

# Bruno Maranda

## List of Publications by Year in descending order

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Version: 2024-02-01

18  
papers

571  
citations

687363

13  
h-index

839539

18  
g-index

18  
all docs

18  
docs citations

18  
times ranked

1218  
citing authors

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