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· 病例报道 ·

Prenatal ultrasonic diagnosis of plit hand and split foot malformation: a case report

产前超声诊断胎儿裂手、裂足畸形 1 例

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[中图分类号] R445.1; R714.53

[文献标识码] B

孕妇, 30岁, 孕4产2, 孕28⁺周, 无致畸药物服用史、放射接触和病毒感染史、吸烟饮酒等不良嗜好及相关家族史, 有孕早期自然流产史, 此次妊娠孕期末行定期产前检查, 今来我院行常规产前检查。超声检查: 胎位左骶后位, 双顶径6.90 cm, 头围25.70 cm, 腹围22.90 cm, 股骨长4.90 cm, 胎盘着床于前壁, 成熟度 I 级, 羊水最大深度5.40 cm, 胎心率133次/min, 律齐, 估测胎儿体质量(1028.00±154.00)g, 胎儿头部、胸部、腹部、四肢常规检查未见明显异常, 胎儿颈部皮肤可见“U”形压迹。胎儿双手、双足部分骨性结构未显示, 双手均仅见2根手指回

声, 双足均仅见2根足趾回声, 双手、双足形态异常, 双足呈蟹钳状(图1, 2)。超声提示: 宫内妊娠, 单活胎, 臀位, 胎儿大小相当于27⁺周; 胎儿双手、双足异常。后经引产证实(图3)。引产标本经X线检查: 双手部分掌骨、指骨缺失, 双足部分跖骨、趾骨缺失(图4)。

讨论: 胎儿裂手、裂足畸形(SHFM)是一种罕见的先天性畸形, 因指/趾或掌骨/跖骨缺失, 表现为手或足的尺侧与桡侧或胫侧与腓侧不同形式的分离, 本病多为常染色体显性遗传, 偶有染色体检查未见异常文献^[1]报道, 染色体畸变及基因突变均可



图1 二维超声显示胎儿一手仅见2根手指回声, 桡侧缺如(A); 一足呈“蟹钳”样分裂(B)

图2 胎儿双足三维重建图像显示双足均仅见2根足趾回声, 中心分裂

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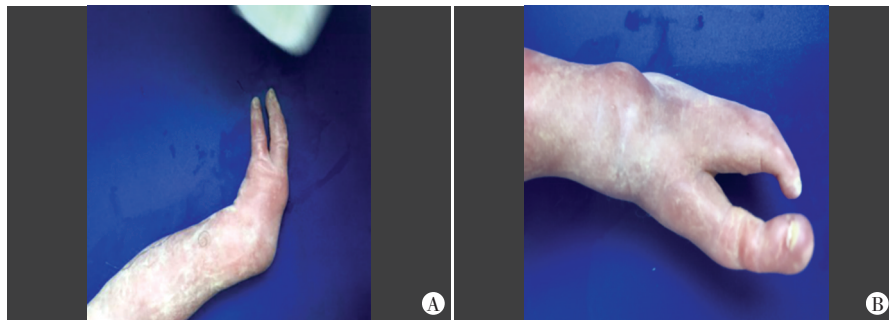


图3 引产标本一手(A)及一足(B)图片清晰显示畸形特点,与产前超声成像相仿



图4 引产标本正位X线片:手部分掌指骨、足部分跖趾骨缺如,软组织成像符合SHFM

导致该病。SHFM可单独发生,亦可作为综合征的一部分存在,我国围产儿中单发SHFM发生率为万分之0.64,综合征SHFM发生率为万分之1.0,其中单发SHFM无城乡差别,综合征SHFM的乡村发生率高于城镇,男性单发及综合征SHFM的发生率均高于女性^[2]。SHFM属于肢体末端畸形,超声对此类畸形排查受限条件较多,产前诊断仍有一定困难,易漏、误诊。本例孕妇为初次产前超声检查,对于胎儿畸形产前筛查的认识不足,导致来检时孕周偏大,引产后标本及孕妇夫妻双方均未行相关基因学检查,无法定位相关遗传基因。本例超声检查清晰显示了胎儿手足,明确诊断此畸形。对于SHFM的预后判断,也因其有无其他合并症而有所不同,综合征型预后较差,本病

例属于非综合征型裂手裂足畸形,并非致死性畸形,但因其双手桡侧手指缺失,仅存尺侧二指,对于双手精细功能会有较大影响,出生后矫治手术效果欠佳;裂足除影响美观外,对行走的影响较小,一般有裂足者可不予矫治。

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