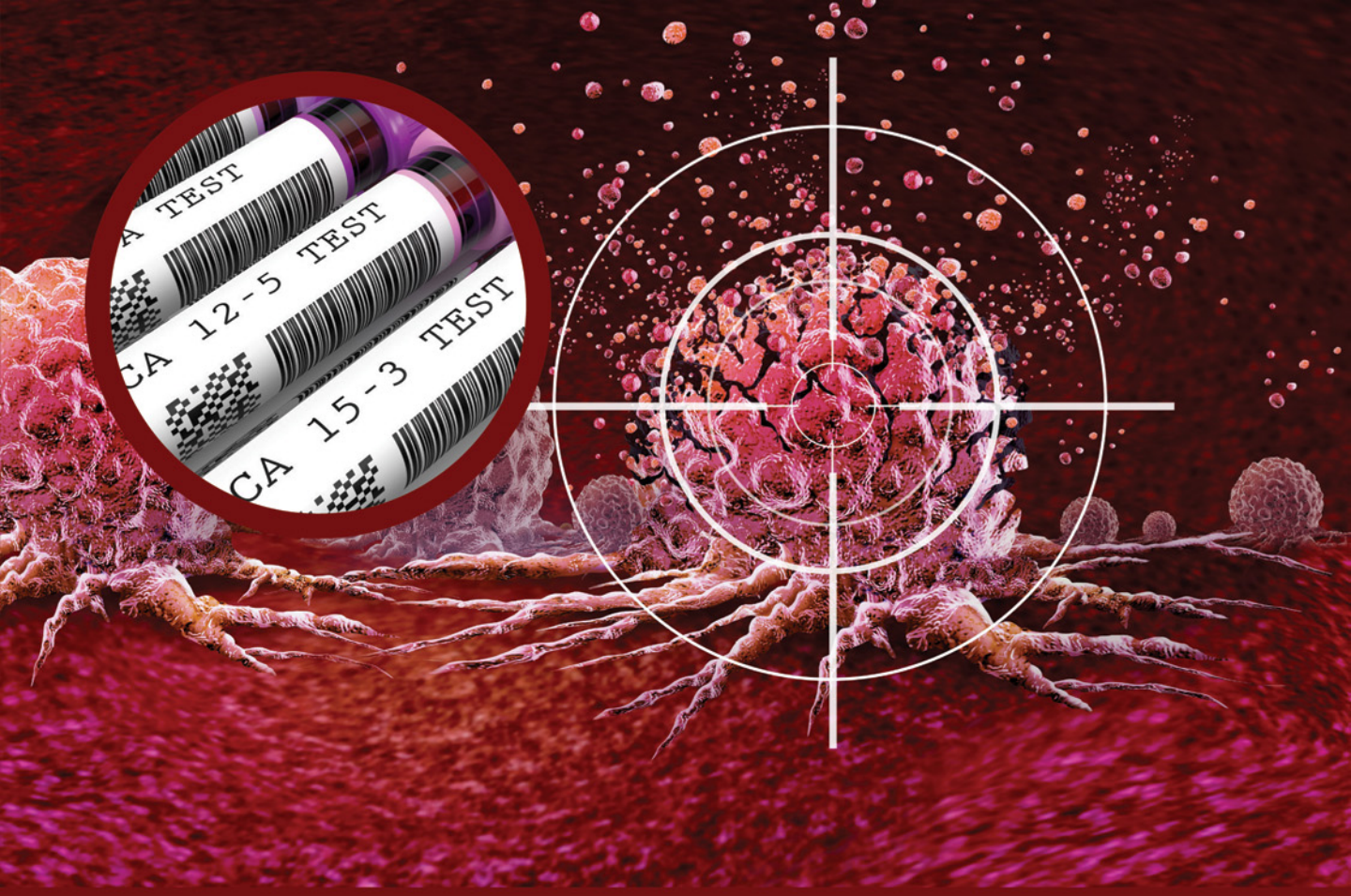

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CHAPTER

10

Genetic biomarkers in cancer

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Abbreviations

ALK	Anaplastic Lymphoma Kinase
APC	Adenomatous Polyposis Coli
BRCA	Breast cancer
CML	Chronic Myeloid Leukemia
CNV	Copy Number Variant
CRC	Colorectal Cancer
CSF	Cerebrospinal fluid
CTC	Circulating Tumor Cell
ctDNA	Circulating Tumor DNA
DNA	Deoxy Ribonucleic Acid
DNMT	DNA Methyltransferase
EGFR	Epidermal Growth Factors Receptors
HER	Human Epidermal Growth Factors Receptors
HRAS	Harvey Rat Sarcoma
KRAS	Kirsten Rat Sarcoma
lnc RNA	Long non-coding RNAs
mi RNA	Micro Ribo Nucleic Acid
MLH	MutL Homolog

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MMR	Mis Match Repair
MRD	Minimal Residual Disease
mRNA	Messenger Ribo nucleic acid
MSH	MutS Homolog
MSI	Micro Satellite Instability
MYC	Myelocytomatosis
ncRNA	Non-Long coding Ribo nucleic Acid
NRAS	Neuroblastoma Rat Sarcoma
NSCLC	Non-Small Cell Lung Cancer
NGS	Next Generation Sequencing
PCR	Polymerase Chain Reaction
qPCR	Quantitative Polymerase Chain Reaction
RNA	Rat Sarcoma (RAS), Ribonucleic Acid
RT-PCR	Real Time-Polymerase Chain Reaction
SNP	Single Nucleotide Polymorphism
TMB	Tumor Mutational Burden
TP	Truncated Peptides
UTRs	Untranslated Region

10.1 Introduction

A biomarker is any chemical substance that can be quantified in body fluids, blood, or tissues and indicates the existence of typical or aberrant biological states, disorders, or diseases. [1]. In oncology, a biomarker of cancer is distinctly separate from cancer characteristics, ideally exhibiting high accuracy and dependability, quantitatively assessed by sensitivity and specificity [2]. Biomarkers for cancer also find applications beyond just identifying the kind of cancer patients are suffering from. Certainly, after identification, tumor markers are able to give invaluable information regarding the probable progression of the illness, including repetition likelihood as well as likely treatment outcome. Biomarkers in cancer are important in setting a disease's prognosis, despite any kind of therapeutic intervention (also called prognostic biomarkers) or in the ability to foresee how a cancer would respond to a particular treatment, thereby leading to the ability to forecast treatment outcomes (also called predictive biomarkers) [3].

A genetic marker is a DNA polymorphism employed as a standard for identifying single-gene disorders and complex trait genes in molecular and quantitative genetics [4]. A number of biomarkers for cancer have been identified and are mainly grouped based on the incidence of proteins in multiple practical classifications, including hormones, receptors, enzymes, and antigens [1,5]. Genetic modification in cancer, such as translocation, mutation, and enhancement at the specific gene stages, or generating genetic outlines using microarrays, generates typical genetic markers. These alterations allow the detection and characterization of markers of cancer, hence advancing the comprehension and control of cancer [6].

The National Cancer Institute has established biomarkers as biological molecules in body fluids, tissues, or blood that mark disease or a biological process or state—such as cancer—whether within normal parameters or not. These molecules are important for distinguishing individuals with the disease from those without it, and changes in their concentrations are