Maria F Bedeschi

List of Publications by Year
in descending order

9	209	1162889 8	1474057 9
papers	citations	h-index	g-index

10	10	10	464
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Childhood-onset dystonia-causing KMT2B variants result in a distinctive genomic hypermethylation profile. Clinical Epigenetics, 2021, 13, 157.	1.8	22
2	Biallelic MADD variants cause a phenotypic spectrum ranging from developmental delay to a multisystem disorder. Brain, 2020, 143, 2437-2453.	3.7	21
3	A Genetics-First Approach Revealed Monogenic Disorders in Patients With ARM and VACTERL Anomalies. Frontiers in Pediatrics, 2020, 8, 310.	0.9	17
4	(Epi)genetic profiling of extraembryonic and postnatal tissues from female monozygotic twins discordant for Beckwithâ€“Wiedemann syndrome. Molecular Genetics & Genomic Medicine, 2020, 8, e1386.	0.6	9
5	Phenotype delineation of ZNF462 related syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 2075-2082.	0.7	23
6	The spatiotemporal organization of episodic memory and its disruption in a neurodevelopmental disorder. Scientific Reports, 2019, 9, 18447.	1.6	13
7	STAR syndrome plus: The first description of a female patient with the lethal form. American Journal of Medical Genetics, Part A, 2017, 173, 3226-3230.	0.7	8
8	Clinical Characterization, Genetics, and Long-Term Follow-up of a Large Cohort of Patients With Agenesis of the Corpus Callosum. Journal of Child Neurology, 2017, 32, 60-71.	0.7	34
9	Moebius syndrome: clinical features, diagnosis, management and early intervention. Italian Journal of Pediatrics, 2016, 42, 56.	1.0	62