GUIDELINES

ISUOG 实践指南（更新）：胎儿心脏超声筛查

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引言

本文是 ISUOG 中孕期胎儿心脏超声筛查指南[1]的更新修订版，它引入了当前产前检测先天性心脏病(CHD)的最新知识。与其他最近发表的指南及建议一致[2-5]，本指南根据最新循证证据，在原有四腔心切面常规筛查的基础上，增加了心脏流出道切面的扫查内容。

CHD 是婴幼儿死亡的主要原因，在活产儿中的发病率约为 4-13/1000[6-8]。根据世界卫生组织在 1950 - 1994 年间的数据，42%的婴幼儿死亡是因心脏缺陷所致[9]。心脏结构异常也是产前超声检查最常漏诊的先天性异常之一[10,11]。对某些类型的 CHD 而言，如果能够在产前及时发现，有望改善患者的结局[2-16]。但由于检查者经验、产妇肥胖、换能器频率、腹部疤痕、胎龄、羊水量及胎儿位置等多种因素的影响[18,19]，CHD 的产前检出率存在较大差异[17]。持续反馈式医疗专业培训的进行、低门槛的超声心动图转诊指证的建立以及胎儿心脏专科医生诊疗便捷通道的设立，对改善筛查效果尤为重要[8,20]。譬如，北英格兰一医疗机构经过一项为期 2 年的培训计划后，其严重心脏畸形的检出率增加了一倍[21]。

制定胎儿心脏筛查指南的目的，是要最大限度地提高中孕期超声胎儿心脏异常的检出率[22]。指南可作为常规产前检查的一部份[23-25]，用于低风险胎儿的评估；也有助于发现遗传综合征高危胎儿，为患者咨询、产科处理和多学科会诊提供有利信息。疑似心脏畸形者须以胎儿超声心动图进行更为详尽的评估[26]。

一般注意事项

尽管四腔心切面和流出道切面在发现胎儿 CHD 中的作用已得到充份证实，导致漏诊的潜在诊断陷阱仍须引起注意[27-29]。执行详细的心脏筛查流程，认识四腔心切面检查并非单纯的数出四个心腔，明白部分病变只能在妊娠后期才能发现，知道某些类型的异常（如大动脉转位或主动脉缩窄）单从四腔心切面不易发现等，是提高 CHD 检出率的重要因素。因此四腔心切面结合流出道切面进行心脏筛查，是改进 CHD 检测的重要步骤。

胎龄

虽然妊娠 22 周后很多结构的检测效果仍然令人满意，心脏筛查的最佳时间为月经龄妊娠 18-22 周。部分异常，尤其是颈项透明层增厚的情况下，也可于早孕后期及中孕初期识别[30-35]。虽然患者多希望在孕早期发现重大缺陷[36]，但选择在 20-22 周筛查胎儿结构可减少重复超声检查的次数。

技术因素

超声换能器

高频率探头可提高分辨率以增加细微缺陷的检出率，缺点是声波穿透力下降。检查时应尽可能使用高频率探头，但须意识到穿透力和分辨率的关系。谐波成像可改善超声图像，这在孕晚期腹壁增厚的孕妇尤为明显[37]。

成像参数

横断面灰阶超声成像技术仍是进行可靠胎儿心脏扫查的基础。系统设置方面应强调高帧频，提高对比度和分辨率，使用低平滑设置、单聚焦区，并采用相对窄的图像区域取图。

缩放和连续影像循环

检测时应把图像放大，直至心脏占据至少 1/3 到 1/2 的屏幕。可采用图像回放来辅助正常心脏结构的实时评估，例如，使用该功能以确定心脏瓣膜在整个心动周期的运动情况。放大图像和使用回放图像有助于识别心脏异常。

心脏检查

自最初的 ISUOG 指南[1]公布至今已经过了一段时间，考虑到最近文献证据，建议目前心脏筛查的内容应包括四腔心及流出道两个切面的检查[38 - 46]。

四腔心切面

四腔心切面检查应根据具体标准仔细评估，而绝非简单地数出四个心腔。表 1、图 1 和图 2 显示了主要检查要素。评估心脏位置，首先要决定胎儿的左右侧，尔后确定胃和心脏位于胎儿左侧。正常心脏的大小通常不会超过胸腔面积的 1/3。某些角度可能会发现胎儿心脏周围出现薄薄的低回声边缘，以致误诊心包积液；事实上若无合并其他异常发现，通常可认为是正常变异[47,48]。

心脏位置主要位于左侧胸腔，心轴通常朝向左侧，角度约为 45 ± 20°(2 SD)[49](图 1)。须留意心轴和心脏位置是否正常；即使四腔心切面的图像不够理想，心轴和心脏位置的评估通常都不会有太大的困难[50]。当发现胎儿心脏和/或胃不在左侧，须怀疑心脏位置异常的可能。心轴异常会增加心脏畸形的风险，特别是流出道相关的畸形；它也可能与染色体异常有关。如心脏自正常左前位移位，可能是先天性隔疝或肺囊性腺瘤样变等占位性病变所致。位置异常也可由胎儿肺部发育不全或缺如引起。心脏轴左移可与胎儿腹裂和脐膨出同时出现。

要确定心率正常和心律规则。胎儿正常心率介乎每分钟 120-160 次(bpm)。轻度和短暂的心动过缓也可在正常的孕中期胎儿观察到。顽固性心动过缓，特别是心率维持在每分钟 110 次以下[51]的情况，须由胎儿心脏专家进行及时检查，评估心脏传导阻滞的可能性。孕晚期反复出现的胎儿心动过缓，可能系胎儿缺氧所致。偶发的停搏通常不会增加胎儿心脏结构畸形的风险，且多为良性，可自行消退。但部分情况下可出现具临床意义的心律失常，此时须进一步行胎儿超声心动图检查[52-54]。作为选择，增加多普勒扫描帧频、排除心包积液可能并确认胎儿心脏视图正常，也可以帮助确认及减少漏诊机会。胎动时出现的轻度心动过速(> 160 bpm)是正常变异。而持续性心动过速(≥ 180 bpm)[55]须进一步检查，以评估胎儿缺氧或更严重的心动过速的可能。

正常情况下，两个心房的大小大致相等；卵圆孔瓣开向左心房；原发隔，即房间隔组织下缘应存在，构成心脏“十字交叉”的一部分，即房间隔下部和室间隔上部连接处，亦为房室瓣插入点。一般可见肺静脉汇入左心房。如技术可行，建议至少要看到两条肺静脉。

调节束，系近心尖部、横跨右心室腔的一束特殊肌肉组织，可用以识别形态右心室。左心室心尖部内壁光滑，形成心尖。两个心室大小应大致相等，室壁无增厚迹象。虽然晚孕期时轻度心室不对称可为正常变异，但如中孕期出现明显左右不对称则须作进一步检查明确[56]；左心梗阻性病变，如主动脉缩窄和发展中的左心发育不良综合征等均为其中重要原因[57,58]。

室间隔检查，应详细扫查自心尖至“十字交叉”部有无缺损；但有时不易发现。当超声声束与室间隔垂直时，最易观察缺损。当声束与室间隔平行时，可能出现衰减伪像，致使误诊室间隔缺损。如超声成像系统横向分辨率不足，则不易发现细小的室间隔缺损(1 - 2 mm)，该情况在胎儿大小及位置均不满意时尤为明显。但细小室间隔缺损的临床重要性大多有限，很多甚至可宫内自然闭合[59,60]。

心脏的两个房室瓣（右边：三尖瓣，左边：二尖瓣）应能各自自如地开放。

三尖瓣附着点较二尖瓣近心尖（即正常偏移）。房室瓣对位异常可成为房室隔缺损等心脏异常的重要超声线索。

流出道切面

左、右心室流出道(LVOT 和 RVOT)切面是胎儿心脏筛查的必要组成。确认两条大血管正常非常重要，须检查其相应的心室连接，两血管相对大小及位置，以及动脉瓣充分打开的情况等。如未能确认正常，应建议进一步评估。

流出道检查最基本的要求是，两条大血管的大小大致相等，且二者起始部，即从相应心室发出时，彼此相交呈直角（正常“交叉”，附录 S1 图组 1)。一项包括了逾 18 000 例胎儿的产科超声大样本调查，评估了将心脏四腔心切面结合流出道切面检查（技术可行的情况下）纳入常规 30 分钟超声检查的可行性[61]。绝大部分(93%)四腔心切面评估满意的案例，亦可获得满意的流出道切面。不显影率分别为：LVOT 4.2%，RVOT 1.6%，双流出道 1.3%。

此外，自右室流出道起始端开始连续扫描，可获得包括三血管(3V)和三血管气管(3VT)切面在内的横切面图像（附录 S1 图组 2)，可用来显示大血管不同层面及其周围结构。一个纳入近 3000 例低风险妊娠的研究发现，常规超声筛查时，如在标准四腔心切面基础上增加 3V 切面和 3VT 切面检查，平均额外增加的时间仅为两分多钟（135 秒; SD 20 秒)。但大约有 1/3 的个案，由于胎位欠佳（脊椎前位）[46]，须耗费 15-20 分钟待胎位好转后继续心脏超声检查。

较之单纯的四腔心切面，流出道切面评估可增加重大心脏畸形的检出率[20,40,42,62,63]。加入流出道切面可以更容易识别圆锥动脉干畸形，如法洛四联症，大动脉转位，右室双出口和永存动脉干等异常[43 - 46,64 - 69]。

超声技术

横切面扫描系从胎儿腹部（标准腹围平面）向头部方向移动探头，经四腔心切面后，继续向上纵隔方向移动探头的扫描手法（扫查法)，可系统评估胎儿心脏，获得多幅超声图像，从而确定 LVOT，RVOT，3V 和 3VT 等流出道切面是否正常[70]，（图 3)。理想情况下各流出道切面较易显示。但在常规筛查时显示所有胎儿的所有流出道切面，技术上未必可行。因此，最好能熟悉各流出道切面。

将探头向胎儿头部滑动（或倾斜）可获得左、右室流出道切面（扫查法)（图 4)，先由四腔心切面获取主、肺动脉起始部的正常交叉。也可观察具体肺动脉分叉（附录 S1 图组 1,2)。此外，旋转技术也可以用以评估胎儿流出道切面[41]（附录 S2 图组 1)。方法是将探头自四腔心切面向胎儿右肩旋转。该技术在声束与室间隔垂直时较易操作。较之扫查法，其技术要求略高，但可更好显示 LVOT，特别利于观察室间隔与主动脉的连续性。它还能显示整段升主动脉，而扫查法仅能显示主动脉的近端部分。无论何种技术，取得 LVOT 切面后，应将探头向胎儿头部倾斜，直到观察到肺动脉与主动脉近乎垂直为止。

将探头进一步从右室流出道向胎儿头部滑动或倾斜可获得包括 3V 和 3VT 切面在内的其他主、肺动脉切面。这些切面可显示两条大动脉与上腔静脉、气管之间的关系，也可同时观察动脉导管弓和主动脉弓的情况[64-67]。

*左室流出道(LVOT)切面：*检查须确认主动脉从形态左心室发出（图 5），室间隔与主动脉前壁连续。主动脉瓣活动自如、瓣膜无增厚。可沿主动脉，追踪主动脉弓、及其走向颈部的三条动脉分支。但识别主动脉弓的血管分支，并非常规心脏检查的组成部分。LVOT 切面有助于发现出口室间隔缺损和圆锥动脉干异常等，这都是单纯四腔心切面检查无法观察到的。

*右室流出道(RVOT)切面。*检查须确认肺动脉从形态右心室发出（图 6）：肺动脉自右室发出后，朝向位于其后方的主动脉左侧走行，并跨越升主动脉起始部，与主动脉几乎呈直角。在胎儿期，肺动脉通常比主动脉根部稍粗。如图 6 所示，在该平面通常可在主动脉右侧发现上腔静脉，这与 Yoo 等描述的 3V 切面相似[64]。

肺动脉瓣须活动自如、瓣膜无增厚。从右室流出道发出的血管，须在发出后不久即向左右分支，据此可确认肺动脉。左右分支的先后顺序为，先发出右肺动脉，再发出左肺动脉。但因胎位影响，肺动脉分支的观察可能存在困难。正常肺动脉将继续向左侧远端行走，入动脉导管后，与降主动脉连结（图 6 和附录 S1）。

*三血管(3V)切面和三血管气管(3VT)切面：*推荐观察这两个切面。尽管技术上观察所有病例的 3V 和 3VT 切面未必可行，但应尽量将它们纳入到常规心脏筛查中。

3V 和 3VT 切面显示三条血管、血管之间及气道（气管）的相互关系。Yoo 等[64]描述了如何根据 3V 切面评估肺动脉、升主动脉、上腔静脉，及它们的相对大小和关系（图 7）。简单来说，评估包括血管数量、大小、走向和排列。自左及右，排列的血管依次为肺动脉、主动脉和上腔静脉。肺动脉最靠前，上腔静脉最靠后。血管直径从左至右依次变窄，即肺动脉比主动脉粗，主动脉又比上腔静脉粗。某些四腔心切面表现正常的典型心脏异常，如完全性大动脉转位，法洛四联症和肺动脉闭锁伴室间隔缺损等，其 3V 切面多数有异常发现。Yagel 等[67]随后描述的 3VT 切面，比 3V 切面更接近头部，可更清楚地显示主动脉横弓（’主动脉弓切面’），并提示其与气管的关系。气管常表现为围绕液性区域的、小的高回声环。正常情况下，动脉导管弓和主动脉弓均位于气管左侧，二者在汇入降主动脉时呈“V”形（图 8）。主动脉弓的位置高于动脉导管弓（近胎儿头部），因此须调整探头角度、离开与四腔心切面平行的平面，方能在图像中同时见到这两个结构。3VT 切面对检出如主动脉缩窄、右位主动脉弓、双主动脉弓、血管环等病变有很大帮助。

彩色多普勒

虽然本指南未将彩色多普勒超声列为必须的检查项目，但鼓励掌握其应用并将之纳入常规筛查[71]。彩色多普勒血流成像技术是胎儿超声心动图不可或缺的部分，对诊断 CHD 的作用不容低估。如操作者能胜任该技术，也可在常规筛查中运用。

该技术有助于显示各心脏结构、提示异常血流信号；它是针对肥胖孕妇， 用来评估胎儿心脏结构的宝贵工具[72]；亦利于进一步增加低风险妊娠中胎儿重大 CHD 的检出率[46,73]。

理想的彩色多普勒超声设置，包括使用较小的取样框（目标区域）宽度以减少对帧频的影响，选择适当的脉冲重复频率、低彩色余辉和足够的增益设定，以显示流经瓣膜和血管的血流（见附录 S2）。

胎儿超声心动图

当怀疑胎儿合并 CHD，或超声筛查未见正常四腔心及流出道切面，又或者存在胎儿 CHD 的高危因素时，须进行胎儿超声心动图检查。有关该检查的具体细节，此前已有发表[26]，本文不作赘述。很多产前检出的 CHD 个案并未合并高危因素或心脏外异常[63]；筛查的重要性由此可见。医疗从业者尚须熟知转诊进行全面胎儿心脏评估的指证[74]。例如，若胎儿颈项透明层在 11-14 孕周时厚度大于 3.5 毫米，即使其在随后的检查中恢复至正常，也应为胎儿安排详细的心脏评估[75-78]。

胎儿超声心动图检查应由熟悉 CHD 产前诊断的专家进行。除基本筛查提供的相关信息外，还须进一步提供包括内脏心房位置、体静脉和肺静脉连接、卵圆孔、房室连接、心室大动脉连接、大血管关系，以及主动脉弓和动脉导管弓矢状切面等在内的详细的心脏结构及功能分析。

其他传统超声技术也可用于胎儿心脏检查。例如，多普勒超声检查可用于测量血流速度或检出流经瓣膜或心腔的异常血流信号。M 型超声心动图亦是分析心脏节律、心功能及室壁厚度的重要手段。新出现推广的技术，如组织多普勒和容积超声（三维/四维/时间空间相关成像（STIC））等，也可用于较详细的胎儿心脏结构及功能评估。已证实 4D 胎儿超声心动图有助于包括圆锥动脉干畸形、主动脉弓畸形和肺静脉异位引流在内的多种复杂心脏畸形的诊断评估[79-81]。还有一些超声检查模式，如斑点追踪技术等，目前主要用于研究，将来可能成为胎儿心脏功能评估的重要临床工具。

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