Benoit Funalot

List of Publications by Year
in descending order

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53	2,529	257450	206112
papers	citations	24 h-index	48 g-index

57	57	57	5315
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	SCN10A variants associated with congenital Harlequin Syndrome. British Journal of Dermatology, 2022, , .	1.5	1
2	Prevalence and determinants of iron deficiency in cardiac amyloidosis. ESC Heart Failure, 2022, 9, 1314-1327.	3.1	4
3	Natural history and impact of treatment with tafamidis on major cardiovascular outcomeâ€free survival time in a cohort of patients with transthyretin amyloidosis. European Journal of Heart Failure, 2021, 23, 264-274.	7.1	30
4	NKX2.1 (TTF1) germline mutation associated with pulmonary fibrosis and lung cancer. ERJ Open Research, 2021, 7, 00356-2021.	2.6	8
5	History of extracardiac/cardiac events in cardiac amyloidosis: prevalence and time from initial onset to diagnosis. ESC Heart Failure, 2021, 8, 5501-5512.	3.1	11
6	Central nervous system complications in adult cystinosis patients. Journal of Inherited Metabolic Disease, 2020, 43, 348-356.	3.6	14
7	Optimized Protocol to Generate Spinal Motor Neuron Cells from Induced Pluripotent Stem Cells from Charcot Marie Tooth Patients. Brain Sciences, 2020, 10, 407.	2.3	7
8	Hereditary sensory autonomic neuropathy type II: Report of two novel mutations in the FAM134B gene. Journal of the Peripheral Nervous System, 2019, 24, 354-358.	3.1	9
9	High predictive value of brain MRI imaging in primary mitochondrial respiratory chain deficiency. Journal of Medical Genetics, 2018, 55, 378-383.	3.2	21
10	Pitfalls in molecular diagnosis of Friedreich ataxia. European Journal of Medical Genetics, 2018, 61, 455-458.	1.3	5
11	Reply: The expanding neurological phenotype of DNM1L-related disorders. Brain, 2018, 141, e29-e29.	7.6	5
12	Refine penetrance estimates in the main pathogenic variants of transthyretin hereditary (familial) amyloid polyneuropathy (TTR-FAP) using a new non-parametric approach (NPSE). Amyloid: the International Journal of Experimental and Clinical Investigation: the Official Journal of the International Society of Amyloidosis, 2017, 24, 115-116.	3.0	5
13	Heterogeneity of lung disease associated with NK2 homeobox 1 mutations. Respiratory Medicine, 2017, 129, 16-23.	2.9	54
14	Genetic diagnosis of Mendelian disorders via RNA sequencing. Nature Communications, 2017, 8, 15824.	12.8	432
15	Mutations in DNM1L, as in OPA1, result in dominant optic atrophy despite opposite effects on mitochondrial fusion and fission. Brain, 2017, 140, 2586-2596.	7.6	100
16	Mutations in Complex I Assembly Factor TMEM126B Result in Muscle Weakness and Isolated Complex I Deficiency. American Journal of Human Genetics, 2016, 99, 208-216.	6.2	51
17	Low Serum Insulin-Like Growth Factor-I Predicts Cognitive Decline in Alzheimerâ€™s Disease. Journal of Alzheimer's Disease, 2016, 52, 641-649.	2.6	28
18	Recessive and Dominant De Novo ITPR1 Mutations Cause Gillespie Syndrome. American Journal of Human Genetics, 2016, 98, 971-980.	6.2	113

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19	New Method for Sorting Endothelial and Neural Progenitors from Human Induced Pluripotent Stem Cells by Sedimentation Field Flow Fractionation. Analytical Chemistry, 2016, 88, 6696-6702.	6.5	15
20	Insulin-Like Growth Factor-I, Insulin-Like Growth factor Binding Protein-3 and Blood Hemoglobin Concentration in an Elderly Population. Journals of Gerontology - Series A Biological Sciences and Medical Sciences, 2015, 70, 854-859.	3.6	4
21	Ehlersâ€“Danlos syndrome in rheumatology: Diagnostic and therapeutic challenges. Joint Bone Spine, 2015, 82, 305-307.	1.6	4
22	Neuroprotective Effect of Erythropoietin against Pressure Ulcer in a Mouse Model of Small Fiber Neuropathy. PLoS ONE, 2014, 9, e113454.	2.5	4
23	Characterization of Endoneurial Fibroblast-like Cells from Human and Rat Peripheral Nerves. Journal of Histochemistry and Cytochemistry, 2014, 62, 424-435.	2.5	49
24	In vitro 3D angiogenesis assay in egg white matrix: comparison to Matrigel, compatibility to various species, and suitability for drug testing. Laboratory Investigation, 2014, 94, 340-349.	3.7	23
25	Loss-of-Function Mutations in WDR73 Are Responsible for Microcephaly and Steroid-Resistant Nephrotic Syndrome: Galloway-Mowat Syndrome. American Journal of Human Genetics, 2014, 95, 637-648.	6.2	108
26	Neuropathologic Characterization ofINF2-Related Charcot-Marie-Tooth Disease: Evidence for a Schwann Cell Actinopathy. Journal of Neuropathology and Experimental Neurology, 2014, 73, 223-233.	1.7	25
27	An exploratory randomised double-blind and placebo-controlled phase 2 study of a combination of baclofen, naltrexone and sorbitol (PXT3003) in patients with Charcot-Marie-Tooth disease type 1A. Orphanet Journal of Rare Diseases, 2014, 9, 199.	2.7	94
28	Insulin-Like Growth Factor I, Insulin-like Growth factor Binding Protein 3, and Atrial Fibrillation in the Elderly. Journals of Gerontology - Series A Biological Sciences and Medical Sciences, 2014, 69, 1025-1032.	3.6	27
29	A reversible functional sensory neuropathy model. Neuroscience Letters, 2014, 571, 39-44.	2.1	11
30	Peripheral Myelin Protein 22 gene duplication with atypical presentations: A new example of the wide spectrum of Charcot-Marie-Tooth 1A disease. Neuromuscular Disorders, 2014, 24, 524-528.	0.6	18
31	<scp>CMT4D</scp> (<scp>NDRG1</scp> mutation): genotypeâ€“phenotype correlations. Journal of the Peripheral Nervous System, 2013, 18, 261-265.	3.1	12
32	Two novel mutations of the calcium-sensing receptor gene affecting the same amino acid position lead to opposite phenotypes and reveal the importance of p.N802 on receptor activity. European Journal of Endocrinology, 2013, 168, K27-K34.	3.7	15
33	The various Charcotâ€“Marieâ€“Tooth diseases. Current Opinion in Neurology, 2013, 26, 473-480.	3.6	48
34	Massive and exclusive pontocerebellar damage in mitochondrial disease and NUBPL mutations. Neurology, 2012, 79, 391-391.	1.1	27
35	Early onset Parkinsonism associated with an intronic SOD1 mutation. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2012, 13, 315-317.	2.1	3
36	Endoneurial Fibroblast-Like Cells. Journal of Neuropathology and Experimental Neurology, 2012, 71, 938-947.	1.7	48

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37	Insulin-Like Growth Factor-I and Insulin-Like Growth Factor Binding Protein-3 in Alzheimer's Disease. Journal of Clinical Endocrinology and Metabolism, 2012, 97, 4673-4681.	3.6	63
38	Fingolimod inhibits PDGF-B-induced migration of vascular smooth muscle cell by down-regulating the S1PR1/S1PR3 pathway. Biochimie, 2012, 94, 2523-2531.	2.6	14
39	Homozygous deletion of an EGR2 enhancer in congenital amyelinating neuropathy. Annals of Neurology, 2012, 71, 719-723.	5.3	17
40	Fingolimod potentiates the effects of sunitinib malate in a rat breast cancer model. Breast Cancer Research and Treatment, 2012, 134, 31-40.	2.5	24
41	INF2Mutations in Charcotâ€“Marieâ€“Tooth Disease with Glomerulopathy. New England Journal of Medicine, 2011, 365, 2377-2388.	27.0	235
42	Novel mutations in the PRX and the MTMR2 genes are responsible for unusual Charcot-Marie-Tooth disease phenotypes. Neuromuscular Disorders, 2011, 21, 543-550.	0.6	30
43	Nerve biopsy: requirements for diagnosis and clinical value. Acta Neuropathologica, 2011, 121, 313-326.	7.7	31
44	Segregation of mtDNA throughout human embryofetal development: m.3243A>G as a model system. Human Mutation, 2011, 32, 116-125.	2.5	103
45	A leaky splicing mutation affecting SMN1 exon 7 inclusion explains an unexpected mild case of spinal muscular atrophy. Human Mutation, 2011, 32, 989-994.	2.5	18
46	How can grafted breast cancer models be optimized?. Cancer Biology and Therapy, 2011, 12, 855-864.	3.4	32
47	CNS/PNS Boundary Transgression by Central Glia in the Absence of Schwann Cells or Krox20/Egr2 Function. Journal of Neuroscience, 2010, 30, 5958-5967.	3.6	54
48	High metabolic level in patients with familial amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2009, 10, 113-117.	2.1	135
49	Genotype-Phenotype Correlations in Charcot-Marie-Tooth Disease Type 2 Caused by Mitofusin 2 Mutations. Archives of Neurology, 2009, 66, 1511-6.	4.5	102
50	A Mutation that Creates a Pseudoexon in SOD1 Causes Familial ALS. Annals of Human Genetics, 2009, 73, 652-657.	0.8	32
51	Histopathological Findings in Hereditary Motor and Sensory Neuropathy of Axonal Type With Onset in Early Childhood Associated WithMitofusin 2Mutations. Journal of Neuropathology and Experimental Neurology, 2008, 67, 1097-1102.	1.7	81
52	Genes encoding endothelin-converting enzyme-1 and endothelin-1 interact to influence blood pressure in women. Journal of Hypertension, 2004, 22, 739-743.	0.5	37
53	Leigh-like encephalopathy complicating Leber's hereditary optic neuropathy. Annals of Neurology, 2002, 52, 374-377.	5.3	64