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多个遗传标记情形下的RFLP的可诊断率估计[1)]

金 力, 刘祖洞

复旦大学遗传所，上海

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摘要 限制性酶切片段长度多态性（RFLP）作为共显性的遗传标记，已广泛应用于遗传病的产前诊 断。为评价各遗传标记的适用性，本文给出了在多个遗传标记情形下的RFLP的可诊断率的估 计方法，即在使用若干个遗传标记时，群体中可被诊断后代罹病与否的婚配类型的比例的估 计方法，包括致病基因为常染色体显性、常染色体隐性、X连锁显性和X连锁隐性的情况，并 认为增加遗传标记的个数和选择具较多等位基因的遗传标记，是提高产前诊断可诊断率的有 效途径。同时，根据，各遗传标记在群体中的多态性分布，可估计各遗传标记及其各种不同组合的可诊断率，以此选择在该群体中最为适合的遗传标记或其组合，以指导RFLP在产前诊 断中的应用。

关键词 RFLP,产前诊断,可诊断率,多个遗传标记

分类号

The Use of Restrictioin Fragment Length Polymorphisms for Diagnosis:The Estimati on of Diagnosable Rate of More Than One Genetic Markers

Jin Li,Liu Zu Dong

Institute of Genetics,Fudan University,Shanghai

Abstract

As a codominant genetic marker,restriction fragment length polymorphisms,referrd as RFLPs,has been widely applied to the prenatal diagnosis of some genetic dise ases.To evaluate the usefulness of the genetic markers in prenatal diagnosis,a p arameter,the diagnosable rate or the proportion of diagnosable matings is propos ed in the cases of two or more genetic markers.The estimation is applicable to t he cases of autosomal recessive and dominant disease,as well as those of X-linke d recessive and dominant diseases .The diagnosable rate of two or more genetic m arkers is based on the distribution of haplotypes.Thus in the case when n geneti c markers being used ,the disagnosable rate,or the fraction of matings in which the A2A2 fetus could be confirmed or ruled out by the fetal restriction fragment patterns in the A1A2×A1A2 matings,is expressed as follows: where A2 is a recessive deleterious gene,yi is the frequency of the genotype of ith type individuals ,and M is the total number of combination of n markers. It is a valuable strategy to use more than one genetic markers in order to incre ase the diagnosable rate.By using the data of the distribution of haplotypes,it is easy to find the most usable genetic markers or their combinations.

Key words RFLP prenatal diagnosis diagnosable rate multiple genetic markers

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