OMICS International
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Christiane Auray-Blais et al., J Chromatogr Sep Tech 2015, 6:4
http://dx.doi.org/10.4172/2157-7064.S1.007

International Summit on Current Trends in Mass Spectrometry

July 13-15, 2015 New Orleans, USA

Mass spectrometry approaches for translational research: From biomarker discovery to clinical applications

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In order to increase the number of treatable disorders screened by the Mass Urinary Screening Program in the Province of Quebec, we developed and validated a tandem mass spectrometry multiplex method for creatine synthesis and transport disorders, Triple H syndrome and ornithine transcarbamylase deficiency. We targeted specific-related biomarkers such as creatine, guanidineacetate, uracil, orotic acid and creatinine using urine collected on filter paper. This method showed good linearity with a mean coefficient of regression at 0.9985 for all metabolites, as well as precision and accuracy for the intraday and interday assays at <15%. All abnormal patients were well discriminated from controls. We have shown that this methodology is feasible formass or high-risk screening with high sensitivity and reproducibility. Regarding lysosomal storage disorders, we have performed a time of flight mass spectrometry metabolomic study for biomarker discovery for Fabry disease, a multisystemic, X-linked complex disorder. We detected novel analogues of a glycosphingolipid, globotriaosylsphingosine (lyso-Gb_3), which were found to be increased in Fabry patients presenting the cardiac variant mutation of the disease. Robust tandem mass spectrometry methods were devised and validated for both urine and plasma. We have also devised a UPLC-MS/MS method for methylated and non-methylated forms of globoriaosylceramide (Gb_3), another biomarker detected in Fabry disease patients. These latter methods allowed the quantification of biomarkers in biological fluids of affected Fabrypatients and controls, and are aimed at the evaluation of correlations to determine disease-severity and progression.

Biography

ChristianeAuray-Blais is the Director of the Quebec Mass Neonatal Urinary Screening Program for hereditary metabolic disorders. She holds a PhD in Radiobiology from the Université de Sherbrooke and Postdoctoral studies from Duke University Medical Center, NC. She has a Master's degree in Health Law from the Faculty of Law at the Université de Sherbrooke. She is the author of 200 publications, abstracts and articles. She is a Professor in the Medical Genetics Division in the Pediatrics department at the Faculty of Medicine and Health Sciences in Sherbrooke. She is the Scientific Director for the Waters-CHUS Expertise Centre in Clinical Mass Spectrometry.

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