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	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
2	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
3	Targeting TGF-Î² for treatment of osteogenesis imperfecta. Journal of Clinical Investigation, 2022, 132, .	8.2	26
4	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
5	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
6	DDRGK1 is required for the proper development and maintenance of the growth plate cartilage. Human Molecular Genetics, 2022, 31, 2820-2830.	2.9	5
7	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
8	<scp>miRNAâ€34c</scp> Suppresses Osteosarcoma Progression In Vivo by Targeting Notch and <scp>E2F</scp>. JBMR Plus, 2022, 6, e10623.	2.7	8
9	The microRNA processorDROSHAis a candidate gene for a severe progressive neurological disorder. Human Molecular Genetics, 2022, 31, 2934-2950.	2.9	6
10	Yap and Taz promote osteogenesis in neural crest cells by preventing chondrogenesis. FASEB Journal, 2022, 36, .	0.5	1
11	Widespread disturbance in extracellular matrix collagen biomarker responses to teriparatide therapy in osteogenesis imperfecta. Bone, 2021, 142, 115703.	2.9	4
12	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
13	Transcriptome-directed analysis for Mendelian disease diagnosis overcomes limitations of conventional genomic testing. Journal of Clinical Investigation, 2021, 131, .	8.2	87
14	<scp>Healthâ€related</scp> quality of life in adults with osteogenesis imperfecta. Clinical Genetics, 2021, 99, 772-779.	2.0	4
15	Nitric oxide modulates bone anabolism through regulation of osteoblast glycolysis and differentiation. Journal of Clinical Investigation, 2021, 131, .	8.2	39
16	Evaluation of extracorporeal cardiopulmonary resuscitation eligibility criteria for out-of-hospital cardiac arrest patients. BMC Research Notes, 2021, 14, 139.	1.4	1
17	Heterozygous variants in SPTBN1 cause intellectual disability and autism. American Journal of Medical Genetics, Part A, 2021, 185, 2037-2045.	1.2	9
18	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

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19	Tendon and motor phenotypes in the Crtap-/- mouse model of recessive osteogenesis imperfecta. ELife, 2021, 10, .	6.0	11
20	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
21	Yap and Taz function as the osteochondrogenic determinant in neural crest cells. FASEB Journal, 2021, 35, .	0.5	0
22	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
23	Pregnancy in women with osteogenesis imperfecta: pregnancy characteristics, maternal, and neonatal outcomes. American Journal of Obstetrics & Gynecology MFM, 2021, 3, 100362.	2.6	11
24	COPB2 loss of function causes a coatopathy with osteoporosis and developmental delay. American Journal of Human Genetics, 2021, 108, 1710-1724.	6.2	18
25	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
26	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
27	Using the Delphi method to identify cliniciansâ€™ perceived importance of pediatric exome sequencing results. Genetics in Medicine, 2020, 22, 69-76.	2.4	11
28	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
29	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
30	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
31	Assessment of longitudinal bone growth in osteogenesis imperfecta using metacarpophalangeal pattern profiles. Bone, 2020, 140, 115547.	2.9	8
32	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
33	ATRAID regulates the action of nitrogen-containing bisphosphonates on bone. Science Translational Medicine, 2020, 12, .	12.4	15
34	The crucial p53-dependent oncogenic role of JAB1 in osteosarcoma in vivo. Oncogene, 2020, 39, 4581-4591.	5.9	22
35	A global Slc7a7 knockout mouse model demonstrates characteristic phenotypes of human lysinuric protein intolerance. Human Molecular Genetics, 2020, 29, 2171-2184.	2.9	15
36	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

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37	Loss- or Gain-of-Function Mutations in ACOX1 Cause Axonal Loss via Different Mechanisms. Neuron, 2020, 106, 589-606.e6.	8.1	71
38	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
39	Notch Signaling in Skeletal Development, Homeostasis and Pathogenesis. Biomolecules, 2020, 10, 332.	4.0	43
40	Fracture Healing in Collagenâ€Related Preclinical Models of Osteogenesis Imperfecta. Journal of Bone and Mineral Research, 2020, 35, 1132-1148.	2.8	13
41	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
42	Sites of Cre-recombinase activity in mouse lines targeting skeletal cells. Journal of Bone and Mineral Research, 2020, 36, 1661-1679.	2.8	24
43	Chronic liver disease and impaired hepatic glycogen metabolism in argininosuccinate lyase deficiency. JCI Insight, 2020, 5, .	5.0	10
44	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
45	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
46	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
47	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
48	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
49	Bmi1 Suppresses Adipogenesis in the Hematopoietic Stem Cell Niche. Stem Cell Reports, 2019, 13, 545-558.	4.8	28
50	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
51	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
52	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
53	Impact of Diagnosis and Therapy on Cognitive Function in Urea Cycle Disorders. Annals of Neurology, 2019, 86, 116-128.	5.3	42
54	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

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55	Mobility in osteogenesis imperfecta: a multicenter North American study. Genetics in Medicine, 2019, 21, 2311-2318.	2.4	15
56	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
57	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
58	Identification of Functionally Distinct Mx1+Î±SMA+ Periosteal Skeletal Stem Cells. Cell Stem Cell, 2019, 25, 784-796.e5.	11.1	128
59	Osteogenesis imperfecta: advancements in genetics and treatment. Current Opinion in Pediatrics, 2019, 31, 708-715.	2.0	84
60	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
61	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
62	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
63	Oro-dental and cranio-facial characteristics of osteogenesis imperfecta type V. European Journal of Medical Genetics, 2019, 62, 103606.	1.3	11
64	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
65	Alterations in non-type I collagen biomarkers in osteogenesis imperfecta. Bone, 2019, 120, 70-74.	2.9	11
66	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
67	Autosomal recessive Noonan syndrome associated with biallelic LZTR1 variants. Genetics in Medicine, 2018, 20, 1175-1185.	2.4	133
68	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
69	Gene therapy for repair and regeneration of bone and cartilage. Current Opinion in Pharmacology, 2018, 40, 59-66.	3.5	56
70	TGF-Î² Family Signaling in Mesenchymal Differentiation. Cold Spring Harbor Perspectives in Biology, 2018, 10, a022202.	5.5	175
71	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
72	Pathogenic Variants in Fucokinase Cause a Congenital Disorder of Glycosylation. American Journal of Human Genetics, 2018, 103, 1030-1037.	6.2	18

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73	Heterozygous WNT1 variant causing a variable bone phenotype. American Journal of Medical Genetics, Part A, 2018, 176, 2419-2424.	1.2	11
74	Effect of Genetic Diagnosis on Patients with Previously Undiagnosed Disease. New England Journal of Medicine, 2018, 379, 2131-2139.	27.0	261
75	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
76	A multicenter study to evaluate pulmonary function in osteogenesis imperfecta. Clinical Genetics, 2018, 94, 502-511.	2.0	33
77	Mechanical properties of infant bone. Bone, 2018, 113, 151-160.	2.9	18
78	2017 Victor A. McKusick Leadership Award Introduction: Arthur L. Beaudet. American Journal of Human Genetics, 2018, 102, 359-360.	6.2	0
79	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
80	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
81	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
82	Prospects of Gene Therapy for Skeletal Diseases. , 2018, , 119-137.		1
83	Arginase overexpression in neurons and its effect on traumatic brain injury. Molecular Genetics and Metabolism, 2018, 125, 112-117.	1.1	22
84	Argininosuccinate Lyase Deficiency Causes an Endothelial-Dependent Form of Hypertension. American Journal of Human Genetics, 2018, 103, 276-287.	6.2	39
85	Atypical Alexander disease with dystonia, retinopathy, and a brain mass mimicking astrocytoma. Neurology: Genetics, 2018, 4, e248.	1.9	7
86	IRF2BPL Is Associated with Neurological Phenotypes. American Journal of Human Genetics, 2018, 103, 245-260.	6.2	69
87	The Undiagnosed Diseases Network: Accelerating Discovery about Health and Disease. American Journal of Human Genetics, 2017, 100, 185-192.	6.2	142
88	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
89	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
90	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

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91	Fkbp10 Deletion in Osteoblasts Leads to Qualitative Defects in Bone. Journal of Bone and Mineral Research, 2017, 32, 1354-1367.	2.8	16
92	Genetic causes and mechanisms of Osteogenesis Imperfecta. Bone, 2017, 102, 40-49.	2.9	82
93	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
94	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
95	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
96	MicroRNA miR-23a cluster promotes osteocyte differentiation by regulating TGF-Î² signalling in osteoblasts. Nature Communications, 2017, 8, 15000.	12.8	91
97	Generalized metabolic bone disease and fracture risk in Rothmund-Thomson syndrome. Human Molecular Genetics, 2017, 26, 3046-3055.	2.9	13
98	Sclt1 deficiency causes cystic kidney by activating ERK and STAT3 signaling. Human Molecular Genetics, 2017, 26, 2949-2960.	2.9	28
99	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
100	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
101	A Syndromic Neurodevelopmental Disorder Caused by De Novo Variants in EBF3. American Journal of Human Genetics, 2017, 100, 128-137.	6.2	96
102	Use of Exome Sequencing for Infants in Intensive Care Units. JAMA Pediatrics, 2017, 171, e173438.	6.2	348
103	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
104	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
105	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
106	Therapeutic Antibody Targeting Tumor- and Osteoblastic Niche-Derived Jagged1 Sensitizes Bone Metastasis to Chemotherapy. Cancer Cell, 2017, 32, 731-747.e6.	16.8	133
107	mTORC1 Signaling is a Critical Regulator of Postnatal Tendon Development. Scientific Reports, 2017, 7, 17175.	3.3	19
108	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

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109	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
110	Heterozygous variants inACTL6A, encoding a component of the BAF complex, are associated with intellectual disability. Human Mutation, 2017, 38, 1365-1371.	2.5	27
111	Corner fracture type spondylometaphyseal dysplasia: Overlap with type II collagenopathies. American Journal of Medical Genetics, Part A, 2017, 173, 733-739.	1.2	8
112	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
113	Osteocyte-specific WNT1 regulates osteoblast function during bone homeostasis. Journal of Clinical Investigation, 2017, 127, 2678-2688.	8.2	143
114	Clinically severe CACNA1A alleles affect synaptic function and neurodegeneration differentially. PLoS Genetics, 2017, 13, e1006905.	3.5	80
115	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
116	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
117	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
118	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
119	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
120	Dissatisfaction with maintenance of certification in academic pediatrics. Pediatric Research, 2016, 79, 240-242.	2.3	12
121	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
122	Glutamine and hyperammonemic crises in patients with urea cycle disorders. Molecular Genetics and Metabolism, 2016, 117, 27-32.	1.1	16
123	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
124	Adult presentation of Xâ€linked Conradiâ€HÃ¼nermannâ€Happle syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 1309-1314.	1.2	6
125	The Osteogenic Niche Promotes Early-Stage Bone Colonization of Disseminated Breast Cancer Cells. Cancer Cell, 2015, 27, 193-210.	16.8	308
126	Losartan increases bone mass and accelerates chondrocyte hypertrophy in developing skeleton. Molecular Genetics and Metabolism, 2015, 115, 53-60.	1.1	19

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127	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
128	Autosomal-Dominant Multiple Pterygium Syndrome Is Caused by Mutations in MYH3. American Journal of Human Genetics, 2015, 96, 841-849.	6.2	55
129	RECQL4 Regulates p53 Function In Vivo During Skeletogenesis. Journal of Bone and Mineral Research, 2015, 30, 1077-1089.	2.8	30
130	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
131	Human recombinant arginase enzyme reduces plasma arginine in mouse models of arginase deficiency. Human Molecular Genetics, 2015, 24, 6417-6427.	2.9	40
132	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
133	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
134	Abstract 050: Loss of Argininosuccinate Lyase Leads to Nitric Oxide Deficiency, Endothelial Dysfunction, Impaired Angiogenesis, and Hypertension. Hypertension, 2015, 66, .	2.7	0
135	SERPINF1 as a Cause of Osteogenesis Imperfecta Type VI. , 2014, , 167-172.		1
136	The swaying mouse as a model of osteogenesis imperfecta caused by WNT1 mutations. Human Molecular Genetics, 2014, 23, 4035-4042.	2.9	66
137	Dual Role of the Trps1 Transcription Factor in Dentin Mineralization. Journal of Biological Chemistry, 2014, 289, 27481-27493.	3.4	27
138	Differential Effects of Collagen Prolyl 3-Hydroxylation on Skeletal Tissues. PLoS Genetics, 2014, 10, e1004121.	3.5	31
139	Recessive Osteogenesis Imperfecta Due to Mutations in CRTAP, LEPRE1 and PPIB. , 2014, , 141-150.		0
140	The genetic basis of DOORS syndrome: an exome-sequencing study. Lancet Neurology, The, 2014, 13, 44-58.	10.2	108
141	Notch Signaling in Skeletal Stem Cells. Calcified Tissue International, 2014, 94, 68-77.	3.1	42
142	Branched-chain amino acid metabolism: from rare Mendelian diseases to more common disorders. Human Molecular Genetics, 2014, 23, R1-R8.	2.9	234
143	Connective tissue alterations in Fkbp10â^'/â^' mice. Human Molecular Genetics, 2014, 23, 4822-4831.	2.9	54
144	Excessive transforming growth factor-Î² signaling is a common mechanism in osteogenesis imperfecta. Nature Medicine, 2014, 20, 670-675.	30.7	237

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145	Notch Activation as a Driver of Osteogenic Sarcoma. Cancer Cell, 2014, 26, 390-401.	16.8	115
146	Genotypeâ€“Phenotype Correlation â€” Promiscuity in the Era of Next-Generation Sequencing. New England Journal of Medicine, 2014, 371, 593-596.	27.0	86
147	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
148	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
149	Mice Expressing Mutant Trpv4 Recapitulate the Human TRPV4 Disorders. Journal of Bone and Mineral Research, 2014, 29, 1815-1822.	2.8	18
150	Lysinuric protein intolerance presenting with multiple fractures. Molecular Genetics and Metabolism Reports, 2014, 1, 176-183.	1.1	20
151	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
152	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
153	Combinatorial treatment with oncolytic adenovirus and helper-dependent adenovirus augments adenoviral cancer gene therapy. Molecular Therapy - Oncolytics, 2014, 1, 14008.	4.4	19
154	Evaluation of teriparatide treatment in adults with osteogenesis imperfecta. Journal of Clinical Investigation, 2014, 124, 491-498.	8.2	140
155	Next-generation sequencing for disorders of low and high bone mineral density. Osteoporosis International, 2013, 24, 2253-2259.	3.1	46
156	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
157	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
158	WNT1 Mutations in Early-Onset Osteoporosis and Osteogenesis Imperfecta. New England Journal of Medicine, 2013, 368, 1809-1816.	27.0	308
159	Ammonia control and neurocognitive outcome among urea cycle disorder patients treated with glycerol phenylbutyrate. Hepatology, 2013, 57, 2171-2179.	7.3	83
160	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
161	Early childhood presentation of Czech dysplasia. Clinical Dysmorphology, 2013, 22, 76-80.	0.3	9
162	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

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163	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
164	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
165	Response to Srilatha et al.. Genetics in Medicine, 2012, 14, 627-628.	2.4	0
166	Optimizing therapy for argininosuccinic aciduria. Molecular Genetics and Metabolism, 2012, 107, 10-14.	1.1	15
167	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
168	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
169	Assessment of bone mineral status in children with Marfan syndrome. American Journal of Medical Genetics, Part A, 2012, 158A, 2221-2224.	1.2	26
170	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
171	miRNA-34c regulates Notch signaling during bone development. Human Molecular Genetics, 2012, 21, 2991-3000.	2.9	210
172	Mutations in KAT6B, Encoding a Histone Acetyltransferase, Cause Genitopatellar Syndrome. American Journal of Human Genetics, 2012, 90, 282-289.	6.2	112
173	Nitric-Oxide Supplementation for Treatment of Long-Term Complications in Argininosuccinic Aciduria. American Journal of Human Genetics, 2012, 90, 836-846.	6.2	73
174	One NOTCH Further: Jagged 1 in Bone Metastasis. Cancer Cell, 2011, 19, 159-161.	16.8	14
175	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
176	Mutations in FKBP10 cause recessive osteogenesis imperfecta and bruck syndrome. Journal of Bone and Mineral Research, 2011, 26, 666-672.	2.8	149
177	Mutations in SERPINF1 cause osteogenesis imperfecta type VI. Journal of Bone and Mineral Research, 2011, 26, 2798-2803.	2.8	164
178	Phenylbutyrate therapy for maple syrup urine disease. Human Molecular Genetics, 2011, 20, 631-640.	2.9	77
179	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
180	Requirement of argininosuccinate lyase for systemic nitric oxide production. Nature Medicine, 2011, 17, 1619-1626.	30.7	189

#	Article	IF	Citations
181	Arginase II reduces arginine availability and nitric oxide production during endotoxemia. FASEB Journal, 2011, 25, 983.20.	0.5	0
182	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
183	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
184	Osteosclerosis owing to Notch gain of function is solely Rbpj-dependent. Journal of Bone and Mineral Research, 2010, 25, 2175-2183.	2.8	65
185	Alteration of Notch signaling in skeletal development and disease. Annals of the New York Academy of Sciences, 2010, 1192, 257-268.	3.8	59
186	Reply to Ligthart-Melis et al.. American Journal of Physiology - Endocrinology and Metabolism, 2010, 299, E684-E684.	3.5	3
187	NOTCHing the bone: Insights into multi-functionality. Bone, 2010, 46, 274-280.	2.9	71
188	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
189	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
190	Signaling Pathways in Human Skeletal Dysplasias. Annual Review of Genomics and Human Genetics, 2010, 11, 189-217.	6.2	55
191	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
192	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
193	Self-Complementary AAV Vectors Cause a Substantially Heightened TLR9-Dependent Innate Immune Response In the Liver. Blood, 2010, 116, 252-252.	1.4	1
194	Generalized Connective Tissue Disease in Crtap-/- Mouse. PLoS ONE, 2010, 5, e10560.	2.5	52
195	Glutamine: precursor or nitrogen donor for the synthesis of citrulline?. FASEB Journal, 2010, 24, 740.14.	0.5	1
196	Precursors for the synthesis of citrulline in mice fed arginine free diets. FASEB Journal, 2010, 24, 740.26.	0.5	0
197	Arginine utilization for citrulline synthesis in arginase II knockout mice. FASEB Journal, 2010, 24, .	0.5	0
198	Notch signaling contributes to the pathogenesis of human osteosarcomas. Human Molecular Genetics, 2009, 18, 1464-1470.	2.9	157

#	Article	IF	Citations
199	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
200	CRTAPandLEPRE1mutations in recessive osteogenesis imperfecta. Human Mutation, 2008, 29, 1435-1442.	2.5	196
201	Brachyâ€“syndactyly caused by loss of Sfrp2 function. Journal of Cellular Physiology, 2008, 217, 127-137.	4.1	61
202	Dimorphic effects of Notch signaling in bone homeostasis. Nature Medicine, 2008, 14, 299-305.	30.7	361
203	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
204	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
205	Phenylbutyrate reduces plasma leucine concentrations without affecting the flux of leucine. FASEB Journal, 2007, 21, A335.	0.5	0
206	CRTAP Is Required for Prolyl 3- Hydroxylation and Mutations Cause Recessive Osteogenesis Imperfecta. Cell, 2006, 127, 291-304.	28.9	465
207	Deficiency of Cartilage-Associated Protein in Recessive Lethal Osteogenesis Imperfecta. New England Journal of Medicine, 2006, 355, 2757-2764.	27.0	307
208	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
209	Vestigial ornithine transcarbamylase activity does not impair ureagenesis in Otc spfâ€ash mice. FASEB Journal, 2006, 20, A1093.	0.5	0
210	Ornithine restores ureagenesis capacity and mitigates hyperammonemia in Otc ^{spfâ€ash} mice. FASEB Journal, 2006, 20, A1092.	0.5	0
211	Considerations in the Difficult-to-Manage Urea Cycle Disorder Patient. Critical Care Clinics, 2005, 21, S19-S25.	2.6	13
212	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
213	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
214	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
215	Long-term correction of urea cycle disorders. Journal of Pediatrics, 2001, 138, S62-S71.	1.8	61
216	A natural history of cleidocranial dysplasia. American Journal of Medical Genetics Part A, 2001, 104, 1-6.	2.4	179

#	Article	IF	Citations
217	Transcriptional dysregulation in skeletal malformation syndromes. American Journal of Medical Genetics Part A, 2001, 106, 258-271.	2.4	23
218	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
219	The long and the short of it: developmental genetics of the skeletal dysplasias. Clinical Genetics, 1999, 57, 50-59.	2.0	0
220	Trisomy 16q in a female newborn with a de novo X;16 translocation and hypoplastic left heart. , 1999, 82, 128-131.		20
221	Mutations in LMX1B cause abnormal skeletal patterning and renal dysplasia in nail patella syndrome. Nature Genetics, 1998, 19, 47-50.	21.4	471
222	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
223	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
224	The long and the short of it: developmental genetics of the skeletal dysplasias. Clinical Genetics, 1998, 54, 464-473.	2.0	27
225	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
226	Linkage of Marfan syndrome and a phenotypically related disorder to two different fibrillin genes. Nature, 1991, 352, 330-334.	27.8	676