Alice Costantini

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/7855268/publications.pdf

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23 papers	354 citations	840585 11 h-index	839398 18 g-index
23 all docs	23 docs citations	23 times ranked	690 citing authors

#	Article	IF	Citations
1	New Insights Into Monogenic Causes of Osteoporosis. Frontiers in Endocrinology, 2019, 10, 70.	1.5	56
2	Spondyloocular Syndrome: Novel Mutations in XYLT2 Gene and Expansion of the Phenotypic Spectrum. Journal of Bone and Mineral Research, 2016, 31, 1577-1585.	3.1	43
3	PLS3 Deletions Lead to Severe Spinal Osteoporosis and Disturbed Bone Matrix Mineralization. Journal of Bone and Mineral Research, 2017, 32, 2394-2404.	3.1	41
4	A novel MYT1L mutation in a patient with severe earlyâ€onset obesity and intellectual disability. American Journal of Medical Genetics, Part A, 2018, 176, 1972-1975.	0.7	24
5	Biomarkers in WNT1 and PLS3 Osteoporosis: Altered Concentrations of DKK1 and FGF23. Journal of Bone and Mineral Research, 2020, 35, 901-912.	3.1	24
6	Rare Copy Number Variants in Array-Based Comparative Genomic Hybridization in Early-Onset Skeletal Fragility. Frontiers in Endocrinology, 2018, 9, 380.	1.5	20
7	A novel frameshift deletion in PLS3 causing severe primary osteoporosis. Journal of Human Genetics, 2018, 63, 923-926.	1.1	20
8	PLS3 Mutations Cause Severe Age and Sex-Related Spinal Pathology. Frontiers in Endocrinology, 2020, 11, 393.	1.5	15
9	Value of rare low bone mass diseases for osteoporosis genetics. BoneKEy Reports, 2016, 5, 773.	2.7	13
10	Novel fibronectin mutations and expansion of the phenotype in spondylometaphyseal dysplasia with â€œcorner fracturesâ€. Bone, 2019, 121, 163-171.	1.4	13
11	Unique, Genderâ€Dependent Serum <scp>microRNA</scp> Profile in <scp>PLS3</scp> Geneâ€Related Osteoporosis. Journal of Bone and Mineral Research, 2020, 35, 1962-1973.	3.1	12
12	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
13	High bone mass due to novel LRP5 and AMER1 mutations. European Journal of Medical Genetics, 2017, 60, 675-679.	0.7	10
14	Autosomal Recessive Osteogenesis Imperfecta Caused by a Novel Homozygous COL1A2 Mutation. Calcified Tissue International, 2018, 103, 353-358.	1.5	9
15	â€˜Metaphyseal dysplasia without hypotrichosisâ€™ can present with late-onset extraskeletal manifestations. Journal of Medical Genetics, 2020, 57, 18-22.	1.5	7
16	New gene discoveries in skeletal diseases with short stature. Endocrine Connections, 2021, 10, R160-R174.	0.8	7
17	Novel form of rhizomelic skeletal dysplasia associated with a homozygous variant in GNPNAT1. Journal of Medical Genetics, 2021, 58, 351-356.	1.5	6
18	Oligogenic Inheritance of Monoallelic TRIP11, FKBP10, NEK1, TBX5, and NBAS Variants Leading to a Phenotype Similar to Odontochondrodysplasia. Frontiers in Genetics, 2021, 12, 680838.	1.1	6

#	Article	IF	Citations
19	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.	0.7	4
20	Exome Sequencing Reveals a Phenotype Modifying Variant in <scp>ZNF528</scp> in Primary Osteoporosis With a <scp>COL1A2</scp> Deletion. Journal of Bone and Mineral Research, 2020, 35, 2381-2392.	3.1	4
21	An<scp>ARHGAP25</scp>variant links aberrant<scp>Rac1</scp>function to earlyâ€onset skeletal fragility. JBMR Plus, 2021, 5, e10509.	1.3	4
22	CRTAP variants in earlyâ€onset osteoporosis and recurrent fractures. American Journal of Medical Genetics, Part A, 2017, 173, 806-808.	0.7	2
23	Mosaic Deletions of Known Genes Explain Skeletal Dysplasias With High and Low Bone Mass. JBMR Plus, 2022, 6, .	1.3	2