

Castilla-Vallmanya Laura

List of Publications by Year in descending order

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Version: 2024-02-01

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	De Novo PORCN and ZIC2 Mutations in a Highly Consanguineous Family. International Journal of Molecular Sciences, 2021, 22, 1549.	4.1	4
2	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
3	Extending the phenotypic spectrum of Bohring-Opitz syndrome: Mild case confirmed by functional studies. American Journal of Medical Genetics, Part A, 2020, 182, 201-204.	1.2	5
4	Phenotypic spectrum and transcriptomic profile associated with germline variants in TRAF7. Genetics in Medicine, 2020, 22, 1215-1226.	2.4	22
5	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
6	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
7	Understanding the Pathophysiology and Searching for Biomarkers for Rare Genetic Developmental Diseases. Proceedings (mdpi), 2019, 22, 53.	0.2	0
8	Detecting and targeting senescent cells using molecularly imprinted nanoparticles. Nanoscale Horizons, 2019, 4, 757-768.	8.0	67
9	Case report of a child bearing a novel deleterious splicing variant in PIGT. Medicine (United States), 2019, 98, e14524.	1.0	5
10	A De Novo FOXP1 Truncating Mutation in a Patient Originally Diagnosed as C Syndrome. Scientific Reports, 2018, 8, 694.	3.3	11
11	The <i>ASXL1</i> mutation p.Gly646Trpfs*12 found in a Turkish boy with Bohring-Opitz Syndrome. Clinical Case Reports (discontinued), 2018, 6, 1452-1456.	0.5	6