GENE MUTATION
data analysis

Mahsa Ehsanifard <♥>

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If(like > 0){
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Print("enjoy")
}
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COURSE'S REFERENCES

1. Introduction

- Quick mention of mutation

- Mutation in cancer

  - role of mutation in cancer

  - key terms and vocabulary

  - types of mutation in cancer

- Biomedical strategies

2. In-silico approaches and techniques

- Data analysis with R

- Methodologies, databases

- AI and technologies for mutation

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GENE MUTATION

❖ <u>Importance</u>:

genetic testing

- Identifying inherited mutated genes -> predicting disease susceptibility.

- Early detection and personalized treatment plans

❖ <u>Types</u>:

- somatic -> occurring in body cells and not inherited

- germline -> Occur in cells producing gametes -> typically passed to half the offspring.

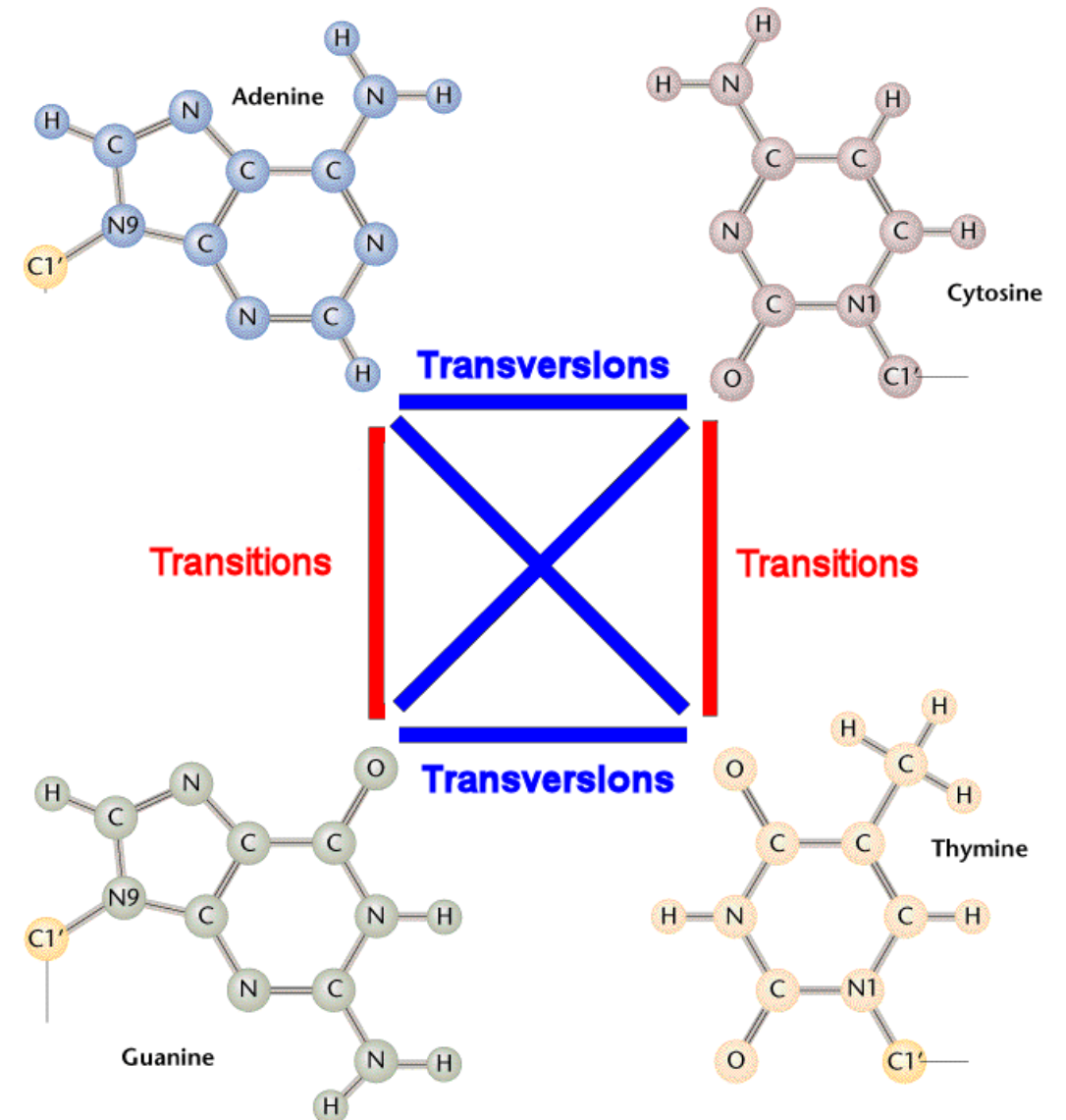☺ genecan693

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MECHANISMS

♠ Transition:

Interchanges of two-ring purines (A <-> G)

Interchanges of one-ring pyrimidines (C <-> T)

♠ Transversion:

Interchanges of purine for pyrimidine bases

(exchange of **one-ring** & **two-ring** structures)
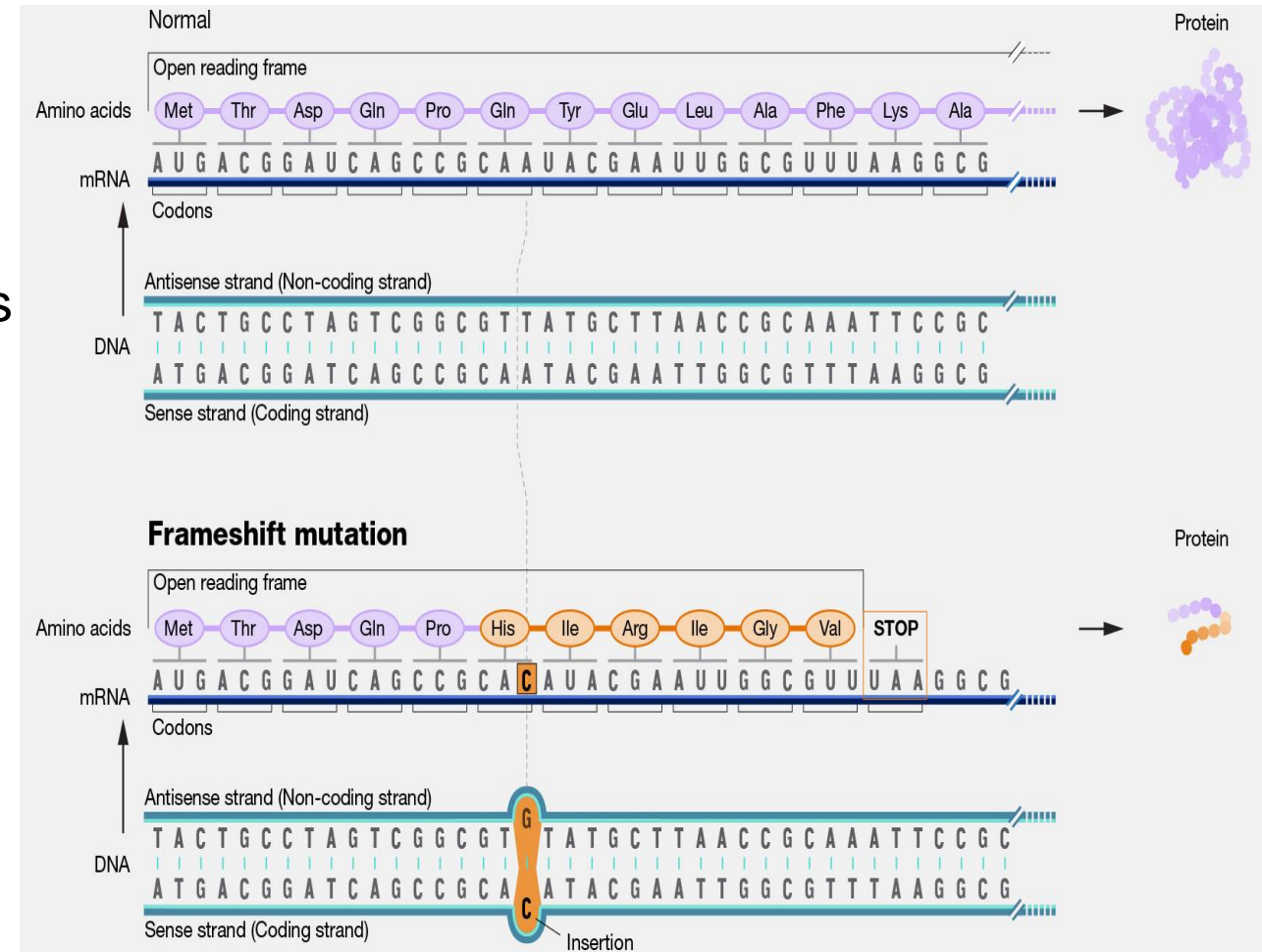
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STRUCTURAL CHANGES

♠ <u>Frameshift</u>: Disrupts codon patterns.

  Insertion or deletion of nucleotide bases

  not in multiple 3 bases

♠ <u>In-frame</u> insertions and deletions

  bases divisible by 3
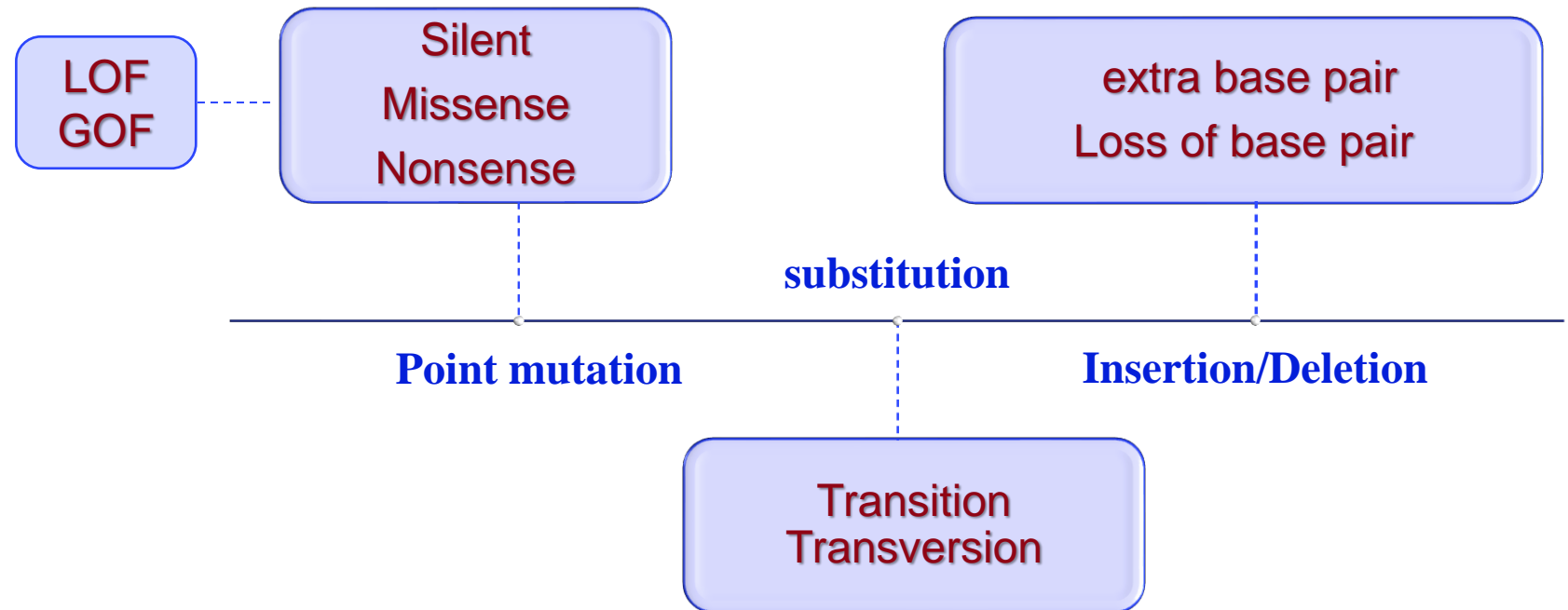
CONSEQUENCES
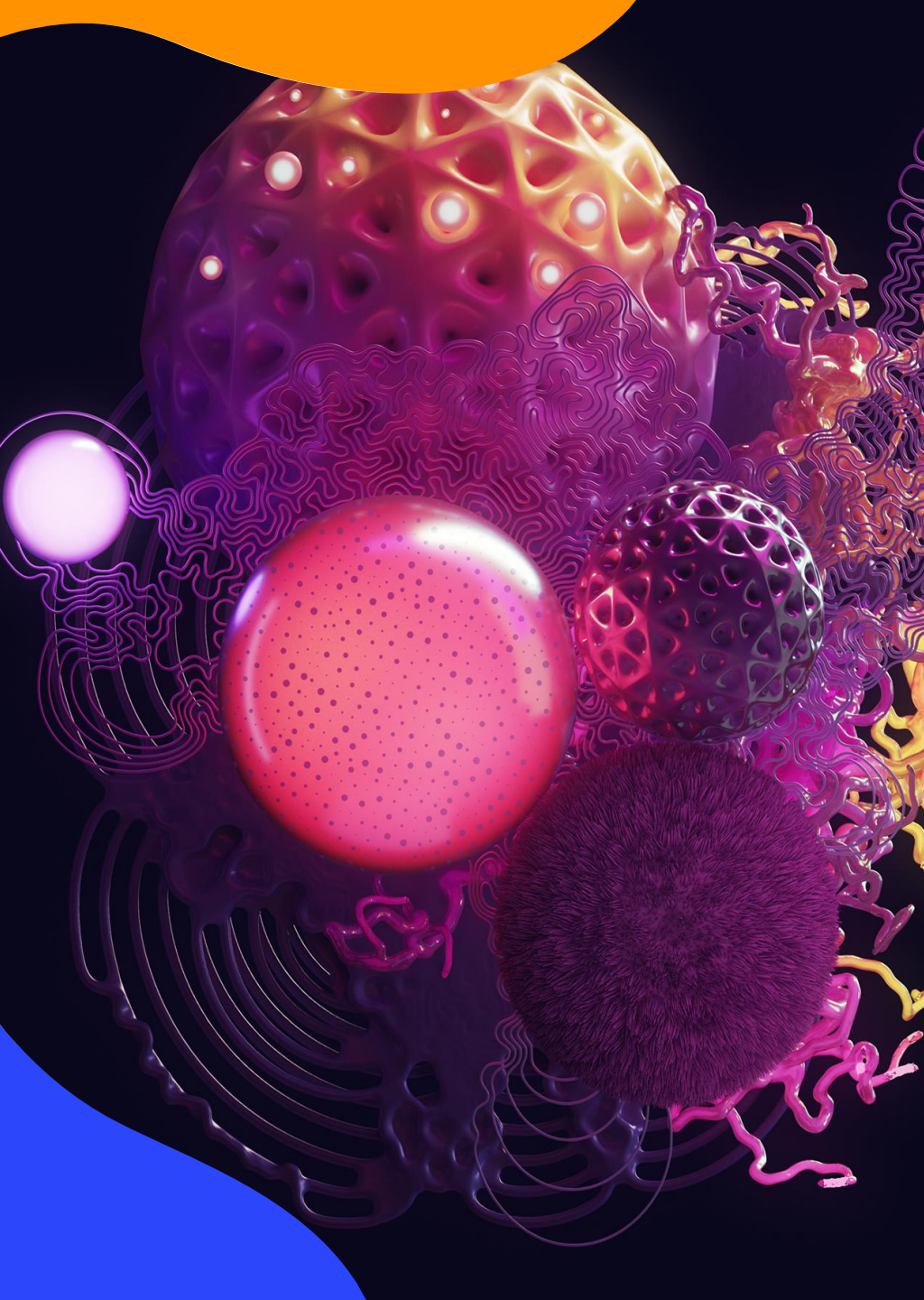
- Point mutation
- substitution
- Insertion
- Deletion
- Frameshift

Different DNA nt. mutation types

LOF
GOF

Silent
Missense
Nonsense

extra base pair
Loss of base pair

Point mutation

substitution

Insertion/Deletion

Transition
Transversion

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CANCER ♋

One of the most common disease characterized by gene mutation and abnormal gene expression.

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KEY TERMS & VOCABULARY

☻ Oncogene

Mutated (changed) forms of normal genes causing cell growth.

mutation

proto-oncogenes ⚡➔ out of control cell growth ➔ Cancer (Gain Of Function)

Mutation
Chromosome translocation
Gene amplification
Retroviral insertion

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☻ Tumor suppressor genes

(protective genes). Limit cell growth
 - Slowing down cell division
 - Repairing mismatched DNA
 - Monitoring cell division
 - Enhancing apoptosis

(Loss Of Function) ---> development of cancer.

☻ Mutagen

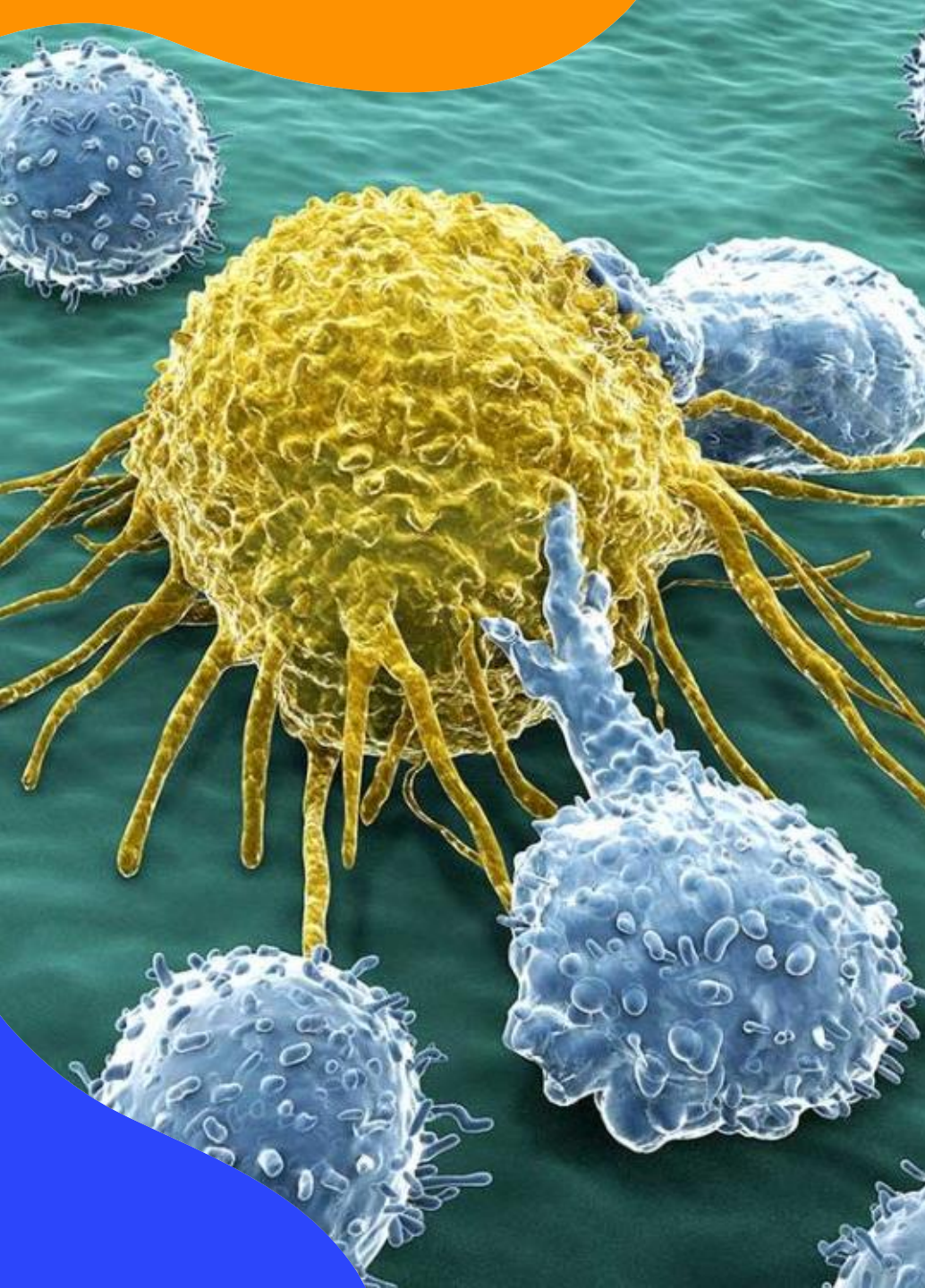A mutation-causing substance

Cancer Mutation In Biomedicine

❖ Cancer Mutations: A Central Theme in <u>Modern Biomedical Research</u>

▶ Understanding the genesis and progression of cancer.

'deep' (exome) sequencing technique -> identify all the somatic mutations in an individual tumor (somatic genotype).

▶ Gene therapy strategies

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CANCER BIOMEDICINE

- understanding the molecular mechanisms of cancer <u>progression</u>, <u>predisposition</u>, and <u>resistance to treatment</u>.

- understand the complex interactions between cancer cells and their environment.

- The development of new cancer treatments, diagnostic tools, and prevention strategies.

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Statistical Methods for Biomedicine:

advanced statistical techniques for analyzing biomedical data, including those related to cancer research

Immunodiagnostic approaches
Immunotherapy
Immunoinfilteration
Computational immunology / genomics

Cancer Prevention & Treatment in Biomedicine

The Paul-Ehrlich-Institut supports the development of new vaccines and biomedicines. Part of the Institute's responsibilities include ensuring their quality, safety, and effectiveness.

illustration monoclonal antibodies
source: ustas7777777/shutterstock.com

Paul-Ehrlich-Institut

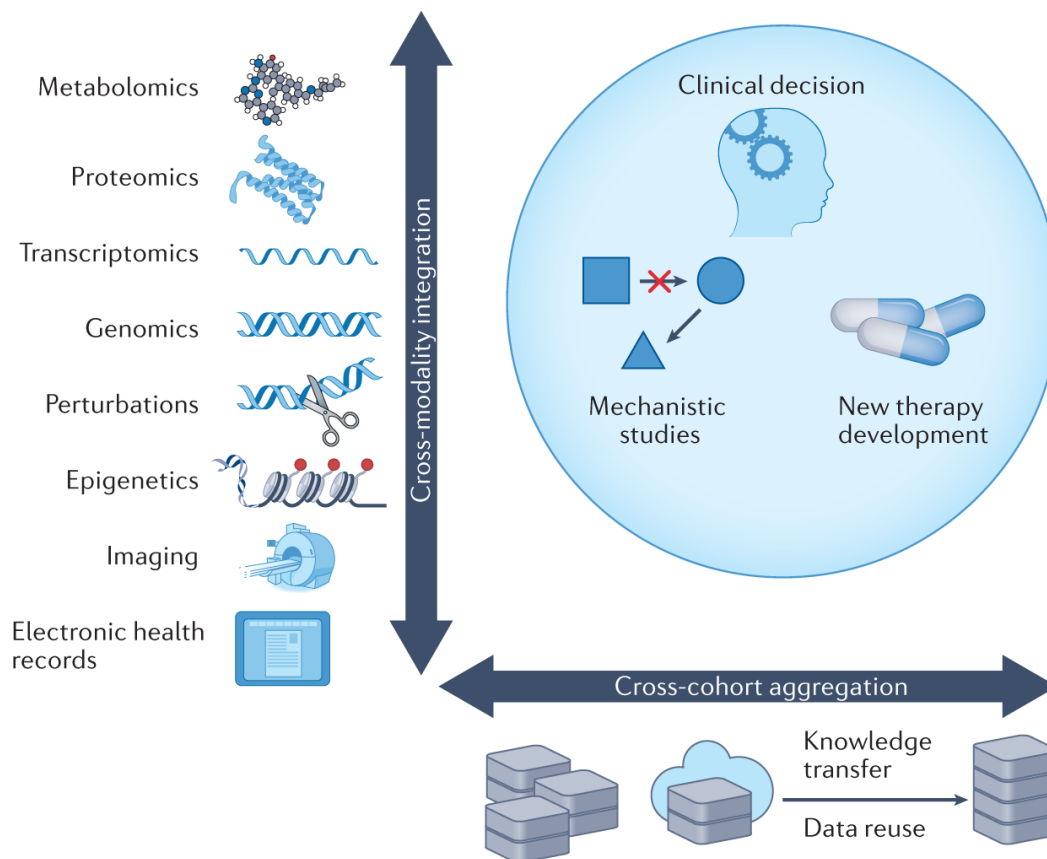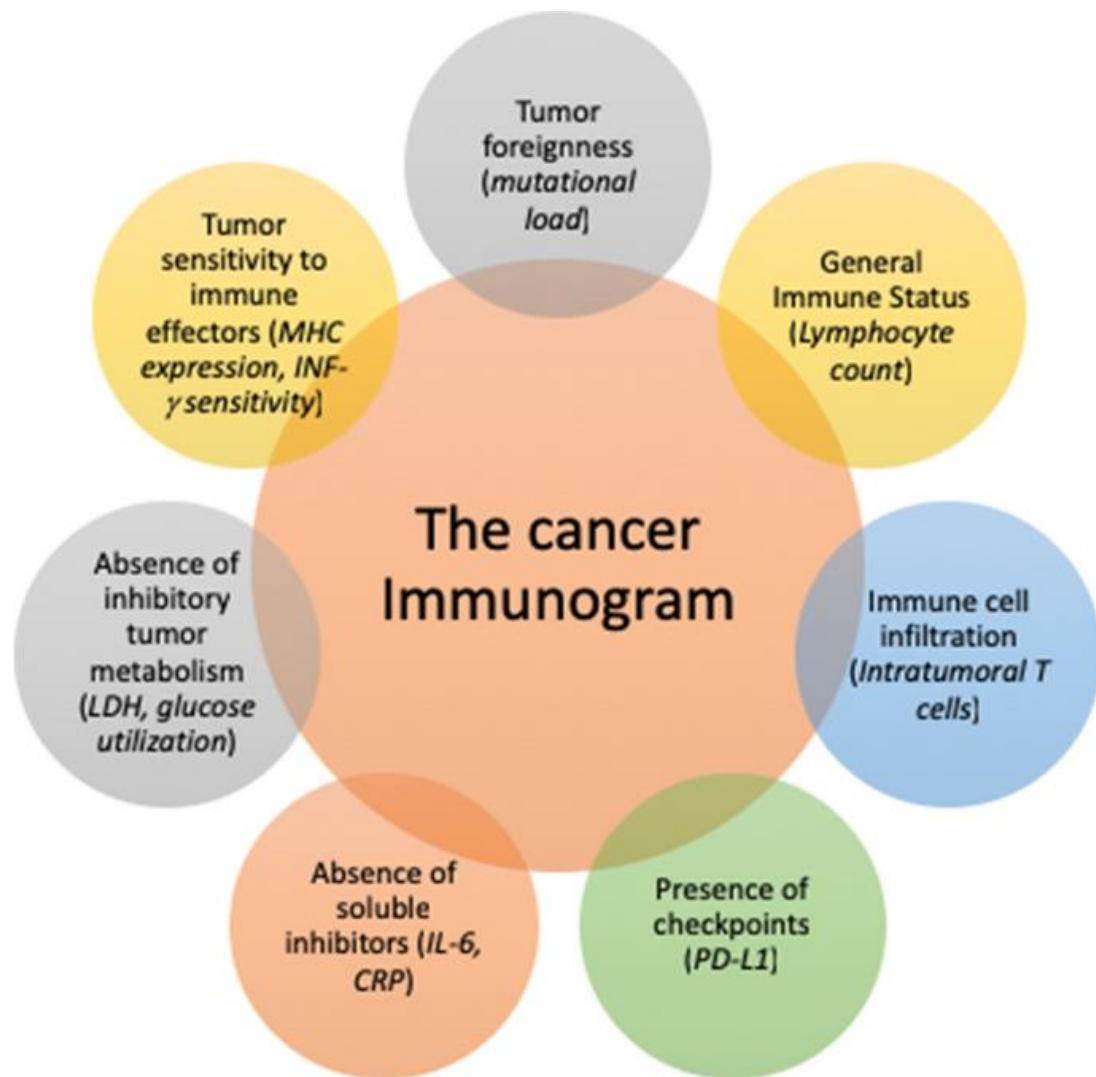Monoclonal antibodies, e.g. for the treatment of colon cancer

CAR T-cell drugs, e.g. for the treatment of blood cancer

Vaccines, e.g. for protection against HPV infection, which can cause diseases such as cervical cancer

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❖ Computational Models

▶ Localization of genetic mutations as biomarkers

- Development of new targeted therapies

(https://breakthroughsforphysicians.nm.org/urology-research-article-biomarker-cancer-genetic-mutation-urology.html)

- Novel molecular mechanisms

- Improving patient treatment

▶ Big data to target gene mutations

- Profiling patients with and without mutations

- Finding effective treatments and understand resistance mechanisms

(https://healthsciences.arizona.edu/news/stories/using-big-data-target-gene-mutations-cancer-tumor-cells)

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IN-SILICO DATA ANALYSIS

- **Databases**

- **Programming approaches**

- **AI tools**

- **Computational genomic techniques**

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DATABASES

♠ Cosmic

♠ cBioPortal

♠ dpSNP (NCBI)

♠ FireBrowse

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Ai-based method to identify cancer mutations

Variant Network (VarNet): An AI Solution :

- deep learning to detect cancer mutations

- Large Datasets

(Singapore)

(https://www.eurekalert.org/news-releases/963144)

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