

# 利用孕妇血浆DNA检测胎儿性别的研究 Application of Fetal DNA in Maternal Plasma for Non-invasive Prenatal Diagnosis of Fetal Sex

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**摘要** 本文探讨应用孕妇血浆中游离DNA进行无创性产前性别诊断的可行性。用柱分离法提取73例孕妇血浆中DNA, 用巢式PCR技术检测其胎儿SRY基因。结果73位孕妇血浆DNA含量为0.0062~0.3399μg/μL。巢式PCR检测胎儿SRY基因的灵敏度为97.37% (37/38), 假阴性率2.86%(1/35), 特异度85.71% (30/35), 假阳性率13.16% (5/38), 总符合率91.78% (67/73)。采用孕妇血浆胎儿DNA和巢式PCR技术可以快速简便的进行无创性产前性别诊断, 诊断结果的准确率为91.8%, 对性连锁遗传病的预防具有重要意义。

**Abstract:**To investigate the feasibility and possibility of application of fetal DNA from maternal plasma for noninvasive prenatal diagnosis of fetal sex, plasma DNAs in blood samples of 73 pregnant women at the gestational period of 26 to 41 weeks were extracted by column separation and nested polymerase chain reaction were employed to amplify the SRY gene.A comparison was made between the amplification results and the real sex of the fetus after their delivery.The concordance rate of SRY gene amplification results of plasma free DNA with real fetal sex was 91.78% (67/73),the sensitivity rate was 97.37% (37/38),and the specific rate was 85.71% (30/35).The cell-free fetal DNA in maternal blood can be one of the valuable material sources for noninvasive prenatal diagnosis and the method of nested PCR could be useful for fetal sex determination.The specific rate of the test was 91.78%.It is of significance to prevent sex-linked inheritant diseases.

**关键词** [巢式PCR](#) [血浆DNA](#) [性别鉴定](#) [产前诊断](#) **Key words** [nested-PCR](#) [plasma DNA](#) [sex determination](#) [prenatal diagnosis](#)

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## Abstract

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