

John A Bernat

List of Publications by Year in descending order

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Version: 2024-02-01

15
papers

1,104
citations

759233

12
h-index

1058476

14
g-index

15
all docs

15
docs citations

15
times ranked

2164
citing authors

#	ARTICLE	IF	CITATIONS
1	The diagnosis and management of Gaucher disease in pediatric patients: Where do we go from here?. <i>Molecular Genetics and Metabolism</i> , 2022, 136, 4-21.	1.1	18
2	Ensuring best practice in genomics education and evaluation: reporting item standards for education and its evaluation in genomics (RISE2 Genomics). <i>Genetics in Medicine</i> , 2021, 23, 1356-1365.	2.4	17
3	Genotype-phenotype analysis of 523 patients by genetics evaluation and clinical exome sequencing. <i>Pediatric Research</i> , 2020, 87, 735-739.	2.3	28
4	Niemann-Pick disease type C presenting as very early onset inflammatory bowel disease. <i>BMJ Case Reports</i> , 2019, 12, e229780.	0.5	8
5	Genotype-phenotype correlations in individuals with pathogenic RERE variants. <i>Human Mutation</i> , 2018, 39, 666-675.	2.5	34
6	Genetic analysis of CHARGE syndrome identifies overlapping molecular biology. <i>Genetics in Medicine</i> , 2018, 20, 1022-1029.	2.4	43
7	WDR26 Haploinsufficiency Causes a Recognizable Syndrome of Intellectual Disability, Seizures, Abnormal Gait, and Distinctive Facial Features. <i>American Journal of Human Genetics</i> , 2017, 101, 139-148.	6.2	45
8	Recurrent deletions and duplications of chromosome 2q11.2 and 2q13 are associated with variable outcomes. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 2664-2673.	1.2	42
9	Functional analysis of candidate genes in 2q13 deletion syndrome implicates FBLN7 and TMEM87B deficiency in congenital heart defects and FBLN7 in craniofacial malformations. <i>Human Molecular Genetics</i> , 2014, 23, 4272-4284.	2.9	41
10	Breast fibroadenomas in the pediatric population: common and uncommon sonographic findings. <i>Pediatric Radiology</i> , 2010, 40, 1681-1689.	2.0	50
11	Genome-wide mapping of DNase hypersensitive sites using massively parallel signature sequencing (MPSS). <i>Genome Research</i> , 2006, 16, 123-131.	5.5	431
12	Distant conserved sequences flanking endothelial-specific promoters contain tissue-specific DNase-hypersensitive sites and over-represented motifs. <i>Human Molecular Genetics</i> , 2006, 15, 2098-2105.	2.9	20
13	Comparative Genomic Analysis of the 5'-Upstream Sequences of von Willebrand Factor and 27 Other Endothelial-Specific Genes Identifies Conserved Noncoding Sequences That Are DNase Hypersensitive.. <i>Blood</i> , 2005, 106, 2650-2650.	1.4	0
14	Bleeding due to disruption of a cargo-specific ER-to-Golgi transport complex. <i>Nature Genetics</i> , 2003, 34, 220-225.	21.4	282
15	Heterochromatin protein 2 (HP2), a partner of HP1 in Drosophila heterochromatin. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2002, 99, 14332-14337.	7.1	45