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· 多学科诊疗研究 ·

直肠子宫内膜异位症多学科诊治1例报告并文献复习

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摘 要

深部浸润型子宫内膜异位症(DIE)是指子宫内膜异位病灶浸润腹膜下 ≥ 5 mm, 常累及宫骶韧带、阴道直肠隔、阴道穹隆及直肠壁等部位, 因临床表现缺乏特异性、影像学特征不典型而易误诊。本研究报告1例术前经内镜超声引导下细针穿刺活检(EUS-FNA)确诊的直肠DIE患者, 结合文献回顾, 总结其多学科团队(MDT)诊治经验。患者为35岁女性, 因排便困难入院, 经MRI及EUS-FNA确诊为直肠DIE。经结直肠肛门外科、妇科及泌尿外科联合讨论后实施腹腔镜直肠病变切除、乙状结肠-直肠吻合及预防性回肠造口术, 术后病理证实切缘阴性, 4个月后成功还纳造口。随访21个月, 患者症状明显缓解, 未见复发。结果提示, EUS-FNA在DIE的早期诊断中具有重要价值, 而以结直肠肛门外科为主导、妇科与泌尿外科协同的MDT模式可显著提高手术安全性和根治性, 对复杂盆腔内异症的精准诊治具有重要指导意义。

关键词

子宫内膜异位症; 直肠; 内镜超声引导细针穿刺; 直肠切除术

中图分类号: R657.1

Multidisciplinary management of rectal endometriosis: a case report and literature review

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Abstract

Deep infiltrating endometriosis (DIE) is defined as endometriotic lesions infiltrating ≥ 5 mm beneath the peritoneum, commonly affecting the uterosacral ligaments, rectovaginal septum, vaginal vault, and rectal wall. Due to nonspecific clinical manifestations and atypical imaging features, DIE is often misdiagnosed. This study reports a case of rectal DIE diagnosed preoperatively by endoscopic ultrasound-guided fine-needle aspiration (EUS-FNA) and summarizes the multidisciplinary treatment experience in conjunction with a literature review. A 35-year-old woman was admitted for defecation difficulty. MRI and EUS-FNA confirmed rectal DIE. After multidisciplinary team (MDT) evaluation involving colorectal, gynecologic, and urologic specialists, laparoscopic resection of the rectal lesion,

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sigmoid-rectal anastomosis, and protective ileostomy were performed. Pathology confirmed rectal DIE with negative margins. The stoma was successfully reversed 4 months later, and no recurrence was observed during 21 months of follow-up. These findings highlight the pivotal role of EUS-FNA in early diagnosis and demonstrate that an MDT approach led by colorectal surgeons can significantly enhance surgical safety and completeness, providing valuable guidance for the individualized management of complex pelvic endometriosis.

Key words

Endometriosis; Rectum; Endoscopic Ultrasound-Guided Fine Needle Aspiration; Proctectomy

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子宫内膜异位症即子宫内膜的腺体和间质出现在子宫腔以外的部位,可能累及多器官的一种常见妇科良性疾病。目前认为子宫内膜异位症的病因不明,可能与子宫内膜播散、血管新生、上皮间质转化、孕激素抵抗、炎症等多种因素密切相关。深部浸润型子宫内膜异位症(deep infiltrating endometriosis, DIE)是指子宫内膜异位症病灶浸润腹膜下的深度 ≥ 5 mm,包括位于宫骶韧带、阴道直肠隔、阴道穹隆、直肠或者结肠壁等位置的病灶。子宫内膜异位症患者累及结直肠的发生率约为8%~12%^[1],其临床表现缺乏特异性,以腹痛、腹泻、便血、排便困难等消化道症状为主(部分患者的症状可能和月经周期相关)。此外,子宫内膜异位组织具有一定的浸润性,可通过反复炎症、纤维增生导致肠黏膜糜烂、溃疡、出血、形成肿块,因此与炎症性肠病、肠癌等鉴别困难,经验不足的消化内科、胃肠外科医师往往会发生误诊或延迟诊断。本例患者的术前诊断、手术思路有一定新颖性,特与同行分享。

1 病例介绍

患者 女,35岁。因“排便困难伴腹胀3个月余”于2023年8月28日收入武汉大学中南医院。患者主要表现为进行性加重的排便费力,无明显腹痛、发热及黏液脓血便。既往有痛经及“卵巢巧克力囊肿”病史。外院肠镜结果示距肛门5 cm肠腔狭窄,活检示慢性炎症。入院查体:腹稍膨隆,叩诊鼓音;肛门指检于距肛门约5 cm处触及环周质硬肿物,活动度差,直肠管腔狭窄;三合诊提示病变与阴道后壁关系密切。实验室检查提示血红蛋白102 g/L、血钾2.91 mmol/L、CA-125 173.5 U/mL。

2 多学科团队(multi-disciplinary team, MDT)诊疗过程

2.1 术前诊断阶段

放射科通过MRI精准评估病灶范围及与周围脏器关系,腹盆腔增强CT及直肠肿瘤MRI均显示直肠中段管壁环形增厚伴管腔狭窄,病灶与子宫后壁分界不清(图1A-B)。消化内科行结肠镜联合内镜超声(endoscopic ultrasonography, EUS)进一步确认距肛门6 cm处环形狭窄,EUS引导下细针穿刺活检(endoscopic ultrasound-guided fine-needle aspiration, EUS-FNA)取得组织送检(图1C),病理见子宫内膜腺体及间质,免疫组化ER(+),PAX8(+),确诊为直肠DIE(图2)。妇科根据患者痛经及卵巢巧克力囊肿病史,协助确认子宫内膜异位症背景及激素管理方案。

2.2 手术规划与实施

经结直肠肛门外科、妇科、泌尿外科共同参与MDT讨论,认为患者已出现不可逆性直肠狭窄伴不全肠梗阻,具备明确手术指征。术中由妇科医师协助放置举宫器,辅助术中定位;泌尿外科术中解剖辨认并保护双侧输尿管,特别是在病变与右侧输尿管粘连致密区域;结直肠肛门外科主导实施腹腔镜下病变直肠切除及吻合。手术采取“由外周向中心”的策略:先游离直肠后间隙及肛提肌上间隙,再拓展双侧侧方间隙,注意保护盆神经及输尿管;随后于病变远端寻找正常直肠阴道隔,打开阴道后穹隆作为解剖引导,逆向分离直肠与阴道(图3)。最终完成病变直肠切除+乙状结肠-直肠吻合+预防性回肠造口。

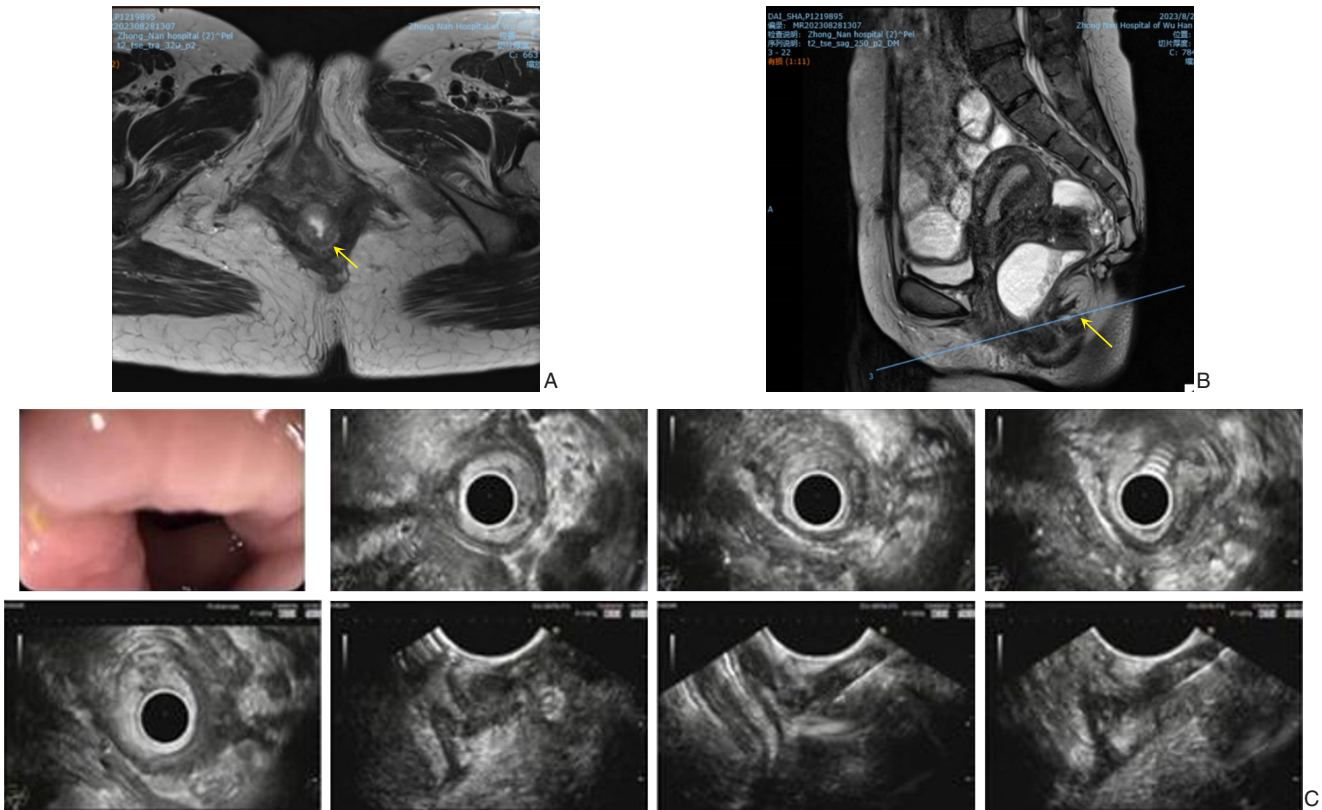


图 1 患者术前影像资料 A-B: 分别为盆腔增强 MR T2 轴位及 T2 矢状位,提示直肠中段管壁局限性环形增厚伴管腔明显狭窄,下缘距离肛缘约 75 mm,病变范围长约 20 mm,病灶与子宫后壁分界不清,局部子宫后壁呈混杂信号,内见小点状 T2 高信号影,其中橙色箭头指示处为病灶; C: EUS 示直肠距肛门 8 cm 可见环形狭窄,内镜可勉强通过,探头置直肠狭窄处可见直肠右侧壁不均匀增厚,管壁层次结构消失,最厚处 10 mm,突破固有肌层,可见明显回声欠均匀低回声囊肿样结构,与直肠壁分界尚清,给予 22 G 穿刺针在增厚直肠壁内抽吸组织条 3 次送检

Figure 1 Preoperative imaging findings of the patient A-B: Pelvic enhanced MRI (T2-weighted axial and sagittal views) showing focal circumferential thickening of the mid-rectum with significant luminal stenosis, the lower margin of the lesion is approximately 75 mm from the anal verge, spanning about 20 mm in length, the lesion shows ill-defined borders with the posterior uterine wall, which displays mixed signal intensity with scattered punctate T2 hyperintense foci (orange arrows indicate the lesion); C: EUS demonstrates a circumferential stenosis 8 cm from the anal verge, barely allowing endoscope passage, the right rectal wall appears irregularly thickened with loss of the normal layered structure (up to 10 mm), breaching the muscularis propria, a heterogeneous hypoechoic cystic area is observed, and three passes of fine-needle aspiration using a 22-gauge needle were performed for pathological sampling

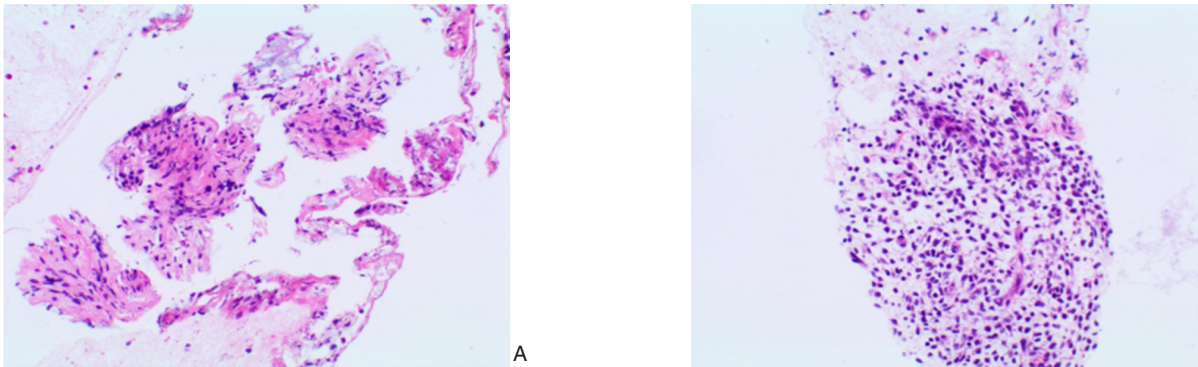


图 2 直肠 EUS-FNA 组织学检查可见少许子宫内膜腺体及间质 (HE×200) A: ER 阳性; B: PAX8 阳性

Figure 2 Histological examination of rectal EUS-FNA showing scant endometrial glands and stroma (HE×200) A: ER positive; B: PAX8 positive

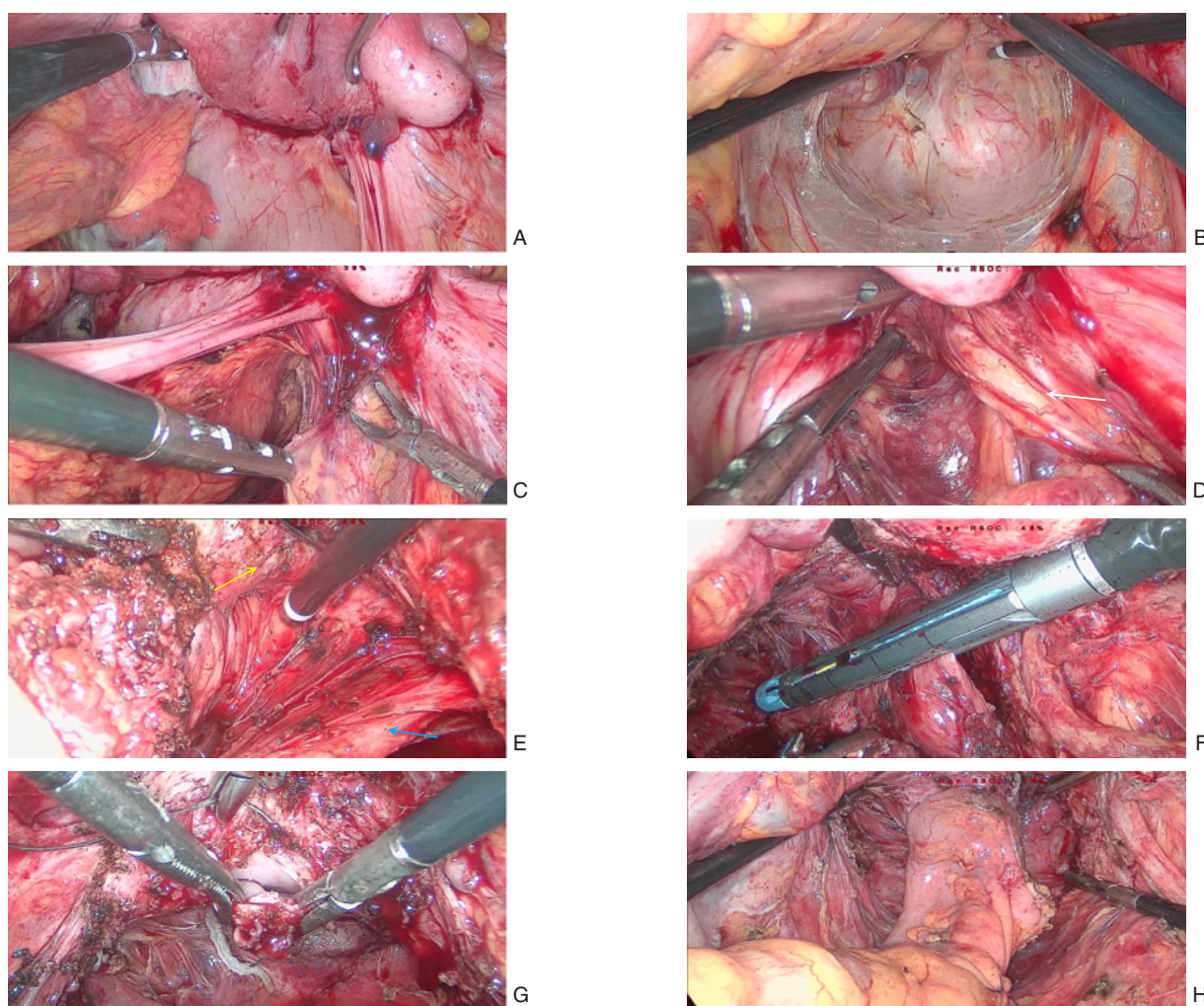


图3 手术过程 A: 显示病灶位于直肠阴道间隔, 直肠与阴道后穹隆、子宫颈形成极其致密的粘连, 并且与右侧输尿管关系不清; B: 先充分游离拓展直肠后间隙、肛提肌上间隙; C: 游离直肠右侧间隙, 从右侧输尿管腹下神经层面外侧拓展; D: 游离并保护右侧输尿管 (白色箭头), 跨越至输尿管腹下神经层面内侧进行直肠右侧间隙拓展; E: 反向自下而上切除病灶, 分离阴道 (黄色箭头) 和直肠 (蓝色箭头); F: 充分裸化直肠后予直线切割闭合器离断直肠; G: 倒刺线妥善缝合关闭阴道; H: 行结肠-直肠端-端吻合

Figure 3 Intraoperative procedure A: The lesion was located in the rectovaginal septum, forming extremely dense adhesions between the rectum, posterior vaginal fornix, and cervix, with an indistinct relationship to the right ureter; B: Dissection of the retrorectal and supraleator spaces; C: Mobilization of the right pararectal space, extending laterally from the ureter-hypogastric nerve plane; D: The right ureter (white arrow) was identified and protected, and the dissection continued medially across the ureter-hypogastric nerve plane; E: Retrograde dissection and separation of the vagina (yellow arrow) and rectum (blue arrow); F: After full mobilization, the rectum was transected using a linear stapler; G: The vaginal defect was closed securely with barbed sutures; H: An end-to-end colorectal anastomosis was performed

2.3 术后管理

结直肠肛门外科负责肠道功能恢复与造口护理; 妇科指导术后药物抑制内异症复发 (如 GnRH-a); 营养科纠正术前贫血与低钾; 护理团队实施疼痛管理与快速康复流程。患者术后 1 周出院, 病理确诊为直肠 DIE, 切缘阴性 (图 4)。术后 4 个月顺利还纳造口。截至末次随访 (造口还纳后 21 个月), 患者排便功能良好, 生活质量满意, 未见复发。

2.4 患者诊疗流程总结

本例患者的诊疗历程包括术前影像学及 EUS-FNA 明确诊断、MDT 团队联合制定手术方案、腹腔镜手术实施及术后综合管理等环节。通过多学科协作, 团队在保证病灶根治性切除的同时, 有效降低了手术风险, 实现了快速康复和长期随访下的良好预后。患者完整的诊疗时间轴见图 5。

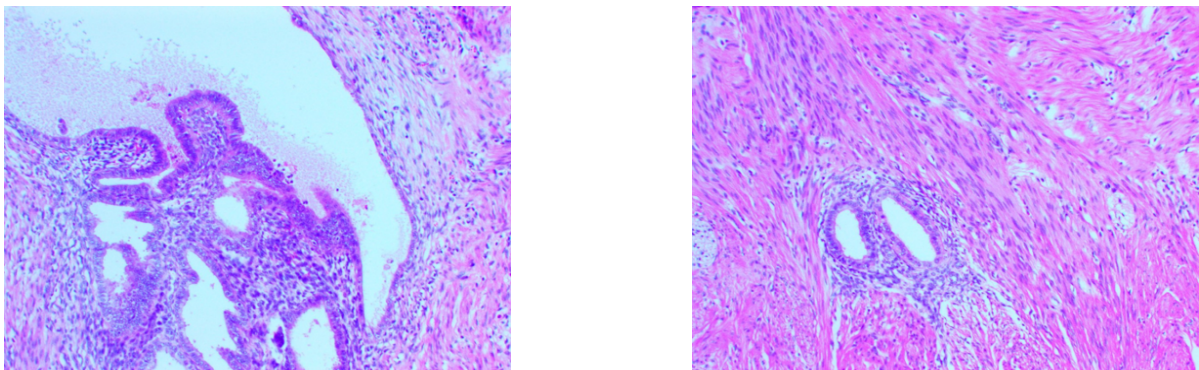


图 4 手术标本病理检查结果 (HE×100)

Figure 4 Pathological findings of the surgical specimen (HE×100)

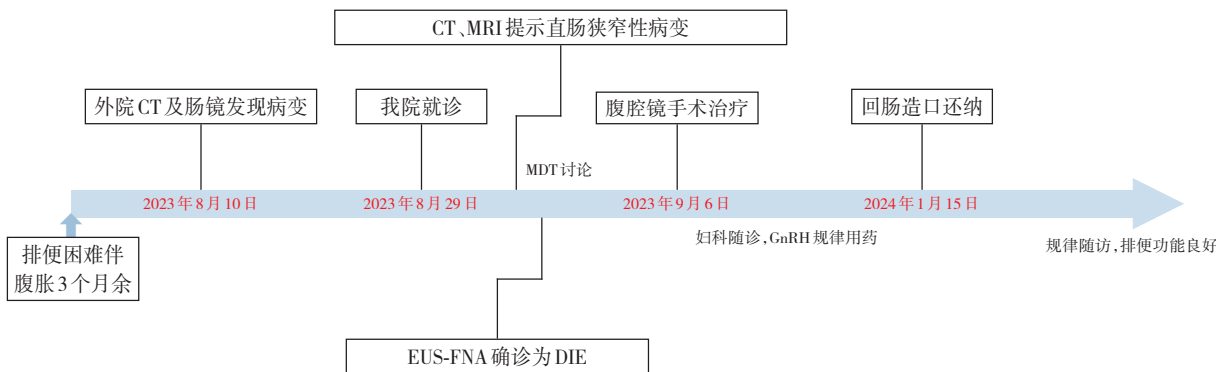


图 5 患者诊疗时间轴

Figure 5 Diagnostic and therapeutic timeline of the patient

3 讨 论

3.1 DIE 的流行病学特点

子宫内膜异位症是育龄期女性常见的妇科疾病，总体患病率约为 5%~10%，而 DIE 约占子宫内膜异位症患者的 8%~30%。常见受累部位包括子宫骶韧带、阴道直肠隔及直肠乙状结肠，肠道受累见于 3.8%~37% 的病例，其中直肠乙状结肠最为常见^[1-3]。DIE 患者多在 20~40 岁之间发病，临床症状缺乏特异性，诊断常延迟多年，误诊率较高。

3.2 辅助检查与 EUS-FNA 的诊断价值

目前子宫内膜异位症的诊断主要是在详细询问病史和仔细专科检查的基础上，结合超声、MRI、结肠镜甚至腹腔镜检查等多种辅助方法进行综合判断。超声检查包括经阴道超声（transvaginal ultrasound，TVS）、经直肠超声（transrectal ultrasound，TRUS），其对操作者经验要求较高，诊断具有一定的主观局限性^[4-10]。MRI 对 DIE 的敏感度和特异度均较高，可全面评估盆腔并辨析病灶与周围结构的关系，是泌尿系和肠道 DIE 的首选影像学检查方法^[11-12]。结肠镜虽可直观观察并行活

检，但由于病灶多位于肠壁浆膜及肌层，表现缺乏特异性，病理常提示慢性炎症，因此对肠道内异症的诊断准确率较低^[13]。

对于体格检查和影像学检查未发现异常但仍高度怀疑 DIE 的病例，可谨慎行诊断性腹腔镜明确病理学依据^[14]。近年来，EUS-FNA 在直肠 DIE 的诊断中显示独特优势。本例患者即通过 EUS-FNA 术前明确诊断，为精准治疗决策提供了关键依据。文献^[15-18]报道，EUS-FNA 对直肠 DIE 的诊断准确率可达 85%~95%，显著优于单纯影像学。因此，对于怀疑直肠（阴道隔）子宫内膜异位症的病例，除 MRI 等检查外，应常规考虑 EUS-FNA，该方法创伤小、风险低，既能判断病灶浸润深度，又可提供病理学证据，为手术方式和范围的确定提供重要参考。

3.3 手术治疗与 MDT 的价值

DIE 的治疗遵循个体化原则，主要包括药物治疗与外科手术。药物治疗（如 GnRH-a、孕激素等）的核心目标是控制疼痛症状，亦可用于术前辅助治疗以缩小病灶体积、降低手术难度，或作为术后长期管理策略，旨在延缓复发、控制远期症

状^[1]。对于已引发明显症状（如肠梗阻、严重疼痛或出血）的肠道DIE，外科手术被视为首选且最有效的根治性手段，其核心目标在于最大限度实现病灶的完整切除^[19]。

由于DIE病灶主要源于肠腔外的异位内膜组织侵袭浆膜及肌层，常规的内镜下球囊扩张或狭窄切开等方法往往难以触及并清除深层病灶，治疗效果有限。目前，针对肠道DIE的手术方式主要根据病灶浸润肠壁的深度和范围进行选择，主要包括：病灶削切术（shaving），适用于浅表或局灶性浸润；蝶形切除术（disc excision），适用于局灶性全层浸润但范围较小者；肠段切除吻合术（segmental excision），适用于环周性或长段（通常>3 cm）浸润以及合并肠腔狭窄的病例^[3,20]。

大量临床研究证实，这些术式总体安全有效，能显著改善患者症状与生活质量。然而，手术本身伴随的并发症发生风险不容忽视，其中出血、吻合口漏、直肠阴道瘘是最具代表性的术后并发症。多项研究对并发症发生率进行了报道，在接受肠段切除的DIE患者中，主要并发症发生率约为5%~10%，其中直肠阴道瘘的发生率在部分研究中可达2%~5%^[21-22]。另有前瞻性研究进一步指出，相较于蝶形切除，肠段切除吻合术虽然能更彻底地切除病灶，但其术后吻合口漏的风险相对较高（约1%~4%），而病灶削切术则可能因病灶残留导致更高的复发风险^[23-24]。腹腔镜手术因创伤小、恢复快，是目前首选^[22]。而术式的选择必须基于术前精准的影像学评估和术中细致的探查，在追求病灶根治性切除的同时，审慎权衡不同术式的获益与风险。

然而，DIE病灶多样，几乎“千人千面”，尤其当合并输尿管梗阻、肠狭窄时，常需输尿管吻合、肠段切除等联合手术，难度和风险显著增加，其手术方式需根据肠道浸润深度和范围进行个体化选择。Ferrero等^[25]强调，根据病灶的深度和范围，手术方式可从蝶形切除到节段性肠管切除，这要求术者具备全面的评估和决策能力。Vercellini等^[26]提出，对复杂DIE应采取“以病理机制为导向”的个体化手术策略，强调根据病灶浸润特点和解剖关系设计术式。MDT是提高手术安全性的重要保障。有研究表明，妇科、结直肠肛门外科和泌尿外科联合手术可显著降低术后并发症发生率，提高病灶完整切除率^[27-29]。在本例中，因病灶

累及低位直肠，由结直肠肛门外科主导手术，妇科及泌尿外科团队协作。术中采用“先易后难”的策略：先游离直肠后方及侧方，在病变下方找到正常直肠与阴道间隙，再反向突围切除病灶，犹如“农村包围城市”。该策略不仅完整切除了病灶，还较好保护了正常直肠和阴道，降低了手术难度与并发症发生率，体现了MDT协作的重要价值。

结合本例患者的诊疗过程可见，EUS-FNA在术前确诊中起到了关键作用，为制定个体化治疗方案奠定了基础；在此基础上，MDT模式有效降低了手术难度及相关并发症风险；而术中采取“农村包围城市”式的分离策略，也有助于保护重要组织结构，提升手术整体安全性。上述经验为类似复杂病例的临床处理提供了有益参考。

作者贡献声明：陈文豪、曾海刚、郝立政负责病史采集、整理，论文撰写；王细文提供妇科专业指导意见；肖军提供消化内镜EUS专业意见；江从庆负责论文审阅和修改。

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