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	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
2	Clinical spectrum of 4H leukodystrophy caused by POLR3A and POLR3B mutations. Neurology, 2014, 83, 1898-1905.	1.1	170
3	Spectrum of Visual Disorders in Children With Cerebral Visual Impairment. Journal of Child Neurology, 2007, 22, 294-301.	1.4	156
4	Neuroâ€ophthalmological disorders in cerebral palsy: ophthalmological, oculomotor, and visual aspects. Developmental Medicine and Child Neurology, 2012, 54, 730-736.	2.1	137
5	Aicardi-Goutieres syndrome. British Medical Bulletin, 2008, 89, 183-201.	6.9	75
6	Aicardi-GoutiÃ¨res Syndrome: Neuroradiologic Findings and Follow-Up. American Journal of Neuroradiology, 2009, 30, 1971-1976.	2.4	72
7	Diffuse hypomyelination is not obligate for POLR3-related disorders. Neurology, 2016, 86, 1622-1626.	1.1	65
8	Neuroradiologic patterns and novel imaging findings in Aicardi-GoutiÃ¨res syndrome. Neurology, 2016, 86, 28-35.	1.1	59
9	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
10	Brain Magnetic Resonance Imaging (MRI) Pattern Recognition in Pol III-Related Leukodystrophies. Journal of Child Neurology, 2014, 29, 214-220.	1.4	47
11	SPG7 mutations explain a significant proportion of French Canadian spastic ataxia cases. European Journal of Human Genetics, 2016, 24, 1016-1021.	2.8	46
12	COL4A1 Mutations Associated with a Characteristic Pattern of Intracranial Calcification. Neuropediatrics, 2011, 42, 227-233.	0.6	38
13	A novel frameshift mutation in FGF14 causes an autosomal dominant episodic ataxia. Neurogenetics, 2015, 16, 233-236.	1.4	34
14	Early-Onset Aicardi-GoutiÃ¨res Syndrome. Journal of Child Neurology, 2015, 30, 1343-1348.	1.4	33
15	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
16	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
17	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
18	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

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19	Paediatric arterial ischaemic stroke and cerebral sinovenous thrombosis. Thrombosis and Haemostasis, 2015, 113, 1270-1277.	3.4	28
20	Aicardiâ€“GoutiÃ¨res syndrome presenting atypically as a sub-acute leukoencephalopathy. European Journal of Paediatric Neurology, 2008, 12, 408-411.	1.6	27
21	Adult-Onset Vanishing White Matter Disease Due to a Novel EIF2B3 Mutation. Archives of Neurology, 2012, 69, 765-68.	4.5	27
22	Autosomal recessive cerebellar ataxia caused by a homozygous mutation inPMPCA. Brain, 2016, 139, e19-e19.	7.6	27
23	Interferonâ€Related Transcriptome Alterations in the Cerebrospinal Fluid Cells of Aicardiâ€GoutiÃ¨res Patients^{â€}. Brain Pathology, 2009, 19, 650-660.	4.1	26
24	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
25	Brain MRI features and scoring of leukodystrophy in adult-onset Krabbe disease. Neurology, 2019, 93, e647-e652.	1.1	25
26	POLR3A and POLR3B Mutations in Unclassified Hypomyelination. Neuropediatrics, 2015, 46, 221-228.	0.6	22
27	Recovery of visual functions after early acquired occipital damage. Developmental Medicine and Child Neurology, 2008, 50, 311-315.	2.1	20
28	COL4A1-Related Disease: Raised Creatine Kinase and Cerebral Calcification as Useful Pointers. Neuropediatrics, 2012, 43, 283-288.	0.6	20
29	A novel mutation in the CSF1R gene causes a variable leukoencephalopathy with spheroids. Neurogenetics, 2014, 15, 289-294.	1.4	20
30	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
31	4H Leukodystrophy: A Brain Magnetic Resonance Imaging Scoring System. Neuropediatrics, 2017, 48, 152-160.	0.6	20
32	Mutations in GALC cause late-onset Krabbe disease with predominant cerebellar ataxia. Neurogenetics, 2016, 17, 137-141.	1.4	16
33	Assessment of clot length with multiphase CT angiography in patients with acute ischemic stroke. Neuroradiology Journal, 2017, 30, 593-599.	1.2	13
34	Neural function in DCC mutation carriers with and without mirror movements. Annals of Neurology, 2019, 85, 433-442.	5.3	12
35	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
36	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

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37	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
38	Spinal Cord Calcification in an Early-Onset Progressive Leukoencephalopathy. Journal of Child Neurology, 2011, 26, 876-880.	1.4	9
39	Spastic Paraparesis and Marked Improvement of Leukoencephalopathy in Aicardiâ€“GoutiÃ¨res Syndrome. Neuropediatrics, 2014, 45, 406-410.	0.6	9
40	Brain functional organization and structure in patients with arteriovenous malformations. Neuroradiology, 2019, 61, 1047-1054.	2.2	9
41	Advances in the diagnosis of leukodystrophies. Future Neurology, 2012, 7, 595-612.	0.5	7
42	Neurological Involvement in Glycogen Storage Disease Type IXa due to PHKA2 Mutation. Canadian Journal of Neurological Sciences, 2020, 47, 400-403.	0.5	7
43	POLR3-related leukodystrophy: How do mutations affecting RNA polymerase III subunits cause hypomyelination?. Faculty Reviews, 2021, 10, 12.	3.9	7
44	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
45	Novel SIL1 mutations cause cerebellar ataxia and atrophy in a French-Canadian family. Neurogenetics, 2015, 16, 315-318.	1.4	5
46	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
47	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
48	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
49	Intraventricular Localization of an Anaplastic Oligodedendroglioma: A Rare Event. Canadian Journal of Neurological Sciences, 2012, 39, 649-651.	0.5	3
50	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
51	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
52	Reply:. American Journal of Neuroradiology, 2010, 31, E64-E64.	2.4	0
53	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
54	Calcifying leukoencephalopathies: New overlapping phenotypes. American Journal of Medical Genetics, Part A, 2012, 158A, 964-965.	1.2	0

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55	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