

# Paola Sabrina Buonuomo

## List of Publications by Year in descending order

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Version: 2024-02-01

13  
papers

110  
citations

1478505

6  
h-index

1372567

10  
g-index

13  
all docs

13  
docs citations

13  
times ranked

191  
citing authors

#	ARTICLE	IF	CITATIONS
1	Genotypes and phenotypes heterogeneity in PIK3CA-related overgrowth spectrum and overlapping conditions: 150 novel patients and systematic review of 1007 patients with PIK3CA pathogenetic variants. <i>Journal of Medical Genetics</i> , 2023, 60, 163-173.	3.2	15
2	Expanding the novel <i>MAPKAPK5</i> related developmental disorder's genotype-phenotype correlation: patient report and 19 months follow-up. <i>Clinical Genetics</i> , 2022, , .	2.0	2
3	Infantile myofibromatosis: a rare cause of subcutaneous nodules in an infant. <i>Archives of Disease in Childhood</i> , 2021, 106, 428-428.	1.9	1
4	Evolocumab in the management of children <10 years of age affected by homozygous familial hypercholesterolemia. <i>Atherosclerosis</i> , 2021, 324, 148-150.	0.8	2
5	Langerhans cell histiocytosis in a young patient with Pitt-Hopkins syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 2746-2750.	1.2	1
6	Diffuse infantile hepatic hemangiomas in a patient with Beckwith-Wiedemann syndrome: A new association?. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 1972-1976.	1.2	6
7	New Insights into the Role of Lipoprotein(a) as Predictor of Early Onset of Cardiovascular Disease in Pediatric Familial Hypercholesterolemia (FH). <i>Pediatric Cardiology</i> , 2020, 41, 1242-1243.	1.3	1
8	Treatment of homozygous familial hypercholesterolaemia in paediatric patients: A monocentric experience. <i>European Journal of Preventive Cardiology</i> , 2018, 25, 1098-1105.	1.8	5
9	Lipid profiles in a large cohort of Italian children with Down syndrome. <i>European Journal of Medical Genetics</i> , 2016, 59, 392-395.	1.3	20
10	Necrosis of the tongue as first symptom of Polyarteritis Nodosa (PAN): unusual presentation of a rare disease in children. <i>Rheumatology International</i> , 2013, 33, 1071-1073.	3.0	8
11	Acute rheumatic fever with chorea. <i>Archives of Disease in Childhood</i> , 2013, 98, 203-203.	1.9	0
12	Infliximab therapy in pediatric Takayasu's arteritis: report of two cases. <i>Rheumatology International</i> , 2011, 31, 93-95.	3.0	24
13	Relapsing polychondritis: new therapeutic strategies with biological agents. <i>Rheumatology International</i> , 2010, 30, 691-693.	3.0	25