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Mutational spectrum of phenylketonuria in Jiangsu province: Genotype–phenotype correlations and genotype-based predictions of BH4-responsiveness

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Phenylketonuria (PKU), an inherited congenital metabolic disease in newborns, is caused by variants in the phenylalanine hydroxylase (*PAH*) gene. A comprehensive analysis for phenylketonuria (PKU) variants can elucidate the mutational spectrum in patients from Jiangsu province, China. A total of 31 unrelated patients with PKU and their parents in Jiangsu, corresponding to 62 independent alleles, were investigated. Patients with BH4-cofactor deficiency were excluded. The patients were mainly from Suqian city in north of Jiangsu and Wuxi city in south of Jiangsu. These samples were collected from 2005-2012 via newborn screening program. All the patients studied were diagnosed as hyperphenylalaninemia at the Wuxi Maternal and Child Health Hospital, where phenylalanine levels on dried blood spots were quantified using the fluorescence test and tandem mass spectrometry. We systematically investigated 13 exons and their flanking introns of the phenylalanine hydroxylase (*PAH*) gene in 31 unrelated patients and their parents using the next-generation sequencing (NGS) technology. A total of 33 different variants were identified in 58 of 62 mutant *PAH* alleles. The variants c.721C>T, c.1068C>A, c.611A>G, c.1197A>T, c.728G>A, c.331C>T, c.442-1G>A were the prevalent variants with a relative frequency of 5% or more. One novel variant was identified in this study: c.699C>G. We studied genotype–phenotype correlation using the Guldberg AV system and revealed consistency rate of 38% (8/21) among the 21 predicted phenotypes. Genotype-based prediction of BH4-responsiveness was also evaluated and 14 patients (45.2%) were predicted to be BH4-responsive. The spectrum of *PAH* variants were presented in Jiangsu province and genotype-based prediction of BH4-responsiveness provides a useful tool in planning and management of future clinical trials using BH4.

Biography

Xin-Ye Jiang has completed her Bachelor's degree at Nanjing Medical University in 2003 and Master's degree in Pediatrics at Suzhou University in 2010. She is the Chief Physician of Child Hygiene in Wuxi Maternity and Child Care Hospital affiliated to Nanjing Medical University. She is currently serving as a Member of Child Health Professional Committee of Jiangsu Preventive Medicine Association and a Member of Child Health Group of Pediatrics Branch of Jiangsu Medical Association. She is mainly engaged in children's health clinical work and scientific research. For the last 5 years, she has headed and participated in six national and provincial research projects and has successively published 17 papers by the first author or the communication author, including three SCI papers and two Chinese papers.

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