MSR14

Unleashing the Potential of Artificial Intelligence in Genomic Biomarker Testing for Precision Oncology: A Scoping Review

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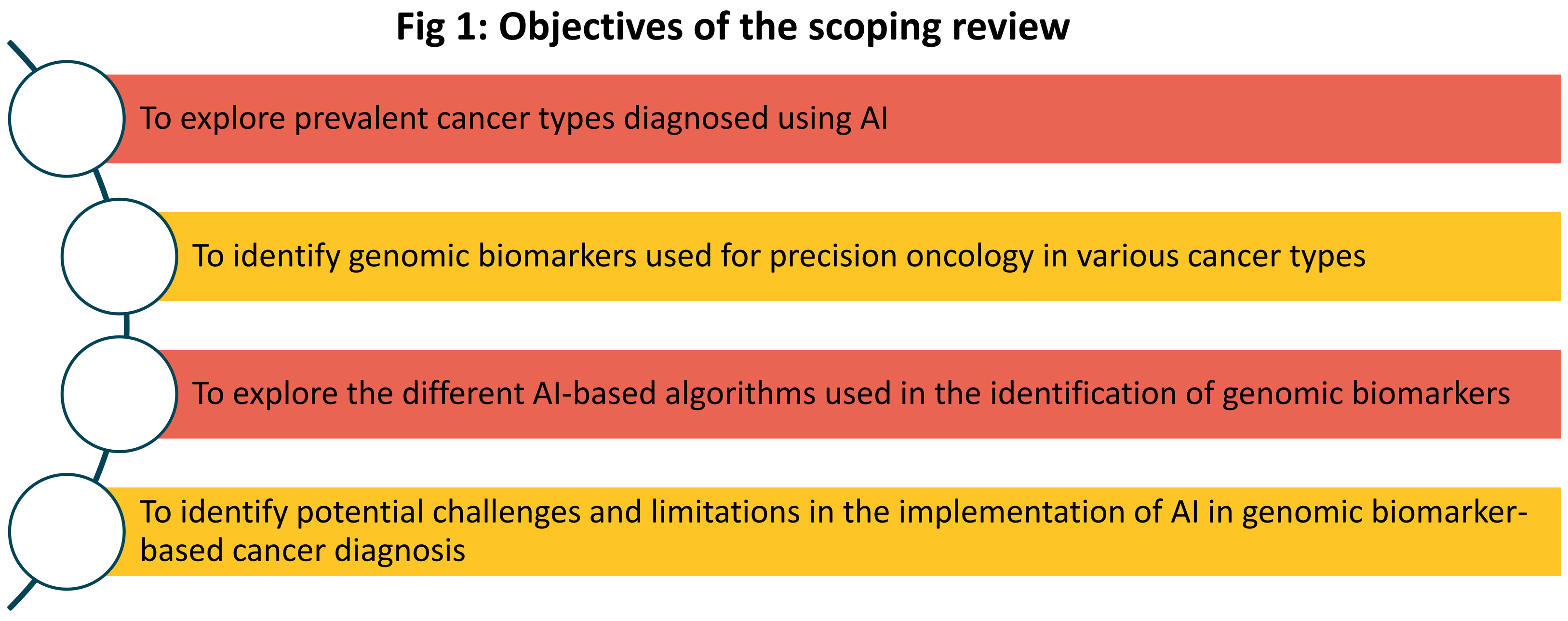
INTRODUCTION

- Genomic biomarker testing represents a cutting-edge approach with superior precision in oncology compared to traditional biomarker testing [1].
- Precision oncology greatly benefits from the advanced molecular insights provided by genomic biomarker testing [1].
- Currently, the manual genomic biomarker testing process faces challenges such as the need for specialized skills, managing extensive volumes of genomic data, and various logistical obstacles [2].
- The integration of Artificial Intelligence and Machine Learning into the healthcare ecosystem holds the promise of addressing these manual genomic biomarker testing challenges. This integration can enhance the precision of genomic biomarker testing for oncology, making it more effective [3].
- This scoping review explores the pivotal role of Artificial Intelligence aided genomic biomarker diagnosis, aiming for a comprehensive understanding that can drive its effective deployment in precision oncology.

OBJECTIVES

Fig 1: Objectives of the scoping review



METHODOLOGY

Search Strategy: A systematic search was conducted on databases PubMed, and Google using an artificial intelligence (AI)-powered evidence synthesis tool MaiA using predefined search terms related to AI, genomic biomarker testing, and precision oncology. The search strategy was designed to capture relevant studies published from January 2013 to June 2023. Boolean operators (AND, OR) and truncation were used to optimize search results. The final studies included consisted of the ones on cancer patients with genomic biomarkers using AI algorithms in the past 10 years. The studies related to other diagnostic techniques and those published before 2013 were excluded. (Figure 2).

Fig 2: The PCC Framework

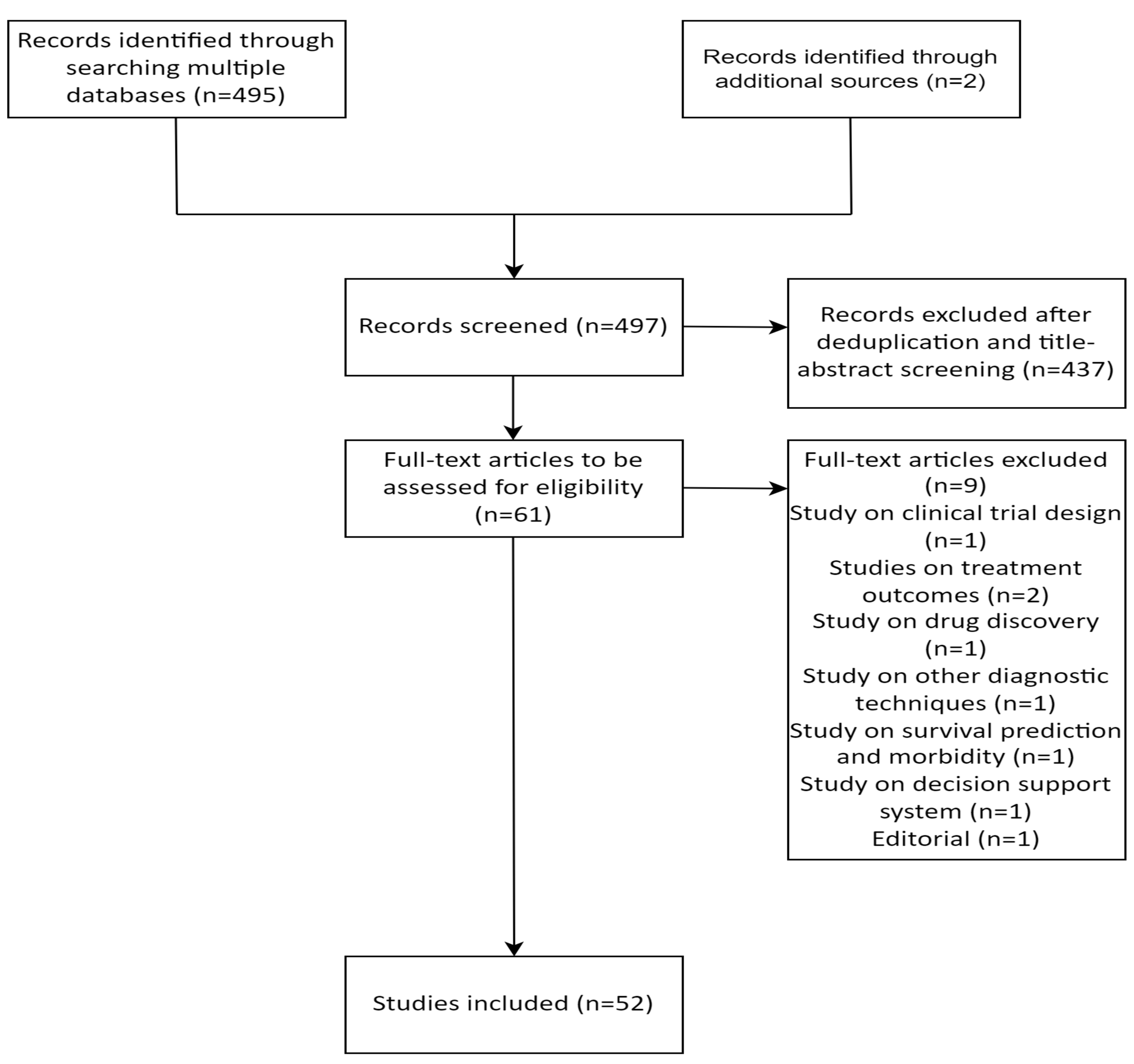
POPULATION	CONCEPT	CONTEXT
Inclusion Criteria: Patients with different cancer types diagnosed with genomic biomarkers **Exclusion Criteria:** Patients diagnosed using other diagnostic techniques	**Inclusion Criteria:**Different techniques used in the identification of genomic biomarkers and associated challenges for personalized biomarker diagnosis **Exclusion Criteria:** Diagnostic techniques in oncology other than genomic biomarker testing	**Inclusion Criteria:**Literature published on the use of AI for the identification of genomic biomarkers for personalized cancer diagnosis in the past 10 years **Exclusion Criteria:** Literature in languages other than English and those published before 2013

Data Extraction and Synthesis: Two independent reviewers screened the retrieved studies based on titles and abstracts for relevance to the research question. The selected articles after the title-abstract screening were screened for full-text relevancy. 52 full-text articles were included in the review after the final screening.

RESULTS

The selected articles after the title-abstract screening were screened for full-text relevancy. 52 full-text articles were included in the review after level 1 and level 2 screening. Figure 3 depicts the PRISMA flowchart of the process.

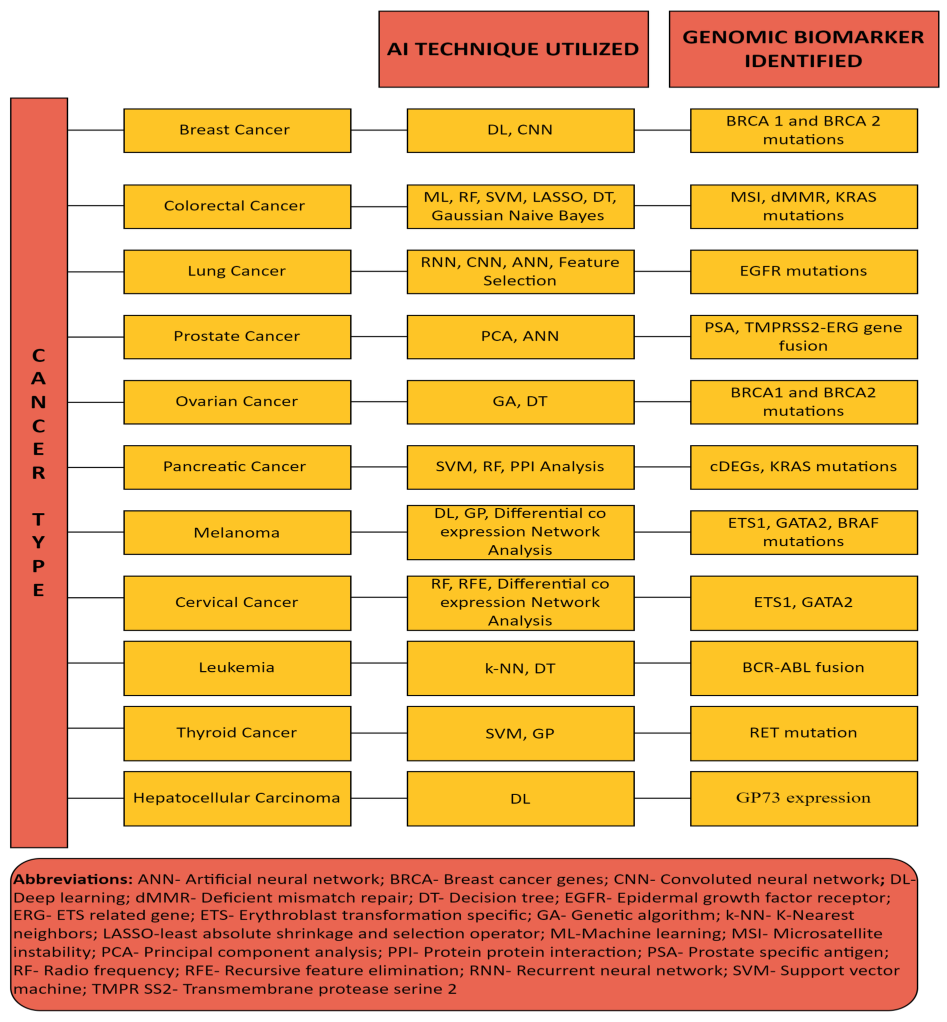
Fig 3: The PRISMA Flowchart



RESULTS (Contd..)

- The combination of AI by quickly analyzing extensive genomic data, identifying complex patterns, and predicting treatment responses based on individual genetic profiles and genomic biomarker testing shows great potential for enhancing the precision, speed, and overall efficacy of cancer treatment selection.
- Numerous prevalent cancer types are utilizing AI methods for the identification of genomic biomarkers (Figure 4).

Fig 4: Cancer types and the associated AI techniques utilized.

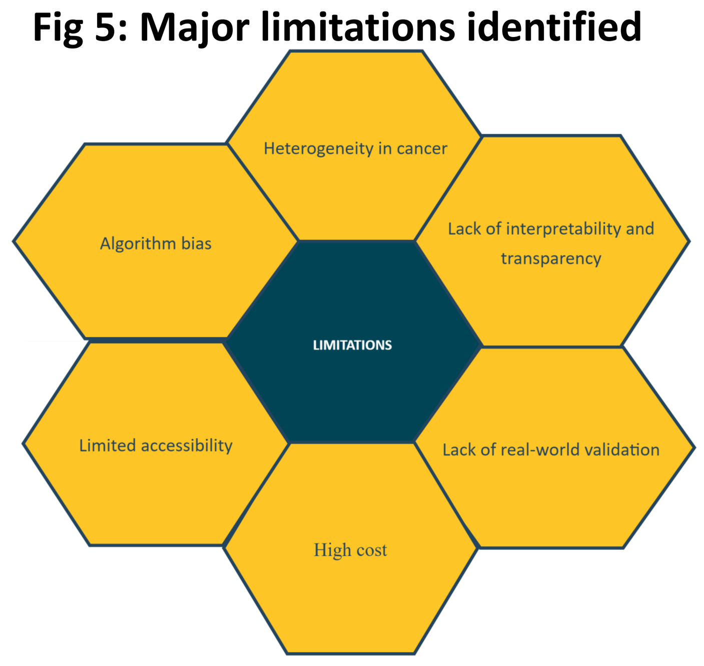


DISCUSSION

- The application of AI in genomic biomarker testing is set to move personalized medicine forward, customizing cancer diagnoses and therapies based on an individual's distinct genetic makeup.
- AI offers the potential to excel in identifying new genomic biomarkers with unparalleled precision, potentially facilitating earlier cancer detection and the development of precise treatment strategies.
- AI-driven genomic testing may enable instantaneous cancer diagnosis and ongoing monitoring, delivering timely information for adjusting treatment strategies as needed.

The major limitations that were identified in the implementation of AI in genomic biomarker-based cancer diagnosis are:

Fig 5: Major limitations identified



FUTURE DIRECTIONS & NEXT STEPS

- Identification of genes as biomarkers that contribute to drug resistance to cancer treatment, which can hinder successful cancer treatment.
- Implementation of global guidelines on the usage of big data to prevent ethical concerns.
- Development of more robust cost-effective identification models that can include different types of cancers including less common ones such as albeit mutations in ocular melanoma followed by cross-validation.
- Identification of the possibility of applying predictive methods to real-world oncology data.

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