

山东省假性肥大型肌营养不良的RFLP分析1.可疑携带者的基因型确定*

郭亦寿, 魏建军, 陈丙玺, 蔡富强, 杨亚萍, 龚瑶琴

山东医科大学医学遗传教研室 250012

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摘要 本文选用10个DMD基因内部和桥探针,对山东省9个地区的21个DMD/BMD家系的173名成员进行RFLP分析,其中可疑携带者55人。多数家系应用1—3个探针,有基因重组家庭选用4—5个探针,便可完成多态分析。RFLP分析可以确定85.45%的可疑携带者(≥95%可信限),即13人确定为携带者,30人排除携带者,另有4人风险大幅提高。通过这些探针的群体多态检测和不同家庭结构和类型的应用分析,我们提出了在中国人群中DMD/BMD基因诊断的基本程序

关键词 [DMD/BMD,RFLP分析,Southern 杂交,携带者检出,风险估计](#)

分类号

Application of RFLP Analysis to Duchenne and Becker Muscular Dystrophy in Shandong*

Guo Yishou, Wei Jianjun, Chen Bingxi, Cai Fuqiang, Yang Yaping, Gong Yaoqin

Department of Medical Genetics, Shandong Medical University, Jinan 250012

Abstract

There were eight intragenic and two flanking bridge DNA polymorphic markers used in the RFLP analysis in 21 Duchenne and Becker muscular dystrophy selected from nine different districts in Shandong Province. A total number of 173 individuals of the families were accepted for gene tracking and carrier detection, as well as prenatal diagnosis. Based on the frequency of the probe polymorphism tested in our study of 53 unrelated individuals, one to three high polymorphic probes were sequentially used and all families presented the informativeness in RFLP analysis. Some more intragenic probes were supplied for assessment of mutated sites and carrier status with the help of gene phases, if recombinations were found. Our results presented a ratio of 85.45% of carrier detection at risk females with 95% or more reliability, including 17 carriers and 30 non-carriers. It was not able to ascertain the carrier status of 8 females because of DNA absence from key family members. Finally, we proposed a diagnostic procedure for the RFLP analysis to DMD/BMD families.

Key words [DMD/BMD](#) [RFLP](#) [Southern blot](#) [Carrier detection](#) [Risk estimation](#)

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