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438例胎儿脐静脉血染色体核型的临床特征分析(PDF)

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Title: Clinical characteristics of chromosomal karyotypes in 438 cases of fetal umbilical venous blood

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关键词: 脐静脉穿刺; 产前诊断; 核型; 21-三体; 18-三体

Keywords: cordocentesis; prenatal diagnosis; karyotype; trisomy 21; trisomy 18

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摘要: 目的 探讨产前诊断中脐静脉血染色体核型的临床特征。 方法 对我院妇产科产前诊断中心2006年1月至2011年12月438例具备指征而行脐静脉穿刺诊断的胎儿脐静脉血染色体核型结果进行统计分析；比较系统超声提示各系统及器官异常时其脐带血核型异常率差异。 结果 各穿刺指征中，系统超声提示异常和高龄妊娠组中异常核型百分率分别为18.2%和12.9%；三体征（21/18/13）百分率分别为10.7%和8.1%；三体征异常核型百分率分别为58.9%和62.5%。系统超声提示异常时，消化系统异常和骨骼系统异常的21-三体检出率分别为33.3%和35.0%，21-三体异常核型百分率分别为75.0%和87.5%；神经系统异常和心血管系统异常的18-三体检出率分别为21.4%和9.7%，18-三体异常核型百分率分别为69.2%和50.0%。 结论 ①系统超声提示异常和高龄妊娠孕妇其发生染色体核型异常的风险较大，对这类孕妇应进一步行羊水穿刺或脐静脉血穿刺进行诊断；②消化系统异常、骨骼系统异常、神经系统异常和心血管系统异常与染色体核型异常的关系较为密切。21-三体者发生消化系统、骨骼系统异常的概率较大，而18-三体者发生神经系统异常、心血管系统异常的概率较大。

Abstract: Objective To investigate the clinical characteristics of chromosomal karyotypes in fetal umbilical venous blood during prenatal diagnosis. Methods The chromosomal karyotypes in fetal umbilical venous blood of 438 patients who were admitted in our hospital from January 2006 to December 2011 and underwent cordocentesis were analyzed. The chromosomal karyotypes were

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compared in different organ and system abnormalities indicated by systematic ultrasonography. Results In the patients with systematic ultrasonography-indicated fetal abnormalities and elder pregnant patients, the rates of abnormal chromosomal karyotypes were 18.2% and 12.9%, the rates of trisomy (21/18/13) were 10.7% and 8.1%, and the rates of abnormal chromosomal karyotypes with trisomy (21/18/13) were 58.9% and 62.5%. In the patients with fetal abnormalities of digestive system and skeletal system, the rates of trisomy 21 were 33.3% and 35.0%, and the rates of abnormal chromosomal karyotypes with trisomy 21 were 75.0% and 87.5%. As for the patients with fetal abnormalities of nervous system and cardiovascular system, the rates of trisomy 18 were 21.4% and 9.7%, and the rates of abnormal chromosomal karyotypes with trisomy 18 were 69.2% and 50.0%. Conclusion (1) The patients with systematic ultrasonography-indicated fetal abnormalities and the elder pregnant women have a relatively high incidence of abnormal chromosomal karyotypes, and amniocentesis or cordocentesis should be performed in prenatal diagnosis. (2) The fetal abnormalities in digestive system, skeletal system, nervous system and cardiovascular system are closely related to chromosomal karyotype abnormalities. Abnormalities of digestive or skeletal system are more possibly found in fetus with trisomy 21, while those of nervous or cardiovascular system are more possibly found in fetus with trisomy 18.

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