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论文

中国汉族人群*S*-美芬妥英4'-羟化酶的表型与基因分析

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摘要：

目的：研究中国汉族人群S-美芬妥英4'-羟化代谢遗传多态性。方法：以美芬妥英为探针药物采用手性毛细管气相色谱法测定尿中*S*-/*R*-MP浓度比值， 对90名志愿者进行了表型分型测定，应用PCR技术对其中的26名志愿者进行了*S*-美芬妥英4'-羟化酶（CYP2C19）基因分析。结果：表型分析结果，11人属慢代谢者（PM），*S*/*R*比值0.95；基因分析结果，6人为野生型纯合子（wt/wt）；10人为杂合子（wt/m_1和wt/m_2），9人为CYP2C19m_1突变型纯合子（m_1/m_1），1人为两突变型杂合子（m_1/m_2）。结论：表型分析与基因分析结果显示了很好的相关性，本实验测得慢代谢者的频发率为12.2%，与文献报道相符。

关键词： 美芬妥英 *S*-美芬妥英4'-羟化酶(CYP2C19) 表型测定 基因分析

THE PHENOTYPE AND GENOTYPE ANALYSIS OF *S*-MEPHENYTOIN HYDROXYLASE (CYP2C19) IN CHINESE SUBJECTS

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Abstract:

AIM: To assess the phenotype and genotype pattern of *S*-mephenytoin 4'-hydroxylation in Chinese population. METHODS: The phenotypes of ninety healthy subjects were analyzed with *S*/*R* mephenytoin ratio in urine after an oral dose of 100 mg racemic mephenytoin by chiral GC-FID method. The genotypes of twenty-six among the 90 subjects were analyzed with identifying the wild-type(wt) gene and two mutations, CYP2C19m1 and CYP2C19m2 by PCR method. RESULTS: Of the 90 subjects eleven were identified as poor metabolizers with the *S*/*R* ratio of 0.95. Among the 26 genotyped subjects six were homozygous for wild-type (wt/wt); nine were homozygous for CYP2C19m_1(m_1/m_1); seven were heterozygous for the CYP2C19m1(wt/m_1); three were heterozygous for the CYP2C19m_2(wt/m_2); one was the heterozygous for the two defects (m_1/m_2). CONCLUSION: The result of CYP2C19 genotype analysis was in agreement with that of phenotype analysis. The frequency of PM by phenotype analysis was 12.2%.

Keywords: *S*-mephenytoin hydroxylase(CYP2C19) phenotype analysis genotype analysis mephenytoin

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