Peter H Byers

List of Publications by Year
in descending order

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		8181	11939
240	20,457	76	134
papers	citations	h-index	g-index

248	248	248	11690
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Aortic dissection in pregnancy and the postpartum period. Seminars in Vascular Surgery, 2022, 35, 60-68.	2.8	5
2	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.	2.8	8
3	Gregor Mendel and the concepts of dominance and recessiveness. Nature Reviews Genetics, 2022, 23, 387-388.	16.3	11
4	Abnormal Bone Collagen Crossâ€Linking in Osteogenesis Imperfecta/Bruck Syndrome Caused by Compound Heterozygous <scp>PLOD2</scp> Mutations. JBMR Plus, 2021, 5, e10454.	2.7	17
5	Extrathoracic subclavian artery aneurysm in a patient with suspected genetic arteriopathy. Journal of Vascular Surgery Cases and Innovative Techniques, 2021, 7, 46-50.	0.6	2
6	Marfan syndrome resulting from a rare pathogenic FBN1 Âvariant, ascertained through a proband with IgG4 â€relatedÂarteriopathy. American Journal of Medical Genetics, Part A, 2021, 185, 2180-2189.	1.2	0
7	Subtle differences in autonomic symptoms in people diagnosed with hypermobile Ehlersâ€“Danlos syndrome and hypermobility spectrum disorders. American Journal of Medical Genetics, Part A, 2021, 185, 2012-2025.	1.2	7
8	2020 McKusick Award address. American Journal of Human Genetics, 2021, 108, 761-763.	6.2	0
9	Caffey disease is associated with distinct arginine to cysteine substitutions in the proÎ±1(I) chain of type I procollagen. Genetics in Medicine, 2021, 23, 2378-2385.	2.4	1
10	A call for direct sequencing of full-length RNAs to identify all modifications. Nature Genetics, 2021, 53, 1113-1116.	21.4	33
11	Genetic counseling and screening of consanguineous couples and their offspring practice resource: Focused Revision. Journal of Genetic Counseling, 2021, 30, 1354-1357.	1.6	1
12	True radial artery aneurysm in a patient with somatic mosaicism for a mutation in platelet-derived growth factor receptor Î² gene. Journal of Vascular Surgery Cases and Innovative Techniques, 2021, 7, 567-571.	0.6	1
13	Biallelic variants in MESD, which encodes a WNT-signaling-related protein, in four new families with recessively inherited osteogenesis imperfecta. Human Genetics and Genomics Advances, 2021, 2, 100051.	1.7	3
14	A multi-institutional experience in vascular Ehlers-Danlos syndrome diagnosis. Journal of Vascular Surgery, 2020, 71, 149-157.	1.1	28
15	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.	1.2	17
16	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.	0.9	14
17	Orthopaedic Conditions Associated with Aneurysms. JBJS Reviews, 2020, 8, e0122-e0122.	2.0	3
18	The Ehlersâ€“Danlos syndromes. Nature Reviews Disease Primers, 2020, 6, 64.	30.5	144

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19	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.	1.7	10
20	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.	1.1	15
21	Introduction to Osteogenesis Imperfecta. , 2020, , 3-9.		0
22	Growth characteristics in individuals with osteogenesis imperfecta in North America: results from a multicenter study. Genetics in Medicine, 2019, 21, 275-283.	2.4	34
23	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.	2.7	22
24	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.	1.2	6
25	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.	1.1	39
26	Mobility in osteogenesis imperfecta: a multicenter North American study. Genetics in Medicine, 2019, 21, 2311-2318.	2.4	15
27	Trends over 42 years in the Adult Medical Genetics Clinic at the University of Washington. Genetics in Medicine, 2019, 21, 1457-1461.	2.4	6
28	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.	2.8	21
29	Testing patterns for genetically triggered aortic and arterial aneurysms and dissections at an academic center. Journal of Vascular Surgery, 2018, 68, 701-711.	1.1	20
30	Monoallelic and biallelic CREB3L1 variant causes mild and severe osteogenesis imperfecta, respectively. Genetics in Medicine, 2018, 20, 411-419.	2.4	47
31	Heterozygous WNT1 variant causing a variable bone phenotype. American Journal of Medical Genetics, Part A, 2018, 176, 2419-2424.	1.2	11
32	A multicenter study to evaluate pulmonary function in osteogenesis imperfecta. Clinical Genetics, 2018, 94, 502-511.	2.0	33
33	Substitutions for arginine at position 780 in triple helical domain of the Î±1(I) chain alter folding of the type I procollagen molecule and cause osteogenesis imperfecta. PLoS ONE, 2018, 13, e0200264.	2.5	16
34	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.	3.2	34
35	6q25.1 (TAB2) microdeletion syndrome: Congenital heart defects and cardiomyopathy. American Journal of Medical Genetics, Part A, 2017, 173, 1848-1857.	1.2	22
36	Endovascular Repair of Internal Mammary Artery Aneurysms in 2 Sisters with SMAD3 Mutation. Annals of Vascular Surgery, 2017, 41, 283.e5-283.e9.	0.9	10

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37	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.	1.6	239
38	Osteogenesis imperfecta. Nature Reviews Disease Primers, 2017, 3, 17052.	30.5	481
39	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.	6.2	100
40	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, 514-519.	0.9	7
41	COL1A1 and COL1A2 sequencing results in cohort of patients undergoing evaluation for potential child abuse. American Journal of Medical Genetics, Part A, 2016, 170, 1858-1862.	1.2	19
42	Cesarean delivery is not associated with decreased at-birth fracture rates in osteogenesis imperfecta. Genetics in Medicine, 2016, 18, 570-576.	2.4	39
43	The challenge of comprehensive and consistent sequence variant interpretation between clinical laboratories. Genetics in Medicine, 2016, 18, 20-24.	2.4	63
44	Molecular Outcome, Prediction, and Clinical Consequences of Splice Variants inCOL1A1, Which Encodes the proÎ±1(I) Chains of Type I Procollagen. Human Mutation, 2015, 36, 728-739.	2.5	22
45	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.	1.2	44
46	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.	1.6	31
47	A crossâ€sectional multicenter study ofÂosteogenesis imperfecta in North AmericaÂâ€“Âresults from the linked clinical research centers. Clinical Genetics, 2015, 87, 133-140.	2.0	59
48	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.	2.8	33
49	Haploinsufficiency for Mutations in Type I Collagen Genes: Mechanisms and Clinical Effects. , 2014, , 125-127.		1
50	Pre- and Postnatal Transplantation of Fetal Mesenchymal Stem Cells in Osteogenesis Imperfecta: A Two-Center Experience. Stem Cells Translational Medicine, 2014, 3, 255-264.	3.3	162
51	Pregnancy-related deaths and complications in women with vascular Ehlersâ€“Danlos syndrome. Genetics in Medicine, 2014, 16, 874-880.	2.4	127
52	Survival is affected by mutation type and molecular mechanism in vascular Ehlersâ€“Danlos syndrome (EDS type IV). Genetics in Medicine, 2014, 16, 881-888.	2.4	217
53	FKBP14-related Ehlers-Danlos syndrome: Expansion of the phenotype to include vascular complications. , 2014, 164, 1750-1755.		23
54	Ehlersâ€“Danlos syndrome: A showcase of conditions that lead to understanding matrix biology. Matrix Biology, 2014, 33, 10-15.	3.6	57

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55	Refining the structure and content of clinical genomic reports. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2014, 166, 85-92.	1.6	37
56	Ehlersâ€“Danlos Syndrome. , 2013, , 1-23.		2
57	WNT1 Mutations in Families Affected by Moderately Severe and Progressive Recessive Osteogenesis Imperfecta. American Journal of Human Genetics, 2013, 92, 590-597.	6.2	179
58	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.	2.9	135
59	Allelic background of LEPRE1 mutations that cause recessive forms of osteogenesis imperfecta in different populations. Molecular Genetics & Genomic Medicine, 2013, 1, 194-205.	1.2	18
60	Heritable Collagen Disorders: The Paradigm of the Ehlersâ€”Danlos Syndrome. Journal of Investigative Dermatology, 2012, 132, E6-E11.	0.7	45
61	Characterization of Tissue-Specific and Developmentally Regulated Alternative Splicing of Exon 64 in theCOL5A1Gene. Connective Tissue Research, 2012, 53, 267-276.	2.3	7
62	EMQN best practice guidelines for the laboratory diagnosis of osteogenesis imperfecta. European Journal of Human Genetics, 2012, 20, 11-19.	2.8	107
63	Loss-of-function mutations in TGFB2 cause a syndromic presentation of thoracic aortic aneurysm. Nature Genetics, 2012, 44, 922-927.	21.4	391
64	Recessively Inherited Forms of Osteogenesis Imperfecta. Annual Review of Genetics, 2012, 46, 475-497.	7.6	94
65	Haploinsufficiency of SF3B4, a Component of the Pre-mRNA Spliceosomal Complex, Causes Nager Syndrome. American Journal of Human Genetics, 2012, 90, 925-933.	6.2	188
66	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.	2.9	118
67	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.	2.4	98
68	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.	2.4	64
69	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.	6.2	291
70	Mutations in the Gene Encoding the RER Protein FKBP65 Cause Autosomal-Recessive Osteogenesis Imperfecta. American Journal of Human Genetics, 2010, 86, 551-559.	6.2	278
71	Mutations in the Gene Encoding the RER Protein FKBP65 Cause Autosomal-Recessive Osteogenesis Imperfecta. American Journal of Human Genetics, 2010, 87, 572-573.	6.2	13
72	Generalized Connective Tissue Disease in Crtap-/- Mouse. PLoS ONE, 2010, 5, e10560.	2.5	52

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73	Mutation and polymorphism spectrum in osteogenesis imperfecta type II: implications for genotype-phenotype relationships. Human Molecular Genetics, 2009, 18, 1893-1895.	2.9	3
74	Mutation and polymorphism spectrum in osteogenesis imperfecta type II: implications for genotypeâ€“phenotype relationships. Human Molecular Genetics, 2009, 18, 463-471.	2.9	107
75	Successful Endovascular Repair of Acute Type B Aortic Dissection in Undiagnosed Ehlersâ€“Danlos Syndrome Type IV. European Journal of Vascular and Endovascular Surgery, 2009, 38, 608-609.	1.5	22
76	Molecular mechanisms of classical Ehlers-Danlos syndrome (EDS). Human Mutation, 2009, 30, 995-1002.	2.5	70
77	Analysis of multigenerational families with thoracic aortic aneurysms and dissections due to TGFBR1 or TGFBR2 mutations. Journal of Medical Genetics, 2009, 46, 607-613.	3.2	172
78	CRTAPandLEPRE1mutations in recessive osteogenesis imperfecta. Human Mutation, 2008, 29, 1435-1442.	2.5	196
79	Natural variation in four human collagen genes across an ethnically diverse population. Genomics, 2008, 91, 307-314.	2.9	47
80	The bicuspid aortic valve: an integrated phenotypic classification of leaflet morphology and aortic root shape. Heart, 2008, 94, 1634-1638.	2.9	364
81	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.	3.4	57
82	Gene Targeting of Mutant COL1A2 Alleles in Mesenchymal Stem Cells From Individuals With Osteogenesis Imperfecta. Molecular Therapy, 2008, 16, 187-193.	8.2	78
83	Type XXVII collagen at the transition of cartilage to bone during skeletogenesis. Bone, 2007, 41, 535-542.	2.9	67
84	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.	2.5	620
85	Usefulness of Bicuspid Aortic Valve Phenotype to Predict Elastic Properties of the Ascending Aorta. American Journal of Cardiology, 2007, 99, 686-690.	1.6	138
86	If Only We Spoke the Same Languageâ€”We Would Have So Much to Discuss*. American Journal of Human Genetics, 2006, 78, 368-372.	6.2	1
87	Introductory Speech for Joseph D. McInerney*. American Journal of Human Genetics, 2006, 78, 373.	6.2	0
88	CRTAP Is Required for Prolyl 3- Hydroxylation and Mutations Cause Recessive Osteogenesis Imperfecta. Cell, 2006, 127, 291-304.	28.9	465
89	The role of genomics in medicine â€” Past, present and future. Journal of Zhejiang University: Science B, 2006, 7, 159-160.	2.8	0
90	Genetic evaluation of suspected osteogenesis imperfecta (OI). Genetics in Medicine, 2006, 8, 383-388.	2.4	78

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91	Aneurysm Syndromes Caused by Mutations in the TGF-Î² Receptor. New England Journal of Medicine, 2006, 355, 788-798.	27.0	1,490
92	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, 348-352.	2.5	21
93	Novel missense mutations in the TRPS1 transcription factor define the nuclear localization signal. European Journal of Human Genetics, 2004, 12, 121-126.	2.8	52
94	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.	2.5	95
95	Gene Targeting in Stem Cells from Individuals with Osteogenesis Imperfecta. Science, 2004, 303, 1198-1201.	12.6	271
96	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.	6.2	147
97	Determination of the molecular basis of Marfan syndrome: a growth industry. Journal of Clinical Investigation, 2004, 114, 161-163.	8.2	36
98	Determination of the molecular basis of Marfan syndrome: a growth industry. Journal of Clinical Investigation, 2004, 114, 161-163.	8.2	16
99	Neurological presentation of Ehlers-Danlos syndrome type IV in a family with parental mosaicism. Clinical Genetics, 2003, 63, 510-515.	2.0	27
100	Identification, characterization and expression analysis of a new fibrillar collagen gene, COL27A1. Matrix Biology, 2003, 22, 3-14.	3.6	112
101	Testing for osteogenesis imperfecta in cases of suspected non-accidental injury. Journal of Medical Genetics, 2002, 39, 382-386.	3.2	72
102	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.	0.8	74
103	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.	3.2	42
104	A variant of osteogenesis imperfecta type IV with resolving kyphomelia is caused by a novel COL1A2 mutation. Journal of Medical Genetics, 2002, 39, 128-132.	3.2	7
105	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.	6.2	100
106	Killing the messenger: new insights into nonsense-mediated mRNA decay. Journal of Clinical Investigation, 2002, 109, 3-6.	8.2	114
107	Killing the messenger: new insights into nonsense-mediated mRNA decay. Journal of Clinical Investigation, 2002, 109, 3-6.	8.2	73
108	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.	6.2	168

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109	Osteogenesis imperfecta: mode of delivery and neonatal outcome. Obstetrics and Gynecology, 2001, 97, 66-69.	2.4	44
110	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.	2.5	38
111	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.	3.2	51
112	Folding defects in fibrillar collagens. Philosophical Transactions of the Royal Society B: Biological Sciences, 2001, 356, 151-158.	4.0	49
113	An Exception to the Rule. New England Journal of Medicine, 2001, 345, 1203-1205.	27.0	9
114	Osteogenesis imperfecta: perspectives and opportunities. Current Opinion in Pediatrics, 2000, 12, 603-609.	2.0	50
115	Collagens: building blocks at the end of the development line. Clinical Genetics, 2000, 58, 270-279.	2.0	43
116	Partial COL1A2 gene duplication produces features of osteogenesis imperfecta and Ehlers-Danlos syndrome type VII. Human Genetics, 2000, 106, 19-28.	3.8	37
117	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.	6.2	122
118	Clinical and Genetic Features of Ehlersâ€“Danlos Syndrome Type IV, the Vascular Type. New England Journal of Medicine, 2000, 342, 673-680.	27.0	1,219
119	Multiple Vascular and Bowel Ruptures in an Adolescent Male with Sporadic Ehlers-Danlos Syndrome Type IV. Pediatric and Developmental Pathology, 1999, 2, 86-93.	1.0	26
120	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
121	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.	6.2	348
122	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.	6.2	62
123	Molecular Genetic Pathology. Journal of Molecular Diagnostics, 1999, 1, 3-4.	2.8	5
124	Molecular Genetic Pathology. American Journal of Pathology, 1999, 155, 673-674.	3.8	4
125	Large kindred with Ehlersâ€Đanlos 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. American Journal of Medical Genetics Part A, 1999, 82, 305-311.	2.4	1
126	Pedigreesâ€”Publish? or Perish the Thought?. American Journal of Human Genetics, 1998, 63, 678-681.	6.2	14

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127	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.	2.9	65
128	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.	6.2	73
129	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.	0.7	71
130	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
131	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
132	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.	2.3	11
133	Studies of collagen synthesis and structure in the differentiation of child abuse from osteogenesis imperfecta. Journal of Pediatrics, 1996, 128, 542-547.	1.8	78
134	A dimorphic Alu Sb-like insertion in COL3A1 is ethnic-specific. Journal of Molecular Evolution, 1996, 42, 117-123.	1.8	12
135	Delineation of the Marfan phenotype associated with mutations in exons 23â€“32 of theFBN1 gene. , 1996, 62, 233-242.		120
136	A translocation interrupts the COL5A1 gene in a patient with Ehlersâ€“Danlos syndrome and hypomelanosis of Ito. Nature Genetics, 1996, 13, 361-365.	21.4	116
137	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.	1.1	93
138	Substitutions of aspartic acid for glycine-220 and of arginine for glycine-664 in the triple helix of the pro Î±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, 815-820.	3.7	25
139	Cerebrovascular complications in Ehlers-Danlos syndrome type IV. Annals of Neurology, 1995, 38, 960-964.	5.3	176
140	A Gly238Ser substitution in the ?2 chain of type I collagen results in osteogenesis imperfecta type III. Human Genetics, 1995, 95, 215-8.	3.8	10
141	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.	3.4	129
142	Frontiers in Rehabilitation Medicine: Osteogenesis Imperfecta, Overview of a Conference. Connective Tissue Research, 1995, 31, 253-255.	2.3	3
143	Etiology of Osteogenesis Imperfecta: An Overview of Biochemical and Molecular Genetic Analyses. Connective Tissue Research, 1995, 31, 257-259.	2.3	5
144	Ehlers-Danlos Syndrome: Recent Advances and Current Understanding of the Clinical and Genetic Heterogeneity.. Journal of Investigative Dermatology, 1994, 103, 47S-52S.	0.7	57

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145	A Gly859Ser substitution in the triple helical domain of the Î±2 chain of type I collagen resulting in osteogenesis imperfecta type III in two unrelated individuals. Human Mutation, 1994, 3, 391-394.	2.5	14
146	MOLECULAR BASIS OF HEREDITARY DISORDERS OF CONNECTIVE TISSUE. Annual Review of Medicine, 1994, 45, 149-163.	12.2	37
147	Sequence of the Coding Region of the Bovine Fibrillin cDNA and Localization to Bovine Chromosome 10. Genomics, 1994, 23, 480-485.	2.9	26
148	Molecular genetics of chondrodysplasias, including clues to development, structure, and function. Current Opinion in Rheumatology, 1994, 6, 345.	4.3	13
149	Osteogenesis imperfecta type I: molecular heterogeneity for COL1A1 null alleles of type I collagen. American Journal of Human Genetics, 1994, 55, 638-47.	6.2	107
150	Molecular heterogeneity in osteogenesis imperfecta type I. American Journal of Medical Genetics Part A, 1993, 45, 223-227.	2.4	44
151	Homology-mediated recombination between type I collagen gene exons results in an internal tandem duplication and lethal osteogenesis imperfecta. Human Mutation, 1993, 2, 21-27.	2.5	11
152	A single amino acid deletion in the ?2(I) chain of type I collagen produces osteogenesis imperfecta type III. Human Genetics, 1993, 90, 621-8.	3.8	15
153	First-trimester prenatal diagnosis of osteogenesis imperfecta type II by DNA analysis and sonography. Prenatal Diagnosis, 1993, 13, 589-596.	2.3	40
154	Sequence and Characterization of the Complete Human Thrombospondin 2 cDNA: Potential Regulatory Role for the 3â€² Untranslated Region. Genomics, 1993, 17, 225-229.	2.9	32
155	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.	3.2	60
156	A novel glycine to glutamic acid substitution at position 343 in the Î±2 chain of type I collagen in an individual with lethal osteogenesis imperfecta. Human Molecular Genetics, 1993, 2, 2175-2177.	2.9	14
157	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.	2.9	45
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