Brendan Lee

List of Publications by Citations

201 papers	12,641 citations	58 h-index	107 g-index
203 ext. papers	14,202 ext. citations	9.3 avg, IF	5.72 L-index

#	Paper	IF	Citations
201	Linkage of Marfan syndrome and a phenotypically related disorder to two different fibrillin genes. *Nature*, **1991**, 352, 330-4	50.4	605
200	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
199	Recurrent reciprocal 1q21.1 deletions and duplications associated with microcephaly or macrocephaly and developmental and behavioral abnormalities. *Nature Genetics*, **2008**, 40, 1466-71	36.3	457
198	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
197	Mutations in LMX1B cause abnormal skeletal patterning and renal dysplasia in nail patella syndrome. *Nature Genetics*, **1998**, 19, 47-50	36.3	419
196	CRTAP is required for prolyl 3- hydroxylation and mutations cause recessive osteogenesis imperfecta. *Cell*, **2006**, 127, 291-304	56.2	394
195	Cloning of the putative tumour suppressor gene for hereditary multiple exostoses (EXT1). *Nature Genetics*, **1995**, 11, 137-43	36.3	361
194	Dimorphic effects of Notch signaling in bone homeostasis. *Nature Medicine*, **2008**, 14, 299-305	50.5	322
193	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
192	Deficiency of cartilage-associated protein in recessive lethal osteogenesis imperfecta. *New England Journal of Medicine*, **2006**, 355, 2757-64	59.2	255
191	Type X collagen gene regulation by Runx2 contributes directly to its hypertrophic chondrocyte-specific expression in vivo. *Journal of Cell Biology*, **2003**, 162, 833-42	7.3	249
190	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
189	The osteogenic niche promotes early-stage bone colonization of disseminated breast cancer cells. *Cancer Cell*, **2015**, 27, 193-210	24.3	235
188	Genetic linkage of the Marfan syndrome, ectopia lentis, and congenital contractural arachnodactyly to the fibrillin genes on chromosomes 15 and 5. The International Marfan Syndrome Collaborative Study. *New England Journal of Medicine*, **1992**, 326, 905-9	59.2	232
187	Excessive transforming growth factor-□signaling is a common mechanism in osteogenesis imperfecta. *Nature Medicine*, **2014**, 20, 670-5	50.5	172
186	CRTAP and LEPRE1 mutations in recessive osteogenesis imperfecta. *Human Mutation*, **2008**, 29, 1435-42	4.7	172
185	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

184	Requirement of argininosuccinate lyase for systemic nitric oxide production. *Nature Medicine*, **2011**, 17, 1619-26	50.5	161
183	Use of a liver-specific promoter reduces immune response to the transgene in adenoviral vectors. *Human Gene Therapy*, **1999**, 10, 1773-81	4.8	160
182	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
181	Diagnosis, symptoms, frequency and mortality of 260 patients with urea cycle disorders from a 21-year, multicentre study of acute hyperammonaemic episodes. *Acta Paediatrica, International Journal of Paediatrics*, **2008**, 97, 1420-5	3.1	153
180	The genome of self-complementary adeno-associated viral vectors increases Toll-like receptor 9-dependent innate immune responses in the liver. *Blood*, **2011**, 117, 6459-68	2.2	148
179	A natural history of cleidocranial dysplasia. *American Journal of Medical Genetics Part A*, **2001**, 104, 1-6		147
178	Mutation analysis of LMX1B gene in nail-patella syndrome patients. *American Journal of Human Genetics*, **1998**, 63, 1651-8	11	142
177	Mutations in SERPINF1 cause osteogenesis imperfecta type VI. *Journal of Bone and Mineral Research*, **2011**, 26, 2798-803	6.3	141
176	Notch signaling contributes to the pathogenesis of human osteosarcomas. *Human Molecular Genetics*, **2009**, 18, 1464-70	5.6	140
175	Long-term stable correction of low-density lipoprotein receptor-deficient mice with a helper-dependent adenoviral vector expressing the very low-density lipoprotein receptor. *Circulation*, **2001**, 103, 1274-81	16.7	126
174	Toll-like receptor 9 triggers an innate immune response to helper-dependent adenoviral vectors. *Molecular Therapy*, **2007**, 15, 378-85	11.7	125
173	Mutations in FKBP10 cause recessive osteogenesis imperfecta and Bruck syndrome. *Journal of Bone and Mineral Research*, **2011**, 26, 666-72	6.3	123
172	Lifelong elimination of hyperbilirubinemia in the Gunn rat with a single injection of helper-dependent adenoviral vector. *Proceedings of the National Academy of Sciences of the United States of America*, **2005**, 102, 3930-5	11.5	118
171	Transcriptional induction of slit diaphragm genes by Lmx1b is required in podocyte differentiation. *Journal of Clinical Investigation*, **2002**, 109, 1065-1072	15.9	113
170	TGF-Family Signaling in Mesenchymal Differentiation. *Cold Spring Harbor Perspectives in Biology*, **2018**, 10,	10.2	110
169	Toxicological comparison of E2a-deleted and first-generation adenoviral vectors expressing alpha1-antitrypsin after systemic delivery. *Human Gene Therapy*, **1998**, 9, 1587-98	4.8	108
168	Evaluation of teriparatide treatment in adults with osteogenesis imperfecta. *Journal of Clinical Investigation*, **2014**, 124, 491-8	15.9	107
167	Therapeutic Antibody Targeting Tumor- and Osteoblastic Niche-Derived Jagged1 Sensitizes Bone Metastasis to Chemotherapy. *Cancer Cell*, **2017**, 32, 731-747.e6	24.3	97

166	Interaction of TGF and BMP signaling pathways during chondrogenesis. *PLoS ONE*, **2011**, 6, e16421	3.7	97
165	Suppression of neuropil aggregates and neurological symptoms by an intracellular antibody implicates the cytoplasmic toxicity of mutant huntingtin. *Journal of Cell Biology*, **2008**, 181, 803-16	7.3	97
164	Unmasked adult-onset urea cycle disorders in the critical care setting. *Critical Care Clinics*, **2005**, 21, S1-8	4.5	96
163	Determinants of vascular permeability in the kidney glomerulus. *Journal of Biological Chemistry*, **2002**, 277, 31154-62	5.4	92
162	Inborn errors of metabolism: the flux from Mendelian to complex diseases. *Nature Reviews Genetics* , **2006**, 7, 449-60	30.1	87
161	Consequences of mutations in the non-coding RMRP RNA in cartilage-hair hypoplasia. *Human Molecular Genetics*, **2005**, 14, 3723-40	5.6	84
160	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
159	Autosomal recessive Noonan syndrome associated with biallelic LZTR1 variants. *Genetics in Medicine*, **2018**, 20, 1175-1185	8.1	80
158	Clinical, biochemical, and molecular spectrum of hyperargininemia due to arginase I deficiency. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2006**, 142C, 113-20	3.1	77
157	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
156	Genome-wide oligonucleotide-based array comparative genome hybridization analysis of non-isolated congenital diaphragmatic hernia. *Human Molecular Genetics*, **2007**, 16, 424-30	5.6	71
155	Reduced inflammation and improved airway expression using helper-dependent adenoviral vectors with a K18 promoter. *Molecular Therapy*, **2003**, 7, 649-58	11.7	69
154	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
153	NOTCHing the bone: insights into multi-functionality. *Bone*, **2010**, 46, 274-80	4.7	65
152	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
151	Mouse model reveals the role of SOX7 in the development of congenital diaphragmatic hernia associated with recurrent deletions of 8p23.1. *Human Molecular Genetics*, **2012**, 21, 4115-25	5.6	63
150	Ammonia control and neurocognitive outcome among urea cycle disorder patients treated with glycerol phenylbutyrate. *Hepatology*, **2013**, 57, 2171-9	11.2	61
149	Urea cycle disorders: clinical presentation outside the newborn period. *Critical Care Clinics*, **2005**, 21, S9-17	4.5	61

148	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
147	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
146	Argininosuccinate lyase deficiency. *Genetics in Medicine*, **2012**, 14, 501-7	8.1	59
145	Genetic causes and mechanisms of Osteogenesis Imperfecta. *Bone*, **2017**, 102, 40-49	4.7	58
144	Phenylbutyrate therapy for maple syrup urine disease. *Human Molecular Genetics*, **2011**, 20, 631-40	5.6	58
143	Nitric-oxide supplementation for treatment of long-term complications in argininosuccinic aciduria. *American Journal of Human Genetics*, **2012**, 90, 836-46	11	56
142	Immune response to helper dependent adenoviral mediated liver gene therapy: challenges and prospects. *Current Gene Therapy*, **2007**, 7, 297-305	4.3	55
141	Developmental outcomes with early orthotopic liver transplantation for infants with neonatal-onset urea cycle defects and a female patient with late-onset ornithine transcarbamylase deficiency. *Pediatrics*, **2004**, 114, e523-6	7.4	55
140	Osteosclerosis owing to Notch gain of function is solely Rbpj-dependent. *Journal of Bone and Mineral Research*, **2010**, 25, 2175-83	6.3	54
139	Long-term correction of urea cycle disorders. *Journal of Pediatrics*, **2001**, 138, S62-71	3.6	54
138	The phenotype of recurrent 10q22q23 deletions and duplications. *European Journal of Human Genetics*, **2011**, 19, 400-8	5.3	52
137	Long-term correction of ornithine transcarbamylase deficiency by WPRE-mediated overexpression using a helper-dependent adenovirus. *Molecular Therapy*, **2004**, 10, 492-9	11.7	52
136	Runx2 contributes to murine Col10a1 gene regulation through direct interaction with its cis-enhancer. *Journal of Bone and Mineral Research*, **2011**, 26, 2899-910	6.3	50
135	Brachy-syndactyly caused by loss of Sfrp2 function. *Journal of Cellular Physiology*, **2008**, 217, 127-37	7	50
134	Transcriptional induction of slit diaphragm genes by Lmx1b is required in podocyte differentiation. *Journal of Clinical Investigation*, **2002**, 109, 1065-72	15.9	50
133	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
132	Alteration of Notch signaling in skeletal development and disease. *Annals of the New York Academy of Sciences*, **2010**, 1192, 257-68	6.5	49
131	Signaling pathways in human skeletal dysplasias. *Annual Review of Genomics and Human Genetics*, **2010**, 11, 189-217	9.7	49

130	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
129	Phenylbutyrate therapy for pyruvate dehydrogenase complex deficiency and lactic acidosis. *Science Translational Medicine*, **2013**, 5, 175ra31	17.5	46
128	RMRP mutations in cartilage-hair hypoplasia. *American Journal of Medical Genetics, Part A*, **2006**, 140, 2121-30	2.5	46
127	Generation of helper-dependent adenoviral vectors by homologous recombination. *Molecular Therapy*, **2002**, 5, 204-10	11.7	46
126	Generalized connective tissue disease in Crtap-/- mouse. *PLoS ONE*, **2010**, 5, e10560	3.7	45
125	Insights into the pathogenesis and treatment of cancer from inborn errors of metabolism. *American Journal of Human Genetics*, **2011**, 88, 402-21	11	44
124	Glutamine: precursor or nitrogen donor for citrulline synthesis?. *American Journal of Physiology - Endocrinology and Metabolism*, **2010**, 299, E69-79	6	44
123	Neonatal helper-dependent adenoviral vector gene therapy mediates correction of hemophilia A and tolerance to human factor VIII. *Proceedings of the National Academy of Sciences of the United States of America*, **2011**, 108, 2082-7	11.5	44
122	Mutations and promoter SNPs in RUNX2, a transcriptional regulator of bone formation. *Molecular Genetics and Metabolism*, **2005**, 86, 257-68	3.7	44
121	Helper-dependent adenoviral vector-mediated long-term expression of human apolipoprotein A-I reduces atherosclerosis in apo E-deficient mice. *Gene*, **2004**, 327, 153-60	3.8	43
120	The fibrillin-Marfan syndrome connection. *BioEssays*, **1993**, 15, 589-94	4.1	43
119	High-frequency detection of deletions and variable rearrangements at the ornithine transcarbamylase (OTC) locus by oligonucleotide array CGH. *Molecular Genetics and Metabolism*, **2009**, 96, 97-105	3.7	42
118	Clinically severe CACNA1A alleles affect synaptic function and neurodegeneration differentially. *PLoS Genetics*, **2017**, 13, e1006905	6	42
117	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
116	aP2-Cre-mediated inactivation of acetyl-CoA carboxylase 1 causes growth retardation and reduced lipid accumulation in adipose tissues. *Proceedings of the National Academy of Sciences of the United States of America*, **2009**, 106, 17576-81	11.5	41
115	Correction of murine hemophilia A and immunological differences of factor VIII variants delivered by helper-dependent adenoviral vectors. *Molecular Therapy*, **2007**, 15, 2080-7	11.7	41
114	Dysregulation of chondrogenesis in human cleidocranial dysplasia. *American Journal of Human Genetics*, **2005**, 77, 305-12	11	40
113	IRF2BPL Is Associated with Neurological Phenotypes. *American Journal of Human Genetics*, **2018**, 103, 245-260	11	39

112	Connective tissue alterations in Fkbp10-/- mice. *Human Molecular Genetics*, **2014**, 23, 4822-31	5.6	39
111	Unique Roles of TLR9- and MyD88-Dependent and -Independent Pathways in Adaptive Immune Responses to AAV-Mediated Gene Transfer. *Journal of Innate Immunity*, **2015**, 7, 302-14	6.9	38
110	Differential type I interferon-dependent transgene silencing of helper-dependent adenoviral vs. adeno-associated viral vectors in vivo. *Molecular Therapy*, **2013**, 21, 796-805	11.7	36
109	Ovotestes and XY sex reversal in a female with an interstitial 9q33.3-q34.1 deletion encompassing NR5A1 and LMX1B causing features of Genitopatellar syndrome. *American Journal of Medical Genetics, Part A*, **2007**, 143A, 1071-81	2.5	36
108	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
107	Nutritional management of urea cycle disorders. *Critical Care Clinics*, **2005**, 21, S27-35	4.5	34
106	An integrated approach to the diagnosis and prospective management of partial ornithine transcarbamylase deficiency. *Pediatrics*, **2002**, 109, 150-2	7.4	34
105	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
104	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
103	Loss- or Gain-of-Function Mutations in ACOX1 Cause Axonal Loss via Different Mechanisms. *Neuron*, **2020**, 106, 589-606.e6	13.9	32
102	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
101	Ammonia control in children ages 2 months through 5 years with urea cycle disorders: comparison of sodium phenylbutyrate and glycerol phenylbutyrate. *Journal of Pediatrics*, **2013**, 162, 1228-34, 1234.e1	3.6	32
100	Deficiency of FRAS1-related extracellular matrix 1 (FREM1) causes congenital diaphragmatic hernia in humans and mice. *Human Molecular Genetics*, **2013**, 22, 1026-38	5.6	32
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	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
97	Transcriptome-directed analysis for Mendelian disease diagnosis overcomes limitations of conventional genomic testing. *Journal of Clinical Investigation*, **2021**, 131,	15.9	28
96	Short-term correction of arginase deficiency in a neonatal murine model with a helper-dependent adenoviral vector. *Molecular Therapy*, **2009**, 17, 1155-63	11.7	27
95	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

94	Coordinated and unique functions of the E-selectin ligand ESL-1 during inflammatory and hematopoietic recruitment in mice. *Blood*, **2013**, 122, 3993-4001	2.2	26
93	MyD88-dependent silencing of transgene expression during the innate and adaptive immune response to helper-dependent adenovirus. *Human Gene Therapy*, **2010**, 21, 325-36	4.8	26
92	An allelic series of mice reveals a role for RERE in the development of multiple organs affected in chromosome 1p36 deletions. *PLoS ONE*, **2013**, 8, e57460	3.7	25
91	Insight into podocyte differentiation from the study of human genetic disease: nail-patella syndrome and transcriptional regulation in podocytes. *Pediatric Research*, **2002**, 51, 551-8	3.2	25
90	Urea-cycle disorders as a paradigm for inborn errors of hepatocyte metabolism. *Trends in Molecular Medicine*, **2002**, 8, 583-9	11.5	25
89	Osteogenesis imperfecta: advancements in genetics and treatment. *Current Opinion in Pediatrics*, **2019**, 31, 708-715	3.2	25
88	E-selectin ligand 1 regulates bone remodeling by limiting bioactive TGF-β in the bone microenvironment. *Proceedings of the National Academy of Sciences of the United States of America*, **2013**, 110, 7336-41	11.5	24
87	RECQL4 Regulates p53 Function In Vivo During Skeletogenesis. *Journal of Bone and Mineral Research*, **2015**, 30, 1077-89	6.3	23
86	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
85	Toxicity and adaptive immune response to intracellular transgenes delivered by helper-dependent vs. first generation adenoviral vectors. *Molecular Genetics and Metabolism*, **2005**, 84, 278-88	3.7	23
84	Lmx1b expression during joint and tendon formation: localization and evaluation of potential downstream targets. *Gene Expression Patterns*, **2004**, 4, 397-405	1.5	23
83	Urea Cycle Disorders. *Current Treatment Options in Neurology*, **2003**, 5, 309-319	4.4	23
82	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
81	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
80	Localization of the cis-enhancer element for mouse type X collagen expression in hypertrophic chondrocytes in vivo. *Journal of Bone and Mineral Research*, **2009**, 24, 1022-32	6.3	21
79	Capsid-modified adenoviral vectors for improved muscle-directed gene therapy. *Human Gene Therapy*, **2012**, 23, 1065-70	4.8	21
78	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
77	Impact of Diagnosis and Therapy on Cognitive Function in Urea Cycle Disorders. *Annals of Neurology* , **2019**, 86, 116-128	9.4	20

76	The long and the short of it: developmental genetics of the skeletal dysplasias. *Clinical Genetics*, **1998**, 54, 464-73	4	20
75	Transatlantic combined and comparative data analysis of 1095 patients with urea cycle disorders-A successful strategy for clinical research of rare diseases. *Journal of Inherited Metabolic Disease*, **2019**, 42, 93-106	5.4	20
74	A multicenter study to evaluate pulmonary function in osteogenesis imperfecta. *Clinical Genetics*, **2018**, 94, 502-511	4	20
73	Sclt1 deficiency causes cystic kidney by activating ERK and STAT3 signaling. *Human Molecular Genetics*, **2017**, 26, 2949-2960	5.6	19
72	FBN1 contributing to familial congenital diaphragmatic hernia. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 831-6	2.5	19
71	Phenotypic correction of ornithine transcarbamylase deficiency using low dose helper-dependent adenoviral vectors. *Journal of Gene Medicine*, **2008**, 10, 890-6	3.5	19
70	Dendritic cell function after gene transfer with adenovirus-calcium phosphate co-precipitates. *Molecular Therapy*, **2007**, 15, 386-92	11.7	19
69	NOD2 signaling contributes to the innate immune response against helper-dependent adenovirus vectors independently of MyD88 in vivo. *Human Gene Therapy*, **2011**, 22, 1071-82	4.8	18
68	Transcriptional dysregulation in skeletal malformation syndromes. *American Journal of Medical Genetics Part A*, **2001**, 106, 258-271		18
67	Isolation, characterization, and mapping of a zinc finger gene, ZFP95, containing both a SCAN box and an alternatively spliced KRAB A domain. *Genomics*, **1999**, 62, 119-22	4.3	18
66	Combinatorial treatment with oncolytic adenovirus and helper-dependent adenovirus augments adenoviral cancer gene therapy. *Molecular Therapy - Oncolytics*, **2014**, 1, 14008	6.4	17
65	Dual role of the Trps1 transcription factor in dentin mineralization. *Journal of Biological Chemistry*, **2014**, 289, 27481-93	5.4	17
64	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
63	Characterization of a new syndrome that associates craniosynostosis, delayed fontanel closure, parietal foramina, imperforate anus, and skin eruption: CDAGS. *American Journal of Human Genetics* , **2005**, 77, 161-8	11	16
62	Genetic counseling issues in urea cycle disorders. *Critical Care Clinics*, **2005**, 21, S37-44	4.5	16
61	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
60	Mice expressing mutant Trpv4 recapitulate the human TRPV4 disorders. *Journal of Bone and Mineral Research*, **2014**, 29, 1815-1822	6.3	15
59	Gene therapy for inborn errors of liver metabolism. *Molecular Genetics and Metabolism*, **2005**, 86, 13-24	3.7	15

58	Losartan increases bone mass and accelerates chondrocyte hypertrophy in developing skeleton. *Molecular Genetics and Metabolism*, **2015**, 115, 53-60	3.7	14
57	Enteral arginase II provides ornithine for citrulline synthesis. *American Journal of Physiology - Endocrinology and Metabolism*, **2011**, 300, E188-94	6	14
56	Transcriptional repression of the Dspp gene leads to dentinogenesis imperfecta phenotype in Col1a1-Trps1 transgenic mice. *Journal of Bone and Mineral Research*, **2012**, 27, 1735-45	6.3	13
55	Plasma arginine and ornithine are the main citrulline precursors in mice infused with arginine-free diets. *Journal of Nutrition*, **2010**, 140, 1432-7	4.1	13
54	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
53	One NOTCH Further: Jagged 1 in Bone Metastasis. *Cancer Cell*, **2011**, 19, 159-61	24.3	12
52	In vivo urea kinetic studies in conscious mice. *Journal of Nutrition*, **2006**, 136, 202-6	4.1	12
51	Nitric oxide modulates bone anabolism through regulation of osteoblast glycolysis and differentiation. *Journal of Clinical Investigation*, **2021**, 131,	15.9	12
50	The crucial p53-dependent oncogenic role of JAB1 in osteosarcoma in vivo. *Oncogene*, **2020**, 39, 4581-4591	9.1	11
49	Interaction between murine spf-ash mutation and genetic background yields different metabolic phenotypes. *American Journal of Physiology - Endocrinology and Metabolism*, **2007**, 293, E1764-71	6	11
48	De novo three-way chromosome translocation 46,XY,t(4;6;21)(p16;p21.1;q21) in a male with cleidocranial dysplasia. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 453-8	2.5	11
47	Considerations in the difficult-to-manage urea cycle disorder patient. *Critical Care Clinics*, **2005**, 21, S19-25	4.5	11
46	Expression profiling of human fetal growth plate cartilage by EST sequencing. *Matrix Biology*, **2005**, 24, 530-8	11.4	11
45	Ornithine restores ureagenesis capacity and mitigates hyperammonemia in Otc(spf-ash) mice. *Journal of Nutrition*, **2006**, 136, 1834-8	4.1	11
44	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
43	Reduced ornithine transcarbamylase activity does not impair ureagenesis in Otc(spf-ash) mice. *Journal of Nutrition*, **2006**, 136, 1017-20	4.1	10
42	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
41	Impairment of cognitive function in ornithine transcarbamylase deficiency is global rather than domain-specific and is associated with disease onset, sex, maximum ammonium, and number of hyperammonemic events. *Journal of Inherited Metabolic Disease*, **2019**, 42, 243-253	5.4	9

40	Fracture Healing in Collagen-Related Preclinical Models of Osteogenesis Imperfecta. *Journal of Bone and Mineral Research*, **2020**, 35, 1132-1148	6.3	8
39	Optimizing therapy for argininosuccinic aciduria. *Molecular Genetics and Metabolism*, **2012**, 107, 10-4	3.7	8
38	Cytokine-conditioned dendritic cells induce humoral tolerance to protein therapy in mice. *Human Gene Therapy*, **2012**, 23, 769-80	4.8	8
37	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
36	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
35	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
34	Generalized metabolic bone disease and fracture risk in Rothmund-Thomson syndrome. *Human Molecular Genetics*, **2017**, 26, 3046-3055	5.6	6
33	Mobility in osteogenesis imperfecta: a multicenter North American study. *Genetics in Medicine*, **2019**, 21, 2311-2318	8.1	6
32	Differential utilization of systemic and enteral ammonia for urea synthesis in control subjects and ornithine transcarbamylase deficiency carriers. *American Journal of Clinical Nutrition*, **2003**, 78, 749-55	7	6
31	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
30	A global Slc7a7 knockout mouse model demonstrates characteristic phenotypes of human lysinuric protein intolerance. *Human Molecular Genetics*, **2020**, 29, 2171-2184	5.6	5
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