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	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
2	MTM1 mutations in X-linked myotubular myopathy. Human Mutation, 2000, 15, 393-409.	2.5	199
3	Genotypeâ€“phenotype correlations in X-linked myotubular myopathy. Neuromuscular Disorders, 2002, 12, 939-946.	0.6	122
4	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
5	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
6	â€œNecklaceâ€•fibers, a new histological marker of late-onset MTM1-related centronuclear myopathy. Acta Neuropathologica, 2009, 117, 283-291.	7.7	106
7	Expanding the clinical, pathological and MRI phenotype of DNM2-related centronuclear myopathy. Neuromuscular Disorders, 2010, 20, 229-237.	0.6	100
8	Adult-onset autosomal dominant centronuclear myopathy due to BIN1 mutations. Brain, 2014, 137, 3160-3170.	7.6	76
9	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
10	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
11	Common and variable clinical, histological, and imaging findings of recessive RYR1-related centronuclear myopathy patients. Neuromuscular Disorders, 2017, 27, 975-985.	0.6	34
12	Novel molecular diagnostic approaches for X-linked centronuclear (myotubular) myopathy reveal intronic mutations. Neuromuscular Disorders, 2010, 20, 375-381.	0.6	33
13	Clinical utility gene card for: Centronuclear and myotubular myopathies. European Journal of Human Genetics, 2012, 20, 1101-1101.	2.8	28
14	Extensive morphological and immunohistochemical characterization in myotubular myopathy. Brain and Behavior, 2013, 3, 476-486.	2.2	25
15	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
16	Integrative Data Mining Highlights Candidate Genes for Monogenic Myopathies. PLoS ONE, 2014, 9, e110888.	2.5	16
17	Some DNM2 mutations cause extremely severe congenital myopathy and phenocopy myotubular myopathy. Acta Neuropathologica Communications, 2018, 6, 93.	5.2	14
18	Clinical, histological, and genetic characterization of PYROXD1-related myopathy. Acta Neuropathologica Communications, 2019, 7, 138.	5.2	14

#	Article	IF	Citations
19	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
20	Selective loss of a LAP1 isoform causes a muscle-specific nuclear envelopathy. Neurogenetics, 2021, 22, 33-41.	1.4	3
21	Fragile X Disease. , 2004, 92, 157-182.		1
22	A recurrent RYR1 mutation associated with early-onset hypotonia and benign disease course. Acta Neuropathologica Communications, 2021, 9, 155.	5.2	1