

• 综述 •

人工智能在消化道早癌诊疗中的应用进展

阙煜轩, 周晓颖*

南京医科大学第一附属医院消化内科, 江苏 南京 210029

[摘要] 消化道早癌(early gastrointestinal cancer, EGC)的早期症状不明显且筛查率低, 导致多数患者确诊时已为中晚期, 预后较差。因此, 早期诊断和及时治疗对改善患者预后至关重要。近年来, 人工智能(artificial intelligence, AI)技术能够通过高精度的图像分析和数据处理, 提高早期病变的检出率和诊断准确性, 从而实现早发现、早诊断、早治疗, 改善患者预后。但同时, AI也面临训练数据不足、算法偏差、各地区发展不均衡等问题, 需要进一步发展改进。文章全面回顾了AI在EGC诊疗中的应用现状, 探讨其优势、挑战及未来发展方向, 以期对相关领域的研究者和临床医生提供有价值的参考。

[关键词] 人工智能; 消化道早癌; 内镜检查; 诊断; 治疗

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Application progress of artificial intelligence in the diagnosis and treatment of early gastrointestinal cancers

QUE Yuxuan, ZHOU Xiaoying*

Department of Gastroenterology, the First Affiliated Hospital of Nanjing Medical University, Nanjing 210029, China

[Abstract] Early gastrointestinal cancer (EGC) has subtle early symptoms and low screening rates, which leads to most patients being diagnosed at the middle or advanced stages, resulting in poor prognoses. Therefore, early diagnosis and timely treatment are crucial for improving patient outcomes. In recent years, artificial intelligence (AI) technology has been able to enhance the detection rate and diagnostic accuracy of early lesions through high-precision image analysis and data processing, thereby achieving early detection, early diagnosis, and early treatment to improve patient prognoses. However, AI also faces challenges such as insufficient training data, algorithm bias, and uneven development across regions, which need to be addressed in future advancements. This review aims to provide a comprehensive overview of the current applications of AI in the diagnosis and treatment of EGC, exploring its advantages, challenges, and future directions to offer valuable references for researchers and clinicians in related fields.

[Key words] artificial intelligence; early gastrointestinal cancer; endoscopy; diagnosis; treatment

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消化道早癌(early gastrointestinal cancer, EGC)主要包括食管癌、胃癌和结直肠癌(colorectal cancer, CRC)的早期病变, 其患病率逐年上升, 且起病隐匿、早期症状通常不明显, 或仅表现出一些非特异性症状, 如腹胀、嗝气等, 待出现显著症状时多已发展为

中晚期, 错过最佳治疗时机^[1]。在我国, 部分消化道肿瘤如食管癌、胃癌的发病率很高, 约占全球的一半。EGC若未及时发现和治疗, 往往进展迅速, 导致患者医疗负担沉重, 生活质量下降且死亡率升高。早期筛查和诊断对于提高患者的生存率至关重要。

近年来, 人工智能(artificial intelligence, AI)技术广泛应用于病理学、影像学等医学领域, 已成为临床工作的重要辅助^[2-3]。AI能够从大量的医学影像和病理数据中学习早期癌症的特征, 识别出微小

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* 通信作者 (Corresponding author), E-mail: zhouxiaoying0926@njmu.edu.cn (ORCID: 0000-0002-6529-0243)

的病变区域,显著提高早期食管癌、胃癌和结直肠癌的诊断准确率并辅助临床决策^[4-6]。AI结合内镜检查等方法,提高EGC筛查敏感性和效率的同时处理和分析大量的临床数据,有助于发现新的生物标志物和疾病相关因素,协助医生更早发现潜在的癌变^[7-9]。AI为提高EGC的诊断水平和治疗效果提供了有力的技术支持。

1 AI技术概述

AI是指计算机系统能够模拟人类智能行为的技术和理论,是能够自主学习、适应环境变化并做出智能决策的系统,其核心技术主要包括:机器学习(machine learning, ML),深度学习(deep learning, DL),自然语言处理(natural language processing, NLP),计算机视觉(computer vision)等。

ML指计算机系统通过数据学习并做出预测或决策,而不需要显式编程,通常依赖于输入数据特征和算法进行训练,并完成分类、回归等任务。

DL是ML的一个子领域,基于多层神经网络进行学习和推理,特别适用于处理复杂的数据类型,如图像、声音和文本等,能够自动提取特征并做出高精度的预测^[10],主要模型有:卷积神经网络(convolutional neural network, CNN)、生成对抗网络(generative adversarial network, GAN)、深度信念网络(deep belief network, DBN)、Transformer模型等,其在图像识别和数据分析中的优势使其成为许多领域的关键技术。

近年来,随着计算机硬件性能的大幅提升、大数据技术的成熟、社会需求的日益增加以及国家政策的支持引导,AI技术得到极大发展。AI在医学领域的应用更呈现出多维度、深层次的发展趋势,在医疗管理、疾病诊断、精准治疗、远程健康管理、跨领域融合与创新等方面不断取得突破^[11-14]。

2 AI在EGC筛查中的应用

2.1 内镜检查中的应用

AI辅助内镜技术通过计算机视觉和DL,能够自动识别潜在病变,提高早期癌症的检出率。CNN专门用于处理具有网格结构的数据,通过模仿人类大脑的神经元连接方式,能够自动从图像中提取高级特征。CNN作为目前应用最广泛的DL算法之一,其计算机辅助检测(computer-aided detection, CADe)常用于胃肠内镜检查中,在结肠息肉检测中的灵敏度可达95.1%,准确率为96.8%,且能够检测

到小至5 mm的病变,灵敏度为93%^[15]。CADe能够自动检测内镜图像中的异常区域,如息肉、溃疡或肿瘤,提醒内镜医师行进一步检查或活检,显著提高了腺瘤和息肉的检出率^[16-19]。Wallace等^[20]研究表明,AI辅助的CRC筛查可以将病变的漏诊率降低50%。

Transformer是一种基于自注意力机制的DL模型,它摒弃了传统的循环和卷积结构,采用编码器-解码器架构,主要由多头自注意力机制(multi-head self-attention)和前馈神经网络(feed-forward network)组成,并通过残差连接和层归一化(residual connection & layer norm)来稳定训练,其核心优势包括并行计算能力强、长距离依赖建模能力出色,以及适用于文本、图像、语音等多模态数据的灵活扩展性。Transformer在处理复杂背景和小病变方面有着出色的能力,且在不同数据集上展现出强大的泛化能力,能够很好地适应不同医院的图像质量和设备配置变化。在诊断Barrett食管癌变方面,Transformer模型的性能与CNN相当,甚至在某些情况下略优于CNN^[21]。基于Transformer架构的MViTX模型,融合了定制化数据增强与外部注意力机制,在涵盖胃肠道疾病、乳腺癌、脑肿瘤等多种器官病变的医学图像数据集(含内镜图像)上展现出卓越性能,测试准确率处于94.1%至99.1%的高水平区间,相较传统CNN模型更具优势^[22]。

2.2 非内镜筛查方法

生物信息学与液体活检技术在EGC筛查中发挥着关键作用,通过对大量基因表达、蛋白质组学及临床数据的分析,ML算法能够挖掘出与EGC密切相关的潜在生物标志物,从而精准锁定高风险个体,进而优化配置有限的内镜资源。中山大学科研团队成功研制出VSOLassoBag算法,该算法在处理基因组、转录组、表观组学等高维组学数据时具有卓越表现,能够从有限样本中高效且稳定地挖掘出更具诊断和预后价值的核心标志物^[23]。

液体活检技术结合生物信息学分析,还能够检测血液中的循环肿瘤细胞(circulating tumor cell, CTC)、外泌体RNA和lncRNA-GCI等生物标志物,研究发现外泌体中的特定miRNA(如miR-1246)可以作为胃癌早期诊断的潜在标志物^[24-26]。在CRC筛查领域,引入新的检测靶点对提升筛查的特异性和检出率具有重要意义。Wisse等^[27]研究出一种新型的CRC筛查方法:相比传统粪便免疫化学检测(fecal immunochemical test, FIT)的阳性率(4.08%)和检出率(1.21%),多靶点FIT(mtFIT),显示出更高的阳性

率(9.11%)和进展期新生物检出率(2.27%)。Zhao等^[28]研究,甲基化糖蛋白2(methylated syndecan-2, mSDC2)作为一种新兴的检测靶点,能够有效增强CRC筛查的精准度,mSDC2检测呈阳性的患者,其腺瘤(adenomatosis of colon and rectum, ACN)、CRC及早期CRC的检出率均显著高于单独使用FIT的情况($P < 0.001$)。这种多层次的筛查策略减少了不必要的侵入性检查,提升了疾病早期发现的几率和后期诊断的精准率。

3 AI在EGC诊断中的应用

3.1 影像学诊断

实现EGC的早期诊断,对于提升患者的生存率和改善预后至关重要。传统影像学诊断方法(如CT、MRI)在病变检测方面虽有一定优势,但存在依赖医生经验、主观性强等局限。近年来,AI技术已广泛融入CT、MRI等医学影像处理领域,借助DL算法进行图像分割、特征提取与病变识别,有效提升了诊断及分期的精准度^[29-31]。

AI可以从CT、MRI影像中提取病变的形态特征(如大小、形状、边界)与纹理(如灰度共生矩阵中的对比度和相关性)特征,以区分良恶性病变^[32]。传统的CNN,例如UNet,在局部特征提取方面表现出色,但在全局特征提取上存在不足。相比之下,Transformer模型因其强大的全局信息捕获能力,能够更有效地处理图像中的长距离依赖关系,在图像分割任务中表现出了显著的优势^[33]。胃肠道间质瘤(gastrointestinal stromal tumor, GIST)具有恶性潜力,而当前的成像技术难以区分GIST和平滑肌瘤。宛新建教授团队成功开发并验证了一种基于超声内镜图像的实时AI系统^[34],该系统在区分GIST和平滑肌瘤时表现出色,其曲线下面积(area under the curve, AUC)值高达0.948,准确率为91.7%,敏感性为90.3%,特异性为93.0%,对于直径 ≤ 20 mm的小病变,诊断准确率更是高达93.5%。在前瞻性实时临床应用中,该AI系统在诊断性能上显著优于内镜医师。

3.2 病理诊断

AI技术在病理组织图像分析中的应用,如CNN等,为病理学家提供了有力的辅助,有效提高了诊断的准确性和效率^[35-37]。在病理图像分析中,CNN借助图像分割技术与多级注意力机制,有效提升对关键病理特征的提取效率,从而助力病理学家精准锁定组织内的病变部位^[38-39]。Alotaibi等^[40]提出了

一种创新的基于生物学图像的CRC诊断方法:最佳深度特征融合方法(CCD-ODFFBI)。该方法整合了MobileNet、SqueezeNet和SE-ResNet 3个DL模型来进行特征提取,利用Osprey优化算法确定DL模型的超参数,最后采用DBN模型进行CRC分类,结果表明与现有技术相比,CCD-ODFFBI方法具有99.39%的卓越准确率。此外,PathChat和CONCH等新型模型结合了视觉和文本数据,能够摄取组织病理学图像并结合临床背景信息,模拟病理学家的工作流程,提供更全面的诊断支持和鉴别诊断建议^[36,41]。

3.3 多模态数据融合

AI可以将不同来源和类型的医学数据(如内镜图像、病理切片、基因检测等)进行整合和分析,以实现更全面、准确的疾病诊断^[42]。其优势是:第一,提供多维度信息,从不同角度反映病变的特征。哈佛医学院等机构开发的CHIEF AI模型,通过融合病理图像和基因组学数据,能够诊断19种癌症,准确率接近94%,该模型还能识别与癌症生长和抑制相关的基因模式,预测患者的生存率^[43]。第二,弥补单一模态数据的局限性,提高诊断的可靠性。Nguyen等^[44]基于DNA特异性甲基化和片段组学特征开发了SPOT-MAS模型,可以为早期CRC检测提供高准确性:该模型在检测CRC时获得的AUC为0.989,敏感性为96.8%,特异性为97%,外部验证AUC为0.96。多模态数据融合AI技术在癌症早期诊断中展现出巨大潜力,能够显著提高诊断的准确性和可靠性,为癌症的精准医疗提供有力支持。

4 AI在EGC治疗中的应用

4.1 内镜下治疗

AI能够精确识别内镜下的病变位置,并确定切除范围,从而显著提升内镜治疗的精准度与安全性^[45-46]。有研究显示,基于AI技术内镜能精准识别并描绘肿瘤边界,为医生提供切除范围的建议,在病变特征不明显或位置较为隐蔽的情况下,可有效降低因医生经验差异而产生的误诊与漏诊风险,并且可最大程度保留周围健康组织,同时降低肿瘤患者在内镜切除术后接受额外手术的概率^[47-49]。在内镜治疗过程中,AI系统还能实时监控手术器械的位置和操作情况,及时发现偏差并发出预警,这种实时监测功能可以帮助医生及时调整手术策略,确保手术的安全性^[50-51]。此外,AI技术还可以实时评估治疗效果,优化治疗决策,从而提高治疗成功率,改善患者的长期预后,并推动个性化医疗的发展^[52]。

4.2 手术治疗

AI增强现实的三维解剖叠加、改进的肿瘤切除可视化以及AI格式的内窥镜和机器人手术引导等方法,为增强外科医生的感知做出了重大贡献^[53]。AI在手术中的应用仍处于起步阶段,目前的研发重点是AI辅助外科医生在手术过程中做出判断,例如解剖导航、手术机器人等,让AI替代医生做出术中决策是不切实际的^[54-55]。

术前,AI帮助外科医生精准规划手术方案,结合患者个性化数据和医生经验,预测手术风险并提供实时指导。英国帝国理工学院的研究团队利用组织的漫反射光谱信息训练机器学习,用于手术中识别上消化道肿瘤边界;结果显示,该方法分辨胃癌的准确度达到93.86%,分辨食管癌的准确度为96.22%,有效辅助手术医师对上消化道肿瘤手术边界的识别,提升手术精度^[56]。在机器人手术领域,AI建模使外科医生能够获得更精细的术中指标,如力量和触觉测量、阳性手术边缘的增强检测等,甚至允许某些手术步骤的完全自动化^[57]。AI分析患者的病史、生理指标、实验室检查及影像学数据等,整合运用ML与DL算法,构建数据驱动的预测模型,可精准预测术后并发症的发生^[58]。但也有研究^[59]发现,ML模型未能显著提高临床医生的预测能力,表明ML在围术期风险评估中的作用仍需进一步研究。

5 AI在EGC预后评估中的应用

5.1 预后预测模型

AI通过对患者临床、影像学及病理等各类数据的深入分析,构建出精准的预后预测模型,进而辅助医护人员更准确地评估患者预后情况。临床数据通常以电子病历形式存储,NLP技术可提取其中关键信息,如肿瘤转移部位和治疗反应等;常见的影像学检查(CT、MRI、超声等)能提供肿瘤大小、位置、形态及与周围组织关系等信息;结合病理图像中的细胞形态和组织结构等微观特征,AI对这些不同类型的数据进行特征提取与融合,构建更全面的特征集,再采用不同模型(如CNN、DBN、XGBoost、支持向量机等)进行DL,从而建立EGC的预后预测模型^[42,60]。Wang等^[61]和Zhang等^[62]分别针对食管癌和胃癌进行临床指标、影像组学的深度融合学习,构建出具有良好预后预测能力的模型。通过内部验证(如交叉验证)和外部验证(在独立数据集上验证)来评估模型性能,实现模型优化。例如,CHIEF

模型在32个独立数据集上进行验证,展示了其在癌症检测、生存预测等方面的强大能力^[43]。预后预测模型能够挖掘人类难以察觉的细微特征,提高预后评估的准确性,帮助医生评估患者的病情进展和治疗反应,从而选择更合适的治疗方案^[60]。

5.2 复发和转移预测

整合多组学数据,AI模型可以更全面评估肿瘤生物学特征,预测复发和转移风险。例如,运用DL和ML算法分析患者基因表达数据、蛋白质标志物等,能够精准预测CRC患者复发风险^[63]。Shaukat等^[64]研究开发的基于血液检测与多组学分析的ML算法,在平均风险人群中具有较高的灵敏度(79.2%)和特异性(91.5%),可用于CRC的早期筛查和复发监测。AI通过分析免疫组化染色的全切片图像,能够量化肿瘤细胞与免疫细胞之间的空间关联,从而评估患者的预后。例如,Morisita-Horn生态指数通过DL技术分割肿瘤细胞核、免疫细胞核和基质细胞核,以此来评估CRC患者的免疫状态和生存预后^[65]。

6 AI在EGC诊疗中的挑战

尽管AI在疾病诊疗中展现了巨大的潜力,但仍面临一些挑战。首先,收集高质量且经专家标注的数据是一项复杂且耗时的工作^[66]。AI模型如CNN的训练依赖于高质量的大规模数据集,然而,手动标注像素级的病变区域不仅费时费力,还导致了高质量数据的匮乏,这使得CNN的训练面临困难^[67]。如果数据存在偏差或不完整,可能导致模型的准确性和可靠性下降。例如,某些地区的患者数据可能无法充分代表全球范围内的患者群体,这无疑会对模型的广泛适用性造成不利影响^[68]。其次,EGC的临床数据来源多样,涵盖内镜图像、病理报告、基因测序数据等,这些数据的格式不统一、质量参差不齐,给数据整合和处理带来挑战^[69]。第三,不同地区医疗资源分配不均,各医院的内镜设备和成像技术存在差异,导致图像质量不一致,给AI模型的训练和应用带来影响^[70]。最后,AI在医疗领域的应用涉及患者的隐私保护、数据安全以及责任归属等伦理和法律问题。如果AI模型的诊断结果导致误诊,责任由谁承担是一个需要解决的问题。

针对以上挑战,可以制定“医疗AI数据标注指南”,要求标注团队通过一致性测试方可上岗,尽量使标注质量标准化;可将内镜图像、基因测序、病理报告等异构数据映射到统一本体,实现跨模态关联

分析,以解决多源数据的整合难题;尽量选择地区、规模差异小的医院进行多中心AI模型的训练,开发“域适应”模型,通过风格迁移技术将低质量内镜图像转换为AI可识别的标准域;AI应用中嵌入各级责任归属设置,以加强患者隐私保护,并建立AI诊断双签制度,即AI结果+高年资医生复核。

7 结语与展望未来

AI在EGC的诊断和治疗方面展现出了许多优势,并且有着极大的潜力,但要实现其临床广泛应用,还需解决数据、技术、法律法规等多方面的难题。未来,AI在EGC诊疗领域将朝着技术创新与优化、多模态数据融合、远程医疗、个性化医疗等方向发展,其临床应用也会更加广泛和深入。

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所有作者单位都隶属于南京医科大学第一附属医院(江苏省人民医院),无利益冲突。

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