Maria Francesca Bedeschi

List of Publications by Year
in descending order

22 papers	320 citations	1162889 8 h-index	887953 17 g-index
23 all docs	23 docs citations	23 times ranked	555 citing authors

#	Article	IF	Citations
1	(Epi)genotypeâ€“phenotype correlations in Beckwithâ€“Wiedemann syndrome. European Journal of Human Genetics, 2016, 24, 183-190.	1.4	113
2	Tubulin-related cerebellar dysplasia: definition of a distinct pattern of cerebellar malformation. European Radiology, 2017, 27, 5080-5092.	2.3	36
3	Association of syndromic mental retardation with an Xq12q13.1 duplication encompassing the oligophrenin 1 gene. American Journal of Medical Genetics, Part A, 2008, 146A, 1718-1724.	0.7	20
4	Assessment of copy number variations in 120 patients with Poland syndrome. BMC Medical Genetics, 2016, 17, 89.	2.1	20
5	Impairment of different protein domains causes variable clinical presentation within Pitt-Hopkins syndrome and suggests intragenic molecular syndromology of TCF4. European Journal of Medical Genetics, 2017, 60, 565-571.	0.7	18
6	Smith-Magenis Syndromeâ€”Clinical Review, Biological Background and Related Disorders. Genes, 2022, 13, 335.	1.0	18
7	Prenatal ultrasound factors and genetic disorders in pregnancies complicated by polyhydramnios. Prenatal Diagnosis, 2016, 36, 726-730.	1.1	13
8	Not Only Diagnostic Yield: Whole-Exome Sequencing in Infantile Cardiomyopathies Impacts on Clinical and Family Management. Journal of Cardiovascular Development and Disease, 2022, 9, 2.	0.8	12
9	Exon-Trapping Assay Improves Clinical Interpretation of COL11A1 and COL11A2 Intronic Variants in Stickler Syndrome Type 2 and Otospondylomegaepiphyseal Dysplasia. Genes, 2020, 11, 1513.	1.0	11
10	Melorheostosis and Osteopoikilosis Clinical and Molecular Description of an Italian Case Series. Calcified Tissue International, 2019, 105, 215-221.	1.5	7
11	Increased RISK for 47,XXY on cellâ€free DNA screen: Not always Klinefelter syndrome. Prenatal Diagnosis, 2021, 41, 1255-1257.	1.1	7
12	Unusual prenatal presentation of Rubinsteinâ€“Taybi syndrome: A case report. American Journal of Medical Genetics, Part A, 2014, 164, 2663-2666.	0.7	6
13	A case series of CHARGE syndrome: identification of key features for a neonatal diagnosis. Italian Journal of Pediatrics, 2020, 46, 53.	1.0	6
14	Impaired glucose metabolism in subjects with the Williams-Beuren syndrome: A five-year follow-up cohort study. PLoS ONE, 2017, 12, e0185371.	1.1	6
15	Expanding the Molecular Spectrum of ANKRD11 Gene Defects in 33 Patients with a Clinical Presentation of KBG Syndrome. International Journal of Molecular Sciences, 2022, 23, 5912.	1.8	6
16	Molecular cytogenetic characterization of a 2q35â€q37 duplication and a 4q35.1â€q35.2 deletion in two cousins: A genotypeâ€“phenotype analysis. American Journal of Medical Genetics, Part A, 2015, 167, 1551-1559.	0.7	4
17	Rare interstitial deletion of chromosome 2p11.2p12. Report of a new patient with developmental delay and unusual clinical features. European Journal of Medical Genetics, 2016, 59, 39-42.	0.7	4
18	Prenatal upper-limb mesomelia and 2q31.1 microdeletions affecting the regulatory genome. Genetics in Medicine, 2018, 20, 1483-1484.	1.1	4

#	Article	IF	Citations
19	Sequence variants identification at the KCNQ1OT1:TSS differentially Methylated region in isolated omphalocele cases. BMC Medical Genetics, 2017, 18, 115.	2.1	3
20	An additional piece in the <scp>TBX6</scp> gene dosage model: A novel nonsense variant in a fetus with severe spondylocostal dysostosis. Clinical Genetics, 2020, 98, 628-629.	1.0	2
21	Biopsy-proven multiple sclerosis in an adult patient with atypical craniometaphyseal dysplasia. BMJ Case Reports, 2018, 2018, bcr-2017-223390.	0.2	1
22	Neuroimaging appearance of hypothalamic hamartomas in monozygotic twins with Pallister-Hall syndrome: case report and review of the literature. BMC Neurology, 2022, 22, 118.	0.8	0