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List of Publications by Year
in descending order

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94 papers	14,520 citations	41258 49 h-index	49773 87 g-index
96 all docs	96 docs citations	96 times ranked	14929 citing authors

#	Article	IF	Citations
1	LDL Receptor-Related Protein 5 (LRP5) Affects Bone Accrual and Eye Development. Cell, 2001, 107, 513-523.	13.5	2,055
2	Preparation of PCR-Quality Mouse Genomic DNA with Hot Sodium Hydroxide and Tris (HotSHOT). BioTechniques, 2000, 29, 52-54.	0.8	1,290
3	Vascular Dysmorphogenesis Caused by an Activating Mutation in the Receptor Tyrosine Kinase TIE2. Cell, 1996, 87, 1181-1190.	13.5	734
4	Nosology and classification of genetic skeletal disorders: 2010 revision. American Journal of Medical Genetics, Part A, 2011, 155, 943-968.	0.7	573
5	Six Novel Missense Mutations in the LDL Receptor-Related Protein 5 (LRP5) Gene in Different Conditions with an Increased Bone Density. American Journal of Human Genetics, 2003, 72, 763-771.	2.6	522
6	The secreted glycoprotein lubricin protects cartilage surfaces and inhibits synovial cell overgrowth. Journal of Clinical Investigation, 2005, 115, 622-631.	3.9	461
7	Lrp5-independent activation of Wnt signaling by lithium chloride increases bone formation and bone mass in mice. Proceedings of the National Academy of Sciences of the United States of America, 2005, 102, 17406-17411.	3.3	455
8	Nosology and classification of genetic skeletal disorders: 2015 revision. American Journal of Medical Genetics, Part A, 2015, 167, 2869-2892.	0.7	453
9	Somatic Mosaic Activating Mutations in PIK3CA Cause CLOVES Syndrome. American Journal of Human Genetics, 2012, 90, 1108-1115.	2.6	447
10	Nosology and classification of genetic skeletal disorders: 2019 revision. American Journal of Medical Genetics, Part A, 2019, 179, 2393-2419.	0.7	431
11	Lymphatic and Other Vascular Malformative/Overgrowth Disorders AreÂCaused by Somatic Mutations in PIK3CA. Journal of Pediatrics, 2015, 166, 1048-1054.e5.	0.9	429
12	Lrp5 functions in bone to regulate bone mass. Nature Medicine, 2011, 17, 684-691.	15.2	404
13	The Wnt Co-receptor LRP5 Is Essential for Skeletal Mechanotransduction but Not for the Anabolic Bone Response to Parathyroid Hormone Treatment. Journal of Biological Chemistry, 2006, 281, 23698-23711.	1.6	364
14	Improving the DNA specificity and applicability of base editing through protein engineering and protein delivery. Nature Communications, 2017, 8, 15790.	5.8	343
15	Mutations in the Transmembrane Natriuretic Peptide Receptor NPR-B Impair Skeletal Growth and Cause Acromesomelic Dysplasia, Type Maroteaux. American Journal of Human Genetics, 2004, 75, 27-34.	2.6	325
16	Decreased BMD and Limb Deformities in Mice Carrying Mutations in Both Lrp5 and Lrp6. Journal of Bone and Mineral Research, 2004, 19, 2033-2040.	3.1	321
17	CACP, encoding a secreted proteoglycan, is mutated in camptodactyly-arthropathy-coxa vara-pericarditis syndrome. Nature Genetics, 1999, 23, 319-322.	9.4	286
18	Mutations in CDMP1 cause autosomal dominant brachydactyly type C. Nature Genetics, 1997, 17, 18-19.	9.4	255

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19	A type X collagen mutation causes Schmid metaphyseal chondrodysplasia. Nature Genetics, 1993, 5, 79-82.	9.4	253
20	Reduced Affinity to and Inhibition by DKK1 Form a Common Mechanism by Which High Bone Mass-Associated Missense Mutations in LRP5 Affect Canonical Wnt Signaling. Molecular and Cellular Biology, 2005, 25, 4946-4955.	1.1	244
21	Association between friction and wear in diarthrodial joints lacking lubricin. Arthritis and Rheumatism, 2007, 56, 3662-3669.	6.7	215
22	Somatic MAP2K1 Mutations Are Associated with Extracranial Arteriovenous Malformation. American Journal of Human Genetics, 2017, 100, 546-554.	2.6	215
23	A mutation in the gene encoding the Î±2 chain of the fibril-associated collagen IX, COL9A2, causes multiple epiphyseal dysplasia (EDM2). Nature Genetics, 1996, 12, 103-105.	9.4	206
24	Role of lubricin and boundary lubrication in the prevention of chondrocyte apoptosis. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, 5852-5857.	3.3	187
25	Somatic Activating Mutations in GNAQ and GNA11 Are Associated with Congenital Hemangioma. American Journal of Human Genetics, 2016, 98, 789-795.	2.6	144
26	The Binding Between Sclerostin and LRP5 is Altered by DKK1 and by High-Bone Mass LRP5 Mutations. Calcified Tissue International, 2008, 82, 445-453.	1.5	128
27	Loss of cartilage structure, stiffness, and frictional properties in mice lacking PRG4. Arthritis and Rheumatism, 2010, 62, 1666-1674.	6.7	125
28	Genetic mapping of cleidocranial dysplasia and evidence of a microdeletion in one family. Human Molecular Genetics, 1995, 4, 71-75.	1.4	114
29	FGFR3 promotes synchondrosis closure and fusion of ossification centers through the MAPK pathway. Human Molecular Genetics, 2008, 18, 227-240.	1.4	112
30	AKT hyper-phosphorylation associated with PI3K mutations in lymphatic endothelial cells from a patient with lymphatic malformation. Angiogenesis, 2015, 18, 151-162.	3.7	110
31	A Somatic MAP3K3 Mutation Is Associated with Verrucous Venous Malformation. American Journal of Human Genetics, 2015, 96, 480-486.	2.6	109
32	Loss-of-Function Mutations in PTPN11 Cause Metachondromatosis, but Not Ollier Disease or Maffucci Syndrome. PLoS Genetics, 2011, 7, e1002050.	1.5	104
33	A somatic GNA11 mutation is associated with extremity capillary malformation and overgrowth. Angiogenesis, 2017, 20, 303-306.	3.7	97
34	Bone Brittleness Varies with Genetic Background in A/J and C57BL/6J Inbred Mice. Journal of Bone and Mineral Research, 2001, 16, 1854-1862.	3.1	96
35	The camptodactyly-arthropathy-coxa vara-pericarditis syndrome: Clinical features and genetic mapping to human chromosome 1. Arthritis and Rheumatism, 1998, 41, 730-735.	6.7	92
36	Superficial cells are selfâ€renewing chondrocyte progenitors, which form the articular cartilage in juvenile mice. FASEB Journal, 2017, 31, 1067-1084.	0.2	92

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37	PIK3CA Activating Mutations in Facial Infiltrating Lipomatosis. Plastic and Reconstructive Surgery, 2014, 133, 12e-19e.	0.7	87
38	Endothelial Cells from Capillary Malformations Are Enriched for Somatic GNAQ Mutations. Plastic and Reconstructive Surgery, 2016, 137, 77e-82e.	0.7	87
39	Gain-of-function mutation of microRNA-140 in human skeletal dysplasia. Nature Medicine, 2019, 25, 583-590.	15.2	86
40	An osteocalcin-deficient mouse strain without endocrine abnormalities. PLoS Genetics, 2020, 16, e1008361.	1.5	81
41	Efficient Mapping and Cloning of Mutations in Zebrafish by Low-Coverage Whole-Genome Sequencing. Genetics, 2012, 190, 1017-1024.	1.2	77
42	Newly recognized autosomal dominant disorder with craniosynostosis. American Journal of Medical Genetics Part A, 1993, 46, 444-449.	2.4	76
43	Sclerostin Inhibition Reverses Skeletal Fragility in an Lrp5-Deficient Mouse Model of OPPG Syndrome. Science Translational Medicine, 2013, 5, 211ra158.	5.8	76
44	An RNA-seq protocol to identify mRNA expression changes in mouse diaphyseal bone: Applications in mice with bone property altering Lrp5 mutations. Journal of Bone and Mineral Research, 2013, 28, 2081-2093.	3.1	76
45	Targeting the LRP5 Pathway Improves Bone Properties in a Mouse Model of Osteogenesis Imperfecta. Journal of Bone and Mineral Research, 2014, 29, 2297-2306.	3.1	72
46	Induced superficial chondrocyte death reduces catabolic cartilage damage in murine posttraumatic osteoarthritis. Journal of Clinical Investigation, 2016, 126, 2893-2902.	3.9	72
47	A Normative Study of the Synovial Fluid Proteome from Healthy Porcine Knee Joints. Journal of Proteome Research, 2014, 13, 4377-4387.	1.8	68
48	High-bone-mass-producing mutations in the Wnt signaling pathway result in distinct skeletal phenotypes. Bone, 2011, 49, 1010-1019.	1.4	62
49	Tendon fascicle gliding in wild type, heterozygous, and lubricin knockout mice. Journal of Orthopaedic Research, 2011, 29, 384-389.	1.2	54
50	SHP2 Regulates Chondrocyte Terminal Differentiation, Growth Plate Architecture and Skeletal Cell Fates. PLoS Genetics, 2014, 10, e1004364.	1.5	52
51	Mechanotransduction in bone tissue: The A214V and G171V mutations in Lrp5 enhance load-induced osteogenesis in a surface-selective manner. Bone, 2012, 51, 459-465.	1.4	51
52	Somatic mutations in intracranial arteriovenous malformations. PLoS ONE, 2019, 14, e0226852.	1.1	51
53	Singleâ€Cell RNA Sequencing of Calvarial and Longâ€Bone Endocortical Cells. Journal of Bone and Mineral Research, 2020, 35, 1981-1991.	3.1	40
54	Anti-Lubricin Monoclonal Antibodies Created Using Lubricin-Knockout Mice Immunodetect Lubricin in Several Species and in Patients with Healthy and Diseased Joints. PLoS ONE, 2015, 10, e0116237.	1.1	36

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55	SHP2 regulates skeletal cell fate by modifying SOX9 expression and transcriptional activity. Bone Research, 2018, 6, 12.	5.4	33
56	Cyclic loading increases friction and changes cartilage surface integrity in lubricinâ€mutant mouse knees. Arthritis and Rheumatism, 2012, 64, 465-473.	6.7	32
57	Bone mineral properties in growing Col1a2+/G610C mice, an animal model of osteogenesis imperfecta. Bone, 2016, 87, 120-129.	1.4	29
58	Somatic PIK3CA mutations are present in multiple tissues of facial infiltrating lipomatosis. Pediatric Research, 2017, 82, 850-854.	1.1	28
59	SHP2 Regulates the Osteogenic Fate of Growth Plate Hypertrophic Chondrocytes. Scientific Reports, 2017, 7, 12699.	1.6	27
60	Reply to Lrp5 regulation of bone mass and gut serotonin synthesis. Nature Medicine, 2014, 20, 1229-1230.	15.2	26
61	Lubricin Restoration in a Mouse Model of Congenital Deficiency. Arthritis and Rheumatology, 2015, 67, 3070-3081.	2.9	26
62	Intramuscular fast-flow vascular anomaly contains somatic MAP2K1 and KRAS mutations. Angiogenesis, 2019, 22, 547-552.	3.7	26
63	Unique and non-redundant function of csf1r paralogues in regulation and evolution of post-embryonic development of the zebrafish. Development (Cambridge), 2020, 147, .	1.2	23
64	Mechanistic and therapeutic insights gained from studying rare skeletal diseases. Bone, 2015, 76, 67-75.	1.4	22
65	High Bone Massâ€“Causing Mutant LRP5 Receptors Are Resistant to Endogenous Inhibitors In Vivo. Journal of Bone and Mineral Research, 2015, 30, 1822-1830.	3.1	20
66	Causal somatic mutations in urine DNA from persons with the CLOVES subgroup of the PIK3CAâ€related overgrowth spectrum. Clinical Genetics, 2018, 93, 1075-1080.	1.0	20
67	Critical Endothelial Regulation by LRP5 during Retinal Vascular Development. PLoS ONE, 2016, 11, e0152833.	1.1	20
68	CRISPR/CAS9 Technologies. Journal of Bone and Mineral Research, 2017, 32, 883-888.	3.1	19
69	Arteriovenous malformation associated with a HRAS mutation. Human Genetics, 2019, 138, 1419-1421.	1.8	19
70	Enhanced Wnt signaling improves bone mass and strength, but not brittleness, in the Col1a1 +/mov13 mouse model of type I Osteogenesis Imperfecta. Bone, 2016, 90, 127-132.	1.4	18
71	The mechanical properties of tail tendon fascicles from lubricin knockout, wild type and heterozygous mice. Journal of Structural Biology, 2011, 176, 41-45.	1.3	16
72	The skeletal phenotype of Achondrogenesis type 1A is caused exclusively by cartilage defects. Development (Cambridge), 2018, 145, .	1.2	12

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73	Induction of Lrp5 HBM-causing mutations in Cathepsin-K expressing cells alters bone metabolism. Bone, 2019, 120, 166-175.	1.4	12
74	Asporin-deficient mice have tougher skin and altered skin glycosaminoglycan content and structure. PLoS ONE, 2017, 12, e0184028.	1.1	12
75	Proteolysis and cartilage development are activated in the synovium after surgical induction of post traumatic osteoarthritis. PLoS ONE, 2020, 15, e0229449.	1.1	11
76	Co-deletion of Lrp5 and Lrp6 in the skeleton severely diminishes bone gain from sclerostin antibody administration. Bone, 2021, 143, 115708.	1.4	11
77	Expression of a Degradation-Resistant Î²-Catenin Mutant in Osteocytes Protects the Skeleton From Mechanodeprivation-Induced Bone Wasting. Journal of Bone and Mineral Research, 2019, 34, 1964-1975.	3.1	10
78	Sensitive detection of Cre-mediated recombination using droplet digital PCR reveals Tg(BGLAP-Cre) and Tg(DMP1-Cre) are active in multiple non-skeletal tissues. Bone, 2021, 142, 115674.	1.4	10
79	Clcn7F318L/+ as a new mouse model of Albers-SchÃ¶nberg disease. Bone, 2017, 105, 253-261.	1.4	9
80	Facial Infiltrating Lipomatosis Contains Somatic PIK3CA Mutations in Multiple Tissues. Plastic and Reconstructive Surgery, 2015, 136, 72-73.	0.7	8
81	Combination therapy in the Col1a2G610C mouse model of Osteogenesis Imperfecta reveals an additive effect of enhancing LRP5 signaling and inhibiting TGFÎ² signaling on trabecular bone but not on cortical bone. Bone, 2020, 131, 115084.	1.4	6
82	Mice maintain predominantly maternal GÎ±s expression throughout life in brown fat tissue (BAT), but not other tissues. Bone, 2017, 103, 177-187.	1.4	5
83	Presphenoidal synchondrosis fusion in DBA/2J mice. Mammalian Genome, 2013, 24, 54-62.	1.0	3
84	Independent validation of experimental results requires timely and unrestricted access to animal models and reagents. PLoS Genetics, 2020, 16, e1008940.	1.5	3
85	RNAseq and RNA molecular barcoding reveal differential gene expression in cortical bone following hindlimb unloading in female mice. PLoS ONE, 2021, 16, e0250715.	1.1	3
86	Clinical and molecular studies of brachydactyly type D. , 1999, 85, 413-418.		2
87	Ocular Manifestations of Chordin-like 1 Knockout Mice. Cornea, 2020, 39, 1145-1150.	0.9	2
88	Cell depleted areas do not repopulate after diphtheria toxin-induced killing of mandibular cartilage chondrocytes. Osteoarthritis and Cartilage, 2021, 29, 1474-1484.	0.6	1
89	Essential Genes in the Development and Maintenance of the Temporomandibular Joint. FASEB Journal, 2013, 27, 319.5.	0.2	0
90	Transiently increased serotonin has modest or no effects on bone mass accrual in growing female C57BL6/J or growing male and female Lrp5A214V mice. Bone, 2022, 158, 116307.	1.4	0

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91	An osteocalcin-deficient mouse strain without endocrine abnormalities. , 2020, 16, e1008361.		0
92	An osteocalcin-deficient mouse strain without endocrine abnormalities. , 2020, 16, e1008361.		0
93	An osteocalcin-deficient mouse strain without endocrine abnormalities. , 2020, 16, e1008361.		0
94	An osteocalcin-deficient mouse strain without endocrine abnormalities. , 2020, 16, e1008361.		0