

John H Livingston

List of Publications by Year in descending order

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Version: 2024-02-01

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	Mutations in the gene encoding the 3'→5' DNA exonuclease TREX1 cause Aicardi-Goutières syndrome at the AGS1 locus. <i>Nature Genetics</i> , 2006, 38, 917-920.	9.4	752
2	Mutations in ADAR1 cause Aicardi-Goutières syndrome associated with a type I interferon signature. <i>Nature Genetics</i> , 2012, 44, 1243-1248.	9.4	712
3	Gain-of-function mutations in IFIH1 cause a spectrum of human disease phenotypes associated with upregulated type I interferon signaling. <i>Nature Genetics</i> , 2014, 46, 503-509.	9.4	490
4	Assessment of interferon-related biomarkers in Aicardi-Goutières syndrome associated with mutations in TREX1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, and ADAR: a case-control study. <i>Lancet Neurology</i> , The, 2013, 12, 1159-1169.	4.9	473
5	Mutations in CTC1, encoding conserved telomere maintenance component 1, cause Coats plus. <i>Nature Genetics</i> , 2012, 44, 338-342.	9.4	234
6	Intracranial calcification in childhood: a review of aetiologies and recognizable phenotypes. <i>Developmental Medicine and Child Neurology</i> , 2014, 56, 612-626.	1.1	132
7	Neurologic Phenotypes Associated with Mutations in TREX1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, ADAR1, and IFIH1: Aicardi-Goutières Syndrome and Beyond. <i>Neuropediatrics</i> , 2016, 47, 355-360.	0.3	127
8	A type I interferon signature identifies bilateral striatal necrosis due to mutations in <i>ADAR1</i> . <i>Journal of Medical Genetics</i> , 2014, 51, 76-82.	1.5	118
9	Mutations in SNORD118 cause the cerebral microangiopathy leukoencephalopathy with calcifications and cysts. <i>Nature Genetics</i> , 2016, 48, 1185-1192.	9.4	114
10	cGAS-mediated induction of type I interferon due to inborn errors of histone pre-mRNA processing. <i>Nature Genetics</i> , 2020, 52, 1364-1372.	9.4	105
11	Severe type I interferonopathy and unrestrained interferon signaling due to a homozygous germline mutation in <i>STAT2</i> . <i>Science Immunology</i> , 2019, 4, .	5.6	80
12	Mutations in ADAR1, IFIH1, and RNASEH2B Presenting As Spastic Paraplegia. <i>Neuropediatrics</i> , 2014, 45, 386-391.	0.3	72
13	Spondyloenchondrodysplasia Due to Mutations in ACP5: A Comprehensive Survey. <i>Journal of Clinical Immunology</i> , 2016, 36, 220-234.	2.0	71
14	Treatments in Aicardi-Goutières syndrome. <i>Developmental Medicine and Child Neurology</i> , 2020, 62, 42-47.	1.1	70
15	Recognizable phenotypes associated with intracranial calcification. <i>Developmental Medicine and Child Neurology</i> , 2013, 55, 46-57.	1.1	68
16	Genetic and phenotypic spectrum associated with IFIH1 gain-of-function. <i>Human Mutation</i> , 2020, 41, 837-849.	1.1	63
17	Genetic, Phenotypic, and Interferon Biomarker Status in ADAR1-Related Neurological Disease. <i>Neuropediatrics</i> , 2017, 48, 166-184.	0.3	62
18	Bi-allelic JAM2 Variants Lead to Early-Onset Recessive Primary Familial Brain Calcification. <i>American Journal of Human Genetics</i> , 2020, 106, 412-421.	2.6	47

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19	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. <i>Journal of Child Neurology</i> , 2009, 24, 503-508.	0.7	41
20	Leukoencephalopathy with Calcifications and Cysts: A Purely Neurological Disorder Distinct from Coats Plus. <i>Neuropediatrics</i> , 2014, 45, 175-182.	0.3	41
21	Early-Onset Aicardi-Goutières Syndrome. <i>Journal of Child Neurology</i> , 2015, 30, 1343-1348.	0.7	33
22	Clinical, radiological and possible pathological overlap of cystic leukoencephalopathy without megalencephaly and Aicardi-Goutières syndrome. <i>European Journal of Paediatric Neurology</i> , 2016, 20, 604-610.	0.7	29
23	COL4A1-Related Disease: Raised Creatine Kinase and Cerebral Calcification as Useful Pointers. <i>Neuropediatrics</i> , 2012, 43, 283-288.	0.3	20
24	Cerebral hypomyelination associated with biallelic variants of <i>FIG4</i> . <i>Human Mutation</i> , 2019, 40, 619-630.	1.1	18
25	Basal Ganglia Calcification in a Patient With Beta-Propeller Protein-Associated Neurodegeneration. <i>Pediatric Neurology</i> , 2014, 51, 843-845.	1.0	17
26	Biallelic mutations in <i>NRROS</i> cause an early onset lethal microgliopathy. <i>Acta Neuropathologica</i> , 2020, 139, 947-951.	3.9	17
27	Leukoencephalopathy with calcifications and cysts: Genetic and phenotypic spectrum. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 15-25.	0.7	15
28	Intracranial calcification in early infantile Krabbe disease: nothing new under the sun. <i>Developmental Medicine and Child Neurology</i> , 2012, 54, 376-379.	1.1	14
29	Autosomal-dominant early-onset spastic paraparesis with brain calcification due to <i>IFIH1</i> gain-of-function. <i>Human Mutation</i> , 2018, 39, 1076-1080.	1.1	8
30	Clinical and radiological characterization of novel <i>FIG4</i> -related combined system disease with neuropathy. <i>Clinical Genetics</i> , 2020, 98, 147-154.	1.0	8
31	An unusual neuroimaging finding and response to immunotherapy in a child with genetically confirmed vanishing white matter disease. <i>European Journal of Paediatric Neurology</i> , 2017, 21, 410-413.	0.7	7
32	Surveillance for variant CJD: should more children with neurodegenerative diseases have autopsies?. <i>Archives of Disease in Childhood</i> , 2019, 104, 360-365.	1.0	7
33	Bradycardia without associated hypertension: a common sign of ventriculo-peritoneal shunt malfunction. <i>Child's Nervous System</i> , 2011, 27, 729-733.	0.6	6
34	Whole Exon Deletion in the <i>GFAP</i> Gene Is a Novel Molecular Mechanism Causing Alexander Disease. <i>Neuropediatrics</i> , 2018, 49, 118-122.	0.3	6
35	Missense mutation of <i>MAL</i> causes a rare leukodystrophy similar to Pelizaeus-Merzbacher disease. <i>European Journal of Human Genetics</i> , 2022, 30, 860-864.	1.4	4
36	Biallelic Mutations in <i>MTPAP</i> Associated with a Lethal Encephalopathy. <i>Neuropediatrics</i> , 2020, 51, 178-184.	0.3	3

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37	Leukoencephalopathy with calcification and cysts: A cerebral microangiopathy caused by mutations in SNORD118. <i>Journal of the Neurological Sciences</i> , 2017, 372, 443.	0.3	2
38	Poems by children as patient-reported outcomes. <i>Developmental Medicine and Child Neurology</i> , 2012, 54, 52-53.	1.1	1
39	Common pathways of intracranial calcification and the role of the pericyte: insights from neuropathology. <i>Developmental Medicine and Child Neurology</i> , 2014, 56, 924-925.	1.1	1
40	CNS infections. <i>Journal of Pediatric Neurology</i> , 2015, 08, 089-090.	0.0	0
41	Next generation child neurologists. <i>Developmental Medicine and Child Neurology</i> , 2015, 57, 4-5.	1.1	0