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List of Publications by Year
in descending order

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21 papers	2,292 citations	516710 16 h-index	713466 21 g-index
21 all docs	21 docs citations	21 times ranked	2230 citing authors

#	Article	IF	Citations
1	Consortium for osteogenesis imperfecta mutations in the helical domain of type I collagen: regions rich in lethal mutations align with collagen binding sites for integrins and proteoglycans. Human Mutation, 2007, 28, 209-221.	2.5	620
2	Prolyl 3-hydroxylase 1 deficiency causes a recessive metabolic bone disorder resembling lethal/severe osteogenesis imperfecta. Nature Genetics, 2007, 39, 359-365.	21.4	429
3	In vivo base editing rescues Hutchinsonâ€“Gilford progeria syndrome in mice. Nature, 2021, 589, 608-614.	27.8	275
4	Lack of Cyclophilin B in Osteogenesis Imperfecta with Normal Collagen Folding. New England Journal of Medicine, 2010, 362, 521-528.	27.0	158
5	Mutations Near Amino End of Î±1(I) Collagen Cause Combined Osteogenesis Imperfecta/Ehlers-Danlos Syndrome by Interference with N-propeptide Processing. Journal of Biological Chemistry, 2005, 280, 19259-19269.	3.4	118
6	Null mutations in LEPRE1 and CRTAP cause severe recessive osteogenesis imperfecta. Cell and Tissue Research, 2010, 339, 59-70.	2.9	108
7	Abnormal Type I Collagen Post-translational Modification and Crosslinking in a Cyclophilin B KO Mouse Model of Recessive Osteogenesis Imperfecta. PLoS Genetics, 2014, 10, e1004465.	3.5	98
8	Structural Heterogeneity of Type I Collagen Triple Helix and Its Role in Osteogenesis Imperfecta. Journal of Biological Chemistry, 2008, 283, 4787-4798.	3.4	81
9	Prolyl 3-hydroxylase 1 and CRTAP are mutually stabilizing in the endoplasmic reticulum collagen prolyl 3-hydroxylation complex. Human Molecular Genetics, 2010, 19, 223-234.	2.9	73
10	Somatic activating mutations in MAP2K1 cause melorheostosis. Nature Communications, 2018, 9, 1390.	12.8	56
11	A targeted antisense therapeutic approach for Hutchinsonâ€“Gilford progeria syndrome. Nature Medicine, 2021, 27, 536-545.	30.7	55
12	Cyclophilin-B Modulates Collagen Cross-linking by Differentially Affecting Lysine Hydroxylation in the Helical and Telopeptidyl Domains of Tendon Type I Collagen. Journal of Biological Chemistry, 2016, 291, 9501-9512.	3.4	46
13	Type I Collagen Triplet Duplication Mutation in Lethal Osteogenesis Imperfecta Shifts Register of Î± Chains throughout the Helix and Disrupts Incorporation of Mutant Helices into Fibrils and Extracellular Matrix. Journal of Biological Chemistry, 2003, 278, 10006-10012.	3.4	29
14	Genetic reduction of mTOR extends lifespan in a mouse model of Hutchinsonâ€Gilford Progeria syndrome. Aging Cell, 2021, 20, e13457.	6.7	27
15	Somatic SMAD3-activating mutations cause melorheostosis by up-regulating the TGF-Î²/SMAD pathway. Journal of Experimental Medicine, 2020, 217, .	8.5	24
16	Evaluation of musculoskeletal phenotype of the G608G progeria mouse model with lonafarnib, pravastatin, and zoledronic acid as treatment groups. Proceedings of the National Academy of Sciences of the United States of America, 2020, 117, 12029-12040.	7.1	20
17	Cyclophilin B control of lysine post-translational modifications of skin type I collagen. PLoS Genetics, 2019, 15, e1008196.	3.5	18
18	COL1A1 C-propeptide mutations cause ER mislocalization of procollagen and impair C-terminal procollagen processing. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2019, 1865, 2210-2223.	3.8	18

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19	Melorheostotic Bone Lesions Caused by Somatic Mutations in MAP2K1 Have Deteriorated Microarchitecture and Periosteal Reaction. Journal of Bone and Mineral Research, 2019, 34, 883-895.	2.8	16
20	Cyclophilin B Deficiency Causes Abnormal Dentin Collagen Matrix. Journal of Proteome Research, 2017, 16, 2914-2923.	3.7	12
21	Substitution of murine type I collagen A1 3-hydroxylation site alters matrix structure but does not recapitulate osteogenesis imperfecta bone dysplasia. Matrix Biology, 2020, 90, 20-39.	3.6	11