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(收稿日期:2016-10-28)

·病例报道·

Fetus anophthalmia diagnosed by ultrasonography:a case report

超声诊断胎儿无眼畸形 1 例

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[中图法分类号]R714.5;R445.1 [文献标识码] B

孕妇,30 岁,孕 1 产 0,孕 23 周,既往无不良孕产史,唐氏筛查无异常。系统超声检查:宫内单活胎,双顶径 6.11 cm,头围 21.98 cm,小脑横径 2.34 cm,腹围 18.90 cm,肱骨 3.57 cm,股骨 4.30 cm。前壁胎盘,成熟度Ⅰ级,羊水指数 20.13 cm。胎儿头颅光环完整,颅内结构正常,透明隔腔存在,侧脑室前角及后角均无增宽,小脑形态正常,蚓部存在。胎儿心脏、腹部及四肢扫查均未见异常声像。胎儿上唇及硬腭正中部分回声中断,上唇部断端宽约 0.56 cm,硬腭部断端宽约 0.66 cm。冠状面扫查胎儿眼部可探及线状睑裂回声,横向扫查胎儿眼部,双侧眼眶内未探及正常眼球结构,仅可见类圆形无回声小囊,其内可见不规则团状偏强回声,回声不均且与周围眼眶结构分界不清(图 1)。超声提示:①宫内单活胎;②胎儿双眼发育异常,考虑无眼畸形;③胎儿唇腭裂(Ⅲ度)。后行 MRI 检查提示胎儿无眼畸形。羊水穿刺结果提示胎儿染色体核型和染色体微缺失结果均正常。孕妇后引产出一男性死婴,尸检所见:死婴上下眼睑及眼裂存在,眼眶内无眼球,仅见少量脂肪组织,颜面部Ⅲ度唇腭裂(图 2)。

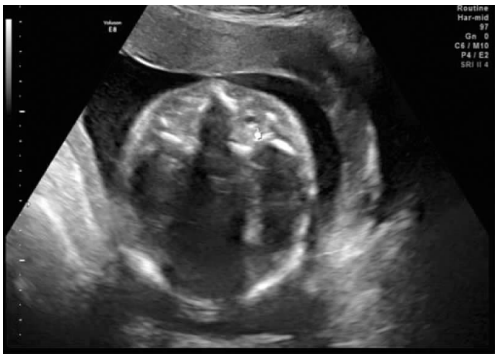

图 1 胎儿无眼畸形的超声图像

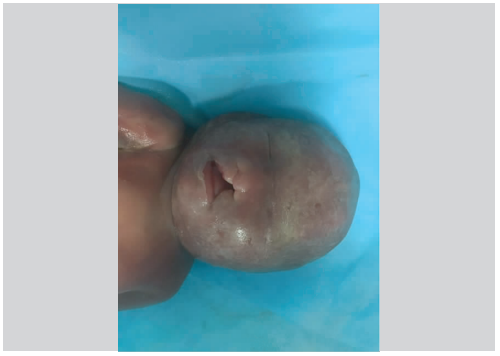

图 2 胎儿无眼畸形的引产图

讨论:对称性无眼畸形极为罕见,发病率约十万分之一[1],超声筛查易被遗漏。此畸形位于体表,一旦出生对家长及患儿均会造成巨大影响。临床检查时在眼部扫查中眼眶可见的情况下还需仔细分辨眼内结构,明确有无眼内附属结构,与小眼畸形相鉴别。小眼畸形多表现为眼眶及眼球的明显缩小,双眼受累者少见,严重者与无眼畸形在临床上难以区分。本例胎儿可显示部分眼眶,眶内除偏高回声外似可见小的类圆形无回声区,若误认为晶状体则极易误诊。MRI 对早期胎儿眼内附属物诊断有明确优势,本例孕妇后行 MRI 检查证实为无眼畸形,故产前超声结合 MRI 检查更有利于明确诊断。

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(收稿日期:2017-02-27)

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