

Marina Macchiaiolo

List of Publications by Year in descending order

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Version: 2024-02-01

13
papers

241
citations

1163117

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h-index

1281871

11
g-index

13
all docs

13
docs citations

13
times ranked

443
citing authors

#	ARTICLE	IF	CITATIONS
1	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
2	Clinical and molecular characterization of patients with adenylosuccinate lyase deficiency. Orphanet Journal of Rare Diseases, 2021, 16, 112.	2.7	12
3	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
4	Very mild isolated intellectual disability caused by adenylosuccinate lyase deficiency: a new phenotype. Molecular Genetics and Metabolism Reports, 2020, 23, 100592.	1.1	4
5	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
6	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
7	Diagnosis and management in Pittâ€™Hopkins syndrome: First international consensus statement. Clinical Genetics, 2019, 95, 462-478.	2.0	63
8	Nomenclature and definition in asymmetric regional body overgrowth. American Journal of Medical Genetics, Part A, 2017, 173, 1735-1738.	1.2	36
9	Cover Image, Volume 173A, Number 7, July 2017. American Journal of Medical Genetics, Part A, 2017, 173, i.	1.2	0
10	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
11	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
12	Homozygous familial hypercholesterolaemia. Lancet, The, 2012, 379, 1330.	13.7	37
13	An unusual case of anisocoria by vegetal intoxication: a case report. Italian Journal of Pediatrics, 2010, 36, 50.	2.6	14