Fabiana Fattori

List of Publications by Year
in descending order

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65 papers	1,109 citations	471477 17 h-index	434170 31 g-index
66 all docs	66 docs citations	66 times ranked	2169 citing authors

#	Article	IF	Citations
1	POPDC1S201F causes muscular dystrophy and arrhythmia by affecting protein trafficking. Journal of Clinical Investigation, 2015, 126, 239-253.	8.2	85
2	Clinical, histological and genetic characterisation of patients with tubular aggregate myopathy caused by mutations in STIM1. Journal of Medical Genetics, 2014, 51, 824-833.	3.2	72
3	Pontocerebellar hypoplasia type 6 caused by mutations in RARS2: definition of the clinical spectrum and molecular findings in five patients. Journal of Inherited Metabolic Disease, 2013, 36, 43-53.	3.6	70
4	Supratentorial and pontine <scp>MRI</scp> abnormalities characterize recessive spastic ataxia of <scp>C</scp>harlevoixâ€<scp>S</scp>aguenay. A comprehensive study of an <scp>I</scp>talian series. European Journal of Neurology, 2013, 20, 138-146.	3.3	57
5	Childhood onset tubular aggregate myopathy associated with de novo STIM1 mutations. Journal of Neurology, 2014, 261, 870-876.	3.6	56
6	MRI in sarcoglycanopathies: a large international cohort study. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 72-77.	1.9	55
7	Centronuclear myopathy related to dynamin 2 mutations: Clinical, morphological, muscle imaging and genetic features of an Italian cohort. Neuromuscular Disorders, 2013, 23, 229-238.	0.6	53
8	Centronuclear myopathies: genotypeâ€“phenotype correlation and frequency of defined genetic forms in an Italian cohort. Journal of Neurology, 2015, 262, 1728-1740.	3.6	51
9	A novel gainâ€ofâ€function mutation in ORAI1 causes lateâ€onset tubular aggregate myopathy and congenital miosis. Clinical Genetics, 2017, 91, 780-786.	2.0	50
10	Progressive cavitating leukoencephalopathy associated with respiratory chain complex I deficiency and a novel mutation in NDUFS1. Neurogenetics, 2011, 12, 9-17.	1.4	43
11	Brownâ€“Vialettoâ€“van Laere and Fazioâ€“Londe overlap syndromes: A clinical, biochemical and genetic study. Neuromuscular Disorders, 2012, 22, 1075-1082.	0.6	36
12	A new method for analysis of mitochondrial DNA point mutations and assess levels of heteroplasmy. Biochemical and Biophysical Research Communications, 2006, 342, 387-393.	2.1	33
13	Comparative Analysis and Functional Mapping of SACS Mutations Reveal Novel Insights into Sacsin Repeated Architecture. Human Mutation, 2013, 34, 525-537.	2.5	31
14	Defective kinesin binding of TUBB2A causes progressive spastic ataxia syndrome resembling sacsinopathy. Human Molecular Genetics, 2018, 27, 1892-1904.	2.9	29
15	Insights from genotypeâ€“phenotype correlations by novel SPEG mutations causing centronuclear myopathy. Neuromuscular Disorders, 2017, 27, 836-842.	0.6	27
16	Broad phenotypic spectrum and genotype-phenotype correlations in GMPPB-related dystroglycanopathies: an Italian cross-sectional study. Orphanet Journal of Rare Diseases, 2018, 13, 170.	2.7	26
17	Novel findings associated with MTM1 suggest a higher number of female symptomatic carriers. Neuromuscular Disorders, 2016, 26, 292-299.	0.6	25
18	A new de novo missense mutation in MYH2 expands clinical and genetic findings in hereditary myosin myopathies. Neuromuscular Disorders, 2013, 23, 437-440.	0.6	17

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19	New Mutations in NEB Gene Discovered by Targeted Next-Generation Sequencing in Nemaline Myopathy Italian Patients. Journal of Molecular Neuroscience, 2016, 59, 351-359.	2.3	17
20	Start codon mutation of GYG1 causing late-onset polyglucosan body myopathy with nemaline rods. Journal of Neurology, 2016, 263, 2133-2135.	3.6	17
21	Further pitfalls in the diagnosis of mtDNA mutations: homoplasmic mt-tRNA mutations. Journal of Medical Genetics, 2007, 45, 55-61.	3.2	15
22	Somatic mosaicism in TPM2-related myopathy with nemaline rods and cap structures. Acta Neuropathologica, 2013, 125, 169-171.	7.7	15
23	Muscle magnetic resonance imaging and histopathology in ACTA1â€related congenital nemaline myopathy. Muscle and Nerve, 2014, 50, 1011-1016.	2.2	15
24	Identification of a new mtDNA mutation (14724G>A) associated with mitochondrial leukoencephalopathy. Biochemical and Biophysical Research Communications, 2007, 354, 937-941.	2.1	14
25	Molecular diagnosis of known recessive ataxias by homozygosity mapping with SNP arrays. Journal of Neurology, 2011, 258, 56-67.	3.6	14
26	A new phenotypic variant in cleidocranial dysplasia (CCD) associated with mutation c.391C>T of the RUNX2 gene. BMJ Case Reports, 2012, 2012, bcr1220115422-bcr1220115422.	0.5	14
27	Mitochondrial Neurogastrointestinal Encephalomyopathy Presenting as Anorexia Nervosa. Journal of Adolescent Health, 2016, 59, 729-731.	2.5	14
28	Clinical and imaging hallmarks of the MYH7â€related myopathy with severe axial involvement. Muscle and Nerve, 2018, 58, 224-234.	2.2	14
29	The NDUFB11 gene is not a modifier in Leber hereditary optic neuropathy. Biochemical and Biophysical Research Communications, 2007, 355, 181-187.	2.1	13
30	Muscle imaging in patients with tubular aggregate myopathy caused by mutations in STIM1. Neuromuscular Disorders, 2015, 25, 898-903.	0.6	13
31	â€˜Amish Nemaline Myopathyâ€™ in 2 Italian siblings harbouring a novel homozygous mutation in Troponin-I gene. Neuromuscular Disorders, 2019, 29, 766-770.	0.6	13
32	Core-rod myopathy due to a novel mutation in BTB/POZ domain of KBTBD13 manifesting as late onset LGMD. Acta Neuropathologica Communications, 2018, 6, 94.	5.2	12
33	Expanding the histopathological spectrum of CFL2â€related myopathies. Clinical Genetics, 2018, 93, 1234-1239.	2.0	11
34	Infantile Mitochondrial Disorders. Bioscience Reports, 2007, 27, 105-112.	2.4	10
35	A novel mutation in the SACS gene associated with a complicated form of spastic ataxia. Journal of Neurology, 2008, 255, 1429-1431.	3.6	9
36	Acute myeloid leukemia in a 3 years old child with cleidocranial dysplasia. Leukemia and Lymphoma, 2016, 57, 2189-2191.	1.3	9

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37	A new mtDNAâ€“tRNAGlu mutation (14728T>C) presenting a late-onset mitochondrial encephalomyopathy. Mitochondrion, 2007, 7, 396-398.	3.4	8
38	Copy Number Variants Account for a Tiny Fraction of Undiagnosed Myopathic Patients. Genes, 2018, 9, 524.	2.4	7
39	Novel Dominant Mutation in BIN1 Gene Causing Mild Centronuclear Myopathy Revealed by Myalgias and CK Elevation. Journal of Neuromuscular Diseases, 2016, 3, 111-114.	2.6	6
40	Mitochondrial myopathy in a child with a muscle-restricted mutation in the mitochondrial transfer RNAAsn gene. Biochemical and Biophysical Research Communications, 2011, 412, 518-521.	2.1	5
41	Diagnostic interest of whole-body MRI in early- and late-onset LAMA2 muscular dystrophies: a large international cohort. Journal of Neurology, 2022, 269, 2414-2429.	3.6	5
42	Blood malignancies presenting with mutations at equivalent residues in RUNX1â€“2 suggest a common leukemogenic pathway. Leukemia and Lymphoma, 2017, 58, 2002-2004.	1.3	4
43	Somatic mosaicism represents an underestimated event underlying collagen 6-related disorders. European Journal of Paediatric Neurology, 2017, 21, 873-883.	1.6	4
44	Novel ACTA1 mutation causes late-presenting nemaline myopathy with unusual dark cores. Neuromuscular Disorders, 2021, 31, 139-148.	0.6	4
45	Neuromyopathy with congenital cataracts and glaucoma: a distinct syndrome caused by POLG variants. European Journal of Human Genetics, 2018, 26, 367-373.	2.8	3
46	Expanding the clinical-pathological and genetic spectrum of RYR1-related congenital myopathies with cores and minicores: an Italian population study. Acta Neuropathologica Communications, 2022, 10, 54.	5.2	3
47	Clinical and audiological follow up of a family with the 8363G>A mutation in the mitochondrial DNA. Neuromuscular Disorders, 2009, 19, 291-296.	0.6	1
48	P3.1 Brownâ€“Vialettoâ€“Van Laere and Fazio Londe overlap sindromes: A clinical, biochemical and genetic study in 6 patients. Neuromuscular Disorders, 2011, 21, 682.	0.6	1
49	Tubular aggregate myopathy with miosis caused by a novel mutation in ORAI1. Neuromuscular Disorders, 2016, 26, S193.	0.6	1
50	EP.126Congenital fiber type disproportion related to novel autosomal dominant mutation in TNNT1. Neuromuscular Disorders, 2019, 29, S204.	0.6	1
51	M.P.1.04 A novel mtDNA mutation in COIII impairs assembly of cytochrome c oxidase in a MELAS patient. Neuromuscular Disorders, 2007, 17, 768-769.	0.6	0
52	P2.26 Clinical, histological and molecular genetic features of a congenital severe infantile rimmed vacuolar myopathy. Neuromuscular Disorders, 2010, 20, 626.	0.6	0
53	P.9.15 Centronuclear myopathies: The experience of Italian Network for congenital myopathies. Neuromuscular Disorders, 2013, 23, 788.	0.6	0
54	P.9.9 A novel de novo mutation in ACTA1 causes a congenital myopathy with misleading type 1 fiber predominance and a peculiar MRI. Neuromuscular Disorders, 2013, 23, 786.	0.6	0

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55	Late-onset congenital myopathies: Clinical and molecular features. Neuromuscular Disorders, 2015, 25, S277-S278.	0.6	0
56	Muscle imaging in STIM1-mutated tubular aggregate myopathy patients. Neuromuscular Disorders, 2015, 25, S289-S290.	0.6	0
57	X-linked myotubular myopathy in females. Neuromuscular Disorders, 2015, 25, S274.	0.6	0
58	Dominant BIN1-related centronuclear myopathy (CNM) revealed by lower limb myalgia and moderate CK elevation. Neuromuscular Disorders, 2015, 25, S275.	0.6	0
59	Clinical, pathology and imaging heterogeneity in autosomal recessive RYR1-related myopathy. Neuromuscular Disorders, 2015, 25, S258.	0.6	0
60	Neuromyopathy with cataracts and glaucoma: A novel syndrome caused by recessive mutations in POLG1. Neuromuscular Disorders, 2016, 26, S174.	0.6	0
61	Novel GYG1 mutation causing late-onset polyglucosan body myopathy with nemaline rods. Neuromuscular Disorders, 2016, 26, S199-S200.	0.6	0
62	CONGENITAL MYOPATHIES: NEMALINE AND TITINOPATHIES. Neuromuscular Disorders, 2018, 28, S101.	0.6	0
63	CONGENITAL MYOPATHIES: NEMALINE AND TITINOPATHIES. Neuromuscular Disorders, 2018, 28, S102.	0.6	0
64	EP.131Novel ACTA1 mutation causes late-onset nemaline myopathy with fuzzy-dark cores. Neuromuscular Disorders, 2019, 29, S205-S206.	0.6	0
65	EP.111Identification of novel biallelic mutations in SPTBN4 in a child with NEDHND featuring a spinal muscular atrophy phenotype. Neuromuscular Disorders, 2019, 29, S200.	0.6	0