Berge A Minassian

List of Publications by Year
in descending order

85 papers	4,701 citations	109321 35 h-index	106344 65 g-index
90 all docs	90 docs citations	90 times ranked	4882 citing authors

#	Article	IF	Citations
1	Mutations in a gene encoding a novel protein tyrosine phosphatase cause progressive myoclonus epilepsy. Nature Genetics, 1998, 20, 171-174.	21.4	499
2	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2017, 101, 664-685.	6.2	337
3	Mutations in NHLRC1 cause progressive myoclonus epilepsy. Nature Genetics, 2003, 35, 125-127.	21.4	294
4	PTG Depletion Removes Lafora Bodies and Rescues the Fatal Epilepsy of Lafora Disease. PLoS Genetics, 2011, 7, e1002037.	3.5	185
5	Laforin is a glycogen phosphatase, deficiency of which leads to elevated phosphorylation of glycogen in vivo. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 19262-19266.	7.1	181
6	Brain Dopamineâ€“Serotonin Vesicular Transport Disease and Its Treatment. New England Journal of Medicine, 2013, 368, 543-550.	27.0	176
7	Laforaâ€™s disease: towards a clinical, pathologic, and molecular synthesis. Pediatric Neurology, 2001, 25, 21-29.	2.1	174
8	Abnormal Metabolism of Glycogen Phosphate as a Cause for Lafora Disease. Journal of Biological Chemistry, 2008, 283, 33816-33825.	3.4	153
9	Novel glycogen synthase kinase 3 and ubiquitination pathways in progressive myoclonus epilepsy. Human Molecular Genetics, 2005, 14, 2727-2736.	2.9	146
10	Lafora disease. Epileptic Disorders, 2016, 18, 38-62.	1.3	127
11	VMA21 deficiency prevents vacuolar ATPase assembly and causes autophagic vacuolar myopathy. Acta Neuropathologica, 2013, 125, 439-457.	7.7	119
12	Lafora disease â€” from pathogenesis to treatment strategies. Nature Reviews Neurology, 2018, 14, 606-617.	10.1	110
13	Laforin preferentially binds the neurotoxic starch-like polyglucosans, which form in its absence in progressive myoclonus epilepsy. Human Molecular Genetics, 2004, 13, 1117-1129.	2.9	101
14	Glycogen hyperphosphorylation underlies lafora body formation. Annals of Neurology, 2010, 68, 925-933.	5.3	96
15	Inhibiting glycogen synthesis prevents lafora disease in a mouse model. Annals of Neurology, 2013, 74, 297-300.	5.3	91
16	LGI2 Truncation Causes a Remitting Focal Epilepsy in Dogs. PLoS Genetics, 2011, 7, e1002194.	3.5	88
17	Hyperphosphorylation of Glucosyl C6 Carbons and Altered Structure of Glycogen in the Neurodegenerative Epilepsy Lafora Disease. Cell Metabolism, 2013, 17, 756-767.	16.2	80
18	Early-onset Lafora body disease. Brain, 2012, 135, 2684-2698.	7.6	76

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19	PTG protein depletion rescues malinâ€deficient Lafora disease in mouse. Annals of Neurology, 2014, 75, 442-446.	5.3	76
20	Efficacy and tolerability of perampanel in ten patients with Lafora disease. Epilepsy and Behavior, 2016, 62, 132-135.	1.7	76
21	Lafora disease offers a unique window into neuronal glycogen metabolism. Journal of Biological Chemistry, 2018, 293, 7117-7125.	3.4	71
22	From Genetic Testing to Precision Medicine in Epilepsy. Neurotherapeutics, 2020, 17, 609-615.	4.4	62
23	Lafora progressive myoclonus epilepsy mutation database-EPM2A and NHLRC1 (EMP2B) genes. Human Mutation, 2005, 26, 397-397.	2.5	59
24	Abnormal glycogen chain length pattern, not hyperphosphorylation, is critical in Lafora disease. EMBO Molecular Medicine, 2017, 9, 906-917.	6.9	59
25	Structural basis of glycogen branching enzyme deficiency and pharmacologic rescue by rational peptide design. Human Molecular Genetics, 2015, 24, 5667-5676.	2.9	58
26	Both gainâ€ofâ€function and lossâ€ofâ€function de novo <scp>CACNA</scp>1A mutations cause severe developmental epileptic encephalopathies in the spectrum of Lennoxâ€Gastaut syndrome. Epilepsia, 2019, 60, 1881-1894.	5.1	57
27	The best evidence for progressive myoclonic epilepsy: A pathway to precision therapy. Seizure: the Journal of the British Epilepsy Association, 2019, 71, 247-257.	2.0	57
28	Pathogenesis of Lafora Disease: Transition of Soluble Glycogen to Insoluble Polyglucosan. International Journal of Molecular Sciences, 2017, 18, 1743.	4.1	55
29	Genome annotation for clinical genomic diagnostics: strengths and weaknesses. Genome Medicine, 2017, 9, 49.	8.2	51
30	Transition from glycogen to starch metabolism in Archaeplastida. Trends in Plant Science, 2014, 19, 18-28.	8.8	48
31	Typical progression of myoclonic epilepsy of the Lafora type: a case report. Nature Clinical Practice Neurology, 2008, 4, 106-111.	2.5	47
32	IQSEC2-related encephalopathy in males and females: a comparative study including 37 novel patients. Genetics in Medicine, 2019, 21, 837-849.	2.4	47
33	Increased Laforin and Laforin Binding to Glycogen Underlie Lafora Body Formation in Malin-deficient Lafora Disease. Journal of Biological Chemistry, 2012, 287, 25650-25659.	3.4	44
34	Efficient Delivery of Structurally Diverse Protein Cargo into Mammalian Cells by a Bacterial Toxin. Molecular Pharmaceutics, 2015, 12, 2962-2971.	4.6	40
35	Skeletal Muscle Glycogen Chain Length Correlates with Insolubility in Mouse Models of Polyglucosan-Associated Neurodegenerative Diseases. Cell Reports, 2019, 27, 1334-1344.e6.	6.4	40
36	Progressive myoclonus epilepsy. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2013, 113, 1731-1736.	1.8	36

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37	Glycogen metabolism in tissues from a mouse model of Lafora disease. Archives of Biochemistry and Biophysics, 2007, 457, 264-269.	3.0	35
38	X-linked myopathy with excessive autophagy: a failure of self-eating. Acta Neuropathologica, 2015, 129, 383-390.	7.7	33
39	Deep IntronicGBE1Mutation in Manifesting Heterozygous Patients With Adult Polyglucosan Body Disease. JAMA Neurology, 2015, 72, 441.	9.0	33
40	Identification of genomic deletions spanning the PCDH19 gene in two unrelated girls with intellectual disability and seizures. Clinical Genetics, 2012, 82, 540-545.	2.0	30
41	Gys1antisense therapy rescues neuropathological bases of murine Lafora disease. Brain, 2021, 144, 2985-2993.	7.6	30
42	Lafora disease in miniature Wirehaired Dachshunds. PLoS ONE, 2017, 12, e0182024.	2.5	27
43	Targeting Gys1 with AAVâ€SaCas9 Decreases Pathogenic Polyglucosan Bodies and Neuroinflammation in Adult Polyglucosan Body and Lafora Disease Mouse Models. Neurotherapeutics, 2021, 18, 1414-1425.	4.4	26
44	Non-coding VMA21 deletions cause X-linked Myopathy with Excessive Autophagy. Neuromuscular Disorders, 2015, 25, 207-211.	0.6	25
45	Cardiac autophagic vacuolation in severe X-linked myopathy with excessive autophagy. Neuromuscular Disorders, 2017, 27, 185-187.	0.6	23
46	An inducible glycogen synthase-1 knockout halts but does not reverse Lafora disease progression in mice. Journal of Biological Chemistry, 2021, 296, 100150.	3.4	23
47	Update on Pharmacological Treatment of Progressive Myoclonus Epilepsies. Current Pharmaceutical Design, 2018, 23, 5662-5666.	1.9	23
48	Laforin Prevents Stress-Induced Polyglucosan Body Formation and Lafora Disease Progression in Neurons. Molecular Neurobiology, 2013, 48, 49-61.	4.0	19
49	Lafora Disease: A Review of Molecular Mechanisms and Pathology. Neuropediatrics, 2018, 49, 357-362.	0.6	19
50	Congenital autophagic vacuolar myopathy is allelic to X-linked myopathy with excessive autophagy. Neurology, 2015, 84, 1714-1716.	1.1	18
51	Dimeric Quaternary Structure of Human Laforin. Journal of Biological Chemistry, 2015, 290, 4552-4559.	3.4	18
52	Nationwide genetic testing towards eliminating Lafora disease from Miniature Wirehaired Dachshunds in the United Kingdom. Canine Genetics and Epidemiology, 2018, 5, 2.	2.8	18
53	Late adultâ€onset of Xâ€linked myopathy with excessive autophagy. Muscle and Nerve, 2014, 50, 138-144.	2.2	17
54	The 5th International Lafora Epilepsy Workshop: Basic science elucidating therapeutic options and preparing for therapies in the clinic. Epilepsy and Behavior, 2020, 103, 106839.	1.7	17

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55	GYS1 or PPP1R3C deficiency rescues murine adult polyglucosan body disease. Annals of Clinical and Translational Neurology, 2020, 7, 2186-2198.	3.7	16
56	Lafora disease: Current biology and therapeutic approaches. Revue Neurologique, 2022, 178, 315-325.	1.5	16
57	The progressive myoclonus epilepsies. Progress in Brain Research, 2014, 213, 113-122.	1.4	15
58	Unverricht-Lundborg Progressive Myoclonus Epilepsy in Oman. Pediatric Neurology, 2008, 38, 252-255.	2.1	14
59	Deficiency of a Glycogen Synthase-associated Protein, Epm2aip1, Causes Decreased Glycogen Synthesis and Hepatic Insulin Resistance. Journal of Biological Chemistry, 2013, 288, 34627-34637.	3.4	14
60	A novel image-based high-throughput screening assay discovers therapeutic candidates for adult polyglucosan body disease. Biochemical Journal, 2017, 474, 3403-3420.	3.7	14
61	AAV-Mediated Artificial miRNA Reduces Pathogenic Polyglucosan Bodies and Neuroinflammation in Adult Polyglucosan Body and Lafora Disease Mouse Models. Neurotherapeutics, 2022, 19, 982-993.	4.4	14
62	Ppp1r3d deficiency preferentially inhibits neuronal and cardiac Lafora body formation in a mouse model of the fatal epilepsy Lafora disease. Journal of Neurochemistry, 2021, 157, 1897-1910.	3.9	13
63	Alleviation of a polyglucosan storage disorder by enhancement of autophagic glycogen catabolism. EMBO Molecular Medicine, 2021, 13, e14554.	6.9	13
64	No cardiomyopathy in X-linked myopathy with excessive autophagy. Neuromuscular Disorders, 2015, 25, 485-487.	0.6	12
65	Glycogen synthase downregulation rescues the amylopectinosis of murine RBCK1 deficiency. Brain, 2022, 145, 2361-2377.	7.6	12
66	The role of common genetic variation in presumed monogenic epilepsies. EBioMedicine, 2022, 81, 104098.	6.1	12
67	Defining the phenotype of FHF1 developmental and epileptic encephalopathy. Epilepsia, 2020, 61, e71-e78.	5.1	11
68	Phosphorylation prevents polyglucosan transport in Lafora disease. Neurology, 2012, 79, 100-102.	1.1	7
69	Dominant LMAN2L mutation causes intellectual disability with remitting epilepsy. Annals of Clinical and Translational Neurology, 2019, 6, 807-811.	3.7	7
70	Ketogenic diet reduces Lafora bodies in murine Lafora disease. Neurology: Genetics, 2020, 6, e533.	1.9	7
71	Diabetes Mellitus in a Patient With Lafora Disease: Possible Links With Pancreatic Î²-Cell Dysfunction and Insulin Resistance. Frontiers in Pediatrics, 2019, 6, 424.	1.9	6
72	Genotypes and phenotypes of patients with Lafora disease living in Germany. Neurological Research and Practice, 2019, 1, .	2.0	6

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73	Exploiting the diphtheria toxin internalization receptor enhances delivery of proteins to lysosomes for enzyme replacement therapy. Science Advances, 2020, 6, .	10.3	6
74	Sensitive quantification of Î±-glucans in mouse tissues, cell cultures, and human cerebrospinal fluid. Journal of Biological Chemistry, 2020, 295, 14698-14709.	3.4	5
75	SGK1 (glucose transport), dishevelled2 (wnt signaling), LC3/p62 (autophagy) and p53 (apoptosis) proteins are unaltered in Lafora disease. The All Results Journal Biol, 2016, 7, 28-33.	2.0	5
76	Retinal alterations in patients with Lafora disease. American Journal of Ophthalmology Case Reports, 2021, 23, 101146.	0.7	4
77	Extraneurological sparing in longâ€lived typical Lafora disease. Epilepsia Open, 2018, 3, 295-298.	2.4	3
78	Gene Therapy: Novel Approaches to Targeting Monogenic Epilepsies. Frontiers in Neurology, 0, 13, .	2.4	3
79	Assessment of burden and segregation profiles of <scp>CNVs</scp> in patients with epilepsy. Annals of Clinical and Translational Neurology, 2022, 9, 1050-1058.	3.7	2
80	Animal Models of Lafora Disease. Microscopy and Microanalysis, 2006, 12, 12-13.	0.4	1
81	Fatal hepatic failure and pontine and extrapontine myelinolysis in XMEA. Neurology, 2016, 87, 1417-1419.	1.1	1
82	EPM2A in-frame deletion slows neurological decline in Lafora Disease. Seizure: the Journal of the British Epilepsy Association, 2021, 91, 97-98.	2.0	1
83	Thrombocytopathy and leukocytopathy in X-linked Myopathy with Excessive Autophagy (XMEA). Microscopy and Microanalysis, 2008, 14, 1524-1525.	0.4	0
84	The antioxidant MnTBAP does not effectively downregulate CD4 expression in T cells in vivo. Journal of Neuroimmunology, 2021, 354, 577544.	2.3	0
85	Lafora Disease: A Perspective in Molecular Mechanism and Pathology. Neuropediatrics, 2018, 49, S1-S12.	0.6	0