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中、晚孕期超声筛查胎儿染色体异常

Ultrasonography screening for fetal chromosomal abnormalities during the second and third trimesters of gestation

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英文关键词: [Amniocentesis](#) [Cordocentesis](#) [Karyotyping](#) [Ultrasonography, prenatal](#) [Chromosomal abnormality](#)

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中文摘要:

目的 探讨中、晚孕期超声筛查胎儿染色体异常的价值。方法 在妊娠中期和晚期,分别对有产前诊断指征的2955名和173名孕妇行羊水和脐血穿刺术检查染色体核型,比较不同指征孕妇的异常核型检出率,并分析染色体异常与超声异常的关系。结果 接受羊水穿刺的2955名孕妇中,异常核型150名,异常率5.08%;超声异常91名,异常核型11名,异常率12.09%;超声异常组染色体异常检出率(12.09%)明显高于唐氏高危组(4.46%)、高龄孕妇组(3.89%)、不良孕产史组(2.74%)($P < 0.05$)。173名脐血染色体核型分析中,26名为异常核型,异常率15.03%;超声异常118名,异常核型16名,异常率13.56%。结论 中、晚孕期超声筛查胎儿染色体异常有很大价值。

英文摘要:

Objective To investigate the value of ultrasonography screening for fetal chromosomal abnormalities during the second and third trimesters. **Methods** Amniocentesis and cordocentesis were performed on 2955 and 173 pregnant women respectively with indications for prenatal diagnosis to detect karyotype of the fetus during second trimester and late pregnancy. The detection rate of the chromosomal abnormality was compared in pregnant women of different indications. The relationship between ultrasonographic abnormalities and abnormal chromosomal karyotypes was analyzed. **Results** In chromosomal karyotypes analysis of 2955 pregnant women with amniocentesis, 150 abnormal chromosomal karyotypes were detected, the abnormal rate was 5.08%. There were 91 in 2955 pregnant women with ultrasonographic abnormalities, 11 abnormal chromosomal karyotypes were found within them and the abnormal rate was 12.09%. The detection rate of fetal chromosomal abnormality detected with ultrasound (12.09%) was higher than that of the Down's syndrome high risk group (4.46%), advanced age group (3.89%), as well as the history of abnormal deliveries group (2.74%) ($P < 0.05$). Within 173 pregnant women of chromosomal karyotypes analysis by cordocentesis, 26 abnormal chromosomal karyotypes were detected and the abnormal rate was 15.03%. There were 118 in 173 pregnant women with ultrasonography abnormalities, 16 abnormal chromosomal karyotypes were found within them and the abnormal rate was 13.56%. **Conclusion** Ultrasonography has great value in screening for fetal chromosomal abnormalities during the second and third trimesters.

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