Roberta La Piana

List of Publications by Year
in descending order

55 papers	2,014 citations	304743 22 h-index	254184 43 g-index
55 all docs	55 docs citations	55 times ranked	3348 citing authors

#	Article	IF	Citations
1	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
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	POLR3-related leukodystrophy: How do mutations affecting RNA polymerase III subunits cause hypomyelination?. Faculty Reviews, 2021, 10, 12.	3.9	7
4	The epileptology of Aicardi-GoutiÃ¨res syndrome: electro-clinical-radiological findings. Seizure: the Journal of the British Epilepsy Association, 2021, 86, 197-209.	2.0	2
5	COVID-19 and disease-modifying therapies in patients with demyelinating diseases of the central nervous system: A systematic review. Multiple Sclerosis and Related Disorders, 2021, 50, 102800.	2.0	58
6	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
7	Neurodevelopmental outcome of preterm very low birth weight infants admitted to an Italian tertiary center over an 11-year period. Scientific Reports, 2021, 11, 16316.	3.3	11
8	Neurological Involvement in Glycogen Storage Disease Type IXa due to PHKA2 Mutation. Canadian Journal of Neurological Sciences, 2020, 47, 400-403.	0.5	7
9	Brain MRI features and scoring of leukodystrophy in adult-onset Krabbe disease. Neurology, 2019, 93, e647-e652.	1.1	25
10	Neural function in DCC mutation carriers with and without mirror movements. Annals of Neurology, 2019, 85, 433-442.	5.3	12
11	Brain functional organization and structure in patients with arteriovenous malformations. Neuroradiology, 2019, 61, 1047-1054.	2.2	9
12	Molecular Genetics and Interferon Signature in the Italian Aicardi GoutiÃ¨res Syndrome Cohort: Report of 12 New Cases and Literature Review. Journal of Clinical Medicine, 2019, 8, 750.	2.4	29
13	Long-Standing Psychiatric Features as the Only Clinical Presentation of Vanishing White Matter Disease. Journal of Neuropsychiatry and Clinical Neurosciences, 2019, 31, 276-279.	1.8	5
14	3T MRI study discloses high intrafamilial variability in CADASIL due to a novel NOTCH3 mutation. Journal of Clinical Neuroscience, 2018, 58, 25-29.	1.5	2
15	Identification and functional characterization of a novel MTFMT mutation associated with selective vulnerability of the visual pathway and a mild neurological phenotype. Neurogenetics, 2017, 18, 97-103.	1.4	11
16	4H Leukodystrophy: A Brain Magnetic Resonance Imaging Scoring System. Neuropediatrics, 2017, 48, 152-160.	0.6	20
17	Assessment of clot length with multiphase CT angiography in patients with acute ischemic stroke. Neuroradiology Journal, 2017, 30, 593-599.	1.2	13
18	Diffuse hypomyelination is not obligate for POLR3-related disorders. Neurology, 2016, 86, 1622-1626.	1.1	65

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19	Mutations in GALC cause late-onset Krabbe disease with predominant cerebellar ataxia. Neurogenetics, 2016, 17, 137-141.	1.4	16
20	SPG7 mutations explain a significant proportion of French Canadian spastic ataxia cases. European Journal of Human Genetics, 2016, 24, 1016-1021.	2.8	46
21	Autosomal recessive cerebellar ataxia caused by a homozygous mutation inPMPCA. Brain, 2016, 139, e19-e19.	7.6	27
22	Neuroradiologic patterns and novel imaging findings in Aicardi-GoutiÃ¨res syndrome. Neurology, 2016, 86, 28-35.	1.1	59
23	Paediatric arterial ischaemic stroke and cerebral sinovenous thrombosis. Thrombosis and Haemostasis, 2015, 113, 1270-1277.	3.4	28
24	Early-Onset Aicardi-GoutiÃ¨res Syndrome. Journal of Child Neurology, 2015, 30, 1343-1348.	1.4	33
25	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
26	A novel frameshift mutation in FGF14 causes an autosomal dominant episodic ataxia. Neurogenetics, 2015, 16, 233-236.	1.4	34
27	POLR3A and POLR3B Mutations in Unclassified Hypomyelination. Neuropediatrics, 2015, 46, 221-228.	0.6	22
28	Novel SIL1 mutations cause cerebellar ataxia and atrophy in a French-Canadian family. Neurogenetics, 2015, 16, 315-318.	1.4	5
29	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
30	Spastic Paraparesis and Marked Improvement of Leukoencephalopathy in Aicardiâ€“GoutiÃ¨res Syndrome. Neuropediatrics, 2014, 45, 406-410.	0.6	9
31	Brain Magnetic Resonance Imaging (MRI) Pattern Recognition in Pol III-Related Leukodystrophies. Journal of Child Neurology, 2014, 29, 214-220.	1.4	47
32	A novel mutation in the CSF1R gene causes a variable leukoencephalopathy with spheroids. Neurogenetics, 2014, 15, 289-294.	1.4	20
33	Clinical spectrum of 4H leukodystrophy caused by POLR3A and POLR3B mutations. Neurology, 2014, 83, 1898-1905.	1.1	170
34	Bilateral striatal necrosis in two subjects with Aicardiâ€“GoutiÃ¨res syndrome due to mutations in ADAR1 (AGS6). American Journal of Medical Genetics, Part A, 2014, 164, 815-819.	1.2	30
35	Brain Reorganization after Endovascular Treatment in a Patient with a Large Arteriovenous Malformation: The Role of Diagnostic and Functional Neuroimaging Techniques. Interventional Neuroradiology, 2013, 19, 329-338.	1.1	6
36	Adult-Onset Vanishing White Matter Disease Due to a Novel EIF2B3 Mutation. Archives of Neurology, 2012, 69, 765-68.	4.5	27

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37	COL4A1-Related Disease: Raised Creatine Kinase and Cerebral Calcification as Useful Pointers. Neuropediatrics, 2012, 43, 283-288.	0.6	20
38	Intraventricular Localization of an Anaplastic Oligodedendroglioma: A Rare Event. Canadian Journal of Neurological Sciences, 2012, 39, 649-651.	0.5	3
39	Advances in the diagnosis of leukodystrophies. Future Neurology, 2012, 7, 595-612.	0.5	7
40	Neuroâ€ophthalmological disorders in cerebral palsy: ophthalmological, oculomotor, and visual aspects. Developmental Medicine and Child Neurology, 2012, 54, 730-736.	2.1	137
41	Neurodevelopmental outcome of preterm very low birth weight infants born from 2005 to 2007. European Journal of Paediatric Neurology, 2012, 16, 716-723.	1.6	28
42	Different Mutations in Three Prime Repair Exonuclease 1 and Ribonuclease H2 Genes Affect Clinical Features in Aicardi-GoutiÃ¨res Syndrome. Journal of Child Neurology, 2012, 27, 51-60.	1.4	4
43	Calcifying leukoencephalopathies: New overlapping phenotypes. American Journal of Medical Genetics, Part A, 2012, 158A, 964-965.	1.2	0
44	The Aicardiâ€“GoutiÃ¨res syndrome. Molecular and clinical features of RNAse deficiency and microRNA overload. Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis, 2011, 717, 99-108.	1.0	30
45	COL4A1 Mutations Associated with a Characteristic Pattern of Intracranial Calcification. Neuropediatrics, 2011, 42, 227-233.	0.6	38
46	Response to Correspondence on â€œSpinal Cord Calcification in an Early-Onset Progressive Leukoencephalopathyâ€. Journal of Child Neurology, 2011, 26, 1058-1058.	1.4	0
47	Spinal Cord Calcification in an Early-Onset Progressive Leukoencephalopathy. Journal of Child Neurology, 2011, 26, 876-880.	1.4	9
48	Reply:. American Journal of Neuroradiology, 2010, 31, E64-E64.	2.4	0
49	Aicardi-GoutiÃ¨res Syndrome: Neuroradiologic Findings and Follow-Up. American Journal of Neuroradiology, 2009, 30, 1971-1976.	2.4	72
50	Interferonâ€Related Transcriptome Alterations in the Cerebrospinal Fluid Cells of Aicardiâ€GoutiÃ¨res Patients^{â€}. Brain Pathology, 2009, 19, 650-660.	4.1	26
51	Recovery of visual functions after early acquired occipital damage. Developmental Medicine and Child Neurology, 2008, 50, 311-315.	2.1	20
52	Aicardiâ€“GoutiÃ¨res syndrome presenting atypically as a sub-acute leukoencephalopathy. European Journal of Paediatric Neurology, 2008, 12, 408-411.	1.6	27
53	Aicardi-Goutieres syndrome. British Medical Bulletin, 2008, 89, 183-201.	6.9	75
54	BRAIN DAMAGE AS DETECTED BY cDNA-MICROARRAY IN THE SPINAL FLUID OF PATIENTS WITH AICARDI-GOUTIÈ€RES SYNDROME. Neurology, 2008, 71, 610-612.	1.1	10

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55	Spectrum of Visual Disorders in Children With Cerebral Visual Impairment. Journal of Child Neurology, 2007, 22, 294-301.	1.4	156