

Types of Biomarkers

Essential Classification for Precision Medicine

Diagnostic

Identify presence or absence of disease

Prognostic

Predict disease outcome independent of treatment

Predictive

Forecast response to specific therapy

Pharmacodynamic

Measure biological response to treatment

Safety

Detect potential adverse effects or toxicity

Each biomarker type serves distinct clinical purposes in precision medicine workflows

Detailed Biomarker Classification

Diagnostic biomarkers are used to detect or confirm the presence of a disease or condition, or to identify individuals with a subtype of the disease. They help clinicians make accurate diagnoses and distinguish between different disease states.

Clinical Examples:

- ▶ **Troponin:** Elevated levels indicate myocardial infarction (heart attack)
- ▶ **HbA1c:** Diagnoses diabetes mellitus ($\geq 6.5\%$ indicates diabetes)
- ▶ **PSA (Prostate-Specific Antigen):** Screening tool for prostate cancer
- ▶ **HIV Antibodies:** Confirms HIV infection
- ▶ **BRCA1/BRCA2 mutations:** Identifies hereditary breast and ovarian cancer risk

Key Characteristics:

- ✓ High sensitivity and specificity
- ✓ Distinguishes diseased from healthy individuals
- ✓ May identify disease subtypes

Patient Presents
with Symptoms



Diagnostic Biomarker
Testing



Positive
Result

Negative
Result



Disease
Confirmed/Ruled Out

Diagnostic Workflow

Prognostic biomarkers provide information about the likely course of disease in an individual patient, independent of treatment. They help predict clinical outcomes such as disease recurrence, progression, or survival, informing treatment intensity decisions.

Clinical Examples:

- ▶ **Oncotype DX score:** Predicts breast cancer recurrence risk (low/intermediate/high)
- ▶ **Tumor Grade & Stage:** Higher grade/stage correlates with worse prognosis
- ▶ **TP53 mutations:** Associated with poor prognosis in various cancers
- ▶ **NT-proBNP levels:** Predicts heart failure outcomes and mortality
- ▶ **Ki-67 index:** High proliferation marker indicates aggressive tumor behavior

Key Characteristics:

Disease Diagnosed



Prognostic Biomarker Assessment



Good Prognosis

Poor Prognosis



Risk Stratification
Treatment Planning

Prognostic Assessment Workflow

- ✓ Independent of treatment effects
- ✓ Stratifies patients by risk level
- ✓ Guides treatment intensity decisions

Predictive biomarkers indicate the likelihood of a patient responding to a specific therapeutic intervention. They guide personalized treatment selection by identifying which patients will benefit from particular therapies and who should avoid treatments unlikely to work or likely to cause harm.

Clinical Examples:

- ▶ **HER2 amplification:** Predicts response to trastuzumab (Herceptin) in breast cancer
- ▶ **EGFR mutations:** Identifies lung cancer patients for EGFR inhibitor therapy
- ▶ **KRAS mutations:** Predicts non-response to anti-EGFR therapy in colorectal cancer
- ▶ **PD-L1 expression:** Predicts response to immune checkpoint inhibitors
- ▶ **BCR-ABL fusion:** Indicates responsiveness to imatinib in CML

Key Characteristics:

Treatment Selection
Required

Predictive Biomarker
Testing

Marker
Positive

Marker
Negative

Targeted Therapy
Selection

Predictive Testing Workflow

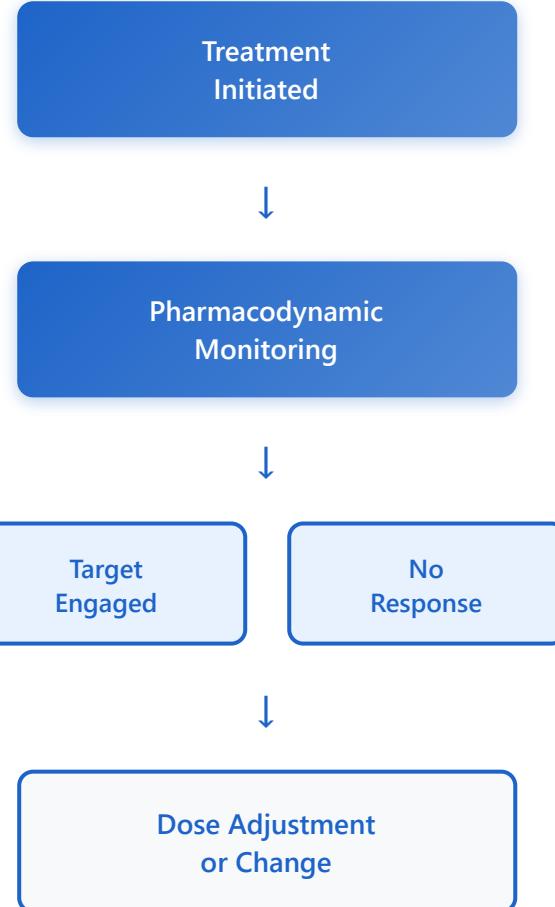
- ✓ Treatment-specific indication
- ✓ Enables precision medicine approach
- ✓ Prevents ineffective treatments

Pharmacodynamic biomarkers measure the biological response to therapeutic intervention, showing whether a drug is having its intended effect on target tissues or pathways. They are essential for dose optimization and confirming target engagement during drug development and clinical use.

Clinical Examples:

- ▶ **Blood pressure reduction:** Response to antihypertensive drugs
- ▶ **HbA1c levels:** Monitoring glucose control with diabetes medications
- ▶ **Tumor shrinkage (imaging):** Response to chemotherapy or targeted therapy
- ▶ **Cytokine levels:** Biological response to immunotherapy
- ▶ **Target protein inhibition:** Confirms kinase inhibitor activity

Key Characteristics:



Pharmacodynamic Monitoring Workflow

- ✓ Demonstrates drug mechanism of action
- ✓ Guides dose optimization
- ✓ Confirms target engagement

Safety biomarkers detect potential adverse effects or toxicity related to therapeutic interventions. They enable early identification of organ damage or dysfunction, allowing for timely intervention to prevent serious complications and guide safe dosing strategies.

Clinical Examples:

- ▶ **Liver enzymes (ALT/AST):** Detect drug-induced liver injury
- ▶ **Creatinine/eGFR:** Monitor kidney function and nephrotoxicity
- ▶ **Troponin elevation:** Detects cardiotoxicity from chemotherapy
- ▶ **Complete blood count:** Monitors for bone marrow suppression
- ▶ **QT interval (ECG):** Identifies cardiac arrhythmia risk

Key Characteristics:

- ✓ Early warning of toxicity
- ✓ Enables dose modification
- ✓ Protects patient safety

Treatment
Ongoing



Safety Biomarker
Monitoring



Normal
Range

Abnormal
Values



Continue or
Modify Treatment

Safety Monitoring Workflow

Integration in Clinical Practice

In modern precision medicine, multiple biomarker types are often used in combination throughout the patient journey—from initial diagnosis to treatment selection, monitoring, and safety surveillance. Understanding these distinct classifications enables clinicians to optimize therapeutic strategies and improve patient outcomes through data-driven, personalized care approaches.