Newborn Screening, Inborn Errors of Metabolism

AB056. Biochemical and molecular investigation of patients with methylmalonic acidemia in Thailand

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Background: Methylmalonic acidemia (MMA) is an autosomal recessively inherited disorder characterized by accumulation of methylmalonyl-CoA and methylmalonic acid in body fluids. MMA results from a functional defect in the methylmalonyl-CoA mutase (MCM) due to either the defect of *MUT* gene encoding MCM enzyme or of *MMAA* and *MMAB* genes which encode proteins involving the synthesis of adenosylcobalamin (AdoCbl), a coenzyme of MCM. MMA is a rare disease and usually cannot be detected by standard screening tests available in hospitals. We, therefore, have established a biochemical assay and mutational detection protocols of MMA since 1997.

Methods: Herein, we review the biochemical and mutational analysis of 19 Thai patients with isolated MMA

Results: There were 10 *MUT* patients, 2 *MMAA* (cblA) patients and 7 *MMAB* (cblB) mutations. We found that patients with *MUT* defective subtypes had none or a very little MCM activity and protein expression level while patients carrying the AdoCbl gene defects had a high MCM activity and protein expression levels in comparison to those in their parents and normal controls. From 19 mutations discovered, 13 (7 in the *MUT* gene, 1 in the *MMAA* gene, and 5 in the *MMAB* gene) were novel mutations firstly found in Thailand.

Conclusions: This finding provides an expanded mutation spectrum of MMA deficiency in global and Thailand. Early detections of MMA will help pediatricians to treat MMA patients properly and in time and provide counselling to the family members.

Keywords: Methylmalonic acidemia (MMA); methylmalonyl-CoA mutase (MCM); *MUT*; *MMAA*; *MMAB*

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