

Miguel Fernandez-Burriel

List of Publications by Citations

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

9

papers

176

citations

5

h-index

10

g-index

10

ext. papers

212

ext. citations

4.6

avg, IF

1.41

L-index

#	Paper	IF	Citations
9	Mutation update of spinal muscular atrophy in Spain: molecular characterization of 745 unrelated patients and identification of four novel mutations in the SMN1 gene. <i>Human Genetics</i> , 2009 , 125, 29-39	6.3	104
8	MLPA as first screening method for the detection of microduplications and microdeletions in patients with X-linked mental retardation. <i>Genetics in Medicine</i> , 2007 , 9, 117-22	8.1	29
7	High-throughput sequencing for the molecular diagnosis of Usher syndrome reveals 42 novel mutations and consolidates CEP250 as Usher-like disease causative. <i>Scientific Reports</i> , 2018 , 8, 17113	4.9	20
6	Xq26.2-q26.3 microduplication in two brothers with intellectual disabilities: clinical and molecular characterization. <i>Journal of Human Genetics</i> , 2010 , 55, 822-6	4.3	10
5	Detection of the fragile X syndrome protein for the evaluation of FMR1 intermediate alleles. <i>Human Genetics</i> , 2000 , 107, 195-6	6.3	6
4	A novel delins mutation in the alpha-TTP gene in a family segregating ataxia with isolated vitamin E deficiency. <i>Pediatric Research</i> , 2008 , 64, 262-4	3.2	4
3	A simple method of screening for the common connexin-26 gene 35delG mutation in nonsyndromic neurosensory autosomal recessive deafness. <i>Genetic Testing and Molecular Biomarkers</i> , 2003 , 7, 147-9		1
2	Polyvariant Mutant Genes: different haplotypes determining different alterations causing azoospermia. <i>Gene Function & Disease</i> , 2000 , 1, 189-193		1
1	TBL1XR1 associated intellectual disability, a new missense variant with dysmorphic features plus autism: Expanding the phenotypic spectrum. <i>Clinical Genetics</i> , 2021 , 99, 812-817	4	1