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广西壮族人群骨桥蛋白基因的单核苷酸多态性研究*

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[摘要] **目的** 分析骨桥蛋白基因单核苷酸多态性(SNP)位点 rs11728697 和 rs9138 的基因型和等位基因型在广西壮族人群中的分布频率,对比其不同国家或地区人群间的分布差异。**方法** 利用 SNaPshot SNP 分型技术检测 150 例广西壮族骨桥蛋白基因 SNP 位点 rs11728697 和 rs9138 的基因型,对比国际人类基因组单倍型图谱计划(HapMap)上 4 个人群(欧洲人群、非洲人群、日本人群和中国北京人群)的 SNP 分型数据,分析 5 个人群的骨桥蛋白基因 SNP 位点 rs11728697 和 rs9138 的基因型和等位基因频率差异。**结果** 在广西壮族人群中,骨桥蛋白基因 rs11728697 位点的 CC 基因型最常见,约为 42.7%;C 等位基因的频率最高,约为 62.7%。rs9138 位点的 CA 基因型最常见,约为 51.3%;C 等位基因的频率最高,约为 63.0%。骨桥蛋白基因型及等位基因频率男女组间比较差异无统计学意义($P>0.05$)。广西壮族人群骨桥蛋白基因 rs11728697 位点的基因型和等位基因频率与欧洲人群、日本人群和非洲人群的差异有统计学意义($P<0.05$),而与中国北京人群的差异无统计学意义($P>0.05$)。广西壮族人群骨桥蛋白基因 rs9138 位点的基因型和等位基因频率与欧洲人群和非洲人群的差异有统计学意义($P<0.05$),而与日本人群和中国北京人群的差异无统计学意义($P>0.05$)。**结论** 骨桥蛋白基因 SNP 位点 rs11728697 和 rs9138 基因型和等位基因在广西壮族人群中的分布频率与不同国家或地区人群相比存在差异,这种差异可能是导致某些疾病在不同国家或地区间发病率和临床表现存在差异的因素之一。

[关键词] 骨桥蛋白;多态性;单核苷酸;人群

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Single nucleotide polymorphisms research of osteopontin gene in Zhuang populations in Guangxi*

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[Abstract] **Objective** To investigate the genotype and allele frequencies of osteopontin gene single nucleotide polymorphisms (SNP) rs11728697 and rs9138 in Zhuang populations in Guangxi, and to compare the distribution of osteopontin polymorphisms among different races. **Methods** The osteopontin gene rs11728697 and rs9138 polymorphisms were detected by SNaPshot SNP genotyping technique in 150 Zhuang populations in Guangxi, the genotype and allele frequencies of osteopontin gene rs11728697 and rs9138 polymorphisms were analyzed in Zhuang populations compared with the other four populations (HapMap-CEU, HapMap-YRI, HapMap-JPT, HapMap-HCB) from HapMap database. **Results** The most common genotype and allele of osteopontin gene rs11728697 polymorphism in Zhuang populations in Guangxi were CC(42.7%) and C(62.7%), and the most common genotype and allele of osteopontin gene rs9138 polymorphism were CA(51.3%) and C(63.0%). There were no significant differences in the genotype and allele frequencies of osteopontin gene rs11728697 and rs9138 polymorphisms between male and female groups ($P>0.05$). The genotype and allele frequencies of osteopontin gene rs11728697 polymorphism were significantly different compared with HapMap-CEU, HapMap-JPT and HapMap-YRI($P<0.05$), but were not significantly different compared with HapMap-HCB ($P>0.05$). The genotype and allele frequencies of osteopontin gene rs9138 polymorphism were significantly different compared with HapMap-CEU and HapMap-YRI($P<0.05$), but had no significantly different compared with HapMap-JPT and HapMap-HCB($P>0.05$). **Conclusion** There are significant differences in the genotype and allele frequencies of osteopontin gene rs11728697 and rs9138 polymorphisms between Zhuang populations and other ethnic populations, and this variation might contribute for a variety of clinical manifestation and morbidity of some osteopontin related diseases.

[Key words] osteopontin; polymorphism, single nucleotide; population

骨桥蛋白是由一种具有多种生物学活性的细胞黏附分子,其广泛分布于多种组织和细胞中。目前研究认为骨桥蛋白在细胞黏附、迁移、细胞存活、血管生成和浸润转移中起重要作用,与肿瘤等疾病的发生、发展密切相关^[1-3]。近年来随着分子

生物学及分子流行病学的不断发展,新的研究发现,骨桥蛋白基因存在多种功能性多态性位点,且这些多态性位点也可能参与甲状腺乳头状癌、尿路结石和缺血性脑卒中等疾病的发展进程^[4-6]。因此,准确分析骨桥蛋白基因单核苷酸多态性(SNP)

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呈显著负相关,幸福感高的老人,其社会交往、心理承受能力、看待事情的态度、满意度都较高,产生孤独情绪较少。而幸福感低的老人,由于在社会交往过程中获得的支持较少,看待问题的角度比较偏激,心理承受能力差等原因,对孤独的感受比较强烈^[8]。老年人生活满意度与其抑郁情绪的发生也有一定的影响,抑郁情绪发生率较高的老年人生活满意度较低^[9]。本次调查也显示,孤独和抑郁量表得分越高的老年人,幸福度得分越低。部分老人存在较严重的抑郁症状和孤独感,这可能与其从原来重要工作岗位退休、空巢家庭、子女探望次数少、家庭关系紧张及老人身体健康状况不佳、生活自理能力下降、参与社会交往少等因素有关。

3.3 根据老人幸福度的影响因素制订个性化照护计划 养老机构照护团队根据老人入住前的幸福度评估结果,结合其医疗护理状况、日常生活自理能力、心理情绪状态和社会交往的等不同方面的需求制定个性化照护计划。

首先,帮助老人接受衰老是人生自然进程,逐步适应角色转变,找到新的生活目标减轻退休综合征。鼓励老人结识新朋友,重新找到共度晚年生活的伙伴和倾诉对象。其次,提供相应的辅助设施和工具,让老人参与完成各项日常活动,维护老人的尊严。同时,进行适当的康复训练,延缓自理能力丧失。最后,针对不良情绪照护团队、家属和老人共同寻找原因,及时给予心理支持和慰藉,必要时心理咨询和干预。照护过程中,对老人进行动态评估修订照护计划。

综上所述,了解老年人情绪状况及影响因素,结合其生理、心理健康和社会交往的特点,提供个性化照护服务,提升老人的幸福度,是养老服务管理中需要不断探索和研究的问题。

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