Valerie Biancalana

List of Publications by Year
in descending order

22 papers	1,483 citations	567281 15 h-index	713466 21 g-index
22 all docs	22 docs citations	22 times ranked	1329 citing authors

#	Article	IF	Citations
1	Selective loss of a LAP1 isoform causes a muscle-specific nuclear envelopathy. Neurogenetics, 2021, 22, 33-41.	1.4	3
2	A recurrent RYR1 mutation associated with early-onset hypotonia and benign disease course. Acta Neuropathologica Communications, 2021, 9, 155.	5.2	1
3	Clinical, histological, and genetic characterization of PYROXD1-related myopathy. Acta Neuropathologica Communications, 2019, 7, 138.	5.2	14
4	Some DNM2 mutations cause extremely severe congenital myopathy and phenocopy myotubular myopathy. Acta Neuropathologica Communications, 2018, 6, 93.	5.2	14
5	Common and variable clinical, histological, and imaging findings of recessive RYR1-related centronuclear myopathy patients. Neuromuscular Disorders, 2017, 27, 975-985.	0.6	34
6	Affected female carriers of MTM1 mutations display a wide spectrum of clinical and pathological involvement: delineating diagnostic clues. Acta Neuropathologica, 2017, 134, 889-904.	7.7	42
7	A Study of a Cohort of X-Linked Myotubular Myopathy at the Clinical, Histologic, and Genetic Levels. Pediatric Neurology, 2016, 58, 107-112.	2.1	13
8	Integrative Data Mining Highlights Candidate Genes for Monogenic Myopathies. PLoS ONE, 2014, 9, e110888.	2.5	16
9	Adult-onset autosomal dominant centronuclear myopathy due to BIN1 mutations. Brain, 2014, 137, 3160-3170.	7.6	76
10	Extensive morphological and immunohistochemical characterization in myotubular myopathy. Brain and Behavior, 2013, 3, 476-486.	2.2	25
11	Clinical utility gene card for: Centronuclear and myotubular myopathies. European Journal of Human Genetics, 2012, 20, 1101-1101.	2.8	28
12	Mutation spectrum in the large GTPase dynamin 2, and genotype-phenotype correlation in autosomal dominant centronuclear myopathy. Human Mutation, 2012, 33, 949-959.	2.5	115
13	Expanding the clinical, pathological and MRI phenotype of DNM2-related centronuclear myopathy. Neuromuscular Disorders, 2010, 20, 229-237.	0.6	100
14	Novel molecular diagnostic approaches for X-linked centronuclear (myotubular) myopathy reveal intronic mutations. Neuromuscular Disorders, 2010, 20, 375-381.	0.6	33
15	â€œNecklaceâ€•fibers, a new histological marker of late-onset MTM1-related centronuclear myopathy. Acta Neuropathologica, 2009, 117, 283-291.	7.7	106
16	Diagnosis of myotubular myopathy in the oldest known manifesting female carrier: A clinical and genetic study. Neuromuscular Disorders, 2007, 17, 180-185.	0.6	24
17	Subtle central and peripheral nervous system abnormalities in a family with centronuclear myopathy and a novel dynamin 2 gene mutation. Neuromuscular Disorders, 2007, 17, 955-959.	0.6	51
18	Mutations in amphiphysin 2 (BIN1) disrupt interaction with dynamin 2 and cause autosomal recessive centronuclear myopathy. Nature Genetics, 2007, 39, 1134-1139.	21.4	353

#	Article	IF	Citations
19	Fragile X Disease. , 2004, 92, 157-182.		1
20	Characterisation of mutations in 77 patients with X-linked myotubular myopathy, including a family with a very mild phenotype. Human Genetics, 2003, 112, 135-142.	3.8	113
21	Genotypeâ€“phenotype correlations in X-linked myotubular myopathy. Neuromuscular Disorders, 2002, 12, 939-946.	0.6	122
22	MTM1 mutations in X-linked myotubular myopathy. Human Mutation, 2000, 15, 393-409.	2.5	199