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ADVANCES IN THE DIAGNOSIS OF HEAD AND NECK CANCERS: AN OVERVIEW OF CONTEMPORARY METHODS AND TECHNOLOGIES

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ABSTRACT

Diagnostics of head and neck cancers have rapidly advanced in recent years due to improvements in imaging, endoscopic techniques, molecular diagnostics and digital technologies. New tools including narrow-band imaging (NBI), advanced PET/CT and PET/MR modalities, radiomics, next generation sequencing (NGS), and liquid biopsy assays (ctDNA, HPV-cfDNA) have enhanced detection sensitivity, localisation accuracy and disease monitoring. Artificial intelligence and machine learning are integrating imaging and molecular data, enabling early biomarkers and refined therapy selection. This review synthesises major advances from 2020–2025 in head and neck cancer diagnostics and outlines future research priorities.

Literature review

Head and neck cancers (HNC) are a significant oncological problem due to the complexity of the anatomy, histological diversity and the impact of the disease and treatment on speech, swallowing and aesthetics. Effective diagnosis is the foundation of cancer treatment from early detection, through accurate staging and localisation, to monitoring response to therapy, prognosis and early detection of recurrence. The last five years have seen significant improvements in diagnostic technologies: functional imaging (PET/CT, PET/MR), endoscopic techniques (NBI, autofluorescence), molecular diagnostic tools (NGS, HPV tests, liquid biopsy) and digital tools (radiomics, AI).

Materials and methods

A literature review was conducted using PubMed, Scopus, PMC and Google Scholar to assess current advances in the diagnosis of head and neck cancer. Studies published between 2020 and 2025 were considered. The search included original articles, review articles, current guidelines for the diagnosis of head and neck cancer, and meta-analyses. Studies of low methodological quality, individual case reports, and reports published before 2020 were excluded unless they were of significant historical importance. Approximately 30 studies representing key achievements in the field were selected for the final reference list.

Results and discussion
1. Advanced endoscopy: Narrow-band imaging (NBI) and other techniques

Narrow-band imaging (NBI) has become a widely used tool in the diagnosis of oral, throat and laryngeal mucosal lesions. NBI improves the 2isualization of vascular networks and enables better detection of precancerous lesions and early squamous cell carcinomas compared to conventional white light endoscopy [1, 2, 3]. Systematic reviews and meta-analyses published in recent years confirm the high sensitivity and specificity of NBI in detecting cancerous lesions, and this technique is particularly useful in the follow-up of patients after treatment, where it can be difficult to distinguish between radiotherapy scars and recurrence [4, 5].

2. Functional and hybrid imaging: PET/CT, PET/MR

18F-FDG PET/CT remains a key method in assessing the stage of HNC and detecting lymph node metastases and distant metastases. New analyses indicate higher accuracy of PET/CT in detecting nodal metastases compared to CT and MRI [6]. Hybrid PET/MR combines the advantages of anatomical MRI imaging with the metabolic values of PET, offering better assessment of soft tissue structures in the head and neck area and potentially lower radiation doses. Reviews from 2022–2025 show the growing use of PET/MR in staging and assessing residual disease, although availability and costs limit its widespread use [7, 8].

3. Radiomics and artificial intelligence (AI)

Radiomics, i.e. the extraction of multidimensional features from medical images, and machine learning methods are changing the approach to image interpretation. Radiomic models show promising results in predicting treatment response, recurrence risk, and overall prognosis, especially when combined with clinical and molecular data [9, 10, 11]. The work of Tortora et al. 2023 provides an overview of radiomics applications in HNC and points to the need for standardisation of methodology and external validation of models [9]. AI is also being investigated in the automation of endoscopic image assessment and prediction of lymphatic spread [12].

4. Molecular diagnostics and liquid biopsy (ctDNA, HPV-cfDNA)

Liquid biopsy using circulating tumour DNA (ctDNA) detection and HPV-cfDNA detection in serum/plasma and saliva is a rapidly developing tool in the diagnosis and monitoring of HNC, especially HPV-dependent throat cancers. Cohort and multicentre studies have demonstrated the high specificity of HPV-ctDNA detection in monitoring residual disease after treatment and in the early detection of recurrence [13, 14, 15]. Recent studies from 2024–2025 suggest the possibility of detecting HPV-ctDNA several years before the clinical manifestation of the disease, which could revolutionise early screening in high-risk individuals [16].

5. Next-generation sequencing (NGS) and molecular profiling

NGS enables comprehensive analysis of mutations, amplification changes and gene markers relevant to clinical trials and targeted therapies. NGS panels for HNC help identify mutations in the TP53, NOTCH1 and PIK3CA genes and HPV markers, which influences eligibility for experimental trials and therapies [17, 18]. The integration of NGS with radiological data (radiogenomics) and liquid biopsy data paves the way for both personalised monitoring and therapy.

6. Sentinel lymph node biopsy (SLNB) and lymph node assessment

Sentinel node biopsy has become an increasingly accepted method for assessing occult metastases in patients with clinical stage N0 oral cancer, providing an alternative to elective neck dissection in selected cases. Systematic reviews in recent years confirm the high diagnostic accuracy of SLNB and its potential to reduce the extent of surgical procedures [19, 20].

7. Endoscopic techniques and transoral surgery: TORS and intraoperative imaging

Transoral robotic surgery (TORS) has changed the approach to the treatment of selected oral and throat tumours, enabling precise resections while preserving organ function. Although TORS is primarily a therapeutic technique, its growing role influences diagnostic decisions and further treatment planning; the integration of preoperative imaging (MRI) with TORS planning improves patient prognosis [21, 22, 23].

8. Challenges and limitations

Despite advances, barriers to the implementation of modern methods remain: the cost of technology (PET/MR, NGS), limited availability in less developed countries, the need to standardise radiomics and AI methods, and the need for multicentre validation of liquid biopsy biomarkers. In addition, the heterogeneity of HNC and aetiological differences (HPV+ vs HPV−) require a differentiated diagnostic and interpretative approach [24, 25, 26].

In recent years, the diagnosis of head and neck cancer has been developing dynamically, integrating molecular, imaging and digital methods. Research on circulating markers confirms the growing role of liquid biopsy in HPV-dependent cancers – Kumari et al. (2025) discuss the use of ct-HPV DNA in the diagnosis, prognosis and monitoring of recurrence in patients with throat cancer, which opens up prospects for much less invasive clinical surveillance. In parallel, next-generation sequencing (NGS) enables increasingly accurate molecular profiling of tumours, as illustrated, among others, by Demirci et al. (2025) in the context of therapeutic applications. On the other hand, advanced endoscopic techniques with assisted imaging, such as narrow-band imaging (NBI), significantly improve the detectability of precancerous lesions within the mucous membranes. Finally, the emergence of artificial intelligence in the analysis of diagnostic data raises new legal and ethical challenges: Gerke, Minssen and Cohen (2020) point to the need for transparency of algorithms, protection of patient privacy and accountability for AI-supported decisions. [27, 28, 29, 30]

Discussion

The diagnosis of head and neck cancer has become a field of integration of imaging, molecular and digital technologies. Advanced endoscopy (NBI), PET/CT and PET/MR, NGS and liquid biopsy are complementary tools which, when combined with AI algorithms, increase the accuracy of detection and monitoring of head and neck cancer. The main challenges are standardisation, external validation of radiomic models and AI algorithms, and ensuring the availability of modern tests in everyday clinical practice.

The proposed research directions include: multicentre prospective validation of radiomic biomarkers, integration of NGS and radiogenomics in clinical trials, standardisation of liquid biopsy protocols and establishment of clinical decision thresholds for ctDNA/HPV-cfDNA, and cost-effectiveness assessment of PET/MR and AI tool implementations.

Conclusions

1. The combination of advanced endoscopy, molecular diagnostics, hybrid imaging, and AI improves diagnostic accuracy and monitoring of HNC.

2. Liquid biopsy (ctDNA, HPV-cfDNA) has great potential in monitoring minimal residual disease, but requires further validation.

3. Radiomics and AI can enhance the predictive capabilities of imaging, provided they are standardised and validated.

4. Multicentre studies, standardisation, and assessment of the costs of implementing new technologies are crucial.

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