

Benoit Funalot

List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/647172/publications.pdf>

Version: 2024-02-01

53
papers

2,529
citations

257101

24
h-index

205818

48
g-index

57
all docs

57
docs citations

57
times ranked

5315
citing authors

#	ARTICLE	IF	CITATIONS
1	Genetic diagnosis of Mendelian disorders via RNA sequencing. <i>Nature Communications</i> , 2017, 8, 15824.	5.8	432
2	<i>INF2</i> Mutations in Charcot-Marie-Tooth Disease with Glomerulopathy. <i>New England Journal of Medicine</i> , 2011, 365, 2377-2388.	13.9	235
3	High metabolic level in patients with familial amyotrophic lateral sclerosis. <i>Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders</i> , 2009, 10, 113-117.	2.3	135
4	Recessive and Dominant De Novo <i>ITPR1</i> Mutations Cause Gillespie Syndrome. <i>American Journal of Human Genetics</i> , 2016, 98, 971-980.	2.6	113
5	Loss-of-Function Mutations in <i>WDR73</i> Are Responsible for Microcephaly and Steroid-Resistant Nephrotic Syndrome: Galloway-Mowat Syndrome. <i>American Journal of Human Genetics</i> , 2014, 95, 637-648.	2.6	108
6	Segregation of mtDNA throughout human embryofetal development: m.3243A>G as a model system. <i>Human Mutation</i> , 2011, 32, 116-125.	1.1	103
7	Genotype-Phenotype Correlations in Charcot-Marie-Tooth Disease Type 2 Caused by <i>Mitofusin 2</i> Mutations. <i>Archives of Neurology</i> , 2009, 66, 1511-6.	4.9	102
8	Mutations in <i>DNM1L</i> , as in <i>OPA1</i> , result in dominant optic atrophy despite opposite effects on mitochondrial fusion and fission. <i>Brain</i> , 2017, 140, 2586-2596.	3.7	100
9	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. <i>Orphanet Journal of Rare Diseases</i> , 2014, 9, 199.	1.2	94
10	Histopathological Findings in Hereditary Motor and Sensory Neuropathy of Axonal Type With Onset in Early Childhood Associated With <i>Mitofusin 2</i> Mutations. <i>Journal of Neuropathology and Experimental Neurology</i> , 2008, 67, 1097-1102.	0.9	81
11	Leigh-like encephalopathy complicating Leber's hereditary optic neuropathy. <i>Annals of Neurology</i> , 2002, 52, 374-377.	2.8	64
12	Insulin-Like Growth Factor-I and Insulin-Like Growth Factor Binding Protein-3 in Alzheimer's Disease. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2012, 97, 4673-4681.	1.8	63
13	CNS/PNS Boundary Transgression by Central Glia in the Absence of Schwann Cells or <i>Krox20/Egr2</i> Function. <i>Journal of Neuroscience</i> , 2010, 30, 5958-5967.	1.7	54
14	Heterogeneity of lung disease associated with <i>NK2 homeobox 1</i> mutations. <i>Respiratory Medicine</i> , 2017, 129, 16-23.	1.3	54
15	Mutations in Complex I Assembly Factor <i>TMEM126B</i> Result in Muscle Weakness and Isolated Complex I Deficiency. <i>American Journal of Human Genetics</i> , 2016, 99, 208-216.	2.6	51
16	Characterization of Endoneurial Fibroblast-like Cells from Human and Rat Peripheral Nerves. <i>Journal of Histochemistry and Cytochemistry</i> , 2014, 62, 424-435.	1.3	49
17	Endoneurial Fibroblast-Like Cells. <i>Journal of Neuropathology and Experimental Neurology</i> , 2012, 71, 938-947.	0.9	48
18	The various Charcot-Marie-Tooth diseases. <i>Current Opinion in Neurology</i> , 2013, 26, 473-480.	1.8	48

#	ARTICLE	IF	CITATIONS
19	Genes encoding endothelin-converting enzyme-1 and endothelin-1 interact to influence blood pressure in women. <i>Journal of Hypertension</i> , 2004, 22, 739-743.	0.3	37
20	A Mutation that Creates a Pseudoexon in <i>SOD1</i> Causes Familial ALS. <i>Annals of Human Genetics</i> , 2009, 73, 652-657.	0.3	32
21	How can grafted breast cancer models be optimized?. <i>Cancer Biology and Therapy</i> , 2011, 12, 855-864.	1.5	32
22	Nerve biopsy: requirements for diagnosis and clinical value. <i>Acta Neuropathologica</i> , 2011, 121, 313-326.	3.9	31
23	Novel mutations in the PRX and the MTMR2 genes are responsible for unusual Charcot-Marie-Tooth disease phenotypes. <i>Neuromuscular Disorders</i> , 2011, 21, 543-550.	0.3	30
24	Natural history and impact of treatment with tafamidis on major cardiovascular outcome—free survival time in a cohort of patients with transthyretin amyloidosis. <i>European Journal of Heart Failure</i> , 2021, 23, 264-274.	2.9	30
25	Low Serum Insulin-Like Growth Factor-I Predicts Cognitive Decline in Alzheimer's Disease. <i>Journal of Alzheimer's Disease</i> , 2016, 52, 641-649.	1.2	28
26	Massive and exclusive pontocerebellar damage in mitochondrial disease and <i>NUBPL</i> mutations. <i>Neurology</i> , 2012, 79, 391-391.	1.5	27
27	Insulin-Like Growth Factor I, Insulin-like Growth factor Binding Protein 3, and Atrial Fibrillation in the Elderly. <i>Journals of Gerontology - Series A Biological Sciences and Medical Sciences</i> , 2014, 69, 1025-1032.	1.7	27
28	Neuropathologic Characterization of <i>INF2</i> -Related Charcot-Marie-Tooth Disease: Evidence for a Schwann Cell Actinopathy. <i>Journal of Neuropathology and Experimental Neurology</i> , 2014, 73, 223-233.	0.9	25
29	Fingolimod potentiates the effects of sunitinib malate in a rat breast cancer model. <i>Breast Cancer Research and Treatment</i> , 2012, 134, 31-40.	1.1	24
30	In vitro 3D angiogenesis assay in egg white matrix: comparison to Matrigel, compatibility to various species, and suitability for drug testing. <i>Laboratory Investigation</i> , 2014, 94, 340-349.	1.7	23
31	High predictive value of brain MRI imaging in primary mitochondrial respiratory chain deficiency. <i>Journal of Medical Genetics</i> , 2018, 55, 378-383.	1.5	21
32	A leaky splicing mutation affecting SMN1 exon 7 inclusion explains an unexpected mild case of spinal muscular atrophy. <i>Human Mutation</i> , 2011, 32, 989-994.	1.1	18
33	Peripheral Myelin Protein 22 gene duplication with atypical presentations: A new example of the wide spectrum of Charcot-Marie-Tooth 1A disease. <i>Neuromuscular Disorders</i> , 2014, 24, 524-528.	0.3	18
34	Homozygous deletion of an <i>EGR2</i> enhancer in congenital amyelinating neuropathy. <i>Annals of Neurology</i> , 2012, 71, 719-723.	2.8	17
35	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. <i>European Journal of Endocrinology</i> , 2013, 168, K27-K34.	1.9	15
36	New Method for Sorting Endothelial and Neural Progenitors from Human Induced Pluripotent Stem Cells by Sedimentation Field Flow Fractionation. <i>Analytical Chemistry</i> , 2016, 88, 6696-6702.	3.2	15

#	ARTICLE	IF	CITATIONS
37	Fingolimod inhibits PDGF-B-induced migration of vascular smooth muscle cell by down-regulating the S1PR1/S1PR3 pathway. <i>Biochimie</i> , 2012, 94, 2523-2531.	1.3	14
38	Central nervous system complications in adult cystinosis patients. <i>Journal of Inherited Metabolic Disease</i> , 2020, 43, 348-356.	1.7	14
39	<scp>CMT4D</scp> (<scp>NDRG1</scp> mutation): genotypeâ€™phenotype correlations. <i>Journal of the Peripheral Nervous System</i> , 2013, 18, 261-265.	1.4	12
40	A reversible functional sensory neuropathy model. <i>Neuroscience Letters</i> , 2014, 571, 39-44.	1.0	11
41	History of extracardiac/cardiac events in cardiac amyloidosis: prevalence and time from initial onset to diagnosis. <i>ESC Heart Failure</i> , 2021, 8, 5501-5512.	1.4	11
42	Hereditary sensory autonomic neuropathy type II: Report of two novel mutations in the FAM134B gene. <i>Journal of the Peripheral Nervous System</i> , 2019, 24, 354-358.	1.4	9
43	<i>NKX2.1</i> (TTF1) germline mutation associated with pulmonary fibrosis and lung cancer. <i>ERJ Open Research</i> , 2021, 7, 00356-2021.	1.1	8
44	Optimized Protocol to Generate Spinal Motor Neuron Cells from Induced Pluripotent Stem Cells from Charcot Marie Tooth Patients. <i>Brain Sciences</i> , 2020, 10, 407.	1.1	7
45	Refine penetrance estimates in the main pathogenic variants of transthyretin hereditary (familial) amyloid polyneuropathy (TTR-FAP) using a new non-parametric approach (NPSE). <i>Amyloid: the International Journal of Experimental and Clinical Investigation: the Official Journal of the International Society of Amyloidosis</i> , 2017, 24, 115-116.	1.4	5
46	Pitfalls in molecular diagnosis of Friedreich ataxia. <i>European Journal of Medical Genetics</i> , 2018, 61, 455-458.	0.7	5
47	Reply: The expanding neurological phenotype of DNM1L-related disorders. <i>Brain</i> , 2018, 141, e29-e29.	3.7	5
48	Neuroprotective Effect of Erythropoietin against Pressure Ulcer in a Mouse Model of Small Fiber Neuropathy. <i>PLoS ONE</i> , 2014, 9, e113454.	1.1	4
49	Insulin-Like Growth Factor-I, Insulin-Like Growth factor Binding Protein-3 and Blood Hemoglobin Concentration in an Elderly Population. <i>Journals of Gerontology - Series A Biological Sciences and Medical Sciences</i> , 2015, 70, 854-859.	1.7	4
50	Ehlersâ€™Danlos syndrome in rheumatology: Diagnostic and therapeutic challenges. <i>Joint Bone Spine</i> , 2015, 82, 305-307.	0.8	4
51	Prevalence and determinants of iron deficiency in cardiac amyloidosis. <i>ESC Heart Failure</i> , 2022, 9, 1314-1327.	1.4	4
52	Early onset Parkinsonism associated with an intronic SOD1 mutation. <i>Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders</i> , 2012, 13, 315-317.	2.3	3
53	SCN10A variants associated with congenital Harlequin Syndrome. <i>British Journal of Dermatology</i> , 2022, , .	1.4	1