

Pilar Camañó

List of Publications by Year in descending order

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Version: 2024-02-01

18
papers

1,854
citations

687220

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940416

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times ranked

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