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List of Publications by Year in descending order

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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	New microdeletion and microduplication syndromes: a comprehensive review. <i>Genetics and Molecular Biology</i> , 2014, 37, 210-219.	0.6	84
2	<i>FOXP1</i>-related intellectual disability syndrome: a recognisable entity. <i>Journal of Medical Genetics</i> , 2017, 54, 613-623.	1.5	48
3	Refining the phenotype associated with <i>GNB1</i> mutations: Clinical data on 18 newly identified patients and review of the literature. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 2259-2275.	0.7	47
4	Novel diagnostic DNA methylation epesignatures expand and refine the epigenetic landscapes of Mendelian disorders. <i>Human Genetics and Genomics Advances</i> , 2022, 3, 100075.	1.0	42
5	Characterization of a 8q21.11 Microdeletion Syndrome Associated with Intellectual Disability and a Recognizable Phenotype. <i>American Journal of Human Genetics</i> , 2011, 89, 295-301.	2.6	37
6	<i>mTOR</i> mutations in Smithâ€Kingsmore syndrome: Four additional patients and a review. <i>Clinical Genetics</i> , 2018, 93, 762-775.	1.0	36
7	A New Overgrowth Syndrome is due to Mutations in <i>RNF125</i>. <i>Human Mutation</i> , 2014, 35, 1436-1441.	1.1	33
8	Customized high resolution CGHâ€array for clinical diagnosis reveals additional genomic imbalances in previously wellâ€defined pathological samples. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 1950-1960.	0.7	32
9	Truncating SRCAP variants outside the Floating-Harbor syndrome locus cause a distinct neurodevelopmental disorder with a specific DNA methylation signature. <i>American Journal of Human Genetics</i> , 2021, 108, 1053-1068.	2.6	31
10	Clinical and molecular analyses of Beckwithâ€Wiedemann syndrome: Comparison between spontaneous conception and assisted reproduction techniques. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 2740-2749.	0.7	30
11	PIAS4 is associated with macro/microcephaly in the novel interstitial 19p13.3 microdeletion/microduplication syndrome. <i>European Journal of Human Genetics</i> , 2015, 23, 1615-1626.	1.4	29
12	Variability in Phelan-McDermid Syndrome in a Cohort of 210 Individuals. <i>Frontiers in Genetics</i> , 2022, 13, 652454.	1.1	27
13	<i>MAGEL2</i>-related disorders: A study and case series. <i>Clinical Genetics</i> , 2019, 96, 493-505.	1.0	26
14	Biallelic MADD variants cause a phenotypic spectrum ranging from developmental delay to a multisystem disorder. <i>Brain</i> , 2020, 143, 2437-2453.	3.7	21
15	Eye coloboma and complex cardiac malformations belong to the clinical spectrum of <i>PUF60</i> variants. <i>Clinical Genetics</i> , 2017, 92, 350-351.	1.0	18
16	Further delineation of neuropsychiatric findings in Tatton-Brown-Rahman syndrome due to disease-causing variants in DNMT3A: seven new patients. <i>European Journal of Human Genetics</i> , 2020, 28, 469-479.	1.4	16
17	Phenotypic expansion of CACNA1C-associated disorders to include isolated neurological manifestations. <i>Genetics in Medicine</i> , 2021, 23, 1922-1932.	1.1	16
18	A parallel study of different array-CGH platforms in a set of Spanish patients with developmental delay and intellectual disability. <i>Gene</i> , 2013, 521, 82-86.	1.0	15

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19	Direct tandem duplication in chromosome 19q characterized by array CGH. <i>European Journal of Medical Genetics</i> , 2008, 51, 257-263.	0.7	13
20	Skin and nails abnormalities in a patient with ZTTK syndrome and a de novo mutation in <i>SON</i> . <i>Pediatric Dermatology</i> , 2020, 37, 517-519.	0.5	11
21	ZTTK syndrome: Clinical and molecular findings of 15 cases and a review of the literature. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 3740-3753.	0.7	11
22	Pitfalls of trio-based exome sequencing: imprinted genes and parental mosaicism—MAGEL2 as an example. <i>Genetics in Medicine</i> , 2017, 19, 1283-1285.	1.1	10
23	The Brain-Lung-Thyroid syndrome (BLTS): A novel deletion in chromosome 14q13.2-q21.1 expands the phenotype to humoral immunodeficiency. <i>European Journal of Medical Genetics</i> , 2018, 61, 393-398.	0.7	10
24	Array CGH Analysis of Paired Blood and Tumor Samples from Patients with Sporadic Wilms Tumor. <i>PLoS ONE</i> , 2015, 10, e0136812.	1.1	8
25	Variable expressivity of syndromic BMP4-related eye, brain, and digital anomalies: A review of the literature and description of three new cases. <i>European Journal of Human Genetics</i> , 2019, 27, 1379-1388.	1.4	8
26	Expanding the Phenotypic Spectrum of PAX6 Mutations: From Congenital Cataracts to Nystagmus. <i>Genes</i> , 2021, 12, 707.	1.0	8
27	Exome sequencing in Crisponi/cold-induced sweating syndrome-like individuals reveals unpredicted alternative diagnoses. <i>Clinical Genetics</i> , 2019, 95, 607-614.	1.0	7
28	Pathogenic variants in <i>KPTN</i> , a rare cause of macrocephaly and intellectual disability. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 2222-2225.	0.7	6
29	Mosaic Variegated Aneuploidy syndrome 2 caused by biallelic variants in CEP57, two new cases and review of the phenotype. <i>European Journal of Medical Genetics</i> , 2021, 64, 104338.	0.7	5
30	Copy number variants of Ras/MAPK pathway genes in patients with isolated cryptorchidism. <i>Andrology</i> , 2017, 5, 923-930.	1.9	4
31	Delineation of the clinical and radiological features of Stuve-Wiedemann syndrome childhood survivors, four new cases and review of the literature. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 856-865.	0.7	4
32	Heterozygous variants in <i>PRPF8</i> are associated with neurodevelopmental disorders. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 2750-2759.	0.7	4
33	Familial imbalance in 16p13.11 leads to a dosage compensation rearrangement in an unaffected carrier. <i>BMC Medical Genetics</i> , 2014, 15, 116.	2.1	3
34	Molecular and histologic insights on early onset cardiomyopathy in Danon disease females. <i>Clinical Genetics</i> , 2021, 99, 481-483.	1.0	3
35	Implementation of chromosomal microarrays in a cohort of patients with intellectual disability at the Argentinean public health system. <i>Molecular Biology Reports</i> , 2020, 47, 6863-6878.	1.0	2
36	In-frame Variants in FLNA Proximal Rod 1 Domain Associate With a Predominant Cardiac Valvular Phenotype. <i>Revista Espanola De Cardiologia (English Ed)</i> , 2018, 71, 545-552.	0.4	1