MarÃ-a Palomares-Bralo

List of Publications by Year
in descending order

36 papers	707 citations	566801 15 h-index	610482 24 g-index
37 all docs	37 docs citations	37 times ranked	1605 citing authors

#	Article	IF	Citations
1	Novel diagnostic DNA methylation episignatures expand and refine the epigenetic landscapes of Mendelian disorders. Human Genetics and Genomics Advances, 2022, 3, 100075.	1.0	42
2	Heterozygous variants in PRPF8 are associated with neurodevelopmental disorders. American Journal of Medical Genetics, Part A, 2022, 188, 2750-2759.	0.7	4
3	Variability in Phelan-McDermid Syndrome in a Cohort of 210 Individuals. Frontiers in Genetics, 2022, 13, 652454.	1.1	27
4	Molecular and histologic insights on early onset cardiomyopathy in Danon disease females. Clinical Genetics, 2021, 99, 481-483.	1.0	3
5	Delineation of the clinical and radiological features of <scp>Stuveâ€“Wiedemann</scp> syndrome childhood survivors, four new cases and review of the literature. American Journal of Medical Genetics, Part A, 2021, 185, 856-865.	0.7	4
6	Expanding the Phenotypic Spectrum of PAX6 Mutations: From Congenital Cataracts to Nystagmus. Genes, 2021, 12, 707.	1.0	8
7	Truncating SRCAP variants outside the Floating-Harbor syndrome locus cause a distinct neurodevelopmental disorder with a specific DNA methylation signature. American Journal of Human Genetics, 2021, 108, 1053-1068.	2.6	31
8	Phenotypic expansion of CACNA1C-associated disorders to include isolated neurological manifestations. Genetics in Medicine, 2021, 23, 1922-1932.	1.1	16
9	ZTTK syndrome: Clinical and molecular findings ofÂ15 cases and a review of the literature. American Journal of Medical Genetics, Part A, 2021, 185, 3740-3753.	0.7	11
10	Mosaic Variegated Aneuploidy syndrome 2 caused by biallelic variants in CEP57, two new cases and review of the phenotype. European Journal of Medical Genetics, 2021, 64, 104338.	0.7	5
11	Further delineation of neuropsychiatric findings in Tatton-Brown-Rahman syndrome due to disease-causing variants in DNMT3A: seven new patients. European Journal of Human Genetics, 2020, 28, 469-479.	1.4	16
12	Biallelic MADD variants cause a phenotypic spectrum ranging from developmental delay to a multisystem disorder. Brain, 2020, 143, 2437-2453.	3.7	21
13	Pathogenic variants in <scp>KPTN</scp>, a rare cause of macrocephaly and intellectual disability. American Journal of Medical Genetics, Part A, 2020, 182, 2222-2225.	0.7	6
14	Implementation of chromosomal microarrays in a cohort of patients with intellectual disability at the Argentinean public health system. Molecular Biology Reports, 2020, 47, 6863-6878.	1.0	2
15	Skin and nails abnormalities in a patient with ZTTK syndrome and a de novo mutation in SON. Pediatric Dermatology, 2020, 37, 517-519.	0.5	11
16	MAGEL2â€related disorders: A study and case series. Clinical Genetics, 2019, 96, 493-505.	1.0	26
17	Variable expressivity of syndromic BMP4-related eye, brain, and digital anomalies: A review of the literature and description of three new cases. European Journal of Human Genetics, 2019, 27, 1379-1388.	1.4	8
18	Exome sequencing in Crisponi/coldâ€induced sweating syndromeâ€“like individuals reveals unpredicted alternative diagnoses. Clinical Genetics, 2019, 95, 607-614.	1.0	7

#	Article	IF	Citations
19	The Brain-Lung-Thyroid syndrome (BLTS): A novel deletion in chromosome 14q13.2-q21.1 expands the phenotype to humoral immunodeficiency. European Journal of Medical Genetics, 2018, 61, 393-398.	0.7	10
20	mTOR mutations in Smithâ€Kingsmore syndrome: Four additional patients and a review. Clinical Genetics, 2018, 93, 762-775.	1.0	36
21	In-frame Variants in FLNA Proximal Rod 1 Domain Associate With a Predominant Cardiac Valvular Phenotype. Revista Espanola De Cardiologia (English Ed), 2018, 71, 545-552.	0.4	1
22	Refining the phenotype associated with GNB1 mutations: Clinical data on 18 newly identified patients and review of the literature. American Journal of Medical Genetics, Part A, 2018, 176, 2259-2275.	0.7	47
23	Eye coloboma and complex cardiac malformations belong to the clinical spectrum of <scp>PUF60</scp> variants. Clinical Genetics, 2017, 92, 350-351.	1.0	18
24	Copy number variants of Ras/<scp>MAPK</scp> pathway genes in patients with isolated cryptorchidism. Andrology, 2017, 5, 923-930.	1.9	4
25	FOXP1-related intellectual disability syndrome: a recognisable entity. Journal of Medical Genetics, 2017, 54, 613-623.	1.5	48
26	Pitfalls of trio-based exome sequencing: imprinted genes and parental mosaicismâ€"MAGEL2 as an example. Genetics in Medicine, 2017, 19, 1283-1285.	1.1	10
27	Clinical and molecular analyses of Beckwithâ€"Wiedemann syndrome: Comparison between spontaneous conception and assisted reproduction techniques. American Journal of Medical Genetics, Part A, 2016, 170, 2740-2749.	0.7	30
28	Array CGH Analysis of Paired Blood and Tumor Samples from Patients with Sporadic Wilms Tumor. PLoS ONE, 2015, 10, e0136812.	1.1	8
29	PIAS4 is associated with macro/microcephaly in the novel interstitial 19p13.3 microdeletion/microduplication syndrome. European Journal of Human Genetics, 2015, 23, 1615-1626.	1.4	29
30	New microdeletion and microduplication syndromes: a comprehensive review. Genetics and Molecular Biology, 2014, 37, 210-219.	0.6	84
31	Familial imbalance in 16p13.11 leads to a dosage compensation rearrangement in an unaffected carrier. BMC Medical Genetics, 2014, 15, 116.	2.1	3
32	A New Overgrowth Syndrome is due to Mutations inRNF125. Human Mutation, 2014, 35, 1436-1441.	1.1	33
33	A parallel study of different array-CGH platforms in a set of Spanish patients with developmental delay and intellectual disability. Gene, 2013, 521, 82-86.	1.0	15
34	Customized high resolution CGHâ€array for clinical diagnosis reveals additional genomic imbalances in previous wellâ€defined pathological samples. American Journal of Medical Genetics, Part A, 2013, 161, 1950-1960.	0.7	32
35	Characterization of a 8q21.11 Microdeletion Syndrome Associated with Intellectual Disability and a Recognizable Phenotype. American Journal of Human Genetics, 2011, 89, 295-301.	2.6	37
36	Direct tandem duplication in chromosome 19q characterized by array CGH. European Journal of Medical Genetics, 2008, 51, 257-263.	0.7	13