Joan C Marini

List of Publications by Year
in descending order

101 papers	8,216 citations	53939 47 h-index	54771 88 g-index
109 all docs	109 docs citations	109 times ranked	5765 citing authors

#	Article	IF	Citations
1	Osteogenesis Imperfecta: Mechanisms and Signaling Pathways Connecting Classical and Rare OI Types. Endocrine Reviews, 2022, 43, 61-90.	8.9	58
2	Osteogenesis Imperfecta: The Impact of Genotype and Clinical Phenotype on Adiposity and Resting Energy Expenditure. Journal of Clinical Endocrinology and Metabolism, 2022, 107, 67-76.	1.8	5
3	Dissecting the phenotypic variability of osteogenesis imperfecta. DMM Disease Models and Mechanisms, 2022, 15, .	1.2	16
4	Alterations of bone material properties in growing Ifitm5/BRIL p.S42 knock-in mice, a new model for atypical type VI osteogenesis imperfecta. Bone, 2022, 162, 116451.	1.4	8
5	SMAD3 mutation in LDS3 causes bone fragility by impairing the TGF-Î² pathway and enhancing osteoclastogenesis. Bone Reports, 2022, 17, 101603.	0.2	3
6	Stress Shielding in the Setting of Osteogenesis Imperfecta and the Effect of Downsizing an Intramedullary Rod. JBJS Case Connector, 2021, 11, .	0.1	0
7	Distribution and Functional Consequences of Somatic MAP2K1 Variants in Affected Skin Associated with Bone Lesions in Melorheostosis. Journal of Investigative Dermatology, 2021, 141, 688-692.e11.	0.3	3
8	Targeting cellular stress in vitro improves osteoblast homeostasis, matrix collagen content and mineralization in two murine models of osteogenesis imperfecta. Matrix Biology, 2021, 98, 1-20.	1.5	19
9	Bruck syndrome 2 variant lacking congenital contractures and involving a novel compound heterozygous PLOD2 mutation. Bone, 2020, 130, 115047.	1.4	14
10	Somatic SMAD3-activating mutations cause melorheostosis by up-regulating the TGF-Î²/SMAD pathway. Journal of Experimental Medicine, 2020, 217, .	4.2	24
11	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.	1.5	11
12	Antagonism Between PEDF and TGF-Î² Contributes to Type VI Osteogenesis Imperfecta Bone and Vascular Pathogenesis. Journal of Bone and Mineral Research, 2020, 37, 925-937.	3.1	7
13	Distinct Clinical and Pathological Features of Melorheostosis Associated With Somatic MAP2K1 Mutations. Journal of Bone and Mineral Research, 2019, 34, 145-156.	3.1	22
14	Sclerostin Antibodyâ€“Induced Changes in Bone Mass Are Site Specific in Developing Crania. Journal of Bone and Mineral Research, 2019, 34, 2301-2310.	3.1	9
15	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.	3.1	16
16	Bone biology: insights from osteogenesis imperfecta and related rare fragility syndromes. FEBS Journal, 2019, 286, 3033-3056.	2.2	35
17	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.	1.8	18
18	Longitudinal growth curves for children with classical osteogenesis imperfecta (types III and IV) caused by structural pathogenic variants in type I collagen. Genetics in Medicine, 2019, 21, 1233-1239.	1.1	22

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19	4-PBA ameliorates cellular homeostasis in fibroblasts from osteogenesis imperfecta patients by enhancing autophagy and stimulating protein secretion. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2018, 1864, 1642-1652.	1.8	55
20	Somatic activating mutations in MAP2K1 cause melorheostosis. Nature Communications, 2018, 9, 1390.	5.8	56
21	Low Dose of Bisphosphonate Enhances Sclerostin Antibody-Induced Trabecular Bone Mass Gains in Brtl/+ Osteogenesis Imperfecta Mouse Model. Journal of Bone and Mineral Research, 2018, 33, 1272-1282.	3.1	13
22	Phenotypic Spectrum in Osteogenesis Imperfecta Due to Mutations in TMEM38B: Unraveling a Complex Cellular Defect. Journal of Clinical Endocrinology and Metabolism, 2017, 102, 2019-2028.	1.8	27
23	Osteogenesis imperfecta: new genes reveal novel mechanisms in bone dysplasia. Translational Research, 2017, 181, 27-48.	2.2	78
24	Hypermineralization and High Osteocyte Lacunar Density in Osteogenesis Imperfecta Type V Bone Indicate Exuberant Primary Bone Formation. Journal of Bone and Mineral Research, 2017, 32, 1884-1892.	3.1	55
25	Osteogenesis imperfecta. Nature Reviews Disease Primers, 2017, 3, 17052.	18.1	481
26	Cyclophilin B Deficiency Causes Abnormal Dentin Collagen Matrix. Journal of Proteome Research, 2017, 16, 2914-2923.	1.8	12
27	Absence of the ER Cation Channel TMEM38B/TRIC-B Disrupts Intracellular Calcium Homeostasis and Dysregulates Collagen Synthesis in Recessive Osteogenesis Imperfecta. PLoS Genetics, 2016, 12, e1006156.	1.5	49
28	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.	1.6	46
29	Single dose of bisphosphonate preserves gains in bone mass following cessation of sclerostin antibody in Brtl/+ osteogenesis imperfecta model. Bone, 2016, 93, 79-85.	1.4	24
30	Non-Lethal Type VIII Osteogenesis Imperfecta Has Elevated Bone Matrix Mineralization. Journal of Clinical Endocrinology and Metabolism, 2016, 101, 3516-3525.	1.8	28
31	MBTPS2 mutations cause defective regulated intramembrane proteolysis in X-linked osteogenesis imperfecta. Nature Communications, 2016, 7, 11920.	5.8	112
32	Bone mineral properties in growing Col1a2+/G610C mice, an animal model of osteogenesis imperfecta. Bone, 2016, 87, 120-129.	1.4	29
33	Effect of anti-sclerostin therapy and osteogenesis imperfecta on tissue-level properties in growing and adult mice while controlling for tissue age. Bone, 2016, 84, 222-229.	1.4	33
34	Osteogenesis imperfecta. Lancet, The, 2016, 387, 1657-1671.	6.3	668
35	Tissue level material composition and mechanical properties in Brtl/+ mouse model of Osteogenesis Imperfecta after sclerostin antibody treatment. , 2015, , .		0
36	Type V OI Primary Osteoblasts Display Increased Mineralization Despite DecreasedCOL1A1Expression. Journal of Clinical Endocrinology and Metabolism, 2015, 100, E325-E332.	1.8	32

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37	Genetic Defects in TAPT1 Disrupt Ciliogenesis and Cause a Complex Lethal Osteochondrodysplasia. American Journal of Human Genetics, 2015, 97, 521-534.	2.6	39
38	Altered cytoskeletal organization characterized lethal but not surviving Brtl^{+/â˜}mice: insight on phenotypic variability in osteogenesis imperfecta. Human Molecular Genetics, 2015, 24, 6118-6133.	1.4	29
39	Rapidly growing Brtl/+ mouse model of osteogenesis imperfecta improves bone mass and strength with sclerostin antibody treatment. Bone, 2015, 71, 115-123.	1.4	71
40	Heritable connective tissue disorders. , 2015, , 1715-1723.		0
41	Allele-specific Col1a1 silencing reduces mutant collagen in fibroblasts from Brtl mouse, a model for classical osteogenesis imperfecta. European Journal of Human Genetics, 2014, 22, 667-674.	1.4	21
42	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.	1.5	98
43	A Novel IFITM5 Mutation in Severe Atypical Osteogenesis Imperfecta Type VI Impairs Osteoblast Production of Pigment Epithelium-Derived Factor. Journal of Bone and Mineral Research, 2014, 29, 1402-1411.	3.1	63
44	Osteogenesis imperfecta due to mutations in non-collagenous genes. Current Opinion in Pediatrics, 2014, 26, 500-507.	1.0	115
45	Sclerostin antibody improves skeletal parameters in a Brtl/+ mouse model of osteogenesis imperfecta. Journal of Bone and Mineral Research, 2013, 28, 73-80.	3.1	111
46	Mutations in WNT1 Cause Different Forms of Bone Fragility. American Journal of Human Genetics, 2013, 92, 565-574.	2.6	240
47	New Genes in Bone Development: What's New in Osteogenesis Imperfecta. Journal of Clinical Endocrinology and Metabolism, 2013, 98, 3095-3103.	1.8	163
48	Mineral and Matrix Changes in Brtl/+Teeth Provide Insights into Mineralization Mechanisms. BioMed Research International, 2013, 2013, 1-9.	0.9	14
49	Kuskokwim Syndrome, a Recessive Congenital Contracture Disorder, Extends the Phenotype ofFKBP10â€‰ Mutations. Human Mutation, 2013, 34, 1279-1288.	1.1	53
50	A founder mutation in LEPRE1 carried by 1.5% of West Africans and 0.4% of African Americans causes lethal recessive osteogenesis imperfecta. Genetics in Medicine, 2012, 14, 543-551.	1.1	49
51	Cardiopulmonary dysfunction in the Osteogenesis imperfecta mouse model Aga2 and human patients are caused by bone-independent mechanisms. Human Molecular Genetics, 2012, 21, 3535-3545.	1.4	57
52	Effects of tissue hydration on nanoscale structural morphology and mechanics of individual Type I collagen fibrils in the Brtl mouse model of Osteogenesis Imperfecta. Journal of Structural Biology, 2012, 180, 428-438.	1.3	45
53	Differential response to intracellular stress in the skin from osteogenesis imperfecta Brtl mice with lethal and non lethal phenotype: A proteomic approach. Journal of Proteomics, 2012, 75, 4717-4733.	1.2	19
54	Increased susceptibility to microdamage in Brtl/+ mouse model for osteogenesis imperfecta. Bone, 2012, 50, 784-791.	1.4	16

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55	Impaired Osteoblastogenesis in a Murine Model of Dominant Osteogenesis Imperfecta: A New Target for Osteogenesis Imperfecta Pharmacological Therapy. Stem Cells, 2012, 30, 1465-1476.	1.4	59
56	Absence ofFKBP10in recessive type XI osteogenesis imperfecta leads to diminished collagen cross-linking and reduced collagen deposition in extracellular matrix. Human Mutation, 2012, 33, 1589-1598.	1.1	86
57	A Novel Mutation in LEPRE1 That Eliminates Only the KDEL ER- Retrieval Sequence Causes Non-Lethal Osteogenesis Imperfecta. PLoS ONE, 2012, 7, e36809.	1.1	28
58	New perspectives on osteogenesis imperfecta. Nature Reviews Endocrinology, 2011, 7, 540-557.	4.3	556
59	Nanoscale morphology of Type I collagen is altered in the Brtl mouse model of Osteogenesis Imperfecta. Journal of Structural Biology, 2011, 173, 146-152.	1.3	74
60	COL1 C-propeptide cleavage site mutations cause high bone mass osteogenesis imperfecta. Human Mutation, 2011, 32, 598-609.	1.1	119
61	Variable bone fragility associated with an Amish COL1A2 variant and a knock-in mouse model. Journal of Bone and Mineral Research, 2010, 25, 247-261.	3.1	98
62	Null mutations in LEPRE1 and CRTAP cause severe recessive osteogenesis imperfecta. Cell and Tissue Research, 2010, 339, 59-70.	1.5	108
63	Near-infrared fluorescent probe traces bisphosphonate delivery and retention in vivo. Journal of Bone and Mineral Research, 2010, 25, 1748-1758.	3.1	72
64	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.	1.4	73
65	Lack of Cyclophilin B in Osteogenesis Imperfecta with Normal Collagen Folding. New England Journal of Medicine, 2010, 362, 521-528.	13.9	158
66	Use of bisphosphonates in childrenâ€"proceed with caution. Nature Reviews Endocrinology, 2009, 5, 241-243.	4.3	36
67	Alendronate Treatment of the Brtl Osteogenesis Imperfecta Mouse Improves Femoral Geometry and Load Response Before Fracture but Decreases Predicted Material Properties and Has Detrimental Effects on Osteoblasts and Bone Formation. Journal of Bone and Mineral Research, 2009, 24, 849-859.	3.1	57
68	In utero transplantation of adult bone marrow decreases perinatal lethality and rescues the bone phenotype in the knockin murine model for classical, dominant osteogenesis imperfecta. Blood, 2009, 114, 459-468.	0.6	93
69	Popcorn calcification in osteogenesis imperfecta: Incidence, progression, and molecular correlation. American Journal of Medical Genetics, Part A, 2008, 146A, 2725-2732.	0.7	36
70	Cellular Mechanism of Decreased Bone in Brtl Mouse Model of OI: Imbalance of Decreased Osteoblast Function and Increased Osteoclasts and Their Precursors. Journal of Bone and Mineral Research, 2008, 23, 1983-1994.	3.1	75
71	Candidate Cell and Matrix Interaction Domains on the Collagen Fibril, the Predominant Protein of Vertebrates. Journal of Biological Chemistry, 2008, 283, 21187-21197.	1.6	244
72	Structural Heterogeneity of Type I Collagen Triple Helix and Its Role in Osteogenesis Imperfecta. Journal of Biological Chemistry, 2008, 283, 4787-4798.	1.6	81

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73	Mapping of SPARC/BM-40/Osteonectin-binding Sites on Fibrillar Collagens. Journal of Biological Chemistry, 2008, 283, 19551-19560.	1.6	87
74	Components of the Collagen Prolyl 3-Hydroxylation Complex are Crucial for Normal Bone Development. Cell Cycle, 2007, 6, 1675-1681.	1.3	107
75	Selective retention and degradation of molecules with a single mutant Î±1(I) chain in the Brtl IV mouse model of OI. Matrix Biology, 2007, 26, 604-614.	1.5	52
76	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.	1.1	620
77	Differential expression of both extracellular and intracellular proteins is involved in the lethal or nonlethal phenotypic variation of BrtlIV, a murine model for osteogenesis imperfecta. Proteomics, 2007, 7, 1877-1891.	1.3	51
78	Prolyl 3-hydroxylase 1 deficiency causes a recessive metabolic bone disorder resembling lethal/severe osteogenesis imperfecta. Nature Genetics, 2007, 39, 359-365.	9.4	429
79	Y-position cysteine substitution in type I collagen (Î±1(I) R888C/p.R1066C) is associated with osteogenesis imperfecta/Ehlers-Danlos syndrome phenotype. Human Mutation, 2007, 28, 396-405.	1.1	63
80	Should children with osteogenesis imperfecta be treated with bisphosphonates?. Nature Clinical Practice Endocrinology and Metabolism, 2006, 2, 14-15.	2.9	11
81	Deficiency of Cartilage-Associated Protein in Recessive Lethal Osteogenesis Imperfecta. New England Journal of Medicine, 2006, 355, 2757-2764.	13.9	307
82	Molecular Mechanism of Î±1(I)-Osteogenesis Imperfecta/Ehlers-Danlos Syndrome. Journal of Biological Chemistry, 2006, 281, 6463-6470.	1.6	77
83	Controlled Trial of Pamidronate in Children With Types III and IV Osteogenesis Imperfecta Confirms Vertebral Gains but Not Short-Term Functional Improvement. Journal of Bone and Mineral Research, 2005, 20, 977-986.	3.1	176
84	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.	1.6	118
85	Brittle IV Mouse Model for Osteogenesis Imperfecta IV Demonstrates Postpubertal Adaptations to Improve Whole Bone Strength. Journal of Bone and Mineral Research, 2004, 19, 614-622.	3.1	118
86	Structure, stability and interactions of type I collagen with GLY349-CYS substitution in Î±1(I) chain in a murine Osteogenesis Imperfecta model. Matrix Biology, 2004, 23, 101-112.	1.5	32
87	Positive Linear Growth and Bone Responses to Growth Hormone Treatment in Children With Types III and IV Osteogenesis Imperfecta: High Predictive Value of the Carboxyterminal Propeptide of Type I Procollagen. Journal of Bone and Mineral Research, 2003, 18, 237-243.	3.1	93
88	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.	1.6	29
89	Do Bisphosphonates Make Children's Bones Better or Brittle?. New England Journal of Medicine, 2003, 349, 423-426.	13.9	150
90	Osteogenesis Imperfecta: Prospects for Molecular Therapeutics. Molecular Genetics and Metabolism, 2000, 71, 225-232.	0.5	58

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91	Use of the Cre/lox Recombination System to Develop a Non-lethal Knock-in Murine Model for Osteogenesis Imperfecta with an Î±1(I) G349C Substitution. Journal of Biological Chemistry, 1999, 274, 37923-37931.	1.6	125
92	An Î±2(I) glycine to aspartate substitution is responsible for the presence of a kink in type I collagen in a lethal case of osteogenesis imperfecta. Matrix Biology, 1998, 17, 575-584.	1.5	20
93	Phenotypic Comparison of an Osteogenesis Imperfecta Type IV Proband with ade NovoÎ±2(I) Gly922 â†' Ser Substitution in Type I Collagen and an Unrelated Patient with an Identical Mutation. Biochemical and Molecular Medicine, 1997, 62, 26-35.	1.5	15
94	Endocrine Aspects of Growth Deficiency in OI. Connective Tissue Research, 1995, 31, s55-s57.	1.1	8
95	Neurologic Profile in Osteogenesis Imperfecta. Connective Tissue Research, 1995, 31, s23-s26.	1.1	10
96	Moderately severe osteogenesis imperfecta associated with substitutions of serine for glycine in the Î±1(I) chain of type I collagen. American Journal of Medical Genetics Part A, 1993, 45, 241-245.	2.4	4
97	Evaluation of growth hormone axis and responsiveness to growth stimulation of short children with osteogenesis imperfecta. American Journal of Medical Genetics Part A, 1993, 45, 261-264.	2.4	49
98	A de novo G+1 â†' a mutation at the Î±2(I) exon 16 splice donor site causes skipping of exon 16 in the cDNA of one allele of an OI Type IV proband. Human Mutation, 1993, 2, 380-388.	1.1	13
99	New autosomal recessive syndrome of sparse hair, osteopenia, and mental retardation in Mennonite sisters. American Journal of Medical Genetics Part A, 1992, 43, 983-988.	2.4	4
100	Intractable vasculitis, resorptive osteolysis, and immunity to type i collagen in type viii ehlersâ€danlos syndrome. Arthritis and Rheumatism, 1991, 34, 1466-1475.	6.7	16
101	Analysis Of cultured chorionic villi in a case of osteogenesis imperfecta type II: Implications for prenatal diagnosis. American Journal of Medical Genetics Part A, 1990, 36, 258-264.	2.4	4