JÃ¸rgen Erik Nielsen

List of Publications by Year
in descending order

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65	2,337	394421	223800
papers	citations	19 h-index	46 g-index

72	72	72	4415
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Mutations in the endosomal ESCRTIII-complex subunit CHMP2B in frontotemporal dementia. Nature Genetics, 2005, 37, 806-808.	21.4	752
2	Frontotemporal dementia and its subtypes: a genome-wide association study. Lancet Neurology, The, 2014, 13, 686-699.	10.2	302
3	Neurons derived from sporadic Alzheimerâ€™s disease iPSCs reveal elevated TAU hyperphosphorylation, increased amyloid levels, and GSK3B activation. Alzheimer's Research and Therapy, 2017, 9, 90.	6.2	161
4	Potential genetic modifiers of disease risk and age at onset in patients with frontotemporal lobar degeneration and GRN mutations: a genome-wide association study. Lancet Neurology, The, 2018, 17, 548-558.	10.2	97
5	Frontotemporal dementia caused by CHMP2B mutation is characterised by neuronal lysosomal storage pathology. Acta Neuropathologica, 2015, 130, 511-523.	7.7	79
6	Patient iPSC-Derived Neurons for Disease Modeling of Frontotemporal Dementia with Mutation in CHMP2B. Stem Cell Reports, 2017, 8, 648-658.	4.8	65
7	Hereditary cerebral small vessel disease and stroke. Clinical Neurology and Neurosurgery, 2017, 155, 45-57.	1.4	62
8	Reduction in mitochondrial DNA copy number in peripheral leukocytes after onset of Huntington's disease. Mitochondrion, 2014, 17, 14-21.	3.4	54
9	Selected CSF biomarkers indicate no evidence of early neuroinflammation in Huntington disease. Neurology: Neuroimmunology and NeuroInflammation, 2016, 3, e287.	6.0	53
10	Reduced ceramide synthase 2 activity causes progressive myoclonic epilepsy. Annals of Clinical and Translational Neurology, 2014, 1, 88-98.	3.7	50
11	A clinical classification acknowledging neuropsychiatric and cognitive impairment in Huntingtonâ€™s disease. Orphanet Journal of Rare Diseases, 2014, 9, 114.	2.7	50
12	Evidence of oxidative stress and mitochondrial dysfunction in spinocerebellar ataxia type 2 (SCA2) patient fibroblasts: Effect of coenzyme Q10 supplementation on these parameters. Mitochondrion, 2017, 34, 103-114.	3.4	42
13	Do I misconstrue? Sarcasm detection, emotion recognition, and theory of mind in Huntington disease.. Neuropsychology, 2016, 30, 181-189.	1.3	39
14	Glutamate-glutamine homeostasis is perturbed in neurons and astrocytes derived from patient iPSC models of frontotemporal dementia. Molecular Brain, 2020, 13, 125.	2.6	36
15	Defining active progressive multiple sclerosis. Multiple Sclerosis Journal, 2017, 23, 1727-1735.	3.0	34
16	Induced pluripotent stem cell - derived neurons for the study of spinocerebellar ataxia type 3. Stem Cell Research, 2016, 17, 306-317.	0.7	27
17	Genotypeâ€phenotype correlations, dystonia and disease progression in spinocerebellar ataxia type 14. Movement Disorders, 2018, 33, 1119-1129.	3.9	26
18	Assessing Impairment of Executive Function and Psychomotor Speed in Premanifest and Manifest Huntingtonâ€™s Disease Gene-expansion Carriers. Journal of the International Neuropsychological Society, 2015, 21, 193-202.	1.8	25

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19	Early Intrathecal T Helper 17.1 Cell Activity in Huntington Disease. Annals of Neurology, 2020, 87, 246-255.	5.3	24
20	Astrocytic reactivity triggered by defective autophagy and metabolic failure causes neurotoxicity in frontotemporal dementia type 3. Stem Cell Reports, 2021, 16, 2736-2751.	4.8	23
21	YKL-40 in cerebrospinal fluid in Huntington's disease â€“ A role in pathology or a nonspecific response to inflammation?. Parkinsonism and Related Disorders, 2014, 20, 1301-1303.	2.2	20
22	Epigenetic remodelling and dysregulation of DLGAP4 is linked with early-onset cerebellar ataxia. Human Molecular Genetics, 2014, 23, 6163-6176.	2.9	19
23	Generation of an isogenic, gene-corrected control cell line of the spinocerebellar ataxia type 2 patient-derived iPSC line H266. Stem Cell Research, 2016, 16, 202-205.	0.7	17
24	SCA28: Novel Mutation in the AFG3L2 Proteolytic Domain Causes a Mild Cerebellar Syndrome with Selective Type-1 Muscle Fiber Atrophy. Cerebellum, 2017, 16, 62-67.	2.5	16
25	Generation of an isogenic, gene-corrected control cell line of the spinocerebellar ataxia type 2 patient-derived iPSC line H271. Stem Cell Research, 2016, 16, 180-183.	0.7	15
26	A Novel TTBK2 De Novo Mutation in a Danish Family with Early-Onset Spinocerebellar Ataxia. Cerebellum, 2017, 16, 268-271.	2.5	15
27	Antisense Gene Silencing: Therapy for Neurodegenerative Disorders?. Genes, 2013, 4, 457-484.	2.4	14
28	Characterization of energy and neurotransmitter metabolism in cortical glutamatergic neurons derived from human induced pluripotent stem cells: A novel approach to study metabolism in human neurons. Neurochemistry International, 2017, 106, 48-61.	3.8	14
29	Liver function in Huntington's disease assessed by blood biochemical analyses in a clinical setting. Journal of the Neurological Sciences, 2016, 362, 326-332.	0.6	12
30	CCGâ€¢CGG interruptions in highâ€penetrance SCA8 families increase RAN translation and protein toxicity. EMBO Molecular Medicine, 2021, 13, e14095.	6.9	12
31	CSF neurofilament light concentration is increased in presymptomatic CHMP2B mutation carriers. Neurology, 2018, 90, e157-e163.	1.1	11
32	Peripheral neuropathy in hereditary spastic paraplegia caused by REEP1 variants. Journal of Neurology, 2019, 266, 735-744.	3.6	11
33	Enhancement of Autophagy and Solubilization of Ataxin-2 Alleviate Apoptosis in Spinocerebellar Ataxia Type 2 Patient Cells. Cerebellum, 2020, 19, 165-181.	2.5	11
34	Exploring Genetic Factors Involved in Huntington Disease Age of Onset: E2F2 as a New Potential Modifier Gene. PLoS ONE, 2015, 10, e0131573.	2.5	11
35	Increased Intrathecal Activity of Follicular Helper T Cells in Patients With Relapsing-Remitting Multiple Sclerosis. Neurology: Neuroimmunology and NeuroInflammation, 2022, 9, .	6.0	11
36	Generation of an isogenic, gene-corrected control cell line of the spinocerebellar ataxia type 2 patient-derived iPSC line H196. Stem Cell Research, 2016, 16, 162-165.	0.7	10

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37	Inflammatory markers of CHMP2B-mediated frontotemporal dementia. Journal of Neuroimmunology, 2018, 324, 136-142.	2.3	10
38	Generation of spinocerebellar ataxia type 3 patient-derived induced pluripotent stem cell line SCA3.B11. Stem Cell Research, 2016, 16, 589-592.	0.7	9
39	Quantitative Measurements of Motor Function in Alzheimerâ€™s Disease, Frontotemporal Dementia, and Dementia with Lewy Bodies: A Proof-of-Concept Study. Dementia and Geriatric Cognitive Disorders, 2018, 46, 168-179.	1.5	9
40	Decreased CSF oxytocin relates to measures of social cognitive impairment in Huntington's disease patients. Parkinsonism and Related Disorders, 2022, 99, 23-29.	2.2	8
41	ATXN2 with intermediate-length CAG/CAA repeats does not seem to be a risk factor in hereditary spastic paraplegia. Journal of the Neurological Sciences, 2012, 321, 100-102.	0.6	7
42	Generation of spinocerebellar ataxia type 3 patient-derived induced pluripotent stem cell line SCA3.A11. Stem Cell Research, 2016, 16, 553-556.	0.7	7
43	Social Cognition, Executive Functions and Self-Report of Psychological Distress in Huntingtonâ€™s Disease. PLOS Currents, 2016, 8, .	1.4	7
44	Personality traits in Huntington's disease: An exploratory study of gene expansion carriers and nonâ€carriers. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2016, 171, 1153-1160.	1.7	6
45	Generation of a human induced pluripotent stem cell line via CRISPR-Cas9 mediated integration of a site-specific heterozygous mutation in CHMP2B. Stem Cell Research, 2016, 17, 148-150.	0.7	6
46	Sporadic Creutzfeldt-Jakob Disease in a Woman Married Into a Gerstmann-StrÃussler-Scheinker Family: An Investigation of Prions Transmission via Microchimerism. Journal of Neuropathology and Experimental Neurology, 2018, 77, 673-684.	1.7	6
47	Generation of a human induced pluripotent stem cell line via CRISPR-Cas9 mediated integration of a site-specific homozygous mutation in CHMP2B. Stem Cell Research, 2016, 17, 151-153.	0.7	5
48	Generation of spinocerebellar ataxia type 2 patient-derived iPSC line H196. Stem Cell Research, 2016, 16, 199-201.	0.7	4
49	TMEM106B and ApoE polymorphisms in CHMP2B-mediated frontotemporal dementia (FTD-3). Neurobiology of Aging, 2017, 59, 221.e1-221.e7.	3.1	4
50	Six generations of CHMP2B â€mediated Frontotemporal Dementia: Clinical features, predictive testing, progression, and survival. Acta Neurologica Scandinavica, 2022, 145, 529-540.	2.1	4
51	Generation of spinocerebellar ataxia type 2 patient-derived iPSC line H271. Stem Cell Research, 2016, 16, 159-161.	0.7	3
52	Generation of spinocerebellar ataxia type 2 patient-derived iPSC line H266. Stem Cell Research, 2016, 16, 166-169.	0.7	3
53	Mania triggered by levodopa treatment in a patient with frontotemporal dementia caused by A C9orf72 repeat expansion: A case report. Clinical Neurology and Neurosurgery, 2020, 198, 106147.	1.4	3
54	Endophenotypical drift in Huntingtonâ€™s disease: a 5-year follow-up study. Orphanet Journal of Rare Diseases, 2021, 16, 340.	2.7	3

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55	Peripheral helper T cells in the pathogenesis of multiple sclerosis. Multiple Sclerosis Journal, 2022, 28, 1340-1350.	3.0	3
56	Impairments of social cognition significantly predict the progression of functional decline in Huntingtonâ€™s disease: A 6-year follow-up study. Applied Neuropsychology Adult, 2022, , 1-10.	1.2	3
57	Beneficial effect of intravenous immunoglobulin treatment in a patient with antiphospholipid syndrome associated chorea. Journal of the Neurological Sciences, 2018, 390, 52-53.	0.6	2
58	Paroxysmal Cranial Dyskinesia and Nailâ€Patella Syndrome Caused by a Novel Variant in the LMX1B Gene. Movement Disorders, 2020, 35, 2343-2347.	3.9	2
59	Intellectual Curiosity and Action Initiation are Subtypes of Apathy Affected in Huntington Disease Gene Expansion Carriers. Cognitive and Behavioral Neurology, 2021, 34, 295-302.	0.9	2
60	Widening the spectrum of spinocerebellar ataxia autosomal recessive type 10 (SCAR10). BMJ Case Reports, 2022, 15, e248228.	0.5	1
61	Novel Homozygous Truncating Variant Widens the Spectrum of Early-Onset Multisystemic SYNE1 Ataxia. Cerebellum, 2021, , 1.	2.5	0
62	F21â€…On the association between apathy and deficits of social cognition and executive functions in huntingtonâ€™s disease. , 2021, , .		0
63	D03â€…Quality control for plasma and cerebrospinal fluid samples using mass spectrometry. , 2018, , .		0
64	Cortical Frontoparietal Network Dysfunction in -Frontotemporal Dementia. Frontiers in Aging Neuroscience, 2021, 13, 714220.	3.4	0
65	Intellectual curiosity and action initiation are subtypes of apathy affected in Huntingtonâ€™s disease gene expansion carriers. Alzheimer's and Dementia, 2021, 17, .	0.8	0