Newborn Screening, Inborn Errors of Metabolism

AB049. Diagnosis and treatment of phenylketonuria in Taiwan- experience from a national newborn screening confirmatory center

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Background: Newborn screening for phenylketonuria has been started in Taiwan since 1985. Until December 2016, a total of >3 million newborns had been screened by our team.

Methods: Data from newborn screening, follow up test, treatment and clinical outcome were reviewed. Mutation analysis was performed as indicated.

Results: The incidence of hyperphenylalaninemia patients who need treatment is around 1/58,000 in Taiwan. The most common mutations of *PAH* gene are R241C and R408Q which accounts for 23.2% and 12.0% of the mutant alleles, respectively. Both mutations have been suggested to be a mild and being BH4 responsive. Under the therapy of PKU special formula or BH4, most of our PAH PKU patients reach a normal IQ. The average IQ score of them is 98. The incidence of 6-pyruvoyl-tetrahydroterin synthase (PTPS) deficiency is around 1/116,000. N52S (52%) and P87S (30%) were the two most common mutations in our population for this disease. All patients received tetrahydrobiopterin replacement in a daily dosage between approximately 2 and 4 mg/kg. The dosages of levodopa replacement were 10–15 mg/kg/d, which is considerably higher than the typically recommended dosages of less than 7 mg/kg/d for patients aged <2 years and 8–10 mg/kg/d for patients aged ≥2 years . Replacement with 5-hydroxytryptophan varied widely among patients. The mean (SD) IQ score of our PTPS-deficient patients was 96.7 (9.7; range 86–119), which was considerably higher than previous reports of other populations of PTPS-deficient patients.

Conclusions: Local data of each country is essential for designing treatment strategy best suit for specific population.

Keywords: Phenylketonuria; PAH; 6-pyruvoyl-tetrahydroterin synthase (PTPS); tetrahydrobiopterin; newborn screening

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