Biomarker/genomic testing–guided systemic therapy in U.S. cancer care: a scoping review of real-world evidence

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Background

- Predictive biomarkers (e.g., genetic mutations, proteins) have transformed cancer care by enabling personalized treatment and improving patient outcomes.
- These biomarkers help estimate how likely a patient is to respond to specific therapies, guiding treatment selection.
- Many cancers now have biomarker or genomic testing available to support precision medicine and targeted therapies.
- Despite their benefits, testing is not always performed or properly used in treatment decisions.
- A scoping review was conducted to characterize biomarker testing practices, treatment utilization, and real-world patterns of care

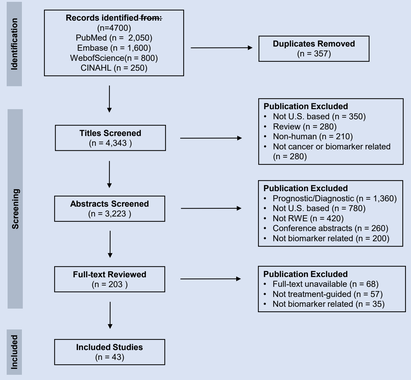
Objectives

Objective: To systematically map existing research on how and how often cancer biomarker/genomic/genetic testing is conducted and incorporated into treatment decisions.

Methods

- We conducted a scoping review, following PRISMA guidelines, to identify studies evaluating the use of biomarker, genomic, or genetic testing to guide chemotherapy treatment in real-world settings. Peer-reviewed, English-language studies published between 2010 and 2025 were included if they included adult populations in the U.S. and assessed the use of such testing in chemotherapy decision-making, including the impact on clinical outcomes, resource utilization, and cost of care.
- Studies focused solely on prognosis, diagnosis, or response to chemotherapy were excluded.
- Searches were conducted in CINAHL, Embase, PubMed, and Web of Science using the terms "chemotherapy AND (biomarker OR genetic testing) AND (real world OR real-world OR realworld)," with additional reference list checks. Two reviewers, DH and SC, independently screened and extracted data, focusing on study characteristics, testing utilization, and outcomes.
- Disagreement was resolved through consensus.

Figure 1: PRISMA Flow Diagram



Results

Table 1: Distribution of Included Studies by Cancer Type

Cancer Type	Count	Percentage
Non-Small Cell Lung Cancer (NSCLC)	20	47%
Breast Cancer	11	26%
Colorectal Cancer	3	7%
Acute Myeloid Leukemia	2	5%
Other Cancers	7	16%

Table 2: Distribution of Included Studies by Data Source

Data Source	Count	Percentage
Flatiron Health	25	58%
IKnowMed	7	16%
Other EHR Databases	11	26%

Table 3: Distribution of Targeted Treatment Strategies in NSCLC Studies

Drug Class	Count	Percentage
Anaplastic Lymphoma Kinase Tyrosine Kinase Inhibitor (ALK TKI)	1	5%
Epidermal Growth Factor Receptor Tyrosine Kinase Inhibitor (EGFR TKI)	4	20%
Immune Checkpoint Inhibitors (ICI)	4	20%
Various Combinations of EGFR inhibitors, ALK inhibitors, B-Raf proto-oncogene, serine/threonine kinase (BRAF) inhibitors, and Mitogen-Activated Protein Kinase Kinase (MET) inhibitors	4	20%
Other	7	35%

Table 4: Distribution of Targeted Treatment Strategies in Breast Cancer Studies

Drug Class	Count	Percentage
Human Epidermal Growth Factor Receptor 2 (HER2) Antagonists	5	45%
Endocrine Therapy + Cyclin-Dependent Kinase 4 and 6 (CDK4/6) inhibitor	4	36%
Other (Chemotherapy)	2	18%

Table 5: Common Biomarkers and Treatment Patterns

Biomarker	Studies that Tested Biomarker (n = 43)	Cancer Type	Targeted Therapy (Drug Class)[1]	Common Drugs in the Drug Class[1]
EGFR	28%	NSCLC	EGFR TKIs	Osimertinib, Erlotinib
ALK	21%	NSCLC	ALK Inhibitors	Alectinib, Crizotinib
PD-L1	30%	NSCLC	ICIs	Pembrolizumab, Nivolumab
HER2	33%	Breast Cancer	HER2-Targeted Therapy	Trastuzumab

Limitations

- **Heterogeneity across studies:** Included studies varied in cancer types, biomarkers, data sources, and outcome measures, limiting direct comparability.
- **Inconsistent reporting of key outcomes:** Not all studies reported biomarker testing rates, targeted therapy use, or survival outcomes, which may affect aggregated estimates.
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- **Rapidly evolving treatment landscape:** Changes in biomarker testing practices and targeted therapies over time may limit the applicability of older studies to current practice.
- **Sample size:** Small sample size in some studies may limit the reliability of findings

Conclusion

- 27 out of 43 studies (63%) reported overall survival, 18 out of 43 studies (42%) reported progression-free survival, and 41 out of 43 studies (95%) reported at least one outcome measure
- Although several studies did not report specific biomarker testing rates, they consistently described a trend of increasing testing utilization over the study period
- Among the 15 studies reporting biomarker testing rates (35%), the average testing rate was 56%. This indicates that biomarker testing is not universally performed, even in populations where it is clinically relevant.
- Across all 43 studies, an average of 52% of patients received targeted therapy, while 46% did not, highlighting a notable gap between biomarker testing and treatment in real-world practice.
- Biomarker testing is frequently used to guide treatment selection; however, a gap remains between testing and the use of targeted therapies in real-world practice.
- Real-world evidence demonstrates variability in the use of biomarker-informed treatment strategies, with inconsistencies in how testing results are translated into clinical decision-making.

References

1. National Comprehensive Cancer Network. *NCCN Guidelines for Patients®* https://www.nccn.org/patientresources/patientresources/guidelines-for-patients. Accessed March 30, 2026.

Acknowledgement

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