

Development and Implementation of Amino Acid Quantitation by Liquid Chromatography-Tandem Mass Spectrometry at BC Children's Hospital

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Amino acid disorders are a class of inherited metabolic disorders which can be attributed to an enzyme deficiency in metabolic pathways, resulting in the toxic accumulation of some substrates and the deficiency of others. Quantitative analysis of amino acids in biological fluids is utilized in the diagnosis of these inborn errors of metabolism. The most widely used method for amino acid analysis is ion-exchange chromatography coupled with post-column ninhydrin derivitization.¹ This technique was utilized by our laboratory, but the combination of low throughput (maximum of eight samples a day) and increasing sample numbers (5% per year) led to a backlog and unacceptable turn-around-times. Thus began our endeavour to develop a high throughput liquid chromatography-tandem mass spectrometry (LC-MS/MS) method for amino acid quantitation.

In recent years, methods utilizing LC-MS/MS for amino acid analysis have been published.^{2, 3, 4, 5} We referred to the method by Waterval and colleagues⁵ as a guideline and made significant changes to the sample preparation and chromatography, with minor modifications to the mass spectrometry conditions. The method was validated and, as of Spring 2014, we implemented LC-MS/MS for plasma amino acid analysis. Urine and cerebrospinal fluid amino acid analyses are anticipated to be implemented in Spring 2015. In this presentation, we will compare our current platform with our previous methodology, with respect to method performance, testing capacity, turn-around-times, and laboratory workflows.

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