P 029 Proc 27th Annual Symposium of the Society for the Study of Inborn Errors of Metabolism, Munich, 1989;p.029.

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Title : **Phenylketonuria caused by tetrahydrobiopterin synthesis defficiency in Taiwan**

In Caucasian population, the cases of tetrahydrobiopterin (BH4) deficient phenylketonuria (PKU) count only 1.5-2.0% of total cases of PKU. From Jan. 1984 to Dec. 1988, 202,106 newborns and 13,375 high risk patients were screened for congenital metabolic diseases in Taiwan, 24 cases of PKU including 11 cases of BH4 synthesis deficiency were found. The BH4 synthesis deficient PKU was diagnosed by BH4 oral loading test, urinary pterins analysis and dihydropteridine reductase (DHPR) determination. Two cases were found to be mild form and did not receive BH4 nor neurotransmitters treatment. All the other 9 cases received treatment with BH4 (0.6-3.6 mg/kg/D), L-dopa (7.5-10 mg/kg/D), Carbidopa (0.75-1.0 mg/kg/D) and 5-hydroxytryptophan (1-3 mg/kg/D). One case died of aspiration pneumonia and another refused to continue after several months of therapy. The psychomotor development of the two cases (4 years; 6 months) detected from neonatal screening are normal. Five cases from high risk group have great improvement in their neurological symptoms, behavior problems, psychomotor development and IQ. In conclusion, the incidence of BH4 deficient PKU in Chinese is much higher than that in Caucasian. Early detection and treatment with BH4 and neurotransmitters may effectively prevent neurological sequelae in the PKU caused by BH4 synthesis deficiency.

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