A SURVEY FOR THE INCIDENCE OF PHENYLKETONURIA IN GUANGDONG, CHINA

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Abstract. A multicenter cooperative investigated the incidence of Phenylketonuria (PKU) in the central, southern and western areas of Guangdong province and its surrounding districts. Tests to measure phenylalanine (Phe) on dried blood spots on filter paper cards used BIA and the fluorescence assay. Four hundred sixty-one thousand eight hundred five (461,805) newborns were screened and 14 cases of persistent hyperphenylalaninemia (PHPA) were detected. The incidence of PHPA was 1/ 33,000, including 1/77,000 for classical PKU; 1 in 66,000 for hyperphenylalaninemia (HPA) and 1 in 461,805 for BH4 deficiency. A high variation in PKU incidence in the different districts was observed. The differences may be attributed to the variations in gene frequency of PKU in the different districts and to bioethical consideration especially as regards reproduction in different areas.

INTRODUCTION

Phenylketonuria (PKU) is one of the most severe inborn errors of metabolism. Worldwide, it is now screened for and treated in the early neonatal stages. The incidence of PKU should be investigated in every district. It is very important for the government to implement a newborn screening program.

METHODS AND RESULTS

Four hundred sixty-one thousand eight hundred five newborns from the six cities of Guangdong province, namely Guangzhou, Shengzhen, Zhanjiang, Fushan, Zhongshan and Zhuhai, were screened.The results of newborn screening were collected through multiple centers. Dried blood spot on S&S903 filter paper cards were collected from the newborn's heel three days after birth and after six feedings. The BIA or the fluorescence assay was used to determine the phenylalanine (phe) concentration of the dried blood sample. The dried blood sample collection was repeated when the phe level was >120 μmol/l (2mg%). Persistent hyperphenylalaninemia (PHPA) was further evaluated together with urinary pterins analysis and a blood dihydropteridine reductase (DHPR) determination was done for differential diagnosis. Among 461,805 newborns, there were 14 babies with PHPA, 6 babies with classical PKU (Phe 1200 μmol/ l), 7 babies with hyperphenylalaninemia (HPA, Phe 120-1200 μmol/l) and 1 baby with BH4 deficiency. The incidence of PHPA was 1 in 33,000; classical PKU, 1 in 77,000 and HPA, 1 in 66,000.

DISCUSSION AND CONCLUSION

PKU should be screened and treated during the early neonatal stage because of the consequent severe brain damage to the baby if it is left untreated. The incidence of PHPA in the different districts of Guangdong, China was almost equal to that in Taiwan (1 in 31,000), and lower than that in northern China (1 in 7,000) and the average level of China (1 in 15,000). It was different from other countries or districts (1 in 70,000 in Japan); 1 in 8,250 in EU and 1 in 15,000 in the USA. A high incidence variation of PKU in the different districts was observed. These differences may be attributable to the variations of gene frequency of PKU in the different districts and to bioethical considerations, with regards to reproduction in different countries. The same standards for diagnosis and differential diagnosis of PKU should be used so that the different incidences can be compared.

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