

Christiane Auray-Blais

List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

90
papers

2,353
citations

28
h-index

45
g-index

105
ext. papers

2,696
ext. citations

5
avg, IF

4.87
L-index

#	Paper	IF	Citations
90	Methylmalonic acid analysis using urine filter paper samples to screen for metabolic vitamin B deficiency in older adults.. <i>Bioanalysis</i> , 2022 , 14, 615-626	2.1	0
89	Globotriaosylsphingosine (lyso-Gb) and analogues in plasma and urine of patients with Fabry disease and correlations with long-term treatment and genotypes in a nationwide female Danish cohort. <i>Journal of Medical Genetics</i> , 2021 , 58, 692-700	5.8	4
88	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,	2.6	2
87	Assessment of plasma lyso-Gb for clinical monitoring of treatment response in migalastat-treated patients with Fabry disease. <i>Genetics in Medicine</i> , 2021 , 23, 192-201	8.1	6
86	Lentivirus-mediated gene therapy for Fabry disease. <i>Nature Communications</i> , 2021 , 12, 1178	17.4	21
85	ATP-binding cassette transporters mediate differential biosynthesis of glycosphingolipid species. <i>Journal of Lipid Research</i> , 2021 , 100128	6.3	3
84	Identification of a Reliable Biomarker Profile for the Diagnosis of Gaucher Disease Type 1 Patients Using a Mass Spectrometry-Based Metabolomic Approach. <i>International Journal of Molecular Sciences</i> , 2020 , 21,	6.3	3
83	Effects of Orally Delivered Alpha-Galactosidase A on Gastrointestinal Symptoms in Patients With Fabry Disease. <i>Gastroenterology</i> , 2020 , 159, 1602-1604	13.3	6
82	Therapeutic challenges in two adolescent male patients with Fabry disease and high antibody titres. <i>Molecular Genetics and Metabolism Reports</i> , 2020 , 24, 100618	1.8	1
81	The clinical utility of total concentration of urinary globotriaosylsphingosine plus its analogues in the diagnosis of Fabry disease. <i>Clinica Chimica Acta</i> , 2020 , 500, 120-127	6.2	11
80	High-risk screening for Fabry disease in a Canadian cohort of chronic kidney disease patients. <i>Clinica Chimica Acta</i> , 2020 , 501, 234-240	6.2	6
79	Assessing the role of glycosphingolipids in the phenotype severity of Fabry disease mouse model. <i>Journal of Lipid Research</i> , 2020 , 61, 1410-1423	6.3	3
78	Mass spectrometry analysis of urinary methylmalonic acid to screen for metabolic vitamin B deficiency in older adults. <i>Bioanalysis</i> , 2020 , 12, 693-705	2.1	2
77	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	6.2	11
76	Mutation-specific Fabry disease patient-derived cell model to evaluate the amenability to chaperone therapy. <i>Journal of Medical Genetics</i> , 2019 , 56, 548-556	5.8	29
75	FACTs Fabry gene therapy clinical trial: Two-year data. <i>Molecular Genetics and Metabolism</i> , 2019 , 126, S99	3.7	8
74	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	5.4	16

73	Distribution of heparan sulfate and dermatan sulfate in mucopolysaccharidosis type II mouse tissues pre- and post-enzyme-replacement therapy determined by UPLC-MS/MS. <i>Bioanalysis</i> , 2019 , 11, 727-740	2.1	4
72	Altered immune phenotypes in subjects with Fabry disease and responses to switching from agalsidase alfa to agalsidase beta. <i>American Journal of Translational Research (discontinued)</i> , 2019 , 11, 1683-1696	3	3
71	Combined malonic and methylmalonic aciduria due to ACSF3 mutations: Benign clinical course in an unselected cohort 2019 , 42, 107		1
70	Neonatal Mass Urine Screening Approach for Early Detection of Mucopolysaccharidoses by UPLC-MS/MS. <i>Diagnostics</i> , 2019 , 9,	3.8	7
69	Globotriaosylsphingosine (Lyso-Gb) as a biomarker for cardiac variant (N215S) Fabry disease. <i>Journal of Inherited Metabolic Disease</i> , 2018 , 41, 239-247	5.4	16
68	Contribution of tandem mass spectrometry to the diagnosis of lysosomal storage disorders. <i>Journal of Inherited Metabolic Disease</i> , 2018 , 41, 457-477	5.4	14
67	Analysis of globotriaosylceramide (Gb) isoforms/analogs in unfractionated leukocytes, B lymphocytes and monocytes from Fabry patients using ultra-high performance liquid chromatography/tandem mass spectrometry. <i>Analytica Chimica Acta</i> , 2018 , 1015, 35-49	6.6	6
66	UPLC-MS/MS analysis of keratan sulfate from urine samples collected on filter paper for monitoring & follow-up of Morquio A patients. <i>Bioanalysis</i> , 2018 , 10, 1181-1192	2.1	3
65	Glutaric Aciduria Type 3: Three Unrelated Canadian Cases, with Different Routes of Ascertainment. <i>JIMD Reports</i> , 2018 , 39, 89-96	1.9	3
64	The lysosomal enzyme alpha-Galactosidase A is deficient in Parkinson's disease brain in association with the pathologic accumulation of alpha-synuclein. <i>Neurobiology of Disease</i> , 2018 , 110, 68-81	7.5	25
63	Elevated Inflammatory Plasma Biomarkers in Patients With Fabry Disease: A Critical Link to Heart Failure With Preserved Ejection Fraction. <i>Journal of the American Heart Association</i> , 2018 , 7, e009098	6	29
62	Biomarkers associated with clinical manifestations in Fabry disease patients with a late-onset cardiac variant mutation. <i>Clinica Chimica Acta</i> , 2017 , 466, 185-193	6.2	35
61	High-Risk Screening for Fabry Disease: Analysis by Tandem Mass Spectrometry of Globotriaosylceramide (Gb) in Urine Collected on Filter Paper. <i>Current Protocols in Human Genetics</i> , 2017 , 93, 17.26.1-17.26.12	3.2	8
60	Lentivector Iterations and Pre-Clinical Scale-Up/Toxicity Testing: Targeting Mobilized CD34 Cells for Correction of Fabry Disease. <i>Molecular Therapy - Methods and Clinical Development</i> , 2017 , 5, 241-258	6.4	28
59	Separation and Analysis of Lactosylceramide, Galabiosylceramide, and Globotriaosylceramide by LC-MS/MS in Urine of Fabry Disease Patients. <i>Analytical Chemistry</i> , 2017 , 89, 13382-13390	7.8	21
58	Fabry Disease Biomarkers: Analysis of Urinary Lyso-Gb3 and Seven Related Analogs Using Tandem Mass Spectrometry. <i>Current Protocols in Human Genetics</i> , 2016 , 90, 17.22.1-17.22.12	3.2	5
57	UPLC-MS/MS detection of disaccharides derived from glycosaminoglycans as biomarkers of mucopolysaccharidoses. <i>Analytica Chimica Acta</i> , 2016 , 936, 139-48	6.6	45
56	Relative distribution of Gb3 isoforms/analogs in NOD/SCID/Fabry mice tissues determined by tandem mass spectrometry. <i>Bioanalysis</i> , 2016 , 8, 1793-807	2.1	12

55	High-Risk Screening of Fabry Disease: Analysis of Fifteen Urinary Methylated and Non-Methylated Gb Isoforms Using Tandem Mass Spectrometry. <i>Current Protocols in Human Genetics</i> , 2016 , 91, 17.24.1-17.24.11 ⁰	3.2	11 ⁰
54	Diagnosis of late-onset Pompe disease and other muscle disorders by next-generation sequencing. <i>Orphanet Journal of Rare Diseases</i> , 2016 , 11, 8	4.2	33
53	Tandem Mass Spectrometry Multiplex Analysis of Glucosylceramide and Galactosylceramide Isoforms in Brain Tissues at Different Stages of Parkinson Disease. <i>Analytical Chemistry</i> , 2016 , 88, 1856-63 ⁸	7.8	52
52	Sulfatide Analysis by Mass Spectrometry for Screening of Metachromatic Leukodystrophy in Dried Blood and Urine Samples. <i>Clinical Chemistry</i> , 2016 , 62, 279-86	5.5	44
51	Tandem mass spectrometry multiplex analysis of methylated and non-methylated urinary Gb3 isoforms in Fabry disease patients. <i>Clinica Chimica Acta</i> , 2016 , 452, 191-8	6.2	17
50	Glycosphingolipid storage in Fabry mice extends beyond globotriaosylceramide and is affected by ABCB1 depletion. <i>Future Science OA</i> , 2016 , 2, FSO147	2.7	5
49	Evaluation of urinary keratan sulfate disaccharides in MPS IVA patients using UPLC-MS/MS. <i>Bioanalysis</i> , 2016 , 8, 179-91	2.1	16
48	Tandem Mass Spectrometry Quantitation of Lyso-Gb3 and Six Related Analogs in Plasma for Fabry Disease Patients. <i>Current Protocols in Human Genetics</i> , 2016 , 90, 17.23.1-17.23.9	3.2	12
47	Maternal inhaled fluticasone propionate intake during pregnancy is detected in neonatal cord blood. <i>Bioanalysis</i> , 2016 , 8, 1441-1450	2.1	2
46	Variations in the GLA gene correlate with globotriaosylceramide and globotriaosylsphingosine analog levels in urine and plasma. <i>Clinica Chimica Acta</i> , 2015 , 447, 96-104	6.2	17
45	Assessment of urinary metabolite excretion after rat acute exposure to perfluorooctanoic acid and other peroxisomal proliferators. <i>Archives of Environmental Contamination and Toxicology</i> , 2015 , 68, 148-58 ²	3.2	6
44	Urinary biomarker investigation in children with Fabry disease using tandem mass spectrometry. <i>Clinica Chimica Acta</i> , 2015 , 438, 195-204	6.2	48
43	Metabolomic discovery of novel urinary galabiosylceramide analogs as Fabry disease biomarkers. <i>Journal of the American Society for Mass Spectrometry</i> , 2015 , 26, 499-510	3.5	31
42	Multiplex tandem mass spectrometry analysis of novel plasma lyso-Gb3-related analogues in Fabry disease. <i>Analytical Chemistry</i> , 2014 , 86, 3476-83	7.8	61
41	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, 249-55	6.2	11
40	Glycation of fetal hemoglobin reflects hyperglycemia exposure in utero. <i>Diabetes Care</i> , 2014 , 37, 2830-314.6	14.6	3
39	A metabolomic study to identify new globotriaosylceramide-related biomarkers in the plasma of Fabry disease patients. <i>Analytical Chemistry</i> , 2013 , 85, 9039-48	7.8	47
38	Multiplex analysis of novel urinary lyso-Gb3-related biomarkers for Fabry disease by tandem mass spectrometry. <i>Analytical Chemistry</i> , 2013 , 85, 1743-52	7.8	62

37	Mutations in the sarcosine dehydrogenase gene in patients with sarcosinemia. <i>Human Genetics</i> , 2012 , 131, 1805-10	6.3	15
36	LC-MS/MS analysis of plasma lyso-Gb3 in Fabry disease. <i>Clinica Chimica Acta</i> , 2012 , 414, 273-80	6.2	46
35	Urinary globotriaosylsphingosine-related biomarkers for Fabry disease targeted by metabolomics. <i>Analytical Chemistry</i> , 2012 , 84, 2745-53	7.8	69
34	An improved method for glycosaminoglycan analysis by LC-MS/MS of urine samples collected on filter paper. <i>Clinica Chimica Acta</i> , 2012 , 413, 771-8	6.2	56
33	Novel gb(3) isoforms detected in urine of fabry disease patients: a metabolomic study. <i>Current Medicinal Chemistry</i> , 2012 , 19, 3241-52	4.3	39
32	Improved ways to screen for patients with Fabry disease, involving optometry in a multidisciplinary approach 2012 , 74, 25		7
31	Clinical validation of cutoff target ranges in newborn screening of metabolic disorders by tandem mass spectrometry: a worldwide collaborative project. <i>Genetics in Medicine</i> , 2011 , 13, 230-54	8.1	250
30	Efficient analysis of urinary glycosaminoglycans by LC-MS/MS in mucopolysaccharidoses type I, II and VI. <i>Molecular Genetics and Metabolism</i> , 2011 , 102, 49-56	3.7	86
29	Analysis of trace metals in single droplet of urine by laser ablation inductively coupled plasma mass spectrometry. <i>International Journal of Mass Spectrometry</i> , 2011 , 307, 174-181	1.9	28
28	Mass spectrometry analysis of metals, other elements and lipids in urine samples of Fabry disease patients. <i>International Journal of Mass Spectrometry</i> , 2011 , 307, 163-173	1.9	
27	Metabolomics and preterm birth: What biomarkers in cervicovaginal secretions are predictive of high-risk pregnant women?. <i>International Journal of Mass Spectrometry</i> , 2011 , 307, 33-38	1.9	23
26	Determination of glycated and acetylated hemoglobins in cord blood by time-of-flight mass spectrometry. <i>Analytical Chemistry</i> , 2011 , 83, 5245-52	7.8	3
25	Analysis of glycosaminoglycans in cerebrospinal fluid from patients with mucopolysaccharidoses by isotope-dilution ultra-performance liquid chromatography-tandem mass spectrometry. <i>Clinical Chemistry</i> , 2011 , 57, 1005-12	5.5	73
24	Loss-of-function mutations in the glutamate transporter SLC1A1 cause human dicarboxylic aminoaciduria. <i>Journal of Clinical Investigation</i> , 2011 , 121, 446-53	15.9	98
23	Biomarkers of Fabry disease nephropathy. <i>Clinical Journal of the American Society of Nephrology: CJASN</i> , 2010 , 5, 360-4	6.9	46
22	How well does urinary lyso-Gb3 function as a biomarker in Fabry disease?. <i>Clinica Chimica Acta</i> , 2010 , 411, 1906-14	6.2	83
21	Biomarkers in fabry disease. <i>Clinical Therapeutics</i> , 2009 , 31, S24-S25	3.5	4
20	Proposed high-risk screening protocol for Fabry disease in patients with renal and vascular disease. <i>Journal of Inherited Metabolic Disease</i> , 2009 , 32, 303-8	5.4	26

19	Gb(3)/creatinine biomarkers for Fabry disease: issues to consider. <i>Molecular Genetics and Metabolism</i> , 2009 , 97, 237	3.7	18
18	Fabry disease urinary globotriaosylceramide/creatinine biomarker evaluation by liquid chromatography-tandem mass spectrometry in healthy infants from birth to 6 months. <i>Molecular Genetics and Metabolism</i> , 2009 , 97, 278-83	3.7	18
17	Urinary globotriaosylceramide excretion correlates with the genotype in children and adults with Fabry disease. <i>Molecular Genetics and Metabolism</i> , 2008 , 93, 331-40	3.7	84
16	Further evidence for allelic heterogeneity in Hartnup disorder. <i>Human Mutation</i> , 2008 , 29, 1217-21	4.7	25
15	Iminoglycinuria and hyperglycinuria are discrete human phenotypes resulting from complex mutations in proline and glycine transporters. <i>Journal of Clinical Investigation</i> , 2008 , 118, 3881-92	15.9	80
14	Persistence of the common Hartnup disease D173N allele in populations of European origin. <i>Annals of Human Genetics</i> , 2007 , 71, 755-61	2.2	12
13	Quebec neonatal mass urinary screening programme: from micromolecules to macromolecules. <i>Journal of Inherited Metabolic Disease</i> , 2007 , 30, 515-21	5.4	57
12	Biobanking primer: down to basics. <i>Science</i> , 2007 , 316, 830	33.3	5
11	A biobank management model applicable to biomedical research. <i>BMC Medical Ethics</i> , 2006 , 7, E4	2.9	27
10	Newborn urine screening programme in the province of Quebec: an update of 30 years experience. <i>Journal of Inherited Metabolic Disease</i> , 2003 , 26, 393-402	5.4	26
9	Outcome of individuals with low-moderate methylmalonic aciduria detected through a neonatal screening program. <i>Journal of Pediatrics</i> , 1999 , 134, 675-80	3.6	48
8	Screening for Neuroblastoma at 3 Weeks of Age: Methods and Preliminary Results From the Quebec Neuroblastoma Screening Project. <i>Pediatrics</i> , 1990 , 86, 765-773	7.4	25
7	Thin-layer chromatography of urinary homovanillic acid and vanillylmandelic acid for large-scale neuroblastoma mass screening. <i>Medical and Pediatric Oncology</i> , 1989 , 17, 364-7		15
6	Neuroblastoma screening: the Canadian experience. <i>Medical and Pediatric Oncology</i> , 1989 , 17, 379-81		10
5	Determination of urinary homovanillic and vanillylmandelic acids from dried filter paper samples: assessment of potential methods for neuroblastoma screening. <i>Clinical Biochemistry</i> , 1987 , 20, 173-7	3.5	25
4	Ontogeny modifies manifestations of cystinuria genes: implications for counseling. <i>Journal of Pediatrics</i> , 1985 , 106, 411-6	3.6	31
3	Diet and medications giving positive ninhydrin reactions on TLC in a newborn urinary screening program. <i>Clinical Biochemistry</i> , 1980 , 13, 103-5	3.5	4
2	Rapid thin-layer chromatographic method for the detection of urinary methylmalonic acid. <i>Clinical Biochemistry</i> , 1979 , 12, 43-5	3.5	14

- 1 Simple and rapid system for screening and identification of reducing sugars in urine. *Clinical Biochemistry*, **1978**, 11, 235-7 3.5 6