Rebecca C Ahrens-Nicklas

List of Publications by Year
in descending order

36 papers	543 citations	687363 13 h-index	713466 21 g-index
37 all docs	37 docs citations	37 times ranked	1036 citing authors

#	Article	IF	Citations
1	Stimulation of entorhinal cortexâ€“dentate gyrus circuitry is antidepressive. Nature Medicine, 2018, 24, 658-666.	30.7	83
2	Precision therapy for a new disorder of AMPA receptor recycling due to mutations in ATAD1. Neurology: Genetics, 2017, 3, e130.	1.9	40
3	Brain Branched-Chain Amino Acids in Maple Syrup Urine Disease: Implications for Neurological Disorders. International Journal of Molecular Sciences, 2020, 21, 7490.	4.1	33
4	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.	4.1	32
5	Complex care of individuals with multiple sulfatase deficiency: Clinical cases and consensus statement. Molecular Genetics and Metabolism, 2018, 123, 337-346.	1.1	31
6	Disruption of cardiac thin filament assembly arising from a mutation in LMOD2 : A novel mechanism of neonatal dilated cardiomyopathy. Science Advances, 2019, 5, eaax2066.	10.3	29
7	Utility of genetic evaluation in infants with congenital heart defects admitted to the cardiac intensive care unit. American Journal of Medical Genetics, Part A, 2016, 170, 3090-3097.	1.2	26
8	ECHS1 Deficiency as a Cause of Severe Neonatal Lactic Acidosis. JIMD Reports, 2016, 30, 33-37.	1.5	26
9	Newborn Screening for Pompe Disease: Pennsylvania Experience. International Journal of Neonatal Screening, 2020, 6, 89.	3.2	24
10	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.	3.6	23
11	Clinical utility of exome sequencing in infantile heart failure. Genetics in Medicine, 2020, 22, 423-426.	2.4	17
12	Adolescent Presentations of Inborn Errors of Metabolism. Journal of Adolescent Health, 2015, 56, 477-482.	2.5	16
13	Efficacy of early treatment in patients with cobalamin C disease identified by newborn screening: a 16-year experience. Genetics in Medicine, 2017, 19, 926-935.	2.4	16
14	Phenotypic predictors and final diagnoses in patients referred for RASopathy testing by targeted next-generation sequencing. Genetics in Medicine, 2017, 19, 715-718.	2.4	14
15	The variability of <scp>SMARCA4</scp>â€related <scp>Coffinâ€“Siris</scp> syndrome: Do nonsense candidate variants add to milder phenotypes?. American Journal of Medical Genetics, Part A, 2020, 182, 2058-2067.	1.2	14
16	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.	3.6	14
17	Imaging of non-neuronopathic Gaucher disease: recent advances in quantitative imaging and comprehensive assessment of disease involvement. Insights Into Imaging, 2019, 10, 70.	3.4	13
18	Morbidity and mortality among exclusively breastfed neonates with medium-chain acyl-CoA dehydrogenase deficiency. Genetics in Medicine, 2016, 18, 1315-1319.	2.4	11

#	Article	IF	Citations
19	Newborn Screening for X-Linked Adrenoleukodystrophy: Review of Data and Outcomes in Pennsylvania. International Journal of Neonatal Screening, 2022, 8, 24.	3.2	11
20	Variants in <scp>NAA15</scp> cause pediatric hypertrophic cardiomyopathy. American Journal of Medical Genetics, Part A, 2021, 185, 228-233.	1.2	10
21	Consolidation of the clinical and genetic definition of aSOX4-related neurodevelopmental syndrome. Journal of Medical Genetics, 2022, 59, 1058-1068.	3.2	10
22	Genetic variant burden and adverse outcomes in pediatric cardiomyopathy. Pediatric Research, 2021, 89, 1470-1476.	2.3	9
23	Characteristics and outcomes of patients with formiminoglutamic aciduria detected through newborn screening. Journal of Inherited Metabolic Disease, 2019, 42, 140-146.	3.6	7
24	Postnatal Arx transcriptional activity regulates functional properties of PV interneurons. IScience, 2021, 24, 101999.	4.1	7
25	<scp>MYH7</scp> variants cause complex congenital heart disease. American Journal of Medical Genetics, Part A, 2022, 188, 2772-2776.	1.2	7
26	Genotypeâ€“phenotype association by echocardiography offers incremental value in patients with Noonan Syndrome with Multiple Lentigines. Pediatric Research, 2021, 90, 444-451.	2.3	6
27	The Importance of Succinylacetone: Tyrosinemia Type I Presenting with Hyperinsulinism and Multiorgan Failure Following Normal Newborn Screening. International Journal of Neonatal Screening, 2020, 6, 39.	3.2	5
28	Neuronal genetic rescue normalizes brain network dynamics in a lysosomal storage disorder despite persistent storage accumulation. Molecular Therapy, 2022, 30, 2464-2473.	8.2	4
29	Fetal cardiomyopathy in neurofibromatosis type I: Novel phenotype and review of the literature. American Journal of Medical Genetics, Part A, 2019, 179, 1042-1046.	1.2	2
30	Atypical Williams syndrome in an infant with complete atrioventricular canal defect. American Journal of Medical Genetics, Part A, 2015, 167, 3108-3112.	1.2	1
31	Case 2: Severe Hyperammonemia in a Neonate: An Alternate Ending. NeoReviews, 2019, 20, e90-e92.	0.8	1
32	Heal thyself: The promise of autologous hematopoietic stem cell gene therapy in neurometabolic disorders. Molecular Therapy, 2022, 30, 1353-1354.	8.2	1
33	Response to van Rijt et al.. Genetics in Medicine, 2016, 18, 1324.	2.4	0
34	Fine-Tuning 3-Methylglutaconic Aciduria Cutoffs for a Patient with Infantile-Onset Barth Syndrome. Clinical Chemistry, 2022, 68, 365-367.	3.2	0
35	Improved Gene Therapy for Metachromatic Leukodystrophy. Blood, 2021, 138, 3979-3979.	1.4	0
36	Contribution of Mendelian disorders in a population-based pediatric neurodegeneration cohort. Journal of Pediatrics, 2022, , .	1.8	0