Marina Macchiaiolo

List of Publications by Year
in descending order

13	241	1163117 8	1281871 11
papers	citations	h-index	g-index

13	13	13	443
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Diagnosis and management in Pittâ€Hopkins syndrome: First international consensus statement. Clinical Genetics, 2019, 95, 462-478.	2.0	63
2	Growth hormone excess in children with neurofibromatosis typeâ€1 and optic glioma. American Journal of Medical Genetics, Part A, 2017, 173, 2353-2358.	1.2	38
3	Homozygous familial hypercholesterolaemia. Lancet, The, 2012, 379, 1330.	13.7	37
4	Nomenclature and definition in asymmetric regional body overgrowth. American Journal of Medical Genetics, Part A, 2017, 173, 1735-1738.	1.2	36
5	TUBB3 E410K syndrome: Case report and review of the clinical spectrum of TUBB3 mutations. American Journal of Medical Genetics, Part A, 2020, 182, 1977-1984.	1.2	15
6	An unusual case of anisocoria by vegetal intoxication: a case report. Italian Journal of Pediatrics, 2010, 36, 50.	2.6	14
7	Clinical and molecular characterization of patients with adenylosuccinate lyase deficiency. Orphanet Journal of Rare Diseases, 2021, 16, 112.	2.7	12
8	A mild form of adenylosuccinate lyase deficiency in absence of typical brain MRI features diagnosed by whole exome sequencing. Italian Journal of Pediatrics, 2017, 43, 65.	2.6	9
9	Diffuse infantile hepatic hemangiomas in a patient with <scp>Beckwithâ€“Wiedemann</scp> syndrome: A new association?. American Journal of Medical Genetics, Part A, 2020, 182, 1972-1976.	1.2	6
10	A deep phenotyping experience: up to date in management and diagnosis of Malan syndrome in a single center surveillance report. Orphanet Journal of Rare Diseases, 2022, 17, .	2.7	6
11	Very mild isolated intellectual disability caused by adenylosuccinate lyase deficiency: a new phenotype. Molecular Genetics and Metabolism Reports, 2020, 23, 100592.	1.1	4
12	Langerhans cell histiocytosis in a young patient with Pittâ€“Hopkins syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 2746-2750.	1.2	1
13	Cover Image, Volume 173A, Number 7, July 2017. American Journal of Medical Genetics, Part A, 2017, 173, i.	1.2	0