Brendan H Lee

List of Publications by Year in Descending Order

219 papers	10,847 citations	54 h-index	99 g-index
265 ext. papers	12,867 ext. citations	8.7 avg, IF	5.75 L-index

#	Paper	IF	Citations
219	What Has the Undiagnosed Diseases Network Taught Us About the Clinical Applications of Genomic Testing?. *Annual Review of Medicine*, **2022**, 73, 575-585	17.4	2
218	PRUNE1 c.933G>A synonymous variant induces exon 7 skipping, disrupts the DHHA2 domain, and leads to an atypical NMIHBA syndrome presentation: Case report and review of the literature.. *American Journal of Medical Genetics, Part A*, **2022**,	2.5	1
217	Genome sequencing reveals novel noncoding variants in PLA2G6 and LMNB1 causing progressive neurologic disease.. *Molecular Genetics & Genomic Medicine*, **2022**, e1892	2.3	2
216	A novel, de novo intronic variant in POGZ causes White-Sutton syndrome.. *American Journal of Medical Genetics, Part A*, **2022**,	2.5	0
215	Suppresses Osteosarcoma Progression In Vivo by Targeting Notch and E2F.. *JBMR Plus*, **2022**, 6, e10623	3.9	1
214	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*, **2022**, 4, e335-e341	2	0
213	Nitric oxide modulates bone anabolism through regulation of osteoblast glycolysis and differentiation. *Journal of Clinical Investigation*, **2021**, 131,	15.9	12
212	Evaluation of extracorporeal cardiopulmonary resuscitation eligibility criteria for out-of-hospital cardiac arrest patients. *BMC Research Notes*, **2021**, 14, 139	2.3	
211	Heterozygous variants in SPTBN1 cause intellectual disability and autism. *American Journal of Medical Genetics, Part A*, **2021**, 185, 2037-2045	2.5	2
210	A novel de novo intronic variant in ITPR1 causes Gillespie syndrome. *American Journal of Medical Genetics, Part A*, **2021**, 185, 2315-2324	2.5	
209	Tendon and motor phenotypes in the mouse model of recessive osteogenesis imperfecta. *ELife*, **2021**, 10,	8.9	1
208	A Novel Mouse Model for SNP in Steroid Receptor Co-Activator-1 Reveals Role in Bone Density and Breast Cancer Metastasis. *Endocrinology*, **2021**, 162,	4.8	3
207	Pregnancy in women with osteogenesis imperfecta: pregnancy characteristics, maternal, and neonatal outcomes. *American Journal of Obstetrics & Gynecology MFM*, **2021**, 3, 100362	7.4	2
206	Widespread disturbance in extracellular matrix collagen biomarker responses to teriparatide therapy in osteogenesis imperfecta. *Bone*, **2021**, 142, 115703	4.7	1
205	The transcriptional cofactor Jab1/Cops5 is crucial for BMP-mediated mouse chondrocyte differentiation by repressing p53 activity. *Journal of Cellular Physiology*, **2021**, 236, 5686-5697	7	1
204	Transcriptome-directed analysis for Mendelian disease diagnosis overcomes limitations of conventional genomic testing. *Journal of Clinical Investigation*, **2021**, 131,	15.9	28
203	Health-related quality of life in adults with osteogenesis imperfecta. *Clinical Genetics*, **2021**, 99, 772-779	4	0

202	Sites of Cre-recombinase activity in mouse lines targeting skeletal cells. *Journal of Bone and Mineral Research*, **2021**, 36, 1661-1679	6.3	3
201	COPB2 loss of function causes a coatopathy with osteoporosis and developmental delay. *American Journal of Human Genetics*, **2021**, 108, 1710-1724	11	2
200	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	2.5	1
199	Further delineation of the clinical spectrum of KAT6B disorders and allelic series of pathogenic variants. *Genetics in Medicine*, **2020**, 22, 1338-1347	8.1	9
198	regulates the action of nitrogen-containing bisphosphonates on bone. *Science Translational Medicine*, **2020**, 12,	17.5	6
197	The crucial p53-dependent oncogenic role of JAB1 in osteosarcoma in vivo. *Oncogene*, **2020**, 39, 4581-4591	9.1	11
196	A global Slc7a7 knockout mouse model demonstrates characteristic phenotypes of human lysinuric protein intolerance. *Human Molecular Genetics*, **2020**, 29, 2171-2184	5.6	5
195	De novo EIF2AK1 and EIF2AK2 Variants Are Associated with Developmental Delay, Leukoencephalopathy, and Neurologic Decompensation. *American Journal of Human Genetics*, **2020**, 106, 570-583	11	21
194	Loss- or Gain-of-Function Mutations in ACOX1 Cause Axonal Loss via Different Mechanisms. *Neuron*, **2020**, 106, 589-606.e6	13.9	32
193	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	6.2	9
192	Notch Signaling in Skeletal Development, Homeostasis and Pathogenesis. *Biomolecules*, **2020**, 10,	5.9	18
191	Fracture Healing in Collagen-Related Preclinical Models of Osteogenesis Imperfecta. *Journal of Bone and Mineral Research*, **2020**, 35, 1132-1148	6.3	8
190	Exome sequencing compared with standard genetic tests for critically ill infants with suspected genetic conditions. *Genetics in Medicine*, **2020**, 22, 1303-1310	8.1	9
189	Chronic liver disease and impaired hepatic glycogen metabolism in argininosuccinate lyase deficiency. *JCI Insight*, **2020**, 5,	9.9	4
188	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	3	2
187	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	3.9	1
186	Hearing loss in individuals with osteogenesis imperfecta in North America: Results from a multicenter study. *American Journal of Medical Genetics, Part A*, **2020**, 182, 697-704	2.5	9
185	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	8.1	4

184	Assessment of longitudinal bone growth in osteogenesis imperfecta using metacarpophalangeal pattern profiles. *Bone*, **2020**, 140, 115547	4.7	2
183	Using the Delphi method to identify cliniciansQperceived importance of pediatric exome sequencing results. *Genetics in Medicine*, **2020**, 22, 69-76	8.1	7
182	Untargeted metabolomic profiling reveals multiple pathway perturbations and new clinical biomarkers in urea cycle disorders. *Genetics in Medicine*, **2019**, 21, 1977-1986	8.1	28
181	A Multicenter Observational Cohort Study to Evaluate the Effects of Bisphosphonate Exposure on Bone Mineral Density and Other Health Outcomes in Osteogenesis Imperfecta. *JBMR Plus*, **2019**, 3, e10118	3.9	7
180	Trps1 Regulates Development of Craniofacial Skeleton and Is Required for the Initiation of Palatal Shelves Fusion. *Frontiers in Physiology*, **2019**, 10, 513	4.6	0
179	Impact of Diagnosis and Therapy on Cognitive Function in Urea Cycle Disorders. *Annals of Neurology* , **2019**, 86, 116-128	9.4	20
178	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	6.8	19
177	Mobility in osteogenesis imperfecta: a multicenter North American study. *Genetics in Medicine*, **2019**, 21, 2311-2318	8.1	6
176	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	8.1	49
175	Growth characteristics in individuals with osteogenesis imperfecta in North America: results from a multicenter study. *Genetics in Medicine*, **2019**, 21, 275-283	8.1	15
174	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	11	22
173	Bmi1 Suppresses Adipogenesis in the Hematopoietic Stem Cell Niche. *Stem Cell Reports*, **2019**, 13, 545-558	5.8	19
172	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,	0.4	78
171	Caries prevalence and experience in individuals with osteogenesis imperfecta: A cross-sectional multicenter study. *Special Care in Dentistry*, **2019**, 39, 214-219	1.7	6
170	Bi-allelic Variants in TONSL Cause SPONASTRIME Dysplasia and a Spectrum of Skeletal Dysplasia Phenotypes. *American Journal of Human Genetics*, **2019**, 104, 422-438	11	10
169	Identification of Functionally Distinct Mx1+SMA+ Periosteal Skeletal Stem Cells. *Cell Stem Cell*, **2019**, 25, 784-796.e5	18	52
168	Osteogenesis imperfecta: advancements in genetics and treatment. *Current Opinion in Pediatrics*, **2019**, 31, 708-715	3.2	25
167	Microdeletions excluding YWHAE and PAFAH1B1 cause a unique leukoencephalopathy: further delineation of the 17p13.3 microdeletion spectrum. *Genetics in Medicine*, **2019**, 21, 1652-1656	8.1	5

166	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	11	27
165	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	2.5	8
164	Oro-dental and cranio-facial characteristics of osteogenesis imperfecta type V. *European Journal of Medical Genetics*, **2019**, 62, 103606	2.6	3
163	Dental and craniofacial characteristics caused by the p.Ser40Leu mutation in IFITM5. *American Journal of Medical Genetics, Part A*, **2019**, 179, 65-70	2.5	5
162	Alterations in non-type I collagen biomarkers in osteogenesis imperfecta. *Bone*, **2019**, 120, 70-74	4.7	5
161	Autosomal recessive Noonan syndrome associated with biallelic LZTR1 variants. *Genetics in Medicine*, **2018**, 20, 1175-1185	8.1	80
160	Whole-Exome Sequencing Identifies an Intronic Cryptic Splice Site in Causing Osteogenesis Imperfecta Type VI. *JBMR Plus*, **2018**, 2, 235-239	3.9	2
159	Gene therapy for repair and regeneration of bone and cartilage. *Current Opinion in Pharmacology*, **2018**, 40, 59-66	5.1	35
158	TGF-Family Signaling in Mesenchymal Differentiation. *Cold Spring Harbor Perspectives in Biology*, **2018**, 10,	10.2	110
157	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	6.3	7
156	Prospects of Gene Therapy for Skeletal Diseases **2018**, 119-137		1
155	Arginase overexpression in neurons and its effect on traumatic brain injury. *Molecular Genetics and Metabolism*, **2018**, 125, 112-117	3.7	13
154	Argininosuccinate Lyase Deficiency Causes an Endothelial-Dependent Form of Hypertension. *American Journal of Human Genetics*, **2018**, 103, 276-287	11	18
153	Atypical Alexander disease with dystonia, retinopathy, and a brain mass mimicking astrocytoma. *Neurology: Genetics*, **2018**, 4, e248	3.8	5
152	IRF2BPL Is Associated with Neurological Phenotypes. *American Journal of Human Genetics*, **2018**, 103, 245-260	11	39
151	Pathogenic Variants in Fucokinase Cause a Congenital Disorder of Glycosylation. *American Journal of Human Genetics*, **2018**, 103, 1030-1037	11	11
150	Heterozygous WNT1 variant causing a variable bone phenotype. *American Journal of Medical Genetics, Part A*, **2018**, 176, 2419-2424	2.5	4
149	Effect of Genetic Diagnosis on Patients with Previously Undiagnosed Disease. *New England Journal of Medicine*, **2018**, 379, 2131-2139	59.2	129

148	Oral health-related quality of life in children and adolescents with osteogenesis imperfecta: cross-sectional study. *Orphanet Journal of Rare Diseases*, **2018**, 13, 187	4.2	10
147	Human Fetal and Neonatal Bone Development **2018**, 115-122		
146	Osteogenic Osteosarcoma **2018**, 768-774		
145	A multicenter study to evaluate pulmonary function in osteogenesis imperfecta. *Clinical Genetics*, **2018**, 94, 502-511	4	20
144	Mechanical properties of infant bone. *Bone*, **2018**, 113, 151-160	4.7	12
143	2017 Victor A. McKusick Leadership Award Introduction: Arthur L. Beaudet. *American Journal of Human Genetics*, **2018**, 102, 359-360	11	
142	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	11	80
141	Further evidence for the involvement of in a Shwachman-Diamond-like syndrome and expansion of the phenotypic features. *Journal of Physical Education and Sports Management*, **2018**, 4,	2.8	18
140	De Novo Missense Variants in TRAF7 Cause Developmental Delay, Congenital Anomalies, and Dysmorphic Features. *American Journal of Human Genetics*, **2018**, 103, 154-162	11	25
139	The Undiagnosed Diseases Network: Accelerating Discovery about Health and Disease. *American Journal of Human Genetics*, **2017**, 100, 185-192	11	102
138	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	4.3	11
137	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	11	23
136	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	5.4	19
135	Fkbp10 Deletion in Osteoblasts Leads to Qualitative Defects in Bone. *Journal of Bone and Mineral Research*, **2017**, 32, 1354-1367	6.3	11
134	Genetic causes and mechanisms of Osteogenesis Imperfecta. *Bone*, **2017**, 102, 40-49	4.7	58
133	2016 Curt Stern Award Address: From Rare to Common Diseases: Translating Genetic Discovery to Therapy. *American Journal of Human Genetics*, **2017**, 100, 397-400	11	
132	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	6.3	41
131	Correlations Between Bone Mechanical Properties and Bone Composition Parameters in Mouse Models of Dominant and Recessive Osteogenesis Imperfecta and the Response to Anti-TGF-β Treatment. *Journal of Bone and Mineral Research*, **2017**, 32, 347-359	6.3	21

130	MicroRNA miR-23a cluster promotes osteocyte differentiation by regulating TGF-signalling in osteoblasts. *Nature Communications*, **2017**, 8, 15000	17.4	63
129	Generalized metabolic bone disease and fracture risk in Rothmund-Thomson syndrome. *Human Molecular Genetics*, **2017**, 26, 3046-3055	5.6	6
128	Sclt1 deficiency causes cystic kidney by activating ERK and STAT3 signaling. *Human Molecular Genetics*, **2017**, 26, 2949-2960	5.6	19
127	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	11	104
126	An exome sequencing study of Moebius syndrome including atypical cases reveals an individual with CFEOM3A and a mutation. *Journal of Physical Education and Sports Management*, **2017**, 3, a000984	2.8	12
125	A Syndromic Neurodevelopmental Disorder Caused by De Novo Variants in EBF3. *American Journal of Human Genetics*, **2017**, 100, 128-137	11	65
124	Use of Exome Sequencing for Infants in Intensive Care Units: Ascertainment of Severe Single-Gene Disorders and Effect on Medical Management. *JAMA Pediatrics*, **2017**, 171, e173438	8.3	215
123	Phenotypic and molecular characterisation of CDK13-related congenital heart defects, dysmorphic facial features and intellectual developmental disorders. *Genome Medicine*, **2017**, 9, 73	14.4	30
122	Osteocyte-specific WNT1 regulates osteoblast function during bone homeostasis. *Journal of Clinical Investigation*, **2017**, 127, 2678-2688	15.9	113
121	Neonatal fractures as a presenting feature of LMOD3-associated congenital myopathy. *American Journal of Medical Genetics, Part A*, **2017**, 173, 2789-2794	2.5	12
120	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	3.7	10
119	A non-mosaic mutation in a male with severe congenital anomalies overlapping focal dermal hypoplasia. *Molecular Genetics and Metabolism Reports*, **2017**, 12, 57-61	1.8	7
118	Therapeutic Antibody Targeting Tumor- and Osteoblastic Niche-Derived Jagged1 Sensitizes Bone Metastasis to Chemotherapy. *Cancer Cell*, **2017**, 32, 731-747.e6	24.3	97
117	mTORC1 Signaling is a Critical Regulator of Postnatal Tendon Development. *Scientific Reports*, **2017**, 7, 17175	4.9	13
116	Long-term use of angiotensin-converting enzyme inhibitors protects against bone loss in African-American elderly men. *Archives of Osteoporosis*, **2017**, 12, 94	2.9	5
115	Mutations in Fibronectin Cause a Subtype of Spondylometaphyseal Dysplasia with "Corner Fractures". *American Journal of Human Genetics*, **2017**, 101, 815-823	11	22
114	Heterozygous variants in ACTL6A, encoding a component of the BAF complex, are associated with intellectual disability. *Human Mutation*, **2017**, 38, 1365-1371	4.7	17
113	Corner fracture type spondylometaphyseal dysplasia: Overlap with type II collagenopathies. *American Journal of Medical Genetics, Part A*, **2017**, 173, 733-739	2.5	5

112	Clinically severe CACNA1A alleles affect synaptic function and neurodegeneration differentially. *PLoS Genetics*, **2017**, 13, e1006905	6	42
111	Pharmacological and biological therapeutic strategies for osteogenesis imperfecta. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2016**, 172, 367-383	3.1	35
110	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	1.8	10
109	Restoration of the serum level of SERPINF1 does not correct the bone phenotype in Serpinf1 null mice. *Molecular Genetics and Metabolism*, **2016**, 117, 378-82	3.7	12
108	Glutamine and hyperammonemic crises in patients with urea cycle disorders. *Molecular Genetics and Metabolism*, **2016**, 117, 27-32	3.7	13
107	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	5.6	74
106	Sclerostin Antibody Treatment Improves the Bone Phenotype of Crtap(-/-) Mice, a Model of Recessive Osteogenesis Imperfecta. *Journal of Bone and Mineral Research*, **2016**, 31, 1030-40	6.3	50
105	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, 43-7	1.8	17
104	Losartan increases bone mass and accelerates chondrocyte hypertrophy in developing skeleton. *Molecular Genetics and Metabolism*, **2015**, 115, 53-60	3.7	14
103	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, 8613-22	5.4	29
102	Autosomal-Dominant Multiple Pterygium Syndrome Is Caused by Mutations in MYH3. *American Journal of Human Genetics*, **2015**, 96, 841-9	11	36
101	RECQL4 Regulates p53 Function In Vivo During Skeletogenesis. *Journal of Bone and Mineral Research*, **2015**, 30, 1077-89	6.3	23
100	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	3.7	8
99	Human recombinant arginase enzyme reduces plasma arginine in mouse models of arginase deficiency. *Human Molecular Genetics*, **2015**, 24, 6417-27	5.6	28
98	Blood ammonia and glutamine as predictors of hyperammonemic crises in patients with urea cycle disorder. *Genetics in Medicine*, **2015**, 17, 561-8	8.1	23
97	Catel-Manzke Syndrome: Further Delineation of the Phenotype Associated with Pathogenic Variants in. *Molecular Genetics and Metabolism Reports*, **2015**, 4, 89-91	1.8	5
96	A transgenic mouse model of OI type V supports a neomorphic mechanism of the IFITM5 mutation. *Journal of Bone and Mineral Research*, **2015**, 30, 489-98	6.3	21
95	Adult presentation of X-linked Conradi-H□ermann-Happle syndrome. *American Journal of Medical Genetics, Part A*, **2015**, 167, 1309-14	2.5	5

94	The osteogenic niche promotes early-stage bone colonization of disseminated breast cancer cells. *Cancer Cell*, **2015**, 27, 193-210	24.3	235
93	Connective tissue alterations in Fkbp10-/- mice. *Human Molecular Genetics*, **2014**, 23, 4822-31	5.6	39
92	Excessive transforming growth factor-signaling is a common mechanism in osteogenesis imperfecta. *Nature Medicine*, **2014**, 20, 670-5	50.5	172
91	Notch activation as a driver of osteogenic sarcoma. *Cancer Cell*, **2014**, 26, 390-401	24.3	93
90	Genotype-phenotype correlation--promiscuity in the era of next-generation sequencing. *New England Journal of Medicine*, **2014**, 371, 593-6	59.2	68
89	Sodium phenylbutyrate decreases plasma branched-chain amino acids in patients with urea cycle disorders. *Molecular Genetics and Metabolism*, **2014**, 113, 131-5	3.7	40
88	ADAMTS-7 forms a positive feedback loop with TNF-in the pathogenesis of osteoarthritis. *Annals of the Rheumatic Diseases*, **2014**, 73, 1575-84	2.4	50
87	Mice expressing mutant Trpv4 recapitulate the human TRPV4 disorders. *Journal of Bone and Mineral Research*, **2014**, 29, 1815-1822	6.3	15
86	Lysinuric Protein Intolerance Presenting with Multiple Fractures. *Molecular Genetics and Metabolism Reports*, **2014**, 1, 176-183	1.8	15
85	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	3.7	32
84	Diagnosis of ALG12-CDG by exome sequencing in a case of severe skeletal dysplasia. *Molecular Genetics and Metabolism Reports*, **2014**, 1, 213-219	1.8	14
83	Combinatorial treatment with oncolytic adenovirus and helper-dependent adenovirus augments adenoviral cancer gene therapy. *Molecular Therapy - Oncolytics*, **2014**, 1, 14008	6.4	17
82	SERPINF1 as a Cause of Osteogenesis Imperfecta Type VI **2014**, 167-172		0
81	The swaying mouse as a model of osteogenesis imperfecta caused by WNT1 mutations. *Human Molecular Genetics*, **2014**, 23, 4035-42	5.6	51
80	Dual role of the Trps1 transcription factor in dentin mineralization. *Journal of Biological Chemistry*, **2014**, 289, 27481-93	5.4	17
79	Differential effects of collagen prolyl 3-hydroxylation on skeletal tissues. *PLoS Genetics*, **2014**, 10, e1004621		24
78	Recessive Osteogenesis Imperfecta Due to Mutations in CRTAP, LEPRE1 and PPIB **2014**, 141-150		
77	The genetic basis of DOORS syndrome: an exome-sequencing study. *Lancet Neurology, The*, **2014**, 13, 44-58	24.1	96

76	Notch signaling in skeletal stem cells. *Calcified Tissue International*, **2014**, 94, 68-77	3.9	33
75	Branched-chain amino acid metabolism: from rare Mendelian diseases to more common disorders. *Human Molecular Genetics*, **2014**, 23, R1-8	5.6	73
74	Evaluation of teriparatide treatment in adults with osteogenesis imperfecta. *Journal of Clinical Investigation*, **2014**, 124, 491-8	15.9	107
73	Next-generation sequencing for disorders of low and high bone mineral density. *Osteoporosis International*, **2013**, 24, 2253-9	5.3	42
72	Phenotypic variability of osteogenesis imperfecta type V caused by an IFITM5 mutation. *Journal of Bone and Mineral Research*, **2013**, 28, 1523-30	6.3	55
71	Yunis-Var□ syndrome is caused by mutations in FIG4, encoding a phosphoinositide phosphatase. *American Journal of Human Genetics*, **2013**, 92, 781-91	11	101
70	WNT1 mutations in early-onset osteoporosis and osteogenesis imperfecta. *New England Journal of Medicine*, **2013**, 368, 1809-16	59.2	253
69	Ammonia control and neurocognitive outcome among urea cycle disorder patients treated with glycerol phenylbutyrate. *Hepatology*, **2013**, 57, 2171-9	11.2	61
68	Structure-based design and mechanisms of allosteric inhibitors for mitochondrial branched-chain □ketoacid dehydrogenase kinase. *Proceedings of the National Academy of Sciences of the United States of America*, **2013**, 110, 9728-33	11.5	40
67	Early childhood presentation of Czech dysplasia. *Clinical Dysmorphology*, **2013**, 22, 76-80	0.9	5
66	Osteogenesis imperfecta without features of type V caused by a mutation in the IFITM5 gene. *Journal of Bone and Mineral Research*, **2013**, 28, 2333-7	6.3	21
65	Notch gain of function inhibits chondrocyte differentiation via Rbpj-dependent suppression of Sox9. *Journal of Bone and Mineral Research*, **2013**, 28, 649-59	6.3	47
64	E-selectin ligand 1 regulates bone remodeling by limiting bioactive TGF-□n the bone microenvironment. *Proceedings of the National Academy of Sciences of the United States of America*, **2013**, 110, 7336-41	11.5	24
63	Optimizing therapy for argininosuccinic aciduria. *Molecular Genetics and Metabolism*, **2012**, 107, 10-4	3.7	8
62	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, 315-21	3.7	28
61	Whole-exome sequencing identifies mutations in the nucleoside transporter gene SLC29A3 in dysosteosclerosis, a form of osteopetrosis. *Human Molecular Genetics*, **2012**, 21, 4904-9	5.6	63
60	Assessment of bone mineral status in children with Marfan syndrome. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 2221-4	2.5	25
59	WDR35 mutation in siblings with Sensenbrenner syndrome: a ciliopathy with variable phenotype. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 2917-24	2.5	35

58	miRNA-34c regulates Notch signaling during bone development. *Human Molecular Genetics*, **2012**, 21, 2991-3000	5.6	182
57	Mutations in KAT6B, encoding a histone acetyltransferase, cause Genitopatellar syndrome. *American Journal of Human Genetics*, **2012**, 90, 282-9	11	99
56	Nitric-oxide supplementation for treatment of long-term complications in argininosuccinic aciduria. *American Journal of Human Genetics*, **2012**, 90, 836-46	11	56
55	Response to Srilatha et al.. *Genetics in Medicine*, **2012**, 14, 628-628	8.1	
54	One NOTCH Further: Jagged 1 in Bone Metastasis. *Cancer Cell*, **2011**, 19, 159-61	24.3	12
53	Argininosuccinate lyase deficiency-argininosuccinic aciduria and beyond. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2011**, 157C, 45-53	3.1	60
52	Mutations in FKBP10 cause recessive osteogenesis imperfecta and Bruck syndrome. *Journal of Bone and Mineral Research*, **2011**, 26, 666-72	6.3	123
51	Mutations in SERPINF1 cause osteogenesis imperfecta type VI. *Journal of Bone and Mineral Research*, **2011**, 26, 2798-803	6.3	141
50	Phenylbutyrate therapy for maple syrup urine disease. *Human Molecular Genetics*, **2011**, 20, 631-40	5.6	58
49	Phenylbutyrate improves nitrogen disposal via an alternative pathway without eliciting an increase in protein breakdown and catabolism in control and ornithine transcarbamylase-deficient patients. *American Journal of Clinical Nutrition*, **2011**, 93, 1248-54	7	8
48	Requirement of argininosuccinate lyase for systemic nitric oxide production. *Nature Medicine*, **2011**, 17, 1619-26	50.5	161
47	Arginase II reduces arginine availability and nitric oxide production during endotoxemia. *FASEB Journal*, **2011**, 25, 983.20	0.9	
46	Alteration of Notch signaling in skeletal development and disease. *Annals of the New York Academy of Sciences*, **2010**, 1192, 257-68	6.5	49
45	Reply to Ligthart-Melis et al.. *American Journal of Physiology - Endocrinology and Metabolism*, **2010**, 299, E684-E684	6	3
44	NOTCHing the bone: insights into multi-functionality. *Bone*, **2010**, 46, 274-80	4.7	65
43	Establishing a consortium for the study of rare diseases: The Urea Cycle Disorders Consortium. *Molecular Genetics and Metabolism*, **2010**, 100 Suppl 1, S97-105	3.7	60
42	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, 221-8	3.7	67
41	Signaling pathways in human skeletal dysplasias. *Annual Review of Genomics and Human Genetics*, **2010**, 11, 189-217	9.7	49

40	Large-scale production of high-quality helper-dependent adenoviral vectors using adherent cells in cell factories. *Human Gene Therapy*, **2010**, 21, 120-6	4.8	33
39	Mutations in the gene encoding the RER protein FKBP65 cause autosomal-recessive osteogenesis imperfecta. *American Journal of Human Genetics*, **2010**, 86, 551-9	11	238
38	Mutations in the Gene Encoding the RER Protein FKBP65 Cause Autosomal-Recessive Osteogenesis Imperfecta. *American Journal of Human Genetics*, **2010**, 87, 572-573	11	13
37	Osteosclerosis owing to Notch gain of function is solely Rbpj-dependent. *Journal of Bone and Mineral Research*, **2010**, 25, 2175-83	6.3	54
36	E-selectin ligand-1 regulates growth plate homeostasis in mice by inhibiting the intracellular processing and secretion of mature TGF-beta. *Journal of Clinical Investigation*, **2010**, 120, 2474-85	15.9	21
35	Self-Complementary AAV Vectors Cause a Substantially Heightened TLR9-Dependent Innate Immune Response In the Liver. *Blood*, **2010**, 116, 252-252	2.2	1
34	Generalized connective tissue disease in Crtap-/- mouse. *PLoS ONE*, **2010**, 5, e10560	3.7	45
33	Glutamine: precursor or nitrogen donor for the synthesis of citrulline?. *FASEB Journal*, **2010**, 24, 740.14	0.9	
32	Precursors for the synthesis of citrulline in mice fed arginine free diets. *FASEB Journal*, **2010**, 24, 740.26	0.9	
31	Notch signaling contributes to the pathogenesis of human osteosarcomas. *Human Molecular Genetics*, **2009**, 18, 1464-70	5.6	140
30	Systemic hypertension in two patients with ASL deficiency: a result of nitric oxide deficiency?. *Molecular Genetics and Metabolism*, **2009**, 98, 195-7	3.7	33
29	Dimorphic effects of Notch signaling in bone homeostasis. *Nature Medicine*, **2008**, 14, 299-305	50.5	322
28	Cross-sectional multicenter study of patients with urea cycle disorders in the United States. *Molecular Genetics and Metabolism*, **2008**, 94, 397-402	3.7	159
27	Uncoupling of chondrocyte differentiation and perichondrial mineralization underlies the skeletal dysplasia in tricho-rhino-phalangeal syndrome. *Human Molecular Genetics*, **2008**, 17, 2244-54	5.6	73
26	CRTAP and LEPRE1 mutations in recessive osteogenesis imperfecta. *Human Mutation*, **2008**, 29, 1435-42	4.7	172
25	Brachy-syndactyly caused by loss of Sfrp2 function. *Journal of Cellular Physiology*, **2008**, 217, 127-37	7	50
24	Phenylbutyrate reduces plasma leucine concentrations without affecting the flux of leucine. *FASEB Journal*, **2007**, 21, A335	0.9	
23	Deficiency of cartilage-associated protein in recessive lethal osteogenesis imperfecta. *New England Journal of Medicine*, **2006**, 355, 2757-64	59.2	255

22	Dominance of SOX9 function over RUNX2 during skeletogenesis. *Proceedings of the National Academy of Sciences of the United States of America*, **2006**, 103, 19004-9	11.5	266
21	CRTAP is required for prolyl 3- hydroxylation and mutations cause recessive osteogenesis imperfecta. *Cell*, **2006**, 127, 291-304	56.2	394
20	Vestigial ornithine transcarbamylase activity does not impair ureagenesis in Otcspf-ash mice. *FASEB Journal*, **2006**, 20, A1093	0.9	
19	Ornithine restores ureagenesis capacity and mitigates hyperammonemia in Otcspf-ash mice. *FASEB Journal*, **2006**, 20, A1092	0.9	
18	Considerations in the difficult-to-manage urea cycle disorder patient. *Critical Care Clinics*, **2005**, 21, S19-25	4.5	11
17	Clinical consequences of urea cycle enzyme deficiencies and potential links to arginine and nitric oxide metabolism. *Journal of Nutrition*, **2004**, 134, 2775S-2782S; discussion 2796S-2797S	4.1	65
16	Effect of alternative pathway therapy on branched chain amino acid metabolism in urea cycle disorder patients. *Molecular Genetics and Metabolism*, **2004**, 81 Suppl 1, S79-85	3.7	83
15	A natural history of cleidocranial dysplasia. *American Journal of Medical Genetics Part A*, **2001**, 104, 1-6		147
14	Transcriptional dysregulation in skeletal malformation syndromes. *American Journal of Medical Genetics Part A*, **2001**, 106, 258-271		18
13	Regulation of glomerular basement membrane collagen expression by LMX1B contributes to renal disease in nail patella syndrome. *Nature Genetics*, **2001**, 27, 205-8	36.3	172
12	Long-term correction of urea cycle disorders. *Journal of Pediatrics*, **2001**, 138, S62-71	3.6	54
11	The long and the short of it: developmental genetics of the skeletal dysplasias. *Clinical Genetics*, **1999**, 57, 50-59	4	
10	Trisomy 16q in a female newborn with a de novo X;16 translocation and hypoplastic left heart. *American Journal of Medical Genetics Part A*, **1999**, 82, 128-31		17
9	Mutations in LMX1B cause abnormal skeletal patterning and renal dysplasia in nail patella syndrome. *Nature Genetics*, **1998**, 19, 47-50	36.3	419
8	Limb and kidney defects in Lmx1b mutant mice suggest an involvement of LMX1B in human nail patella syndrome. *Nature Genetics*, **1998**, 19, 51-5	36.3	441
7	Cloning, characterization, and chromosomal assignment of the human ortholog of murine Zfp-37, a candidate gene for Nager syndrome. *Mammalian Genome*, **1998**, 9, 458-62	3.2	27
6	The long and the short of it: developmental genetics of the skeletal dysplasias. *Clinical Genetics*, **1998**, 54, 464-73	4	20
5	Missense mutations abolishing DNA binding of the osteoblast-specific transcription factor OSF2/CBFA1 in cleidocranial dysplasia. *Nature Genetics*, **1997**, 16, 307-10	36.3	490

4	Linkage of Marfan syndrome and a phenotypically related disorder to two different fibrillin genes. *Nature*, **1991**, 352, 330-4	50.4	605
3	Mesomelic Dysplasia With Specific Autopodal Synostoses: A Third Observation And Further Delineation Of The Multiple Congenital Anomaly Syndrome		1
2	COPB2haploinsufficiency causes a coatopathy with osteoporosis and developmental delay		1
1	Loss-of-function in IRF2BPL is associated with neurological phenotypes		2