Christiane Auray-Blais

List of Publications by Citations

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	Clinical validation of cutoff target ranges in newborn screening of metabolic disorders by tandem mass spectrometry: a worldwide collaborative project. *Genetics in Medicine*, **2011**, 13, 230-54	8.1	250
89	Loss-of-function mutations in the glutamate transporter SLC1A1 cause human dicarboxylic aminoaciduria. *Journal of Clinical Investigation*, **2011**, 121, 446-53	15.9	98
88	Efficient analysis of urinary glycosaminoglycans by LC-MS/MS in mucopolysaccharidoses type I, II and VI. *Molecular Genetics and Metabolism*, **2011**, 102, 49-56	3.7	86
87	Urinary globotriaosylceramide excretion correlates with the genotype in children and adults with Fabry disease. *Molecular Genetics and Metabolism*, **2008**, 93, 331-40	3.7	84
86	How well does urinary lyso-Gb3 function as a biomarker in Fabry disease?. *Clinica Chimica Acta*, **2010**, 411, 1906-14	6.2	83
85	Iminoglycinuria and hyperglycinuria are discrete human phenotypes resulting from complex mutations in proline and glycine transporters. *Journal of Clinical Investigation*, **2008**, 118, 3881-92	15.9	80
84	Analysis of glycosaminoglycans in cerebrospinal fluid from patients with mucopolysaccharidoses by isotope-dilution ultra-performance liquid chromatography-tandem mass spectrometry. *Clinical Chemistry*, **2011**, 57, 1005-12	5.5	73
83	Urinary globotriaosylsphingosine-related biomarkers for Fabry disease targeted by metabolomics. *Analytical Chemistry*, **2012**, 84, 2745-53	7.8	69
82	Multiplex analysis of novel urinary lyso-Gb3-related biomarkers for Fabry disease by tandem mass spectrometry. *Analytical Chemistry*, **2013**, 85, 1743-52	7.8	62
81	Multiplex tandem mass spectrometry analysis of novel plasma lyso-Gb□-related analogues in Fabry disease. *Analytical Chemistry*, **2014**, 86, 3476-83	7.8	61
80	Quebec neonatal mass urinary screening programme: from micromolecules to macromolecules. *Journal of Inherited Metabolic Disease*, **2007**, 30, 515-21	5.4	57
79	An improved method for glycosaminoglycan analysis by LC-MS/MS of urine samples collected on filter paper. *Clinica Chimica Acta*, **2012**, 413, 771-8	6.2	56
78	Tandem Mass Spectrometry Multiplex Analysis of Glucosylceramide and Galactosylceramide Isoforms in Brain Tissues at Different Stages of Parkinson Disease. *Analytical Chemistry*, **2016**, 88, 1856-63	7.8	52
77	Urinary biomarker investigation in children with Fabry disease using tandem mass spectrometry. *Clinica Chimica Acta*, **2015**, 438, 195-204	6.2	48
76	Outcome of individuals with low-moderate methylmalonic aciduria detected through a neonatal screening program. *Journal of Pediatrics*, **1999**, 134, 675-80	3.6	48
75	A metabolomic study to identify new globotriaosylceramide-related biomarkers in the plasma of Fabry disease patients. *Analytical Chemistry*, **2013**, 85, 9039-48	7.8	47
74	LC-MS/MS analysis of plasma lyso-Gb3 in Fabry disease. *Clinica Chimica Acta*, **2012**, 414, 273-80	6.2	46

73	Biomarkers of Fabry disease nephropathy. *Clinical Journal of the American Society of Nephrology: CJASN*, **2010**, 5, 360-4	6.9	46
72	UPLC-MS/MS detection of disaccharides derived from glycosaminoglycans as biomarkers of mucopolysaccharidoses. *Analytica Chimica Acta*, **2016**, 936, 139-48	6.6	45
71	Sulfatide Analysis by Mass Spectrometry for Screening of Metachromatic Leukodystrophy in Dried Blood and Urine Samples. *Clinical Chemistry*, **2016**, 62, 279-86	5.5	44
70	Novel gb(3) isoforms detected in urine of fabry disease patients: a metabolomic study. *Current Medicinal Chemistry*, **2012**, 19, 3241-52	4.3	39
69	Biomarkers associated with clinical manifestations in Fabry disease patients with a late-onset cardiac variant mutation. *Clinica Chimica Acta*, **2017**, 466, 185-193	6.2	35
68	Diagnosis of late-onset Pompe disease and other muscle disorders by next-generation sequencing. *Orphanet Journal of Rare Diseases*, **2016**, 11, 8	4.2	33
67	Metabolomic discovery of novel urinary galabiosylceramide analogs as Fabry disease biomarkers. *Journal of the American Society for Mass Spectrometry*, **2015**, 26, 499-510	3.5	31
66	Ontogeny modifies manifestations of cystinuria genes: implications for counseling. *Journal of Pediatrics*, **1985**, 106, 411-6	3.6	31
65	Mutation-specific Fabry disease patient-derived cell model to evaluate the amenability to chaperone therapy. *Journal of Medical Genetics*, **2019**, 56, 548-556	5.8	29
64	Elevated Inflammatory Plasma Biomarkers in Patients With Fabry Disease: A Critical Link to Heart Failure With Preserved Ejection Fraction. *Journal of the American Heart Association*, **2018**, 7, e009098	6	29
63	Lentivector Iterations and Pre-Clinical Scale-Up/Toxicity Testing: Targeting Mobilized CD34 Cells for Correction of Fabry Disease. *Molecular Therapy - Methods and Clinical Development*, **2017**, 5, 241-258	6.4	28
62	Analysis of trace metals in single droplet of urine by laser ablation inductively coupled plasma mass spectrometry. *International Journal of Mass Spectrometry*, **2011**, 307, 174-181	1.9	28
61	A biobank management model applicable to biomedical research. *BMC Medical Ethics*, **2006**, 7, E4	2.9	27
60	Proposed high-risk screening protocol for Fabry disease in patients with renal and vascular disease. *Journal of Inherited Metabolic Disease*, **2009**, 32, 303-8	5.4	26
59	Newborn urine screening programme in the province of Quebec: an update of 30 yearsTexperience. *Journal of Inherited Metabolic Disease*, **2003**, 26, 393-402	5.4	26
58	Further evidence for allelic heterogeneity in Hartnup disorder. *Human Mutation*, **2008**, 29, 1217-21	4.7	25
57	Determination of urinary homovanillic and vanillylmandelic acids from dried filter paper samples: assessment of potential methods for neuroblastoma screening. *Clinical Biochemistry*, **1987**, 20, 173-7	3.5	25
56	Screening for Neuroblastoma at 3 Weeks of Age: Methods and Preliminary Results From the Quebec Neuroblastoma Screening Project. *Pediatrics*, **1990**, 86, 765-773	7.4	25

55	The lysosomal enzyme alpha-Galactosidase A is deficient in Parkinson's disease brain in association with the pathologic accumulation of alpha-synuclein. *Neurobiology of Disease*, **2018**, 110, 68-81	7.5	25
54	Metabolomics and preterm birth: What biomarkers in cervicovaginal secretions are predictive of high-risk pregnant women?. *International Journal of Mass Spectrometry*, **2011**, 307, 33-38	1.9	23
53	Separation and Analysis of Lactosylceramide, Galabiosylceramide, and Globotriaosylceramide by LC-MS/MS in Urine of Fabry Disease Patients. *Analytical Chemistry*, **2017**, 89, 13382-13390	7.8	21
52	Lentivirus-mediated gene therapy for Fabry disease. *Nature Communications*, **2021**, 12, 1178	17.4	21
51	Gb(3)/creatinine biomarkers for Fabry disease: issues to consider. *Molecular Genetics and Metabolism*, **2009**, 97, 237	3.7	18
50	Fabry disease urinary globotriaosylceramide/creatinine biomarker evaluation by liquid chromatography-tandem mass spectrometry in healthy infants from birth to 6 months. *Molecular Genetics and Metabolism*, **2009**, 97, 278-83	3.7	18
49	Variations in the GLA gene correlate with globotriaosylceramide and globotriaosylsphingosine analog levels in urine and plasma. *Clinica Chimica Acta*, **2015**, 447, 96-104	6.2	17
48	Tandem mass spectrometry multiplex analysis of methylated and non-methylated urinary Gb3 isoforms in Fabry disease patients. *Clinica Chimica Acta*, **2016**, 452, 191-8	6.2	17
47	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	5.4	16
46	Globotriaosylsphingosine (Lyso-Gb) as a biomarker for cardiac variant (N215S) Fabry disease. *Journal of Inherited Metabolic Disease*, **2018**, 41, 239-247	5.4	16
45	Evaluation of urinary keratan sulfate disaccharides in MPS IVA patients using UPLC-MS/MS. *Bioanalysis*, **2016**, 8, 179-91	2.1	16
44	Mutations in the sarcosine dehydrogenase gene in patients with sarcosinemia. *Human Genetics*, **2012**, 131, 1805-10	6.3	15
43	Thin-layer chromatography of urinary homovanillic acid and vanillylmandelic acid for large-scale neuroblastoma mass screening. *Medical and Pediatric Oncology*, **1989**, 17, 364-7		15
42	Contribution of tandem mass spectrometry to the diagnosis of lysosomal storage disorders. *Journal of Inherited Metabolic Disease*, **2018**, 41, 457-477	5.4	14
41	Rapid thin-layer chromatographic method for the detection of urinary methylmalonic acid. *Clinical Biochemistry*, **1979**, 12, 43-5	3.5	14
40	Relative distribution of Gb3 isoforms/analogs in NOD/SCID/Fabry mice tissues determined by tandem mass spectrometry. *Bioanalysis*, **2016**, 8, 1793-807	2.1	12
39	Persistence of the common Hartnup disease D173N allele in populations of European origin. *Annals of Human Genetics*, **2007**, 71, 755-61	2.2	12
38	Tandem Mass Spectrometry Quantitation of Lyso-Gb3 and Six Related Analogs in Plasma for Fabry Disease Patients. *Current Protocols in Human Genetics*, **2016**, 90, 17.23.1-17.23.9	3.2	12

37	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	6.2	11
36	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, 249-55	6.2	11
35	The clinical utility of total concentration of urinary globotriaosylsphingosine plus its analogues in the diagnosis of Fabry disease. *Clinica Chimica Acta*, **2020**, 500, 120-127	6.2	11
34	Neuroblastoma screening: the Canadian experience. *Medical and Pediatric Oncology*, **1989**, 17, 379-81		10
33	High-Risk Screening for Fabry Disease: Analysis by Tandem Mass Spectrometry of Globotriaosylceramide (Gb) in Urine Collected on Filter Paper. *Current Protocols in Human Genetics*, **2017**, 93, 17.26.1-17.26.12	3.2	8
32	FACTs Fabry gene therapy clinical trial: Two-year data. *Molecular Genetics and Metabolism*, **2019**, 126, S99	3.7	8
31	Improved ways to screen for patients with Fabry disease, involving optometry in a multidisciplinary approach **2012**, 74, 25		7
30	Neonatal Mass Urine Screening Approach for Early Detection of Mucopolysaccharidoses by UPLC-MS/MS. *Diagnostics*, **2019**, 9,	3.8	7
29	Assessment of urinary metabolite excretion after rat acute exposure to perfluorooctanoic acid and other peroxisomal proliferators. *Archives of Environmental Contamination and Toxicology*, **2015**, 68, 148-58	3.2	6
28	Effects of Orally Delivered Alpha-Galactosidase A on Gastrointestinal Symptoms in Patients With Fabry Disease. *Gastroenterology*, **2020**, 159, 1602-1604	13.3	6
27	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. *Analytica Chimica Acta*, **2018**, 1015, 35-49	6.6	6
26	Simple and rapid system for screening and identification of reducing sugars in urine. *Clinical Biochemistry*, **1978**, 11, 235-7	3.5	6
25	High-risk screening for Fabry disease in a Canadian cohort of chronic kidney disease patients. *Clinica Chimica Acta*, **2020**, 501, 234-240	6.2	6
24	Assessment of plasma lyso-Gb for clinical monitoring of treatment response in migalastat-treated patients with Fabry disease. *Genetics in Medicine*, **2021**, 23, 192-201	8.1	6
23	Fabry Disease Biomarkers: Analysis of Urinary Lyso-Gb3 and Seven Related Analogs Using Tandem Mass Spectrometry. *Current Protocols in Human Genetics*, **2016**, 90, 17.22.1-17.22.12	3.2	5
22	Biobanking primer: down to basics. *Science*, **2007**, 316, 830	33.3	5
21	Glycosphingolipid storage in Fabry mice extends beyond globotriaosylceramide and is affected by ABCB1 depletion. *Future Science OA*, **2016**, 2, FSO147	2.7	5
20	Distribution of heparan sulfate and dermatan sulfate in mucopolysaccharidosis type II mouse tissues pre- and post-enzyme-replacement therapy determined by UPLC-MS/MS. *Bioanalysis*, **2019**, 11, 727-740	2.1	4

19	Biomarkers in fabry disease. *Clinical Therapeutics*, **2009**, 31, S24-S25	3.5	4
18	Diet and medications giving positive ninhydrin reactions on TLC in a newborn urinary screening program. *Clinical Biochemistry*, **1980**, 13, 103-5	3.5	4
17	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. *Journal of Medical Genetics*, **2021**, 58, 692-700	5.8	4
16	Identification of a Reliable Biomarker Profile for the Diagnosis of Gaucher Disease Type 1 Patients Using a Mass Spectrometry-Based Metabolomic Approach. *International Journal of Molecular Sciences*, **2020**, 21,	6.3	3
15	UPLC-MS/MS analysis of keratan sulfate from urine samples collected on filter paper for monitoring & follow-up of Morquio A patients. *Bioanalysis*, **2018**, 10, 1181-1192	2.1	3
14	Glutaric Aciduria Type 3: Three Unrelated Canadian Cases, with Different Routes of Ascertainment. *JIMD Reports*, **2018**, 39, 89-96	1.9	3
13	Glycation of fetal hemoglobin reflects hyperglycemia exposure in utero. *Diabetes Care*, **2014**, 37, 2830-3	14.6	3
12	Determination of glycated and acetylated hemoglobins in cord blood by time-of-flight mass spectrometry. *Analytical Chemistry*, **2011**, 83, 5245-52	7.8	3
11	Altered immune phenotypes in subjects with Fabry disease and responses to switching from agalsidase alfa to agalsidase beta. *American Journal of Translational Research (discontinued)*, **2019**, 11, 1683-1696	3	3
10	Assessing the role of glycosphingolipids in the phenotype severity of Fabry disease mouse model. *Journal of Lipid Research*, **2020**, 61, 1410-1423	6.3	3
9	ATP-binding cassette transporters mediate differential biosynthesis of glycosphingolipid species. *Journal of Lipid Research*, **2021**, 100128	6.3	3
8	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,	2.6	2
7	Maternal inhaled fluticasone propionate intake during pregnancy is detected in neonatal cord blood. *Bioanalysis*, **2016**, 8, 1441-1450	2.1	2
6	Mass spectrometry analysis of urinary methylmalonic acid to screen for metabolic vitamin B deficiency in older adults. *Bioanalysis*, **2020**, 12, 693-705	2.1	2
5	Therapeutic challenges in two adolescent male patients with Fabry disease and high antibody titres. *Molecular Genetics and Metabolism Reports*, **2020**, 24, 100618	1.8	1
4	Combined malonic and methylmalonic aciduria due to ACSF3 mutations: Benign clinical course in an unselected cohort **2019**, 42, 107		1
3	High-Risk Screening of Fabry Disease: Analysis of Fifteen Urinary Methylated and Non-Methylated Gb Isoforms Using Tandem Mass Spectrometry. *Current Protocols in Human Genetics*, **2016**, 91, 17.24.1-17.24.11	2.2	0
2	Methylmalonic acid analysis using urine filter paper samples to screen for metabolic vitamin B deficiency in older adults.. *Bioanalysis*, **2022**, 14, 615-626	2.1	0

1	Mass spectrometry analysis of metals, other elements and lipids in urine samples of Fabry disease patients. *International Journal of Mass Spectrometry*, **2011**, 307, 163-173	1.9