· 论著 ·

二维及三维超声产前诊断末端肢体横向缺失的应用

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【摘要】 **目的**　探讨应用二维及三维超声产前诊断末端肢体横向缺失的临床应用。**方法**　对一孕龄为 27 孕周的 21 岁初产妇行产前超声检查。超声仪器为 Philips iU22 (Philips Ultrasound, Bothell, WA),应用 9-4 MHz 腹部探头及 6-2 MHz 三维探头为胎儿行二维及三维超声检查。**结果**　在二维超声检查时发现胎儿左下肢呈末端呈一大小约 1.7 cm×1.2 cm 不规则形实性团块,未见明显血流信号。三维超声显示一不规则形实性团块连于左腿末端。正常左足未显示。**结论**　以单足整只缺失为表现的末端肢体横向缺失是一种非常罕见的先天畸形。同时应用二维及三维超声对临床处理更有价值,并且是产科诊疗的有用工具。

【关键词】 末端肢体横向缺失;　二维;　三维;　超声

Prenatal Diagnosis of Terminal Transverse Deficiency With 2- and 3-Dimensional Ultrasound: A Case Report

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【**Abstract**】 **Objective**　To explore the prenatal diagnosis and delineate the terminal transverse deficiency (TLD) by using both two- (2D) and three-dimensional (3D) sonography. **Methods**　A 21-year-old primigravida underwent ultrasound (US) examination at 27 weeks′ gestation. The US machine used was a Philips iU22 (Philips Ultrasound, Bothell, WA) with a transabdominal 9-4 MHz and a 3D 6-2 MHz transducers. 2D and 3D US examination of the fetus were performed. **Results**　In 2D sonography, a solid mass with irregular shape, measuring 1.7 cm×1.2 cm without Doppler flow, was detected in the distal end of the left lower leg. 3D sonography was then performed and showed a solid irregular lump attached to the distal end of the left lower leg. A normal left foot was not detected. **Conclusions**　TLD with absence of a unilateral whole foot is a rare form of congenital deformity. This case shows the value of using both 2D and 3D US for better clinical management and as a useful tool in obstetric counseling.

【**Key words**】 terminal transverse deficiency;　two-dimensional;　three-dimensional;　ultrasound

1　Case Report

A 21-year-old primigravida underwent an ultrasound (US) examination in Department of Ultrasound, Siping Central Hospital at 27 weeks′ gestation. The US machine was a Philips iU22 (Philips Ultrasound, Bothell, WA) with a transabdominal 9-4 MHz and a three dimensional (3D) 6-2 MHz transducers.

2　Results

Two dimensional (2D) US examination of the fetus was performed. The fetus had grown appropriately for gestational age and had normal findings other than the left lower extremity. A solid mass with irregular shape, measuring 1.7 cm × 1.2 cm without Doppler flow, was detected in the distal end of the left lower leg (Figure 1). A normal left foot was not detected. 3D sonography was then performed and showed a solid irregular lump attached to the distal end of the left lower leg (Figure 2). The woman was healthy. She was a non-smoker and denied alcohol intake. There was no family history of limb defects or exposure to teratogenic medication, irradiation or infectious disease during pregnancy.

The woman requested for termination of pregnancy after counseling. A female fetus with absence of left foot was induced (Figure 3). No other congenital abnormalities were detected. The diagnosis was confirmed as fetal congenital deformity

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with absence of left foot. However, the woman and her relatives disagreed to do autopsy and chromosome examination for further evaluation.

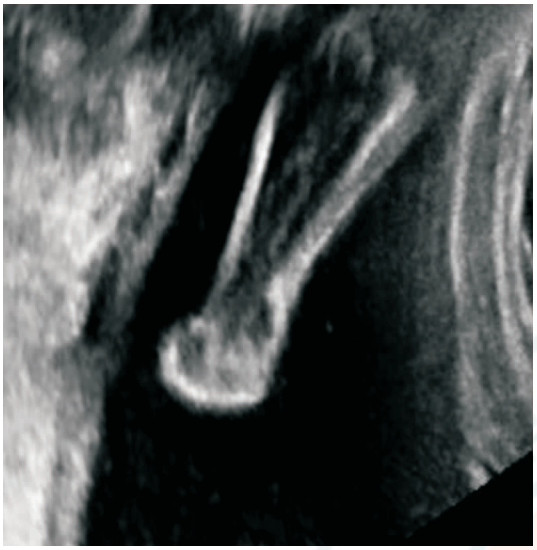

Figure 1 A solid mass with irregular shape, measuring 1.7 x 1.2 cm without Doppler flow, was detected in the distal end of the left lower leg

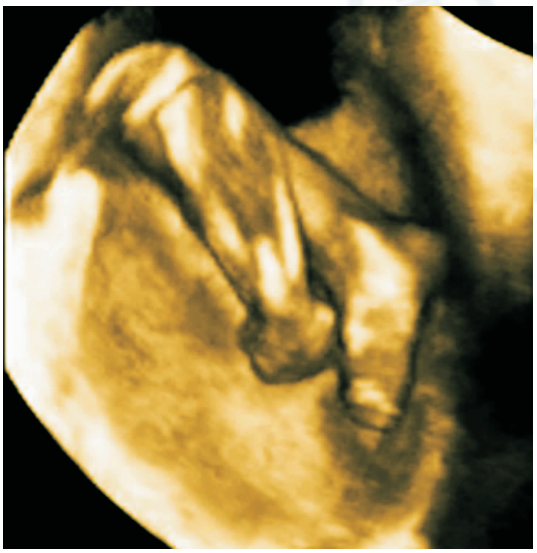

Figure 2 Three-dimensional (3D) sonography was then performed and showed a solid irregular lump attached to the distal end of the left lower leg

3 Discussion

According to the classification of limb defects proposed by the International Clearinghouse for Birth Defects Monitoring Systems, our case of absence of one foot was classified as distal or terminal transverse limb deficiency (TLD)[1]. Cases with TLD can be further categorized as 'isolated TLD' if only limb

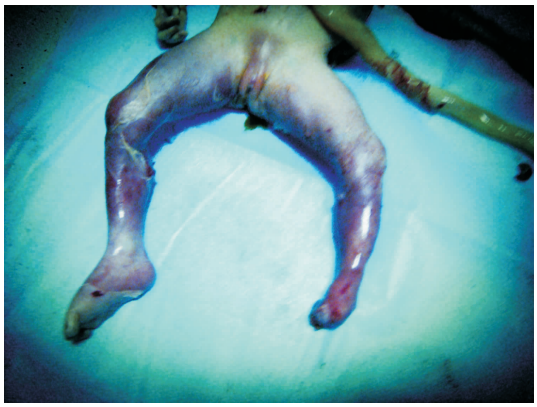

Figure 3 A female fetus with absence of left foot was induced

defects were present[2]. The prevalence rate for all types of limb deficiency was 0.69/1000[3]. While TLD occur less often but was the most common type of isolated limb deficiency with a birth prevalence of 0.124 to 0.15 per 1000 total births[4-5]. In the Hungary study, TLD were not usually associated with non-limb defects, and typically only one limb was affected. Upper limb was more frequently affected than lower. Left side and females were affected more often in upper limbs while lower limb defects were evenly distributed between right and left sides and both sexes. Familiar occurrence was not found[4, 6]. However, Keret *et al* shown that 30% to 45% of limb reduction deficiencies were associated with other malformations instead[7].

Our case was in accordance with the Hungarian study in that there was no association with any non-limb defects, no familial occurrence and only one limb was affected. However, in our case the abnormality was occurred in the lower limb rather than in the more common upper limb. To the best of our knowledge, this type of TLD: absence of a unilateral whole foot is rare and not much case has been reported yet.

The causes for the limb reduction defects were found to be associated with teratogenic exposures during pregnancy, after prenatal diagnosis procedure of chorionic villus sampling, vascular disruption or occlusion, vasoactive exposures included maternal cigarette smoking and use of decongestants, non-steroid anti-inflammatory drugs, and anti-hypertensive drugs in the periconceptional period, hereditary disorders, chromosome abnormalities or specific malformation syndromes[2-6].

However, there were studies did not support genetic causation since there was no familial occurrence[4]. This is supported by our case where there was no family history of TLD.

The mother was a non-smoker, so vascoactive exposures included maternal cigarette smoking is less likely. This was consistent with Werler's finding that maternal cigarette smoking might not play a major role in the pathogenesis of TLD[2].

However, the cause of the TLD in this case could not be found since the mother refused autopsy and chromosome examination. On the other hand, the aforementioned causes did not fit into our case except we were not sure chromosome abnormalities was a contributory factor or not. Therefore the previously mentioned cause could not explain our case.

For the mothers, 3D sonography is a novel means of visualizing and increasing their ability in appreciating fetal abnormalities. Our case shows the value of using both 2D and 3D ultrasound for better clinical management and as a useful tool in obstetric counseling.

(Figure 1-3, please see in No. 6 CD)

参 考 文 献

1 Stoll C, Duboule D, Holmes LB, *et al*. Classification of limb defects[J]. Am J Med Genet 1998, 77: 439-441.

2 Werler MM, Jaclyn LF, Bosco JL, *et al*. Maternal vasoactive exposures, amniotic bands, and terminal transverse limb defects [J]. Birth Defects Res A Clin Mol Teratol, 2009, 85(1): 52-55.

3 McGuirk CK, Westgate MN, Holmes LB. Limb deficiencies in newborn infants[J]. Pediatrics, 2001, 108(4): E64.

4 Czeizel AE, Vitez M, Kodaj I, *et al*. A morphological and family study on isolated terminal transverse type of congential limb deficiency in hungary, 1975-1984[J]. Teratology, 1993, 48(4): 323-327.

5 Harmon JV, Osathanondh R, Holmes LB. Symmetrical terminal transverse limb defects: Report of a twenty-week fetus [J]. Teratology, 1995, 51: 237-242.

6 Evans JA, Vitez M, Czeizel A. Congential abnormalities associated with limb deficiency defects: A population study based on cases from the Hungarian Congenital Malforamtion Registry (1975-1984) [J]. Am J Med Genet, 1994, 49(1): 52-66.

7 Keret D, Bronshtein M, Wientraub S. Prenatal diagnosis of musculoskeletal anomalies[J]. Clin Orthopaed Related Res, 2005, 434: 8-15.

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