

论著

E-Cadherin(CDH1)基因胚系突变与胃癌风险的研究

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摘要 背景与目的: 检测E_Cadherin (CDH1) 基因在家族性胃癌中的突变情况以探讨CDH1基因胚系突变在家族性胃癌中的作用。方法: 采用聚合酶链反应(Polymerase chain reaction, PCR)、变性高效液相色谱分析(Denaturing high_performance liquids chromatography, DHPLC)和直接测序法对CDH1基因进行全基因筛查, 检查80名胃癌患者的CDH1基因突变情况, 其中62例有家族史。同时检测80例正常人CDH1基因, 进行分析比较。结果: 2.5%(2/80)的胃癌患者检出CDH1基因错义突变Thr340Ala(杂合型), 在80例正常人中未检出这一突变; 第13外显子上游(内含子)第13位碱基检出多态位点IVS(13)_13T→C, 这一多态性存在于12.5%(10/80)的胃癌患者和15.0%(12/80)的正常对照, 两组差异无统计学意义; 23.8%(19/80)的胃癌患者中检出同义突变Asn751Asn, 相同的突变存在于8.8%(7/80)的正常对照。统计结果表明, 胃癌患者中CDH1基因 Asn751Asn检出率高于正常人群(P<0.05), 这种差异主要表现在高龄组胃癌患者(P<0.01)。结论: CDH1基因胚系突变在家族性胃癌中不是频发事件, 但多态位点Asn751Asn的存在可使携带者胃癌发病风险增高, 可能是部分癌高发家族癌发生的原因之一。

关键词 [家族性胃癌](#); [胚系突变](#); [CDH1基因](#)

Association of the Germline Mutations of E_Cadherin Gene (CDH1) in Gastric Cancer

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Abstract BACKGROUND & AIM: To detect the germline mutations in E_Cadherin gene (CDH1) and to investigate their possible role in familial gastric cancer. MATERIAL AND METHODS: With PCR, denaturing high_performance liquid chromatography (DHPLC) and direct sequencing, we screened all exons of the E_cadherin gene for mutations in 80 Chinese gastric cancer patients, 62 of whom had positive family history, we investigated 80 healthy individuals and conducted a case_control study to investigate whether such mutations affect the risk of developing gastric carcinoma. RESULTS: Two of 80 gastric cancer patients carried missense mutation Thr340Ala (heterozygosity). This variation was not found in the control population. We confirmed a T→C polymorphism site at position -13 in upstream of exon 13. The polymorphism was identified in 10 of 80 gastric cancer patients and 12 of 80 healthy individuals and the frequency of this polymorphism did not differ between the familial gastric cancer patients and controls. We found a germline E_cadherin nonsense mutation at codon 751 in exon 14 in 19 of 80 patients analyzed. Only 7 of 80 healthy individuals were Asn751Asn carriers, and significant difference existed between gastric cancer patients and control population, especially between the elder patients and controls. CONCLUSION: Though the germline mutations of CDH1 gene did not occur frequently in familial gastric cancer, CDH1 gene Asn751Asn single nucleotide polymorphism might increase the risk of gastric cancer in Chinese population.

Keywords [familial gastric cancer](#) [CDH1 gene](#) [germline mutation](#)

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