Pilar CamaÃ±o

List of Publications by Year
in descending order

18 papers	1,854 citations	687220 13 h-index	940416 16 g-index
18 all docs	18 docs citations	18 times ranked	2020 citing authors

#	Article	IF	Citations
1	A Unifying Genetic Model for Facioscapulohumeral Muscular Dystrophy. Science, 2010, 329, 1650-1653.	6.0	638
2	Clinical features of facioscapulohumeral muscular dystrophy 2. Neurology, 2010, 75, 1548-1554.	1.5	215
3	LGMD2A: genotypeâ€“phenotype correlations based on a large mutational survey on the calpain 3 gene. Brain, 2005, 128, 732-742.	3.7	197
4	Mitochondrial DNA depletion. Neurology, 2002, 59, 1197-1202.	1.5	156
5	Mitochondrial DNA depletion anddGKgene mutations. Annals of Neurology, 2002, 52, 311-317.	2.8	152
6	Inter-individual differences in CpG methylation at D4Z4 correlate with clinical variability in FSHD1 and FSHD2. Human Molecular Genetics, 2015, 24, 659-669.	1.4	130
7	X-Linked Dominant Scapuloperoneal Myopathy Is Due to a Mutation in the Gene Encoding Four-and-a-Half-LIM Protein 1. American Journal of Human Genetics, 2008, 82, 208-213.	2.6	108
8	Clinical outcome in 19 French and Spanish patients with valosin-containing protein myopathy associated with Pagetâ€™s disease of bone and frontotemporal dementia. Neuromuscular Disorders, 2009, 19, 316-323.	0.3	79
9	Patients with a phenotype consistent with facioscapulohumeral muscular dystrophy display genetic and epigenetic heterogeneity. Journal of Medical Genetics, 2012, 49, 41-46.	1.5	55
10	CD24 V/V is an allele associated with the risk of developing multiple sclerosis in the Spanish population. Multiple Sclerosis Journal, 2006, 12, 511-514.	1.4	40
11	Hybridization analysis of D4Z4 repeat arrays linked to FSHD. Chromosoma, 2007, 116, 107-116.	1.0	25
12	Double SMCHD1 variants in FSHD2: the synergistic effect of two SMCHD1 variants on D4Z4 hypomethylation and disease penetrance in FSHD2. European Journal of Human Genetics, 2016, 24, 78-85.	1.4	23
13	Deep phenotyping of facioscapulohumeral muscular dystrophy type 2 by magnetic resonance imaging. European Journal of Neurology, 2020, 27, 2604-2615.	1.7	16
14	Novel valosin containing protein mutation in a Swiss family with hereditary inclusion body myopathy and dementia. Neuromuscular Disorders, 2013, 23, 149-154.	0.3	11
15	Cognitive function in facioscapulohumeral dystrophy correlates with the molecular defect. Genes, Brain and Behavior, 2009, 8, 53-59.	1.1	5
16	Longitudinal Clinical Follow-up of a Large Family With the R357P Twinkle Mutation. JAMA Neurology, 2013, 70, 1425.	4.5	4
17	Erratum to â€˜Clinical outcome in 19 French and Spanish patients with valosin-containing protein myopathy associated with Pagetâ€™s disease of bone and frontotemporal dementiaâ€™ [Neuromuscular Disorders 19 (2009) 316â€“323]. Neuromuscular Disorders, 2011, 21, e1.	0.3	0
18	Rapidly Reversible Winging Scapula. Arthritis and Rheumatology, 2015, 67, 2502-2502.	2.9	0