Miguel Fernandez-Burriel

List of Publications by Year in Descending Order

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