John A Bernat

List of Publications by Year
in descending order

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