

遗传疾病的产前诊断I,羊水细胞培养和产前诊断一百例结果报道1

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摘要 遗传疾病的产前诊断技术在我国才刚刚开展。本文报道总结一百例染色体和甲胎蛋白宫内检查的结果。其中一例平衡易位、4例无脑畸形及两例性边;锁遗传疾病,胎儿被流产。由此表明产前诊断是预防某些遗传疾病的有效措施之一。

关键词

分类号

PRENATAL DIAGNOSIS OF HUMAN GENETIC DISEASES I. REPORT ON RESULTS OF DIAGNOSIS OF 100PREGNANCIES AMNOIOTIC FLUID CULTURE AMNOIOTIC FLUID CULTURE

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Abstract

In China, the research on prenatal diagnosis of human genetic diseases has just begun. The results of diagnosis of 100 pregnancies are presented in this report. Among these cases,71%(71 out of 100 cases) were chumosome analyses. The remainders (29%)were neural tube defects (table 1). Four cases of neural trbe defects ,one case of balanced chromosomal translocation [i.e. 46, XY, t(4;5) (4pter→4q35::5q 13→5qter; 5qter→5q 13::4q 35→4qter)], and two cases of sex-linked diseases were aborted. The prenatal diagnosis of balanced translocation and X-linked diseases were discussed.

Key words

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