Brendan Lee

List of Publications by Year
in descending order

196 papers	15,499 citations	16451 64 h-index	19749 117 g-index
203 all docs	203 docs citations	203 times ranked	15615 citing authors

#	Article	IF	Citations
1	Linkage of Marfan syndrome and a phenotypically related disorder to two different fibrillin genes. Nature, 1991, 352, 330-334.	27.8	676
2	Missense mutations abolishing DNA binding of the osteoblast-specific transcription factor OSF2/CBFA1 in cleidocranial dysplasia. Nature Genetics, 1997, 16, 307-310.	21.4	548
3	Recurrent reciprocal 1q21.1 deletions and duplications associated with microcephaly or macrocephaly and developmental and behavioral abnormalities. Nature Genetics, 2008, 40, 1466-1471.	21.4	535
4	Limb and kidney defects in Lmx1b mutant mice suggest an involvement of LMX1B in human nail patella syndrome. Nature Genetics, 1998, 19, 51-55.	21.4	499
5	Mutations in LMX1B cause abnormal skeletal patterning and renal dysplasia in nail patella syndrome. Nature Genetics, 1998, 19, 47-50.	21.4	471
6	CRTAP Is Required for Prolyl 3- Hydroxylation and Mutations Cause Recessive Osteogenesis Imperfecta. Cell, 2006, 127, 291-304.	28.9	465
7	Cloning of the putative tumour suppressor gene for hereditary multiple exostoses (EXT1). Nature Genetics, 1995, 11, 137-143.	21.4	413
8	Dimorphic effects of Notch signaling in bone homeostasis. Nature Medicine, 2008, 14, 299-305.	30.7	361
9	Dominance of SOX9 function over RUNX2 during skeletogenesis. Proceedings of the National Academy of Sciences of the United States of America, 2006, 103, 19004-19009.	7.1	325
10	The Osteogenic Niche Promotes Early-Stage Bone Colonization of Disseminated Breast Cancer Cells. Cancer Cell, 2015, 27, 193-210.	16.8	308
11	Deficiency of Cartilage-Associated Protein in Recessive Lethal Osteogenesis Imperfecta. New England Journal of Medicine, 2006, 355, 2757-2764.	27.0	307
12	Type X collagen gene regulation by Runx2 contributes directly to its hypertrophic chondrocyteâ€“specific expression in vivo. Journal of Cell Biology, 2003, 162, 833-842.	5.2	287
13	Mutations in the Gene Encoding the RER Protein FKBP65 Cause Autosomal-Recessive Osteogenesis Imperfecta. American Journal of Human Genetics, 2010, 86, 551-559.	6.2	278
14	Genetic Linkage of the Marfan Syndrome, Ectopia Lentis, and Congenital Contractural Arachnodactyly to the Fibrillin Genes on Chromosomes 15 and 5. New England Journal of Medicine, 1992, 326, 905-909.	27.0	257
15	Excessive transforming growth factor-Î² signaling is a common mechanism in osteogenesis imperfecta. Nature Medicine, 2014, 20, 670-675.	30.7	237
16	CRTAPandLEPRE1mutations in recessive osteogenesis imperfecta. Human Mutation, 2008, 29, 1435-1442.	2.5	196
17	Regulation of glomerular basement membrane collagen expression by LMX1B contributes to renal disease in nail patella syndrome. Nature Genetics, 2001, 27, 205-208.	21.4	189
18	Cross-sectional multicenter study of patients with urea cycle disorders in the United States. Molecular Genetics and Metabolism, 2008, 94, 397-402.	1.1	189

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19	Requirement of argininosuccinate lyase for systemic nitric oxide production. Nature Medicine, 2011, 17, 1619-1626.	30.7	189
20	Diagnosis, symptoms, frequency and mortality of 260 patients with urea cycle disorders from a 21â€year, multicentre study of acute hyperammonaemic episodes. Acta Paediatrica, International Journal of Paediatrics, 2008, 97, 1420-1425.	1.5	188
21	The genome of self-complementary adeno-associated viral vectors increases Toll-like receptor 9â€“dependent innate immune responses in the liver. Blood, 2011, 117, 6459-6468.	1.4	187
22	A natural history of cleidocranial dysplasia. American Journal of Medical Genetics Part A, 2001, 104, 1-6.	2.4	179
23	TGF-Î² Family Signaling in Mesenchymal Differentiation. Cold Spring Harbor Perspectives in Biology, 2018, 10, a022202.	5.5	175
24	Use of a Liver-Specific Promoter Reduces Immune Response to the Transgene in Adenoviral Vectors. Human Gene Therapy, 1999, 10, 1773-1781.	2.7	174
25	Mutation Analysis of LMX1B Gene in Nail-Patella Syndrome Patients. American Journal of Human Genetics, 1998, 63, 1651-1658.	6.2	166
26	Mutations in SERPINF1 cause osteogenesis imperfecta type VI. Journal of Bone and Mineral Research, 2011, 26, 2798-2803.	2.8	164
27	Notch signaling contributes to the pathogenesis of human osteosarcomas. Human Molecular Genetics, 2009, 18, 1464-1470.	2.9	157
28	Mutations in FKBP10 cause recessive osteogenesis imperfecta and bruck syndrome. Journal of Bone and Mineral Research, 2011, 26, 666-672.	2.8	149
29	Long-Term Stable Correction of Low-Density Lipoprotein Receptorâ€“Deficient Mice With a Helper-Dependent Adenoviral Vector Expressing the Very Low-Density Lipoprotein Receptor. Circulation, 2001, 103, 1274-1281.	1.6	146
30	Evaluation of teriparatide treatment in adults with osteogenesis imperfecta. Journal of Clinical Investigation, 2014, 124, 491-498.	8.2	140
31	Therapeutic Antibody Targeting Tumor- and Osteoblastic Niche-Derived Jagged1 Sensitizes Bone Metastasis to Chemotherapy. Cancer Cell, 2017, 32, 731-747.e6.	16.8	133
32	Autosomal recessive Noonan syndrome associated with biallelic LZTR1 variants. Genetics in Medicine, 2018, 20, 1175-1185.	2.4	133
33	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, 3930-3935.	7.1	132
34	Toll-like Receptor 9 Triggers an Innate Immune Response to Helper-dependent Adenoviral Vectors. Molecular Therapy, 2007, 15, 378-385.	8.2	130
35	Transcriptional induction of slit diaphragm genes by Lmx1b is required in podocyte differentiation. Journal of Clinical Investigation, 2002, 109, 1065-1072.	8.2	122
36	Unmasked Adult-Onset Urea Cycle Disorders in the Critical Care Setting. Critical Care Clinics, 2005, 21, S1-S8.	2.6	120

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37	Interaction of TGFÎ² and BMP Signaling Pathways during Chondrogenesis. PLoS ONE, 2011, 6, e16421.	2.5	120
38	Toxicological Comparison of E2a-Deleted and First-Generation Adenoviral Vectors ExpressingÎ±₁-Antitrypsin after Systemic Delivery. Human Gene Therapy, 1998, 9, 1587-1598.	2.7	118
39	Inborn errors of metabolism: the flux from Mendelian to complex diseases. Nature Reviews Genetics, 2006, 7, 449-459.	16.3	113
40	Determinants of Vascular Permeability in the Kidney Glomerulus. Journal of Biological Chemistry, 2002, 277, 31154-31162.	3.4	108
41	Suppression of neuropil aggregates and neurological symptoms by an intracellular antibody implicates the cytoplasmic toxicity of mutant huntingtin. Journal of Cell Biology, 2008, 181, 803-816.	5.2	106
42	Consequences of mutations in the non-coding RMRP RNA in cartilage-hair hypoplasia. Human Molecular Genetics, 2005, 14, 3723-3740.	2.9	94
43	Effect of alternative pathway therapy on branched chain amino acid metabolism in urea cycle disorder patients. Molecular Genetics and Metabolism, 2004, 81, 79-85.	1.1	93
44	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, 113-120.	1.6	91
45	Uncoupling of chondrocyte differentiation and perichondrial mineralization underlies the skeletal dysplasia in tricho-rhino-phalangeal syndrome. Human Molecular Genetics, 2008, 17, 2244-2254.	2.9	90
46	Transcriptome-directed analysis for Mendelian disease diagnosis overcomes limitations of conventional genomic testing. Journal of Clinical Investigation, 2021, 131, .	8.2	87
47	Osteogenesis imperfecta: advancements in genetics and treatment. Current Opinion in Pediatrics, 2019, 31, 708-715.	2.0	84
48	Argininosuccinate lyase deficiency. Genetics in Medicine, 2012, 14, 501-507.	2.4	83
49	Ammonia control and neurocognitive outcome among urea cycle disorder patients treated with glycerol phenylbutyrate. Hepatology, 2013, 57, 2171-2179.	7.3	83
50	Genetic causes and mechanisms of Osteogenesis Imperfecta. Bone, 2017, 102, 40-49.	2.9	82
51	Transcriptional induction of slit diaphragm genes by Lmx1b is required in podocyte differentiation. Journal of Clinical Investigation, 2002, 109, 1065-1072.	8.2	82
52	Clinically severe CACNA1A alleles affect synaptic function and neurodegeneration differentially. PLoS Genetics, 2017, 13, e1006905.	3.5	80
53	Genome-wide oligonucleotide-based array comparative genome hybridization analysis of non-isolated congenital diaphragmatic hernia. Human Molecular Genetics, 2007, 16, 424-430.	2.9	79
54	Urea Cycle Disorders: Clinical Presentation Outside the Newborn Period. Critical Care Clinics, 2005, 21, S9-S17.	2.6	78

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55	Phase 2 comparison of a novel ammonia scavenging agent with sodium phenylbutyrate in patients with urea cycle disorders: Safety, pharmacokinetics and ammonia control. Molecular Genetics and Metabolism, 2010, 100, 221-228.	1.1	78
56	Argininosuccinate lyase deficiencyâ€"Argininosuccinic aciduria and beyond. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2011, 157, 45-53.	1.6	78
57	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, 4115-4125.	2.9	78
58	Phenylbutyrate therapy for maple syrup urine disease. Human Molecular Genetics, 2011, 20, 631-640.	2.9	77
59	Reduced inflammation and improved airway expression using Helper-Dependent adenoviral vectors with a k18 promoter. Molecular Therapy, 2003, 7, 649-658.	8.2	76
60	Clinical Consequences of Urea Cycle Enzyme Deficiencies and Potential Links to Arginine and Nitric Oxide Metabolism. Journal of Nutrition, 2004, 134, 2775S-2782S.	2.9	76
61	Establishing a consortium for the study of rare diseases: The Urea Cycle Disorders Consortium. Molecular Genetics and Metabolism, 2010, 100, S97-S105.	1.1	73
62	Nitric-Oxide Supplementation for Treatment of Long-Term Complications in Argininosuccinic Aciduria. American Journal of Human Genetics, 2012, 90, 836-846.	6.2	73
63	NOTCHing the bone: Insights into multi-functionality. Bone, 2010, 46, 274-280.	2.9	71
64	Loss- or Gain-of-Function Mutations in ACOX1 Cause Axonal Loss via Different Mechanisms. Neuron, 2020, 106, 589-606.e6.	8.1	71
65	Sclerostin Antibody Treatment Improves the Bone Phenotype of Crtapâ€"/â€" Mice, a Model of Recessive Osteogenesis Imperfecta. Journal of Bone and Mineral Research, 2016, 31, 1030-1040.	2.8	70
66	IRF2BPL Is Associated with Neurological Phenotypes. American Journal of Human Genetics, 2018, 103, 245-260.	6.2	69
67	Runx2 contributes to murine Col10a1 gene regulation through direct interaction with its cis-enhancer. Journal of Bone and Mineral Research, 2011, 26, 2899-2910.	2.8	67
68	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-e526.	2.1	65
69	Osteosclerosis owing to Notch gain of function is solely Rbpj-dependent. Journal of Bone and Mineral Research, 2010, 25, 2175-2183.	2.8	65
70	The phenotype of recurrent 10q22q23 deletions and duplications. European Journal of Human Genetics, 2011, 19, 400-408.	2.8	63
71	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, 302-314.	3.8	62
72	Long-term correction of urea cycle disorders. Journal of Pediatrics, 2001, 138, S62-S71.	1.8	61

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73	Brachyâ€“syndactyly caused by loss of Sfrp2 function. Journal of Cellular Physiology, 2008, 217, 127-137.	4.1	61
74	Notch gain of function inhibits chondrocyte differentiation via Rbpj-dependent suppression of Sox9. Journal of Bone and Mineral Research, 2013, 28, 649-659.	2.8	61
75	Immune Response to Helper Dependent Adenoviral Mediated Liver Gene Therapy: Challenges and Prospects. Current Gene Therapy, 2007, 7, 297-305.	2.0	60
76	Alteration of Notch signaling in skeletal development and disease. Annals of the New York Academy of Sciences, 2010, 1192, 257-268.	3.8	59
77	Phenylbutyrate Therapy for Pyruvate Dehydrogenase Complex Deficiency and Lactic Acidosis. Science Translational Medicine, 2013, 5, 175ra31.	12.4	59
78	Insights into the Pathogenesis and Treatment of Cancer from Inborn Errors of Metabolism. American Journal of Human Genetics, 2011, 88, 402-421.	6.2	58
79	Long-Term Correction of Ornithine Transcarbamylase Deficiency by WPRE-Mediated Overexpression Using a Helper-Dependent Adenovirus. Molecular Therapy, 2004, 10, 492-499.	8.2	57
80	Signaling Pathways in Human Skeletal Dysplasias. Annual Review of Genomics and Human Genetics, 2010, 11, 189-217.	6.2	55
81	Connective tissue alterations in Fkbp10â^'/â^' mice. Human Molecular Genetics, 2014, 23, 4822-4831.	2.9	54
82	Pharmacological and biological therapeutic strategies for osteogenesis imperfecta. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2016, 172, 367-383.	1.6	53
83	RMRPmutations in cartilage-hair hypoplasia. American Journal of Medical Genetics, Part A, 2006, 140A, 2121-2130.	1.2	52
84	Generalized Connective Tissue Disease in Crtap-/- Mouse. PLoS ONE, 2010, 5, e10560.	2.5	52
85	Glutamine: precursor or nitrogen donor for citrulline synthesis?. American Journal of Physiology - Endocrinology and Metabolism, 2010, 299, E69-E79.	3.5	51
86	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, 2082-2087.	7.1	51
87	Helper-dependent adenoviral vector-mediated long-term expression of human apolipoprotein A-I reduces atherosclerosis in apo E-deficient mice. Gene, 2004, 327, 153-160.	2.2	50
88	Mutations and promoter SNPs in RUNX2, a transcriptional regulator of bone formation. Molecular Genetics and Metabolism, 2005, 86, 257-268.	1.1	50
89	A Chaperone Complex Formed by HSP47, FKBP65, and BiP Modulates Telopeptide Lysyl Hydroxylation of Type I Procollagen. Journal of Bone and Mineral Research, 2017, 32, 1309-1319.	2.8	50
90	The fibrillin-marfan syndrome connection. BioEssays, 1993, 15, 589-594.	2.5	49

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91	Generation of Helper-Dependent Adenoviral Vectors by Homologous Recombination. Molecular Therapy, 2002, 5, 204-210.	8.2	49
92	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.	1.1	48
93	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, 17576-17581.	7.1	47
94	Dysregulation of Chondrogenesis in Human Cleidocranial Dysplasia. American Journal of Human Genetics, 2005, 77, 305-312.	6.2	45
95	Correction of Murine Hemophilia A and Immunological Differences of Factor VIII Variants Delivered by Helper-dependent Adenoviral Vectors. Molecular Therapy, 2007, 15, 2080-2087.	8.2	45
96	Ovotestes and XY sex reversal in a female with an interstitial 9q33.3â€q34.1 deletion encompassing NR5A1 and LMX1B causing features of genitopatellar syndrome. American Journal of Medical Genetics, Part A, 2007, 143A, 1071-1081.	1.2	43
97	Nutritional Management of Urea Cycle Disorders. Critical Care Clinics, 2005, 21, S27-S35.	2.6	42
98	Deficiency of FRAS1-related extracellular matrix 1 (FREM1) causes congenital diaphragmatic hernia in humans and mice. Human Molecular Genetics, 2013, 22, 1026-1038.	2.9	42
99	Impact of Diagnosis and Therapy on Cognitive Function in Urea Cycle Disorders. Annals of Neurology, 2019, 86, 116-128.	5.3	42
100	Systemic hypertension in two patients with ASL deficiency: A result of nitric oxide deficiency?. Molecular Genetics and Metabolism, 2009, 98, 195-197.	1.1	41
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, 1228-1234.e1.	1.8	40
102	Differential Type I Interferon-dependent Transgene Silencing of Helper-dependent Adenoviral vs. Adeno-associated Viral Vectors In Vivo. Molecular Therapy, 2013, 21, 796-805.	8.2	40
103	Human recombinant arginase enzyme reduces plasma arginine in mouse models of arginase deficiency. Human Molecular Genetics, 2015, 24, 6417-6427.	2.9	40
104	Nitric oxide modulates bone anabolism through regulation of osteoblast glycolysis and differentiation. Journal of Clinical Investigation, 2021, 131, .	8.2	39
105	Glycerol phenylbutyrate treatment in children with urea cycle disorders: Pooled analysis of short and long-term ammonia control and outcomes. Molecular Genetics and Metabolism, 2014, 112, 17-24.	1.1	38
106	An Integrated Approach to the Diagnosis and Prospective Management of Partial Ornithine Transcarbamylase Deficiency. Pediatrics, 2002, 109, 150-152.	2.1	37
107	Large-Scale Production of High-Quality Helper-Dependent Adenoviral Vectors Using Adherent Cells in Cell Factories. Human Gene Therapy, 2010, 21, 120-126.	2.7	35
108	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.	2.5	35

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109	Transatlantic combined and comparative data analysis of 1095 patients with urea cycle disordersâ€"A successful strategy for clinical research of rare diseases. Journal of Inherited Metabolic Disease, 2019, 42, 93-106.	3.6	35
110	Growth characteristics in individuals with osteogenesis imperfecta in North America: results from a multicenter study. Genetics in Medicine, 2019, 21, 275-283.	2.4	34
111	Urea-cycle disorders as a paradigm for inborn errors of hepatocyte metabolism. Trends in Molecular Medicine, 2002, 8, 583-589.	6.7	33
112	A multicenter study to evaluate pulmonary function in osteogenesis imperfecta. Clinical Genetics, 2018, 94, 502-511.	2.0	33
113	A randomized controlled trial to evaluate the effects of high-dose versus low-dose of arginine therapy on hepatic function tests in argininosuccinic aciduria. Molecular Genetics and Metabolism, 2012, 107, 315-321.	1.1	32
114	E-selectin ligand 1 regulates bone remodeling by limiting bioactive TGF-Î² in the bone microenvironment. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, 7336-7341.	7.1	32
115	Insight into Podocyte Differentiation from the Study of Human Genetic Disease: Nail-Patella Syndrome and Transcriptional Regulation in Podocytes. Pediatric Research, 2002, 51, 551-558.	2.3	31
116	MyD88-Dependent Silencing of Transgene Expression During the Innate and Adaptive Immune Response to Helper-Dependent Adenovirus. Human Gene Therapy, 2010, 21, 325-336.	2.7	31
117	Coordinated and unique functions of the E-selectin ligand ESL-1 during inflammatory and hematopoietic recruitment in mice. Blood, 2013, 122, 3993-4001.	1.4	31
118	A Transgenic Mouse Model of OI Type V Supports a Neomorphic Mechanism of the IFITM5 Mutation. Journal of Bone and Mineral Research, 2015, 30, 489-498.	2.8	30
119	RECQL4 Regulates p53 Function In Vivo During Skeletogenesis. Journal of Bone and Mineral Research, 2015, 30, 1077-1089.	2.8	30
120	Blood ammonia and glutamine as predictors of hyperammonemic crises in patients with urea cycle disorder. Genetics in Medicine, 2015, 17, 561-568.	2.4	30
121	Short-term Correction of Arginase Deficiency in a Neonatal Murine Model With a Helper-dependent Adenoviral Vector. Molecular Therapy, 2009, 17, 1155-1163.	8.2	29
122	Cloning, characterization, and chromosomal assignment of the human ortholog of murine Zfp-37, a candidate gene for Nager syndrome. Mammalian Genome, 1998, 9, 458-462.	2.2	28
123	Lmx1b expression during joint and tendon formation: localization and evaluation of potential downstream targets. Gene Expression Patterns, 2004, 4, 397-405.	0.8	28
124	Sclt1 deficiency causes cystic kidney by activating ERK and STAT3 signaling. Human Molecular Genetics, 2017, 26, 2949-2960.	2.9	28
125	The long and the short of it: developmental genetics of the skeletal dysplasias. Clinical Genetics, 1998, 54, 464-473.	2.0	27
126	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, 1022-1032.	2.8	27

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127	Dual Role of the Trps1 Transcription Factor in Dentin Mineralization. Journal of Biological Chemistry, 2014, 289, 27481-27493.	3.4	27
128	Barriers to drug adherence in the treatment of urea cycle disorders: Assessment of patient, caregiver and provider perspectives. Molecular Genetics and Metabolism Reports, 2016, 8, 43-47.	1.1	27
129	Targeting TGF-Î² for treatment of osteogenesis imperfecta. Journal of Clinical Investigation, 2022, 132, .	8.2	26
130	Capsid-Modified Adenoviral Vectors for Improved Muscle-Directed Gene Therapy. Human Gene Therapy, 2012, 23, 1065-1070.	2.7	25
131	Urea cycle disorders. Current Treatment Options in Neurology, 2003, 5, 309-319.	1.8	24
132	Toxicity and adaptive immune response to intracellular transgenes delivered by helper-dependent vs. first generation adenoviral vectors. Molecular Genetics and Metabolism, 2005, 84, 278-288.	1.1	24
133	FBN1 contributing to familial congenital diaphragmatic hernia. American Journal of Medical Genetics, Part A, 2015, 167, 831-836.	1.2	24
134	Correlations Between Bone Mechanical Properties and Bone Composition Parameters in Mouse Models of Dominant and Recessive Osteogenesis Imperfecta and the Response to Anti-TGF-Î² Treatment. Journal of Bone and Mineral Research, 2017, 32, 347-359.	2.8	24
135	E-selectin ligandâ€“1 regulates growth plate homeostasis in mice by inhibiting the intracellular processing and secretion of mature TGF-Î². Journal of Clinical Investigation, 2010, 120, 2474-2485.	8.2	24
136	Transcriptional dysregulation in skeletal malformation syndromes. American Journal of Medical Genetics Part A, 2001, 106, 258-271.	2.4	23
137	Phenotypic correction of ornithine transcarbamylase deficiency using low dose helperâ€dependent adenoviral vectors. Journal of Gene Medicine, 2008, 10, 890-896.	2.8	22
138	NOD2 Signaling Contributes to the Innate Immune Response Against Helper-Dependent Adenovirus Vectors Independently of MyD88 In Vivo. Human Gene Therapy, 2011, 22, 1071-1082.	2.7	22
139	A Multicenter Observational Cohort Study to Evaluate the Effects of Bisphosphonate Exposure on Bone Mineral Density and Other Health Outcomes in Osteogenesis Imperfecta. JBMR Plus, 2019, 3, e10118.	2.7	22
140	The crucial p53-dependent oncogenic role of JAB1 in osteosarcoma in vivo. Oncogene, 2020, 39, 4581-4591.	5.9	22
141	Dendritic Cell Function After Gene Transfer with Adenovirus-calcium Phosphate Co-precipitates. Molecular Therapy, 2007, 15, 386-392.	8.2	20
142	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, 119-122.	2.9	19
143	Genetic Counseling Issues in Urea Cycle Disorders. Critical Care Clinics, 2005, 21, S37-S44.	2.6	19
144	Combinatorial treatment with oncolytic adenovirus and helper-dependent adenovirus augments adenoviral cancer gene therapy. Molecular Therapy - Oncolytics, 2014, 1, 14008.	4.4	19

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145	Losartan increases bone mass and accelerates chondrocyte hypertrophy in developing skeleton. Molecular Genetics and Metabolism, 2015, 115, 53-60.	1.1	19
146	Oral health-related quality of life in children and adolescents with osteogenesis imperfecta: cross-sectional study. Orphanet Journal of Rare Diseases, 2018, 13, 187.	2.7	19
147	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, 161-168.	6.2	18
148	Plasma Arginine and Ornithine Are the Main Citrulline Precursors in Mice Infused with Arginine-Free Diets , ,. Journal of Nutrition, 2010, 140, 1432-1437.	2.9	18
149	Mice Expressing Mutant Trpv4 Recapitulate the Human TRPV4 Disorders. Journal of Bone and Mineral Research, 2014, 29, 1815-1822.	2.8	18
150	COPB2 loss of function causes a coatopathy with osteoporosis and developmental delay. American Journal of Human Genetics, 2021, 108, 1710-1724.	6.2	18
151	Gene therapy for inborn errors of liver metabolism. Molecular Genetics and Metabolism, 2005, 86, 13-24.	1.1	17
152	Enteral arginase II provides ornithine for citrulline synthesis. American Journal of Physiology - Endocrinology and Metabolism, 2011, 300, E188-E194.	3.5	16
153	Transcriptional repression of the Dspp gene leads to dentinogenesis imperfecta phenotype in Col1a1-Trps1 transgenic mice. Journal of Bone and Mineral Research, 2012, 27, 1735-1745.	2.8	16
154	Optimizing therapy for argininosuccinic aciduria. Molecular Genetics and Metabolism, 2012, 107, 10-14.	1.1	15
155	Impairment of cognitive function in ornithine transcarbamylase deficiency is global rather than domainâ€specific and is associated with disease onset, sex, maximum ammonium, and number of hyperammonemic events. Journal of Inherited Metabolic Disease, 2019, 42, 243-253.	3.6	15
156	Mobility in osteogenesis imperfecta: a multicenter North American study. Genetics in Medicine, 2019, 21, 2311-2318.	2.4	15
157	A global Slc7a7 knockout mouse model demonstrates characteristic phenotypes of human lysinuric protein intolerance. Human Molecular Genetics, 2020, 29, 2171-2184.	2.9	15
158	In Vivo Urea Kinetic Studies in Conscious Mice. Journal of Nutrition, 2006, 136, 202-206.	2.9	14
159	One NOTCH Further: Jagged 1 in Bone Metastasis. Cancer Cell, 2011, 19, 159-161.	16.8	14
160	Protein and calorie intakes in adult and pediatric subjects with urea cycle disorders participating in clinical trials of glycerol phenylbutyrate. Molecular Genetics and Metabolism Reports, 2016, 6, 34-40.	1.1	14
161	Pediatric Outcomes Data Collection Instrument is a Useful Patient-Reported Outcome Measure for Physical Function in Children with Osteogenesis Imperfecta. Genetics in Medicine, 2020, 22, 581-589.	2.4	14
162	Considerations in the Difficult-to-Manage Urea Cycle Disorder Patient. Critical Care Clinics, 2005, 21, S19-S25.	2.6	13

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163	Interaction between murinespf-ashmutation and genetic background yields different metabolic phenotypes. American Journal of Physiology - Endocrinology and Metabolism, 2007, 293, E1764-E1771.	3.5	13
164	De novo threeâ€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, 453-458.	1.2	13
165	Mutations in the Gene Encoding the RER Protein FKBP65 Cause Autosomal-Recessive Osteogenesis Imperfecta. American Journal of Human Genetics, 2010, 87, 572-573.	6.2	13
166	Generalized metabolic bone disease and fracture risk in Rothmund-Thomson syndrome. Human Molecular Genetics, 2017, 26, 3046-3055.	2.9	13
167	Fracture Healing in Collagenâ€Related Preclinical Models of Osteogenesis Imperfecta. Journal of Bone and Mineral Research, 2020, 35, 1132-1148.	2.8	13
168	Serum Sclerostin Levels in Adults With Osteogenesis Imperfecta: Comparison With Normal Individuals and Response to Teriparatide Therapy. Journal of Bone and Mineral Research, 2018, 33, 307-315.	2.8	12
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