Brendan H Lee

List of Publications by Year
in descending order

226 papers	14,451 citations	19657 61 h-index	23533 111 g-index
265 all docs	265 docs citations	265 times ranked	16812 citing authors

#	Article	IF	Citations
1	Linkage of Marfan syndrome and a phenotypically related disorder to two different fibrillin genes. Nature, 1991, 352, 330-334.	27.8	676
2	Missense mutations abolishing DNA binding of the osteoblast-specific transcription factor OSF2/CBFA1 in cleidocranial dysplasia. Nature Genetics, 1997, 16, 307-310.	21.4	548
3	Limb and kidney defects in Lmx1b mutant mice suggest an involvement of LMX1B in human nail patella syndrome. Nature Genetics, 1998, 19, 51-55.	21.4	499
4	Mutations in LMX1B cause abnormal skeletal patterning and renal dysplasia in nail patella syndrome. Nature Genetics, 1998, 19, 47-50.	21.4	471
5	CRTAP Is Required for Prolyl 3- Hydroxylation and Mutations Cause Recessive Osteogenesis Imperfecta. Cell, 2006, 127, 291-304.	28.9	465
6	Dimorphic effects of Notch signaling in bone homeostasis. Nature Medicine, 2008, 14, 299-305.	30.7	361
7	Use of Exome Sequencing for Infants in Intensive Care Units. JAMA Pediatrics, 2017, 171, e173438.	6.2	348
8	Dominance of SOX9 function over RUNX2 during skeletogenesis. Proceedings of the National Academy of Sciences of the United States of America, 2006, 103, 19004-19009.	7.1	325
9	WNT1 Mutations in Early-Onset Osteoporosis and Osteogenesis Imperfecta. New England Journal of Medicine, 2013, 368, 1809-1816.	27.0	308
10	The Osteogenic Niche Promotes Early-Stage Bone Colonization of Disseminated Breast Cancer Cells. Cancer Cell, 2015, 27, 193-210.	16.8	308
11	Deficiency of Cartilage-Associated Protein in Recessive Lethal Osteogenesis Imperfecta. New England Journal of Medicine, 2006, 355, 2757-2764.	27.0	307
12	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
13	Effect of Genetic Diagnosis on Patients with Previously Undiagnosed Disease. New England Journal of Medicine, 2018, 379, 2131-2139.	27.0	261
14	Excessive transforming growth factor-Î² signaling is a common mechanism in osteogenesis imperfecta. Nature Medicine, 2014, 20, 670-675.	30.7	237
15	Branched-chain amino acid metabolism: from rare Mendelian diseases to more common disorders. Human Molecular Genetics, 2014, 23, R1-R8.	2.9	234
16	miRNA-34c regulates Notch signaling during bone development. Human Molecular Genetics, 2012, 21, 2991-3000.	2.9	210
17	CRTAP and LEPRE1 mutations in recessive osteogenesis imperfecta. Human Mutation, 2008, 29, 1435-1442.	2.5	196
18	Regulation of glomerular basement membrane collagen expression by LMX1B contributes to renal disease in nail patella syndrome. Nature Genetics, 2001, 27, 205-208.	21.4	189

#	Article	IF	Citations
19	Cross-sectional multicenter study of patients with urea cycle disorders in the United States. Molecular Genetics and Metabolism, 2008, 94, 397-402.	1.1	189
20	Requirement of argininosuccinate lyase for systemic nitric oxide production. Nature Medicine, 2011, 17, 1619-1626.	30.7	189
21	MARRVEL: Integration of Human and Model Organism Genetic Resources to Facilitate Functional Annotation of the Human Genome. American Journal of Human Genetics, 2017, 100, 843-853.	6.2	181
22	A natural history of cleidocranial dysplasia. American Journal of Medical Genetics Part A, 2001, 104, 1-6.	2.4	179
23	TGF-Î² Family Signaling in Mesenchymal Differentiation. Cold Spring Harbor Perspectives in Biology, 2018, 10, a022202.	5.5	175
24	Mutations in SERPINF1 cause osteogenesis imperfecta type VI. Journal of Bone and Mineral Research, 2011, 26, 2798-2803.	2.8	164
25	Notch signaling contributes to the pathogenesis of human osteosarcomas. Human Molecular Genetics, 2009, 18, 1464-1470.	2.9	157
26	Mutations in FKBP10 cause recessive osteogenesis imperfecta and bruck syndrome. Journal of Bone and Mineral Research, 2011, 26, 666-672.	2.8	149
27	Osteocyte-specific WNT1 regulates osteoblast function during bone homeostasis. Journal of Clinical Investigation, 2017, 127, 2678-2688.	8.2	143
28	The Undiagnosed Diseases Network: Accelerating Discovery about Health and Disease. American Journal of Human Genetics, 2017, 100, 185-192.	6.2	142
29	Evaluation of teriparatide treatment in adults with osteogenesis imperfecta. Journal of Clinical Investigation, 2014, 124, 491-498.	8.2	140
30	Therapeutic Antibody Targeting Tumor- and Osteoblastic Niche-Derived Jagged1 Sensitizes Bone Metastasis to Chemotherapy. Cancer Cell, 2017, 32, 731-747.e6.	16.8	133
31	Autosomal recessive Noonan syndrome associated with biallelic LZTR1 variants. Genetics in Medicine, 2018, 20, 1175-1185.	2.4	133
32	Heterozygous Truncating Variants in POMP Escape Nonsense-Mediated Decay and Cause a Unique Immune Dysregulatory Syndrome. American Journal of Human Genetics, 2018, 102, 1126-1142.	6.2	128
33	Identification of Functionally Distinct Mx1+Î±SMA+ Periosteal Skeletal Stem Cells. Cell Stem Cell, 2019, 25, 784-796.e5.	11.1	128
34	Yunis-VarÃ³n Syndrome Is Caused by Mutations in FIG4, Encoding a Phosphoinositide Phosphatase. American Journal of Human Genetics, 2013, 92, 781-791.	6.2	124
35	Notch Activation as a Driver of Osteogenic Sarcoma. Cancer Cell, 2014, 26, 390-401.	16.8	115
36	Mutations in KAT6B, Encoding a Histone Acetyltransferase, Cause Genitopatellar Syndrome. American Journal of Human Genetics, 2012, 90, 282-289.	6.2	112

#	Article	IF	Citations
37	The genetic basis of DOORS syndrome: an exome-sequencing study. Lancet Neurology, The, 2014, 13, 44-58.	10.2	108
38	A Syndromic Neurodevelopmental Disorder Caused by De Novo Variants in EBF3. American Journal of Human Genetics, 2017, 100, 128-137.	6.2	96
39	Clinical Application of Genome and Exome Sequencing as a Diagnostic Tool for Pediatric Patients: a Scoping Review of the Literature. Genetics in Medicine, 2019, 21, 3-16.	2.4	96
40	Effect of alternative pathway therapy on branched chain amino acid metabolism in urea cycle disorder patients. Molecular Genetics and Metabolism, 2004, 81, 79-85.	1.1	93
41	MicroRNA miR-23a cluster promotes osteocyte differentiation by regulating TGF-Î² signalling in osteoblasts. Nature Communications, 2017, 8, 15000.	12.8	91
42	Uncoupling of chondrocyte differentiation and perichondrial mineralization underlies the skeletal dysplasia in tricho-rhino-phalangeal syndrome. Human Molecular Genetics, 2008, 17, 2244-2254.	2.9	90
43	A recurrent p.Arg92Trp variant in steroidogenic factor-1 (NR5A1) can act as a molecular switch in human sex development. Human Molecular Genetics, 2016, 25, 3446-3453.	2.9	90
44	Transcriptome-directed analysis for Mendelian disease diagnosis overcomes limitations of conventional genomic testing. Journal of Clinical Investigation, 2021, 131, .	8.2	87
45	Genotypeâ€“Phenotype Correlation â€” Promiscuity in the Era of Next-Generation Sequencing. New England Journal of Medicine, 2014, 371, 593-596.	27.0	86
46	Osteogenesis imperfecta: advancements in genetics and treatment. Current Opinion in Pediatrics, 2019, 31, 708-715.	2.0	84
47	Ammonia control and neurocognitive outcome among urea cycle disorder patients treated with glycerol phenylbutyrate. Hepatology, 2013, 57, 2171-2179.	7.3	83
48	Genetic causes and mechanisms of Osteogenesis Imperfecta. Bone, 2017, 102, 40-49.	2.9	82
49	Whole-exome sequencing identifies mutations in the nucleoside transporter gene SLC29A3 in dysosteosclerosis, a form of osteopetrosis. Human Molecular Genetics, 2012, 21, 4904-4909.	2.9	81
50	Clinically severe CACNA1A alleles affect synaptic function and neurodegeneration differentially. PLoS Genetics, 2017, 13, e1006905.	3.5	80
51	Phase 2 comparison of a novel ammonia scavenging agent with sodium phenylbutyrate in patients with urea cycle disorders: Safety, pharmacokinetics and ammonia control. Molecular Genetics and Metabolism, 2010, 100, 221-228.	1.1	78
52	Argininosuccinate lyase deficiencyâ€”Argininosuccinic aciduria and beyond. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2011, 157, 45-53.	1.6	78
53	Phenylbutyrate therapy for maple syrup urine disease. Human Molecular Genetics, 2011, 20, 631-640.	2.9	77
54	Clinical Consequences of Urea Cycle Enzyme Deficiencies and Potential Links to Arginine and Nitric Oxide Metabolism. Journal of Nutrition, 2004, 134, 2775S-2782S.	2.9	76

#	Article	IF	Citations
55	Establishing a consortium for the study of rare diseases: The Urea Cycle Disorders Consortium. Molecular Genetics and Metabolism, 2010, 100, S97-S105.	1.1	73
56	Nitric-Oxide Supplementation for Treatment of Long-Term Complications in Argininosuccinic Aciduria. American Journal of Human Genetics, 2012, 90, 836-846.	6.2	73
57	NOTCHing the bone: Insights into multi-functionality. Bone, 2010, 46, 274-280.	2.9	71
58	Loss- or Gain-of-Function Mutations in ACOX1 Cause Axonal Loss via Different Mechanisms. Neuron, 2020, 106, 589-606.e6.	8.1	71
59	Sclerostin Antibody Treatment Improves the Bone Phenotype of Crtapâ€"/â€" Mice, a Model of Recessive Osteogenesis Imperfecta. Journal of Bone and Mineral Research, 2016, 31, 1030-1040.	2.8	70
60	IRF2BPL Is Associated with Neurological Phenotypes. American Journal of Human Genetics, 2018, 103, 245-260.	6.2	69
61	Phenotypic Variability of Osteogenesis Imperfecta Type V Caused by an IFITM 5 Mutation. Journal of Bone and Mineral Research, 2013, 28, 1523-1530.	2.8	67
62	The swaying mouse as a model of osteogenesis imperfecta caused by WNT1 mutations. Human Molecular Genetics, 2014, 23, 4035-4042.	2.9	66
63	Osteosclerosis owing to Notch gain of function is solely Rbpj-dependent. Journal of Bone and Mineral Research, 2010, 25, 2175-2183.	2.8	65
64	ADAMTS-7 forms a positive feedback loop with TNF-Î± in the pathogenesis of osteoarthritis. Annals of the Rheumatic Diseases, 2014, 73, 1575-1584.	0.9	64
65	Long-term correction of urea cycle disorders. Journal of Pediatrics, 2001, 138, S62-S71.	1.8	61
66	Brachyâ€"syndactyly caused by loss of Sfrp2 function. Journal of Cellular Physiology, 2008, 217, 127-137.	4.1	61
67	Notch gain of function inhibits chondrocyte differentiation via Rbpj-dependent suppression of Sox9. Journal of Bone and Mineral Research, 2013, 28, 649-659.	2.8	61
68	Alteration of Notch signaling in skeletal development and disease. Annals of the New York Academy of Sciences, 2010, 1192, 257-268.	3.8	59
69	Expanding the Spectrum of BAF-Related Disorders: De Novo Variants in SMARCC2 Cause a Syndrome with Intellectual Disability and Developmental Delay. American Journal of Human Genetics, 2019, 104, 164-178.	6.2	59
70	Structure-based design and mechanisms of allosteric inhibitors for mitochondrial branched-chain Î±-ketoacid dehydrogenase kinase. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, 9728-9733.	7.1	58
71	Sodium phenylbutyrate decreases plasma branched-chain amino acids in patients with urea cycle disorders. Molecular Genetics and Metabolism, 2014, 113, 131-135.	1.1	58
72	Gene therapy for repair and regeneration of bone and cartilage. Current Opinion in Pharmacology, 2018, 40, 59-66.	3.5	56

#	Article	IF	Citations
73	De Novo Missense Variants in TRAF7 Cause Developmental Delay, Congenital Anomalies, and Dysmorphic Features. American Journal of Human Genetics, 2018, 103, 154-162.	6.2	56
74	Signaling Pathways in Human Skeletal Dysplasias. Annual Review of Genomics and Human Genetics, 2010, 11, 189-217.	6.2	55
75	Autosomal-Dominant Multiple Pterygium Syndrome Is Caused by Mutations in MYH3. American Journal of Human Genetics, 2015, 96, 841-849.	6.2	55
76	Connective tissue alterations in Fkbp10â^'/â^' mice. Human Molecular Genetics, 2014, 23, 4822-4831.	2.9	54
77	Pharmacological and biological therapeutic strategies for osteogenesis imperfecta. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2016, 172, 367-383.	1.6	53
78	Generalized Connective Tissue Disease in Crtap-/- Mouse. PLoS ONE, 2010, 5, e10560.	2.5	52
79	A Chaperone Complex Formed by HSP47, FKBP65, and BiP Modulates Telopeptide Lysyl Hydroxylation of Type I Procollagen. Journal of Bone and Mineral Research, 2017, 32, 1309-1319.	2.8	50
80	Untargeted metabolomic profiling reveals multiple pathway perturbations and new clinical biomarkers in urea cycle disorders. Genetics in Medicine, 2019, 21, 1977-1986.	2.4	47
81	Next-generation sequencing for disorders of low and high bone mineral density. Osteoporosis International, 2013, 24, 2253-2259.	3.1	46
82	Post-translationally Abnormal Collagens of Prolyl 3-Hydroxylase-2 Null Mice Offer a Pathobiological Mechanism for the High Myopia Linked to Human LEPREL1 Mutations. Journal of Biological Chemistry, 2015, 290, 8613-8622.	3.4	44
83	Notch Signaling in Skeletal Development, Homeostasis and Pathogenesis. Biomolecules, 2020, 10, 332.	4.0	43
84	Notch Signaling in Skeletal Stem Cells. Calcified Tissue International, 2014, 94, 68-77.	3.1	42
85	Mutations in ANAPC1, Encoding a Scaffold Subunit of the Anaphase-Promoting Complex, Cause Rothmund-Thomson Syndrome Type 1. American Journal of Human Genetics, 2019, 105, 625-630.	6.2	42
86	Impact of Diagnosis and Therapy on Cognitive Function in Urea Cycle Disorders. Annals of Neurology, 2019, 86, 116-128.	5.3	42
87	Systemic hypertension in two patients with ASL deficiency: A result of nitric oxide deficiency?. Molecular Genetics and Metabolism, 2009, 98, 195-197.	1.1	41
88	WDR35 mutation in siblings with Sensenbrenner syndrome: A ciliopathy with variable phenotype. American Journal of Medical Genetics, Part A, 2012, 158A, 2917-2924.	1.2	40
89	Human recombinant arginase enzyme reduces plasma arginine in mouse models of arginase deficiency. Human Molecular Genetics, 2015, 24, 6417-6427.	2.9	40
90	Phenotypic and molecular characterisation of CDK13-related congenital heart defects, dysmorphic facial features and intellectual developmental disorders. Genome Medicine, 2017, 9, 73.	8.2	39

#	Article	IF	Citations
91	Argininosuccinate Lyase Deficiency Causes an Endothelial-Dependent Form of Hypertension. American Journal of Human Genetics, 2018, 103, 276-287.	6.2	39
92	Nitric oxide modulates bone anabolism through regulation of osteoblast glycolysis and differentiation. Journal of Clinical Investigation, 2021, 131, .	8.2	39
93	Glycerol phenylbutyrate treatment in children with urea cycle disorders: Pooled analysis of short and long-term ammonia control and outcomes. Molecular Genetics and Metabolism, 2014, 112, 17-24.	1.1	38
94	Mutations in Fibronectin Cause a Subtype of Spondylometaphyseal Dysplasia with â€œCorner Fracturesâ€• American Journal of Human Genetics, 2017, 101, 815-823.	6.2	37
95	De novo EIF2AK1 and EIF2AK2 Variants Are Associated with Developmental Delay, Leukoencephalopathy, and Neurologic Decompensation. American Journal of Human Genetics, 2020, 106, 570-583.	6.2	37
96	Large-Scale Production of High-Quality Helper-Dependent Adenoviral Vectors Using Adherent Cells in Cell Factories. Human Gene Therapy, 2010, 21, 120-126.	2.7	35
97	A Recurrent De Novo Variant in NACC1 Causes a Syndrome Characterized by Infantile Epilepsy, Cataracts, and Profound Developmental Delay. American Journal of Human Genetics, 2017, 100, 343-351.	6.2	35
98	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
99	A multicenter study to evaluate pulmonary function in osteogenesis imperfecta. Clinical Genetics, 2018, 94, 502-511.	2.0	33
100	Effects of Aspirin on Growth Factor Release From Freshly Isolated Leukocyte-Rich Platelet-Rich Plasma in Healthy Men: A Prospective Fixed-Sequence Controlled Laboratory Study. American Journal of Sports Medicine, 2019, 47, 1223-1229.	4.2	33
101	A randomized controlled trial to evaluate the effects of high-dose versus low-dose of arginine therapy on hepatic function tests in argininosuccinic aciduria. Molecular Genetics and Metabolism, 2012, 107, 315-321.	1.1	32
102	E-selectin ligand 1 regulates bone remodeling by limiting bioactive TGF-Î² in the bone microenvironment. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, 7336-7341.	7.1	32
103	P3h3-null and Sc65-null Mice Phenocopy the Collagen Lysine Under-hydroxylation and Cross-linking Abnormality of Ehlers-Danlos Syndrome Type VIA. Journal of Biological Chemistry, 2017, 292, 3877-3887.	3.4	32
104	Differential Effects of Collagen Prolyl 3-Hydroxylation on Skeletal Tissues. PLoS Genetics, 2014, 10, e1004121.	3.5	31
105	A Transgenic Mouse Model of OI Type V Supports a Neomorphic Mechanism of the IFITM5 Mutation. Journal of Bone and Mineral Research, 2015, 30, 489-498.	2.8	30
106	RECQL4 Regulates p53 Function In Vivo During Skeletogenesis. Journal of Bone and Mineral Research, 2015, 30, 1077-1089.	2.8	30
107	Blood ammonia and glutamine as predictors of hyperammonemic crises in patients with urea cycle disorder. Genetics in Medicine, 2015, 17, 561-568.	2.4	30
108	Further evidence for the involvement of EFL1 in a Shwachmanâ€“Diamond-like syndrome and expansion of the phenotypic features. Journal of Physical Education and Sports Management, 2018, 4, a003046.	1.2	29

#	Article	IF	Citations
109	Cloning, characterization, and chromosomal assignment of the human ortholog of murine Zfp-37, a candidate gene for Nager syndrome. Mammalian Genome, 1998, 9, 458-462.	2.2	28
110	Sclt1 deficiency causes cystic kidney by activating ERK and STAT3 signaling. Human Molecular Genetics, 2017, 26, 2949-2960.	2.9	28
111	Bmi1 Suppresses Adipogenesis in the Hematopoietic Stem Cell Niche. Stem Cell Reports, 2019, 13, 545-558.	4.8	28
112	The long and the short of it: developmental genetics of the skeletal dysplasias. Clinical Genetics, 1998, 54, 464-473.	2.0	27
113	Dual Role of the Trps1 Transcription Factor in Dentin Mineralization. Journal of Biological Chemistry, 2014, 289, 27481-27493.	3.4	27
114	Barriers to drug adherence in the treatment of urea cycle disorders: Assessment of patient, caregiver and provider perspectives. Molecular Genetics and Metabolism Reports, 2016, 8, 43-47.	1.1	27
115	Heterozygous variants inACTL6A, encoding a component of the BAF complex, are associated with intellectual disability. Human Mutation, 2017, 38, 1365-1371.	2.5	27
116	Bi-allelic Variants in TONSL Cause SPONASTRIME Dysplasia and a Spectrum of Skeletal Dysplasia Phenotypes. American Journal of Human Genetics, 2019, 104, 422-438.	6.2	27
117	Assessment of bone mineral status in children with Marfan syndrome. American Journal of Medical Genetics, Part A, 2012, 158A, 2221-2224.	1.2	26
118	Osteogenesis imperfecta without features of type V caused by a mutation in the IFITM 5 gene. Journal of Bone and Mineral Research, 2013, 28, 2333-2337.	2.8	26
119	Targeting TGF-Î² for treatment of osteogenesis imperfecta. Journal of Clinical Investigation, 2022, 132, .	8.2	26
120	Further delineation of the clinical spectrum of KAT6B disorders and allelic series of pathogenic variants. Genetics in Medicine, 2020, 22, 1338-1347.	2.4	25
121	Correlations Between Bone Mechanical Properties and Bone Composition Parameters in Mouse Models of Dominant and Recessive Osteogenesis Imperfecta and the Response to Anti-TGF-Î² Treatment. Journal of Bone and Mineral Research, 2017, 32, 347-359.	2.8	24
122	Sites of Cre-recombinase activity in mouse lines targeting skeletal cells. Journal of Bone and Mineral Research, 2020, 36, 1661-1679.	2.8	24
123	E-selectin ligandâ€“1 regulates growth plate homeostasis in mice by inhibiting the intracellular processing and secretion of mature TGF-Î². Journal of Clinical Investigation, 2010, 120, 2474-2485.	8.2	24
124	Transcriptional dysregulation in skeletal malformation syndromes. American Journal of Medical Genetics Part A, 2001, 106, 258-271.	2.4	23
125	Arginase overexpression in neurons and its effect on traumatic brain injury. Molecular Genetics and Metabolism, 2018, 125, 112-117.	1.1	22
126	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

#	Article	IF	Citations
127	The crucial p53-dependent oncogenic role of JAB1 in osteosarcoma in vivo. Oncogene, 2020, 39, 4581-4591.	5.9	22
128	Exome sequencing compared with standard genetic tests for critically ill infants with suspected genetic conditions. Genetics in Medicine, 2020, 22, 1303-1310.	2.4	21
129	Trisomy 16q in a female newborn with a de novo X;16 translocation and hypoplastic left heart. , 1999, 82, 128-131.		20
130	Lysinuric protein intolerance presenting with multiple fractures. Molecular Genetics and Metabolism Reports, 2014, 1, 176-183.	1.1	20
131	Milder clinical and biochemical phenotypes associated with the c.482G > A (p.Arg161Gln) pathogenic variant in cobalamin C disease: Implications for management and screening. Molecular Genetics and Metabolism, 2017, 122, 60-66.	1.1	20
132	Combinatorial treatment with oncolytic adenovirus and helper-dependent adenovirus augments adenoviral cancer gene therapy. Molecular Therapy - Oncolytics, 2014, 1, 14008.	4.4	19
133	Losartan increases bone mass and accelerates chondrocyte hypertrophy in developing skeleton. Molecular Genetics and Metabolism, 2015, 115, 53-60.	1.1	19
134	mTORC1 Signaling is a Critical Regulator of Postnatal Tendon Development. Scientific Reports, 2017, 7, 17175.	3.3	19
135	Oral health-related quality of life in children and adolescents with osteogenesis imperfecta: cross-sectional study. Orphanet Journal of Rare Diseases, 2018, 13, 187.	2.7	19
136	Mice Expressing Mutant Trpv4 Recapitulate the Human TRPV4 Disorders. Journal of Bone and Mineral Research, 2014, 29, 1815-1822.	2.8	18
137	An exome sequencing study of Moebius syndrome including atypical cases reveals an individual with CFEOM3A and a TUBB3 mutation. Journal of Physical Education and Sports Management, 2017, 3, a000984.	1.2	18
138	Pathogenic Variants in Fucokinase Cause a Congenital Disorder of Glycosylation. American Journal of Human Genetics, 2018, 103, 1030-1037.	6.2	18
139	Mechanical properties of infant bone. Bone, 2018, 113, 151-160.	2.9	18
140	COPB2 loss of function causes a coatopathy with osteoporosis and developmental delay. American Journal of Human Genetics, 2021, 108, 1710-1724.	6.2	18
141	Neonatal fractures as a presenting feature of LMOD3â€associated congenital myopathy. American Journal of Medical Genetics, Part A, 2017, 173, 2789-2794.	1.2	17
142	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
143	Leukocyte-dependent effects of platelet-rich plasma on cartilage loss and thermal hyperalgesia in a mouse model of post-traumatic osteoarthritis. Osteoarthritis and Cartilage, 2020, 28, 1385-1393.	1.3	17
144	Diagnosis of ALG12-CDG by exome sequencing in a case of severe skeletal dysplasia. Molecular Genetics and Metabolism Reports, 2014, 1, 213-219.	1.1	16

#	Article	IF	Citations
145	Glutamine and hyperammonemic crises in patients with urea cycle disorders. Molecular Genetics and Metabolism, 2016, 117, 27-32.	1.1	16
146	Fkbp10 Deletion in Osteoblasts Leads to Qualitative Defects in Bone. Journal of Bone and Mineral Research, 2017, 32, 1354-1367.	2.8	16
147	Optimizing therapy for argininosuccinic aciduria. Molecular Genetics and Metabolism, 2012, 107, 10-14.	1.1	15
148	Mobility in osteogenesis imperfecta: a multicenter North American study. Genetics in Medicine, 2019, 21, 2311-2318.	2.4	15
149	ATRAID regulates the action of nitrogen-containing bisphosphonates on bone. Science Translational Medicine, 2020, 12, .	12.4	15
150	A global Slc7a7 knockout mouse model demonstrates characteristic phenotypes of human lysinuric protein intolerance. Human Molecular Genetics, 2020, 29, 2171-2184.	2.9	15
151	One NOTCH Further: Jagged 1 in Bone Metastasis. Cancer Cell, 2011, 19, 159-161.	16.8	14
152	Protein and calorie intakes in adult and pediatric subjects with urea cycle disorders participating in clinical trials of glycerol phenylbutyrate. Molecular Genetics and Metabolism Reports, 2016, 6, 34-40.	1.1	14
153	Kaufman oculo-cerebro-facial syndrome in a child with small and absent terminal phalanges and absent nails. Journal of Human Genetics, 2017, 62, 465-471.	2.3	14
154	Pediatric Outcomes Data Collection Instrument is a Useful Patient-Reported Outcome Measure for Physical Function in Children with Osteogenesis Imperfecta. Genetics in Medicine, 2020, 22, 581-589.	2.4	14
155	4-PBA Treatment Improves Bone Phenotypes in the Aga2 Mouse Model of Osteogenesis Imperfecta. Journal of Bone and Mineral Research, 2020, 37, 675-686.	2.8	14
156	Considerations in the Difficult-to-Manage Urea Cycle Disorder Patient. Critical Care Clinics, 2005, 21, S19-S25.	2.6	13
157	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
158	Generalized metabolic bone disease and fracture risk in Rothmund-Thomson syndrome. Human Molecular Genetics, 2017, 26, 3046-3055.	2.9	13
159	A non-mosaic PORCN mutation in a male with severe congenital anomalies overlapping focal dermal hypoplasia. Molecular Genetics and Metabolism Reports, 2017, 12, 57-61.	1.1	13
160	Long-term use of angiotensin-converting enzyme inhibitors protects against bone loss in African-American elderly men. Archives of Osteoporosis, 2017, 12, 94.	2.4	13
161	Fracture Healing in Collagenâ€Related Preclinical Models of Osteogenesis Imperfecta. Journal of Bone and Mineral Research, 2020, 35, 1132-1148.	2.8	13
162	Self-reported treatment-associated symptoms among patients with urea cycle disorders participating in glycerol phenylbutyrate clinical trials. Molecular Genetics and Metabolism, 2015, 116, 29-34.	1.1	12

#	Article	IF	Citations
163	Dissatisfaction with maintenance of certification in academic pediatrics. Pediatric Research, 2016, 79, 240-242.	2.3	12
164	Restoration of the serum level of SERPINF1 does not correct the bone phenotype in Serpinf1 null mice. Molecular Genetics and Metabolism, 2016, 117, 378-382.	1.1	12
165	Serum Sclerostin Levels in Adults With Osteogenesis Imperfecta: Comparison With Normal Individuals and Response to Teriparatide Therapy. Journal of Bone and Mineral Research, 2018, 33, 307-315.	2.8	12
166	Heterozygous WNT1 variant causing a variable bone phenotype. American Journal of Medical Genetics, Part A, 2018, 176, 2419-2424.	1.2	11
167	Caries prevalence and experience in individuals with osteogenesis imperfecta: A crossâ€sectional multicenter study. Special Care in Dentistry, 2019, 39, 214-219.	0.8	11
168	Recurrent mosaic MTOR c.5930C > T (p.Thr1977Ile) variant causing megalencephaly, asymmetric polymicrogyria, and cutaneous pigmentary mosaicism: Case report and review of the literature. American Journal of Medical Genetics, Part A, 2019, 179, 475-479.	1.2	11
169	Oro-dental and cranio-facial characteristics of osteogenesis imperfecta type V. European Journal of Medical Genetics, 2019, 62, 103606.	1.3	11
170	Alterations in non-type I collagen biomarkers in osteogenesis imperfecta. Bone, 2019, 120, 70-74.	2.9	11
171	Using the Delphi method to identify cliniciansâ€™ perceived importance of pediatric exome sequencing results. Genetics in Medicine, 2020, 22, 69-76.	2.4	11
172	Tendon and motor phenotypes in the Crtap-/- mouse model of recessive osteogenesis imperfecta. ELife, 2021, 10, .	6.0	11
173	Pregnancy in women with osteogenesis imperfecta: pregnancy characteristics, maternal, and neonatal outcomes. American Journal of Obstetrics & Gynecology MFM, 2021, 3, 100362.	2.6	11
174	What Has the Undiagnosed Diseases Network Taught Us About the Clinical Applications of Genomic Testing?. Annual Review of Medicine, 2022, 73, 575-585.	12.2	11
175	Phenylbutyrate improves nitrogen disposal via an alternative pathway without eliciting an increase in protein breakdown and catabolism in control and ornithine transcarbamylaseâ€“deficient patients. American Journal of Clinical Nutrition, 2011, 93, 1248-1254.	4.7	10
176	Chronic liver disease and impaired hepatic glycogen metabolism in argininosuccinate lyase deficiency. JCI Insight, 2020, 5, .	5.0	10
177	Early childhood presentation of Czech dysplasia. Clinical Dysmorphology, 2013, 22, 76-80.	0.3	9
178	Heterozygous variants in SPTBN1 cause intellectual disability and autism. American Journal of Medical Genetics, Part A, 2021, 185, 2037-2045.	1.2	9
179	Corner fracture type spondylometaphyseal dysplasia: Overlap with type II collagenopathies. American Journal of Medical Genetics, Part A, 2017, 173, 733-739.	1.2	8
180	Microdeletions excluding YWHAE and PAFAH1B1 cause a unique leukoencephalopathy: further delineation of the 17p13.3 microdeletion spectrum. Genetics in Medicine, 2019, 21, 1652-1656.	2.4	8

#	Article	IF	Citations
181	Assessment of longitudinal bone growth in osteogenesis imperfecta using metacarpophalangeal pattern profiles. Bone, 2020, 140, 115547.	2.9	8
182	Novel assessment of leukocyte-rich platelet-rich plasma on functional and patient-reported outcomes in knee osteoarthritis: a pilot study. Regenerative Medicine, 2021, 16, 823-832.	1.7	8
183	<scp>miRNAâ€34c</scp> Suppresses Osteosarcoma Progression In Vivo by Targeting Notch and <scp>E2F</scp>. JBMR Plus, 2022, 6, e10623.	2.7	8
184	Catelâ€“Manzke syndrome: Further delineation of the phenotype associated with pathogenic variants in TGDS. Molecular Genetics and Metabolism Reports, 2015, 4, 89-91.	1.1	7
185	Atypical Alexander disease with dystonia, retinopathy, and a brain mass mimicking astrocytoma. Neurology: Genetics, 2018, 4, e248.	1.9	7
186	Dental and craniofacial characteristics caused by the p.Ser40Leu mutation in IFITM5. American Journal of Medical Genetics, Part A, 2019, 179, 65-70.	1.2	7
187	Double-Spin Leukocyte-Rich Platelet-Rich Plasma Is Predominantly Lymphocyte Rich With Notable Concentrations of Other White Blood Cell Subtypes. Arthroscopy, Sports Medicine, and Rehabilitation, 2021, 4, e335-e341.	1.7	7
188	Adult presentation of Xâ€linked Conradiâ€HÃ¼nermannâ€Happle syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 1309-1314.	1.2	6
189	Wholeâ€Exome Sequencing Identifies an Intronic Cryptic Splice Site in SERPINF1 Causing Osteogenesis Imperfecta Type VI. JBMR Plus, 2018, 2, 235-239.	2.7	6
190	The microRNA processorDROSHAis a candidate gene for a severe progressive neurological disorder. Human Molecular Genetics, 2022, 31, 2934-2950.	2.9	6
191	Trps1 Regulates Development of Craniofacial Skeleton and Is Required for the Initiation of Palatal Shelves Fusion. Frontiers in Physiology, 2019, 10, 513.	2.8	5
192	A Novel Mouse Model for SNP in Steroid Receptor Co-Activator-1 Reveals Role in Bone Density and Breast Cancer Metastasis. Endocrinology, 2021, 162, .	2.8	5
193	DDRGK1 is required for the proper development and maintenance of the growth plate cartilage. Human Molecular Genetics, 2022, 31, 2820-2830.	2.9	5
194	Widespread disturbance in extracellular matrix collagen biomarker responses to teriparatide therapy in osteogenesis imperfecta. Bone, 2021, 142, 115703.	2.9	4
195	The transcriptional cofactor Jab1/Cops5 is crucial for BMPâ€mediated mouse chondrocyte differentiation by repressing p53 activity. Journal of Cellular Physiology, 2021, 236, 5686-5697.	4.1	4
196	<scp>Healthâ€related</scp> quality of life in adults with osteogenesis imperfecta. Clinical Genetics, 2021, 99, 772-779.	2.0	4
197	Targeted and sustained Sox9 expression in mouse hypertrophic chondrocytes causes severe and spontaneous osteoarthritis by perturbing cartilage homeostasis. American Journal of Translational Research (discontinued), 2020, 12, 1056-1069.	0.0	4
198	Genome sequencing reveals novel noncoding variants in <scp>PLA2G6</scp> and <scp>LMNB1</scp> causing progressive neurologic disease. Molecular Genetics & Genomic Medicine, 2022, 10, e1892.	1.2	4

#	Article	IF	Citations
199	A novel, de novo intronic variant in <scp>POGZ</scp> causes <scp>Whiteâ€“Sutton</scp> syndrome. American Journal of Medical Genetics, Part A, 2022, 188, 2198-2203.	1.2	4
200	Reply to Ligthart-Melis et al.. American Journal of Physiology - Endocrinology and Metabolism, 2010, 299, E684-E684.	3.5	3
201	Localized chondro-ossification underlies joint dysfunction and motor deficits in the Fkbp10 mouse model of osteogenesis imperfecta. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, .	7.1	3
202	A novel de novo intronic variant in ITPR1 causes Gillespie syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 2315-2324.	1.2	2
203	PRUNE1 c.<scp>933G</scp>>A synonymous variant induces exon 7 skipping, disrupts the <scp>DHHA2</scp> domain, and leads to an atypical <scp>NMIHBA</scp> syndrome presentation: Case report and review of the literature. American Journal of Medical Genetics, Part A, 2022, 188, 1868-1874.	1.2	2
204	SERPINF1 as a Cause of Osteogenesis Imperfecta Type VI. , 2014, , 167-172.		1
205	Prospects of Gene Therapy for Skeletal Diseases. , 2018, , 119-137.		1
206	Genetic Burden Contributing to Extremely Low or High Bone Mineral Density in a Senior Male Population From the Osteoporotic Fractures in Men Study (MrOS). JBMR Plus, 2020, 4, e10335.	2.7	1
207	Evaluation of extracorporeal cardiopulmonary resuscitation eligibility criteria for out-of-hospital cardiac arrest patients. BMC Research Notes, 2021, 14, 139.	1.4	1
208	Mesomelic Dysplasia With Specific Autopodal Synostoses: A Third Observation And Further Delineation Of The Multiple Congenital Anomaly Syndrome. Fetal and Pediatric Pathology, 2003, 22, 23-35.	0.3	1
209	Self-Complementary AAV Vectors Cause a Substantially Heightened TLR9-Dependent Innate Immune Response In the Liver. Blood, 2010, 116, 252-252.	1.4	1
210	Glutamine: precursor or nitrogen donor for the synthesis of citrulline?. FASEB Journal, 2010, 24, 740.14.	0.5	1
211	Molecular alterations due to Col5a1 haploinsufficiency in a mouse model of classic Ehlersâ€“Danlos syndrome. Human Molecular Genetics, 2022, 31, 1325-1335.	2.9	1
212	Yap and Taz promote osteogenesis in neural crest cells by preventing chondrogenesis. FASEB Journal, 2022, 36, .	0.5	1
213	The long and the short of it: developmental genetics of the skeletal dysplasias. Clinical Genetics, 1999, 57, 50-59.	2.0	0
214	Response to Srilatha et al.. Genetics in Medicine, 2012, 14, 627-628.	2.4	0
215	Recessive Osteogenesis Imperfecta Due to Mutations in CRTAP, LEPRE1 and PPIB. , 2014, , 141-150.		0
216	2016 Curt Stern Award Address: From Rare to Common Diseases: Translating Genetic Discovery to Therapy 1. American Journal of Human Genetics, 2017, 100, 397-400.	6.2	0

#	Article	IF	Citations
217	2017 Victor A. McKusick Leadership Award Introduction: Arthur L. Beaudet. American Journal of Human Genetics, 2018, 102, 359-360.	6.2	0
218	Yap and Taz function as the osteochondrogenic determinant in neural crest cells. FASEB Journal, 2021, 35, .	0.5	0
219	Vestigial ornithine transcarbamylase activity does not impair ureagenesis in Otc spfâ€ash mice. FASEB Journal, 2006, 20, A1093.	0.5	0
220	Ornithine restores ureagenesis capacity and mitigates hyperammonemia in Otc ^{spfâ€ash} mice. FASEB Journal, 2006, 20, A1092.	0.5	0
221	Phenylbutyrate reduces plasma leucine concentrations without affecting the flux of leucine. FASEB Journal, 2007, 21, A335.	0.5	0
222	Precursors for the synthesis of citrulline in mice fed arginine free diets. FASEB Journal, 2010, 24, 740.26.	0.5	0
223	Arginine utilization for citrulline synthesis in arginase II knockout mice. FASEB Journal, 2010, 24, .	0.5	0
224	Arginase II reduces arginine availability and nitric oxide production during endotoxemia. FASEB Journal, 2011, 25, 983.20.	0.5	0
225	SAT-LB088 Assessing Metacarpal Cortical Thickness as a Tool to Evaluate Bone Density Compared to DXA in Osteogenesis Imperfecta. Journal of the Endocrine Society, 2019, 3, .	0.2	0
226	Abstract 050: Loss of Argininosuccinate Lyase Leads to Nitric Oxide Deficiency, Endothelial Dysfunction, Impaired Angiogenesis, and Hypertension. Hypertension, 2015, 66, .	2.7	0