Bruno Maranda

List of Publications by Year
in descending order

18	571	687363 13	839539 18
papers	citations	h-index	g-index

18	18	18	1218
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Lysosphingolipid urine screening test using mass spectrometry for theÂearly detection of lysosomal storage disorders. Bioanalysis, 2022, 14, 289-306.	1.5	1
2	Neonatal Urine Screening Program in the Province of Quebec: Technological Upgrade from Thin Layer Chromatography to Tandem Mass Spectrometry. International Journal of Neonatal Screening, 2021, 7, 18.	3.2	4
3	Diurnal Variation of Urinary Fabry Disease Biomarkers during Enzyme Replacement Therapy Cycles. International Journal of Molecular Sciences, 2020, 21, 6114.	4.1	3
4	Outcomes in pediatric studies of medium-chain acyl-coA dehydrogenase (MCAD) deficiency and phenylketonuria (PKU): a review. Orphanet Journal of Rare Diseases, 2020, 15, 12.	2.7	15
5	HSD10 mitochondrial disease: p.Leu122Val variant, mild clinical phenotype, and founder effect in Frenchâ€Canadian patients from Quebec. Molecular Genetics & Genomic Medicine, 2019, 7, e1000.	1.2	8
6	Tandem mass spectrometry analysis of urinary podocalyxin and podocin in the investigation of podocyturia in women with preeclampsia and Fabry disease patients. Clinica Chimica Acta, 2019, 495, 67-75.	1.1	17
7	Combined malonic and methylmalonic aciduria due to ACSF3 mutations: Benign clinical course in an unselected cohort. Journal of Inherited Metabolic Disease, 2019, 42, 107-116.	3.6	23
8	Combined malonic and methylmalonic aciduria due to ACSF3 mutations: benign clinical course in an unselected cohort. Journal of Inherited Metabolic Disease, 2019, 42, 107.	3.6	4
9	Clinical validity of phenotype-driven analysis software PhenoVar as a diagnostic aid for clinical geneticists in the interpretation of whole-exome sequencing data. Genetics in Medicine, 2018, 20, 942-949.	2.4	15
10	Evaluation of urinary keratan sulfate disaccharides in MPS IVA patients using UPLCâ€“MS/MS. Bioanalysis, 2016, 8, 179-191.	1.5	17
11	UPLC-MS/MS detection of disaccharides derived from glycosaminoglycans as biomarkers of mucopolysaccharidoses. Analytica Chimica Acta, 2016, 936, 139-148.	5.4	53
12	Three new cases of terminal deletion of the long arm of chromosome 7 and literature review to correlate genotype and phenotype manifestations. American Journal of Medical Genetics, Part A, 2016, 170, 896-907.	1.2	21
13	Diagnosis of late-onset Pompe disease and other muscle disorders by next-generation sequencing. Orphanet Journal of Rare Diseases, 2016, 11, 8.	2.7	42
14	Retinal lipid and glucose metabolism dictates angiogenesis through the lipid sensor Ffar1. Nature Medicine, 2016, 22, 439-445.	30.7	183
15	Identifying candidate genes for 2p15p16.1 microdeletion syndrome using clinical, genomic, and functional analysis. JCI Insight, 2016, 1, e85461.	5.0	22
16	Guanidinoacetate methyltransferase (GAMT) deficiency: Outcomes in 48 individuals and recommendations for diagnosis, treatment and monitoring. Molecular Genetics and Metabolism, 2014, 111, 16-25.	1.1	111
17	High-throughput tandem mass spectrometry multiplex analysis for newborn urinary screening of creatine synthesis and transport disorders, Triple H syndrome and OTC deficiency. Clinica Chimica Acta, 2014, 436, 249-255.	1.1	14
18	Spinal muscular atrophy: Clinical validation of a single-tube multiplex real time PCR assay for determination of SMN1 and SMN2 copy numbers. Clinical Biochemistry, 2012, 45, 88-91.	1.9	18