Castilla-Vallmanya Laura

List of Publications by Year
in descending order

11 papers	165 citations	1478505 6 h-index	1281871 11 g-index
12 all docs	12 docs citations	12 times ranked	381 citing authors

#	Article	IF	Citations
1	Detecting and targeting senescent cells using molecularly imprinted nanoparticles. Nanoscale Horizons, 2019, 4, 757-768.	8.0	67
2	Phenotypic spectrum and transcriptomic profile associated with germline variants in TRAF7. Genetics in Medicine, 2020, 22, 1215-1226.	2.4	22
3	Five new cases of syndromic intellectual disability due to KAT6A mutations: widening the molecular and clinical spectrum. Orphanet Journal of Rare Diseases, 2020, 15, 44.	2.7	18
4	DPH1 syndrome: two novel variants and structural and functional analyses of seven missense variants identified in syndromic patients. European Journal of Human Genetics, 2020, 28, 64-75.	2.8	15
5	A De Novo FOXP1 Truncating Mutation in a Patient Originally Diagnosed as C Syndrome. Scientific Reports, 2018, 8, 694.	3.3	11
6	Neuronal and Astrocytic Differentiation from Sanfilippo C Syndrome iPSCs for Disease Modeling and Drug Development. Journal of Clinical Medicine, 2020, 9, 644.	2.4	10
7	The ASXL1 mutation p.Gly646Trpfs*12 found in a Turkish boy with Bohringâ€Opitz Syndrome. Clinical Case Reports (discontinued), 2018, 6, 1452-1456.	0.5	6
8	Case report of a child bearing a novel deleterious splicing variant in PIGT. Medicine (United States), 2019, 98, e14524.	1.0	5
9	Extending the phenotypic spectrum of Bohringâ€Opitz syndrome: Mild case confirmed by functional studies. American Journal of Medical Genetics, Part A, 2020, 182, 201-204.	1.2	5
10	De Novo PORCN and ZIC2 Mutations in a Highly Consanguineous Family. International Journal of Molecular Sciences, 2021, 22, 1549.	4.1	4
11	Understanding the Pathophysiology and Searching for Biomarkers for Rare Genetic Developmental Diseases. Proceedings (mdpi), 2019, 22, 53.	0.2	0