Newborn Screening, Inborn Errors of Metabolism

AB102. A pilot newborn screening program for X-linked adrenoleukodystrophy

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Background: X-linked adrenoleukodystrophy (X-ALD) is a rare inherited metabolic disorder and is caused by mutations in the *ABCD1* gene encoding a peroxisomal ABC transporter ALDP. X-ALD can show a variety of phenotypic manifestations from severe childhood cerebral ALD to adult-onset adrenomyeloneuropathy. Treatments for childhood cerebral ALD are available: Lorenzo's oil and stem cell transplantation. Better outcomes are associated with early treatment and a need of newborn screening program has been proposed. Recent studies have showed that C26:0-lysophosphatidylcholine (C26:0-LPC) can be a sensitive biomarker for X-ALD before the onset of manifestations. To investigate the effect, we have conducted a pilot newborn screening program in Taiwan.

Methods: C26:0-LPC levels were measured in dried-blood spots (DBS) after 48 hours at birth. First tier screening is performed using MS/MS and 2nd tier screening is accomplished by LC-MS/MS with a first-tier result of >0.3 μM. The recall DBS were obtained with a 2nd tier result of >0.4 μM. If positive of a recall DBS, further confirmation tests including physical examination and other methodology for ALD confirmation will be provided. To monitor the performance of X-ALD newborn screening program, proficiency testing from Centers for Disease Control and Prevention (CDC, Atlanta, USA) was enrolled.

Results: From Nov 1st, 2016 to May 31st, 2017, 38,058 newborns have been screened for X-ALD. One newborn was recalled and had normal result. In DBS from 38,058 newborns, C26:0-LPC were 0.184±0.051 μM by MS/MS, while C26:0-LPC was 1.056 μM in one retrieved newborn DBS of X-ALD patient.

Conclusions: Preliminary results showed that C26:0-LPC can be detected in DBS. Levels of C26:0-LPC showed significant elevation in DBS of X-ALD patient compared to controls.

Keywords: X-linked adrenoleukodystrophy (X-ALD); Newborn screening; C26:0-lysophosphatidylcholine

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