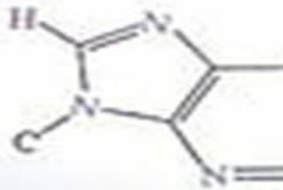


Advances in Biomarkers & Precision Diagnostics in Cancer

Dr Chin Loi Khim
Consultant Pathologist
Hospital Tunku Azizah

10th September 2025



Advances in Biomarkers & Precision Diagnostics in Cancer

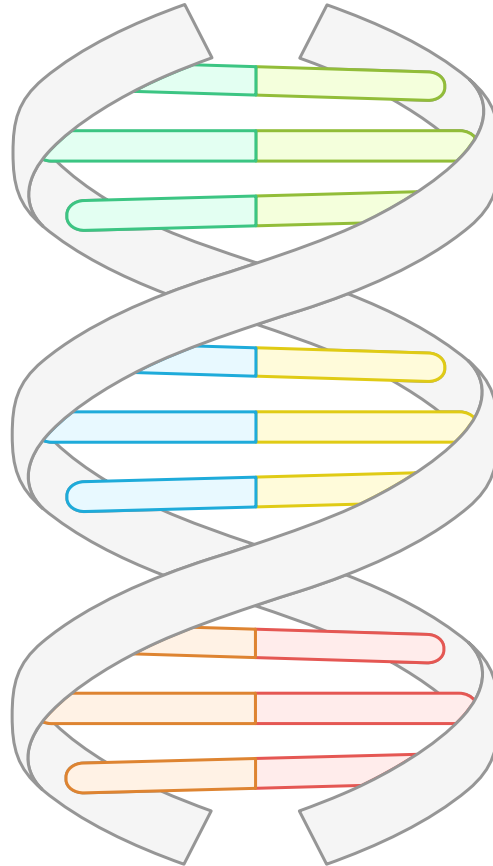
Importance of Biomarkers



AI & Machine Learning



Precision medicine



**Genomic Profiling
Liquid Biopsy**



Omics Technologies

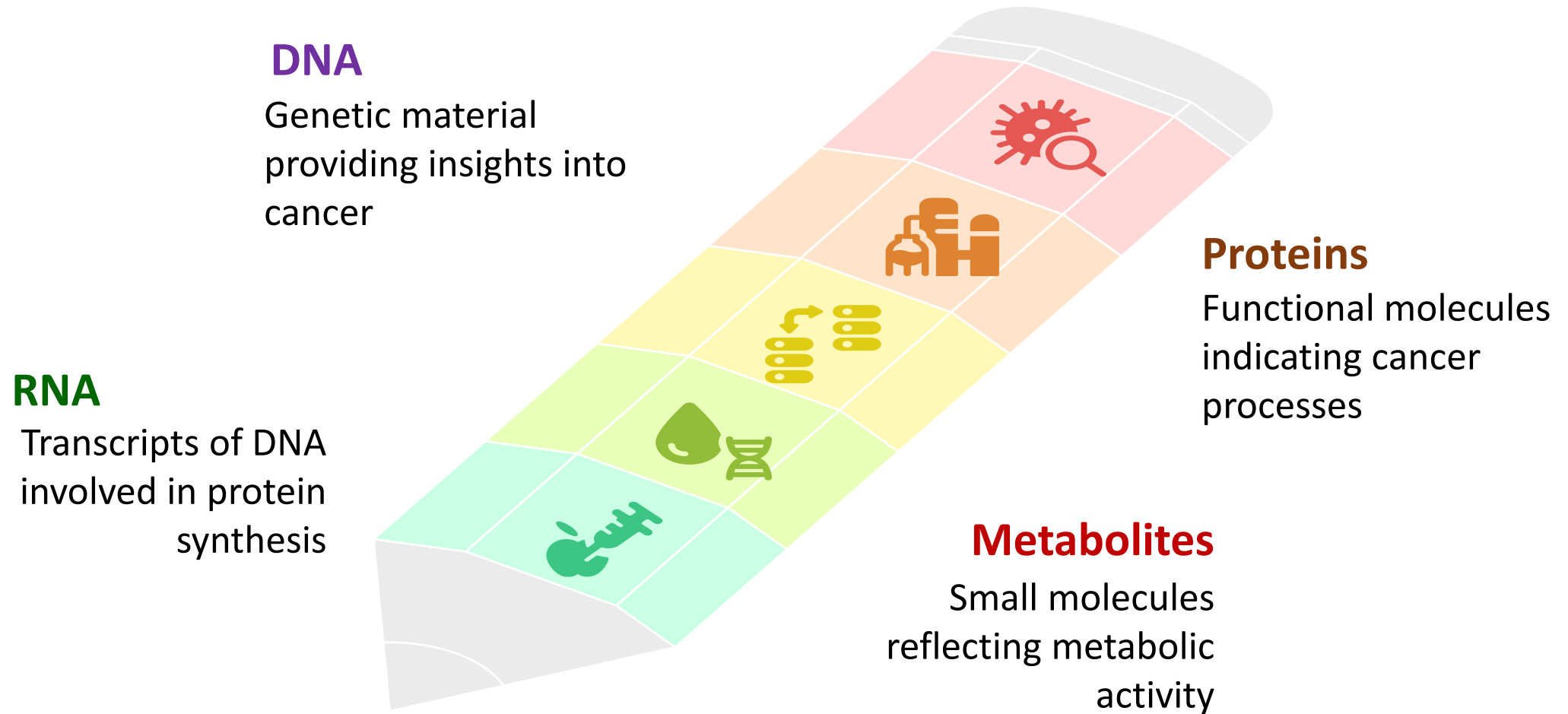


**Advancing Forward
Precision Oncology**



Decoding Biomarkers

- biomarkers are objectively measurable biological indicators





Why Biomarkers Matter



Early detection: Identifying cancer before symptoms appear.



Diagnosis: Confirm the type of cancer & molecular subtyping based on genetic alterations

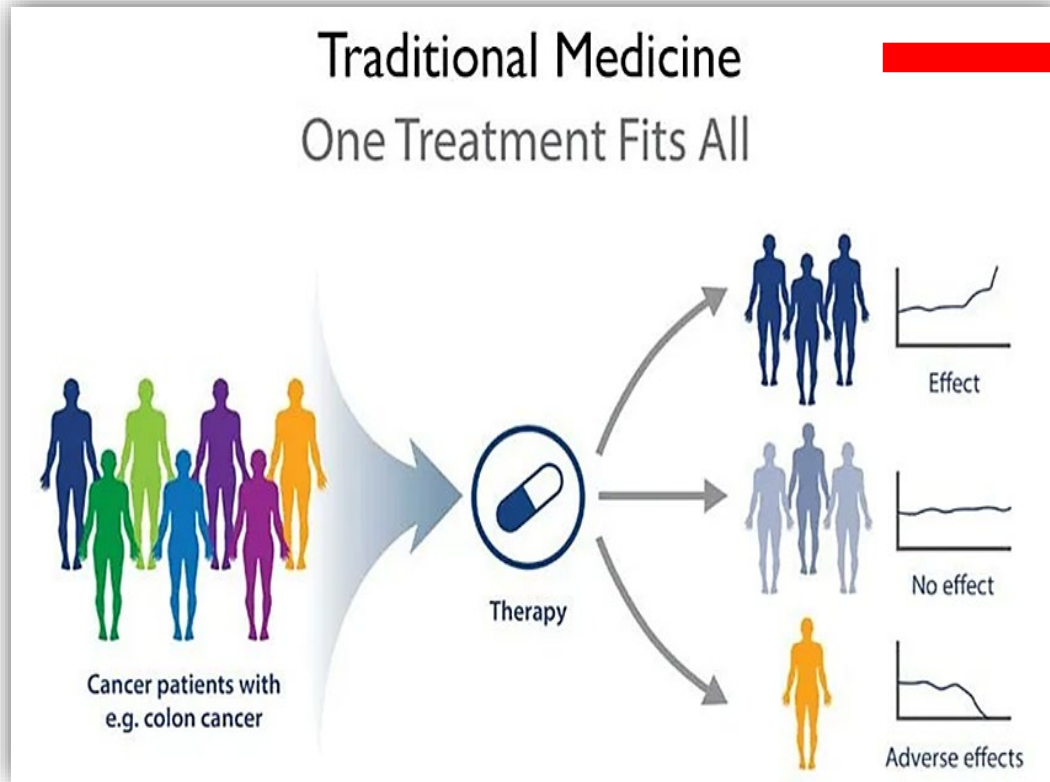


Prognosis: Predict the outcomes of the disease



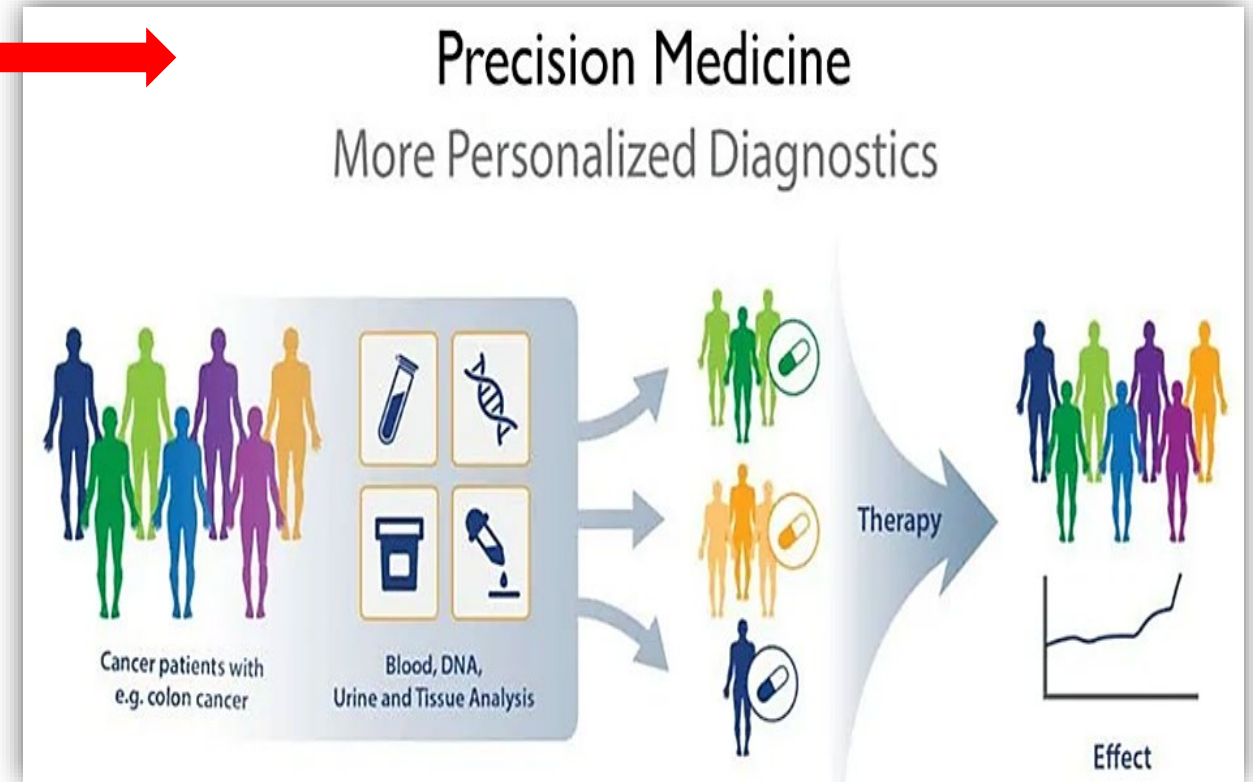
Treatment: Determining the likelihood of response to a specific therapy, eg. EGFR mutations response to EGFR inhibitors.

Paradigm Shift



Traditional therapy: same treatment for all

- Rely on a 'one-size-fits-all' approach, leading to variable efficacy & significant therapy side effects.

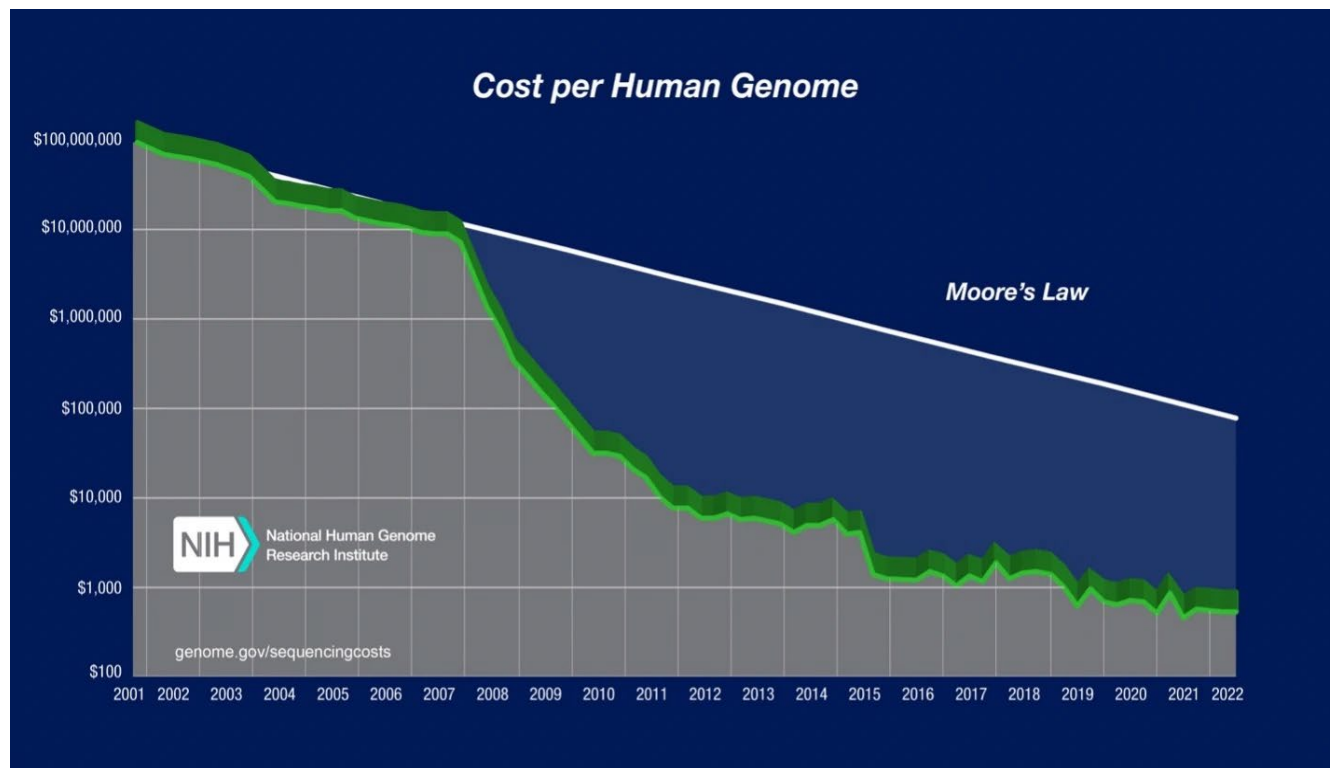


Precision medicine:

- Uses molecular biomarkers to personalise therapy
- Focuses on the specific genetic alterations in each patient's tumour

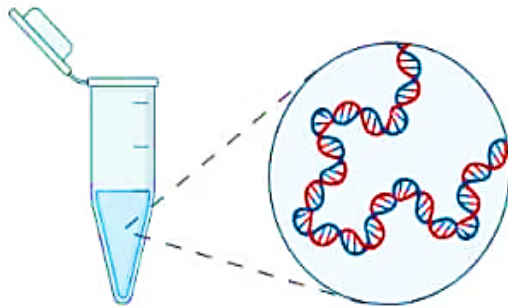
Genomic Profiling

- Next Generation Sequencing (NGS) enables rapid, massively parallel sequencing of DNA & RNA.
- Offering comprehensive analysis of the cancer genome & transcriptome while significantly reducing cost & turnaround time.



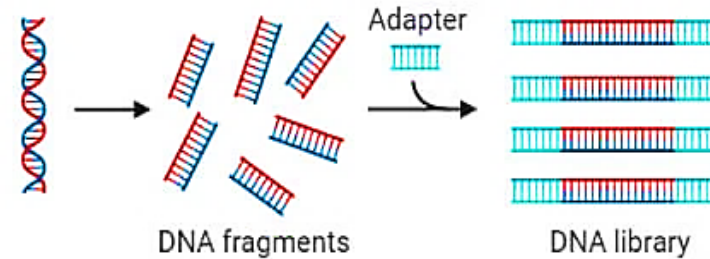
Within just two decades (early 2000s–2022), the cost of sequencing a genome has plummeted dramatically, revolutionising accessibility to genomic medicine

Step 1:
DNA extraction



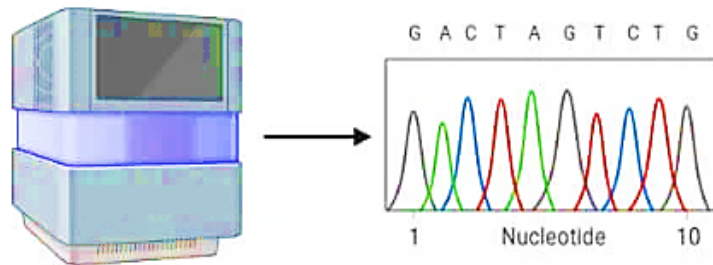
Step 2:

Library preparation



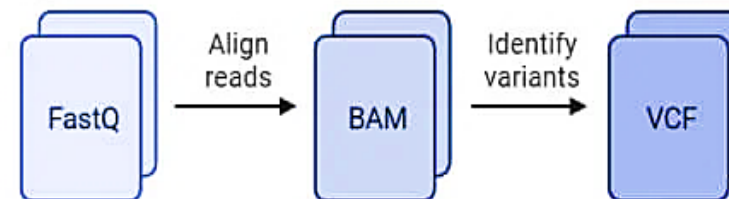
**Next Generation
Sequencing Workflow**

Step 3:
Sequencing



Step 4:

**Bioinformatics data
analysis & interpretation**

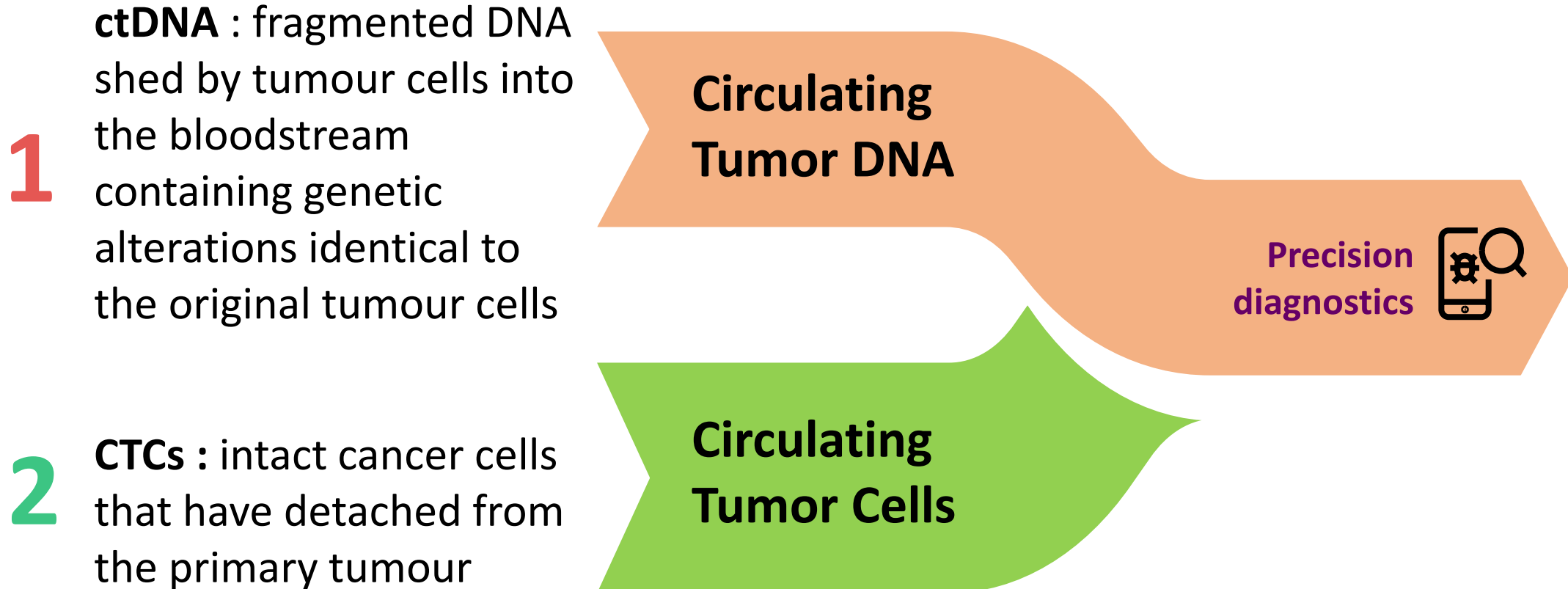


Basic methodology
of NGS includes

- sample processing & nucleic acid extraction,
- library preparation,
- sequencing,
- bioinformatics data analysis & interpretation

Liquid Biopsy

Non-invasive Blood-Based Cancer Biomarkers



Multifaceted Benefits of Liquid Biopsy



Early Detection

- Detects cancer recurrence earlier than traditional methods



Monitoring

- Assesses treatment response in real-time



Minimally invasive

- Provides a less invasive alternative to tissue biopsy

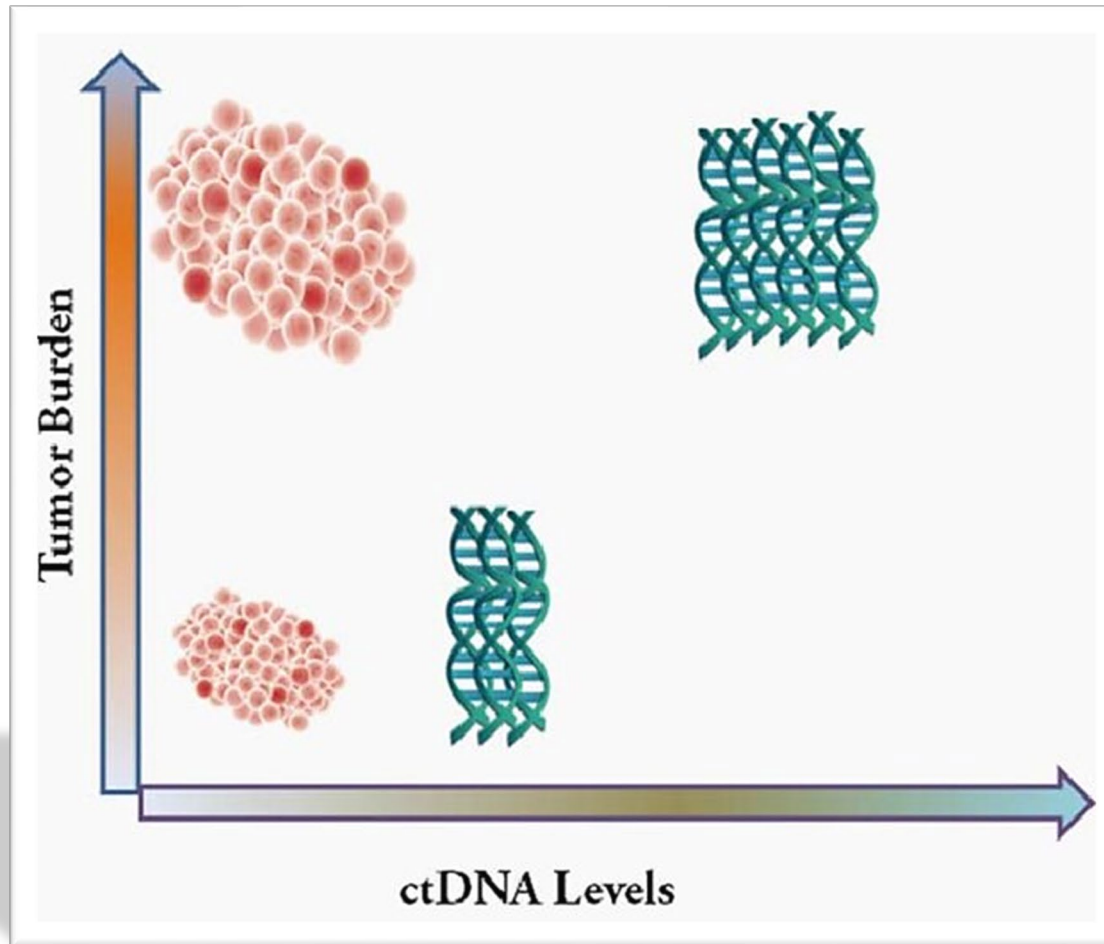


Comprehensive Snapshot

- Gives a broader view of tumour heterogeneity



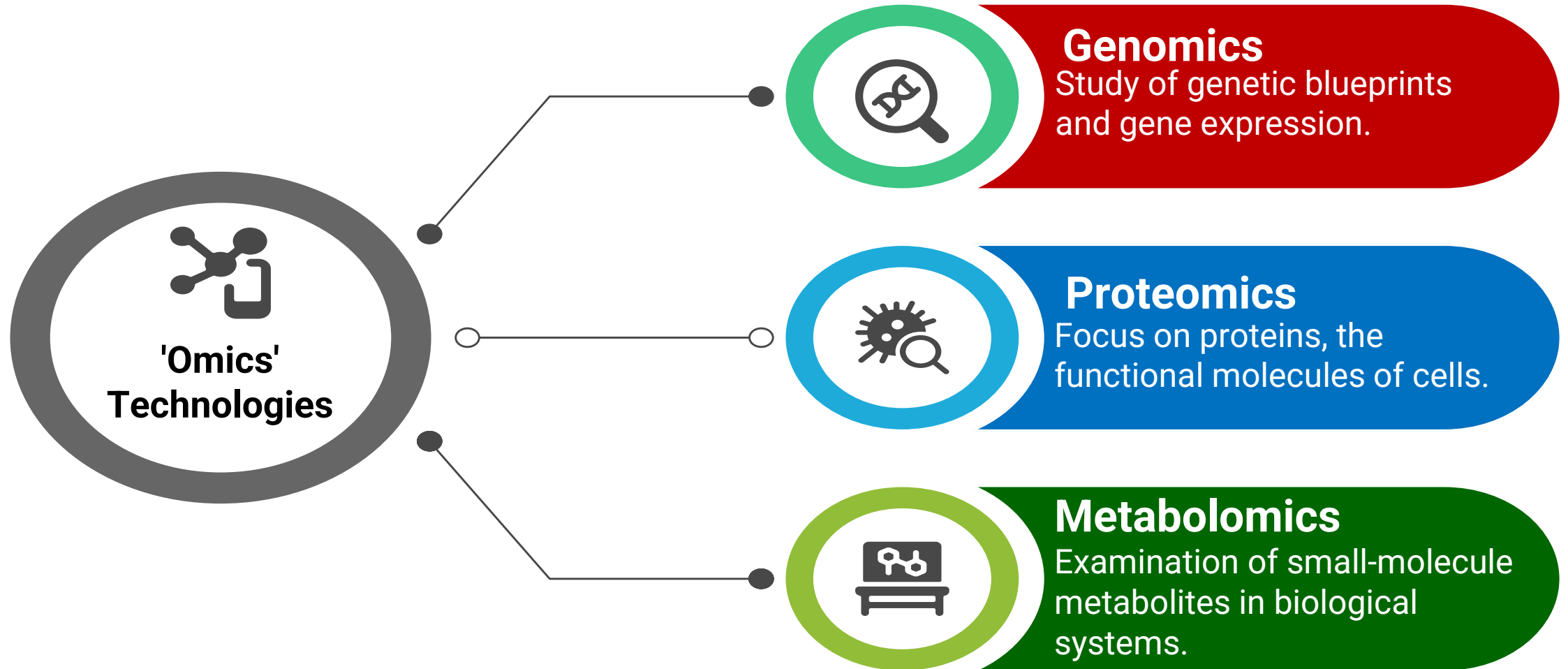
A direct correlation -- tumour burden and plasma ctDNA levels



- ✓ As tumour burden increases, ctDNA accumulates in the plasma.
- ✓ With therapeutic intervention, tumour burden and ctDNA levels decrease.
- ✓ Thus, ctDNA can serve as a surrogate marker of tumour progression & regression.

Comprehensive understanding of cancer through 'Omics'

The 'omics' technologies provide a snapshot of the functional state of cells & tissues





AI in Medicine

Smarter, Faster, Sharper

Novel Biomarker Identification

AI able to uncover new biological markers for disease detection



Disease Progression Insights

AI reveals how diseases progress over time



Prognosis Prediction

AI predicts the likely outcome of a disease



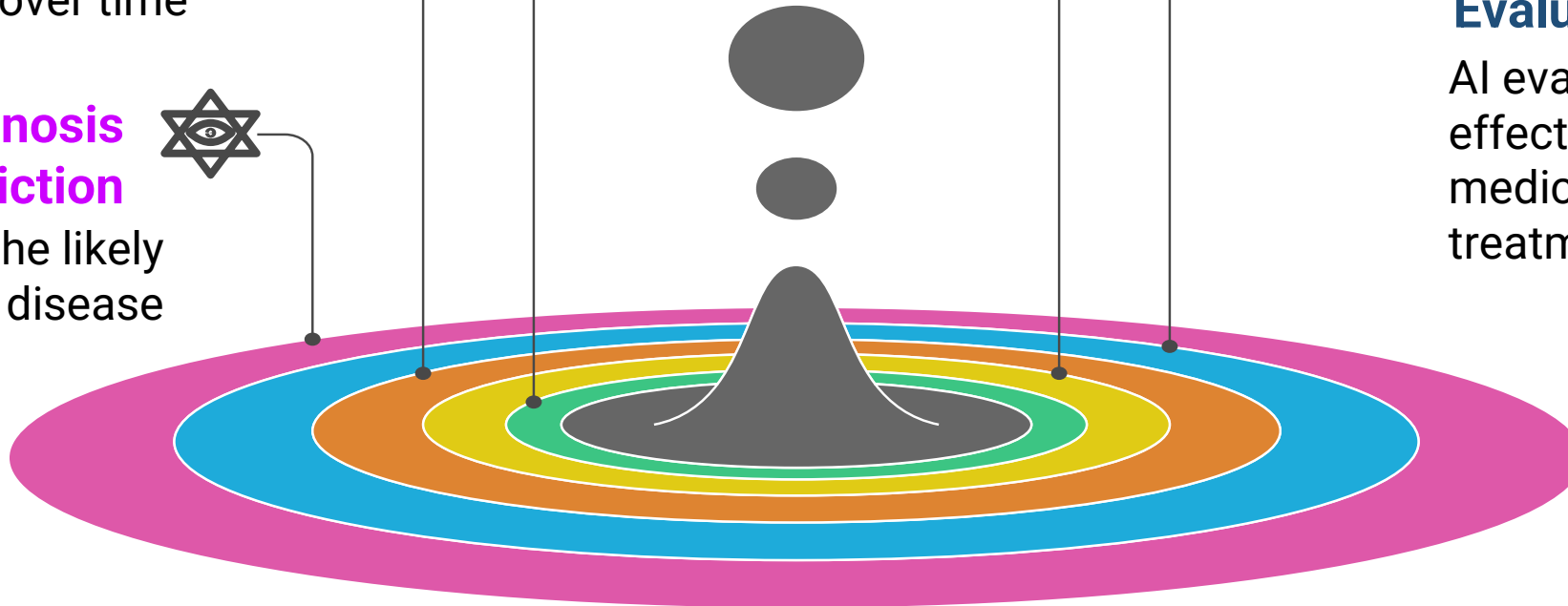
Drug Response Prediction

AI predicts how patients will respond to different drugs



Treatment Response Evaluation

AI evaluates the effectiveness of medical treatments



Clinical Impact of Precision Diagnostics & Targeted Therapy





Precision Diagnostics



Early Detection

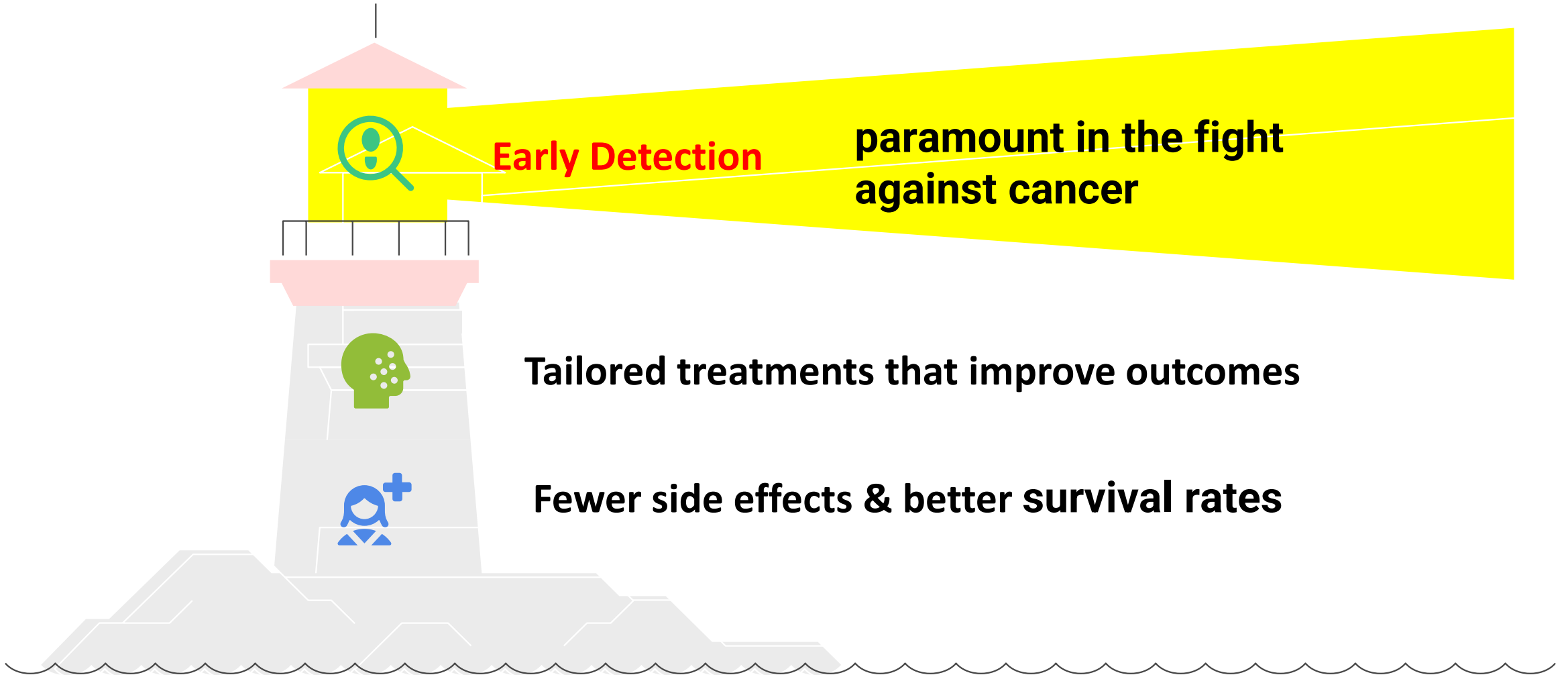
**paramount in the fight
against cancer**



Tailored treatments that improve outcomes



Fewer side effects & better survival rates



How Targeted Therapy Work

- block specific molecules or pathways essential for cancer growth & survival



Small-Molecule Inhibitors

- Block intracellular targets, inhibiting cancer cell growth eg. tyrosine kinases



Monoclonal Antibodies

- Bind to cell surface targets such as receptors, thereby disrupting cancer progression

Targeted Therapies

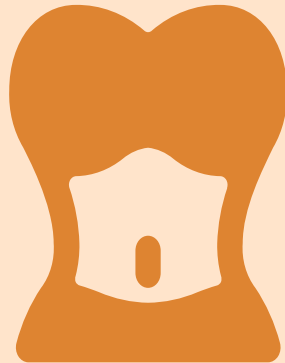
EGFR Inhibitors

Gefitinib targets
EGFR
mutations in
NSCLC



HER2 Inhibitors

Trastuzumab
targets
HER2
overexpression
in breast cancer



PARP Inhibitors

Olaparib targets
BRCA1/2
mutations in
ovarian & breast
cancer



BRAF Inhibitors

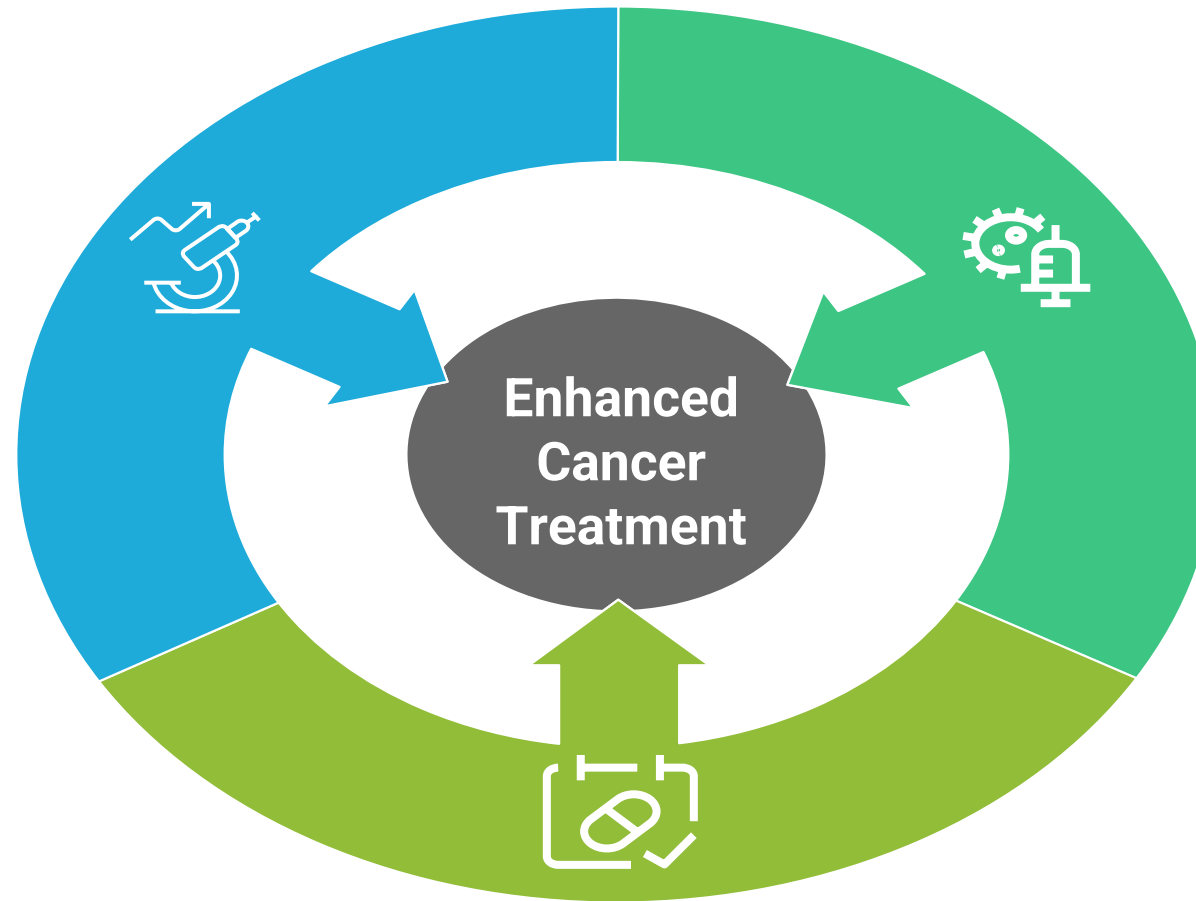
Dabrafenib targets
BRAF V600
mutations
in melanoma





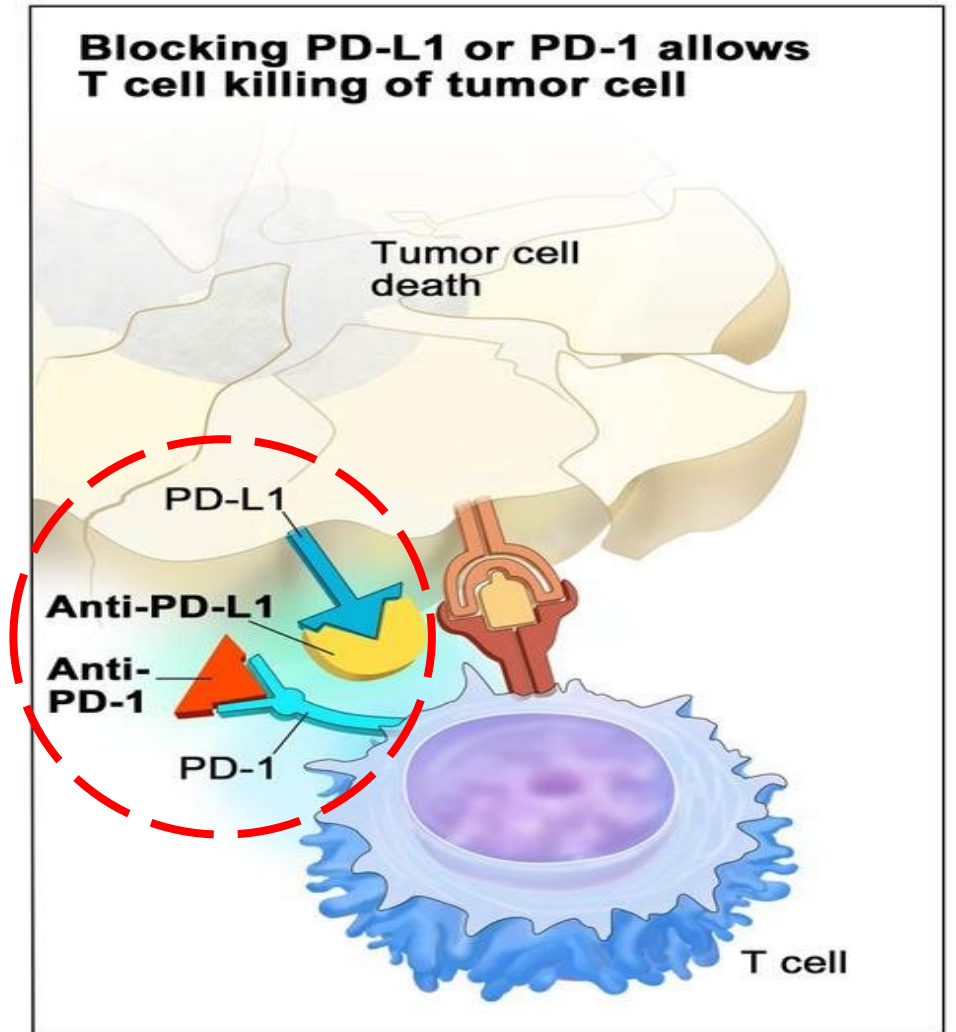
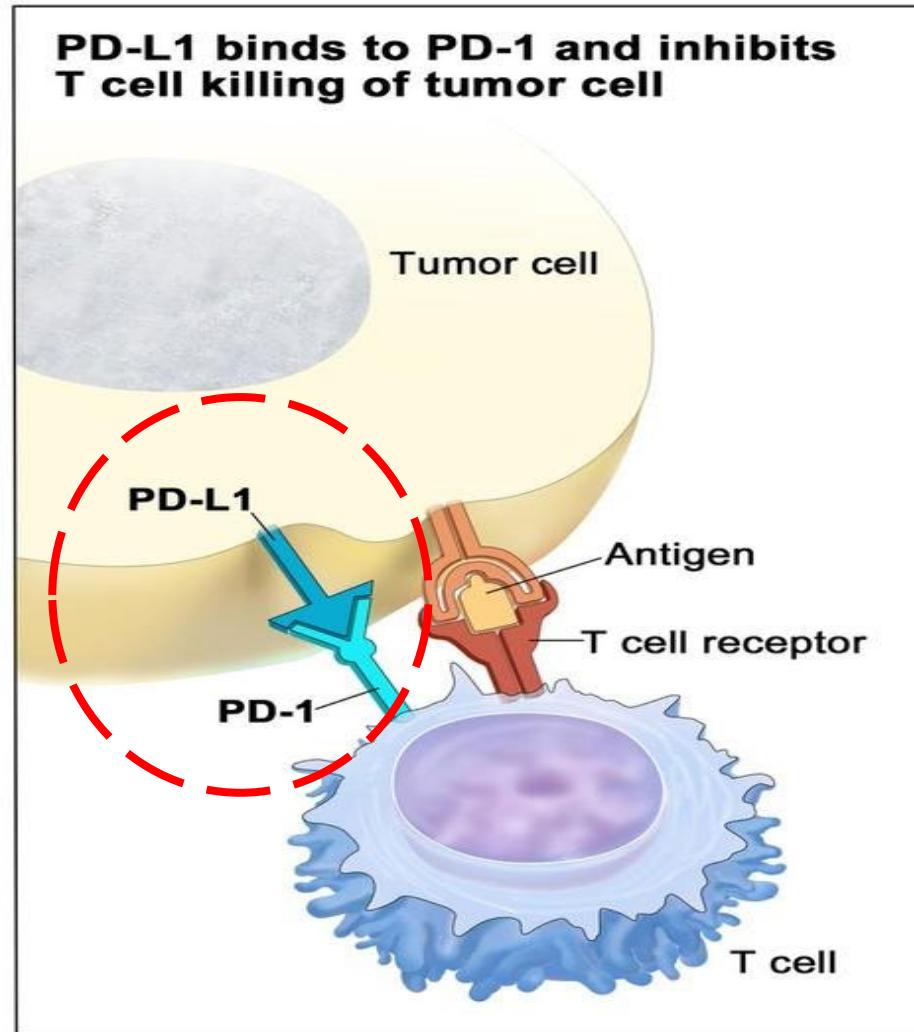
A Game Changer - Immunotherapy

Biomarker Testing
Identifies patients likely to benefit from immunotherapy



Immunotherapy
Stimulates the immune system to attack cancer cells

Immune Checkpoint inhibitors
Drugs (e.g. PD-L1,) block signals that stop our immune cells from attacking cancer cells



Checkpoint proteins --

PD-L1 binding to PD-1 shuts down T cells, allows tumour cells to survive

Immune Checkpoint Inhibitor drug block PD-L1–PD-1 unleashes T cells to kill tumour cells.

Unlocking Precision Medicine Through Multi-Omics

Genomics

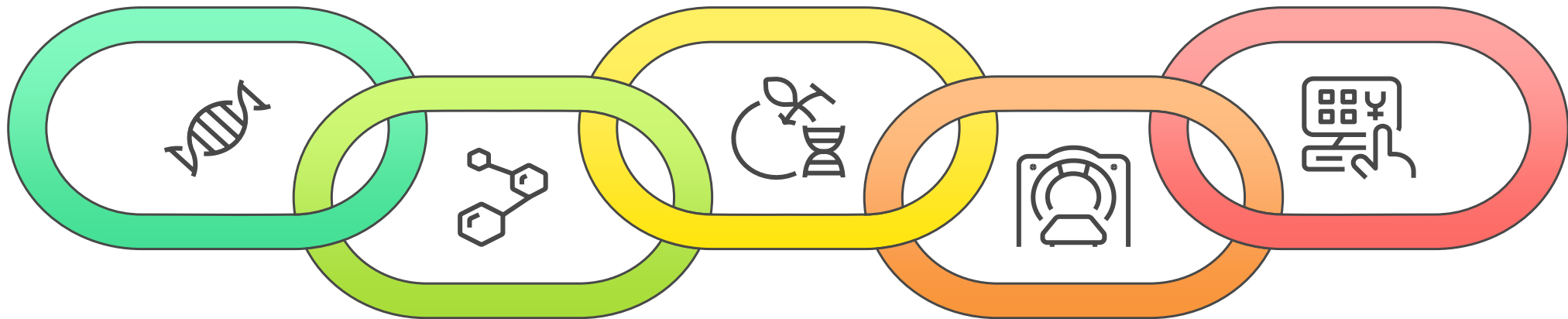
Study of genes and their functions in cancer

Metabolomics

Study of metabolic processes in cancer cells

Transcriptomics

Study of all RNA transcripts to reveal gene activity



Proteomics

Analysis of proteins and their role in cancer

Radiomics

Use of advanced imaging techniques to predict tumour behaviour & response

Future Directions in Precision Oncology





Emerging Biomarkers

Shaping the Future of Precision Oncology

Microbiome-based Biomarkers

Helps predict immunotherapy outcomes & provides insight into cancer development.

Epigenetic Biomarkers

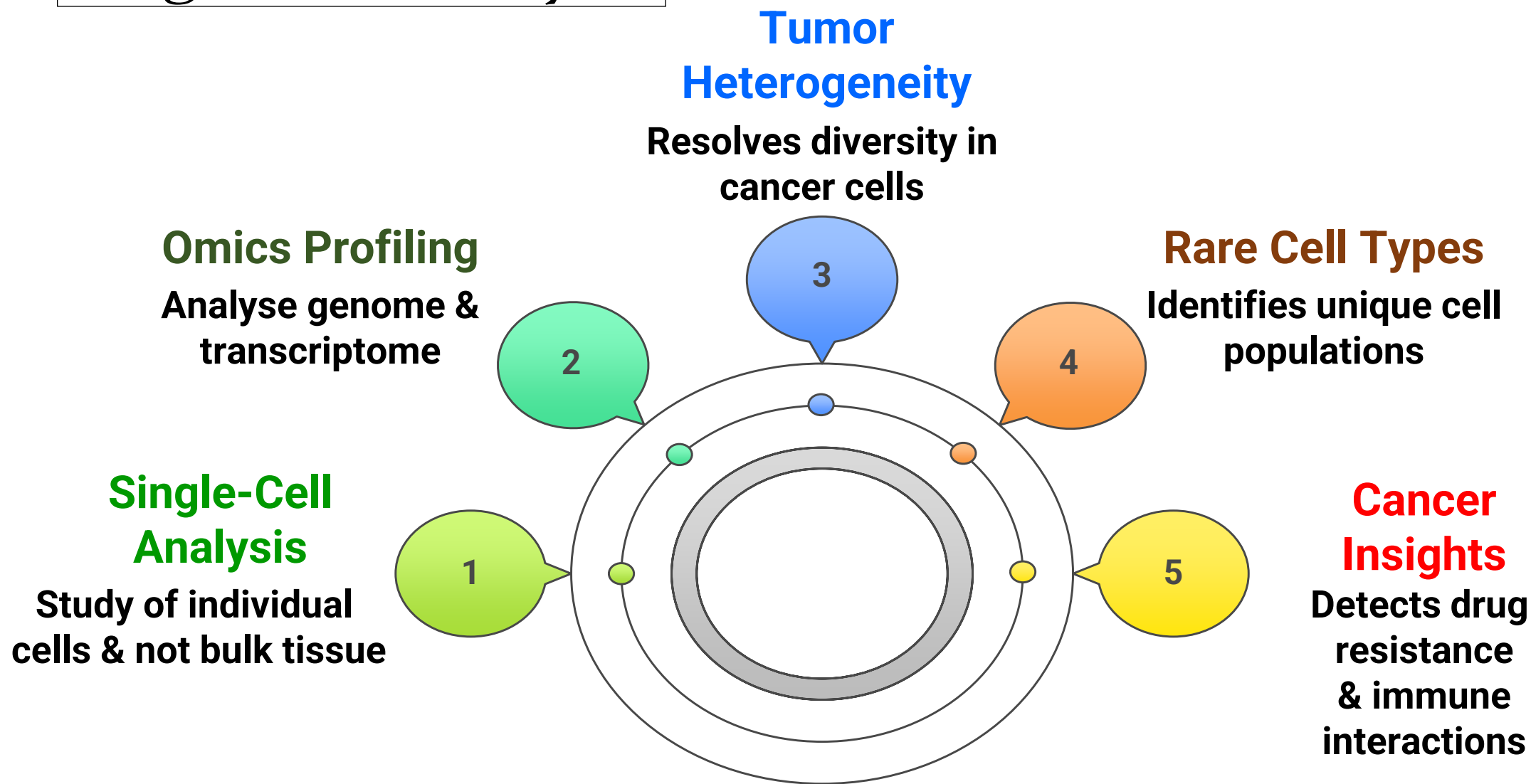
Supports early detection & prognosis through DNA methylation.

Exosomal Biomarkers

Offers non-invasive real-time tumour status monitoring via liquid biopsy.

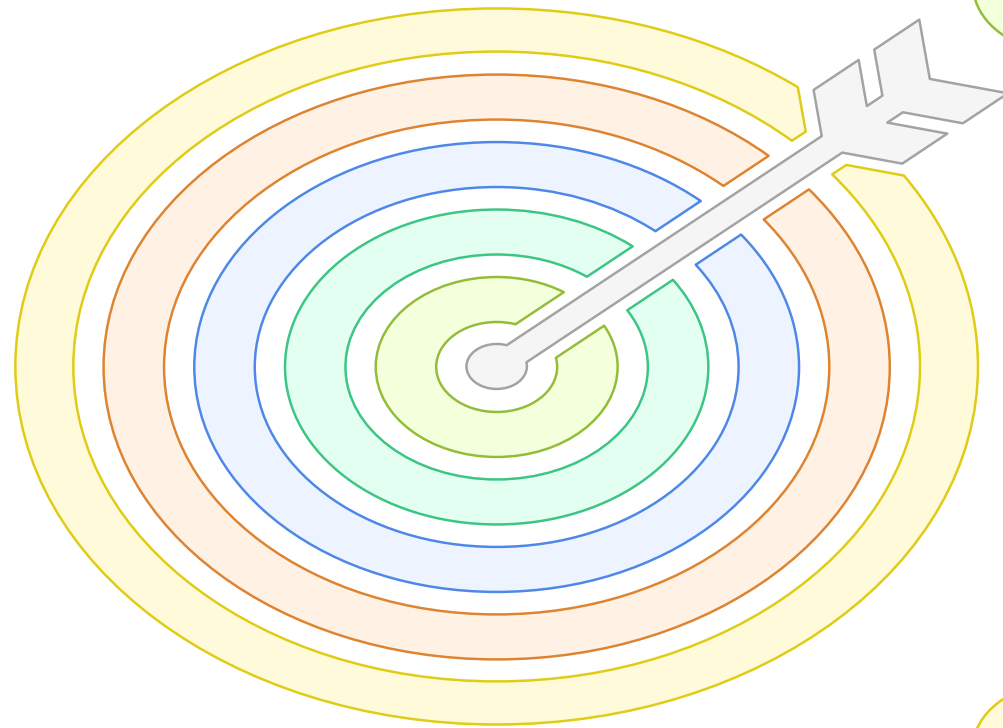


Single-Cell Analysis





Spatial Transcriptomics



Spatial Transcriptomics

Maps gene expression to its physical location in tissue



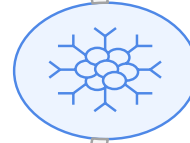
Tissue Architecture Preservation

Maintains tissue context



Tumor-Immune Niches

Defines Tumor-Immune interactions within tumor microenvironment



Immune Hot & Cold Tumors

Explains Immune activity - shapes immunotherapy response



Therapy Response

Links spatial data to treatment outcomes



Barriers to Precision Medicine

Visible hurdles in precision medicine

Assay Reproducibility
Ensuring consistent
biomarker results across labs.



Data Analysis Complexity
Large Omics data requires
advanced bioinformatics.



Ethical Concerns
Addressing privacy &
consent in genetic testing.



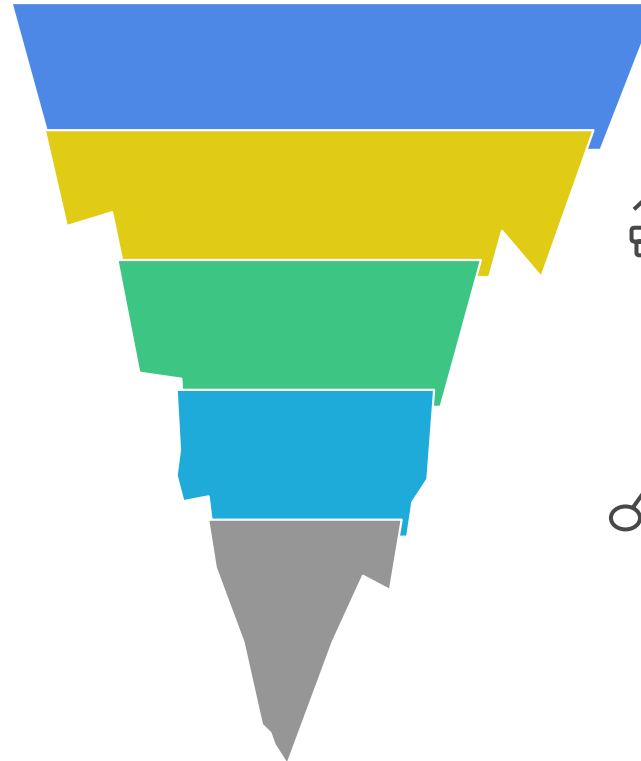
High Costs

Expensive tests limit
accessibility to precision
medicine.



Data Sharing Issues

Better data sharing &
collaboration are essential
to unlock their full potential.



Key to Precision Oncology's Future

Novel Biomarkers

Identifying new biomarkers for early detection & treatment response prediction.

Personalised vaccines

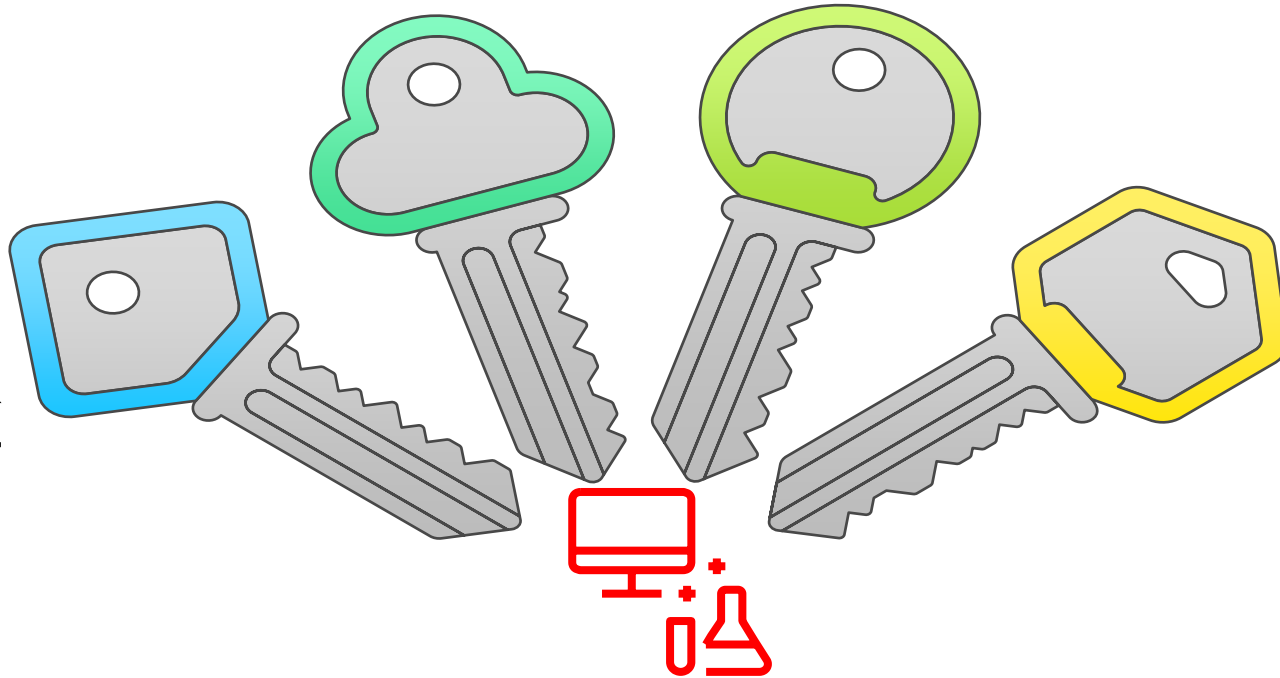
Developing vaccines targeting unique tumour mutations to stimulate immune response.

AI Integration

AI and machine learning to drive personalised treatment & biomarker discovery.

Liquid Biopsies

Expanding the use of liquid biopsies for broader cancer detection & monitoring.



Precision Oncology



Precision Oncology Journey

