Proc 1989 Annual Meeting of the Society for Inherited Metabolic Disorders, Florida, 1989, p.16

Abstract #20

HIGH INCIDENCE OF TETRAHYDROBIOPTERIN DEFICIENT PHENYLKETONURIA IN THE CHINESE POPULATION OF 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], 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. Since 1983, from studying mentally retarded children and pediatric patients in Taiwan, we have detected 16 PKU cases from 12 unrelated families. But half of them, 8 cases from 6 families, were caused by defective synthesis of BH4. From 1984.1 to 1988.12, 202,099 newborns in Taiwan were screened for PKU by analyzing the phenylalanine in dried blood spots collected on filter paper. Five PKU babies (1/40,000) were found. Among them, two cases were identified as having BH4 synthesis deficiency. All of the PKU cases detected by our neonatal screening program were confirmatively diagnosed and treated accordingly within 37 days after birth. The results show that about half of the PKU cases were caused by the BH4 synthesis deficiency in the Chinese population of Taiwan. Such a high incidence of BH4 deficient PKU found in the Chinese indicates that the differential diagnosis between the classical and BH4 deficient forms of PKU is much more important for the Chinese than for the Caucasian.

(99)