

Rebecca C Ahrens-Nicklas

List of Publications by Year in descending order

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36
papers

543
citations

686830

13
h-index

713013

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37
all docs

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docs citations

37
times ranked

1036
citing authors

#	ARTICLE	IF	CITATIONS
1	Fine-Tuning 3-Methylglutaconic Aciduria Cutoffs for a Patient with Infantile-Onset Barth Syndrome. <i>Clinical Chemistry</i> , 2022, 68, 365-367.	1.5	0
2	Consolidation of the clinical and genetic definition of a <i>SOX4</i> -related neurodevelopmental syndrome. <i>Journal of Medical Genetics</i> , 2022, 59, 1058-1068.	1.5	10
3	Newborn Screening for X-Linked Adrenoleukodystrophy: Review of Data and Outcomes in Pennsylvania. <i>International Journal of Neonatal Screening</i> , 2022, 8, 24.	1.2	11
4	Heal thyself: The promise of autologous hematopoietic stem cell gene therapy in neurometabolic disorders. <i>Molecular Therapy</i> , 2022, 30, 1353-1354.	3.7	1
5	Neuronal genetic rescue normalizes brain network dynamics in a lysosomal storage disorder despite persistent storage accumulation. <i>Molecular Therapy</i> , 2022, 30, 2464-2473.	3.7	4
6	<i>MYH7</i> variants cause complex congenital heart disease. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 2772-2776.	0.7	7
7	Contribution of Mendelian disorders in a population-based pediatric neurodegeneration cohort. <i>Journal of Pediatrics</i> , 2022, , .	0.9	0
8	Genotype-phenotype association by echocardiography offers incremental value in patients with Noonan Syndrome with Multiple Lentiginosities. <i>Pediatric Research</i> , 2021, 90, 444-451.	1.1	6
9	Genetic variant burden and adverse outcomes in pediatric cardiomyopathy. <i>Pediatric Research</i> , 2021, 89, 1470-1476.	1.1	9
10	Variants in <i>NAA15</i> cause pediatric hypertrophic cardiomyopathy. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 228-233.	0.7	10
11	Postnatal <i>Arx</i> transcriptional activity regulates functional properties of PV interneurons. <i>iScience</i> , 2021, 24, 101999.	1.9	7
12	Improved Gene Therapy for Metachromatic Leukodystrophy. <i>Blood</i> , 2021, 138, 3979-3979.	0.6	0
13	Clinical utility of exome sequencing in infantile heart failure. <i>Genetics in Medicine</i> , 2020, 22, 423-426.	1.1	17
14	Brain Branched-Chain Amino Acids in Maple Syrup Urine Disease: Implications for Neurological Disorders. <i>International Journal of Molecular Sciences</i> , 2020, 21, 7490.	1.8	33
15	The variability of <i>SMARCA4</i> -related Coffin-Siris syndrome: Do nonsense candidate variants add to milder phenotypes?. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 2058-2067.	0.7	14
16	Newborn Screening for Pompe Disease: Pennsylvania Experience. <i>International Journal of Neonatal Screening</i> , 2020, 6, 89.	1.2	24
17	Natural history of multiple sulfatase deficiency: Retrospective phenotyping and functional variant analysis to characterize an ultra-rare disease. <i>Journal of Inherited Metabolic Disease</i> , 2020, 43, 1298-1309.	1.7	23
18	Multiple Sulfatase Deficiency: A Disease Comprising Mucopolysaccharidosis, Sphingolipidosis, and More Caused by a Defect in Posttranslational Modification. <i>International Journal of Molecular Sciences</i> , 2020, 21, 3448.	1.8	32

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19	The Importance of Succinylacetone: Tyrosinemia Type I Presenting with Hyperinsulinism and Multiorgan Failure Following Normal Newborn Screening. <i>International Journal of Neonatal Screening</i> , 2020, 6, 39.	1.2	5
20	A systematic review and meta-analysis of published cases reveals the natural disease history in multiple sulfatase deficiency. <i>Journal of Inherited Metabolic Disease</i> , 2020, 43, 1288-1297.	1.7	14
21	Imaging of non-neuronopathic Gaucher disease: recent advances in quantitative imaging and comprehensive assessment of disease involvement. <i>Insights Into Imaging</i> , 2019, 10, 70.	1.6	13
22	Disruption of cardiac thin filament assembly arising from a mutation in <i>LMOD2</i> : A novel mechanism of neonatal dilated cardiomyopathy. <i>Science Advances</i> , 2019, 5, eaax2066.	4.7	29
23	Case 2: Severe Hyperammonemia in a Neonate: An Alternate Ending. <i>NeoReviews</i> , 2019, 20, e90-e92.	0.4	1
24	Fetal cardiomyopathy in neurofibromatosis type I: Novel phenotype and review of the literature. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 1042-1046.	0.7	2
25	Characteristics and outcomes of patients with formiminoglutamic aciduria detected through newborn screening. <i>Journal of Inherited Metabolic Disease</i> , 2019, 42, 140-146.	1.7	7
26	Stimulation of entorhinal cortex dentate gyrus circuitry is antidepressive. <i>Nature Medicine</i> , 2018, 24, 658-666.	15.2	83
27	Complex care of individuals with multiple sulfatase deficiency: Clinical cases and consensus statement. <i>Molecular Genetics and Metabolism</i> , 2018, 123, 337-346.	0.5	31
28	Efficacy of early treatment in patients with cobalamin C disease identified by newborn screening: a 16-year experience. <i>Genetics in Medicine</i> , 2017, 19, 926-935.	1.1	16
29	Precision therapy for a new disorder of AMPA receptor recycling due to mutations in <i>ATAD1</i> . <i>Neurology: Genetics</i> , 2017, 3, e130.	0.9	40
30	Phenotypic predictors and final diagnoses in patients referred for RASopathy testing by targeted next-generation sequencing. <i>Genetics in Medicine</i> , 2017, 19, 715-718.	1.1	14
31	Morbidity and mortality among exclusively breastfed neonates with medium-chain acyl-CoA dehydrogenase deficiency. <i>Genetics in Medicine</i> , 2016, 18, 1315-1319.	1.1	11
32	Utility of genetic evaluation in infants with congenital heart defects admitted to the cardiac intensive care unit. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 3090-3097.	0.7	26
33	Response to van Rijt et al.. <i>Genetics in Medicine</i> , 2016, 18, 1324.	1.1	0
34	ECHS1 Deficiency as a Cause of Severe Neonatal Lactic Acidosis. <i>JIMD Reports</i> , 2016, 30, 33-37.	0.7	26
35	Atypical Williams syndrome in an infant with complete atrioventricular canal defect. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 3108-3112.	0.7	1
36	Adolescent Presentations of Inborn Errors of Metabolism. <i>Journal of Adolescent Health</i> , 2015, 56, 477-482.	1.2	16