Newborn Screening, Inborn Errors of Metabolism

AB104. Evaluation of a new non-derivatized MS/MS kit in newborn screening program

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Background: The NeoBase 2 newborn screening kit uses a non-derivatized tandem mass spectrometry (MS/MS) method to measure blood levels of amino acids, free carnitines, acylcarnitines, lysophospholipids, and purine nucleosides in newborns. The abnormal levels of these markers may be associated with inborn errors of metabolism (IEM). The most commonly used method is time-consuming and utilizing hazardous reagents such HCl-butanol. We evaluated a new commercial kit without harmful derivatizing agents and saving sample preparation time.

Methods: The precision and clinical evaluation were performed by two levels of quality controls, proficiency testing samples from Centers for Disease Control and Prevention (CDC, Atlanta, USA) and dried blood spots from 7,545 de-identified newborns.

Results: The precision among these analytes were 5.8±13.6% (CV) and cutoff values were determined with normal neonates and confirmed retrospective specimens. Sample preparation time can be reduced significantly to 60 minutes. To date, we have screened 28,644 neonates and identified 7 cases of IEM; 1 for 3-methylcrotonyl-CoA carboxylase deficiency, 2 for maternal primary carnitine deficiency, 1 for 2-methylbutyryl-CoA dehydrogenase deficiency, 1 for methylmalonic acidemia, 1 for phenylketonuria, and 1 for citrullinemia type II.

Conclusions: The non-derivatized MS/MS assay was demonstrated to be accurate in the detection of newborns with IEM and without the risk of the exposure to highly toxic reagents and requirement of additional equipment for toxic fume evacuation. This new kit could incorporate new markers and enhanced functionality.

Keywords: Non-derivatized MS/MS; newborn screening; inborn errors of metabolism (IEM)

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