Dario Cocciadiferro

List of Publications by Year
in descending order

14	324	1163117 8	1058476 14
papers	citations	h-index	g-index

16	16	16	814
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Expanding the novel <scp> MAPKAPK5 </scp> â€“related developmental disorderâ€™s genotypeâ€“phenotype correlation: patient report and 19 months followâ€up. Clinical Genetics, 2022, , .	2.0	2
2	Clinical presentation and molecular characterization of a novel patient with variant <scp>POC1A</scp>â€related syndrome. Clinical Genetics, 2021, 99, 540-546.	2.0	7
3	A case report of isoniazid adverse drug reaction in a pediatric patient with defective NAT2 gene. Medicine, Case Reports and Study Protocols, 2021, 2, e0043.	0.1	0
4	Expanding the clinical and molecular spectrum of lethal congenital contracture syndrome 8 associated with biallelic variants of ADCY6. Clinical Genetics, 2020, 97, 649-654.	2.0	4
5	COL1â€related overlap disorder: A novel connective tissue disorder incorporating the osteogenesis imperfecta/Ehlersâ€Danlos syndrome overlap. Clinical Genetics, 2020, 97, 396-406.	2.0	27
6	Genetic identification and molecular modeling characterization of a novel POU3F4 variant in two Italian deaf brothers. International Journal of Pediatric Otorhinolaryngology, 2020, 129, 109790.	1.0	8
7	Novel exostosinâ€2 missense variants in a family with autosomal recessive exostosinâ€2â€related syndrome: further evidences on the phenotype. Clinical Genetics, 2019, 95, 165-171.	2.0	3
8	TRIM50 regulates Beclin 1 proautophagic activity. Biochimica Et Biophysica Acta - Molecular Cell Research, 2018, 1865, 908-919.	4.1	39
9	Dissecting KMT2D missense mutations in Kabuki syndrome patients. Human Molecular Genetics, 2018, 27, 3651-3668.	2.9	49
10	Clinical and Neurobehavioral Features of Three Novel Kabuki Syndrome Patients with Mosaic KMT2D Mutations and a Review of Literature. International Journal of Molecular Sciences, 2018, 19, 82.	4.1	19
11	A novel MED12 mutation: Evidence for a fourth phenotype. American Journal of Medical Genetics, Part A, 2016, 170, 2377-2382.	1.2	31
12	GNB5 Mutations Cause an Autosomal-Recessive Multisystem Syndrome with Sinus Bradycardia and Cognitive Disability. American Journal of Human Genetics, 2016, 99, 704-710.	6.2	58
13	TRIM8 downregulation in glioma affects cell proliferation and it is associated with patients survival. BMC Cancer, 2015, 15, 470.	2.6	61
14	Genomic and Genetic Disorders Biobank. Open Journal of Bioresources, 2015, 2, .	1.5	1