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在线全文

胎儿食管闭锁的诊疗挑战：从产前诊断、手术干预到预后管理*

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【摘要】 先天性食管闭锁（esophageal atresia, EA）是严重的消化道发育畸形，其发生率约占活产儿的1/2 500～1/3 000，其特征为食管连续性中断，常伴发食管气管瘘（tracheoesophageal fistula, TEF）。EA病因复杂，随着产前影像学技术、胎儿医学及新生儿外科技术的不断发展，其产前检出率、围产期管理水平及术后预后均得到改善。但EA的临床处理仍存在相应的挑战，主要包括：产前诊断技术难以高效检出最常见的C型EA，个体化手术时机缺乏量化的决策标准，以及预后评估偏重近期解剖重建而忽视远期生活质量。本文系统阐述了EA在病因学、产前诊断策略、外科治疗及预后评估方面的最新研究进展。随着EA患儿整体生存率的显著提升，未来的临床研究与疾病管理亟需发生转变：从单纯关注外科解剖重建，转向突破产前诊断的技术瓶颈、优化个体化手术时机决策，弥合近期解剖重建与远期生活质量之间的鸿沟。此外，应进一步深化多学科协作模式，建立贯穿产前至成年的全生命周期随访与干预规范，以期全面改善患儿的远期生理与心理预后。

【关键词】 食管闭锁 胎儿 产前诊断 围生期综合护理 手术 预后 综述

Clinical Challenges in the Diagnosis and Management of Fetal Esophageal Atresia: From Prenatal Diagnosis and Surgical Intervention to Prognostic Management

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[Abstract] Congenital esophageal atresia (EA), a severe malformation of the digestive tract, has an incidence of approximately 1 in every 2 500 to 3 000 live births. It is characterized by an interruption in the continuity of the esophagus and is often accompanied by tracheoesophageal fistula (TEF) . The etiology of EA is complex. With continuous advancements in prenatal imaging technology, fetal medicine, and neonatal surgery, the prenatal detection rate, perinatal management, and postoperative outcomes of EA have improved. However, the clinical management of EA still faces significant challenges, including, primarily, the inefficiency of prenatal diagnostic techniques in detecting the most common Type C EA, the lack of quantitative decision-making criteria for individualized timing of surgical intervention, and prognostic evaluations that overemphasize short-term anatomical reconstruction while neglecting long-term quality of life. This article systematically reviews the latest research advances in the etiology, prenatal diagnostic strategies, surgical management, and prognostic evaluation of EA. Given the significant improvement in the overall survival rate of pediatric patients with EA, future clinical research and disease management urgently require a paradigm shift—transitioning from a sole focus on surgical anatomical reconstruction to overcoming technical limitations in prenatal diagnosis, optimizing decision-making for individualized surgical timing, and bridging the gap between short-term anatomical reconstruction and long-term quality of life. Furthermore, it is imperative to further strengthen the multidisciplinary collaborative model and establish standardized, whole-life-cycle follow-up and intervention protocols spanning from the prenatal period through adulthood, with the goal of comprehensively improving the long-term physiological and psychological outcomes of these patients.

[Key words] Esophageal atresia Fetus Prenatal diagnosis Perinatal care Surgery Prognosis Review

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食管闭锁(esophageal atresia, EA)是一种严重的先天性消化道畸形,发病率约为活产儿的1/2 500 ~ 1/3 000[1-2],其病理特征为食管连续性中断,常合并食管气管瘘(tracheoesophageal fistula, TEF)[3]。近年来,产前诊断技术的进步使EA的产前检出率提升至10% ~ 40%[4-5],诊断依据亦从“胃泡小”“羊水过多”等非特异性间接征象,逐步转向“食管上段盲袋征”“食管下咽部扩张”等更具特异性的直接征象。同时,磁共振成像(magnetic resonance imaging, MRI)凭借其优越的软组织分辨率,显著提高了产前诊断的准确性。外科技术的持续发展也有效改善了患儿的生存率[6-7]。然而,尽管理论与技术层面不断突破,EA的临床诊疗与研究仍面临诸多挑战,突出表现为漏诊率较高、伴发畸形复杂(如VACTERL联合征[8])及术后并发症多样。当前,该领域亟待解决以下三大核心问题:其一,产前诊断能力与临床需求之间存在明显落差——尽管现代影像学手段日益精密,但临床最常见且最亟需产前干预的C型EA,其产前检出率反而长期处于较低水平;其二,个体化手术时机的决策缺乏客观依据——在外科治疗从传统术式向个体化限期手术转型的过程中,如何精准权衡心肺并发症风险与微创手术适应证,迄今尚无公认的量化决策标准或预测模型;其三,预后评估体系有待拓展——传统疗效评价多聚焦于术后消化道的近期解剖修复及并发症控制,而患儿成年后面临的主要健康负担,往往集中于心理健康、社会融入等长期被忽视的远期生活质量维度。基于上述背景,本文拟系统综述胎儿EA的病因学研究现状、产前诊断策略、外科治疗方式及预后评估体系,并针对三大核心问题进行批判性分析,解构现有文献证据,旨在为EA的早期精准诊疗、个体化手术时机的量化决策依据及覆盖产前至成年期的全生命周期照护提供新的临床参考。

1 遗传咨询与环境归因的临床瓶颈

EA的确切病因尚未完全明确[9],目前认为其发生源于遗传易感性与环境暴露因素之间的复杂交互作用。流行病学资料表明,约50%的EA/TEF患者合并其他先天性异常[10],其中以VACTERL综合征和CHARGE综合征最为密切关联[11-12]。VACTERL综合征的诊断需至少存在三种结构畸形,可累及椎体、肛门、心脏、气管食管及肾脏等多个系统;据统计,50% ~ 80%的VACTERL综合征患者合并EA[13]。CHARGE综合征则是一种单基因疾病,多数病例由*CHD7*基因功能丧失性致病变异所致[14]。因此,对于产前疑似EA的胎儿,建议进行*CHD7*基因测序,以协助明确病因诊断。

鉴于超过50%的EA患儿常合并其他系统畸形或复杂症候群,产前是否应对疑似EA胎儿常规开展全外显子组测序(whole exome sequencing, WES),近年来已成为一个备受争议的议题,核心围绕“诊断效能”与“临床实用性”之间的平衡展开。Zhang等[15]的最新研究指出,家系全外显子组测序(Trio-WES)虽能为超声结构异常的胎儿额外提供约22.7%的单基因致病变异检出率,从而有助于精准遗传咨询,但其在临床常规应用中仍面临检测成本高昂、报告周期较长等现实障碍。该研究在权衡卫生经济学效益与临床获益后,强调应根据超声表型实施“分层诊断策略”:WES在多系统结构畸形胎儿中的诊断阳性率显著更高(可达37.5%)。将这一策略延伸至EA的临床管理,目前多数学者建议:对于影像学高度提示EA且合并其他系统畸形的胎儿,应优先或同步进行WES以排查复杂遗传综合征;而对于孤立性EA胎儿,鉴于染色体微阵列分析(chromosomal microarray analysis, CMA)技术成熟、检测高效且成本效益更优,仍应作为一线诊断工具。常规开展WES的卫生经济学价值,尚有待大样本、多中心的前瞻性队列研究进一步验证。

此外,现有研究资料表明,孕妇饮酒、被动吸烟、孕产妇糖尿病、高龄(> 35岁)以及药物暴露(如外源性激素、甲硫咪唑、除草剂、杀虫剂等)亦与EA的发生存在相关性[16-17]。然而,当前文献多限于现象学层面的关联性描述,对上述危险因素的归因危险度缺乏精准的定量评估。剔除混杂因素后,特定环境暴露的消除究竟能在多大程度上降低EA的发生风险,目前尚缺乏明确答案。尤其值得关注的是,基于中国人群的大样本分析数据严重不足,这一缺口也为未来病因学及公共卫生干预研究指明了核心方向。

胚胎期食管与气管的正常分离及形态构建若受遗传或环境因素的干扰,则可能导致不同类型的EA。目前,临床分型主要采用Gross分型法,依据畸形的解剖关系,分为以下5型:A型,食管有两个盲端,不合并TEF,约占8%;B型,食管近端与气管相通,合并为TEF,盲端在远端,约占1%;C型,食管盲端在近端,远端与气管相通,合并为TEF,此型最常见,约占84%;D型,食管两端均与气管相通,形成两处TEF,约占1%;E型,食管全程通畅,但食管与气管管壁间存在异常交通支,形似“H”,约占4%[12, 18]。

2 常见C型EA的高漏诊率困境

2.1 传统超声筛查的局限性

超声检查是产前筛查EA的主要无创影像学手段[19],但其诊断准确性易受胎儿体位、羊水量及操作者经验等多种因素影响,导致产前漏诊率较高。文献报道,产前超

声对EA的检出率介于13%～65%之间，波动范围较大[5, 20-22]，相当一部分病例直至出生后方可确诊。研究表明，超声的检测敏感性还与EA的解剖类型密切相关。Pollack等[23]回顾性分析费城儿童医院2013年1月—2024年12月收治的产前疑似EA或TEF病例，结果显示产前超声诊断EA的总体灵敏度为48%，其中A型可达100%，而临床最常见的C型仅为36%。这意味着，近70%的C型EA病例在宫内未被识别，产前漏诊问题尤为突出。

剖析C型EA产前漏诊率偏高的原因，主要在于其解剖特征削弱了超声筛查的预警信号。在该型中，因存在TEF，羊水可通过瘘管进入胎儿胃内，致使约10%的C型EA病例胃泡大小表现正常，从而掩盖了“胃泡小或未见显示”这一关键间接征象。近年研究表明，相较于间接征象，直接征象（如食管上盲袋征、下咽部扩张）在EA产前诊断中具有更高的特异性。其中，食管上盲袋征表现为胎儿颈根部喉后区域的囊状无回声结构，系闭锁段以上食管因吞咽羊水而代偿性增厚、扩张所致，且随吞咽动作呈间歇性显现[19, 24]。然而，在缺乏典型间接征象提示的情况下，要求超声医师在常规筛查中精准捕捉上述直接征象，较大程度依赖于操作者经验及羊水透声条件。尽管Tracy等[25]建议对存在高危征象的胎儿延长观察时间至30～50 min，并动态监测胃泡大小、进行多切面反复扫描，但在当前超声门诊高负荷运转的现实条件下，该建议的临床可操作性受到较大限制。

为弥补超声对直接征象显示的不足，临床常借助软组织分辨率更高的胎儿MRI。研究指出，MRI可清晰显示食管下段盲袋状扩张，且其显影不受瘘管干扰，亦是诊断EA的重要影像学依据[26]。然而需注意，在MRI实际扫描中，C型EA远端瘘管所致胃泡假性正常化同样会干扰影像医师的判断；此外，食管盲袋的精准捕捉高度依赖胎儿在有限扫描窗内的间歇性吞咽动作。因此，当超声筛查提示可疑EA而诊断尚不明确时，胎儿MRI不宜仅被视为单纯追求提高“确诊率”的补充检查手段[26-29]。

2.2　胎儿MRI在风险分层中的关键作用

胎儿MRI应被提升为临床关键的风险分层工具，其核心价值不仅在于借助软组织高分辨率明确食管盲袋及食管连续性中断，更在于精准测量“食管盲端间距”这一关键解剖学参数。该指标直接决定患儿出生后是否需接受复杂的长间隙食管延长术（如Foker术）甚至食管替代术。通过将影像学参数与手术可行性及术后并发症风险相关联，胎儿MRI可为产前咨询提供更为客观、精准的预后评估，既为家庭妊娠决策提供重要参考，也为围产期重症医疗资源的合理配置提供循证依据。

传统二维超声对胎儿EA（尤其是最常见的C型）的筛查主要依赖羊水过多或胃泡异常等间接征象，诊断敏感度较为有限。近年来，胸腹部三维容积超声及高级透明渲染技术的应用，推动产前诊断从“间接推断”逐步迈向“直接可视化”。该技术不仅能直观显示食管盲袋征并评估盲端间距，还可联合多平面重建及具有高阴性预测值的胎儿MRI，进一步提升EA解剖分型及合并畸形（如VACTERL综合征）的检出率，从而为高危孕妇的及时转诊、新生儿早期气道保护及精准外科干预提供重要的影像学依据[30-32]。

3　围产期稳定与个体化手术权衡的挑战

3.1　围产期管理

围产期管理的核心目标在于实现患儿从宫内到宫外的平稳生理过渡，为后续外科干预奠定最佳基础。一旦产前高度疑诊EA，应及时组建多学科团队（multidisciplinary team, MDT）进行综合评估，制定个体化分娩与转诊方案，确保患儿在具备高水平新生儿重症监护与小儿外科救治能力的医疗中心出生[33]。

合并EA胎儿终止妊娠时机至今尚无确切共识及指南给出建议，需要依据个体化情况进行临床处理。

出生后的精确诊断与病情稳定是抢救EA患儿的黄金窗口。由于产前检出率仍面临挑战，部分病例宫内未被识别，新生儿娩出后若表现为口腔溢出泡沫样唾液、初次喂养即发生呛咳与发绀，应高度怀疑本病[34]。简易确诊方法为尝试置入鼻胃管，若其在咽喉部（距口鼻约9～11 cm处）受阻，且X线片显示管端在胸廓入口（T2～T4水平）卷曲，即可临床诊断。确诊后应立即经口或鼻置入Replogle管至食管近端盲袋，持续低负压吸引以防止唾液反流所致致命性吸入性肺炎；同时需审慎管理气道，避免常规无创正压通气，防止气体经食管气管瘘进入胃肠道而加重腹胀。在维持水电解质平衡的基础上，需紧急完善超声心动图等检查，重点评估是否合并严重心脏畸形（发生率较高）及明确主动脉弓位置[35-37]。上述围产期稳定措施的目的在于优先处理危及生命的合并症，从而为临床权衡个体化手术时机、决定采用微创腔镜或传统开胸术式，提供关键的决策窗口与生理储备。

3.2　个体化手术时机与微创术式的决策

EA的外科治疗属于复杂性手术范畴，当前治疗策略趋向于微创化与个体化。所有确诊患儿均具有绝对手术指征，手术时机已从传统的急诊手术逐步转变为个体化的限期手术，但最佳手术时间窗目前尚无定论[38-39]。临床实践中，患儿出生后首先进入“稳定期”，此阶段优先处理

危及生命的合并症,尤其是需要干预的严重先天性心脏病,同时积极纠正水电解质紊乱、控制肺炎,并评估其他重要畸形(如VACTERL关联畸形)。待患儿一般状况稳定后,应尽早施行根治性吻合术。对于长间隙EA或极低出生体重儿中病情不稳定者,则常采用分期手术策略[40-42]。

EA的经典外科术式为“一期吻合联合瘘管结扎”。近年来,胸腔镜下食管吻合术因创伤小、美观性好而被广泛应用,并已成为多数医疗中心的首选术式,然而其在并发症控制方面是否具有绝对优势,目前学术界仍存争议[26]。Wang等[38]的最新研究指出,胸腔镜手术虽具微创特性,但学习曲线陡峭,对术者技术要求极高,且部分研究显示其术后吻合口漏与狭窄的发生率甚至高于传统开胸手术。该研究进一步探讨了手术时机的抉择逻辑,明确指出对于合并术前呼吸道感染或严重心血管畸形的C型EA患儿,不宜盲目追求紧急腔镜干预;适度延迟手术(≥5 d)不仅不增加围术期呼吸道感染风险,反而有助于患儿平稳度过生理适应期,同时降低组织含水量、增强食管壁缝合耐受性,从而显著减少术后因重度吻合口狭窄而需行球囊扩张的频率。对于极低出生体重儿(出生体质量<1 500 g),最佳手术方式与时机尚存争议。Shimizu等[41]的多中心队列研究证实,对此类高危患儿采用分期手术策略——先行胃造瘘、瘘管结扎或食管绑扎等姑息性干预,待生理指标稳定且体质量达目标值(约3 000 g)后再行二期根治性吻合——是一种安全、有效且能显著改善总体生存率的方案。尽管未来仍需大规模前瞻性随机对照试验提供更高循证等级证据,但现有结论已充分提示,临床实践中不应盲目追求早期微创,而应建立严谨的决策算法,以筛选禁忌人群、合理规划手术时机与方式。

对于长间隙型EA或多次手术失败的复杂病例,食管磁加压吻合术与分期食管延长术等新兴技术提供了突破性选择,有助于利用自体食管组织实现重建,从而避免替代器官的使用。食管磁加压吻合术由Zaritzky等[43]于2009年首次提出,历经多次技术改良与临床推广,但目前仍面临术后食管顽固性狭窄这一严重并发症,多数患者需反复接受内镜下球囊扩张治疗[43-46]。该术式重建后的食管蠕动功能及远期抗反流效果是否真正优于传统延迟吻合术或机械牵引术(如Foker术),目前尚缺乏高质量随机对照试验证据支持。在长间隙EA的治疗中,已有多种术式被报道,主要包括各类食管替代术与食管延长术。早期治疗多倾向于采用食管替代手术,如利用胃、结肠或小肠等组织代食管;近年来,保留自体食管、优先重建或修复原始食管的理念逐渐获得更多医师的倡导[42, 47]。以延迟吻合与机械牵引(如Foker术)为代表的食管延长技术,可使患者在不依赖替代器官的前提下完成重建,从而有效规避与食管替代术相关的长期并发症[48]。目前,延迟初级修复是长间隙EA最常用的术式之一[49-51]。

在长间隙EA或复杂病例的外科治疗中,胸腔镜手术的核心优势在于可提供高倍放大的手术视野,有利于后纵隔区域的精细解剖。然而,该微创技术的固有局限性在于缺乏直接触觉反馈,且新生儿胸腔操作空间狭小,在应对长间隙EA等需复杂解剖重建的病例时,对术者的技术水平要求极为严苛。相比之下,传统开胸手术不仅能提供直观的触觉反馈与更充裕的操作空间,而且在术中遭遇大出血或血流动力学急剧恶化等危急情况时,便于迅速实施有效抢救。因此,在临床实践中,胸腔镜与传统开胸并非绝对对立的选项,而是互为补充的治疗策略。术式选择应摒弃片面的“唯微创论”,须在综合评估术者腔镜操作经验、患儿局部解剖复杂度及全身心肺耐受能力的基础上审慎决策。临床决策应始终以保障患儿生命安全为首要前提,在确保医疗安全的基础上追求创伤最小化。当胸腔镜操作遭遇技术瓶颈或患儿生命体征出现失代偿时,果断中转开胸应被视为保障围手术期安全及改善远期预后的基本原则。

4 近期解剖重建与远期生活质量的鸿沟

合并EA/TEF患儿的临床结局主要取决于是否伴发其他先天性异常,而非EA/TEF本身[12]。其中,严重心脏畸形是导致死亡率增加的最显著因素,染色体异常次之[52-53]。此外,低出生体质量亦为重要危险因素,出生体质量越低,死亡风险越高[52, 54]。

EA外科治疗的围手术期并发症风险较高,主要包括吻合口狭窄、吻合口漏及瘘管复发,文献报道其发生率分别为32% ~ 59%、5% ~ 20%和3% ~ 10%[55]。Kum等[56]对1997—2022年间接受手术修复的31例EA患者进行长期随访,结果显示一期修复成功率达90.3%,早期严重并发症(如吻合口漏)发生率较低(6.45%),但长期预后面临显著挑战:其一,术后并发症负担较重,需反复扩张治疗的食管狭窄占35.5%,胃食管反流症状发生率为35.5%,其中22.6%因药物控制不佳最终接受胃底折叠术;其二,健康相关生活质量全面受损,采用儿童生活质量量表(PedsQL)及胃肠生活质量指数(GIQLI)评估发现,患者总体评分显著低于健康人群,且心理社会领域损害(得分63.3)较身体领域更为突出。

随着多MDT模式的推广及外科手术技术的不断进步,EA患儿的生存率已显著提高[53]。在此背景下,EA的治疗终点不应再局限于吻合口的愈合,而应延伸至患儿

成年后能否实现正常的进食功能与社会参与。然而，该类患儿的全程管理仍面临诸多挑战，不仅需应对围手术期短期并发症，还需处理远期并发症，包括胃食管反流病、食管运动功能障碍、食管化生及食管癌风险，以及反复呼吸道感染、慢性咳嗽、持续性肺功能异常和哮喘等胃肠道与呼吸系统问题[57-58]。鉴于EA常作为复杂症候群的一部分，其预后管理需超越食管本身，突破传统外科随访的局限，构建系统化的远期过渡期管理模式。应常态化整合儿童心理科、临床营养科及康复医学科等多专业力量，制定覆盖新生儿期、学龄期至青春期的全生命周期随访规范，从而切实缩小近期外科疗效与患儿远期生活质量之间的差距。

5　小结及展望

胎儿EA是一种源于前肠发育异常的复杂先天性畸形，其病因涉及遗传因素与环境暴露的交互作用。在产前诊断中，超声作为一线筛查手段，需综合直接与间接征象进行系统评估；对于疑难病例，胎儿MRI不仅能提供高分辨率的定性诊断，还能客观测量“食管盲端间距”，并系统排查心血管及泌尿系统等严重伴发畸形。这一精准的影像学预判，为产前优生咨询、围产期重症资源配置及新生儿期个体化手术方案的制定提供了不可替代的解剖学依据。未来，随着超声与MRI技术的深度融合，以及三维容积超声与高级透明渲染技术的广泛推广，产前诊断的敏感性与特异性有望进一步提升。

在外科治疗方面，建立个体化处理策略是基础，胸腔镜下“一期吻合联合瘘管结扎”作为经典术式可改善预后，但术后吻合口狭窄、吻合口漏及胃食管反流等并发症仍是长期管理的难点。针对长间隙等复杂病例，食管延长术与磁加压吻合术为利用自体食管实现原位重建、规避替代器官所致远期功能障碍提供了新的选择。需注意的是，个体化手术时机的抉择仍缺乏可量化的风险预测工具。如何权衡心肺合并症、低出生体质量及微创适应证等多重变量，精准确定最佳手术窗口，是当前临床决策的最大痛点。为此，亟需依托多中心前瞻性队列，构建包含解剖参数、生理指标及术者经验在内的决策预测模型，为“何时做、如何做”提供循证依据。

在预后管理层面，现代医学对EA的认知与管理应超越传统的“外科解剖重建”范式，转向贯穿“产前—新生儿—学龄期—成年期”的全生命周期系统管理工程，未来应建立常态化的MDT协作机制，整合心理、营养、康复等专业力量，制定从新生儿期延续至成年的过渡期照护计划，弥合近期解剖重建与远期生活质量之间的鸿沟，最终实现患儿心身的全面康复。

*　　*　　*

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