GeneviÃ¨ve Bernard

List of Publications by Year
in descending order

124 papers	5,121 citations	147801 31 h-index	106344 65 g-index
130 all docs	130 docs citations	130 times ranked	7250 citing authors

#	Article	IF	Citations
1	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.	10.2	473
2	Characterization of human disease phenotypes associated with mutations in TREX1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, ADAR, and IFIH1. American Journal of Medical Genetics, Part A, 2015, 167, 296-312.	1.2	447
3	Utility of wholeâ€exome sequencing for those near the end of the diagnostic odyssey: time to address gaps in care. Clinical Genetics, 2016, 89, 275-284.	2.0	323
4	Mutations of POLR3A Encoding a Catalytic Subunit of RNA Polymerase Pol III Cause a Recessive Hypomyelinating Leukodystrophy. American Journal of Human Genetics, 2011, 89, 415-423.	6.2	219
5	Exosomes Induce Fibroblast Differentiation into Cancer-Associated Fibroblasts through TGFÎ² Signaling. Molecular Cancer Research, 2018, 16, 1196-1204.	3.4	200
6	Exome Sequencing Identifies FUS Mutations as a Cause of Essential Tremor. American Journal of Human Genetics, 2012, 91, 313-319.	6.2	176
7	Clinical spectrum of 4H leukodystrophy caused by POLR3A and POLR3B mutations. Neurology, 2014, 83, 1898-1905.	1.1	170
8	A clinical approach to the diagnosis of patients with leukodystrophies and genetic leukoencephelopathies. Molecular Genetics and Metabolism, 2015, 114, 501-515.	1.1	163
9	Mutations in the Mitochondrial Methionyl-tRNA Synthetase Cause a Neurodegenerative Phenotype in Flies and a Recessive Ataxia (ARSAL) in Humans. PLoS Biology, 2012, 10, e1001288.	5.6	147
10	Recessive Mutations in POLR3B, Encoding the Second Largest Subunit of Pol III, Cause a Rare Hypomyelinating Leukodystrophy. American Journal of Human Genetics, 2011, 89, 652-655.	6.2	139
11	Hypomyelinating leukodystrophies: Translational research progress and prospects. Annals of Neurology, 2014, 76, 5-19.	5.3	132
12	Recessive mutations in POLR1C cause a leukodystrophy by impairing biogenesis of RNA polymerase III. Nature Communications, 2015, 6, 7623.	12.8	127
13	Whole exome sequencing in patients with white matter abnormalities. Annals of Neurology, 2016, 79, 1031-1037.	5.3	116
14	Mutations inPOLR3AandPOLR3Bare a major cause of hypomyelinating leukodystrophies with or without dental abnormalities and/or hypogonadotropic hypogonadism. Journal of Medical Genetics, 2013, 50, 194-197.	3.2	109
15	Plasticity of locomotor sensorimotor interactions after peripheral and/or spinal lesions. Brain Research Reviews, 2008, 57, 228-240.	9.0	81
16	Genome-wide association study in essential tremor identifies three new loci. Brain, 2016, 139, 3163-3169.	7.6	78
17	Channelopathies: A Review. Pediatric Neurology, 2008, 38, 73-85.	2.1	69
18	Diffuse hypomyelination is not obligate for POLR3-related disorders. Neurology, 2016, 86, 1622-1626.	1.1	65

#	Article	IF	Citations
19	Bi-allelic Mutations in EPRS, Encoding the Glutamyl-Prolyl-Aminoacyl-tRNA Synthetase, Cause a Hypomyelinating Leukodystrophy. American Journal of Human Genetics, 2018, 102, 676-684.	6.2	58
20	4H Syndrome With Late-Onset Growth Hormone Deficiency Caused by POLR3A Mutations. Archives of Neurology, 2012, 69, 920-3.	4.5	56
21	TUBB4A de novo mutations cause isolated hypomyelination. Neurology, 2014, 83, 898-902.	1.1	52
22	Diversity of ARSACS Mutations in French-Canadians. Canadian Journal of Neurological Sciences, 2013, 40, 61-66.	0.5	51
23	Longitudinal Outcomes in the 2014 Acute Flaccid Paralysis Cluster in Canada. Journal of Child Neurology, 2017, 32, 301-307.	1.4	50
24	Brain Magnetic Resonance Imaging (MRI) Pattern Recognition in Pol III-Related Leukodystrophies. Journal of Child Neurology, 2014, 29, 214-220.	1.4	47
25	Tissue-engineered human 3D model of bladder cancer for invasion study and drug discovery. Biomaterials, 2017, 145, 233-241.	11.4	47
26	Disease specific therapies in leukodystrophies and leukoencephalopathies. Molecular Genetics and Metabolism, 2015, 114, 527-536.	1.1	45
27	Revised consensus statement on the preventive and symptomatic care of patients with leukodystrophies. Molecular Genetics and Metabolism, 2017, 122, 18-32.	1.1	42
28	Recovery From Central Nervous System Acute Demyelination in Children. Pediatrics, 2015, 136, e115-e123.	2.1	40
29	Tremorâ€“ataxia with central hypomyelination (TACH) leukodystrophy maps to chromosome 10q22.3â€“10q23.31. Neurogenetics, 2010, 11, 457-464.	1.4	39
30	Matchmaking facilitates the diagnosis of an autosomal-recessive mitochondrial disease caused by biallelic mutation of the tRNA isopentenyltransferase (TRIT1) gene. Human Mutation, 2017, 38, 511-516.	2.5	39
31	Leukodystrophy-associated POLR3A mutations down-regulate the RNA polymerase III transcript and important regulatory RNA BC200. Journal of Biological Chemistry, 2019, 294, 7445-7459.	3.4	39
32	Clinical spectrum of POLR3-related leukodystrophy caused by biallelic POLR1C pathogenic variants. Neurology: Genetics, 2019, 5, e369.	1.9	38
33	Surgical Option for the Correction of Peyronie's Disease: An Autologous Tissue-Engineered Endothelialized Graft. Journal of Sexual Medicine, 2011, 8, 3227-3235.	0.6	33
34	Absence of neurological abnormalities in mice homozygous for the Polr3a G672E hypomyelinating leukodystrophy mutation. Molecular Brain, 2017, 10, 13.	2.6	33
35	Insights on the Interactions of Synthetic Amphipathic Peptides with Model Membranes as Revealed by 31P and 2H Solid-State NMR and Infrared Spectroscopies. Biophysical Journal, 2006, 90, 4071-4084.	0.5	32
36	Consensus statement on preventive and symptomatic care of leukodystrophy patients. Molecular Genetics and Metabolism, 2015, 114, 516-526.	1.1	29

#	Article	IF	Citations
37	Adipose-derived stromal cells for the reconstruction of a human vesical equivalent. Journal of Tissue Engineering and Regenerative Medicine, 2015, 9, E135-E143.	2.7	28
38	Biallelic mutations in valyl-tRNA synthetase gene VARS are associated with a progressive neurodevelopmental epileptic encephalopathy. Nature Communications, 2019, 10, 707.	12.8	28
39	RNA Polymerase III Subunit Mutations in Genetic Diseases. Frontiers in Molecular Biosciences, 2021, 8, 696438.	3.5	28
40	Adult-Onset Vanishing White Matter Disease Due to a Novel EIF2B3 Mutation. Archives of Neurology, 2012, 69, 765-68.	4.5	27
41	More Than Hypomyelination in Pol-III Disorder. Journal of Neuropathology and Experimental Neurology, 2013, 72, 67-75.	1.7	27
42	AlteredPLP1splicing causes hypomyelination of early myelinating structures. Annals of Clinical and Translational Neurology, 2015, 2, 648-661.	3.7	27
43	A recessive ataxia diagnosis algorithm for the next generation sequencing era. Annals of Neurology, 2017, 82, 892-899.	5.3	27
44	Acute Combined Central and Peripheral Nervous System Demyelination in Children. Pediatric Neurology, 2008, 39, 307-316.	2.1	26
45	Genome sequencing in persistently unsolved white matter disorders. Annals of Clinical and Translational Neurology, 2020, 7, 144-152.	3.7	26
46	Endocrine and Growth Abnormalities in 4H Leukodystrophy Caused by Variants in POLR3A, POLR3B, and POLR1C. Journal of Clinical Endocrinology and Metabolism, 2021, 106, e660-e674.	3.6	26
47	Simultaneous Guillain-BarrÃ© Syndrome and Acute Disseminated Encephalomyelitis in the Pediatric Population. Journal of Child Neurology, 2008, 23, 752-757.	1.4	25
48	A treatable new cause of chorea: Betaâ€ketothiolase deficiency. Movement Disorders, 2013, 28, 1054-1056.	3.9	25
49	Postzygotic inactivating mutations of RHOA cause a mosaic neuroectodermal syndrome. Nature Genetics, 2019, 51, 1438-1441.	21.4	25
50	POLR3A variants with striatal involvement and extrapyramidal movement disorder. Neurogenetics, 2020, 21, 121-133.	1.4	24
51	LINGO1 Variants in the French-Canadian Population. PLoS ONE, 2011, 6, e16254.	2.5	23
52	POLR3A and POLR3B Mutations in Unclassified Hypomyelination. Neuropediatrics, 2015, 46, 221-228.	0.6	22
53	Dosage Effect of a Dominant CLCN1 Mutation: A Novel Syndrome. Journal of Child Neurology, 2008, 23, 163-166.	1.4	21
54	Lysophosphatidic acid enhances collagen deposition and matrix thickening in engineered tissue. Journal of Tissue Engineering and Regenerative Medicine, 2015, 9, E65-E75.	2.7	21

#	Article	IF	Citations
55	Study of Cutaneous Reflex Compensation During Locomotion After Nerve Section in the Cat. Journal of Neurophysiology, 2007, 97, 4173-4185.	1.8	20
56	Effects of Serum-Free Culture at the Airâ€“Liquid Interface in a Human Tissue-Engineered Skin Substitute. Tissue Engineering - Part A, 2011, 17, 877-888.	3.1	20
57	Myelination Delay and Allan-Herndon-Dudley Syndrome Caused by a Novel Mutation in the SLC16A2 Gene. Journal of Child Neurology, 2015, 30, 1371-1374.	1.4	20
58	4H Leukodystrophy: A Brain Magnetic Resonance Imaging Scoring System. Neuropediatrics, 2017, 48, 152-160.	0.6	20
59	Expanding the phenotypic and molecular spectrum of RNA polymerase IIIâ€“related leukodystrophy. Neurology: Genetics, 2020, 6, e425.	1.9	20
60	De novo variants in POLR3B cause ataxia, spasticity, and demyelinating neuropathy. American Journal of Human Genetics, 2021, 108, 186-193.	6.2	19
61	Large exonic deletions in POLRB gene cause POLR3-related leukodystrophy. Orphanet Journal of Rare Diseases, 2015, 10, 69.	2.7	18
62	Characterization of a psoriatic skin model produced with involved or uninvolved cells. Journal of Tissue Engineering and Regenerative Medicine, 2015, 9, 789-798.	2.7	18
63	RARS1â€related hypomyelinating leukodystrophy: Expanding the spectrum. Annals of Clinical and Translational Neurology, 2020, 7, 83-93.	3.7	18
64	RNA Polymerases I and III in development and disease. Seminars in Cell and Developmental Biology, 2023, 136, 49-63.	5.0	18
65	POLR3A variants in hereditary spastic paraplegia and ataxia. Brain, 2018, 141, e1-e1.	7.6	17
66	Randomized Clinical Trial of <scp>Firstâ€Line</scp> Genome Sequencing in Pediatric White Matter Disorders. Annals of Neurology, 2020, 88, 264-273.	5.3	17
67	Movement disorders in MCT8 deficiency/Allan-Herndon-Dudley Syndrome. Molecular Genetics and Metabolism, 2022, 135, 109-113.	1.1	17
68	Association of Essential Tremor With Novel Risk Loci. JAMA Neurology, 2022, 79, 185.	9.0	17
69	Refractory and lethal status epilepticus in a patient with ring chromosome 20 syndrome. Epileptic Disorders, 2008, 10, 254-259.	1.3	17
70	The Wobbly Child: An Approach to Inherited Ataxias. Seminars in Pediatric Neurology, 2008, 15, 194-208.	2.0	16
71	Demonstration of the direct impact of ketamine on urothelium using a tissue engineered bladder model. Canadian Urological Association Journal, 2015, 9, 613.	0.6	16
72	A variant of neonatal progeroid syndrome, or Wiedemannâ€“Rautenstrauch syndrome, is associated with a nonsense variant in POLR3GL. European Journal of Human Genetics, 2020, 28, 461-468.	2.8	16

#	Article	IF	Citations
73	Increased Prevalence of Non-motor Symptoms in Essential Tremor. Tremor and Other Hyperkinetic Movements, 2014, 4, 162.	2.0	16
74	Dystonia in RNA Polymerase IIIâ€Related Leukodystrophy. Movement Disorders Clinical Practice, 2019, 6, 155-159.	1.5	14
75	Increased Prevalence of Non-motor Symptoms in Essential Tremor. Tremor and Other Hyperkinetic Movements, 2020, 4, 162.	2.0	14
76	A Case of Secondary Dystonia Responding to Levodopa. Journal of Child Neurology, 2010, 25, 780-781.	1.4	13
77	A homozygous mutation in the NDUFS1 gene presents with a mild cavitating leukoencephalopathy. Neurogenetics, 2014, 15, 161-164.	1.4	12
78	Recessive Mutations in POLR3B Encoding RNA Polymerase III Subunit Causing Diffuse Hypomyelination in Patients with 4H Leukodystrophy with Polymicrogyria and Cataracts. Clinical Neuroradiology, 2017, 27, 213-220.	1.9	12
79	RMND1-Related Leukoencephalopathy With Temporal Lobe Cysts and Hearing Lossâ€"Another Mendelian Mimicker of Congenital Cytomegalovirus Infection. Pediatric Neurology, 2017, 66, 59-62.	2.1	12
80	4H Leukodystrophy: Lessons from 3T Imaging. Neuropediatrics, 2018, 49, 112-117.	0.6	12
81	Therapy Trial Design in Vanishing White Matter. Neurology: Genetics, 2022, 8, e657.	1.9	12
82	Ataxia-Telangiectasia Presenting With a Novel Immunodeficiency. Pediatric Neurology, 2012, 46, 322-324.	2.1	11
83	Endocrine Aspects of 4H Leukodystrophy: A Case Report and Review of the Literature. Case Reports in Endocrinology, 2015, 2015, 1-6.	0.4	10
84	Recessive mutations in <scp>NDUFA2</scp> cause mitochondrial leukoencephalopathy. Clinical Genetics, 2018, 93, 396-400.	2.0	10
85	Patient-Derived Stem Cells, Another in vitro Model, or the Missing Link Toward Novel Therapies for Autism Spectrum Disorders?. Frontiers in Pediatrics, 2019, 7, 225.	1.9	10
86	Abnormal Myelination in Ring Chromosome 18 Syndrome. Journal of Child Neurology, 2012, 27, 1042-1047.	1.4	9
87	Spastic Paraparesis and Marked Improvement of Leukoencephalopathy in Aicardiâ€"GoutiÃ¨res Syndrome. Neuropediatrics, 2014, 45, 406-410.	0.6	9
88	Health-Related Quality of Life for Patients With Genetically Determined Leukoencephalopathy. Pediatric Neurology, 2018, 84, 21-26.	2.1	9
89	Biallelic Loss-of-Function Variants in AIMP1 Cause a Rare Neurodegenerative Disease. Journal of Child Neurology, 2019, 34, 74-80.	1.4	9
90	POLR3-Related Leukodystrophy: Exploring Potential Therapeutic Approaches. Frontiers in Cellular Neuroscience, 2020, 14, 631802.	3.7	9

#	Article	IF	Citations
91	ABHD16A deficiency causes a complicated form of hereditary spastic paraplegia associated with intellectual disability and cerebral anomalies. American Journal of Human Genetics, 2021, 108, 2017-2023.	6.2	9
92	HSD10 mitochondrial disease: p.Leu122Val variant, mild clinical phenotype, and founder effect in Frenchâ€Canadian patients from Quebec. Molecular Genetics & Genomic Medicine, 2019, 7, e1000.	1.2	8
93	Expanded phenotype of AARS1-related white matter disease. Genetics in Medicine, 2021, 23, 2352-2359.	2.4	8
94	Advances in the diagnosis of leukodystrophies. Future Neurology, 2012, 7, 595-612.	0.5	7
95	4H leukodystrophy. Neurology: Genetics, 2020, 6, e409.	1.9	7
96	POLR3-related leukodystrophy: How do mutations affecting RNA polymerase III subunits cause hypomyelination?. Faculty Reviews, 2021, 10, 12.	3.9	7
97	Tremorâ€ataxia with central hypomyelination (TACH): Dystonia as a new clinical feature. Movement Disorders, 2012, 27, 1831-1832.	3.9	6
98	Vanishing White Matter Disease in French-Canadian Patients From Quebec. Pediatric Neurology, 2014, 51, 225-232.	2.1	6
99	Neurogenic bladder and neuroendocrine abnormalities in Pol III-related leukodystrophy. BMC Neurology, 2015, 15, 22.	1.8	6
100	Teaching Neuro Images : Hypomyelinating leukodystrophy with hypodontia due to POLR3B. Neurology, 2013, 81, e145.	1.1	5
101	Novel SIL1 mutations cause cerebellar ataxia and atrophy in a French-Canadian family. Neurogenetics, 2015, 16, 315-318.	1.4	5
102	Novel biallelic variants in NRROS associated with a lethal microgliopathy, brain calcifications, and neurodegeneration. Neurogenetics, 2022, 23, 151-156.	1.4	5
103	TACH Leukodystrophy: Locus Refinement to Chromosome 10q22.3-23.1. Canadian Journal of Neurological Sciences, 2012, 39, 122-123.	0.5	4
104	Strategies to Reconstruct a Functional Urethral Substitute by Self-assembly Method. Procedia Engineering, 2013, 59, 193-200.	1.2	4
105	Pediatric leukodystrophies: The role of the otolaryngologist. International Journal of Pediatric Otorhinolaryngology, 2017, 101, 141-144.	1.0	4
106	Adult Hereditary White Matter Diseases With Psychiatric Presentation: Clinical Pointers and MRI Algorithm to Guide the Diagnostic Process. Journal of Neuropsychiatry and Clinical Neurosciences, 2021, 33, 180-193.	1.8	4
107	The LORIS MyeliNeuroGene rare disease database for natural history studies and clinical trial readiness. Orphanet Journal of Rare Diseases, 2021, 16, 328.	2.7	4
108	Distinguishing severe phenotypes associated with pathogenic variants in POLR3A. American Journal of Medical Genetics, Part A, 2022, 188, 708-712.	1.2	4

#	Article	IF	Citations
109	Neurotransmitter diseases and related conditions. Molecular Genetics and Metabolism, 2007, 92, 189-197.	1.1	3
110	A Novel PLP1 Mutation Further Expands the Clinical Heterogeneity at the Locus. Canadian Journal of Neurological Sciences, 2012, 39, 220-224.	0.5	3
111	Variants in LSM7 impair LSM complexes assembly, neurodevelopment in zebrafish and may be associated with an ultra-rare neurological disease. Human Genetics and Genomics Advances, 2021, 2, 100034.	1.7	3
112	Endocannabinoid dysfunction in neurological disease: neuro-ocular DAGLA-related syndrome. Brain, 2022, 145, 3383-3390.	7.6	3
113	Chylomicron retention disease: Dystonia as a new clinical feature. Movement Disorders, 2010, 25, 1755-1756.	3.9	2
114	Mutations in RNF216 do not cause 4H syndrome. Parkinsonism and Related Disorders, 2015, 21, 1387-1388.	2.2	2
115	The Movement Disorder of Brain-Lung-Thyroid Syndrome Can be Responsive to Methylphenidate. Tremor and Other Hyperkinetic Movements, 2017, 7, 508.	2.0	2
116	Experience of Parents of Children with Genetically Determined Leukoencephalopathies Regarding the Adapted Health Care Services During the COVID-19 Pandemic. Journal of Child Neurology, 2022, 37, 237-245.	1.4	2
117	Oculodentodigital Dysplasia. Neurology, 2022, 98, 675-677.	1.1	2
118	Mutations of POLR3A Encoding a Catalytic Subunit of RNA Polymerase Pol III Cause a Recessive Hypomyelinating Leukodystrophy p415. American Journal of Human Genetics, 2012, 91, 972.	6.2	1
119	Expert opinion and caution are imperative for interpretation of next generation sequencing data. European Journal of Medical Genetics, 2016, 59, 519-521.	1.3	1
120	A Novel De Novo Variant in DYNC1H1 Causes Spinal Muscular Atrophy Lower Extremity Predominant in Identical Twins: A Case Report. Child Neurology Open, 2021, 8, 2329048X2110274.	1.1	1
121	Classifying Hypomyelination: A Critical (White) Matter. Child Neurology Open, 2020, 7, 2329048X2098376.	1.1	1
122	A unique pediatric case of radiation-induced parkinsonism. Journal of Pediatric Neurology, 2015, 09, 123-126.	0.2	0
123	Response to Correspondence on â€œStress in Parents of Children With Genetically Determined Leukoencephalopathies: A Pilot Studyâ€• Journal of Child Neurology, 2021, 36, 245-246.	1.4	0
124	Clinical, neuroradiological, and molecular characterization of patients with atypical Zellweger spectrum disorder caused by PEX16 mutations: a case series. Neurogenetics, 2022, 23, 115-127.	1.4	0