Andrew C Edmondson

List of Publications by Year
in descending order

30 papers	800 citations	623734 14 h-index	526287 27 g-index
33 all docs	33 docs citations	33 times ranked	1529 citing authors

#	Article	IF	Citations
1	Expanding the phenotypic spectrum of ARCN1-related syndrome. Genetics in Medicine, 2022, 24, 1227-1237.	2.4	5
2	Newborn Screening for X-Linked Adrenoleukodystrophy: Review of Data and Outcomes in Pennsylvania. International Journal of Neonatal Screening, 2022, 8, 24.	3.2	11
3	Patient-reported outcomes and quality of life in PMM2-CDG. Molecular Genetics and Metabolism, 2022, 136, 145-151.	1.1	10
4	A rare cause of infantile achalasia: <scp>GMPPAâ€congenital</scp> disorder of glycosylation with two novel compound heterozygous variants. American Journal of Medical Genetics, Part A, 2022, 188, 2438-2442.	1.2	3
5	<scp>ALG8â€CDG</scp>: Molecular and phenotypic expansion suggests clinical management guidelines. Journal of Inherited Metabolic Disease, 2022, 45, 969-980.	3.6	5
6	International consensus guidelines for phosphoglucomutase 1 deficiency (<scp>PGM1â€CDG</scp>): Diagnosis, followâ€up, and management. Journal of Inherited Metabolic Disease, 2021, 44, 148-163.	3.6	27
7	X-linked cellular mosaicism underlies age-dependent occurrence of seizure-like events in mouse models of CDKL5 deficiency disorder. Neurobiology of Disease, 2021, 148, 105176.	4.4	21
8	Spontaneous improvement of carbohydrate-deficient transferrin in PMM2-CDG without mannose observed in CDG natural history study. Orphanet Journal of Rare Diseases, 2021, 16, 102.	2.7	8
9	ALG13 Xâ€linked intellectual disability: New variants, glycosylation analysis, and expanded phenotypes. Journal of Inherited Metabolic Disease, 2021, 44, 1001-1012.	3.6	9
10	Expanding the phenotype, genotype and biochemical knowledge of <scp>ALG3â€CDG</scp>. Journal of Inherited Metabolic Disease, 2021, 44, 987-1000.	3.6	4
11	PIGG variant pathogenicity assessment reveals characteristic features within 19 families. Genetics in Medicine, 2021, 23, 1873-1881.	2.4	5
12	Bi-allelic variants in the ER quality-control mannosidase gene EDEM3 cause a congenital disorder of glycosylation. American Journal of Human Genetics, 2021, 108, 1342-1349.	6.2	9
13	Manifestations and Management of Hepatic Dysfunction in Congenital Disorders of Glycosylation. Clinical Liver Disease, 2021, 18, 54-66.	2.1	0
14	Should patients with Phosphomannomutase 2-CDG (PMM2-CDG) be screened for adrenal insufficiency?. Molecular Genetics and Metabolism, 2021, 133, 397-399.	1.1	3
15	Liver manifestations in a cohort of 39 patients with congenital disorders of glycosylation: pin-pointing the characteristics of liver injury and proposing recommendations for follow-up. Orphanet Journal of Rare Diseases, 2021, 16, 20.	2.7	14
16	Sorbitol Is a Severity Biomarker for <scp>PMM2â€CDG</scp> with Therapeutic Implications. Annals of Neurology, 2021, 90, 887-900.	5.3	22
17	Early infantile epileptic encephalopathy due to biallelic pathogenic variants in <scp>PIGQ</scp>: Report of seven new subjects and review of the literature. Journal of Inherited Metabolic Disease, 2020, 43, 1321-1332.	3.6	15
18	Clinical and biochemical improvement with galactose supplementation in SLC35A2-CDG. Genetics in Medicine, 2020, 22, 1102-1107.	2.4	42

#	Article	IF	Citations
19	Novel congenital disorder of O-linked glycosylation caused by GALNT2 loss of function. Brain, 2020, 143, 1114-1126.	7.6	46
20	Congenital hyperinsulinism as the presenting feature of Kabuki syndrome: clinical and molecular characterization of 10 affected individuals. Genetics in Medicine, 2019, 21, 233-242.	2.4	39
21	The Metabolic Map into the Pathomechanism and Treatment of PGM1-CDG. American Journal of Human Genetics, 2019, 104, 835-846.	6.2	59
22	Increased Clinical Sensitivity and Specificity of Plasma Protein N-Glycan Profiling for Diagnosing Congenital Disorders of Glycosylation by Use of Flow Injectionâ€“Electrospray Ionizationâ€“Quadrupole Time-of-Flight Mass Spectrometry. Clinical Chemistry, 2019, 65, 653-663.	3.2	40
23	A human case of SLC35A3â€related skeletal dysplasia. American Journal of Medical Genetics, Part A, 2017, 173, 2758-2762.	1.2	20
24	Gainâ€ofâ€function mutations in SMAD4 cause a distinctive repertoire of cardiovascular phenotypes in patients with Myhre syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 2617-2631.	1.2	53
25	Missed Newborn Screening Case of Carnitine Palmitoyltransferase-II Deficiency. JIMD Reports, 2016, 33, 93-97.	1.5	17
26	Kabuki syndrome as a cause of nonâ€immune fetal hydrops/ascites. American Journal of Medical Genetics, Part A, 2016, 170, 3333-3337.	1.2	11
27	Loss of Function of GALNT2 Lowers High-Density Lipoproteins in Humans, Nonhuman Primates, and Rodents. Cell Metabolism, 2016, 24, 234-245.	16.2	103
28	ECHS1 Deficiency as a Cause of Severe Neonatal Lactic Acidosis. JIMD Reports, 2016, 30, 33-37.	1.5	26
29	Overgrowth Syndromes. Journal of Pediatric Genetics, 2015, 04, 136-143.	0.7	57
30	Germline gain-of-function mutations in AFF4 cause a developmental syndrome functionally linking the super elongation complex and cohesin. Nature Genetics, 2015, 47, 338-344.	21.4	109