Laura A Adang

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/6441079/publications.pdf

Version: 2024-02-01

36	725	566801	610482
papers	citations	15 h-index	24 g-index

38	38	38	925
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Janus Kinase Inhibition in the Aicardiâ€“GoutiÃ¨res Syndrome. New England Journal of Medicine, 2020, 383, 986-989.	13.9	109
2	Genetic and phenotypic spectrum associated with IFIH1 gainâ€ofâ€function. Human Mutation, 2020, 41, 837-849.	1.1	63
3	Revised consensus statement on the preventive and symptomatic care of patients with leukodystrophies. Molecular Genetics and Metabolism, 2017, 122, 18-32.	0.5	42
4	Developmental Outcomes of Aicardi GoutiÃ¨res Syndrome. Journal of Child Neurology, 2020, 35, 7-16.	0.7	40
5	Progressive Multifocal Leukoencephalopathy. F1000Research, 2015, 4, 1424.	0.8	39
6	<scp>MRI</scp> surveillance of boys with Xâ€linked adrenoleukodystrophy identified by newborn screening: Metaâ€analysis and consensus guidelines. Journal of Inherited Metabolic Disease, 2021, 44, 728-739.	1.7	39
7	Aicardi goutiÃ¨res syndrome is associated with pulmonary hypertension. Molecular Genetics and Metabolism, 2018, 125, 351-358.	0.5	35
8	Multiple Sulfatase Deficiency: A Disease Comprising Mucopolysaccharidosis, Sphingolipidosis, and More Caused by a Defect in Posttranslational Modification. International Journal of Molecular Sciences, 2020, 21, 3448.	1.8	32
9	Complex care of individuals with multiple sulfatase deficiency: Clinical cases and consensus statement. Molecular Genetics and Metabolism, 2018, 123, 337-346.	0.5	31
10	The 2021 European Alliance of Associations for Rheumatology/American College of Rheumatology points to consider for diagnosis and management of autoinflammatory type I interferonopathies: CANDLE/PRAAS, SAVI and AGS. Annals of the Rheumatic Diseases, 2022, 81, 601-613.	0.5	31
11	Development of a neurologic severity scale for Aicardi GoutiÃ¨res Syndrome. Molecular Genetics and Metabolism, 2020, 130, 153-160.	0.5	25
12	Natural history of multiple sulfatase deficiency: Retrospective phenotyping and functional variant analysis to characterize an ultraâ€rare disease. Journal of Inherited Metabolic Disease, 2020, 43, 1298-1309.	1.7	23
13	The 2021 European Alliance of Associations for Rheumatology/American College of Rheumatology Points to Consider for Diagnosis and Management of Autoinflammatory Type I Interferonopathies: <scp>CANDLE</scp>/<scp>PRAAS</scp>, <scp>SAVI</scp>, and <scp>AGS</scp>. Arthritis and Rheumatology, 2022, 74, 735-751.	2.9	23
14	Late-Onset Aicardi-GoutiÃ¨res Syndrome: A Characterization of Presenting Clinical Features. Pediatric Neurology, 2021, 115, 1-6.	1.0	18
15	Leukocyte and Dried Blood Spot Arylsulfatase A Assay by Tandem Mass Spectrometry. Analytical Chemistry, 2020, 92, 6341-6348.	3.2	17
16	Consensus clinical management guideline for betaâ€propeller proteinâ€associated neurodegeneration. Developmental Medicine and Child Neurology, 2021, 63, 1402-1409.	1.1	17
17	Glial cells in the driver seat of leukodystrophy pathogenesis. Neurobiology of Disease, 2020, 146, 105087.	2.1	16
18	Phenotypic and Imaging Spectrum Associated With WDR45. Pediatric Neurology, 2020, 109, 56-62.	1.0	16

#	Article	IF	Citations
19	Understanding the phenotypic spectrum of ASXL â€related disease: Ten cases and a review of the literature. American Journal of Medical Genetics, Part A, 2021, 185, 1700-1711.	0.7	16
20	Mutations in SZT2 result in earlyâ€onset epileptic encephalopathy and leukoencephalopathy. American Journal of Medical Genetics, Part A, 2018, 176, 1443-1448.	0.7	15
21	A systematic review and metaâ€analysis of published cases reveals the natural disease history in multiple sulfatase deficiency. Journal of Inherited Metabolic Disease, 2020, 43, 1288-1297.	1.7	14
22	Newborn Screening for X-Linked Adrenoleukodystrophy: Review of Data and Outcomes in Pennsylvania. International Journal of Neonatal Screening, 2022, 8, 24.	1.2	11
23	Modified Delphi procedure-based expert consensus on endpoints for an international disease registry for Metachromatic Leukodystrophy: The European Metachromatic Leukodystrophy initiative (MLDi). Orphanet Journal of Rare Diseases, 2022, 17, 48.	1.2	10
24	Considerations for the Safe Operation of Schools During the Coronavirus Pandemic. Frontiers in Public Health, 2021, 9, 751451.	1.3	9
25	Hematologic abnormalities in Aicardi GoutiÃ¨res Syndrome. Molecular Genetics and Metabolism, 2022, 136, 324-329.	0.5	8
26	Neuropathological Findings in a Case of IFIH1-Related Aicardiâ€“GoutiÃ¨res Syndrome. Pediatric and Developmental Pathology, 2019, 22, 566-570.	0.5	7
27	Hepatic Involvement in Aicardi-GoutiÃ¨res Syndrome. Neuropediatrics, 2021, 52, 441-447.	0.3	6
28	Reliability of the Telemedicine Application of the Gross Motor Function Measure-88 in Patients With Leukodystrophy. Pediatric Neurology, 2021, 125, 34-39.	1.0	6
29	Validation of a symptom-based questionnaire for pediatric CNS demyelinating diseases. Journal of AAPOS, 2019, 23, 157.e1-157.e7.	0.2	3
30	Early developmental delay in Leigh syndrome spectrum disorders is associated with poor clinical prognosis. Molecular Genetics and Metabolism, 2022, 135, 342-349.	0.5	2
31	Teaching NeuroImages: Atrophy in epileptic encephalopathy. Neurology, 2018, 90, e442-e443.	1.5	1
32	Pediatric Multiple Sclerosis. Current Pediatrics Reports, 2017, 5, 6-12.	1.7	0
33	Teaching NeuroImages: Cranial neuropathies following clival infarction. Neurology, 2019, 93, e1031.	1.5	0
34	Widening the MRI Findings of PLA2G6-Associated Neurodegeneration. Neuropediatrics, 2021, 52, 509-510.	0.3	0
35	Improved Gene Therapy for Metachromatic Leukodystrophy. Blood, 2021, 138, 3979-3979.	0.6	0
36	Clinical, neuroradiological, and molecular characterization of patients with atypical Zellweger spectrum disorder caused by PEX16 mutations: a case series. Neurogenetics, 2022, 23, 115-127.	0.7	0