

临床医学

烟台市汉族女性MTHFR和MTRR基因多态性的分布特征

严倩1, 鲁衍强2, 李瑛2, 石春红1, 姜凌1, 于传亭1

1.烟台市烟台山医院妇产科, 山东 烟台 264000;
2.上海靶向分子医学研究所, 上海 200433

摘要:

目的 分析山东省烟台市汉族女性亚甲基四氢叶酸还原酶(MTHFR)C677T、A1298C及甲硫氨酸合成酶还原酶(MTRR)A66G单核苷酸多态性的分布特征。方法 以烟台市2 670例汉族女性为研究对象, 采集口腔黏膜上皮细胞, 提取基因组DNA, 通过荧光定量PCR方法进行MTHFR和MTRR基因多态性检测。统计分析该地区基因多态性的分布特征, 并与其他地区已报道的数据进行比较。结果 烟台市汉族女性的MTHFR C677T位点CC、CT、TT基因型的频率分别是18.6%、49.2%、32.2%, 其中TT基因型的分布频率低于潍坊市、淄博市、郑州市, 高于镇江市、武汉市、德阳市、昆明市、广州市和琼海市, 且与以上城市的基因多态性分布存在统计学差异(P<0.05)。MTHFR A1298C位点AA、AC、CC基因型的频率分别为72.5%、25.7%、1.8%, 其中CC基因型的分布频率与潍坊市、临沂市、镇江市、武汉市、德阳市、昆明市、广州市和琼海市间的差异均有统计学意义(P<0.05)。MTRR A66G位点AA、AG、GG基因型的频率分别为54.6%、38.1%、7.2%, 其中GG基因型的分布频率仅与潍坊市、淄博市和琼海市间的差异有统计学意义(P<0.05)。结论 烟台市汉族女性MTHFR和MTRR基因多态性分布情况不同于其他地区, 具有地域特异性。

关键词: 亚甲基四氢叶酸还原酶; 甲硫氨酸合成酶还原酶; 单核苷酸多态性

Polymorphisms of MTHFR and MTRR in women of Chinese Han population in Yantai city

YAN Qian1, LU Yan-qiang2, LI Ying2, SHI Chun-hong1, JIANG Ling1, YU Chuan-ting1

1. Department of Gynecology and Obstetrics, Mount Yantai Hospital, Yantai 264000, Shandong, China;
2. Shanghai Institute of Targeted Therapy and Molecular Medicine, Shanghai 200433, China

Abstract:

Objective To explore gene polymorphisms of 5,10-methylenetetrahydrofolate reductase (MTHFR) and 5-methyltetrahydrofolate-homocysteine methyltransferase reductase (MTRR) in women from Chinese Han population in Yantai city. Methods A total of 2 670 Han women were recruited in the study. Their oral epithelial cells were collected, followed by the extraction of genome DNA, in order to detect gene polymorphisms of MTHFR and MTRR using fluorescence quantitative PCR method. The Results were compared with those in other cities of China. Results The frequency of MTHFR 677CC, 677CT and 677TT of Han women in Yantai city were 18.6%, 49.2% and 32.2%, respectively. The frequency of MTHFR 677TT among women in Yantai city was obviously different from Weifang, Zibo, Zhengzhou, Zhenjiang, Wuhan, Deyang, Kunming, Guangzhou and Qionghai (P<0.05). The frequency of MTHFR 1298AA, 1298AC and 1298CC of Han women in Yantai city were 72.5%, 25.7% and 1.8%, respectively. The frequency of MTHFR 1298CC in Yantai was different from Weifang, Zibo, Linyi, Zhenjiang, Wuhan, Deyang, Kunming, Guangzhou and Qionghai (P<0.05). The frequency of MTRR 66AA, 66AG and 66GG of Han women in Yantai city were 54.6%, 38.1% and 7.2%, respectively. The frequency of MTRR 66GG in Yantai was only different from Zibo, Linyi and Qionghai(P<0.05). Conclusion The MTHFR, MTRR polymorphism distribution of Han women in Yantai city is different from other regions, showing region-specific characters.

Keywords: 5,10-methylenetetrahydrofolate reductase; 5-methyltetrahydrofolate-homocysteine methyltransferase reductase; Single nucleotide polymorphism

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通讯作者: 鲁衍强, E-mail: luyanqiang@genechina.com

作者简介:

作者Email:

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