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	TRIM8 downregulation in glioma affects cell proliferation and it is associated with patients survival. BMC Cancer, 2015, 15, 470.	2.6	61
2	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
3	Dissecting KMT2D missense mutations in Kabuki syndrome patients. Human Molecular Genetics, 2018, 27, 3651-3668.	2.9	49
4	TRIM50 regulates Beclin 1 proautophagic activity. Biochimica Et Biophysica Acta - Molecular Cell Research, 2018, 1865, 908-919.	4.1	39
5	A novel MED12 mutation: Evidence for a fourth phenotype. American Journal of Medical Genetics, Part A, 2016, 170, 2377-2382.	1.2	31
6	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
7	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
8	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
9	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
10	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
11	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
12	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
13	Genomic and Genetic Disorders Biobank. Open Journal of Bioresources, 2015, 2, .	1.5	1
14	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