Kimiyo Raymond

List of Publications by Year
in descending order

13	364	1040056 9	1125743 13
papers	citations	h-index	g-index

13	13	13	536
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Precision newborn screening for lysosomal disorders. Genetics in Medicine, 2018, 20, 847-854.	2.4	99
2	Oral D-galactose supplementation in PGM1-CDG. Genetics in Medicine, 2017, 19, 1226-1235.	2.4	55
3	The critical role of psychosine in screening, diagnosis, and monitoring of Krabbe disease. Genetics in Medicine, 2020, 22, 1108-1118.	2.4	39
4	Integrating glycomics and genomics uncovers SLC10A7 as essential factor for bone mineralization by regulating post-Golgi protein transport and glycosylation. Human Molecular Genetics, 2018, 27, 3029-3045.	2.9	37
5	Incorporation of Second-Tier Biomarker Testing Improves the Specificity of Newborn Screening for Mucopolysaccharidosis Type I. International Journal of Neonatal Screening, 2020, 6, 10.	3.2	32
6	Sorbitol Is a Severity Biomarker for <scp>PMM2â€CDG</scp> with Therapeutic Implications. Annals of Neurology, 2021, 90, 887-900.	5.3	22
7	The Combined Impact of CLIR Post-Analytical Tools and Second Tier Testing on the Performance of Newborn Screening for Disorders of Propionate, Methionine, and Cobalamin Metabolism. International Journal of Neonatal Screening, 2020, 6, 33.	3.2	19
8	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
9	NGLY1 Deficiency: A Rare Newly Described Condition with a Typical Presentation. Life, 2021, 11, 187.	2.4	12
10	Multiplex testing for the screening of lysosomal storage disease in urine: Sulfatides and glycosaminoglycan profiles in 40 cases of sulfatiduria. Molecular Genetics and Metabolism, 2020, 129, 106-110.	1.1	10
11	A new <scp>Dâ€galactose</scp> treatment monitoring index for <scp>PGM1â€CDG</scp>. Journal of Inherited Metabolic Disease, 2021, 44, 1263-1271.	3.6	10
12	Defining the mild variant of leukocyte adhesion deficiency type <scp>II</scp> (<scp>SLC35C1</scp>â€congenital disorder of glycosylation) and response to <scp>l</scp>â€fucose therapy: Insights from two new families and review of the literature. American Journal of Medical Genetics, Part A, 2022, 188, 2005-2018.	1.2	10
13	Immune dysfunction in MGAT2â€CDG : A clinical report and review of the literature. American Journal of Medical Genetics, Part A, 2021, 185, 213-218.	1.2	5