遗传学报 1991 18 (1): 6-11 ISSN: 0379-4172 CN: 11-5450/R

中国人LDR152/PstI RFLPs及其在一个强直性肌营养不良症(DM)家系中的连锁分析*

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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 强直性肌营养不良症是一种常染色体显性遗传病，临床上以发病晚、症状表现多样为特征。 本研究应用19号染色体上与DM基因紧密连锁的单拷贝片希LDR152(D19S19)在中国上海地区人 群(61例)及1个DM家系中进行RFLP(Restriction Fragment Length Polymorphism)的连锁分 析，结果表明：(1)等位片段19kb和11kb在人群中的分布频率分别为43.44%和56.56%，其中1 9kb纯合子为22.95%、11kb纯合子为36.07%，19kb和11kb杂合子为40.98%，此结果与国外报 道的明显不同。(2)在我们所分析的这例DM家系中发现DM基因与19kb等位片段相连锁，并呈 孟德尔式遗传。进而，对两例无任何临床症状的DM基因携带者作出了明确的基因诊断，并对 家系中患DM的危险成员进行了DM的风险估计。

关键词 限制性片段长度多态性,强直性肌营养不良症,连锁分析,D19S19

分类号

The RFLP of LDR152/Psti in the Chinese and Its Application to Linkage Analysis i n A Myotonic Dystrophy Family*

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Abstract

Myotonic dystrophy (DM) is inherited as an autosomal dominant trait and is chara cterized by variable expressivity and late age-of onset.In the present paper,the DNA from 61 normal individuals and a DM family with 15 members of 4 generations were collected and digested with PstI,then hybridized with the LDR152 (D19S19). The results showed that the alleles for the PstI polymorphism were 19 and 11kb in size (gene frequencies were 0.4344 and 0.5656 respectively,which are obviously different from the previous data reported.).In this DM family,the carriers who had lived most of their life without knowing that they had been infected with th e disease were detected by the LDR152 and the estimation of DM risk on atrisk-in dividuals was also calculated.

Key words RFLP Linkage analysis Myotonic dystrophy D19S19

DOI:

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