

Biomarkers for disease identification/outcome



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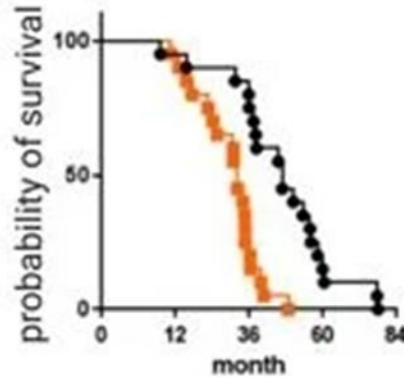
October 10, 2025

Biomarkers

diagnostic



prognostic



predictive



clinical and laboratory features



protein



DNA



metabolome



microbiome



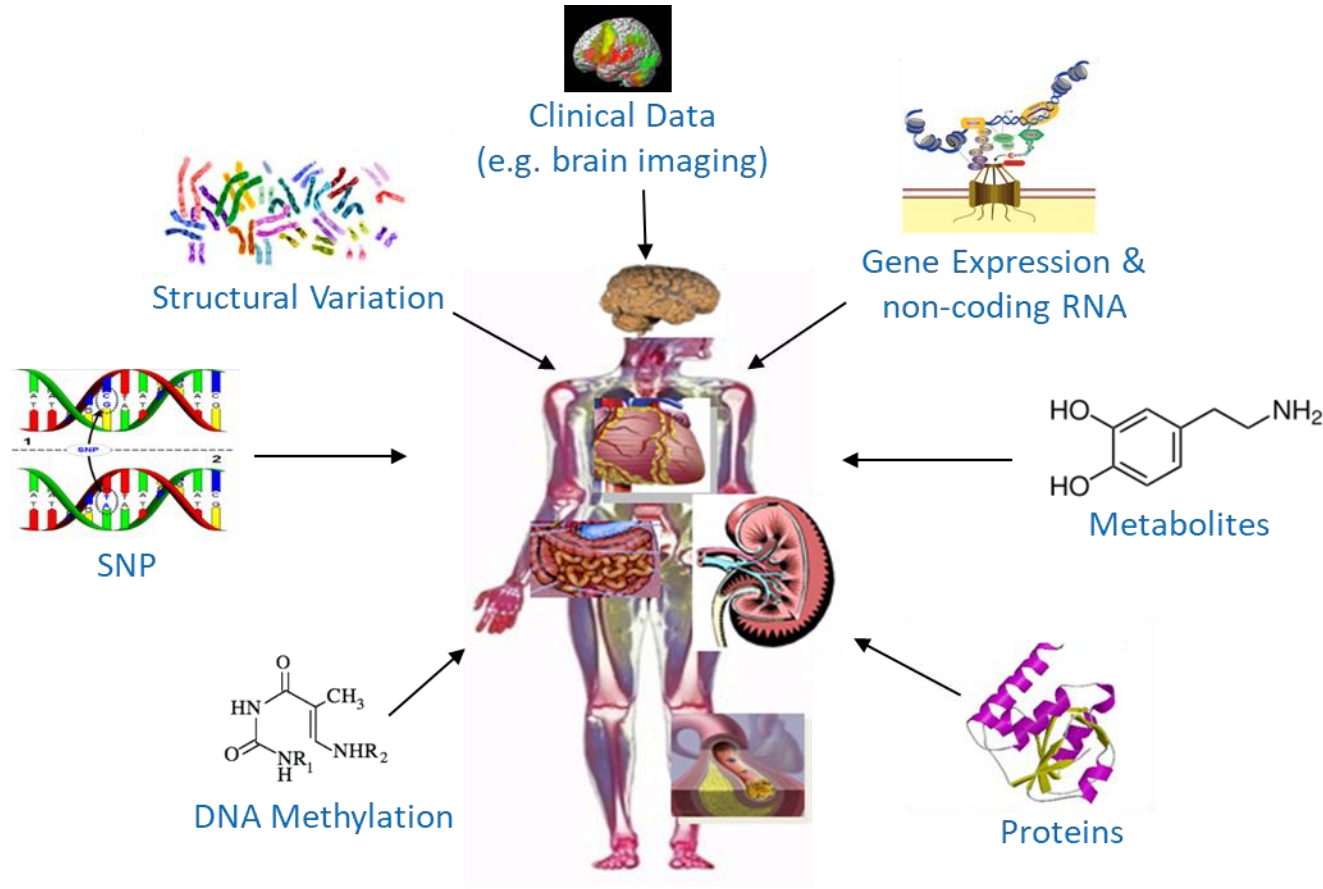
immune /stroma



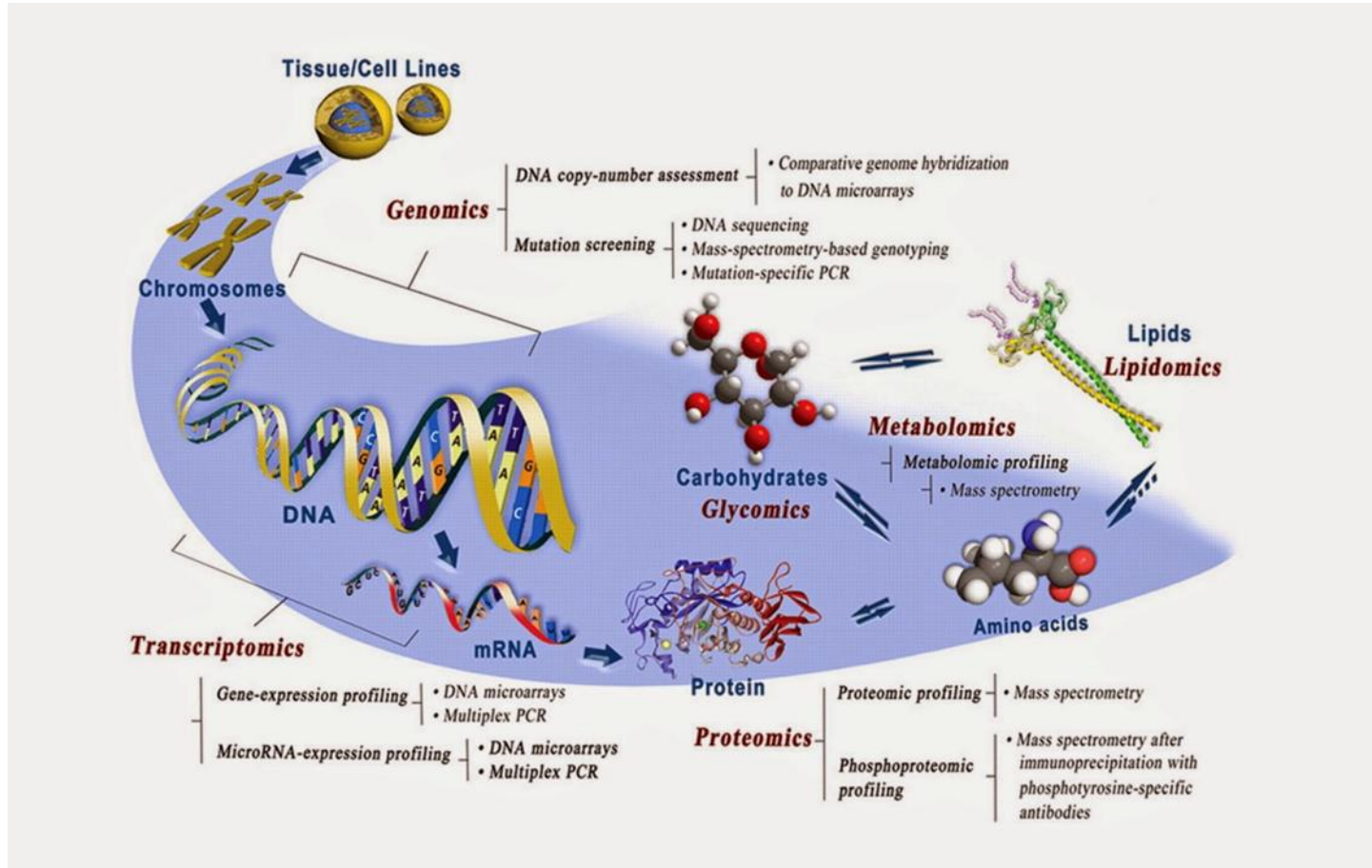
Biomarkers

- Diagnostic biomarker: To identify a disease or disease conditions. Wide range of test of disease identifications and level of severity.
- Prognostic biomarker: To predicting outcome of treatment or survival probability of patients (e.g., risk prediction). Important to understand advantage of a treatment.
- Disease forecasting: Forecasting of a disease or it severity based on biological or environmental factors

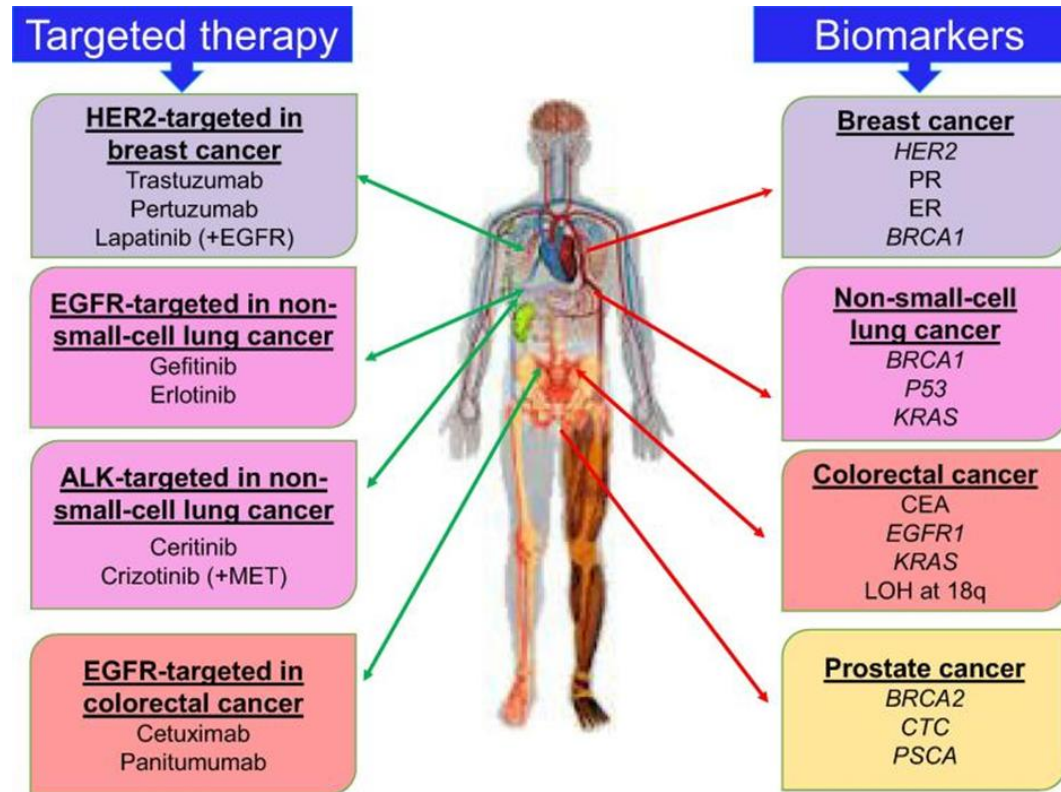
High throughput technologies for biomarker identification



Omics technologies



Importance of biomarkers in cancer treatment

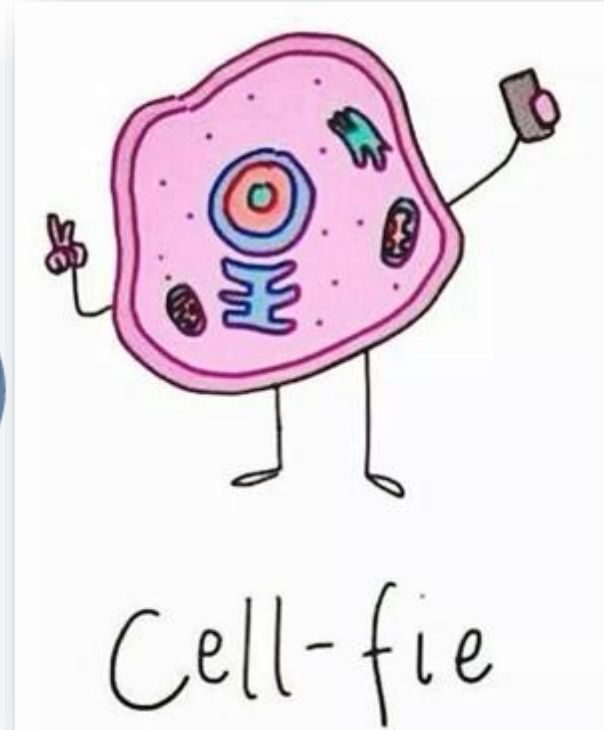
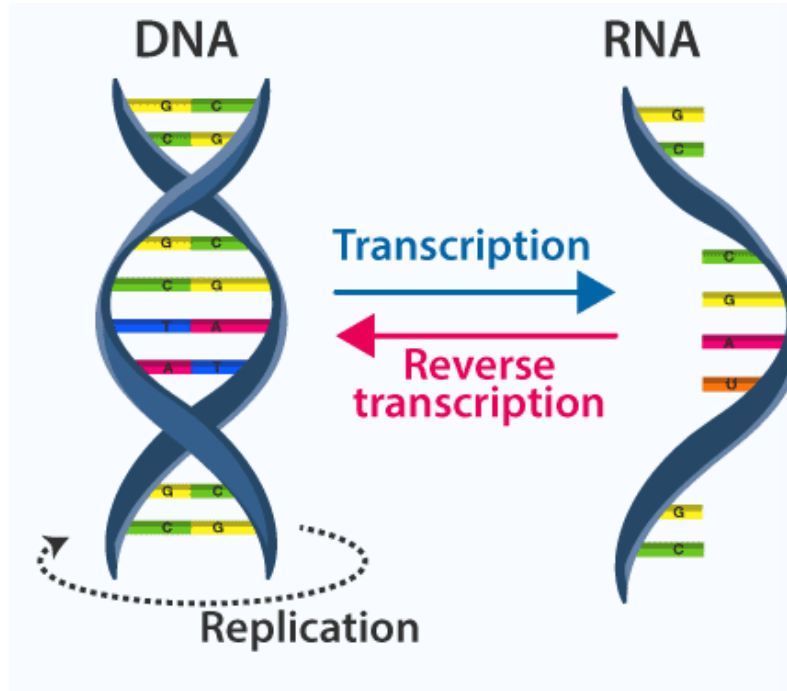


List of Targeted therapies approved for different types of cancer:

<https://www.cancer.gov/about-cancer/treatment/types/targeted-therapies/approved-drug-list>

Gene expression-based biomarker identification

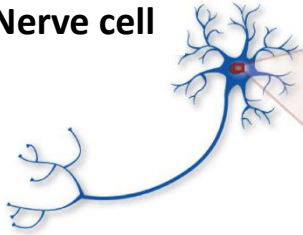
Central Dogma: DNA to RNA to Protein



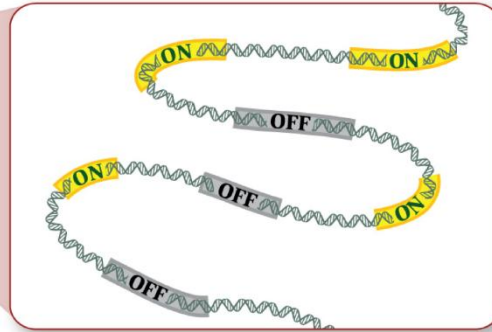
Complete set of RNAs encoded by the genome of a specific cell/organism at a specific time under specific set of conditions₇

Why Transcriptome?

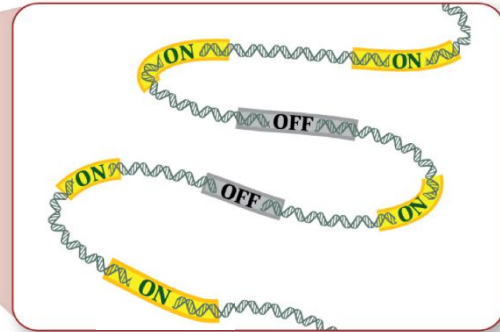
Nerve cell



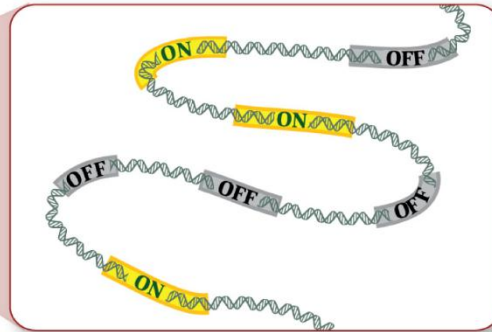
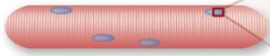
Normal



Diseased



Muscle cell



Differential gene expression analysis

which genes are expressed at different levels and reasonable for the disease ?

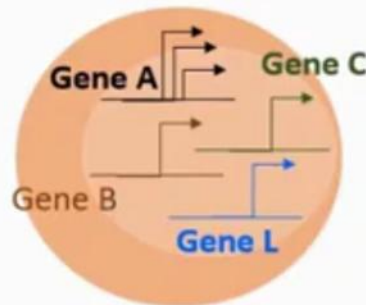
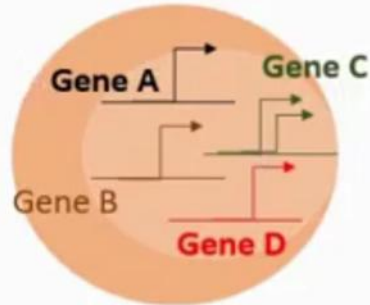


Normal cell

VS



Tumor cell



What are the differentially expressed genes?

Gene A is up regulated
Gene c is down regulated
Gene D is turned off
Gene L is turned on

Why sequence RNA (Versus DNA)?

1. *Functional studies*

Genome may be constant but an experimental condition has a profound effect on the gene expression (differential expression)

Eg. Drug vs. untreated cells

Eg. Wild type vs. knock out mice cells

2. *Predicting transcript sequence from genome sequence is difficult*

3. *Some molecular features can only be observed at the RNA level*

Alternative isoforms, fusion transcripts, RNA editing

4. *Understand allele specific expression*