Antonella Forlino

List of Publications by Year
in descending order

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Version: 2024-02-01

108	6,275	94381	74108
papers	citations	37 h-index	75 g-index

109	109	109	6802
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Osteogenesis imperfecta. Lancet, The, 2016, 387, 1657-1671.	6.3	668
2	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
3	New perspectives on osteogenesis imperfecta. Nature Reviews Endocrinology, 2011, 7, 540-557.	4.3	556
4	Osteogenesis imperfecta. Nature Reviews Disease Primers, 2017, 3, 17052.	18.1	481
5	Constitutive Activation of PKA Catalytic Subunit in Adrenal Cushing's Syndrome. New England Journal of Medicine, 2014, 370, 1019-1028.	13.9	355
6	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
7	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
8	PRKACB and Carney Complex. New England Journal of Medicine, 2014, 370, 1065-1067.	13.9	121
9	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
10	A diastrophic dysplasia sulfate transporter (SLC26A2) mutant mouse: morphological and biochemical characterization of the resulting chondrodysplasia phenotype. Human Molecular Genetics, 2005, 14, 859-871.	1.4	116
11	Flavivirus Antagonism of Type I Interferon Signaling Reveals Prolidase as a Regulator of IFNAR1 Surface Expression. Cell Host and Microbe, 2015, 18, 61-74.	5.1	115
12	Human prolidase and prolidase deficiency: an overview on the characterization of the enzyme involved in proline recycling and on the effects of its mutations. Amino Acids, 2008, 35, 739-752.	1.2	103
13	Glycosaminoglycans show a specific periodic interaction with type I collagen fibrils. Journal of Structural Biology, 2008, 164, 134-139.	1.3	93
14	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
15	Current and emerging treatments for the management of osteogenesis imperfecta. Therapeutics and Clinical Risk Management, 2010, 6, 367.	0.9	87
16	XX males SRY negative: a confirmed cause of infertility. Journal of Medical Genetics, 2011, 48, 710-712.	1.5	86
17	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
18	Zebrafish: A Resourceful Vertebrate Model to Investigate Skeletal Disorders. Frontiers in Endocrinology, 2020, 11, 489.	1.5	74

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19	The chaperone activity of 4PBA ameliorates the skeletal phenotype of Chihuahua, a zebrafish model for dominant osteogenesis imperfecta. Human Molecular Genetics, 2017, 26, 2897-2911.	1.4	68
20	Evaluation of the TiMo12Zr6Fe2 alloy for orthopaedic implants: in vitro biocompatibility study by using primary human fibroblasts and osteoblasts. Biomaterials, 2002, 23, 2863-2869.	5.7	67
21	Severely Impaired Bone Material Quality in Chihuahua Zebrafish Resembles Classical Dominant Human Osteogenesis Imperfecta. Journal of Bone and Mineral Research, 2018, 33, 1489-1499.	3.1	61
22	MCM5: a new actor in the link between DNA replication and Meier-Gorlin syndrome. European Journal of Human Genetics, 2017, 25, 646-650.	1.4	60
23	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
24	Osteogenesis Imperfecta: Prospects for Molecular Therapeutics. Molecular Genetics and Metabolism, 2000, 71, 225-232.	0.5	58
25	Deletions of the PRKAR1A Locus at 17q24.2-q24.3 in Carney Complex: Genotype-Phenotype Correlations and Implications for Genetic Testing. Journal of Clinical Endocrinology and Metabolism, 2014, 99, E183-E188.	1.8	57
26	HEM dysplasia and ichthyosis are likely laminopathies and not due to 3Î²-hydroxysterol Î”14-reductase deficiency. Human Molecular Genetics, 2007, 16, 1176-1187.	1.4	56
27	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
28	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
29	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
30	Activity of Î±1-antitrypsin and cigarette smoking in subarachnoid haemorrhage from ruptured aneurysm. Journal of the Neurological Sciences, 1996, 141, 33-38.	0.3	50
31	Mutation analysis of five new patients affected by prolidase deficiency: the lack of enzyme activity causes necrosis-like cell death in cultured fibroblasts. Human Genetics, 2002, 111, 314-322.	1.8	49
32	Further delineation of CANT1 phenotypic spectrum and demonstration of its role in proteoglycan synthesis. Human Mutation, 2012, 33, 1261-1266.	1.1	47
33	TREX1 C-terminal frameshift mutations in the systemic variant of retinal vasculopathy with cerebral leukodystrophy. Neurological Sciences, 2015, 36, 323-330.	0.9	45
34	Defective proteoglycan sulfation of the growth plate zones causes reduced chondrocyte proliferation via an altered Indian hedgehog signalling. Matrix Biology, 2010, 29, 453-460.	1.5	44
35	Paternal mosaicism for a COL1A1 dominant mutation (Î±1 Ser-415) causes recurrent osteogenesis imperfecta. Human Mutation, 1993, 2, 196-204.	1.1	40
36	Alteration of proteoglycan sulfation affects bone growth and remodeling. Bone, 2013, 54, 83-91.	1.4	40

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37	Human recombinant prolidase from eukaryotic and prokaryotic sources.. FEBS Journal, 2006, 273, 5466-5478.	2.2	38
38	Three novel type I collagen mutations in osteogenesis imperfecta type IV probands are associated with discrepancies between electrophoretic migration of osteoblast and fibroblast collagen. , 1998, 11, 395-403.		37
39	Bone biology: insights from osteogenesis imperfecta and related rare fragility syndromes. FEBS Journal, 2019, 286, 3033-3056.	2.2	35
40	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
41	Characterization of stress response in human retinal epithelial cells. Journal of Cellular and Molecular Medicine, 2013, 17, 103-115.	1.6	32
42	Absence of Dipeptidyl Peptidase 3 Increases Oxidative Stress and Causes Bone Loss. Journal of Bone and Mineral Research, 2019, 34, 2133-2148.	3.1	32
43	Cellular stress due to impairment of collagen prolyl hydroxylation complex is rescued by the chaperone 4-phenylbutyrate. DMM Disease Models and Mechanisms, 2019, 12, .	1.2	32
44	Identifying the structure of the active sites of human recombinant prolidase. European Biophysics Journal, 2010, 39, 935-945.	1.2	30
45	Replenishing Cartilage from Endogenous Stem Cells. New England Journal of Medicine, 2012, 366, 2522-2524.	13.9	30
46	In vivo contribution of amino acid sulfur to cartilage proteoglycan sulfation. Biochemical Journal, 2006, 398, 509-514.	1.7	29
47	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
48	Characterization of a new PEPD allele causing prolidase deficiency in two unrelated patients: natural-occurrent mutations as a tool to investigate structureâ€“function relationship. Journal of Human Genetics, 2004, 49, 500-506.	1.1	28
49	Crtap and p3h1 knock out zebrafish support defective collagen chaperoning as the cause of their osteogenesis imperfecta phenotype. Matrix Biology, 2020, 90, 40-60.	1.5	28
50	Calcium activated nucleotidase 1 (CANT1) is critical for glycosaminoglycan biosynthesis in cartilage and endochondral ossification. Matrix Biology, 2019, 81, 70-90.	1.5	27
51	Deficient expression of the small proteoglycan decorin in a case of severe/lethal osteogenesis imperfecta. , 1996, 63, 161-166.		24
52	Matrix Disruptions, Growth, and Degradation of Cartilage with Impaired Sulfation. Journal of Biological Chemistry, 2012, 287, 22030-22042.	1.6	24
53	Multiple effects of the Na+/H+ antiporter inhibitor HMA on cancer cells. Apoptosis: an International Journal on Programmed Cell Death, 2013, 18, 1586-1598.	2.2	24
54	Mutations in the Neuroblastoma Amplified Sequence gene in a family affected by Acrofrontofacionasal Dysostosis type 1. Bone, 2018, 114, 125-136.	1.4	24

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55	When Functionalization Becomes Useful: Ionic Liquids with a â€œSweetâ€•Appended Moiety Demonstrate Drastically Reduced Toxicological Effects. ACS Sustainable Chemistry and Engineering, 2020, 8, 926-938.	3.2	24
56	Loss-of-FunctionFANCLMutations Associate with Severe Fanconi Anemia Overlapping the VACTERL Association. Human Mutation, 2015, 36, 562-568.	1.1	23
57	Lack of prolidase causes a bone phenotype both in human and in mouse. Bone, 2015, 72, 53-64.	1.4	23
58	N-acetylcysteine treatment ameliorates the skeletal phenotype of a mouse model of diastrophic dysplasia. Human Molecular Genetics, 2015, 24, 5570-5580.	1.4	22
59	Cytoskeleton and nuclear lamina affection in recessive osteogenesis imperfecta: A functional proteomics perspective. Journal of Proteomics, 2017, 167, 46-59.	1.2	22
60	Alternative Splicing in COL1A1 mRNA Leads to a Partial Null Allele and Two In-frame Forms with Structural Defects in Non-lethal Osteogenesis Imperfecta. Journal of Biological Chemistry, 1996, 271, 28617-28623.	1.6	21
61	N-benzyloxycarbonyl-l-proline: An in vitro and in vivo inhibitor of prolidase. Biochimica Et Biophysica Acta - Molecular Cell Research, 2005, 1744, 157-163.	1.9	21
62	Decorin Transfection Induces Proteomic and Phenotypic Modulation in Breast Cancer Cells 8701-BC. Connective Tissue Research, 2008, 49, 30-41.	1.1	21
63	Kinetic and Structural Evidences on Human Prolidase Pathological Mutants Suggest Strategies for Enzyme Functional Rescue. PLoS ONE, 2013, 8, e58792.	1.1	21
64	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
65	Appendage Regeneration in Vertebrates: What Makes This Possible?. Cells, 2021, 10, 242.	1.8	21
66	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
67	A quantitative and qualitative method for direct 2â€ÐE analysis of murine cartilage. Proteomics, 2007, 7, 4003-4007.	1.3	20
68	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
69	Treatment options for osteogenesis imperfecta. Expert Opinion on Orphan Drugs, 2015, 3, 165-181.	0.5	19
70	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
71	More Bone with Less Minerals? The Effects of Dietary Phosphorus on the Post-Cranial Skeleton in Zebrafish. International Journal of Molecular Sciences, 2020, 21, 5429.	1.8	18
72	A Mn(II)â€“Mn(II) center in human prolidase. Biochimica Et Biophysica Acta - Proteins and Proteomics, 2013, 1834, 197-204.	1.1	17

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73	Dissecting the phenotypic variability of osteogenesis imperfecta. DMM Disease Models and Mechanisms, 2022, 15, .	1.2	16
74	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
75	Procollagen with Skipping of Î±1(I) Exon 41 Has Lower Binding Affinity for Î±1(I) C-telopeptide, Impaired in Vitro Fibrillogenesis, and Altered Fibril Morphology. Journal of Biological Chemistry, 2002, 277, 4215-4222.	1.6	15
76	Extracellular Matrix Deposition in Cultured Dermal Fibroblasts from Four Probands Affected by Osteogenesis Imperfecta. Matrix Biology, 1993, 13, 275-280.	1.8	13
77	Improved prolidase activity assay allowed enzyme kinetic characterization and faster prolidase deficiency diagnosis. Clinica Chimica Acta, 2011, 412, 1814-1820.	0.5	13
78	Prolidase deficiency: case reports of two Argentinian brothers. International Journal of Dermatology, 2004, 43, 684-686.	0.5	12
79	Novel RPL13 Variants and Variable Clinical Expressivity in a Human Ribosomopathy With Spondyloepimetaphyseal Dysplasia. Journal of Bone and Mineral Research, 2020, 36, 283-297.	3.1	12
80	Osteogenesis imprefecta and type-I collagen mutations. A lethal variant caused by a Gly910Ala substitution in the alpha1(I) chain. FEBS Journal, 1993, 211, 415-419.	0.2	11
81	Partial Rescue of Biochemical Parameters After Hematopoietic Stem Cell Transplantation in a Patient with Prolidase Deficiency Due to Two Novel PEPD Mutations. JIMD Reports, 2011, 3, 71-77.	0.7	11
82	Altered Signaling in the G1 Phase Deregulates Chondrocyte Growth in a Mouse Model With Proteoglycan Undersulfation. Journal of Cellular Biochemistry, 2014, 115, 1779-1786.	1.2	11
83	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
84	Intracellular and Extracellular Markers of Lethality in Osteogenesis Imperfecta: A Quantitative Proteomic Approach. International Journal of Molecular Sciences, 2021, 22, 429.	1.8	11
85	Alpha1-antitrypsin activity in subarachnoid hemorrhage. Life Sciences, 1996, 59, 15-20.	2.0	10
86	Molecular stability of chemically modified collagen triple helices. FEBS Letters, 2003, 547, 170-176.	1.3	10
87	Improvement of the skeletal phenotype in a mouse model of diastrophic dysplasia after postnatal treatment with N-acetylcysteine. Biochemical Pharmacology, 2021, 185, 114452.	2.0	10
88	New frontiers for dominant osteogenesis imperfecta treatment: gene/cellular therapy approaches. Advances in Regenerative Biology, 2015, 2, 27964.	0.2	9
89	Early Fracture Healing is Delayed in the Col1a2+/G610C Osteogenesis Imperfecta Murine Model. Calcified Tissue International, 2018, 103, 653-662.	1.5	9
90	Deposition of Mutant Type I Collagen in the Extracellular Matrix of Cultured Dermal Fibroblasts in Osteogenesis Imperfecta. Connective Tissue Research, 1993, 29, 41-49.	1.1	8

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91	The Stimulating Effect of Rosmarinic Acid and Extracts from Rosemary and Lemon Balm on Collagen Type I Biosynthesis in Osteogenesis Imperfecta Type I Skin Fibroblasts. Pharmaceutics, 2021, 13, 938.	2.0	8
92	A 931 + 2T â† C transition in one COL1A2 allele causes exon 16 skipping in PROÎ±2(I) mRNA and produces moderately severe OI. Human Mutation, 1995, 6, 268-271.	1.1	6
93	A Rare Cause of Lower Extremity Ulcers. International Journal of Lower Extremity Wounds, 2016, 15, 86-91.	0.6	6
94	Prolidase enzyme is required for extracellular matrix integrity and impacts on postnatal cerebellar cortex development. Journal of Comparative Neurology, 2020, 528, 65-84.	0.9	6
95	Osteoblasts mineralization and collagen matrix are conserved upon specific Col1a2 silencing. Matrix Biology Plus, 2020, 6-7, 100028.	1.9	6
96	Compression Fractures and Partial Phenotype Rescue With a Low Phosphorus Diet in the Chihuahua Zebrafish Osteogenesis Imperfecta Model. Frontiers in Endocrinology, 2022, 13, 851879.	1.5	6
97	Optimization of a capillary electrophoretic method to detect and quantify the Glyâ€“Pro dipeptide in complex matrices from long term cultured prolidase deficiency fibroblasts. Journal of Chromatography B: Analytical Technologies in the Biomedical and Life Sciences, 2003, 795, 133-139.	1.2	5
98	Steady-State and Pulse-Chase Analyses of Fibrillar Collagen. Methods in Molecular Biology, 2019, 1952, 45-53.	0.4	5
99	Testing the Cre-mediated genetic switch for the generation of conditional knock-in mice. PLoS ONE, 2019, 14, e0213660.	1.1	5
100	Knocking out TMEM38B in human foetal osteoblasts hFOB 1.19 by CRISPR/Cas9: A model for recessive OI type XIV. PLoS ONE, 2021, 16, e0257254.	1.1	5
101	Insights from a Transgenic Mouse Model on the Role of SLC26A2 in Health and Disease. Novartis Foundation Symposium, 2008, , 193-212.	1.2	4
102	Recombinant Prolidase Activates EGFR-Dependent Cell Growth in an Experimental Model of Inflammation in HaCaT Keratinocytes. Implication for Wound Healing. Frontiers in Molecular Biosciences, 2022, 9, 876348.	1.6	4
103	Understanding the Role of Estrogen Receptor Status in PRODH/POX-Dependent Apoptosis/Survival in Breast Cancer Cells. Biology, 2021, 10, 1314.	1.3	3
104	Insights from a transgenic mouse model on the role of SLC26A2 in health and disease. Novartis Foundation Symposium, 2006, 273, 193-206; discussion 206-12, 261-4.	1.2	2
105	Phenotypic Characterization of Immortalized Chondrocytes from a Desbuquois Dysplasia Type 1 Mouse Model: A Tool for Studying Defects in Glycosaminoglycan Biosynthesis. International Journal of Molecular Sciences, 2021, 22, 9304.	1.8	1
106	Development of Gene Therapy for Osteogenesis Imperfecta Using Hammerhead Ribozymes and a Knock-In Mouse Model of OI. Pediatric Research, 1999, 45, 139A-139A.	1.1	0
107	Abstract LB-182: Constitutive activation of PRKACA in adrenal Cushing's syndrome. , 2014, , .		0
108	Editorial: Molecular Mechanisms of Heritable Connective Tissue Disorders. Frontiers in Genetics, 2022, 13, 866665.	1.1	0