Analysis of the association between protein C gene single nucleotide polymorphism and pulmonary thromboembolism in Chinese population

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Body: Background: We investigated the role of protein C (PC) polymorphism in patients with PTE in order to find out the correlation between its polymorphism and the susceptibility of the Chinese population to develop PTE. Methods: Sixty three consecutive patients with PTE were enrolled as the investigated group and eighty six healthy people as the control group. C/T at the position of 2405 and A/G at the position of 2418 in the PC gene promoter region were detected through polymerase chain reaction-restriction fragment length polymorphism analysis. Results: (1) The results suggested that the genotype frequencies of the two SNPs when combined together were not significantly different between two group (P > 0.05). However, the allele frequency of the C2405T SNP was significantly different between the case and control group. The frequency of T allele in the PTE group was higher. (2) These results suggested that there were six different kinds of genotype distribution (TA - TA, TA - CA, TA - CG, CG - CG, CA - CG, CA - CA) and three different kinds of haplotype (TA, CG, CA). Conclusions: These results suggest that the two polymorphisms present in the control region of PC gene are associated with an increased susceptibility to PTE in the Chinese population. The 2405T allele may be a possible risk factor for the development of PTE while the C allele may probably be a protective factor of PTE. Moreover, the TA haplotype may also be associated with an increased risk for developing PTE.