Accessibility and Practicality of Actionable Cancer Genetic Testing Guidelines for Primary Care: A Systematic Review

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Introduction

The rapid expansion of germline and somatic genetic testing has transformed cancer prevention and management. Professional societies including the National Comprehensive Cancer Network (NCCN), American Society of Clinical Oncology (ASCO), European Society for Medical Oncology (ESMO), and American College of Medical Genetics and Genomics (ACMG) have published recommendations for cancer genetic testing. As genetic testing expands into primary care settings, non-genetic specialists increasingly rely on published guidelines to guide testing decisions and downstream management. However, the accessibility, clarity, and clinical usability of these guidelines for primary care providers remain unclear.

Aims and Objectives

To systematically evaluate cancer-related genetic testing guidelines with respect to:

1.) Accessibility
2.) Level of detail
3.) Understandability for non-specialists
4.) Practicality for clinical implementation
5.) Alignment with NCCN recommendations

Limitations

Limitations of this study include the use of a scoring rubric that incorporates some subjective interpretation when assessing guideline clarity and practicality. Although screening and data extraction were performed independently by two reviewers, differences in interpretation may influence scoring outcomes. Additionally, some included conference abstracts contained limited methodological detail, which may have affected scoring accuracy.

Methods

A systematic review was conducted using PubMed, Embase, and Scopus to identify cancer genetic testing guidelines published within the past 10 years. Screening of the initial 667 articles was performed independently by two reviewers using Covidence.

Included documents were evaluated using a standardized scoring rubric (1–5) assessing:

- Accessibility
- Level of detail
- Understandability
- Practicality

Discrepancies between reviewers were resolved through consensus.

Final inclusion: 65 guidelines.

Results

Category	Key Finding	Mean Score*
Accessibility	Open-access guidelines most accessible	3.1
Detail	Professional societies most detailed	4.8
Understandability	Group-authored studies easier to read	3.4
Practicality	Clinical consensus guidelines most practical	3.2

*Mean score was out of a maximum of five

Scoring Categories

Score	Accessibility	Level of Detail	Understandability	Practicality
5	Freely accessible online with no login or subscription required	Very detailed; covers technical and practical aspects thoroughly	Clear and easily understood by non-specialists	Highly practical with clear clinical steps
4	Accessible with free registration or institutional login	Detailed with minor omissions	Generally clear with minor complexity	Generally practical with minor clarification needed
3	Abstract available but full text restricted	Moderately detailed; basic aspects covered	Understandable with some effort; occasional jargon	Moderately practical; requires adaptation
2	Subscription or payment required with no open summary	Limited detail; key information missing	Difficult without specialized knowledge	Limited clinical applicability
1	Not publicly accessible or available only internally	Minimal information provided	Highly technical and confusing	Not practical for real-world use

Conclusions

1. Substantial variability exists in the accessibility and usability of cancer genetic testing guidelines.
2. Professional society guidelines provide high-level detail but may present barriers for primary care clinicians due to technical complexity and access restrictions.
3. Improving clarity, public accessibility, and workflow integration may enhance implementation of genomic medicine in non-specialty settings.

References

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