Newborn Screening, Inborn Errors of Metabolism

# AB133. Expanded newborn screening program in Thailand

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**Background:** A pilot expanded newborn screening (NBS) program to detect inborn errors of metabolism (IEM) using tandem mass spectrometry (TMS) began at Siriraj Hospital, Bangkok, Thailand in May, 2014, with funding support from National Health Security Office (NSHO).

**Methods:** During the study period between May 2014 to March 2017. Dried blood spots of 99,234 newborns from 15 public hospitals in Bangkok were screened for amino acids, organic acids and fatty acid metabolism by tandem mass spectrometry (TMS). Dried blood spot (DBS) samples were taken on day 3 from neonates for newborn screening. Samples were analyzed for amino acid and acylcarnitine profiles using tandem mass spectrometry. Samples with abnormal results were repeated and the babies were recalled to confirm the diagnosis with plasma amino acid, plasma acylcarnitine, and urine organic acids.

**Results:** A total of 15 cases were identified and confirmed to have inborn errors of metabolism: 1 case with citrullinemia, 1 case with maple syrup urine diseases (MSUD), 1 case with methylmalonic acidemia (MMA), 5 cases with primary carnitine deficiency, 6 cases with maternal primary carnitine deficiency and 1 case with maternal glutaric aciduria type I.

**Conclusions:** Expanded NBS at Siriraj hospital could early detect many babies born in Bangkok region, and lead to early treatment and prevention of death or disability in these babies. Primary carnitine deficiency is the most common condition detected by expanded NBS (including many asymptomatic maternal cases and asymptomatic siblings). Genotyping study is carried on the identified cases.

**Keywords:** Expanded newborn screening (NBS); Thailand

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