypermobility spectrum disorders. *American Journal of Medical Genetics, Part A*, **2021**, 185, 2012-2025	2.5	1
234	2020 McKusick Award address. *American Journal of Human Genetics*, **2021**, 108, 761-763	11	
233	Caffey disease is associated with distinct arginine to cysteine substitutions in the pro(I) chain of type I procollagen. *Genetics in Medicine*, **2021**, 23, 2378-2385	8.1	0
232	A call for direct sequencing of full-length RNAs to identify all modifications. *Nature Genetics*, **2021**, 53, 1113-1116	36.3	5
231	Genetic counseling and screening of consanguineous couples and their offspring practice resource: Focused Revision. *Journal of Genetic Counseling*, **2021**, 30, 1354-1357	2.5	1
230	Abnormal Bone Collagen Cross-Linking in Osteogenesis Imperfecta/Bruck Syndrome Caused by Compound Heterozygous Mutations. *JBMR Plus*, **2021**, 5, e10454	3.9	5
229	Extrathoracic subclavian artery aneurysm in a patient with suspected genetic arteriopathy. *Journal of Vascular Surgery Cases and Innovative Techniques*, **2021**, 7, 46-50	1.1	0
228	True radial artery aneurysm in a patient with somatic mosaicism for a mutation in platelet-derived growth factor receptor gene. *Journal of Vascular Surgery Cases and Innovative Techniques*, **2021**, 7, 567-571	1.1	
227	Biallelic variants in , which encodes a WNT-signaling-related protein, in four new families with recessively inherited osteogenesis imperfecta.. *Human Genetics and Genomics Advances*, **2021**, 2, 100051	0.8	1
226	6q25.1 (TAB2) microdeletion is a risk factor for hypoplastic left heart: a case report that expands the phenotype. *BMC Cardiovascular Disorders*, **2020**, 20, 137	2.3	8
225	Setting a research agenda for vascular Ehlers-Danlos syndrome using a patient and stakeholder engagement model. *Journal of Vascular Surgery*, **2020**, 72, 1436-1444.e2	3.5	3
224	Introduction to Osteogenesis Imperfecta **2020**, 3-9		
223	Hearing loss in individuals with osteogenesis imperfecta in North America: Results from a multicenter study. *American Journal of Medical Genetics, Part A*, **2020**, 182, 697-704	2.5	9
222	Assessment of the Information Sources and Interest in Research Collaboration Among Individuals with Vascular Ehlers-Danlos Syndrome. *Annals of Vascular Surgery*, **2020**, 62, 326-334	1.7	6

221	Orthopaedic Conditions Associated with Aneurysms. *JBJS Reviews*, **2020**, 8, e0122	2.6	2
220	The Ehlers-Danlos syndromes. *Nature Reviews Disease Primers*, **2020**, 6, 64	51.1	47
219	A multi-institutional experience in vascular Ehlers-Danlos syndrome diagnosis. *Journal of Vascular Surgery*, **2020**, 71, 149-157	3.5	9
218	A Multicenter Observational Cohort Study to Evaluate the Effects of Bisphosphonate Exposure on Bone Mineral Density and Other Health Outcomes in Osteogenesis Imperfecta. *JBMR Plus*, **2019**, 3, e10118	3.9	7
217	Compound heterozygosity for a frameshift mutation and an upstream deletion that reduces expression of SERPINH1 in siblings with a moderate form of osteogenesis imperfecta. *American Journal of Medical Genetics, Part A*, **2019**, 179, 1466-1475	2.5	3
216	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	3.5	12
215	Mobility in osteogenesis imperfecta: a multicenter North American study. *Genetics in Medicine*, **2019**, 21, 2311-2318	8.1	6
214	Growth characteristics in individuals with osteogenesis imperfecta in North America: results from a multicenter study. *Genetics in Medicine*, **2019**, 21, 275-283	8.1	15
213	Trends over 42 years in the Adult Medical Genetics Clinic at the University of Washington. *Genetics in Medicine*, **2019**, 21, 1457-1461	8.1	4
212	Mutations That Alter the Carboxy-Terminal-Propeptide Cleavage Site of the Chains of Type I Procollagen Are Associated With a Unique Osteogenesis Imperfecta Phenotype. *Journal of Bone and Mineral Research*, **2018**, 33, 1260-1271	6.3	20
211	Testing patterns for genetically triggered aortic and arterial aneurysms and dissections at an academic center. *Journal of Vascular Surgery*, **2018**, 68, 701-711	3.5	12
210	Monoallelic and biallelic CREB3L1 variant causes mild and severe osteogenesis imperfecta, respectively. *Genetics in Medicine*, **2018**, 20, 411-419	8.1	28
209	Substitutions for arginine at position 780 in triple helical domain of the α1(I) chain alter folding of the type I procollagen molecule and cause osteogenesis imperfecta. *PLoS ONE*, **2018**, 13, e0200264	3.7	12
208	Heterozygous WNT1 variant causing a variable bone phenotype. *American Journal of Medical Genetics, Part A*, **2018**, 176, 2419-2424	2.5	4
207	A multicenter study to evaluate pulmonary function in osteogenesis imperfecta. *Clinical Genetics*, **2018**, 94, 502-511	4	20
206	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
205	6q25.1 (TAB2) microdeletion syndrome: Congenital heart defects and cardiomyopathy. *American Journal of Medical Genetics, Part A*, **2017**, 173, 1848-1857	2.5	13
204	Endovascular Repair of Internal Mammary Artery Aneurysms in 2 Sisters with SMAD3 Mutation. *Annals of Vascular Surgery*, **2017**, 41, 283.e5-283.e9	1.7	8

203	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
202	Osteogenesis imperfecta. *Nature Reviews Disease Primers*, **2017**, 3, 17052	51.1	292
201	Cesarean delivery is not associated with decreased at-birth fracture rates in osteogenesis imperfecta. *Genetics in Medicine*, **2016**, 18, 570-6	8.1	27
200	The challenge of comprehensive and consistent sequence variant interpretation between clinical laboratories. *Genetics in Medicine*, **2016**, 18, 20-4	8.1	46
199	Current Practices and the Provider Perspectives on Inconclusive Genetic Test Results for Osteogenesis Imperfecta in Children with Unexplained Fractures: ELSI Implications. *Journal of Law, Medicine and Ethics*, **2016**, 44, 514-9	1.2	5
198	COL1A1 and COL1A2 sequencing results in cohort of patients undergoing evaluation for potential child abuse. *American Journal of Medical Genetics, Part A*, **2016**, 170, 1858-62	2.5	15
197	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
196	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
195	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
194	Molecular Outcome, Prediction, and Clinical Consequences of Splice Variants in COL1A1, Which Encodes the pro□(I) Chains of Type I Procollagen. *Human Mutation*, **2015**, 36, 728-39	4.7	16
193	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
192	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
191	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
190	Haploinsufficiency for Mutations in Type I Collagen Genes: Mechanisms and Clinical Effects **2014**, 125-127		1
189	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
188	Pregnancy-related deaths and complications in women with vascular Ehlers-Danlos syndrome. *Genetics in Medicine*, **2014**, 16, 874-80	8.1	86
187	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
186	FKBP14-related Ehlers-Danlos syndrome: expansion of the phenotype to include vascular complications. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 1750-5	2.5	20

185	Ehlers-Danlos syndrome: a showcase of conditions that lead to understanding matrix biology. *Matrix Biology*, **2014**, 33, 10-5	11.4	45
184	EhlersDanlos Syndrome **2013**, 1-23		
183	WNT1 mutations in families affected by moderately severe and progressive recessive osteogenesis imperfecta. *American Journal of Human Genetics*, **2013**, 92, 590-7	11	156
182	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
181	Allelic background of LEPRE1 mutations that cause recessive forms of osteogenesis imperfecta in different populations. *Molecular Genetics & Genomic Medicine*, **2013**, 1, 194-205	2.3	16
180	Recessively inherited forms of osteogenesis imperfecta. *Annual Review of Genetics*, **2012**, 46, 475-97	14.5	78
179	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
178	Heritable collagen disorders: the paradigm of the Ehlers-Danlos syndrome. *Journal of Investigative Dermatology*, **2012**, 132, E6-11	4.3	38
177	Characterization of tissue-specific and developmentally regulated alternative splicing of exon 64 in the COL5A1 gene. *Connective Tissue Research*, **2012**, 53, 267-76	3.3	3
176	EMQN best practice guidelines for the laboratory diagnosis of osteogenesis imperfecta. *European Journal of Human Genetics*, **2012**, 20, 11-9	5.3	99
175	Loss-of-function mutations in TGFB2 cause a syndromic presentation of thoracic aortic aneurysm. *Nature Genetics*, **2012**, 44, 922-7	36.3	323
174	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
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	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
171	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
170	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
169	Mutations in the Gene Encoding the RER Protein FKBP65 Cause Autosomal-Recessive Osteogenesis Imperfecta. *American Journal of Human Genetics*, **2010**, 87, 572-573	11	13
168	Generalized connective tissue disease in Crtap-/- mouse. *PLoS ONE*, **2010**, 5, e10560	3.7	45

167	Mutation and polymorphism spectrum in osteogenesis imperfecta type II: implications for genotype-phenotype relationships. *Human Molecular Genetics*, **2009**, 18, 463-71	5.6	95
166	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
165	Molecular mechanisms of classical Ehlers-Danlos syndrome (EDS). *Human Mutation*, **2009**, 30, 995-1002	4.7	65
164	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
163	Natural variation in four human collagen genes across an ethnically diverse population. *Genomics*, **2008**, 91, 307-14	4.3	35
162	The bicuspid aortic valve: an integrated phenotypic classification of leaflet morphology and aortic root shape. *Heart*, **2008**, 94, 1634-8	5.1	287
161	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
160	Gene targeting of mutant COL1A2 alleles in mesenchymal stem cells from individuals with osteogenesis imperfecta. *Molecular Therapy*, **2008**, 16, 187-93	11.7	73
159	CRTAP and LEPRE1 mutations in recessive osteogenesis imperfecta. *Human Mutation*, **2008**, 29, 1435-42	4.7	172
158	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
157	Usefulness of bicuspid aortic valve phenotype to predict elastic properties of the ascending aorta. *American Journal of Cardiology*, **2007**, 99, 686-90	3	114
156	Type XXVII collagen at the transition of cartilage to bone during skeletogenesis. *Bone*, **2007**, 41, 535-42	4.7	56
155	Genetic evaluation of suspected osteogenesis imperfecta (OI). *Genetics in Medicine*, **2006**, 8, 383-8	8.1	56
154	Aneurysm syndromes caused by mutations in the TGF-beta receptor. *New England Journal of Medicine*, **2006**, 355, 788-98	59.2	1243
153	2005 ASHG Presidential Address. If only we spoke the same language--we would have so much to discuss. *American Journal of Human Genetics*, **2006**, 78, 368-72	11	
152	2005 ASHG Award for Excellence in Human Genetics Education. Introductory speech of Joseph D. McInerney. *American Journal of Human Genetics*, **2006**, 78, 373	11	
151	CRTAP is required for prolyl 3- hydroxylation and mutations cause recessive osteogenesis imperfecta. *Cell*, **2006**, 127, 291-304	56.2	394
150	The role of genomics in medicine--past, present and future. *Journal of Zhejiang University: Science B*, **2006**, 7, 159-60	4.5	

149	Bovine model of Marfan syndrome results from an amino acid change (c.3598G > A, p.E1200K) in a calcium-binding epidermal growth factor-like domain of fibrillin-1. *Human Mutation*, **2005**, 25, 348-52	4.7	18
148	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
147	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
146	Gene targeting in stem cells from individuals with osteogenesis imperfecta. *Science*, **2004**, 303, 1198-201	33.3	244
145	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
144	Determination of the molecular basis of Marfan syndrome: a growth industry. *Journal of Clinical Investigation*, **2004**, 114, 161-163	15.9	32
143	Determination of the molecular basis of Marfan syndrome: a growth industry. *Journal of Clinical Investigation*, **2004**, 114, 161-3	15.9	9
142	Neurological presentation of Ehlers-Danlos syndrome type IV in a family with parental mosaicism. *Clinical Genetics*, **2003**, 63, 510-5	4	26
141	Identification, characterization and expression analysis of a new fibrillar collagen gene, COL27A1. *Matrix Biology*, **2003**, 22, 3-14	11.4	96
140	Testing for osteogenesis imperfecta in cases of suspected non-accidental injury. *Journal of Medical Genetics*, **2002**, 39, 382-6	5.8	64
139	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
138	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
137	A variant of osteogenesis imperfecta type IV with resolving kyphomelia is caused by a novel COL1A2 mutation. *Journal of Medical Genetics*, **2002**, 39, 128-32	5.8	5
136	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
135	Killing the messenger: new insights into nonsense-mediated mRNA decay. *Journal of Clinical Investigation*, **2002**, 109, 3-6	15.9	103
134	Killing the messenger: new insights into nonsense-mediated mRNA decay. *Journal of Clinical Investigation*, **2002**, 109, 3-6	15.9	46
133	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
132	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

131	Folding defects in fibrillar collagens. *Philosophical Transactions of the Royal Society B: Biological Sciences*, **2001**, 356, 151-7; discussion 157-8	5.8	42
130	An exception to the rule. *New England Journal of Medicine*, **2001**, 345, 1203-5	59.2	5
129	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
128	Osteogenesis imperfecta: mode of delivery and neonatal outcome. *Obstetrics and Gynecology*, **2001** , 97, 66-9	4.9	32
127	Osteogenesis imperfecta: perspectives and opportunities. *Current Opinion in Pediatrics*, **2000**, 12, 603-9	3.2	38
126	Collagens: building blocks at the end of the development line. *Clinical Genetics*, **2000**, 58, 270-9	4	35
125	Partial COL1A2 gene duplication produces features of osteogenesis imperfecta and Ehlers-Danlos syndrome type VII. *Human Genetics*, **2000**, 106, 19-28	6.3	33
124	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
123	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
122	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
121	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
120	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
119	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
118	Molecular genetic pathology: coming of age in the molecular world. *Journal of Molecular Diagnostics*, **1999**, 1, 3-4	5.1	2
117	Molecular genetic pathology. Coming of age in the molecular world. *American Journal of Pathology*, **1999**, 155, 673-4	5.8	3
116	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		1
115	Pedigrees-publish? or perish the thought?. *American Journal of Human Genetics*, **1998**, 63, 678-81	11	12
114	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

113	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
112	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
111	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
110	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
109	Strategies and outcomes of prenatal diagnosis for osteogenesis imperfecta: a review of biochemical and molecular studies completed in 129 pregnancies. *Prenatal Diagnosis*, **1997**, 17, 559-70	3.2	10
108	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
107	A dimorphic Alu Sb-like insertion in COL3A1 is ethnic-specific. *Journal of Molecular Evolution*, **1996**, 42, 117-23	3.1	12
106	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
105	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
104	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
103	A Gly238Ser substitution in the alpha 2 chain of type I collagen results in osteogenesis imperfecta type III. *Human Genetics*, **1995**, 95, 215-8	6.3	8
102	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
101	Etiology of osteogenesis imperfecta: an overview of biochemical and molecular genetic analyses. *Connective Tissue Research*, **1995**, 31, 257-9	3.3	5
100	Substitutions of aspartic acid for glycine-220 and of arginine for glycine-664 in the triple helix of the pro alpha 1(I) chain of type I procollagen produce lethal osteogenesis imperfecta and disrupt the ability of collagen fibrils to incorporate crystalline hydroxyapatite. *Biochemical Journal*, **1995**, 311 (Pt 3), 815-20	3.8	20
99	Cerebrovascular complications in Ehlers-Danlos syndrome type IV. *Annals of Neurology*, **1995**, 38, 960-4	9.4	153
98	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
97	A Gly859Ser substitution in the triple helical domain of the alpha 2 chain of type I collagen resulting in osteogenesis imperfecta type III in two unrelated individuals. *Human Mutation*, **1994**, 3, 391-4	4.7	12
96	Molecular basis of hereditary disorders of connective tissue. *Annual Review of Medicine*, **1994**, 45, 149-63	17.4	33

95	Sequence of the coding region of the bovine fibrillin cDNA and localization to bovine chromosome 10. *Genomics*, **1994**, 23, 480-5	4.3	25
94	Molecular genetics of chondrodysplasias, including clues to development, structure, and function. *Current Opinion in Rheumatology*, **1994**, 6, 345-50	5.3	9
93	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
92	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
91	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
90	A novel glycine to glutamic acid substitution at position 343 in the alpha 2 chain of type I collagen in an individual with lethal osteogenesis imperfecta. *Human Molecular Genetics*, **1993**, 2, 2175-7	5.6	11
89	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
88	Molecular heterogeneity in osteogenesis imperfecta type I. *American Journal of Medical Genetics Part A*, **1993**, 45, 223-7		27
87	Homology-mediated recombination between type I collagen gene exons results in an internal tandem duplication and lethal osteogenesis imperfecta. *Human Mutation*, **1993**, 2, 21-7	4.7	10
86	A single amino acid deletion in the alpha 2(I) chain of type I collagen produces osteogenesis imperfecta type III. *Human Genetics*, **1993**, 90, 621-8	6.3	14
85	First-trimester prenatal diagnosis of osteogenesis imperfecta type II by DNA analysis and sonography. *Prenatal Diagnosis*, **1993**, 13, 589-96	3.2	30
84	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
83	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
82	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
81	Abnormal fibrillin metabolism in bovine Marfan syndrome. *American Journal of Pathology*, **1993**, 142, 803-10	5.8	16
80	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
79	Osteogenesis imperfecta. *Annual Review of Medicine*, **1992**, 43, 269-82	17.4	223
78	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

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