Kaja K Selmer

List of Publications by Year
in descending order

59	2,026	279798	265206
papers	citations	23 h-index	42 g-index

65	65	65	4184
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Gain-of-function variants in GABRD reveal a novel pathway for neurodevelopmental disorders and epilepsy. Brain, 2022, 145, 1299-1309.	7.6	34
2	Substantial early changes in bone and calcium metabolism among adult pharmacoresistant epilepsy patients on a modified Atkins diet. Epilepsia, 2022, 63, 880-891.	5.1	11
3	Autosomal Recessive Cerebellar Atrophy and Spastic Ataxia in Patients With Pathogenic Biallelic Variants in GEMIN5. Frontiers in Cell and Developmental Biology, 2022, 10, 783762.	3.7	10
4	Sex-specific disease modifiers in juvenile myoclonic epilepsy. Scientific Reports, 2022, 12, 2785.	3.3	19
5	Genomeâ€wide decrease in <scp>DNA</scp> methylation in adults with epilepsy treated with modified ketogenic diet: A prospective study. Epilepsia, 2022, 63, 2413-2426.	5.1	6
6	Trait impulsivity in Juvenile Myoclonic Epilepsy. Annals of Clinical and Translational Neurology, 2021, 8, 138-152.	3.7	21
7	Loss of function mutations in GEMIN5 cause a neurodevelopmental disorder. Nature Communications, 2021, 12, 2558.	12.8	28
8	A novel somatic mutation inGNB2provides new insights to the pathogenesis of Sturgeâ€“Weber syndrome. Human Molecular Genetics, 2021, 30, 1919-1931.	2.9	15
9	Macrophage migration inhibitory factor: a potential biomarker for chronic low back pain in patients with Modic changes. RMD Open, 2021, 7, e001726.	3.8	7
10	Inherited retinal disease in Norway â€“ a characterization of current clinical and genetic knowledge. Acta Ophthalmologica, 2020, 98, 286-295.	1.1	29
11	Effects of modified Atkins diet on thyroid function in adult patients with pharmacoresistant epilepsy. Epilepsy and Behavior, 2020, 111, 107285.	1.7	7
12	Trait impulsivity correlates with active myoclonic seizures in genetic generalized epilepsy. Epilepsy and Behavior, 2020, 112, 107260.	1.7	8
13	The effect of infliximab in patients with chronic low back pain and Modic changes (the BackToBasic) Tj ETQq1 1 0.784314 rgBT /Overlo Musculoskeletal Disorders, 2020, 21, 698.	1.9	8
14	Elevated hydroxycholesterols in Norwegian patients with hereditary spastic paraplegia SPG5. Journal of the Neurological Sciences, 2020, 419, 117211.	0.6	4
15	Phenotypic spectrum and transcriptomic profile associated with germline variants in TRAF7. Genetics in Medicine, 2020, 22, 1215-1226.	2.4	22
16	Differential Glial Activation in Early Epileptogenesisâ€”Insights From Cell-Specific Analysis of DNA Methylation and Gene Expression in the Contralateral Hippocampus. Frontiers in Neurology, 2020, 11, 573575.	2.4	5
17	Microbiota-gut brain axis involvement in neuropsychiatric disorders. Expert Review of Neurotherapeutics, 2019, 19, 1037-1050.	2.8	116
18	Coexistence of Congenital Adrenal Hyperplasia and Autoimmune Addison's Disease. Frontiers in Endocrinology, 2019, 10, 648.	3.5	2

#	Article	IF	Citations
19	Pharmacokinetic interaction between modified Atkins diet and antiepileptic drugs in adults with drugâ€resistant epilepsy. Epilepsia, 2019, 60, 2235-2244.	5.1	38
20	Psychosocial complications in juvenile myoclonic epilepsy. Epilepsy and Behavior, 2019, 90, 122-128.	1.7	23
21	Identification and characterization of rare toll-like receptor 3 variants in patients with autoimmune Addison's disease. Journal of Translational Autoimmunity, 2019, 1, 100005.	4.0	5
22	A Recurrent Missense Variant in AP2M1 Impairs Clathrin-Mediated Endocytosis and Causes Developmental and Epileptic Encephalopathy. American Journal of Human Genetics, 2019, 104, 1060-1072.	6.2	78
23	Biallelic POLR3A variants confirmed as a frequent cause of hereditary ataxia and spastic paraparesis. Brain, 2019, 142, e12-e12.	7.6	21
24	Neuronal and glial DNA methylation and gene expression changes in early epileptogenesis. PLoS ONE, 2019, 14, e0226575.	2.5	22
25	Neuronal and glial DNA methylation and gene expression changes in early epileptogenesis. , 2019, 14, e0226575.		0
26	Neuronal and glial DNA methylation and gene expression changes in early epileptogenesis. , 2019, 14, e0226575.		0
27	Neuronal and glial DNA methylation and gene expression changes in early epileptogenesis. , 2019, 14, e0226575.		0
28	Neuronal and glial DNA methylation and gene expression changes in early epileptogenesis. , 2019, 14, e0226575.		0
29	Neuronal and glial DNA methylation and gene expression changes in early epileptogenesis. , 2019, 14, e0226575.		0
30	Neuronal and glial DNA methylation and gene expression changes in early epileptogenesis. , 2019, 14, e0226575.		0
31	Aberrant Inclusion of a Poison Exon Causes Dravet Syndrome and Related SCN1A-Associated Genetic Epilepsies. American Journal of Human Genetics, 2018, 103, 1022-1029.	6.2	76
32	Effect of modified Atkins diet in adults with drugâ€resistant focal epilepsy: A randomized clinical trial. Epilepsia, 2018, 59, 1567-1576.	5.1	65
33	NovelUCHL1mutations reveal new insights into ubiquitin processing. Human Molecular Genetics, 2017, 26, ddw391.	2.9	22
34	GLUT1-deficiency syndrome: Report of a four-generation Norwegian family with a mild phenotype. Epilepsy and Behavior, 2017, 70, 1-4.	1.7	16
35	PUF60 variants cause a syndrome of ID, short stature, microcephaly, coloboma, craniofacial, cardiac, renal and spinal features. European Journal of Human Genetics, 2017, 25, 552-559.	2.8	42
36	Prevalence of Renal Angiomyolipomas and Spontaneous Bleeding Related to Angiomyolipomas in Tuberous Sclerosis Complex Patients in France and Norwayâ€"a Questionnaire Study. Urology, 2017, 104, 70-76.	1.0	11

#	Article	IF	Citations
37	Biochemical and genetic characterization of an unusual mild PEX3- related Zellweger spectrum disorder. Molecular Genetics and Metabolism, 2017, 121, 325-328.	1.1	13
38	Prevalence of juvenile myoclonic epilepsy in people <30 years of ageâ€"A populationâ€based study in Norway. Epilepsia, 2017, 58, 105-112.	5.1	28
39	StrÃ¸mme Syndrome Is a Ciliary Disorder Caused by Mutations inCENPF. Human Mutation, 2016, 37, 359-363.	2.5	27
40	Exome Sequencing Fails to Identify the Genetic Cause of Aicardi Syndrome. Molecular Syndromology, 2016, 7, 234-238.	0.8	16
41	Pathogenic variants inKCTD7perturb neuronal K⁺fluxes and glutamine transport. Brain, 2016, 139, 3109-3120.	7.6	31
42	FILTUS: a desktop GUI for fast and efficient detection of disease-causing variants, including a novel autozygosity detector. Bioinformatics, 2016, 32, 1592-1594.	4.1	44
43	Friedreich ataxia in Norway â€" an epidemiological, molecular and clinical study. Orphanet Journal of Rare Diseases, 2015, 10, 108.	2.7	13
44	Aicardi Syndrome: An Epidemiologic and Clinical Study in Norway. Pediatric Neurology, 2015, 52, 182-186.e3.	2.1	19
45	Generalized epilepsy in a family with basal ganglia calcifications and mutations in SLC20A2 and CHRNB2. European Journal of Medical Genetics, 2015, 58, 624-628.	1.3	19
46	A prospective study of the modified Atkins diet for adults with idiopathic generalized epilepsy. Epilepsy and Behavior, 2015, 53, 197-201.	1.7	51
47	Spastic Paraplegia Type 7 Is Associated with Multiple Mitochondrial DNA Deletions. PLoS ONE, 2014, 9, e86340.	2.5	49
48	Does ketogenic diet improve cognitive function in patients with GLUT1-DS? A 6- to 17-month follow-up study. Epilepsy and Behavior, 2014, 39, 111-115.	1.7	39
49	CHD2 mutations in Lennoxâ€"Gastaut syndrome. Epilepsy and Behavior, 2014, 33, 18-21.	1.7	50
50	De Novo Mutations in Synaptic Transmission Genes Including DNM1 Cause Epileptic Encephalopathies. American Journal of Human Genetics, 2014, 95, 360-370.	6.2	388
51	Occurrence of GLUT1 deficiency syndrome in patients treated with ketogenic diet. Epilepsy and Behavior, 2014, 32, 76-78.	1.7	11
52	Good outcome in patients with early dietary treatment of <scp>GLUT</scp>â€1 deficiency syndrome: results from a retrospective Norwegian study. Developmental Medicine and Child Neurology, 2013, 55, 440-447.	2.1	56
53	Copy number variants in adult patients with Lennoxâ€"Gastaut syndrome features. Epilepsy Research, 2013, 105, 110-117.	1.6	35
54	A mild form of Mucopolysaccharidosis IIIB diagnosed with targeted next-generation sequencing of linked genomic regions. European Journal of Human Genetics, 2012, 20, 58-63.	2.8	24

#	Article	IF	Citations
55	A de novo 163Âkb interstitial 1q44 microdeletion in a boy with thin corpus callosum, psychomotor delay and seizures. European Journal of Medical Genetics, 2012, 55, 715-718.	1.3	11
56	Autosomal dominant pericentral retinal dystrophy caused by a novel missense mutation in the TOPORS gene. Acta Ophthalmologica, 2010, 88, 323-328.	1.1	21
57	Genome-wide Linkage Analysis with Clustered SNP Markers. Journal of Biomolecular Screening, 2009, 14, 92-96.	2.6	12
58	SCN1A mutation screening in adult patients with Lennoxâ€“Gastaut syndrome features. Epilepsy and Behavior, 2009, 16, 555-557.	1.7	23
59	SLC9A6 Mutations Cause X-Linked Mental Retardation, Microcephaly, Epilepsy, and Ataxia, a Phenotype Mimicking Angelman Syndrome. American Journal of Human Genetics, 2008, 82, 1003-1010.	6.2	209