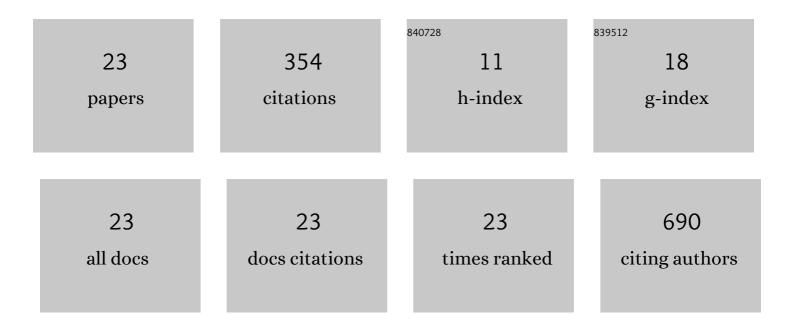
Alice Costantini

List of Publications by Year in descending order

Source: https://exaly.com/author-pdf/7855268/publications.pdf Version: 2024-02-01



#	Article	IF	CITATIONS
1	Mosaic Deletions of Known Genes Explain Skeletal Dysplasias With High and Low Bone Mass. JBMR Plus, 2022, 6, .	2.7	2
2	Novel form of rhizomelic skeletal dysplasia associated with a homozygous variant in GNPNAT1. Journal of Medical Genetics, 2021, 58, 351-356.	3.2	6
3	New gene discoveries in skeletal diseases with short stature. Endocrine Connections, 2021, 10, R160-R174.	1.9	7
4	Oligogenic Inheritance of Monoallelic TRIP11, FKBP10, NEK1, TBX5, and NBAS Variants Leading to a Phenotype Similar to Odontochondrodysplasia. Frontiers in Genetics, 2021, 12, 680838.	2.3	6
5	An <scp><i>ARHGAP25</i></scp> variant links aberrant <scp>Rac1</scp> function to earlyâ€onset skeletal fragility. JBMR Plus, 2021, 5, e10509.	2.7	4
6	Exome Sequencing Reveals a Phenotype Modifying Variant in <scp><i>ZNF528</i></scp> in Primary Osteoporosis With a <scp><i>COL1A2</i></scp> Deletion. Journal of Bone and Mineral Research, 2020, 35, 2381-2392.	2.8	4
7	Unique, Genderâ€Dependent Serum <scp>microRNA</scp> Profile in <scp><i>PLS3</i></scp> Geneâ€Related Osteoporosis. Journal of Bone and Mineral Research, 2020, 35, 1962-1973.	2.8	12
8	PLS3 Mutations Cause Severe Age and Sex-Related Spinal Pathology. Frontiers in Endocrinology, 2020, 11, 393.	3.5	15
9	Biomarkers in WNT1 and PLS3 Osteoporosis: Altered Concentrations of DKK1 and FGF23. Journal of Bone and Mineral Research, 2020, 35, 901-912.	2.8	24
10	â€~Metaphyseal dysplasia without hypotrichosis' can present with late-onset extraskeletal manifestations. Journal of Medical Genetics, 2020, 57, 18-22.	3.2	7
11	Novel RPL13 Variants and Variable Clinical Expressivity in a Human Ribosomopathy With Spondyloepimetaphyseal Dysplasia. Journal of Bone and Mineral Research, 2020, 36, 283-297.	2.8	12
12	New Insights Into Monogenic Causes of Osteoporosis. Frontiers in Endocrinology, 2019, 10, 70.	3.5	56
13	Novel fibronectin mutations and expansion of the phenotype in spondylometaphyseal dysplasia with "corner fractures― Bone, 2019, 121, 163-171.	2.9	13
14	Expansion of the clinical spectrum of frontometaphyseal dysplasia 2 caused by the recurrent mutation p.Pro485Leu in MAP3K7. European Journal of Medical Genetics, 2018, 61, 612-615.	1.3	4
15	Autosomal Recessive Osteogenesis Imperfecta Caused by a Novel Homozygous COL1A2 Mutation. Calcified Tissue International, 2018, 103, 353-358.	3.1	9
16	A novel <i>MYT1L</i> mutation in a patient with severe earlyâ€onset obesity and intellectual disability. American Journal of Medical Genetics, Part A, 2018, 176, 1972-1975.	1.2	24
17	Rare Copy Number Variants in Array-Based Comparative Genomic Hybridization in Early-Onset Skeletal Fragility. Frontiers in Endocrinology, 2018, 9, 380.	3.5	20
18	A novel frameshift deletion in PLS3 causing severe primary osteoporosis. Journal of Human Genetics, 2018, 63, 923-926.	2.3	20

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#	Article	IF	CITATIONS
19	<i>CRTAP</i> variants in earlyâ€onset osteoporosis and recurrent fractures. American Journal of Medical Genetics, Part A, 2017, 173, 806-808.	1.2	2
20	High bone mass due to novel LRP5 and AMER1 mutations. European Journal of Medical Genetics, 2017, 60, 675-679.	1.3	10
21	<i>PLS3</i> Deletions Lead to Severe Spinal Osteoporosis and Disturbed Bone Matrix Mineralization. Journal of Bone and Mineral Research, 2017, 32, 2394-2404.	2.8	41
22	Spondyloocular Syndrome: Novel Mutations in <i>XYLT2</i> Gene and Expansion of the Phenotypic Spectrum. Journal of Bone and Mineral Research, 2016, 31, 1577-1585.	2.8	43
23	Value of rare low bone mass diseases for osteoporosis genetics. BoneKEy Reports, 2016, 5, 773.	2.7	13