Jonathan Baets

List of Publications by Year
in descending order

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41	1,378	361413 20	361022 35
papers	citations	h-index	g-index

41	41	41	2983
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	A Recurrent <scp>KPNA3</scp> Missense Variant Causing Infantile Pure Spastic Paraplegia. Annals of Neurology, 2022, 91, 298-299.	5.3	0
2	Clinical practice guideline for the management of paediatric Charcot-Marie-Tooth disease. Journal of Neurology, Neurosurgery and Psychiatry, 2022, 93, 530-538.	1.9	10
3	De Novo and Dominantly Inherited <scp>SPTAN1</scp> Mutations Cause Spastic Paraplegia and Cerebellar Ataxia. Movement Disorders, 2022, 37, 1175-1186.	3.9	9
4	RFC1 repeat expansions: A recurrent cause of sensory and autonomic neuropathy with cough and ataxia. European Journal of Neurology, 2022, 29, 2156-2161.	3.3	14
5	PCYT2 mutations disrupting etherlipid biosynthesis: phenotypes converging on the CDP-ethanolamine pathway. Brain, 2021, 144, e17-e17.	7.6	6
6	Biallelic variants in HPDL cause pure and complicated hereditary spastic paraplegia. Brain, 2021, 144, 1422-1434.	7.6	22
7	High prevalence of sporadic late-onset nemaline myopathy in a cohort of whole-exome sequencing negative myopathy patients. Neuromuscular Disorders, 2021, 31, 1154-1160.	0.6	4
8	Unrestrained poly-ADP-ribosylation provides insights into chromatin regulation and human disease. Molecular Cell, 2021, 81, 2640-2655.e8.	9.7	52
9	Characterization of HNRNPA1 mutations defines diversity in pathogenic mechanisms and clinical presentation. JCI Insight, 2021, 6, .	5.0	38
10	Biallelic ADPRHL2 mutations in complex neuropathy affect ADP ribosylation and DNA damage response. Life Science Alliance, 2021, 4, e202101057.	2.8	11
11	Assessment of Sacsin Turnover in Patients With ARSACS. Neurology, 2021, 97, e2315-e2327.	1.1	11
12	Multisystem proteinopathy due to a homozygous p.Arg159His VCP mutation. Neurology, 2020, 94, e785-e796.	1.1	15
13	Pathogenic Variants in the Myosin Chaperone UNC-45B Cause Progressive Myopathy with Eccentric Cores. American Journal of Human Genetics, 2020, 107, 1078-1095.	6.2	24
14	The expanding genetic landscape of hereditary motor neuropathies. Brain, 2020, 143, 3540-3563.	7.6	12
15	De Novo and Inherited Variants in GBF1 are Associated with Axonal Neuropathy Caused by Golgi Fragmentation. American Journal of Human Genetics, 2020, 107, 763-777.	6.2	14
16	Reply: A homozygous GDAP2 loss-of-function variant in a patient with adult-onset cerebellar ataxia; and Novel GDAP2 pathogenic variants cause autosomal recessive spinocerebellar ataxia-27 (SCAR27) in a Chinese family. Brain, 2020, 143, e51-e51.	7.6	1
17	Clinico-Genetic, Imaging and Molecular Delineation of COQ8A-Ataxia: A Multicenter Study of 59 Patients. , 2020, 88, 251.		1
18	Reply:De novo SPTAN1mutation in axonal sensorimotor neuropathy and developmental disorder. Brain, 2020, 143, e105-e105.	7.6	1

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19	Safety and efficacy of intravenous bimagrumab in inclusion body myositis (RESILIENT): a randomised, double-blind, placebo-controlled phase 2b trial. Lancet Neurology, The, 2019, 18, 834-844.	10.2	91
20	Nonsense mutations in alpha-II spectrin in three families with juvenile onset hereditary motor neuropathy. Brain, 2019, 142, 2605-2616.	7.6	29
21	Functional characterization of GYG1 variants in two patients with myopathy and glycogenin-1 deficiency. Neuromuscular Disorders, 2019, 29, 951-960.	0.6	8
22	Defects in Axonal Transport in Inherited Neuropathies. Journal of Neuromuscular Diseases, 2019, 6, 401-419.	2.6	23
23	Muscular dystrophy with arrhythmia caused by loss-of-function mutations in BVES. Neurology: Genetics, 2019, 5, e321.	1.9	26
24	Truncating SLC5A7 mutations underlie a spectrum of dominant hereditary motor neuropathies. Neurology: Genetics, 2018, 4, e222.	1.9	19
25	Beyond ALS and FTD: the phenotypic spectrum of TBK1 mutations includes PSP-like and cerebellar phenotypes. Neurobiology of Aging, 2018, 62, 244.e9-244.e13.	3.1	30
26	Detection of variants in dystroglycanopathy-associated genes through the application of targeted whole-exome sequencing analysis to a large cohort of patients with unexplained limb-girdle muscle weakness. Skeletal Muscle, 2018, 8, 23.	4.2	40
27	No supportive evidence for TIA1 gene mutations in a European cohort of ALS-FTD spectrum patients. Neurobiology of Aging, 2018, 69, 293.e9-293.e11.	3.1	15
28	Investigating the role of ALS genes CHCHD10 and TUBA4A in Belgian FTD-ALS spectrum patients. Neurobiology of Aging, 2017, 51, 177.e9-177.e16.	3.1	60
29	Clinical Evidence of Disease Anticipation in Families Segregating a C9orf72 Repeat Expansion. JAMA Neurology, 2017, 74, 445.	9.0	56
30	MultisystemicSYNE1ataxia: confirming the high frequency and extending the mutational and phenotypic spectrum. Brain, 2016, 139, e46-e46.	7.6	40
31	SYNE1 ataxia is a common recessive ataxia with major non-cerebellar features: a large multi-centre study. Brain, 2016, 139, 1378-1393.	7.6	87
32	Clinical features ofTBK1carriers compared withC9orf72,GRNand non-mutation carriers in a Belgian cohort. Brain, 2016, 139, 452-467.	7.6	86
33	Megaconial muscular dystrophy caused by mitochondrial membrane homeostasis defect, new insights from skeletal and heart muscle analyses. Mitochondrion, 2016, 27, 32-38.	3.4	9
34	Transcriptional regulator PRDM12 is essential for human pain perception. Nature Genetics, 2015, 47, 803-808.	21.4	137
35	Loss of function mutations inHARScause a spectrum of inherited peripheral neuropathies. Brain, 2015, 138, 2161-2172.	7.6	71
36	A novel AARS mutation in a family with dominant myeloneuropathy. Neurology, 2015, 84, 2040-2047.	1.1	33

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37	Defects of mutant DNMT1 are linked to a spectrum of neurological disorders. Brain, 2015, 138, 845-861.	7.6	94
38	Truncating and Missense Mutations in IGHMBP2 Cause Charcot-Marie Tooth Disease Type 2. American Journal of Human Genetics, 2014, 95, 590-601.	6.2	75
39	Partial deletion of AFG3L2 causing spinocerebellar ataxia type 28. Neurology, 2014, 82, 2092-2100.	1.1	24
40	Recent advances in Charcotâ€“Marieâ€“Tooth disease. Current Opinion in Neurology, 2014, 27, 532-540.	3.6	60
41	Autosomal Recessive Axonal Neuropathy With Neuromyotonia: A Rare Entity. Pediatric Neurology, 2014, 50, 104-107.	2.1	20