Giulia Pascolini

List of Publications by Year
in descending order

24	271	1306789 7	996533 15
papers	citations	h-index	g-index

26	26	26	495
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Clinical refinement of the <scp>SETD5</scp>â€associated phenotype in a child displaying novel features and <scp>KBG</scp> syndromeâ€like appearance. American Journal of Medical Genetics, Part A, 2022, 188, 1623-1625.	0.7	3
2	Duane retraction syndrome characterized by inner ear agenesis and neurodevelopmental phenotype in an Italian family with a variant in <scp>MAFB</scp>. Clinical Genetics, 2022, 101, 377-378.	1.0	1
3	First Italian experience using the automated craniofacial gestalt analysis on a cohort of pediatric patients with multiple anomaly syndromes. Italian Journal of Pediatrics, 2022, 48, .	1.0	5
4	Koolenâ€de Vries syndrome in the first adulthood patient of Southern India ancestry. American Journal of Medical Genetics, Part A, 2021, 185, 978-981.	0.7	2
5	Disruption of RFX family transcription factors causes autism, attention-deficit/hyperactivity disorder, intellectual disability, and dysregulated behavior. Genetics in Medicine, 2021, 23, 1028-1040.	1.1	34
6	Metacarpophalangeal profile pattern analysis in a further patient with a novel ARID1B variant. Congenital Anomalies (discontinued), 2021, 61, 193-196.	0.3	1
7	Clinical and Molecular Aspects of the Neurodevelopmental Disorder Associated with PAK3 Perturbation. Journal of Molecular Neuroscience, 2021, 71, 2474-2481.	1.1	3
8	Clinical presentation and molecular characterization of a novel patient with variant <scp>POC1A</scp>â€related syndrome. Clinical Genetics, 2021, 99, 540-546.	1.0	7
9	Striking phenotypic overlap between Nicolaides-Baraitser and Coffin-Siris syndromes in monozygotic twins with ARID1B intragenic deletion. European Journal of Medical Genetics, 2020, 63, 103739.	0.7	8
10	Episignatures Stratifying Helsmoortel-Van Der Aa Syndrome Show Modest Correlation with Phenotype. American Journal of Human Genetics, 2020, 107, 555-563.	2.6	32
11	A novel patient with <scp>Whiteâ€“Sutton</scp> syndrome refines the mutational and clinical repertoire of the POGZâ€related phenotype and suggests further observations. American Journal of Medical Genetics, Part A, 2020, 182, 1791-1795.	0.7	10
12	Further delineation of the neurodevelopmental phenotypic spectrum associated to 14q11.2 microduplication. Neurological Sciences, 2020, 41, 3751-3753.	0.9	3
13	Answer to Letter to the Editor regarding the article â€œStriking phenotypic overlap between Nicolaides-Baraitser and Coffin-Siris syndromes in monozygotic twins with ARID1B intragenic deletionâ€. European Journal of Medical Genetics, 2020, 63, 103993.	0.7	0
14	The p.Arg377Trp variant in ACTL6A underlines a recognizable BAFâ€opathy phenotype. Clinical Genetics, 2020, 97, 672-674.	1.0	4
15	Structural modeling of a novel TERC variant in a patient with aplastic anemia and short telomeres. Annals of Hematology, 2019, 98, 805-807.	0.8	1
16	The facial dysmorphology analysis technology in intellectual disability syndromes related to defects in the histones modifiers. Journal of Human Genetics, 2019, 64, 721-728.	1.1	8
17	Autism spectrum disorder in a patient with a genomic rearrangement that only involves the EPHA5 gene. Psychiatric Genetics, 2019, 29, 86-90.	0.6	5
18	Helsmoortel-Van der Aa Syndrome as emerging clinical diagnosis in intellectually disabled children with autistic traits and ocular involvement. European Journal of Paediatric Neurology, 2018, 22, 552-557.	0.7	19

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19	Incidental finding of an Xq microdeletion in a girl with trichorhinophalangeal syndrome type I harboring a novel TRPS1 nonsense mutation. Minerva Pediatrica, 2018, 70, 639-642.	2.6	0
20	Lack of pathogenic mutations in SOS1 gene in phenytoin-induced gingival overgrowth patients. Archives of Oral Biology, 2017, 80, 160-163.	0.8	3
21	Axial skeletogenesis in human autosomal aneuploidies: A radiographic study of 145 second trimester fetuses. American Journal of Medical Genetics, Part A, 2016, 170, 676-687.	0.7	11
22	Microcephaly, ectodermal dysplasia, multiple skeletal anomalies and distinctive facial appearance: Delineation of cerebroâ€dermatoâ€osseousâ€dysplasia. American Journal of Medical Genetics, Part A, 2015, 167, 842-851.	0.7	1
23	Gastrointestinal and nutritional issues in joint hypermobility syndrome/ehlersâ€“danlos syndrome, hypermobility type. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2015, 169, 54-75.	0.7	76
24	Clinical and genetic study of two patients with Zimmermannâ€“Laband syndrome and literature review. European Journal of Medical Genetics, 2013, 56, 570-576.	0.7	32