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	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
2	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
3	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
4	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
5	Newborn Screening for X-Linked Adrenoleukodystrophy: Review of Data and Outcomes in Pennsylvania. International Journal of Neonatal Screening, 2022, 8, 24.	1.2	11
6	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
7	Hematologic abnormalities in Aicardi GoutiÃ¨res Syndrome. Molecular Genetics and Metabolism, 2022, 136, 324-329.	0.5	8
8	Late-Onset Aicardi-GoutiÃ¨res Syndrome: A Characterization of Presenting Clinical Features. Pediatric Neurology, 2021, 115, 1-6.	1.0	18
9	Hepatic Involvement in Aicardi-GoutiÃ¨res Syndrome. Neuropediatrics, 2021, 52, 441-447.	0.3	6
10	<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
11	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
12	Widening the MRI Findings of PLA2G6-Associated Neurodegeneration. Neuropediatrics, 2021, 52, 509-510.	0.3	0
13	Consensus clinical management guideline for betaâ€propeller proteinâ€associated neurodegeneration. Developmental Medicine and Child Neurology, 2021, 63, 1402-1409.	1.1	17
14	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
15	Improved Gene Therapy for Metachromatic Leukodystrophy. Blood, 2021, 138, 3979-3979.	0.6	0
16	Considerations for the Safe Operation of Schools During the Coronavirus Pandemic. Frontiers in Public Health, 2021, 9, 751451.	1.3	9
17	Developmental Outcomes of Aicardi GoutiÃ¨res Syndrome. Journal of Child Neurology, 2020, 35, 7-16.	0.7	40
18	Genetic and phenotypic spectrum associated with IFIH1 gainâ€ofâ€function. Human Mutation, 2020, 41, 837-849.	1.1	63

#	Article	IF	Citations
19	Glial cells in the driver seat of leukodystrophy pathogenesis. Neurobiology of Disease, 2020, 146, 105087.	2.1	16
20	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
21	Janus Kinase Inhibition in the Aicardiâ€“GoutiÃ¨res Syndrome. New England Journal of Medicine, 2020, 383, 986-989.	13.9	109
22	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
23	Phenotypic and Imaging Spectrum Associated With WDR45. Pediatric Neurology, 2020, 109, 56-62.	1.0	16
24	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
25	Leukocyte and Dried Blood Spot Arylsulfatase A Assay by Tandem Mass Spectrometry. Analytical Chemistry, 2020, 92, 6341-6348.	3.2	17
26	Development of a neurologic severity scale for Aicardi GoutiÃ¨res Syndrome. Molecular Genetics and Metabolism, 2020, 130, 153-160.	0.5	25
27	Validation of a symptom-based questionnaire for pediatric CNS demyelinating diseases. Journal of AAPOS, 2019, 23, 157.e1-157.e7.	0.2	3
28	Neuropathological Findings in a Case of IFIH1-Related Aicardiâ€“GoutiÃ¨res Syndrome. Pediatric and Developmental Pathology, 2019, 22, 566-570.	0.5	7
29	Teaching NeuroImages: Cranial neuropathies following clival infarction. Neurology, 2019, 93, e1031.	1.5	0
30	Complex care of individuals with multiple sulfatase deficiency: Clinical cases and consensus statement. Molecular Genetics and Metabolism, 2018, 123, 337-346.	0.5	31
31	Teaching NeuroImages: Atrophy in epileptic encephalopathy. Neurology, 2018, 90, e442-e443.	1.5	1
32	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
33	Aicardi goutiÃ¨res syndrome is associated with pulmonary hypertension. Molecular Genetics and Metabolism, 2018, 125, 351-358.	0.5	35
34	Pediatric Multiple Sclerosis. Current Pediatrics Reports, 2017, 5, 6-12.	1.7	0
35	Revised consensus statement on the preventive and symptomatic care of patients with leukodystrophies. Molecular Genetics and Metabolism, 2017, 122, 18-32.	0.5	42
36	Progressive Multifocal Leukoencephalopathy. F1000Research, 2015, 4, 1424.	0.8	39