# 中国人COL2A1基因座的扩增片段长度多态性

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收稿日期 修回日期 网络版发布日期 接受日期

摘要 用聚合酶链式反应(PCR)、高分辨聚丙烯酰胺凝胶水平电泳及银染技术对位于人类II型胶原基因终止密码下游非转录区1. 3kb处的可变数目串联重复序列(COL2A1 VNTR)进行了研究。制备了由人类不同基因型DNA混合而成的人类等位基因分型参考物,根据实验结果进行了命名。新命名法在一定范围内达到国际标准化。对120名中国汉族无关个体的分型结果表明,该群体样本含有由14个COL2A1等位基因构成的23种基因型,包括4种新的等位基因,基因频率分布与德国白人群体有显著性差异,说明该基因座不仅是重要的法庭血液遗传标记,而且是人类遗传学的有用标记。

关键词 <u>COL2A1基因座</u> 聚合酶链式反应 <u>PCR</u> 可变数目串联重复序列 <u>VNTR</u> 遗传多态性 <u>中国群体</u> 分类号

# Amplified Fragment Length Polymorphism of the VNTR Locus COL2A1 in Chinese Population

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#### Abstract

The amplifiable VNTR polymorphic system COL2A1 has been investigated in a Chinese Han population(n=120)by the polymerase chain reaction(PCR) and PAGE horizontal electrophoresis followed by silver stain. In order to accurately identify COL2A1 alleles, a number of human allele adders prepared by mixing DNAs extracted from different individuals of known COL2A1 genotypes were used. A total of 14 different alleles in 23 genotypes were observed in this Chinese Hanpopulation. Among them, four were new alleles disclosed in the present study. The results imply that COL2A1 locus may be served as a genetic marker in forensic haemogenetics as well as in anthropogenetics.

 Key words
 COL2A1 locus
 Polymerase chain reaction (PCR)
 Variable number tandem repeat

 (VNTR)
 Polymorphism
 Chinese Han population

DOI:

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