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超声检出肱骨和股骨短小对筛查胎儿染色体三体的价值

Value of ultrasonic detection of humerus and femur length shortening in screening of fetal chromosomal trisomy

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中文摘要：

目的 评价超声检出肱骨和股骨短小对诊断胎儿染色体三体的价值。方法 对妊娠中期和中晚期有产前诊断指征的6425名孕妇行羊水和脐血穿刺术检查染色体核型，常规测量胎儿的双顶径、肱骨和股骨长度，计算肱骨和股骨短小对检出胎儿染色体三体的评价指标。结果 接受穿刺的6425名孕妇中，检出染色体三体胎儿98胎，其中肱骨股骨均短小52胎（52/98，53.06%），单纯肱骨短小28胎（28/98，28.57%），单纯股骨短小21胎（21/98，21.43%）。核型正常的6130胎儿中，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%）。染色体三体胎儿中单纯肱骨、股骨短小检出率明显高于正常胎儿（$P<0.05$）。超声检出单纯肱骨短小、股骨短小诊断染色体三体的敏感度分别为60.87%（28/46）、45.65%（21/46），特异度分别为84.68%（3854/4551）、83.74%（3811/4551），阳性预测值分别为3.86%（28/725）、2.76%（21/761），阴性预测值99.54%（3854/3872）、99.35%（3811/3836）。结论 产前超声检出肱骨和股骨短小对筛查胎儿染色体三体有重要价值。

英文摘要：

Objective To explore the value of ultrasonic detection of humerus and femur length (HL, FL) shortening in screening of fetal chromosomal trisomy. **Methods** Amniocentesis and cordocentesis were performed on 6425 pregnant women with indications for prenatal diagnosis to detect karyotype of the fetus during the second trimester and late pregnancy. The fetal biparietal diameter, HL and FL were conventionally measured. Several evaluation indexes about humerus and femur length shortening for the screening of chromosomal trisomy were calculated. **Results** In chromosomal karyotypes analysis of 6425 pregnant women by amniocentesis and cordocentesis, 98 chromosomal trisomy were detected, including 52 (52/98, 53.06%) with both short HL and FL, 28 (28/98, 28.57%) with short HL and 21 (21/98, 21.43%) with short FL. Among 6130 karyotypically normal fetuses,1579 with short HL and FL, 697 (697/6130, 11.37%) showed short HL and 740 (740/6130, 12.07%) showed short FL. The detection rate of short HL and FL of chromosomal trisomy were all significantly higher than that of karyotypically normal fetuses ($P<0.05$). The sensitivity, specificity, positive predictive value, negative predictive value of short HL and FL in detection of chromosomal trisomy was 60.87% (28/46), 45.65% (21/46); 84.68% (3854/4551), 83.74% (3811/4551); 3.86% (28/725), 2.76% (21/761); 99.54% (3854/3872), 99.35% (3811/3836), respectively. **Conclusion** Prenatal ultrasonic detection of short HL and FL has important value in screening of chromosomal trisomy.

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