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	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
2	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
3	NGLY1 Deficiency: A Rare Newly Described Condition with a Typical Presentation. Life, 2021, 11, 187.	2.4	12
4	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
5	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
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	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
8	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
9	The critical role of psychosine in screening, diagnosis, and monitoring of Krabbe disease. Genetics in Medicine, 2020, 22, 1108-1118.	2.4	39
10	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
11	Precision newborn screening for lysosomal disorders. Genetics in Medicine, 2018, 20, 847-854.	2.4	99
12	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
13	Oral D-galactose supplementation in PGM1-CDG. Genetics in Medicine, 2017, 19, 1226-1235.	2.4	55