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	What Has the Undiagnosed Diseases Network Taught Us About the Clinical Applications of Genomic Testing?. Annual Review of Medicine, 2022, 73, 575-585.	12.2	11
2	Targeting TGF-Î² for treatment of osteogenesis imperfecta. Journal of Clinical Investigation, 2022, 132, .	8.2	26
3	Widespread disturbance in extracellular matrix collagen biomarker responses to teriparatide therapy in osteogenesis imperfecta. Bone, 2021, 142, 115703.	2.9	4
4	The transcriptional cofactor Jab1/Cops5 is crucial for BMPâ€mediated mouse chondrocyte differentiation by repressing p53 activity. Journal of Cellular Physiology, 2021, 236, 5686-5697.	4.1	4
5	Transcriptome-directed analysis for Mendelian disease diagnosis overcomes limitations of conventional genomic testing. Journal of Clinical Investigation, 2021, 131, .	8.2	87
6	<scp>Healthâ€related</scp> quality of life in adults with osteogenesis imperfecta. Clinical Genetics, 2021, 99, 772-779.	2.0	4
7	Nitric oxide modulates bone anabolism through regulation of osteoblast glycolysis and differentiation. Journal of Clinical Investigation, 2021, 131, .	8.2	39
8	Osteogenesis imperfecta tooth level phenotype analysis: Cross-sectional study. Bone, 2021, 147, 115917.	2.9	7
9	COPB2 loss of function causes a coatopathy with osteoporosis and developmental delay. American Journal of Human Genetics, 2021, 108, 1710-1724.	6.2	18
10	Genetic Burden Contributing to Extremely Low or High Bone Mineral Density in a Senior Male Population From the Osteoporotic Fractures in Men Study (MrOS). JBMR Plus, 2020, 4, e10335.	2.7	1
11	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
12	Assessment of longitudinal bone growth in osteogenesis imperfecta using metacarpophalangeal pattern profiles. Bone, 2020, 140, 115547.	2.9	8
13	The crucial p53-dependent oncogenic role of JAB1 in osteosarcoma in vivo. Oncogene, 2020, 39, 4581-4591.	5.9	22
14	A global Slc7a7 knockout mouse model demonstrates characteristic phenotypes of human lysinuric protein intolerance. Human Molecular Genetics, 2020, 29, 2171-2184.	2.9	15
15	Loss- or Gain-of-Function Mutations in ACOX1 Cause Axonal Loss via Different Mechanisms. Neuron, 2020, 106, 589-606.e6.	8.1	71
16	Fracture Healing in Collagenâ€Related Preclinical Models of Osteogenesis Imperfecta. Journal of Bone and Mineral Research, 2020, 35, 1132-1148.	2.8	13
17	Chronic liver disease and impaired hepatic glycogen metabolism in argininosuccinate lyase deficiency. JCI Insight, 2020, 5, .	5.0	10
18	Skeletal disorders. , 2020, , 369-379.		0

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19	Targeted and sustained Sox9 expression in mouse hypertrophic chondrocytes causes severe and spontaneous osteoarthritis by perturbing cartilage homeostasis. American Journal of Translational Research (discontinued), 2020, 12, 1056-1069.	0.0	4
20	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
21	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
22	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
23	Trps1 Regulates Development of Craniofacial Skeleton and Is Required for the Initiation of Palatal Shelves Fusion. Frontiers in Physiology, 2019, 10, 513.	2.8	5
24	Impact of Diagnosis and Therapy on Cognitive Function in Urea Cycle Disorders. Annals of Neurology, 2019, 86, 116-128.	5.3	42
25	Mobility in osteogenesis imperfecta: a multicenter North American study. Genetics in Medicine, 2019, 21, 2311-2318.	2.4	15
26	Caries prevalence and experience in individuals with osteogenesis imperfecta: A crossâ€sectional multicenter study. Special Care in Dentistry, 2019, 39, 214-219.	0.8	11
27	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
28	Osteogenesis imperfecta: advancements in genetics and treatment. Current Opinion in Pediatrics, 2019, 31, 708-715.	2.0	84
29	Oro-dental and cranio-facial characteristics of osteogenesis imperfecta type V. European Journal of Medical Genetics, 2019, 62, 103606.	1.3	11
30	Alterations in non-type I collagen biomarkers in osteogenesis imperfecta. Bone, 2019, 120, 70-74.	2.9	11
31	Autosomal recessive Noonan syndrome associated with biallelic LZTR1 variants. Genetics in Medicine, 2018, 20, 1175-1185.	2.4	133
32	Wholeâ€Exome Sequencing Identifies an Intronic Cryptic Splice Site in SERPINF1 Causing Osteogenesis Imperfecta Type VI. JBMR Plus, 2018, 2, 235-239.	2.7	6
33	TGF-Î² Family Signaling in Mesenchymal Differentiation. Cold Spring Harbor Perspectives in Biology, 2018, 10, a022202.	5.5	175
34	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
35	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
36	A multicenter study to evaluate pulmonary function in osteogenesis imperfecta. Clinical Genetics, 2018, 94, 502-511.	2.0	33

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37	IRF2BPL Is Associated with Neurological Phenotypes. American Journal of Human Genetics, 2018, 103, 245-260.	6.2	69
38	Genetic causes and mechanisms of Osteogenesis Imperfecta. Bone, 2017, 102, 40-49.	2.9	82
39	2016 Curt Stern Award Address: From Rare to Common Diseases: Translating Genetic Discovery to Therapy 1. American Journal of Human Genetics, 2017, 100, 397-400.	6.2	0
40	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
41	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
42	Generalized metabolic bone disease and fracture risk in Rothmund-Thomson syndrome. Human Molecular Genetics, 2017, 26, 3046-3055.	2.9	13
43	Sclt1 deficiency causes cystic kidney by activating ERK and STAT3 signaling. Human Molecular Genetics, 2017, 26, 2949-2960.	2.9	28
44	Therapeutic Antibody Targeting Tumor- and Osteoblastic Niche-Derived Jagged1 Sensitizes Bone Metastasis to Chemotherapy. Cancer Cell, 2017, 32, 731-747.e6.	16.8	133
45	Clinically severe CACNA1A alleles affect synaptic function and neurodegeneration differentially. PLoS Genetics, 2017, 13, e1006905.	3.5	80
46	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
47	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
48	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
49	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
50	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
51	The Osteogenic Niche Promotes Early-Stage Bone Colonization of Disseminated Breast Cancer Cells. Cancer Cell, 2015, 27, 193-210.	16.8	308
52	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
53	Losartan increases bone mass and accelerates chondrocyte hypertrophy in developing skeleton. Molecular Genetics and Metabolism, 2015, 115, 53-60.	1.1	19
54	RECQL4 Regulates p53 Function In Vivo During Skeletogenesis. Journal of Bone and Mineral Research, 2015, 30, 1077-1089.	2.8	30

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55	FBN1 contributing to familial congenital diaphragmatic hernia. American Journal of Medical Genetics, Part A, 2015, 167, 831-836.	1.2	24
56	Human recombinant arginase enzyme reduces plasma arginine in mouse models of arginase deficiency. Human Molecular Genetics, 2015, 24, 6417-6427.	2.9	40
57	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
58	Dual Role of the Trps1 Transcription Factor in Dentin Mineralization. Journal of Biological Chemistry, 2014, 289, 27481-27493.	3.4	27
59	Recessive Osteogenesis Imperfecta Due to Mutations in CRTAP, LEPRE1 and PPIB. , 2014, , 141-150.		0
60	Connective tissue alterations in Fkbp10â^'/â^' mice. Human Molecular Genetics, 2014, 23, 4822-4831.	2.9	54
61	Excessive transforming growth factor-Î² signaling is a common mechanism in osteogenesis imperfecta. Nature Medicine, 2014, 20, 670-675.	30.7	237
62	Mice Expressing Mutant Trpv4 Recapitulate the Human TRPV4 Disorders. Journal of Bone and Mineral Research, 2014, 29, 1815-1822.	2.8	18
63	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
64	Combinatorial treatment with oncolytic adenovirus and helper-dependent adenovirus augments adenoviral cancer gene therapy. Molecular Therapy - Oncolytics, 2014, 1, 14008.	4.4	19
65	Evaluation of teriparatide treatment in adults with osteogenesis imperfecta. Journal of Clinical Investigation, 2014, 124, 491-498.	8.2	140
66	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
67	Prospects of Gene Therapy. , 2013, , 133-150.		1
68	Ammonia control and neurocognitive outcome among urea cycle disorder patients treated with glycerol phenylbutyrate. Hepatology, 2013, 57, 2171-2179.	7.3	83
69	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
70	Phenylbutyrate Therapy for Pyruvate Dehydrogenase Complex Deficiency and Lactic Acidosis. Science Translational Medicine, 2013, 5, 175ra31.	12.4	59
71	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
72	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

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73	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
74	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
75	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
76	Argininosuccinate lyase deficiency. Genetics in Medicine, 2012, 14, 501-507.	2.4	83
77	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
78	Capsid-Modified Adenoviral Vectors for Improved Muscle-Directed Gene Therapy. Human Gene Therapy, 2012, 23, 1065-1070.	2.7	25
79	Response to Srilatha et al.. Genetics in Medicine, 2012, 14, 627-628.	2.4	0
80	Optimizing therapy for argininosuccinic aciduria. Molecular Genetics and Metabolism, 2012, 107, 10-14.	1.1	15
81	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
82	Cytokine-Conditioned Dendritic Cells Induce Humoral Tolerance to Protein Therapy in Mice. Human Gene Therapy, 2012, 23, 769-780.	2.7	8
83	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
84	Nitric-Oxide Supplementation for Treatment of Long-Term Complications in Argininosuccinic Aciduria. American Journal of Human Genetics, 2012, 90, 836-846.	6.2	73
85	Interaction of TGFÎ² and BMP Signaling Pathways during Chondrogenesis. PLoS ONE, 2011, 6, e16421.	2.5	120
86	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
87	The phenotype of recurrent 10q22q23 deletions and duplications. European Journal of Human Genetics, 2011, 19, 400-408.	2.8	63
88	One NOTCH Further: Jagged 1 in Bone Metastasis. Cancer Cell, 2011, 19, 159-161.	16.8	14
89	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
90	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

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91	Mutations in FKBP10 cause recessive osteogenesis imperfecta and bruck syndrome. Journal of Bone and Mineral Research, 2011, 26, 666-672.	2.8	149
92	Mutations in SERPINF1 cause osteogenesis imperfecta type VI. Journal of Bone and Mineral Research, 2011, 26, 2798-2803.	2.8	164
93	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
94	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
95	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
96	Phenylbutyrate therapy for maple syrup urine disease. Human Molecular Genetics, 2011, 20, 631-640.	2.9	77
97	Phenylbutyrate improves nitrogen disposal via an alternative pathway without eliciting an increase in protein breakdown and catabolism in control and ornithine transcarbamylaseâ€“deficient patients. American Journal of Clinical Nutrition, 2011, 93, 1248-1254.	4.7	10
98	Requirement of argininosuccinate lyase for systemic nitric oxide production. Nature Medicine, 2011, 17, 1619-1626.	30.7	189
99	Enteral arginase II provides ornithine for citrulline synthesis. American Journal of Physiology - Endocrinology and Metabolism, 2011, 300, E188-E194.	3.5	16
100	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
101	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
102	Osteosclerosis owing to Notch gain of function is solely Rbpj-dependent. Journal of Bone and Mineral Research, 2010, 25, 2175-2183.	2.8	65
103	Alteration of Notch signaling in skeletal development and disease. Annals of the New York Academy of Sciences, 2010, 1192, 257-268.	3.8	59
104	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
105	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
106	Glutamine: precursor or nitrogen donor for citrulline synthesis?. American Journal of Physiology - Endocrinology and Metabolism, 2010, 299, E69-E79.	3.5	51
107	NOTCHing the bone: Insights into multi-functionality. Bone, 2010, 46, 274-280.	2.9	71
108	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

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109	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
110	Signaling Pathways in Human Skeletal Dysplasias. Annual Review of Genomics and Human Genetics, 2010, 11, 189-217.	6.2	55
111	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
112	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
113	Generalized Connective Tissue Disease in Crtap-/- Mouse. PLoS ONE, 2010, 5, e10560.	2.5	52
114	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
115	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
116	Notch signaling contributes to the pathogenesis of human osteosarcomas. Human Molecular Genetics, 2009, 18, 1464-1470.	2.9	157
117	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
118	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
119	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
120	Nail-Patella Syndrome. , 2009, , 545-557.		1
121	Phenotypic correction of ornithine transcarbamylase deficiency using low dose helperâ€dependent adenoviral vectors. Journal of Gene Medicine, 2008, 10, 890-896.	2.8	22
122	CRTAPandLEPRE1mutations in recessive osteogenesis imperfecta. Human Mutation, 2008, 29, 1435-1442.	2.5	196
123	Brachyâ€“syndactyly caused by loss of Sfrp2 function. Journal of Cellular Physiology, 2008, 217, 127-137.	4.1	61
124	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
125	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
126	Dimorphic effects of Notch signaling in bone homeostasis. Nature Medicine, 2008, 14, 299-305.	30.7	361

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127	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
128	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
129	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
130	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
131	Immune Response to Helper Dependent Adenoviral Mediated Liver Gene Therapy: Challenges and Prospects. Current Gene Therapy, 2007, 7, 297-305.	2.0	60
132	Toll-like Receptor 9 Triggers an Innate Immune Response to Helper-dependent Adenoviral Vectors. Molecular Therapy, 2007, 15, 378-385.	8.2	130
133	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
134	Dendritic Cell Function After Gene Transfer with Adenovirus-calcium Phosphate Co-precipitates. Molecular Therapy, 2007, 15, 386-392.	8.2	20
135	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
136	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
137	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
138	CRTAP Is Required for Prolyl 3- Hydroxylation and Mutations Cause Recessive Osteogenesis Imperfecta. Cell, 2006, 127, 291-304.	28.9	465
139	Reduced Ornithine Transcarbamylase Activity Does Not Impair Ureagenesis in Otc Mice. Journal of Nutrition, 2006, 136, 1017-1020.	2.9	10
140	In Vivo Urea Kinetic Studies in Conscious Mice. Journal of Nutrition, 2006, 136, 202-206.	2.9	14
141	Ornithine Restores Ureagenesis Capacity and Mitigates Hyperammonemia in Otc Mice. Journal of Nutrition, 2006, 136, 1834-1838.	2.9	11
142	Inborn errors of metabolism: the flux from Mendelian to complex diseases. Nature Reviews Genetics, 2006, 7, 449-459.	16.3	113
143	RMRPmutations in cartilage-hair hypoplasia. American Journal of Medical Genetics, Part A, 2006, 140A, 2121-2130.	1.2	52
144	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

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145	Deficiency of Cartilage-Associated Protein in Recessive Lethal Osteogenesis Imperfecta. New England Journal of Medicine, 2006, 355, 2757-2764.	27.0	307
146	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
147	Consequences of mutations in the non-coding RMRP RNA in cartilage-hair hypoplasia. Human Molecular Genetics, 2005, 14, 3723-3740.	2.9	94
148	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
149	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
150	Dysregulation of Chondrogenesis in Human Cleidocranial Dysplasia. American Journal of Human Genetics, 2005, 77, 305-312.	6.2	45
151	Considerations in the Difficult-to-Manage Urea Cycle Disorder Patient. Critical Care Clinics, 2005, 21, S19-S25.	2.6	13
152	Genetic Counseling Issues in Urea Cycle Disorders. Critical Care Clinics, 2005, 21, S37-S44.	2.6	19
153	Nutritional Management of Urea Cycle Disorders. Critical Care Clinics, 2005, 21, S27-S35.	2.6	42
154	Unmasked Adult-Onset Urea Cycle Disorders in the Critical Care Setting. Critical Care Clinics, 2005, 21, S1-S8.	2.6	120
155	Expression profiling of human fetal growth plate cartilage by EST sequencing. Matrix Biology, 2005, 24, 530-538.	3.6	11
156	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
157	Mutations and promoter SNPs in RUNX2, a transcriptional regulator of bone formation. Molecular Genetics and Metabolism, 2005, 86, 257-268.	1.1	50
158	Gene therapy for inborn errors of liver metabolism. Molecular Genetics and Metabolism, 2005, 86, 13-24.	1.1	17
159	Urea Cycle Disorders: Clinical Presentation Outside the Newborn Period. Critical Care Clinics, 2005, 21, S9-S17.	2.6	78
160	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
161	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
162	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

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163	Lmx1b expression during joint and tendon formation: localization and evaluation of potential downstream targets. Gene Expression Patterns, 2004, 4, 397-405.	0.8	28
164	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
165	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
166	Urea cycle disorders. Current Treatment Options in Neurology, 2003, 5, 309-319.	1.8	24
167	Crane-Heise syndrome: A second familial case report with elaboration of phenotype. American Journal of Medical Genetics Part A, 2003, 118A, 223-228.	2.4	7
168	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
169	Reduced inflammation and improved airway expression using Helper-Dependent adenoviral vectors with a k18 promoter. Molecular Therapy, 2003, 7, 649-658.	8.2	76
170	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, 749-755.	4.7	7
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