Elsa Rossignol

List of Publications by Year
in descending order

41 papers	2,337 citations	331670 21 h-index	289244 40 g-index
42 all docs	42 docs citations	42 times ranked	5035 citing authors

#	Article	IF	Citations
1	Infantile onset carnitine palmitoyltransferase 2 deficiency: Cortical polymicrogyria, schizencephaly, and gray matter heterotopias in an adolescent with normal development. JIMD Reports, 2022, 63, 3-10.	1.5	1
2	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
3	FBXO28 causes developmental and epileptic encephalopathy with profound intellectual disability. Epilepsia, 2021, 62, e13-e21.	5.1	8
4	Reversing frontal disinhibition rescues behavioural deficits in models of CACNA1A-associated neurodevelopment disorders. Molecular Psychiatry, 2021, 26, 7225-7246.	7.9	16
5	Stress in Parents of Children With Genetically Determined Leukoencephalopathies: A Pilot Study. Journal of Child Neurology, 2020, 35, 901-907.	1.4	7
6	Mutations in PIGB Cause an Inherited GPI Biosynthesis Defect with an Axonal Neuropathy and Metabolic Abnormality in Severe Cases. American Journal of Human Genetics, 2019, 105, 384-394.	6.2	37
7	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
8	Mutations in ACTL6B Cause Neurodevelopmental Deficits and Epilepsy and Lead to Loss of Dendrites in Human Neurons. American Journal of Human Genetics, 2019, 104, 815-834.	6.2	59
9	IQSEC2-related encephalopathy in males and females: a comparative study including 37 novel patients. Genetics in Medicine, 2019, 21, 837-849.	2.4	47
10	The epilepsy-associated protein TBC1D24 is required for normal development, survival and vesicle trafficking in mammalian neurons. Human Molecular Genetics, 2019, 28, 584-597.	2.9	35
11	Recessive mutations in VPS13D cause childhood onset movement disorders. Annals of Neurology, 2018, 83, 1089-1095.	5.3	104
12	Health-Related Quality of Life for Patients With Genetically Determined Leukoencephalopathy. Pediatric Neurology, 2018, 84, 21-26.	2.1	9
13	FRMPD4 mutations cause X-linked intellectual disability and disrupt dendritic spine morphogenesis. Human Molecular Genetics, 2018, 27, 589-600.	2.9	20
14	PHACTRing in actin: actin deregulation in genetic epilepsies. Brain, 2018, 141, 3084-3088.	7.6	0
15	Ex Utero Electroporation and Organotypic Slice Cultures of Embryonic Mouse Brains for Live-Imaging of Migrating GABAergic Interneurons. Journal of Visualized Experiments, 2018, , .	0.3	1
16	Remodeled cortical inhibition prevents motor seizures in generalized epilepsy. Annals of Neurology, 2018, 84, 436-451.	5.3	19
17	Optic nerve hypoplasia in a patient with a de novo KIF1A heterozygous mutation. Canadian Journal of Ophthalmology, 2017, 52, e169-e171.	0.7	14
18	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2017, 101, 664-685.	6.2	337

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19	Involvement of cortical fast-spiking parvalbumin-positive basket cells in epilepsy. Progress in Brain Research, 2016, 226, 81-126.	1.4	74
20	LPIN1 deficiency with severe recurrent rhabdomyolysis and persistent elevation of creatine kinase levels due to chromosome 2 maternal isodisomy. Molecular Genetics and Metabolism Reports, 2015, 5, 85-88.	1.1	20
21	Efficacy and safety of lacosamide as an adjunctive therapy for refractory focal epilepsy in paediatric patients: a retrospective single-centre study. Epileptic Disorders, 2015, 17, 436-443.	1.3	20
22	De Novo Mutations in the Motor Domain of KIF1A Cause Cognitive Impairment, Spastic Paraparesis, Axonal Neuropathy, and Cerebellar Atrophy. Human Mutation, 2015, 36, 69-78.	2.5	114
23	CACNA1A haploinsufficiency causes cognitive impairment, autism and epileptic encephalopathy with mild cerebellar symptoms. European Journal of Human Genetics, 2015, 23, 1505-1512.	2.8	165
24	Normal Cerebrospinal Fluid Pyridoxal 5â€²-Phosphate Level in a PNPO-Deficient Patient with Neonatal-Onset Epileptic Encephalopathy. JIMD Reports, 2015, 22, 67-75.	1.5	21
25	A Gain-of-Function Mutation inNALCNin a Child with Intellectual Disability, Ataxia, and Arthrogryposis. Human Mutation, 2015, 36, 753-757.	2.5	46
26	Bilateral congenital corneal anesthesia in a patient with SCN9A mutation, confirmed primary erythromelalgia, and paroxysmal extreme pain disorder. Journal of AAPOS, 2015, 19, 478-479.	0.3	13
27	An atypical case of SCN9A mutation presenting with global motor delay and a severe pain disorder. Muscle and Nerve, 2014, 49, 134-138.	2.2	20
28	<scp>WONOEP</scp> appraisal: New genetic approaches to study epilepsy. Epilepsia, 2014, 55, 1170-1186.	5.1	13
29	The genetic landscape of infantile spasms. Human Molecular Genetics, 2014, 23, 4846-4858.	2.9	156
30	Vanishing White Matter Disease in French-Canadian Patients From Quebec. Pediatric Neurology, 2014, 51, 225-232.	2.1	6
31	Mutations in DOCK7 in Individuals with Epileptic Encephalopathy and Cortical Blindness. American Journal of Human Genetics, 2014, 94, 891-897.	6.2	44
32	Ca_V2.1 ablation in cortical interneurons selectively impairs fastâ€spiking basket cells and causes generalized seizures. Annals of Neurology, 2013, 74, 209-222.	5.3	95
33	Chronic inflammatory demyelinating polyneuropathy. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2013, 112, 1163-1169.	1.8	8
34	Satb1 Is an Activity-Modulated Transcription Factor Required for the Terminal Differentiation and Connectivity of Medial Ganglionic Eminence-Derived Cortical Interneurons. Journal of Neuroscience, 2012, 32, 17690-17705.	3.6	122
35	Opposing regulation of dopaminergic activity and exploratory motor behavior by forebrain and brainstem cholinergic circuits. Nature Communications, 2012, 3, 1172.	12.8	69
36	Genetics and Function of Neocortical GABAergic Interneurons in Neurodevelopmental Disorders. Neural Plasticity, 2011, 2011, 1-25.	2.2	181

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37	The Cell-Intrinsic Requirement of Sox6 for Cortical Interneuron Development. Neuron, 2009, 63, 466-481.	8.1	194
38	Clinical, electrophysiologic, and genetic study of non-dystrophic myotonia in French-Canadians. Neuromuscular Disorders, 2009, 19, 330-334.	0.6	44
39	Vagus nerve stimulation in pediatric epileptic syndromes. Seizure: the Journal of the British Epilepsy Association, 2009, 18, 34-37.	2.0	73
40	Evolution and Treatment of Childhood Chronic Inflammatory Polyneuropathy. Pediatric Neurology, 2007, 36, 88-94.	2.1	40
41	Pharmacogenetic testing in pediatric neurology: a pragmatic study evaluating clinician and patient perceptions. Personalized Medicine, 0, , .	1.5	0