Pascal Laforet

List of Publications by Year
in descending order

81 papers	3,712 citations	172386 29 h-index	138417 58 g-index
83 all docs	83 docs citations	83 times ranked	3533 citing authors

#	Article	IF	Citations
1	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.	1.8	5
2	Motor and respiratory decline in patients with late onset Pompe disease after cessation of enzyme replacement therapy during COVIDâ€19 pandemic. European Journal of Neurology, 2022, 29, 1181-1186.	1.7	7
3	No effect of resveratrol on fatty acid oxidation or exercise capacity in patients with fatty acid oxidation disorders: A randomized clinical crossâ€over trial. Journal of Inherited Metabolic Disease, 2022, 45, 517-528.	1.7	7
4	Nusinersen treatment in adults with severe spinal muscular atrophy: A real-life retrospective observational cohort study. Revue Neurologique, 2022, 178, 234-240.	0.6	6
5	FDX2 and ISCU Gene Variations Lead to Rhabdomyolysis With Distinct Severity and Iron Regulation. Neurology: Genetics, 2022, 8, e648.	0.9	4
6	No effect of triheptanoin in patients with phosphofructokinase deficiency. Neuromuscular Disorders, 2022, , .	0.3	1
7	Macroglossia: A potentially severe complication of lateâ€onset Pompe disease. European Journal of Neurology, 2022, 29, 2121-2128.	1.7	4
8	Phenotypical variability and atypical presentations in a French cohort of Andersenâ€“Tawil syndrome. European Journal of Neurology, 2022, 29, 2398-2411.	1.7	1
9	Unravelling the impact of frontal lobe impairment for social dysfunction in myotonic dystrophy type 1. Brain Communications, 2022, 4, .	1.5	5
10	The Glycogen Storage Diseases and Related Disorders. , 2022, , 179-200.		2
11	Clinical correlations and longâ€term followâ€up in 100 patients with sarcoglycanopathies. European Journal of Neurology, 2021, 28, 660-669.	1.7	11
12	251st ENMC international workshop: Polyglucosan storage myopathies 13â€“15 December 2019, Hoofddorp, the Netherlands. Neuromuscular Disorders, 2021, 31, 466-477.	0.3	4
13	Deep phenotyping of an international series of patients with lateâ€onset dysferlinopathy. European Journal of Neurology, 2021, 28, 2092-2102.	1.7	9
14	Narrative review of glycogen storage disorder type <scp>III</scp> with a focus on neuromuscular, cardiac and therapeutic aspects. Journal of Inherited Metabolic Disease, 2021, 44, 521-533.	1.7	9
15	A multicenter cross-sectional French study of the impact of COVID-19 on neuromuscular diseases. Orphanet Journal of Rare Diseases, 2021, 16, 450.	1.2	9
16	Safety and efficacy of cipaglucosidase alfa plus miglustat versus alglucosidase alfa plus placebo in late-onset Pompe disease (PROPEL): an international, randomised, double-blind, parallel-group, phase 3 trial. Lancet Neurology, The, 2021, 20, 1027-1037.	4.9	42
17	European muscle MRI study in limb girdle muscular dystrophy type R1/2A (LGMDR1/LGMD2A). Journal of Neurology, 2020, 267, 45-56.	1.8	43
18	Incidence and predictors of total mortality in 267 adults presenting with mitochondrial diseases. Journal of Inherited Metabolic Disease, 2020, 43, 459-466.	1.7	11

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19	A high prevalence of arterial hypertension in patients with mitochondrial diseases. Journal of Inherited Metabolic Disease, 2020, 43, 478-485.	1.7	5
20	Biallelic mutations in Tenascin-X cause classical-like Ehlers-Danlos syndrome with slowly progressive muscular weakness. Neuromuscular Disorders, 2020, 30, 833-838.	0.3	2
21	Creation and implementation of a European registry for patients with McArdle disease and other muscle glycogenoses (EUROMAC registry). Orphanet Journal of Rare Diseases, 2020, 15, 187.	1.2	3
22	Phenotypic Spectrum of Myopathies with Recessive Anoctamin-5 Mutations. Journal of Neuromuscular Diseases, 2020, 7, 443-451.	1.1	9
23	A Proteomics-Based Analysis Reveals Predictive Biological Patterns in Fabry Disease. Journal of Clinical Medicine, 2020, 9, 1325.	1.0	18
24	Urine glucose tetrasaccharide: A good biomarker for glycogenoses type II and III? A study of the French cohort. Molecular Genetics and Metabolism Reports, 2020, 23, 100583.	0.4	17
25	Glycogenin-1 deficiency mimicking limb-girdle muscular dystrophy. Molecular Genetics and Metabolism Reports, 2020, 24, 100597.	0.4	2
26	Ganglionopathies Associated with MERRF Syndrome: An Original Report. Journal of Neuromuscular Diseases, 2020, 7, 419-423.	1.1	4
27	Longâ€term benefit of enzyme replacement therapy with alglucosidase alfa in adults with Pompe disease: Prospective analysis from the French Pompe Registry. Journal of Inherited Metabolic Disease, 2020, 43, 1219-1231.	1.7	21
28	Sensory neuronopathy as a major clinical feature of mitochondrial trifunctional protein deficiency in adults. Revue Neurologique, 2020, 176, 380-386.	0.6	9
29	Guidance for the care of neuromuscular patients during the COVID-19 pandemic outbreak from the French Rare Health Care for Neuromuscular Diseases Network. Revue Neurologique, 2020, 176, 507-515.	0.6	71
30	Late Onset Multiple Acyl-CoA Dehydrogenase Deficiency (MADD) Myopathy Misdiagnosed as Polymyositis. Journal of Clinical Rheumatology, 2020, 26, e125-e127.	0.5	3
31	Deep morphological analysis of muscle biopsies from type III glycogenesis (GSDIII), debranching enzyme deficiency, revealed stereotyped vacuolar myopathy and autophagy impairment. Acta Neuropathologica Communications, 2019, 7, 167.	2.4	17
32	Scapular dyskinesis in myotonic dystrophy type 1: clinical characteristics and genetic investigations. Journal of Neurology, 2019, 266, 2987-2996.	1.8	1
33	X-linked Emeryâ€“Dreifuss muscular dystrophy manifesting with adult onset axial weakness, camptocormia, and minimal joint contractures. Neuromuscular Disorders, 2019, 29, 678-683.	0.3	6
34	Life-threatening lactic acidosis occurring in adults with mitochondrial disorders. Revue Neurologique, 2019, 175, 564-567.	0.6	0
35	Congenital myopathies are mainly associated with a mild cardiac phenotype. Journal of Neurology, 2019, 266, 1367-1375.	1.8	10
36	Large variation in effects during 10 years of enzyme therapy in adults with Pompe disease. Neurology, 2019, 93, e1756-e1767.	1.5	70

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37	Assessment of diaphragm motion using ultrasonography in a patient with facio-scapulo-humeral dystrophy. Medicine (United States), 2019, 98, e13887.	0.4	3
38	Anti-HMGCR myopathy may resemble limb-girdle muscular dystrophy. Neurology: Neuroimmunology and NeuroInflammation, 2019, 6, e523.	3.1	66
39	The spinal and cerebral profile of adult spinal-muscular atrophy: A multimodal imaging study. NeuroImage: Clinical, 2019, 21, 101618.	1.4	54
40	Progress and challenges of gene therapy for Pompe disease. Annals of Translational Medicine, 2019, 7, 287-287.	0.7	35
41	Development and Validation of a New Scoring System to Predict Survival in Patients With Myotonic Dystrophy Type 1. JAMA Neurology, 2018, 75, 573.	4.5	32
42	Rescue of GSDIII Phenotype with Gene Transfer Requires Liver- and Muscle-Targeted GDE Expression. Molecular Therapy, 2018, 26, 890-901.	3.7	24
43	Diaphragm: Pathophysiology and Ultrasound Imaging in Neuromuscular Disorders. Journal of Neuromuscular Diseases, 2018, 5, 1-10.	1.1	57
44	Two new cases of mitochondrial myopathy with exercise intolerance, hyperlactatemia and cardiomyopathy, caused by recessive SLC25A4 mutations. Mitochondrion, 2018, 39, 26-29.	1.6	12
45	The motor unit number index (MUNIX) profile of patients with adult spinal muscular atrophy. Clinical Neurophysiology, 2018, 129, 2333-2340.	0.7	33
46	Lateâ€onset Pompe disease in France: molecular features and epidemiology from a nationwide study. Journal of Inherited Metabolic Disease, 2018, 41, 937-946.	1.7	27
47	Encephalopathy associated with a reversible splenial lesion in riboflavin-responsive multiple acyl-CoA dehydrogenase deficiency. Revue Neurologique, 2018, 174, 747-750.	0.6	1
48	Renal artery fibromuscular dysplasia in Pompe disease: A case report. Molecular Genetics and Metabolism Reports, 2018, 16, 64-65.	0.4	7
49	Efficacy of Rituximab in Refractory Generalized anti-AChR Myasthenia Gravis. Journal of Neuromuscular Diseases, 2018, 5, 241-249.	1.1	31
50	Incidence and predictors of sudden death, major conduction defects and sustained ventricular tachyarrhythmias in 1388 patients with myotonic dystrophy type 1. European Heart Journal, 2017, 38, ehw569.	1.0	59
51	The diagnostic value of hyperammonaemia induced by the non-ischaemic forearm exercise test. Journal of Clinical Pathology, 2017, 70, 896-898.	1.0	4
52	European consensus for starting and stopping enzyme replacement therapy in adult patients with Pompe disease: a 10â€year experience. European Journal of Neurology, 2017, 24, 768.	1.7	118
53	Effect of enzyme replacement therapy with alglucosidase alfa (MyozymeÂ®) in 12 patients with advanced late-onset Pompe disease. Molecular Genetics and Metabolism, 2017, 122, 80-85.	0.5	21
54	Update on new muscle glycogenosis. Current Opinion in Neurology, 2017, 30, 449-456.	1.8	23

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55	Risk for Complications after Pacemaker or Cardioverter Defibrillator Implantations in Patients with Myotonic Dystrophy Type 1. Journal of Neuromuscular Diseases, 2017, 4, 175-181.	1.1	5
56	Rescue of Pompe disease in mice by AAV-mediated liver delivery of secretable acid Î±-glucosidase. Science Translational Medicine, 2017, 9, .	5.8	103
57	Muscle glycogen synthesis and breakdown are both impaired in glycogenin-1 deficiency. Neurology, 2017, 89, 2491-2494.	1.5	13
58	Prediction of longâ€term prognosis by heteroplasmy levels of the m.3243A>G mutation in patients with the <scp>mitochondrial encephalomyopathy, lactic acidosis and strokeâ€like episodes</scp> syndrome. European Journal of Neurology, 2017, 24, 255-261.	1.7	41
59	Long-term exposure to Myozyme results in a decrease of anti-drug antibodies in late-onset Pompe disease patients. Scientific Reports, 2016, 6, 36182.	1.6	22
60	Atrial flutter in myotonic dystrophy type 1: Patient characteristics and clinical outcome. Neuromuscular Disorders, 2016, 26, 227-233.	0.3	19
61	Gender as a Modifying Factor Influencing Myotonic Dystrophy Type 1 Phenotype Severity and Mortality: A Nationwide Multiple Databases Cross-Sectional Observational Study. PLoS ONE, 2016, 11, e0148264.	1.1	113
62	SHOULD patients with asymptomatic pompe disease be treated? A nationwide study in france. Muscle and Nerve, 2015, 51, 884-889.	1.0	19
63	208th ENMC International Workshop: Formation of a European Network to develop a European data sharing model and treatment guidelines for Pompe disease Naarden, The Netherlands, 26â€“28 September 2014. Neuromuscular Disorders, 2015, 25, 674-678.	0.3	24
64	Long-term cardiac prognosis and risk stratification in 260 adults presenting with mitochondrial diseases. European Heart Journal, 2015, 36, 2886-2893.	1.0	71
65	Skeletal muscle metabolism is impaired during exercise in glycogen storage disease type III. Neurology, 2015, 84, 1767-1771.	1.5	26
66	Clinical and genetic spectrum in limb-girdle muscular dystrophy type 2E. Neurology, 2015, 84, 1772-1781.	1.5	50
67	Skeletal muscle quantitative nuclear magnetic resonance imaging followâ€up of adult Pompe patients. Journal of Inherited Metabolic Disease, 2015, 38, 565-572.	1.7	83
68	Minutes of the European POmpe Consortium (EPOC) Meeting March 27 to 28, 2015, Munich, Germany. Acta Myologica, 2015, 34, 141-3.	1.5	5
69	The French Pompe registry. Baseline characteristics of a cohort of 126 patients with adult Pompe disease. Revue Neurologique, 2013, 169, 595-602.	0.6	48
70	Open-label extension study following the Late-Onset Treatment Study (LOTS) of alglucosidase alfa. Molecular Genetics and Metabolism, 2012, 107, 456-461.	0.5	93
71	Whole-body muscle MRI in 20 patients suffering from late onset Pompe disease: Involvement patterns. Neuromuscular Disorders, 2011, 21, 791-799.	0.3	138
72	A Randomized Study of Alglucosidase Alfa in Late-Onset Pompe's Disease. New England Journal of Medicine, 2010, 362, 1396-1406.	13.9	674

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73	Disorders of muscle lipid metabolism: Diagnostic and therapeutic challenges. Neuromuscular Disorders, 2010, 20, 693-700.	0.3	86
74	Analysis of theDYSFmutational spectrum in a large cohort of patients. Human Mutation, 2009, 30, E345-E375.	1.1	97
75	Complete fatty degeneration of lumbar erector spinae muscles caused by a primary dysferlinopathy. Muscle and Nerve, 2008, 37, 410-414.	1.0	32
76	Clinical features of lateâ€onset Pompe disease: A prospective cohort study. Muscle and Nerve, 2008, 38, 1236-1245.	1.0	200
77	Dilative arteriopathy and basilar artery dolichoectasia complicating late-onset Pompe disease. Neurology, 2008, 70, 2063-2066.	1.5	89
78	Phenotypic Study in 40 Patients With Dysferlin Gene Mutations. Archives of Neurology, 2007, 64, 1176.	4.9	230
79	Respiratory insufficiency and limb muscle weakness in adults with Pompe's disease. European Respiratory Journal, 2005, 26, 1024-1031.	3.1	101
80	Mitochondrial DNA transfer RNA gene sequence variations in patients with mitochondrial disorders. Brain, 2001, 124, 984-994.	3.7	78
81	Juvenile and adult-onset acid maltase deficiency in France. Neurology, 2000, 55, 1122-1128.	1.5	197