John H Livingston

List of Publications by Year
in descending order

41 papers	4,091 citations	304602 22 h-index	289141 40 g-index
42 all docs	42 docs citations	42 times ranked	5557 citing authors

#	Article	IF	Citations
1	Missense mutation of MAL causes a rare leukodystrophy similar to Pelizaeus-Merzbacher disease. European Journal of Human Genetics, 2022, 30, 860-864.	1.4	4
2	Leukoencephalopathy with calcifications and cysts: Genetic and phenotypic spectrum. American Journal of Medical Genetics, Part A, 2021, 185, 15-25.	0.7	15
3	Treatments in Aicardiâ€“GoutiÃ¨res syndrome. Developmental Medicine and Child Neurology, 2020, 62, 42-47.	1.1	70
4	Biallelic Mutations in MTPAP Associated with a Lethal Encephalopathy. Neuropediatrics, 2020, 51, 178-184.	0.3	3
5	Genetic and phenotypic spectrum associated with IFIH1 gainâ€ofâ€function. Human Mutation, 2020, 41, 837-849.	1.1	63
6	cGAS-mediated induction of type I interferon due to inborn errors of histone pre-mRNA processing. Nature Genetics, 2020, 52, 1364-1372.	9.4	105
7	Clinical and radiological characterization of novel <scp>FIG4</scp>â€related combined system disease with neuropathy. Clinical Genetics, 2020, 98, 147-154.	1.0	8
8	Bi-allelic JAM2 Variants Lead to Early-Onset Recessive Primary Familial Brain Calcification. American Journal of Human Genetics, 2020, 106, 412-421.	2.6	47
9	Biallelic mutations in NRROS cause an early onset lethal microgliopathy. Acta Neuropathologica, 2020, 139, 947-951.	3.9	17
10	Surveillance for variant CJD: should more children with neurodegenerative diseases have autopsies?. Archives of Disease in Childhood, 2019, 104, 360-365.	1.0	7
11	Cerebral hypomyelination associated with biallelic variants of FIG4. Human Mutation, 2019, 40, 619-630.	1.1	18
12	Severe type I interferonopathy and unrestrained interferon signaling due to a homozygous germline mutation in STAT2. Science Immunology, 2019, 4, .	5.6	80
13	Whole Exon Deletion in the GFAP Gene Is a Novel Molecular Mechanism Causing Alexander Disease. Neuropediatrics, 2018, 49, 118-122.	0.3	6
14	Autosomal-dominant early-onset spastic paraparesis with brain calcification due to IFIH1 gain-of-function. Human Mutation, 2018, 39, 1076-1080.	1.1	8
15	Genetic, Phenotypic, and Interferon Biomarker Status in ADAR1-Related Neurological Disease. Neuropediatrics, 2017, 48, 166-184.	0.3	62
16	Leukoencephalopathy with calcification and cysts: A cerebral microangiopathy caused by mutations in SNORD118. Journal of the Neurological Sciences, 2017, 372, 443.	0.3	2
17	An unusual neuroimaging finding and response to immunotherapy in a child with genetically confirmed vanishing white matter disease. European Journal of Paediatric Neurology, 2017, 21, 410-413.	0.7	7
18	Clinical, radiological and possible pathological overlap of cystic leukoencephalopathy without megalencephaly and Aicardi-GoutiÃ¨res syndrome. European Journal of Paediatric Neurology, 2016, 20, 604-610.	0.7	29

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19	Mutations in SNORD118 cause the cerebral microangiopathy leukoencephalopathy with calcifications and cysts. Nature Genetics, 2016, 48, 1185-1192.	9.4	114
20	Neurologic Phenotypes Associated with Mutations in TREX1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, ADAR1, and IFIH1: Aicardiâ€“GoutiÃ¨res Syndrome and Beyond. Neuropediatrics, 2016, 47, 355-360.	0.3	127
21	Spondyloenchondrodysplasia Due to Mutations in ACP5: A Comprehensive Survey. Journal of Clinical Immunology, 2016, 36, 220-234.	2.0	71
22	CNS infections. Journal of Pediatric Neurology, 2015, 08, 089-090.	0.0	0
23	Early-Onset Aicardi-GoutiÃ¨res Syndrome. Journal of Child Neurology, 2015, 30, 1343-1348.	0.7	33
24	Next generation child neurologists. Developmental Medicine and Child Neurology, 2015, 57, 4-5.	1.1	0
25	Leukoencephalopathy with Calcifications and Cysts: A Purely Neurological Disorder Distinct from Coats Plus. Neuropediatrics, 2014, 45, 175-182.	0.3	41
26	Mutations in ADAR1, IFIH1, and RNASEH2B Presenting As Spastic Paraplegia. Neuropediatrics, 2014, 45, 386-391.	0.3	72
27	Basal Ganglia Calcification in a Patient With Beta-Propeller Protein-Associated Neurodegeneration. Pediatric Neurology, 2014, 51, 843-845.	1.0	17
28	Common pathways of intracranial calcification and the role of the pericyte: insights from neuropathology. Developmental Medicine and Child Neurology, 2014, 56, 924-925.	1.1	1
29	A type I interferon signature identifies bilateral striatal necrosis due to mutations inADAR1. Journal of Medical Genetics, 2014, 51, 76-82.	1.5	118
30	Gain-of-function mutations in IFIH1 cause a spectrum of human disease phenotypes associated with upregulated type I interferon signaling. Nature Genetics, 2014, 46, 503-509.	9.4	490
31	Intracranial calcification in childhood: a review of aetiologies and recognizable phenotypes. Developmental Medicine and Child Neurology, 2014, 56, 612-626.	1.1	132
32	Assessment of interferon-related biomarkers in Aicardi-GoutiÃ¨res syndrome associated with mutations in TREX1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, and ADAR: a case-control study. Lancet Neurology, The, 2013, 12, 1159-1169.	4.9	473
33	Recognizable phenotypes associated with intracranial calcification. Developmental Medicine and Child Neurology, 2013, 55, 46-57.	1.1	68
34	COL4A1-Related Disease: Raised Creatine Kinase and Cerebral Calcification as Useful Pointers. Neuropediatrics, 2012, 43, 283-288.	0.3	20
35	Intracranial calcification in early infantile Krabbe disease: nothing new under the sun. Developmental Medicine and Child Neurology, 2012, 54, 376-379.	1.1	14
36	Mutations in CTC1, encoding conserved telomere maintenance component 1, cause Coats plus. Nature Genetics, 2012, 44, 338-342.	9.4	234

#	Article	IF	Citations
37	Mutations in ADAR1 cause Aicardi-GoutiÃ¨res syndrome associated with a type I interferon signature. Nature Genetics, 2012, 44, 1243-1248.	9.4	712
38	Poems by children as patient-reported outcomes. Developmental Medicine and Child Neurology, 2012, 54, 52-53.	1.1	1
39	Bradycardia without associated hypertension: a common sign of ventriculo-peritoneal shunt malfunction. Child's Nervous System, 2011, 27, 729-733.	0.6	6
40	A Novel Inherited Mutation in the Voltage Sensor Region of SCN1A Is Associated With Panayiotopoulos Syndrome in Siblings and Generalized Epilepsy With Febrile Seizures Plus. Journal of Child Neurology, 2009, 24, 503-508.	0.7	41
41	Mutations in the gene encoding the 3â€²-5â€² DNA exonuclease TREX1 cause Aicardi-GoutiÃ¨res syndrome at the AGS1 locus. Nature Genetics, 2006, 38, 917-920.	9.4	752