

BIOMARKERS AS A DIAGNOSTIC TOOL IN CANCER: A BOON OR A BANE?

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ABSTRACT:

- Cancer Screening is a hotly debated and extensively researched topic.
- There are several screening modalities currently in application as well as being researched with the sole aim to detect cancer early and eventually eradicate it.
- The development of Next Generation Sequencing in genomic analysis and identifying biomarkers has garnered an exponentially growing interest amongst cancer researchers to precisely diagnose the presence of cancer at an early stage, thereby reducing the morbidity and mortality and potentially assisting with the prognostic and predictive profiling.
- The diagnostic and clinical efficacy of these biomarkers are potentially instrumental in helping improve the clinical outcome of cancer and should be researched with this goal in mind.

INTRODUCTION

It is a defined characteristic or a biological observation, intended to predict and/ or confirm the presence of a pathogenic process, or response to a therapeutic intervention [1].

CATEGORIES

Conventional

Gastric [2]	Liver	Ovarian	Medullary Thyroid	B cell lymphoma	Neuroendocrine	Gastrinoma	Carcinoid	Prostate
CEA, CA19-9, AFP, CA-125, CA72-4	AFP	CA125	Calcitonin	CD19 and CD22	Chromogranin A	Gastrin	5-HIAA	PSA

Biomarkers utilizing Next Generation Sequencing

Genetic [3]	Transcriptomic	Epigenetic	Proteomic	Metabolomic	Multiomics
ctDNA, mutation to the driver genes	mRNA, miRNA	Methylated DNA	prognostic	Altered metabolism	Combination of all

DIAGNOSTIC IMPLICATIONS

Pros:

- Early Detection of Cancer before it metastasizes [4]
- Better survival rates with results of diagnostic profiling available as prognostic and predictive markers
- Wider treatment options available when diagnosed earlier
- Personalized patient care with targeted therapy delivery

Cons:

- Limited sensitivity
- Limited specificity
- Overdiagnosis with added emotional and financial burden to patient and healthcare system

Overcoming the Cons:

- Precision Medicine with laser sharp focus
- Improved accuracy with detailed genomic analysis using Next Generation Sequencing
- Designing the trial keeping the demographic with the intent to treat in mind and the prevalence of the cancer in that demographic
- Improving Positive predictive value and Negative predictive value
- Concordant result across the sites in the clinical trial

CONCLUSION

- The novel genomic approaches to identify biomarkers for an earlier detection of cancer is a boon in making.
- However, it comes with its very own challenges in diagnosing cancer with unknown primary source of origin.
- The advanced genomic analysis can be supplemented by cancer risk profiling and guided by diagnostic profile results to deliver improved performance of the test in clinical practice in early detection of cancer.

REFERENCES

1. [BEST \(Biomarkers, EndpointS, and other Tools\) Resource \(nih.gov\)](#)
2. Matsuoka T, Yashiro M. Biomarkers of gastric cancer: Current topics and future perspective. World J Gastroenterol. 2018;24(26):2818-2832. doi:10.3748/wjg.v24.i26.2818
3. [Tumor Markers in Common Use - National Cancer Institute](#)
4. Schiffman JD, Fisher PG, Gibbs P. Early detection of cancer: past, present, and future. Am Soc Clin Oncol Educ Book. 2015;57-65. doi: 10.14694/EdBook_AM.2015.35.57. PMID: 25993143.