

Antonella Forlino

List of Publications by Year in descending order

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108
papers

6,275
citations

94381

37
h-index

74108

75
g-index

109
all docs

109
docs citations

109
times ranked

6802
citing authors

#	ARTICLE	IF	CITATIONS
1	Compression Fractures and Partial Phenotype Rescue With a Low Phosphorus Diet in the Chihuahua Zebrafish Osteogenesis Imperfecta Model. <i>Frontiers in Endocrinology</i> , 2022, 13, 851879.	1.5	6
2	Recombinant Prolidase Activates EGFR-Dependent Cell Growth in an Experimental Model of Inflammation in HaCaT Keratinocytes. Implication for Wound Healing. <i>Frontiers in Molecular Biosciences</i> , 2022, 9, 876348.	1.6	4
3	Editorial: Molecular Mechanisms of Heritable Connective Tissue Disorders. <i>Frontiers in Genetics</i> , 2022, 13, 866665.	1.1	0
4	Dissecting the phenotypic variability of osteogenesis imperfecta. <i>DMM Disease Models and Mechanisms</i> , 2022, 15, .	1.2	16
5	Intracellular and Extracellular Markers of Lethality in Osteogenesis Imperfecta: A Quantitative Proteomic Approach. <i>International Journal of Molecular Sciences</i> , 2021, 22, 429.	1.8	11
6	Improvement of the skeletal phenotype in a mouse model of diastrophic dysplasia after postnatal treatment with N-acetylcysteine. <i>Biochemical Pharmacology</i> , 2021, 185, 114452.	2.0	10
7	Targeting cellular stress in vitro improves osteoblast homeostasis, matrix collagen content and mineralization in two murine models of osteogenesis imperfecta. <i>Matrix Biology</i> , 2021, 98, 1-20.	1.5	19
8	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. <i>Pharmaceutics</i> , 2021, 13, 938.	2.0	8
9	Phenotypic Characterization of Immortalized Chondrocytes from a Desbuquois Dysplasia Type 1 Mouse Model: A Tool for Studying Defects in Glycosaminoglycan Biosynthesis. <i>International Journal of Molecular Sciences</i> , 2021, 22, 9304.	1.8	1
10	Knocking out TMEM38B in human foetal osteoblasts hFOB 1.19 by CRISPR/Cas9: A model for recessive OI type XIV. <i>PLoS ONE</i> , 2021, 16, e0257254.	1.1	5
11	Appendage Regeneration in Vertebrates: What Makes This Possible?. <i>Cells</i> , 2021, 10, 242.	1.8	21
12	Understanding the Role of Estrogen Receptor Status in PRODH/POX-Dependent Apoptosis/Survival in Breast Cancer Cells. <i>Biology</i> , 2021, 10, 1314.	1.3	3
13	Prolidase enzyme is required for extracellular matrix integrity and impacts on postnatal cerebellar cortex development. <i>Journal of Comparative Neurology</i> , 2020, 528, 65-84.	0.9	6
14	When Functionalization Becomes Useful: Ionic Liquids with a "Sweet" Appended Moiety Demonstrate Drastically Reduced Toxicological Effects. <i>ACS Sustainable Chemistry and Engineering</i> , 2020, 8, 926-938.	3.2	24
15	More Bone with Less Minerals? The Effects of Dietary Phosphorus on the Post-Cranial Skeleton in Zebrafish. <i>International Journal of Molecular Sciences</i> , 2020, 21, 5429.	1.8	18
16	Zebrafish: A Resourceful Vertebrate Model to Investigate Skeletal Disorders. <i>Frontiers in Endocrinology</i> , 2020, 11, 489.	1.5	74
17	Osteoblasts mineralization and collagen matrix are conserved upon specific Col1a2 silencing. <i>Matrix Biology Plus</i> , 2020, 6-7, 100028.	1.9	6
18	Crtap and p3h1 knock out zebrafish support defective collagen chaperoning as the cause of their osteogenesis imperfecta phenotype. <i>Matrix Biology</i> , 2020, 90, 40-60.	1.5	28

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19	Substitution of murine type I collagen A1 3-hydroxylation site alters matrix structure but does not recapitulate osteogenesis imperfecta bone dysplasia. <i>Matrix Biology</i> , 2020, 90, 20-39.	1.5	11
20	Novel RPL13 Variants and Variable Clinical Expressivity in a Human Ribosomopathy With Spondyloepimetaphyseal Dysplasia. <i>Journal of Bone and Mineral Research</i> , 2020, 36, 283-297.	3.1	12
21	Absence of Dipeptidyl Peptidase 3 Increases Oxidative Stress and Causes Bone Loss. <i>Journal of Bone and Mineral Research</i> , 2019, 34, 2133-2148.	3.1	32
22	Cellular stress due to impairment of collagen prolyl hydroxylation complex is rescued by the chaperone 4-phenylbutyrate. <i>DMM Disease Models and Mechanisms</i> , 2019, 12, .	1.2	32
23	Bone biology: insights from osteogenesis imperfecta and related rare fragility syndromes. <i>FEBS Journal</i> , 2019, 286, 3033-3056.	2.2	35
24	Steady-State and Pulse-Chase Analyses of Fibrillar Collagen. <i>Methods in Molecular Biology</i> , 2019, 1952, 45-53.	0.4	5
25	Testing the Cre-mediated genetic switch for the generation of conditional knock-in mice. <i>PLoS ONE</i> , 2019, 14, e0213660.	1.1	5
26	Calcium activated nucleotidase 1 (CANT1) is critical for glycosaminoglycan biosynthesis in cartilage and endochondral ossification. <i>Matrix Biology</i> , 2019, 81, 70-90.	1.5	27
27	4-PBA ameliorates cellular homeostasis in fibroblasts from osteogenesis imperfecta patients by enhancing autophagy and stimulating protein secretion. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2018, 1864, 1642-1652.	1.8	55
28	Severely Impaired Bone Material Quality in Chihuahua Zebrafish Resembles Classical Dominant Human Osteogenesis Imperfecta. <i>Journal of Bone and Mineral Research</i> , 2018, 33, 1489-1499.	3.1	61
29	Mutations in the Neuroblastoma Amplified Sequence gene in a family affected by Acrofrontofacionasal Dysostosis type 1. <i>Bone</i> , 2018, 114, 125-136.	1.4	24
30	Early Fracture Healing is Delayed in the Col1a2+/G610C Osteogenesis Imperfecta Murine Model. <i>Calcified Tissue International</i> , 2018, 103, 653-662.	1.5	9
31	MCM5: a new actor in the link between DNA replication and Meier-Gorlin syndrome. <i>European Journal of Human Genetics</i> , 2017, 25, 646-650.	1.4	60
32	The chaperone activity of 4PBA ameliorates the skeletal phenotype of Chihuahua, a zebrafish model for dominant osteogenesis imperfecta. <i>Human Molecular Genetics</i> , 2017, 26, 2897-2911.	1.4	68
33	Osteogenesis imperfecta. <i>Nature Reviews Disease Primers</i> , 2017, 3, 17052.	18.1	481
34	Cytoskeleton and nuclear lamina affection in recessive osteogenesis imperfecta: A functional proteomics perspective. <i>Journal of Proteomics</i> , 2017, 167, 46-59.	1.2	22
35	A Rare Cause of Lower Extremity Ulcers. <i>International Journal of Lower Extremity Wounds</i> , 2016, 15, 86-91.	0.6	6
36	Osteogenesis imperfecta. <i>Lancet, The</i> , 2016, 387, 1657-1671.	6.3	668

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37	New frontiers for dominant osteogenesis imperfecta treatment: gene/cellular therapy approaches. <i>Advances in Regenerative Biology</i> , 2015, 2, 27964.	0.2	9
38	Treatment options for osteogenesis imperfecta. <i>Expert Opinion on Orphan Drugs</i> , 2015, 3, 165-181.	0.5	19
39	TREX1 C-terminal frameshift mutations in the systemic variant of retinal vasculopathy with cerebral leukodystrophy. <i>Neurological Sciences</i> , 2015, 36, 323-330.	0.9	45
40	Flavivirus Antagonism of Type I Interferon Signaling Reveals Prolidase as a Regulator of IFNAR1 Surface Expression. <i>Cell Host and Microbe</i> , 2015, 18, 61-74.	5.1	115
41	Loss-of-Function <i>FANCL</i> Mutations Associate with Severe Fanconi Anemia Overlapping the VACTERL Association. <i>Human Mutation</i> , 2015, 36, 562-568.	1.1	23
42	Altered cytoskeletal organization characterized lethal but not surviving <i>Brtl</i> ^{+/Δ} mice: insight on phenotypic variability in osteogenesis imperfecta. <i>Human Molecular Genetics</i> , 2015, 24, 6118-6133.	1.4	29
43	<i>N</i> -acetylcysteine treatment ameliorates the skeletal phenotype of a mouse model of diastrophic dysplasia. <i>Human Molecular Genetics</i> , 2015, 24, 5570-5580.	1.4	22
44	Lack of prolidase causes a bone phenotype both in human and in mouse. <i>Bone</i> , 2015, 72, 53-64.	1.4	23
45	Allele-specific <i>Col1a1</i> silencing reduces mutant collagen in fibroblasts from <i>Brtl</i> mouse, a model for classical osteogenesis imperfecta. <i>European Journal of Human Genetics</i> , 2014, 22, 667-674.	1.4	21
46	Altered Signaling in the G1 Phase Deregulates Chondrocyte Growth in a Mouse Model With Proteoglycan Undersulfation. <i>Journal of Cellular Biochemistry</i> , 2014, 115, 1779-1786.	1.2	11
47	Deletions of the <i>PRKAR1A</i> Locus at 17q24.2-q24.3 in Carney Complex: Genotype-Phenotype Correlations and Implications for Genetic Testing. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2014, 99, E183-E188.	1.8	57
48	<i>PRKACB</i> and Carney Complex. <i>New England Journal of Medicine</i> , 2014, 370, 1065-1067.	13.9	121
49	Constitutive Activation of PKA Catalytic Subunit in Adrenal Cushing's Syndrome. <i>New England Journal of Medicine</i> , 2014, 370, 1019-1028.	13.9	355
50	Abstract LB-182: Constitutive activation of <i>PRKACA</i> in adrenal Cushing's syndrome. , 2014, , .		0
51	Multiple effects of the Na ⁺ /H ⁺ antiporter inhibitor HMA on cancer cells. <i>Apoptosis: an International Journal on Programmed Cell Death</i> , 2013, 18, 1586-1598.	2.2	24
52	A Mn(II)â€“Mn(II) center in human prolidase. <i>Biochimica Et Biophysica Acta - Proteins and Proteomics</i> , 2013, 1834, 197-204.	1.1	17
53	Alteration of proteoglycan sulfation affects bone growth and remodeling. <i>Bone</i> , 2013, 54, 83-91.	1.4	40
54	Characterization of stress response in human retinal epithelial cells. <i>Journal of Cellular and Molecular Medicine</i> , 2013, 17, 103-115.	1.6	32

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55	Kinetic and Structural Evidences on Human Prolidase Pathological Mutants Suggest Strategies for Enzyme Functional Rescue. PLoS ONE, 2013, 8, e58792.	1.1	21
56	Matrix Disruptions, Growth, and Degradation of Cartilage with Impaired Sulfation. Journal of Biological Chemistry, 2012, 287, 22030-22042.	1.6	24
57	Replenishing Cartilage from Endogenous Stem Cells. New England Journal of Medicine, 2012, 366, 2522-2524.	13.9	30
58	Differential response to intracellular stress in the skin from osteogenesis imperfecta Brl mice with lethal and non lethal phenotype: A proteomic approach. Journal of Proteomics, 2012, 75, 4717-4733.	1.2	19
59	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
60	Further delineation of CANT1 phenotypic spectrum and demonstration of its role in proteoglycan synthesis. Human Mutation, 2012, 33, 1261-1266.	1.1	47
61	New perspectives on osteogenesis imperfecta. Nature Reviews Endocrinology, 2011, 7, 540-557.	4.3	556
62	Improved prolidase activity assay allowed enzyme kinetic characterization and faster prolidase deficiency diagnosis. Clinica Chimica Acta, 2011, 412, 1814-1820.	0.5	13
63	XX males SRY negative: a confirmed cause of infertility. Journal of Medical Genetics, 2011, 48, 710-712.	1.5	86
64	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
65	Identifying the structure of the active sites of human recombinant prolidase. European Biophysics Journal, 2010, 39, 935-945.	1.2	30
66	Current and emerging treatments for the management of osteogenesis imperfecta. Therapeutics and Clinical Risk Management, 2010, 6, 367.	0.9	87
67	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
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	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
70	Cellular Mechanism of Decreased Bone in Brl 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	Glycosaminoglycans show a specific periodic interaction with type I collagen fibrils. Journal of Structural Biology, 2008, 164, 134-139.	1.3	93
72	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

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73	Decorin Transfection Induces Proteomic and Phenotypic Modulation in Breast Cancer Cells 8701-BC. <i>Connective Tissue Research</i> , 2008, 49, 30-41.	1.1	21
74	Insights from a Transgenic Mouse Model on the Role of SLC26A2 in Health and Disease. <i>Novartis Foundation Symposium</i> , 2008, , 193-212.	1.2	4
75	HEM dysplasia and ichthyosis are likely laminopathies and not due to 3 β -hydroxysterol Δ^5 -reductase deficiency. <i>Human Molecular Genetics</i> , 2007, 16, 1176-1187.	1.4	56
76	Selective retention and degradation of molecules with a single mutant $\alpha 1(I)$ chain in the Brl IV mouse model of OI. <i>Matrix Biology</i> , 2007, 26, 604-614.	1.5	52
77	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. <i>Human Mutation</i> , 2007, 28, 209-221.	1.1	620
78	Differential expression of both extracellular and intracellular proteins is involved in the lethal or nonlethal phenotypic variation of BrlIV, a murine model for osteogenesis imperfecta. <i>Proteomics</i> , 2007, 7, 1877-1891.	1.3	51
79	A quantitative and qualitative method for direct ^{35}S analysis of murine cartilage. <i>Proteomics</i> , 2007, 7, 4003-4007.	1.3	20
80	Human recombinant prolidase from eukaryotic and prokaryotic sources. <i>FEBS Journal</i> , 2006, 273, 5466-5478.	2.2	38
81	In vivo contribution of amino acid sulfur to cartilage proteoglycan sulfation. <i>Biochemical Journal</i> , 2006, 398, 509-514.	1.7	29
82	Insights from a transgenic mouse model on the role of SLC26A2 in health and disease. <i>Novartis Foundation Symposium</i> , 2006, 273, 193-206; discussion 206-12, 261-4.	1.2	2
83	N-benzyloxycarbonyl-L-proline: An in vitro and in vivo inhibitor of prolidase. <i>Biochimica Et Biophysica Acta - Molecular Cell Research</i> , 2005, 1744, 157-163.	1.9	21
84	A diastrophic dysplasia sulfate transporter (SLC26A2) mutant mouse: morphological and biochemical characterization of the resulting chondrodysplasia phenotype. <i>Human Molecular Genetics</i> , 2005, 14, 859-871.	1.4	116
85	Prolidase deficiency: case reports of two Argentinian brothers. <i>International Journal of Dermatology</i> , 2004, 43, 684-686.	0.5	12
86	Brittle IV Mouse Model for Osteogenesis Imperfecta IV Demonstrates Postpubertal Adaptations to Improve Whole Bone Strength. <i>Journal of Bone and Mineral Research</i> , 2004, 19, 614-622.	3.1	118
87	Characterization of a new PEPD allele causing prolidase deficiency in two unrelated patients: natural-occurrent mutations as a tool to investigate structure \rightarrow function relationship. <i>Journal of Human Genetics</i> , 2004, 49, 500-506.	1.1	28
88	Structure, stability and interactions of type I collagen with GLY349-CYS substitution in $\alpha 1(I)$ chain in a murine Osteogenesis Imperfecta model. <i>Matrix Biology</i> , 2004, 23, 101-112.	1.5	32
89	Optimization of a capillary electrophoretic method to detect and quantify the Gly \rightarrow Pro dipeptide in complex matrices from long term cultured prolidase deficiency fibroblasts. <i>Journal of Chromatography B: Analytical Technologies in the Biomedical and Life Sciences</i> , 2003, 795, 133-139.	1.2	5
90	Molecular stability of chemically modified collagen triple helices. <i>FEBS Letters</i> , 2003, 547, 170-176.	1.3	10

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91	Procollagen with Skipping of $\hat{I}1(I)$ Exon 41 Has Lower Binding Affinity for $\hat{I}1(I)$ C-telopeptide, Impaired in Vitro Fibrillogenesis, and Altered Fibril Morphology. <i>Journal of Biological Chemistry</i> , 2002, 277, 4215-4222.	1.6	15
92	Evaluation of the TiMo12Zr6Fe2 alloy for orthopaedic implants: in vitro biocompatibility study by using primary human fibroblasts and osteoblasts. <i>Biomaterials</i> , 2002, 23, 2863-2869.	5.7	67
93	Mutation analysis of five new patients affected by prolidase deficiency: the lack of enzyme activity causes necrosis-like cell death in cultured fibroblasts. <i>Human Genetics</i> , 2002, 111, 314-322.	1.8	49
94	Osteogenesis Imperfecta: Prospects for Molecular Therapeutics. <i>Molecular Genetics and Metabolism</i> , 2000, 71, 225-232.	0.5	58
95	Use of the Cre/lox Recombination System to Develop a Non-lethal Knock-in Murine Model for Osteogenesis Imperfecta with an $\hat{I}1(I)$ G349C Substitution. <i>Journal of Biological Chemistry</i> , 1999, 274, 37923-37931.	1.6	125
96	Development of Gene Therapy for Osteogenesis Imperfecta Using Hammerhead Ribozymes and a Knock-In Mouse Model of OI. <i>Pediatric Research</i> , 1999, 45, 139A-139A.	1.1	0
97	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
98	An $\hat{I}2(I)$ glycine to aspartate substitution is responsible for the presence of a kink in type I collagen in a lethal case of osteogenesis imperfecta. <i>Matrix Biology</i> , 1998, 17, 575-584.	1.5	20
99	Phenotypic Comparison of an Osteogenesis Imperfecta Type IV Proband with a Novel $\hat{I}2(I)$ Gly922 \hat{I} Ser Substitution in Type I Collagen and an Unrelated Patient with an Identical Mutation. <i>Biochemical and Molecular Medicine</i> , 1997, 62, 26-35.	1.5	15
100	Activity of $\hat{I}1$ -antitrypsin and cigarette smoking in subarachnoid haemorrhage from ruptured aneurysm. <i>Journal of the Neurological Sciences</i> , 1996, 141, 33-38.	0.3	50
101	Alpha1-antitrypsin activity in subarachnoid hemorrhage. <i>Life Sciences</i> , 1996, 59, 15-20.	2.0	10
102	Deficient expression of the small proteoglycan decorin in a case of severe/lethal osteogenesis imperfecta. , 1996, 63, 161-166.		24
103	Alternative Splicing in COL1A1 mRNA Leads to a Partial Null Allele and Two In-frame Forms with Structural Defects in Non-lethal Osteogenesis Imperfecta. <i>Journal of Biological Chemistry</i> , 1996, 271, 28617-28623.	1.6	21
104	A 931 + 2T \hat{I} C transition in one COL1A2 allele causes exon 16 skipping in PRO $\hat{I}2(I)$ mRNA and produces moderately severe OI. <i>Human Mutation</i> , 1995, 6, 268-271.	1.1	6
105	Paternal mosaicism for a COL1A1 dominant mutation ($\hat{I}1$ Ser-415) causes recurrent osteogenesis imperfecta. <i>Human Mutation</i> , 1993, 2, 196-204.	1.1	40
106	Osteogenesis imperfecta and type-I collagen mutations. A lethal variant caused by a Gly910Ala substitution in the alpha1(I) chain. <i>FEBS Journal</i> , 1993, 211, 415-419.	0.2	11
107	Extracellular Matrix Deposition in Cultured Dermal Fibroblasts from Four Proband Affected by Osteogenesis Imperfecta. <i>Matrix Biology</i> , 1993, 13, 275-280.	1.8	13
108	Deposition of Mutant Type I Collagen in the Extracellular Matrix of Cultured Dermal Fibroblasts in Osteogenesis Imperfecta. <i>Connective Tissue Research</i> , 1993, 29, 41-49.	1.1	8