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产前超声诊断及漏诊胎儿肢体畸形分析

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摘要 目的:探讨胎儿肢体畸形超声特征及诊断价值。方法:采用连续顺序追踪法对 66342 例妊娠 12-40 周孕妇行胎儿四肢畸形筛查。将产前超声诊断结果与引产或产后结果进行对比分析。结果:发生肢体畸形 271 例,发生率为 0.41 % (271/66342),包括四肢短小 5 例,桡骨发育不全 1 例,缺肢畸形 5 例,足内翻 17 例,手掌畸形 3 例,指趾畸形 222 例及骨骼多发畸形 18 例。其中产前诊断胎儿肢体畸形 49 例;漏诊 222 例,包括:足内翻 3 例、指趾畸形 218 例、多发骨骼畸形 1 例。胎儿肢体畸形的出现率和产前检出率分别为:四肢短小 1.84 % (5/271)、100 % (5/5);桡骨发育不全 0.36 % (1/271)、100 % (1/1);缺肢畸形 1.84 % (5/271)、100 % (5/5);足内翻 6.27 % (17/271)、82.35 % (14/17);手掌畸形 1.10 % (3/271)、100 % (3/3);指趾畸形 81.91 % (222/217)、1.8 % (4/222);多发骨骼畸形 6.64 % (18/271)、94.44 % (17/18)。结论:超声对胎儿手掌、脚掌部位以上畸形的检出率较高。指趾畸形出现率最高,但检出率最低。

关键词: 胎儿;超声;肢体畸形

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Analysis of Prenatal Ultrasound Diagnosis and Missed Diagnosis of Fetal Limb Deformities

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ABSTRACT Objective: This study was to investigate the ultrasonic characteristics and its value for fetal limb deformities. **Methods:** Systematic continuous sequence approach was performed to observe fetal limbs and fetal limb deformities for 66,342 cases of pregnancy in 12-40 weeks. The prenatal diagnosis results were analyzed and compared with postnatal results. **Results:** Two hundred and seventy-one cases in 66,342 fetuses occurred limb deformities (incidence rate as 0.41%) including five cases of short limbs, one case of radial hypoplasia, five cases of ectromelia, 17 cases of strephenopodia, 3 cases of palm deformities, 222 cases of fingers and toes deformities, 17 cases of multiple skeletal deformities. Forty-nine cases of fetal limb deformities were diagnosed by prenatal ultrasound and 222 cases were missed including 3 cases of strephenopodia, 218 cases of fingers and toes deformities, 1 case of multiple skeletal deformities. The frequency and prenatal detection rate of fetal limb deformities were: short limbs 1.84 % (5/271) and 100 % (5/5); radial hypoplasia 0.36 % (1/271) and 100 % (1/1); ectromelia 1.84 % (5/271) and 100 % (5/5); strephenopodia 6.27 % (17/271) and 82.35 % (14/17); palm deformity 1.10 % (3/271) and 100 % (3/3); fingers and toes deformities 81.91 % (222/217) and 1.8 % (4/222); multiple skeletal deformities 6.64 % (18/271) and 94.44 % (17/18). **Conclusion:** The detection rate of prenatal ultrasound was very high for the limb deformities above fetal palms and soles. Fingers and toes deformities had a high incidence rate but a very low detection rate.

Key words: Fetus; Ultrasound; Limb deformity

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前言

胎儿肢体畸形并不少见,国外报道发生率约 0.2 %^[1]。国内报道在所有围生儿先天畸形中,肢体畸形的发生率为 26 %^[2]。胎儿肢体畸形种类繁多,受累部位亦多,形成原因复杂^[3,4]。部分严重胎儿肢体畸形严重影响患儿生存率及生存质量,宫内早期诊断及早期干预可减少家庭及社会负担。随着超声诊断仪器分辨率提高及规范化产前超声检查,胎儿肢体畸形检出率明显提

高,但部分胎儿肢体畸形产前检出率仍较低。本研究对我院 2003 年至 2011 年产前超声诊断及漏诊的胎儿肢体畸形进行分析和报道。

1 资料与方法

1.1 一般资料

2003 年 1 月-2011 年 12 月在我院建卡接受产检的孕妇 66342 例,年龄 20-45 岁,平均年龄 30.6±4.3 岁,孕周 12-26 周。

1.2 检查方法及观察指标

仪器采用 GEV 730、HD11DX 及 Envisor C HD 彩色多普勒超声诊断仪。探头频率 2 MHz-5MHz。仪器调节至胎儿条件。孕妇于孕 13-18 周及孕 23-26 周行两次超声检查。孕妇取

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仰卧位。孕 13-18 周行常规检查,测量胎儿双顶径、枕额径、股骨、肱骨、羊水指数,观察胎盘位置、分级。其次观察胎儿结构,包括:颅骨光环、心脏四腔观、胃泡、腹壁、膀胱、脊柱、四肢。胎儿四肢运用连续追踪法^[5,6]检查胎儿肱骨、尺桡骨、手掌、手指及手与前臂的关系,股骨、胫腓骨、脚掌、脚趾及脚与小腿的关系等情况对胎儿肢体情况进行初步筛查。孕 23-26 周再次行常规超声检查,观察胎儿生长发育情况:测量胎儿双顶径、枕额径、头围、腹围、股骨、肱骨、羊水指数、观察胎盘位置、分级、脐血管数目。其次观察胎儿结构,包括:颅骨光环、脑中线、脉络膜、丘脑、小脑、侧脑室、三脑室、透明隔腔、小脑延髓池、眼眶、鼻孔、上唇、脊柱、双肺、心脏四腔、胃泡、肝脏、腹壁、双肾、膀胱、四肢,胎儿四肢再次运用连续顺序追踪超声检测法观察。

2 结果

随访新生儿及畸形引产资料,66342 例胎儿中发生肢体畸形 271 例,胎儿肢体畸形的发生率为 4.08 %。

产前超声诊断胎儿肢体畸形 49 例,包括:四肢短小 5 例、桡骨发育不全 1 例、缺肢畸形 5 例、足内翻 14 例、手掌畸形 3 例、指趾畸形 4 例、多发骨骼畸形 17 例。漏诊 222 例,包括:足内翻 3 例、多发骨骼畸形 1 例、指趾畸形 218 例。胎儿肢体畸形的出现率和检出率见表 1。one case of radial hypoplasia, five cases of ectromelia, 17 cases of strephenopodia, 3 cases of palm deformities, 222 cases of fingers and toes deformities, 17 cases of multiple skeletal deformities.

表 1 胎儿肢体畸形出现率和检出率

Table 1 The rate of incidence and detection of fetal limb deformities

Abnormality types	Case(n)			Incidence rate (%)	Detection rate (%)
	Total	Corrected diagnosis	Missed diagnosis		
Short limbs	5	5	0	1.84	100
Radial hypoplasia	1	1	0	0.36	100
Ectromelia	5	5	0	1.84	100
Strephenopodia	17	14	3	6.26	82.35
Palm deformities	3	3	3	1.10	100
Fingers and toes deformities	222	4	218	81.91	1.8
Multiple skeletal deformities	18	17	1	6.64	94.44

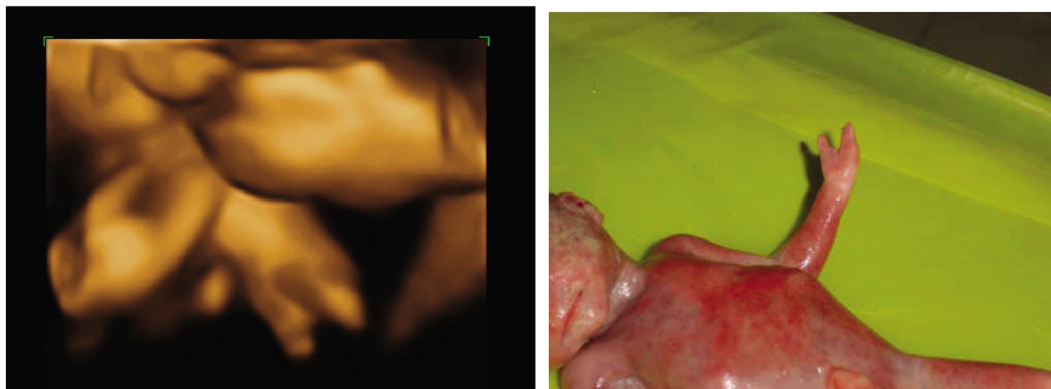


图 1 缺指(三指)畸形

Fig.1 Ectrodactyly (three fingers)

注:左 孕 23 周胎儿缺指(三指)畸形声像图,右 缺指(三指)畸形图引产照片。

Note: Left the ultrasonography of fetal ectrodactyly (three fingers, 23 weeks), Right the picture of ectrodactyly (three fingers) after induced labor.

3 讨论

国内有关产前超声诊断胎儿肢体畸形文献报道不多,各家产前诊断检出率高低不一,从 23-55 %不等。李胜利采用连续追踪法观察胎儿肢体畸形检出率达 87.18 %^[5]。本研究对 66342 例孕妇分别在孕 13-18 周、23-26 周采用该方法观察胎儿肢体,随访新生儿及引产儿资料进行回顾性分析。结果发现不同部位的肢体畸形发生率和检出率有明显差异。手掌、脚掌以上部位的畸形占肢体畸形的 18.09 %,检出率达 82.35-100 %;指趾畸

形占肢体畸形的 81.91 %,而检出率仅为 1.8 %。说明手掌、脚掌部位以上的严重胎儿肢体畸形发生率低但检出率高。在早中孕期规范的产前超声检查能及时准确诊断。指趾畸形发生率最高,但检出率很低。导致胎儿指趾畸形漏诊的原因很多^[7-11],其中临床重视不够也是一个主要原因。尽管单纯指趾畸形出生后对人体功能影响很小,但是部分指趾畸形可合并其它系统畸形或染色体异常。有文献报道拇指发育不全可能为 18- 三体的早期唯一征象^[12],国内学者亦报道胎儿指趾畸形与 18- 三体及 13- 三体等染色体异常具有一定关联^[13]。因而指趾畸形的产



图2 左手赘生物

Fig.2 Vegetation on the left hand

注:左 产前超声检查示胎儿左手拇指外侧见一不均匀低回声(36周),右 新生儿左手大鱼际见一赘生物。

Note: Left a inhomogeneous hypoechoic lesion outside fetal left thumb shown by prenatal ultrasound (36 week), Right a neoplasm at the greater thenar in the left hand of newborn baby.

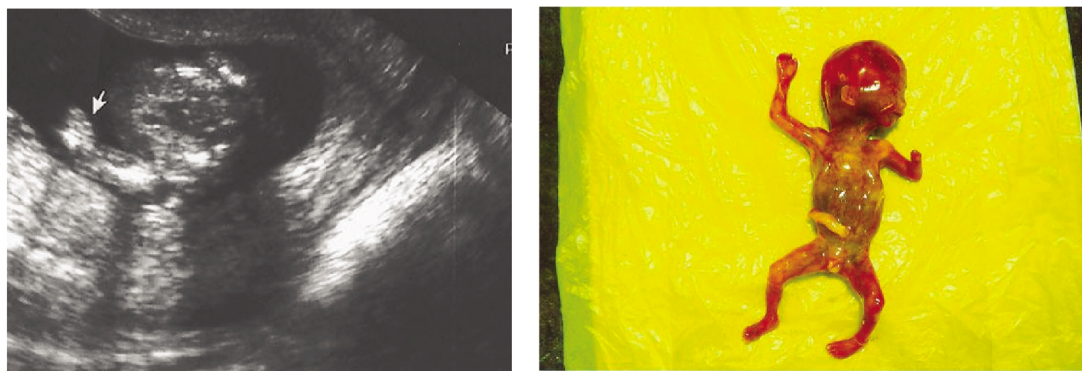


图3 缺肢畸形

Fig.3 ectromelia

注:左 产前超声检查示胎儿左侧手掌缺如,前臂短小,仅见一根骨,右 缺肢畸形引产照片。

Note: Left prenatal ultrasound showed that the left hand was absent and the left antebrachium was short, Right the picture of ectromelia after induced labor.

前超声诊断有利于染色体异常等的宫内发现,具有重要的临床意义。

本研究漏诊4例手掌、脚掌部位以上畸形,包括足内翻3例和膝关节畸形1例。足内翻畸形是一种常见肢体畸形,活婴发病率为0.1-0.3%^[14]。本研究中足内翻出现率较高。足内翻产前检查漏诊的原因可能是未能真正追踪小腿骨骼与足底的相互关系,未能显示有价值的切面而漏诊。羊水过少、孕周过大等因素也可影响胎儿足的观察。足内翻可单独存在,也可与其它畸形并存^[15-17],本研究发现胎儿足内翻17例,其中9例合并其它畸形,占52.9%。产前超声诊断单纯足内翻者,发生染色体畸形的危险性增加6-22%,且仍有10-13%的患儿经新生儿检查发现额外结构或神经异常而成为复杂的足内翻^[18]。因此当超声检查发现足内翻时,应仔细检查胎儿其他结构,并建议进一步行染色体检查。

关节畸形可发生在肩关节、肘关节、腕关节、髋关节、膝关

节、踝关节^[19]。胎儿关节畸形虽然以踝关节和腕关节多见,但其它部位关节也有可能发生^[20]。此例膝关节畸形产前未检出原因就是重点观察了胎儿踝关节和腕关节而忽视了其它关节的检查而导致。关节畸形出生后对人体功能影响较大,因此产前超声检查时应尽量观察到胎儿每一个关节,可疑异常要在胎动后反复多次观察,避免漏诊和误诊的发生。其它严重肢体畸形如四肢短小,桡骨发育不全及缺肢畸形的检出率为100%。

综上,胎儿肢体畸形种类多样,部分畸形可合并其它畸形或染色体异常,部分畸形严重影响患儿生存质量。因而超声诊断医师应提高意识,注意胎儿肢体的检查,避免相关畸形的漏诊。

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