Maria Gnoli

List of Publications by Year
in descending order

11 papers	116 citations	1478505 6 h-index	1474206 9 g-index
11 all docs	11 docs citations	11 times ranked	190 citing authors

#	Article	IF	Citations
1	Genotypeâ€“phenotype correlation study in 364 osteogenesis imperfecta Italian patients. European Journal of Human Genetics, 2019, 27, 1090-1100.	2.8	52
2	Patterns of Novel Alleles and Genotype/Phenotype Correlations Resulting from the Analysis of 108 Previously Undetected Mutations in Patients Affected by Neurofibromatosis Type I. International Journal of Molecular Sciences, 2017, 18, 2071.	4.1	11
3	The natural history of multiple osteochondromas in a large Italian cohort of pediatric patients. Bone, 2020, 139, 115499.	2.9	11
4	COL1-Related Disorders: Case Report and Review of Overlapping Syndromes. Frontiers in Genetics, 2021, 12, 640558.	2.3	9
5	Melorheostosis and Osteopoikilosis Clinical and Molecular Description of an Italian Case Series. Calcified Tissue International, 2019, 105, 215-221.	3.1	7
6	Variable clinical expression of Stickler Syndrome: A case report of a novel COL11A1 mutation. Molecular Genetics & Genomic Medicine, 2020, 8, e1353.	1.2	7
7	Tumor Syndromes That Include Bone Tumors. Surgical Pathology Clinics, 2017, 10, 749-764.	1.7	6
8	The Rizzoli Multiple Osteochondromas Classification revised: describing the phenotype to improve clinical practice. American Journal of Medical Genetics, Part A, 2021, 185, 3466-3475.	1.2	6
9	Evaluation of TP53 Pro72Arg and MDM2 SNP285â€“SNP309 polymorphisms in an Italian cohort of LFS suggestive patients lacking identifiable TP53 germline mutations. Familial Cancer, 2016, 15, 635-643.	1.9	4
10	Familiar osteopoikilosis: Case report with differential diagnosis and review of the literature. Clinical Case Reports (discontinued), 2021, 9, 922-926.	0.5	2
11	An Easy-to-Use Approach to Detect CNV From Targeted NGS Data: Identification of a Novel Pathogenic Variant in MO Disease. Frontiers in Endocrinology, 0, 13, .	3.5	1