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超声检出胎儿肱骨、股骨短小诊断21-三体的价值

Value of prenatal ultrasonic detection of short humerus length and femur length of fetuses in diagnosing trisomy 21

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中文摘要：

目的 探讨超声检出胎儿肱骨、股骨短小对诊断21-三体胎儿的价值。方法 对有产前诊断指征的6425名孕17~37周孕妇行羊水和脐血穿刺术检查染色体核型。超声常规测量胎儿双顶径、股骨和肱骨长度,计算超声对肱骨和股骨短小的检出率,评价超声检查胎儿肱骨、股骨短小对21-三体胎儿的诊断价值。结果 共检出21-三体胎儿66胎,其中肱骨股骨均短小24胎(24/66,36.36%),单纯肱骨短小22胎(22/66,33.33%),股骨短小18胎(18/66,27.27%)。核型正常的6130胎中,肱骨股骨均短小1579胎,单纯肱骨短小697胎(697/6130,11.37%),单纯股骨短小740胎(740/6130,12.07%),21-三体胎儿肱骨、股骨短小检出率明显高于核型正常胎儿(*P*均<0.05)。根据超声检出单纯肱骨短小诊断21-三体的敏感度为52.38%(22/42),特异度为84.68%(3854/4551),阳性预测值为3.06%(22/719),阴性预测值为99.48%(3854/3874);根据超声检出单纯股骨短小诊断21-三体的敏感度为42.86%(18/42),特异度为83.74%(3811/4551),阳性预测值为2.37%(18/758)、阴性预测值为99.37%(3811/3835)。结论 产前超声检出肱骨、股骨短小提示胎儿患21-三体的风险显著增加。

英文摘要：

Objective To investigate the value of prenatal ultrasonic detection of short humerus length (HL) and short femur length (FL) of fetuses in diagnosing trisomy 21. **Methods** Amniocentesis and cordocentesis were performed in 6425 pregnant women who had indication for prenatal diagnosis to detect karyotype of the fetus during 17-37 pregnancy weeks. The fetal biparietal diameter (BPD), HL and FL were conventionally measured. The ultrasonic detection rate of short HL and short FL were calculated, and its value for detecting the trisomy 21 fetuses was evaluated. **Results** In 6425 pregnant women underwent amniocentesis and cordocentesis, 66 fetuses of trisomy 21 were detected, including 24 (24/66, 36.36%) with both short HL and FL, 22 (22/66, 33.33%) with short HL and 18 (18/66, 27.27%) with short FL. Among 6130 karyotypically normal fetuses, 1579 with both short HL and FL, 697 (697/6130, 11.37%) showed short HL and 740 (740/6130, 12.07%) showed short FL. The detection rates of short HL and FL of trisomy 21 fetuses were significantly higher than those in karyotypically normal fetuses (*P*<0.05). The sensitivity, specificity, positive predictive value and negative predictive value of ultrasonic diagnosis of short HL was 52.38% (22/42), 84.68% (3854/4551), 3.06% (22/719) and 99.48% (3854/3874), respectively, while of short FL was 42.86% (18/42), 83.74% (3811/4551), 2.37% (18/758) and 99.37% (3811/3835), respectively. **Conclusion** Prenatal ultrasonic detection of short HL and short FL indicates the increase risk of trisomy 21.

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