Brendan Lee

List of Publications by Year in Descending Order

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	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
200	Nitric oxide modulates bone anabolism through regulation of osteoblast glycolysis and differentiation. *Journal of Clinical Investigation*, **2021**, 131,	15.9	12
199	Osteogenesis imperfecta tooth level phenotype analysis: Cross-sectional study. *Bone*, **2021**, 147, 115917	4.7	1
198	Widespread disturbance in extracellular matrix collagen biomarker responses to teriparatide therapy in osteogenesis imperfecta. *Bone*, **2021**, 142, 115703	4.7	1
197	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
196	Transcriptome-directed analysis for Mendelian disease diagnosis overcomes limitations of conventional genomic testing. *Journal of Clinical Investigation*, **2021**, 131,	15.9	28
195	Health-related quality of life in adults with osteogenesis imperfecta. *Clinical Genetics*, **2021**, 99, 772-779	4	0
194	COPB2 loss of function causes a coatopathy with osteoporosis and developmental delay. *American Journal of Human Genetics*, **2021**, 108, 1710-1724	11	2
193	The crucial p53-dependent oncogenic role of JAB1 in osteosarcoma in vivo. *Oncogene*, **2020**, 39, 4581-4591	9.1	11
192	A global Slc7a7 knockout mouse model demonstrates characteristic phenotypes of human lysinuric protein intolerance. *Human Molecular Genetics*, **2020**, 29, 2171-2184	5.6	5
191	Loss- or Gain-of-Function Mutations in ACOX1 Cause Axonal Loss via Different Mechanisms. *Neuron*, **2020**, 106, 589-606.e6	13.9	32
190	Fracture Healing in Collagen-Related Preclinical Models of Osteogenesis Imperfecta. *Journal of Bone and Mineral Research*, **2020**, 35, 1132-1148	6.3	8
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	Skeletal disorders **2020**, 369-379		
186	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
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	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
182	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
181	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
180	Impact of Diagnosis and Therapy on Cognitive Function in Urea Cycle Disorders. *Annals of Neurology* , **2019**, 86, 116-128	9.4	20
179	Mobility in osteogenesis imperfecta: a multicenter North American study. *Genetics in Medicine*, **2019**, 21, 2311-2318	8.1	6
178	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
177	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
176	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
175	Osteogenesis imperfecta: advancements in genetics and treatment. *Current Opinion in Pediatrics*, **2019**, 31, 708-715	3.2	25
174	Oro-dental and cranio-facial characteristics of osteogenesis imperfecta type V. *European Journal of Medical Genetics*, **2019**, 62, 103606	2.6	3
173	Alterations in non-type I collagen biomarkers in osteogenesis imperfecta. *Bone*, **2019**, 120, 70-74	4.7	5
172	Autosomal recessive Noonan syndrome associated with biallelic LZTR1 variants. *Genetics in Medicine*, **2018**, 20, 1175-1185	8.1	80
171	Whole-Exome Sequencing Identifies an Intronic Cryptic Splice Site in Causing Osteogenesis Imperfecta Type VI. *JBMR Plus*, **2018**, 2, 235-239	3.9	2
170	TGF- Family Signaling in Mesenchymal Differentiation. *Cold Spring Harbor Perspectives in Biology*, **2018**, 10,	10.2	110
169	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
168	IRF2BPL Is Associated with Neurological Phenotypes. *American Journal of Human Genetics*, **2018**, 103, 245-260	11	39
167	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

166	Human Fetal and Neonatal Bone Development **2018**, 115-122		
165	Osteogenic Osteosarcoma **2018**, 768-774		
164	A multicenter study to evaluate pulmonary function in osteogenesis imperfecta. *Clinical Genetics*, **2018**, 94, 502-511	4	20
163	Genetic causes and mechanisms of Osteogenesis Imperfecta. *Bone*, **2017**, 102, 40-49	4.7	58
162	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	
161	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
160	Correlations Between Bone Mechanical Properties and Bone Composition Parameters in Mouse Models of Dominant and Recessive Osteogenesis Imperfecta and the Response to Anti-TGF-□ Treatment. *Journal of Bone and Mineral Research*, **2017**, 32, 347-359	6.3	21
159	Generalized metabolic bone disease and fracture risk in Rothmund-Thomson syndrome. *Human Molecular Genetics*, **2017**, 26, 3046-3055	5.6	6
158	Sclt1 deficiency causes cystic kidney by activating ERK and STAT3 signaling. *Human Molecular Genetics*, **2017**, 26, 2949-2960	5.6	19
157	Therapeutic Antibody Targeting Tumor- and Osteoblastic Niche-Derived Jagged1 Sensitizes Bone Metastasis to Chemotherapy. *Cancer Cell*, **2017**, 32, 731-747.e6	24.3	97
156	Clinically severe CACNA1A alleles affect synaptic function and neurodegeneration differentially. *PLoS Genetics*, **2017**, 13, e1006905	6	42
155	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
154	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
153	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
152	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
151	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
150	Losartan increases bone mass and accelerates chondrocyte hypertrophy in developing skeleton. *Molecular Genetics and Metabolism*, **2015**, 115, 53-60	3.7	14
149	RECQL4 Regulates p53 Function In Vivo During Skeletogenesis. *Journal of Bone and Mineral Research*, **2015**, 30, 1077-89	6.3	23

148	FBN1 contributing to familial congenital diaphragmatic hernia. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 831-6	2.5	19
147	Human recombinant arginase enzyme reduces plasma arginine in mouse models of arginase deficiency. *Human Molecular Genetics*, **2015**, 24, 6417-27	5.6	28
146	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
145	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
144	The osteogenic niche promotes early-stage bone colonization of disseminated breast cancer cells. *Cancer Cell*, **2015**, 27, 193-210	24.3	235
143	Connective tissue alterations in Fkbp10-/- mice. *Human Molecular Genetics*, **2014**, 23, 4822-31	5.6	39
142	Excessive transforming growth factor-signaling is a common mechanism in osteogenesis imperfecta. *Nature Medicine*, **2014**, 20, 670-5	50.5	172
141	Mice expressing mutant Trpv4 recapitulate the human TRPV4 disorders. *Journal of Bone and Mineral Research*, **2014**, 29, 1815-1822	6.3	15
140	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
139	Combinatorial treatment with oncolytic adenovirus and helper-dependent adenovirus augments adenoviral cancer gene therapy. *Molecular Therapy - Oncolytics*, **2014**, 1, 14008	6.4	17
138	Dual role of the Trps1 transcription factor in dentin mineralization. *Journal of Biological Chemistry*, **2014**, 289, 27481-93	5.4	17
137	Recessive Osteogenesis Imperfecta Due to Mutations in CRTAP, LEPRE1 and PPIB **2014**, 141-150		
136	Evaluation of teriparatide treatment in adults with osteogenesis imperfecta. *Journal of Clinical Investigation*, **2014**, 124, 491-8	15.9	107
135	Osteogenic Osteosarcoma **2013**, 702-710		4
134	Human Fetal and Neonatal Bone Development **2013**, 119-126		2
133	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
132	Prospects of Gene Therapy **2013**, 133-150		0
131	Ammonia control and neurocognitive outcome among urea cycle disorder patients treated with glycerol phenylbutyrate. *Hepatology*, **2013**, 57, 2171-9	11.2	61

130	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
129	Phenylbutyrate therapy for pyruvate dehydrogenase complex deficiency and lactic acidosis. *Science Translational Medicine*, **2013**, 5, 175ra31	17.5	46
128	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
127	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
126	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
125	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
124	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
123	Optimizing therapy for argininosuccinic aciduria. *Molecular Genetics and Metabolism*, **2012**, 107, 10-4	3.7	8
122	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
121	Cytokine-conditioned dendritic cells induce humoral tolerance to protein therapy in mice. *Human Gene Therapy*, **2012**, 23, 769-80	4.8	8
120	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
119	Nitric-oxide supplementation for treatment of long-term complications in argininosuccinic aciduria. *American Journal of Human Genetics*, **2012**, 90, 836-46	11	56
118	Argininosuccinate lyase deficiency. *Genetics in Medicine*, **2012**, 14, 501-7	8.1	59
117	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
116	Capsid-modified adenoviral vectors for improved muscle-directed gene therapy. *Human Gene Therapy*, **2012**, 23, 1065-70	4.8	21
115	Response to Srilatha et al.. *Genetics in Medicine*, **2012**, 14, 628-628	8.1	
114	Interaction of TGF□and BMP signaling pathways during chondrogenesis. *PLoS ONE*, **2011**, 6, e16421	3.7	97
113	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

112	The phenotype of recurrent 10q22q23 deletions and duplications. *European Journal of Human Genetics*, **2011**, 19, 400-8	5.3	52
111	One NOTCH Further: Jagged 1 in Bone Metastasis. *Cancer Cell*, **2011**, 19, 159-61	24.3	12
110	Insights into the pathogenesis and treatment of cancer from inborn errors of metabolism. *American Journal of Human Genetics*, **2011**, 88, 402-21	11	44
109	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
108	Mutations in FKBP10 cause recessive osteogenesis imperfecta and Bruck syndrome. *Journal of Bone and Mineral Research*, **2011**, 26, 666-72	6.3	123
107	Mutations in SERPINF1 cause osteogenesis imperfecta type VI. *Journal of Bone and Mineral Research*, **2011**, 26, 2798-803	6.3	141
106	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
105	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
104	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
103	Phenylbutyrate therapy for maple syrup urine disease. *Human Molecular Genetics*, **2011**, 20, 631-40	5.6	58
102	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
101	Requirement of argininosuccinate lyase for systemic nitric oxide production. *Nature Medicine*, **2011**, 17, 1619-26	50.5	161
100	Enteral arginase II provides ornithine for citrulline synthesis. *American Journal of Physiology - Endocrinology and Metabolism*, **2011**, 300, E188-94	6	14
99	Alteration of Notch signaling in skeletal development and disease. *Annals of the New York Academy of Sciences*, **2010**, 1192, 257-68	6.5	49
98	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
97	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
96	Glutamine: precursor or nitrogen donor for citrulline synthesis?. *American Journal of Physiology - Endocrinology and Metabolism*, **2010**, 299, E69-79	6	44
95	NOTCHing the bone: insights into multi-functionality. *Bone*, **2010**, 46, 274-80	4.7	65

94	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
93	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
92	Signaling pathways in human skeletal dysplasias. *Annual Review of Genomics and Human Genetics*, **2010**, 11, 189-217	9.7	49
91	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
90	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
89	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
88	Osteosclerosis owing to Notch gain of function is solely Rbpj-dependent. *Journal of Bone and Mineral Research*, **2010**, 25, 2175-83	6.3	54
87	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
86	Generalized connective tissue disease in Crtap-/- mouse. *PLoS ONE*, **2010**, 5, e10560	3.7	45
85	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
84	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
83	Notch signaling contributes to the pathogenesis of human osteosarcomas. *Human Molecular Genetics*, **2009**, 18, 1464-70	5.6	140
82	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
81	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
80	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
79	Nail-Patella Syndrome **2009**, 545-557		1
78	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
77	Dimorphic effects of Notch signaling in bone homeostasis. *Nature Medicine*, **2008**, 14, 299-305	50.5	322

76	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
75	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
74	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
73	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
72	Phenotypic correction of ornithine transcarbamylase deficiency using low dose helper-dependent adenoviral vectors. *Journal of Gene Medicine*, **2008**, 10, 890-6	3.5	19
71	CRTAP and LEPRE1 mutations in recessive osteogenesis imperfecta. *Human Mutation*, **2008**, 29, 1435-42	4.7	172
70	Brachy-syndactyly caused by loss of Sfrp2 function. *Journal of Cellular Physiology*, **2008**, 217, 127-37	7	50
69	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
68	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
67	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
66	Immune response to helper dependent adenoviral mediated liver gene therapy: challenges and prospects. *Current Gene Therapy*, **2007**, 7, 297-305	4.3	55
65	Toll-like receptor 9 triggers an innate immune response to helper-dependent adenoviral vectors. *Molecular Therapy*, **2007**, 15, 378-85	11.7	125
64	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
63	Dendritic cell function after gene transfer with adenovirus-calcium phosphate co-precipitates. *Molecular Therapy*, **2007**, 15, 386-92	11.7	19
62	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
61	RMRP mutations in cartilage-hair hypoplasia. *American Journal of Medical Genetics, Part A*, **2006**, 140, 2121-30	2.5	46
60	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
59	Deficiency of cartilage-associated protein in recessive lethal osteogenesis imperfecta. *New England Journal of Medicine*, **2006**, 355, 2757-64	59.2	255

58	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
57	CRTAP is required for prolyl 3- hydroxylation and mutations cause recessive osteogenesis imperfecta. *Cell*, **2006**, 127, 291-304	56.2	394
56	Reduced ornithine transcarbamylase activity does not impair ureagenesis in Otc(spf-ash) mice. *Journal of Nutrition*, **2006**, 136, 1017-20	4.1	10
55	In vivo urea kinetic studies in conscious mice. *Journal of Nutrition*, **2006**, 136, 202-6	4.1	12
54	Ornithine restores ureagenesis capacity and mitigates hyperammonemia in Otc(spf-ash) mice. *Journal of Nutrition*, **2006**, 136, 1834-8	4.1	11
53	Inborn errors of metabolism: the flux from Mendelian to complex diseases. *Nature Reviews Genetics* , **2006**, 7, 449-60	30.1	87
52	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
51	Dysregulation of chondrogenesis in human cleidocranial dysplasia. *American Journal of Human Genetics*, **2005**, 77, 305-12	11	40
50	Considerations in the difficult-to-manage urea cycle disorder patient. *Critical Care Clinics*, **2005**, 21, S19-25	4.5	11
49	Genetic counseling issues in urea cycle disorders. *Critical Care Clinics*, **2005**, 21, S37-44	4.5	16
48	Nutritional management of urea cycle disorders. *Critical Care Clinics*, **2005**, 21, S27-35	4.5	34
47	Unmasked adult-onset urea cycle disorders in the critical care setting. *Critical Care Clinics*, **2005**, 21, S1-8	4.5	96
46	Expression profiling of human fetal growth plate cartilage by EST sequencing. *Matrix Biology*, **2005**, 24, 530-8	11.4	11
45	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
44	Mutations and promoter SNPs in RUNX2, a transcriptional regulator of bone formation. *Molecular Genetics and Metabolism*, **2005**, 86, 257-68	3.7	44
43	Gene therapy for inborn errors of liver metabolism. *Molecular Genetics and Metabolism*, **2005**, 86, 13-24	3.7	15
42	Urea cycle disorders: clinical presentation outside the newborn period. *Critical Care Clinics*, **2005**, 21, S9-17	4.5	61
41	Consequences of mutations in the non-coding RMRP RNA in cartilage-hair hypoplasia. *Human Molecular Genetics*, **2005**, 14, 3723-40	5.6	84

40	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
39	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
38	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
37	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
36	Lmx1b expression during joint and tendon formation: localization and evaluation of potential downstream targets. *Gene Expression Patterns*, **2004**, 4, 397-405	1.5	23
35	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
34	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
33	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
32	Reduced inflammation and improved airway expression using helper-dependent adenoviral vectors with a K18 promoter. *Molecular Therapy*, **2003**, 7, 649-58	11.7	69
31	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
30	Urea Cycle Disorders. *Current Treatment Options in Neurology*, **2003**, 5, 309-319	4.4	23
29	Crane-Heise syndrome: a second familial case report with elaboration of phenotype. *American Journal of Medical Genetics Part A*, **2003**, 118A, 223-8		4
28	Determinants of vascular permeability in the kidney glomerulus. *Journal of Biological Chemistry*, **2002**, 277, 31154-62	5.4	92
27	Generation of helper-dependent adenoviral vectors by homologous recombination. *Molecular Therapy*, **2002**, 5, 204-10	11.7	46
26	An integrated approach to the diagnosis and prospective management of partial ornithine transcarbamylase deficiency. *Pediatrics*, **2002**, 109, 150-2	7.4	34
25	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
24	Urea-cycle disorders as a paradigm for inborn errors of hepatocyte metabolism. *Trends in Molecular Medicine*, **2002**, 8, 583-9	11.5	25
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