Newborn Screening, Inborn Errors of Metabolism

AB038. The biochemical and genetic analyses of biotinidase deficiency in the population of Taiwan

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Background: Biotinidase deficiency (BD) is an autosomal recessively inherited disorder of biotin recycling that is associated with neurologic and cutaneous presentations if untreated. Newborn screening for BD can benefit patients especially to those with profound BD (below 10% of mean normal activity). However, the incidence of BD in Taiwan is unknown.

Methods: Dried blood samples were taken from newborns at the age of 2–3 days as other newborn screening items. The biotin activity in dry blood spots was determined using a quantitative fluorescence assay. Newborns who were deficient in biotinidase activity (≤30% of normal mean activity) were referred for *BTD* mutation analysis.

Results: From November 2015 to May 2017, we have screened 94,354 newborns. Nine (0.01%) newborns failed in the first screen and a 2nd dried blood spot was requested. The population mean activity was 160.1 nmol/min/dL [standard deviation (SD) 39.93]. Finally, four newborns were requested to visit our hospital due to BD. Three of them presented each with 2 pathologic *BTD* mutations in trans, confirming the diagnosis of BD. Half of the mutant alleles were c.1250_1251TC>AG (p.V417E). However, all of them were classified as partial BD, and they have biotin supplement only when illness. The incidence of BD in this population was 1 in 31,451 [95% confidence interval (CI), 1 in 10,697 to 92,480].

Conclusions: The incidence of BD is 1 in 31,451, but all belong to partial BD only. The true incidence of profound BD is less than 1 in 94,354 newborns.

Keywords: Biotinidase deficiency (BD); *BTD* mutations; population

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