

应用胎儿特异性抗体HbF (γ链) 标记法无创性产前基因诊断DMD Use of Fetal Specific Antibody—HbF to Detect Fetal Erythroblasts for Non-invasive Prenatal Diagnosis of DMD

刘丽英¹, 金春莲¹, 林长坤¹, 武盈玉², 孙开来¹ LIU Li-Ying¹, JIN Chun-Lian¹, LIN Chang-Kun¹, WU Ying-Yu², SUN Kai-Lai¹

- 1. ¹ 中国医科大学医学遗传学教研室, 沈阳 110001²
- 2. 中国医科大学附属二院儿保室, 沈阳 110001 1. Department of Medical Genetics, China Medical University, Shenyang, 110001, China³
- 2. Affiliated 2nd hospital, China Medical University, Shenyang 110001, China⁴

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摘要 以孕8~26周孕妇外周血为材料, 经过Percoll密度梯度离心初步富集, 胎儿细胞特异性抗体—HbF标记、识别胎儿有核红细胞, 母体和胎儿有核红细胞的精确区分是以胎儿和成人血红蛋白的组成差异为基础的。胎儿细胞胞浆黄染, 而具有成人血红蛋白的母体细胞没有颜色。显微操作法获取全部阳性细胞后, 以其全基因组扩增 (PEP) 的产物为模板, 进行性别检测、DMD基因的多重PCR检测和STR连锁分析。结果, 20名孕妇外周血中均发现与HbF呈阳性反应的胎儿NRBC。并完成7例DMD的产前基因诊断。HbF抗体标记法能有效识别胎儿有核红细胞, 是无创性产前基因诊断中很有应用前景的标记方法。 Abstract: Maternal blood was obtained at 8-26weeks of gestation. After discontinuous density gradient centrifugation with Percoll, HbF antibody was used to identify fetal NRBC. The precise differentiation between fetal and maternal NRBC is based on the constitutional difference between fetal and adult hemoglobin (Hb). Fetal cells appear yellow cytoplasmic staining, while adult cells colorless. NRBCs were collected by micromanipulation and whole genome amplification was performed. DMD was prenatally diagnosed by using the combination of sex determination, multiplex PCR and linkage analysis of several STR sites of dystrophin. NRBCs stained with HbF were found in all of 20 maternal blood samples with gestations, and 7 fetuses with risk of DMD were diagnosed. We concluded that HbF antibody could identify fetal NRBC efficaciously, and this is a kind of more prospective application method.

关键词 [有核红细胞](#) [免疫组化](#) [Duchenne型肌营养不良症 \(DMD\)](#) **Key words** [erythroblasts](#) [immunohistochemistry](#) [Duchenne muscular dystrophy \(DMD\)](#)

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