

一个DMD家系的Bayes分析和RFLP连锁分析*

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摘要 本文对一个DMD家系中先证者之妹的致病基因携带者风险用3种方法进行估计。单纯根据系谱分析,其风险为50%;以CPK值为条件概率作Bayes分析,其风险为25%;用RFLP连锁分析,推断其风险仅为5%。将RFLP连锁分析的结果作为又一个条件概率进行Bayes分析,其风险估计又进一步准确到不超过2%。三者结合,得到了最佳的结果。

关键词 [Duchenne型肌营养不良症](#),[Bayes分析](#),[RFLP连锁分析](#)

分类号

Bayesian and RFLP Linkage Analysis on a DMD Family

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Abstract

Three methods were applied to estimate the carrier risk of the daughter of an obligate carrier of the Duchenne gene in a pedigree of Duchenne muscular dystrophy (DMD). According to Mendel's law of segregation, the daughter (III-3) of the obligate carrier (II-2) has a 50% chance of being a carrier. III-3 is also found to have a normal value in a carrier test, creatine phosphate kinase (CPK), and so the Bayesian analysis in risk estimation shows that the posterior probability that she is a carrier (25%). The restriction fragment length polymorphism (RFLP) linkage analysis indicates that the probability that III-3 is a carrier is less than 5%. This result can be included in the Bayesian analysis to give an even lower risk of less than 2%. Our study demonstrates that the best result of carrier risk estimation can be obtained by a simultaneous application of Mendel's law, RFLP linkage analysis and the Bayesian analysis.

Key words [Duchenne muscular dystrophy](#) [the Bayesian analysis](#) [Restriction fragment length polymorphism linkage analysis](#)

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