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TETRAHYDROBIOPTERIN DEFICIENT PHENYLKETONURIA DETECTED BY NEONATAL SCREENING IN TAIWAN. K.J. Hsiao, S.H. Chiang, T.T. Liu, P.C. Chiu[1] and K.D. Wuu[2]. Depts. of Medical Research and Pediatrics[1], Veterans General Hospital, and Institute of Genetics[2], National Yang-Ming Medical College; Taipei, Taiwan 11217, Republic of China.

Phenylketonuria (PKU) caused by tetrahydrobiopterin (BH4) deficiency is reported to be a rare variant form of PKU. The incidence of BH4 deficiency among hyperphenylalaninemic babies was estimated to be 1.5 - 2% in the Caucasian population. The program for neonatal screening of congenital metabolic diseases in Taiwan was initiated in 1984. From 1984.1 to 1989.3, 222,800 newborns were screened for PKU by analyzing the phenylalanine in dried blood spots collected on filter paper. Six PKU babies (1/38,000) were detected. Among them, two cases were found with defective synthesis of BH4 and one case was caused by dihydropteridine reductase (DHPR) deficiency. All of the PKU cases detected by our neonatal screening program were confirmatively diagnosed by BH4 oral loading test, urinary pterins analysis, and blood DHPR determination, and treated accordingly within 37 days after birth. The results indicated that the incidence of BH4 deficient PKU in Chinese is much higher than that in Caucasian and Japanese. Therefore, the differential diagnosis between different forms of PKU is very important for the Chinese PKU patients.

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PRENATAL DIAGNOSIS OF 6-PYRUVOYL TETRAHYDROPTERIN SYNTHETASE DEFICIENCY IN EAST ASIA. H. Shintaku[1], M. Fujioka[1], G. Isshiki[1], Y. Sawada[2], T. Ohura[3], S. Yamaoka[3], T. Ueda[4], K. Hatanaka[4], M. Suzuki[4], R. Murata[4], M. Matsumoto[4], K.J. Hsiao[5], T.T. Liu[5], R.G. Chen[6], [1]Osaka City University Medical School, [2]Juso Citizens' Hospital, [3]Osaka Municipal Rehabilitation Center for the Disabled, [4]Osaka City Perinatal Center, Osaka, Japan, [5]Veterans General Hospital, Taipei, Taiwan 11217, Republic of China, [6]Shanghai Institute for Pediatric Research, Shanghai, China.

Prenatal diagnosis of 6-pyruvoyl tetrahydropterin synthetase (PTPS) deficiency was performed a total of four times in three families by pteridine analysis of amniotic fluid. Twice (once in Japan and once in China) PTPS activity in erythrocytes of the fetus was also measured. Two Japanese siblings and one Chinese fetus in Shanghai were diagnosed prenatally as being heterozygotes for PTPS deficiency. Another Chinese fetus in Taipei was found to be normal. These diagnoses were comfirmed after birth. Niederwieser et al. reported that the analysis of amniotic fluid pterins is enough for prenatal diagnosis of PTPS deficiency. Our study shows that even heterozygotes for PTPS deficiency can be diagnosed prenatally by analysis of pterins in the amniotic fluid.

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