

研究报告

## MMP-13基因多态性与食管癌、贲门癌遗传易感性的关联研究

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### 摘要

采用病例-对照研究, 分析河北省磁、涉县食管鳞状细胞癌(ESCC)患者和贲门腺癌(GCA)患者与健康对照人群的基质金属蛋白酶-13(MMP-13)基因启动子区转录起始点上游77 bp处A/G多态等位基因和基因型频率分布的差异, 旨在探讨此基因多态性与ESCC、GCA遗传易感性的关系。采用聚合酶链反应-限制性片段长度多态性(PCR-RFLP)技术检测316名ESCC患者、243名GCA患者和609名健康对照的MMP-13 A-77G SNP的基因型。结果表明, MMP-13 SNP的基因型及等位基因型频率在两患者组和健康对照组之间, 其总体分布均无显著性差异( $P>0.05$ )。根据吸烟状况分层分析发现, 吸烟组中GCA患者组与对照组A/A、A/G、G/G基因型频率分别是27.1%、44.1%、28.8%和17.8%、60.5%、21.7%, 两组相比具有显著性差异( $\chi^2=9.01, P=0.01$ ), GCA患者组A/G基因型频率明显低于对照组, 与A/A基因型比较, A/G基因型可能减少吸烟个体GCA的发病风险( $OR=0.48, 95\% CI=0.28\sim 0.83$ )。认为MMP-13基因A-77G SNP可能与河北省磁、涉县人群的食管癌、贲门癌发病风险无关; 但A/G基因型对吸烟人群的GCA发病可能具有保护作用。

关键词 [食管鳞状细胞癌](#) [贲门腺癌](#) [MMP-13](#) [基因多态性](#) [肿瘤易感性](#)

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## The Association of MMP-13 Polymorphism with Susceptibility to Esophageal Squamous Cell Carcinoma and Gastric Cardiac Adenocarcinoma

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### Abstract

<P>Matrix metalloproteinase-13 (<EM>MMP-13</EM>) belongs to a family of enzymes that degrade all components of the extracellular matrix. Polymorphism in the promoter region of the <EM>MMP-13</EM> gene may modify its transcriptional activity and protein level. This study was designed to investigate the correlation of <EM>MMP-13</EM> A-77G single nucleotide polymorphism (SNP) with susceptibility to esophageal squamous cell carcinoma (ESCC) and gastric cardiac carcinoma (GCA) in a population from a high-incidence region of Hebei province. <EM>MMP-13</EM> promoter SNP (A-77G) was genotyped by poly-merase chain reaction-restriction fragment length polymorphism (PCR-RFLP) analysis in 316 ESCC patients, 243 GCA patients and 609 healthy controls. The overall genotype and allelotype distributions of <EM>MMP-13</EM> in ESCC and GCA patients were not significantly different from those in healthy controls (<EM>P</EM>>0.05). When stratified for smoking status, the <EM>A/A, A/G, G/G</EM> genotype frequencies of <EM>MMP-13</EM> in GCA patients and smoker controls group were 27.1%, 44.1%, 28.8% and 17.8%, 60.5%, 21.7% respectively. There was a significant difference between the two groups ( $\chi^2=9.01, P=0.01$ ). The <EM>A/G</EM> genotype frequency in GCA patients was significantly lower than that in the controls, when compared to the <EM>A/A</EM> genotype, suggesting that individuals with the <EM>A/G</EM> genotype may have reduced susceptibility to GCA in the smoker group (<EM>OR</EM>=0.48, 95% <EM>CI</EM>=0.28~0.83). However, the <EM>G/G</EM> genotype showed no significant influence on the risk of developing GCA. Thus, in the region of Hebei Province where there is a high incidence of GCA and ESCC, the <EM>MMP-13</EM> A-77G SNP may not be associated with cancer susceptibility. The A/G genotype may confer protection

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against GCA for smokers. <BR></P>

**Key words** [esophageal squamous cell carcinoma](#) [gastric cardiac adenocarcinoma](#) [MMP-13 polymorphism](#) [susceptibility](#).

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