但其对臂丛神经节前损伤有一定局限性，不能直接显示椎间孔内的神经根撕脱，尤其是断端处于椎间孔内的节前损伤，有待今后进一步研究。

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·病例报道·

Ultrasonic diagnosis of a "bird face" fetus: a case report

超声诊断"鸟面"胎儿1例

戴渝萍 陈小辉

[中图法分类号]R445.1；R714.5 [文献标识码]B

孕妇，26岁，孕1产0，孕期及既往体健，无家族遗传疾病史，无有害物质接触史，孕期实验室检查结果正常。孕13周于我院行常规超声检查：胎儿双顶径2.00 cm，头围8.16 cm，腹围6.24 cm，股骨长1.06 cm，头臀径6.85 cm，颈项透明层厚度（NT）1.1 mm；彩色多普勒示出心血流似仅有一条，颅脑、鼻骨、鼻后三角、四肢等均未见明显异常。超声提示：单活胎，孕13⁺周；心脏结构异常可能，建议3周后复查。复查超声：胎儿双顶径3.41 cm，头围13.70 cm，腹围11.40 cm，股骨长2.24 cm，肱骨长2.14 cm，羊水最大深度3.70 cm，心率157次/min，鼻骨、双眼均可见，下颌斜切面未见明显下颌骨声像，面部矢状切面下颌明显后缩，下唇后移，与下颌形成的"S"形曲线消失几乎呈一直线，上唇突出，持续张口未见闭合，口腔前部未见胎舌声像，嘴部冠状切面呈小"O"形（图1），未见明显上腭回声中断。双耳形态大小尚可，位置似偏低。心脏心轴右偏，四腔心可见，左、右对称，冠状静脉窦无扩张，室间隔上段回声中断约0.20 cm，仅见一条动脉从心底发出，宽约0.26 cm，三血管切面示该动脉左侧见一内径约0.10 cm的血管声像（图2），远程走行欠清晰。腹部横切面示脐静脉入腹后分为两支，一支向右侧弯曲走行，另一支较细直接汇入下腔静脉。静脉导管频谱显示a波反向。胎儿余结构未见明显异常。超声提示：①宫内单活胎，孕16⁺周；②颜面部发育异常，考虑小下颌畸形（下颌骨缺如？下颌骨极度短小伴骨化不良？），小嘴畸形，双耳低位？③复杂先天性心脏病，考虑心

（下转第933页）

作者单位：400020 重庆市人口和计划生育科学技术研究院
通讯作者：陈小辉，Email：wise10004004@163.com

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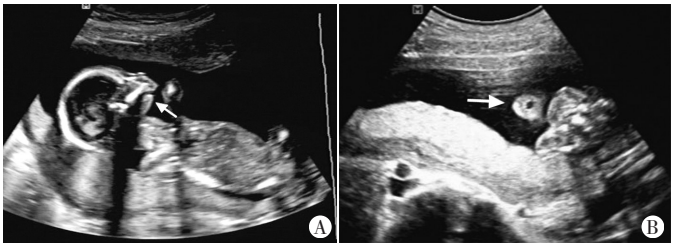
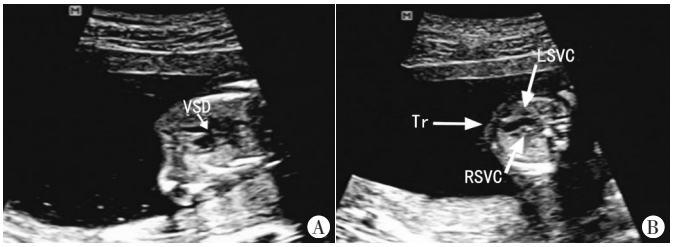
轴右偏,室间隔缺损,永存动脉干,永存左上腔?肺静脉异位引流待排;④脐静脉、下腔静脉异常连接?

孕妇后行引产,大体标本胎儿侧面观见上唇前突,下颌极度后缩,呈"鸟面"样,双耳位置低(图3);正面观见双眼距离较宽,嘴部张开呈小圆形,口腔中后部见后坠的舌头(图4),上腭完整;躯体余部位正常。



A:箭头示明显后缩的小下颌,口腔前部无胎舌;B:箭头示呈"O"形小嘴

图1 孕16⁺周胎儿颜面部超声图像



A:室间隔缺损(箭头示);B:三血管切面示单一动脉干及左侧异常血管横断面(箭头示)。VSD:室间隔缺损;Tr:动脉干;RSVC:右上腔静脉;LSVC:左上腔静脉

图2 孕16⁺周胎儿心脏超声图像

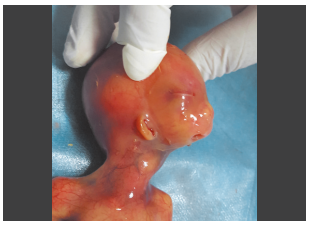


图3 引产后胎儿侧面观,呈"鸟面"面容,上唇突出,下颌小,极度后缩,耳下移

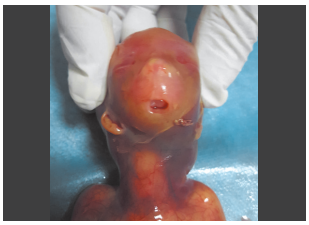


图4 引产后胎儿正面观,眼距增宽,下颌后缩,"O"形张口小嘴,舌头后坠

讨论:胎儿小下颌畸形常伴发于染色体畸形或基因变异综合征、骨骼系统发育不良等疾病。本例胎儿外貌特征主要为严重的下颌后缩、上颌相对突出似鸟样面容,同时伴有复杂心脏结构异常等。引产后考虑Pierre Robin综合征和DIGeorge综合征,前者新生儿发病率为1/5000~1/3000,产后因呼吸道阻塞致死率为30%~65%,与遗传、宫内巨细胞病毒感染等有关[1]。DIGeorge综合征是由胚胎期第3、4腮弓发育异常引起小下颌、眼距宽等鸟面征象,可合并腭裂和耳低位,心脏畸形以永存动脉干为主,是较常见的染色体微缺失综合征,新生儿发病率约1/4000,6%~10%有家族史[2],多伴胸腺、甲状旁腺缺如或发育不全。

临床诊断小下颌伴心脏畸形时应排除Shpintzens综合征,18-三体、13-三体、软骨发育不全等也常伴发小下颌,需鉴别诊断。本例胎儿因孕周较小,上腭、胸腺、心脏等部分组织结构尚未能清晰显示,若继续妊娠可能会出现更多异常超声表现,染色体、基因等遗传学检查则能明确诊断。

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