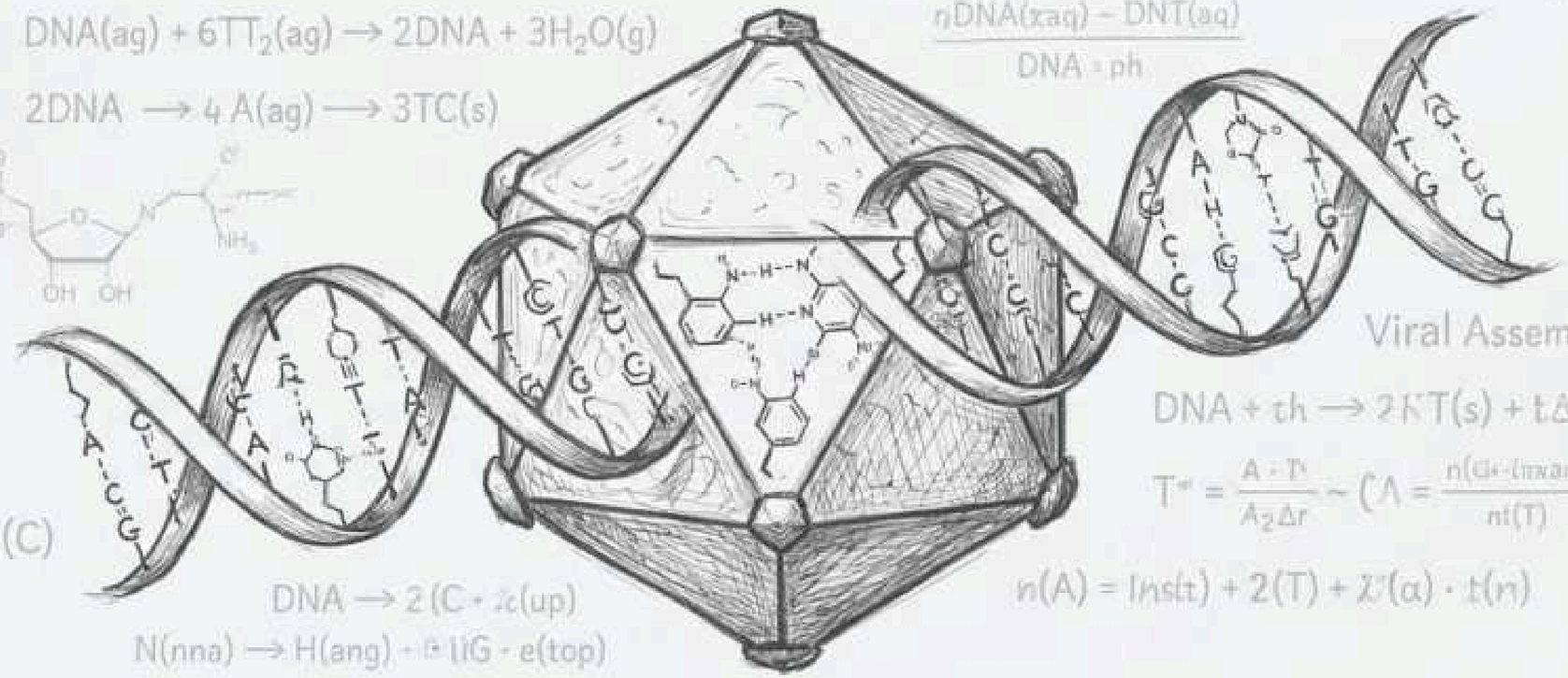
Genetic Testing, Gene Therapy, and Ethics

MED 503 B Lecture 6 · Anderson Academy



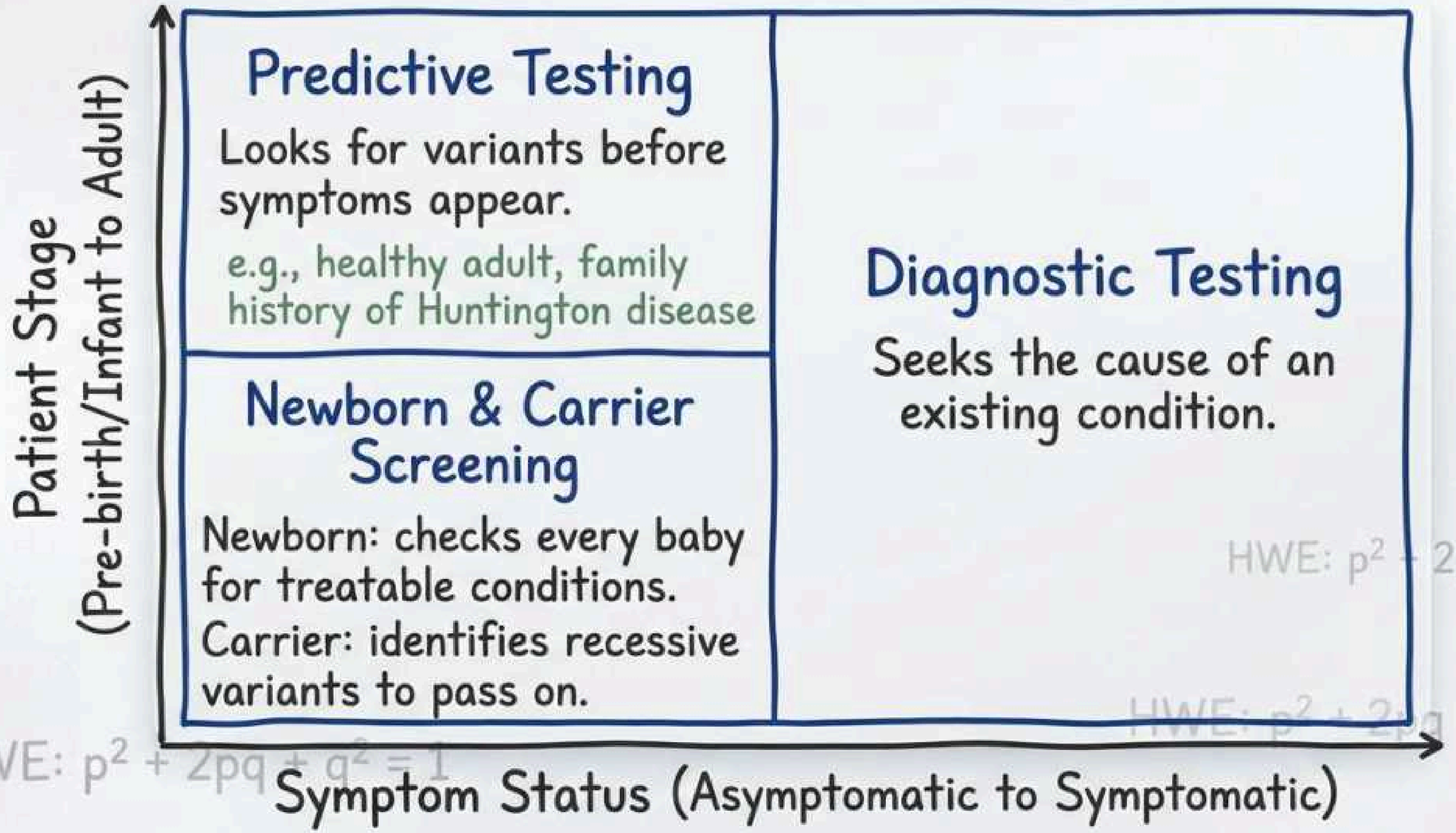
At a glance: Kinds of tests, sequencing, pharmacogenomics, gene editing, counselling, and ethical frameworks.

DNA Methylation: CH_3 + CpG

Mendelian Inheritance: Aa × Aa → 1AA : 2Aa : 1aa

HWE: $p^2 + 2pq + q^2 = 1$

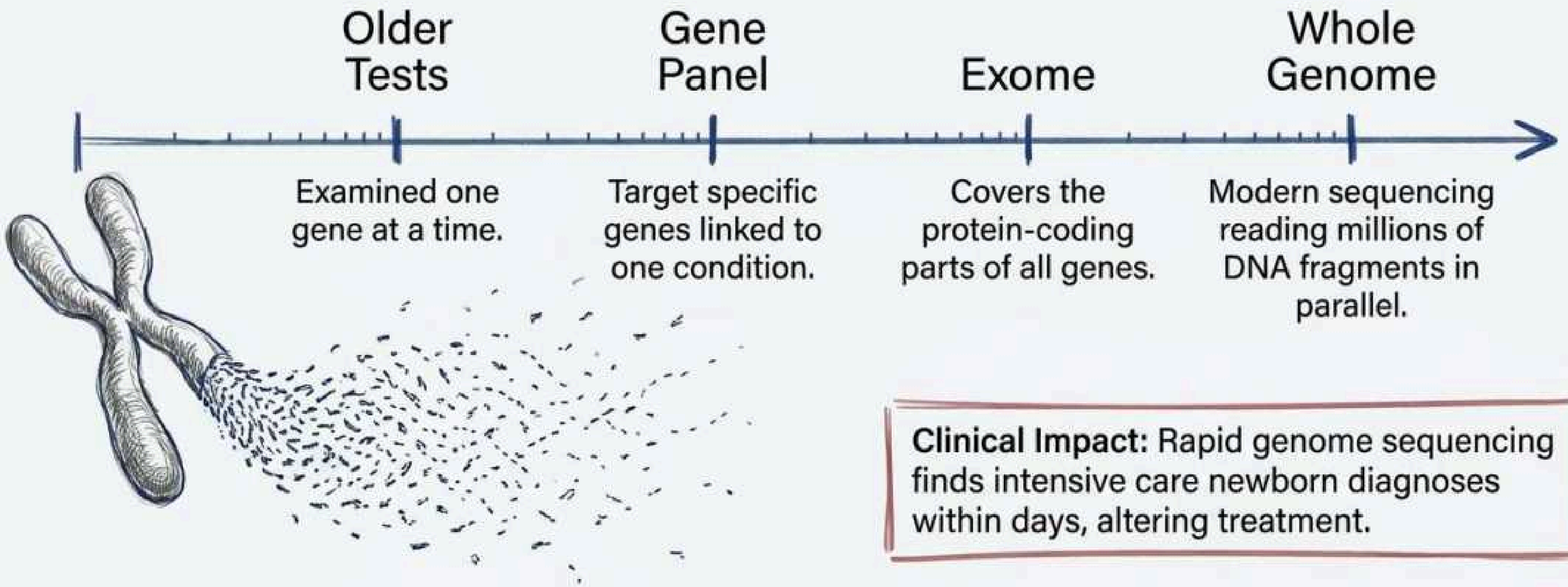
¶ 6.1



HWE: $p^2 + 2pq + q^2 = 1$

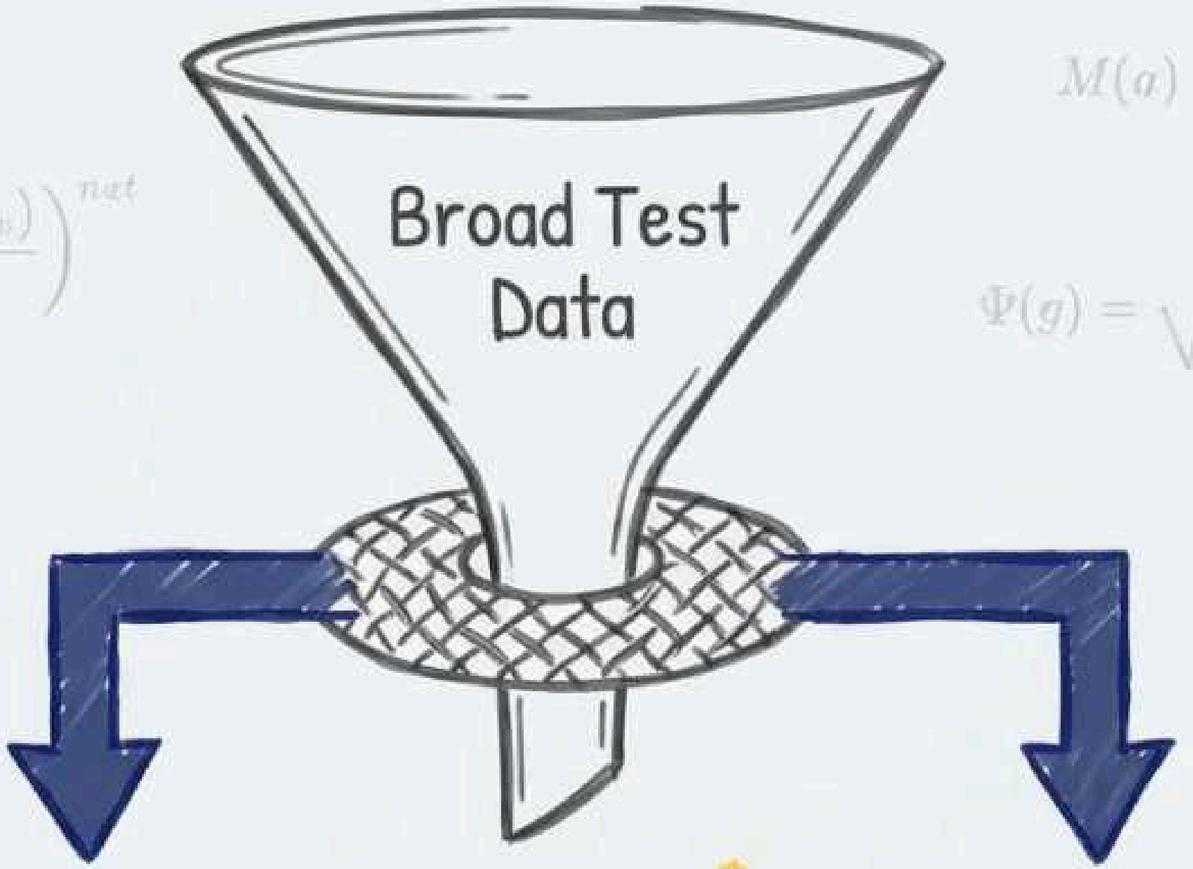
HWE: $p^2 + 2pq + q^2 = 1$

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Clinical Impact: Rapid genome sequencing finds intensive care newborn diagnoses within days, altering treatment.

¶ 6.3



Variants of Uncertain Significance (VUS)

Must not be treated as diagnoses.

Incidental / Secondary Findings

Harmful variants unrelated to the test reason (e.g., cancer risk found during heart evaluation).

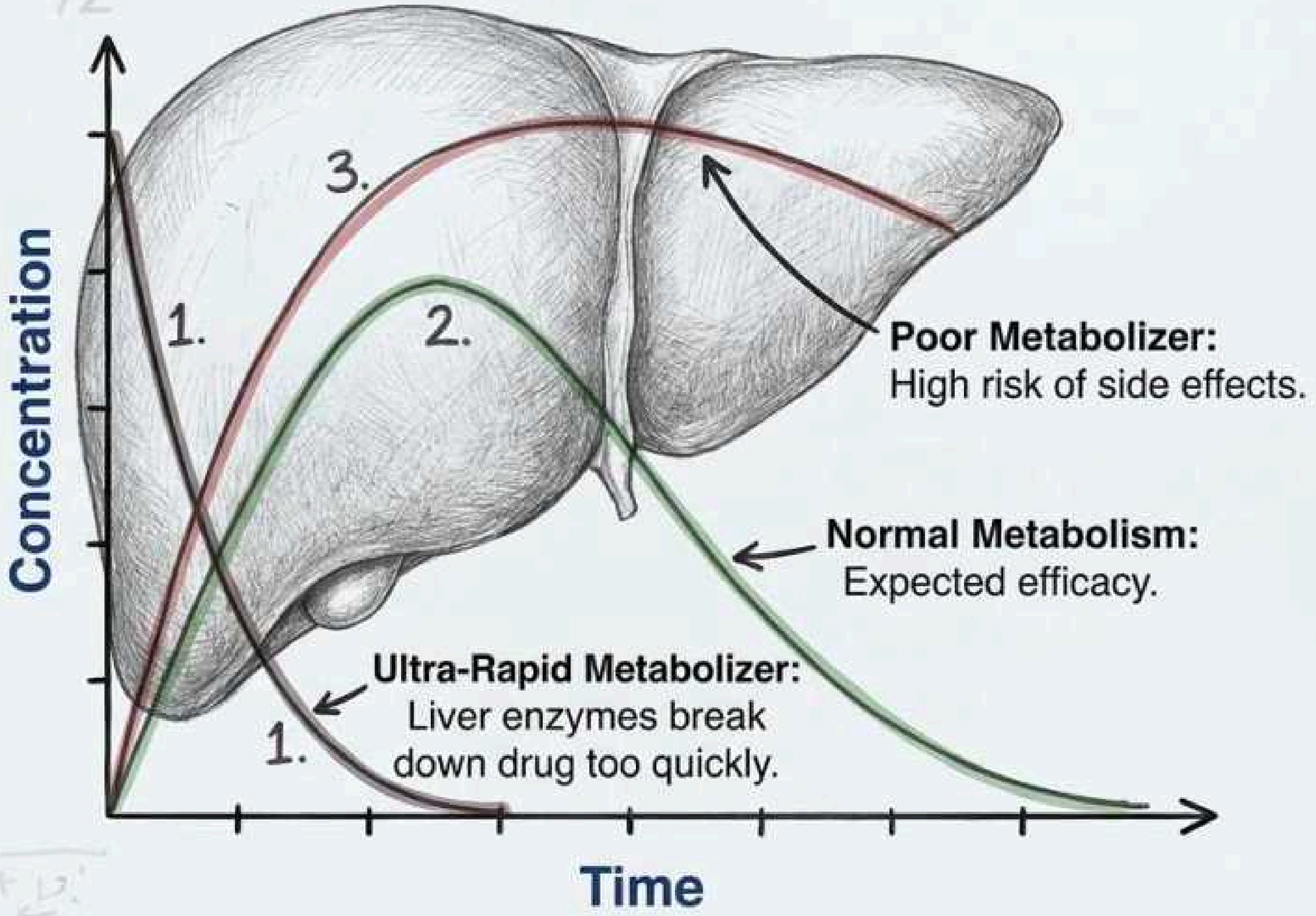
[Clinical Best Practice: Agree beforehand what findings a person wants to learn.]

$$t_{1/2} = \frac{\ln(2)}{k}$$

¶6.4

Pharmacogenomics: Genes shape drug response.

Guidelines advise pre-treatment testing to avoid severe immune-system reactions.

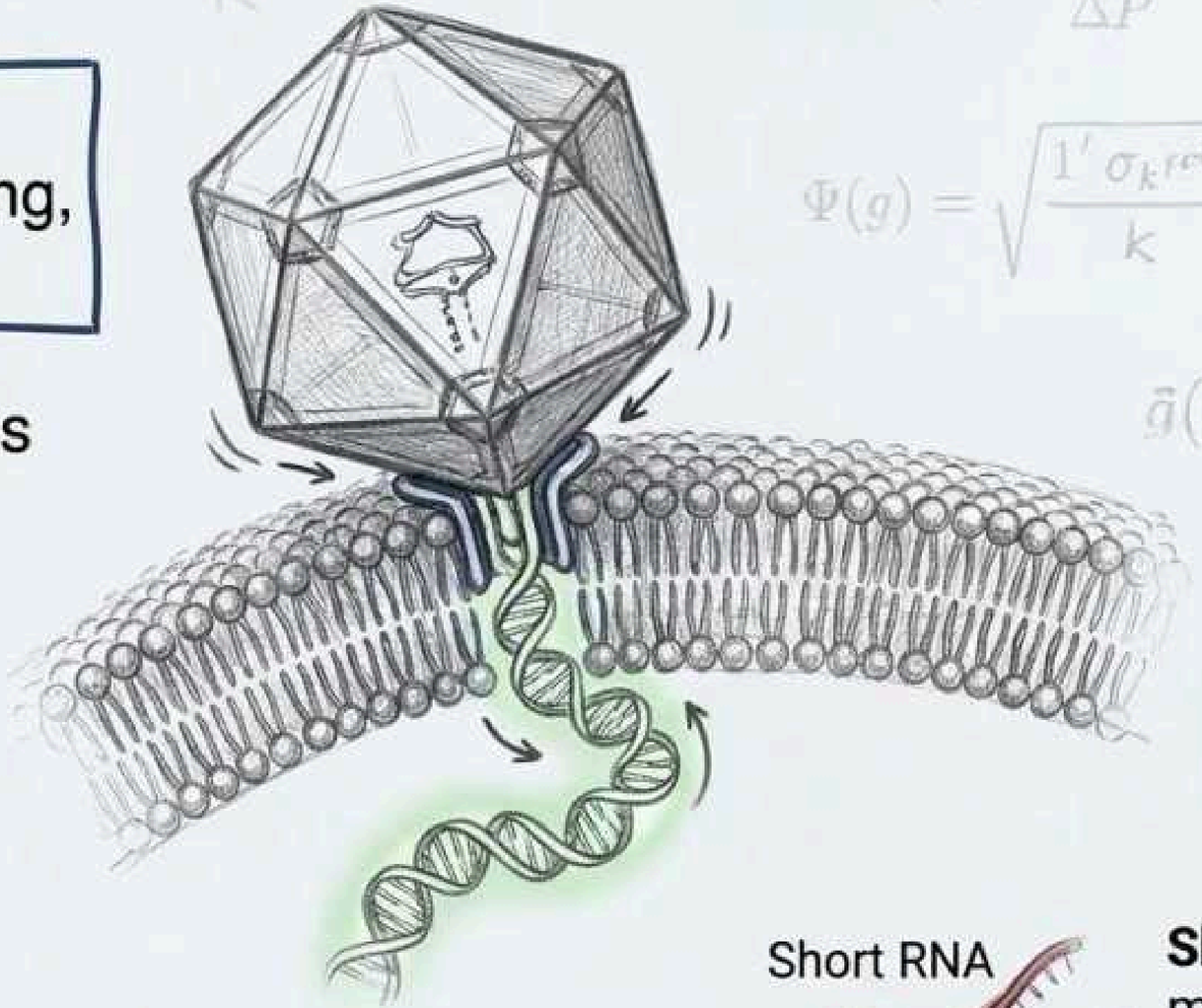


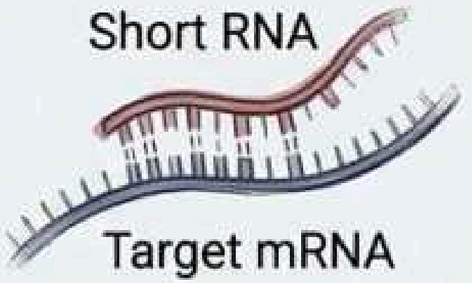
¶6.5

Gene Therapy: Treat disease by adding, replacing, or altering genes.

The Mechanism: Harmless viruses deliver a working gene copy into cells.

Clinical Success: Approved for inherited blindness and spinal muscular atrophy (severe nerve disease in children).



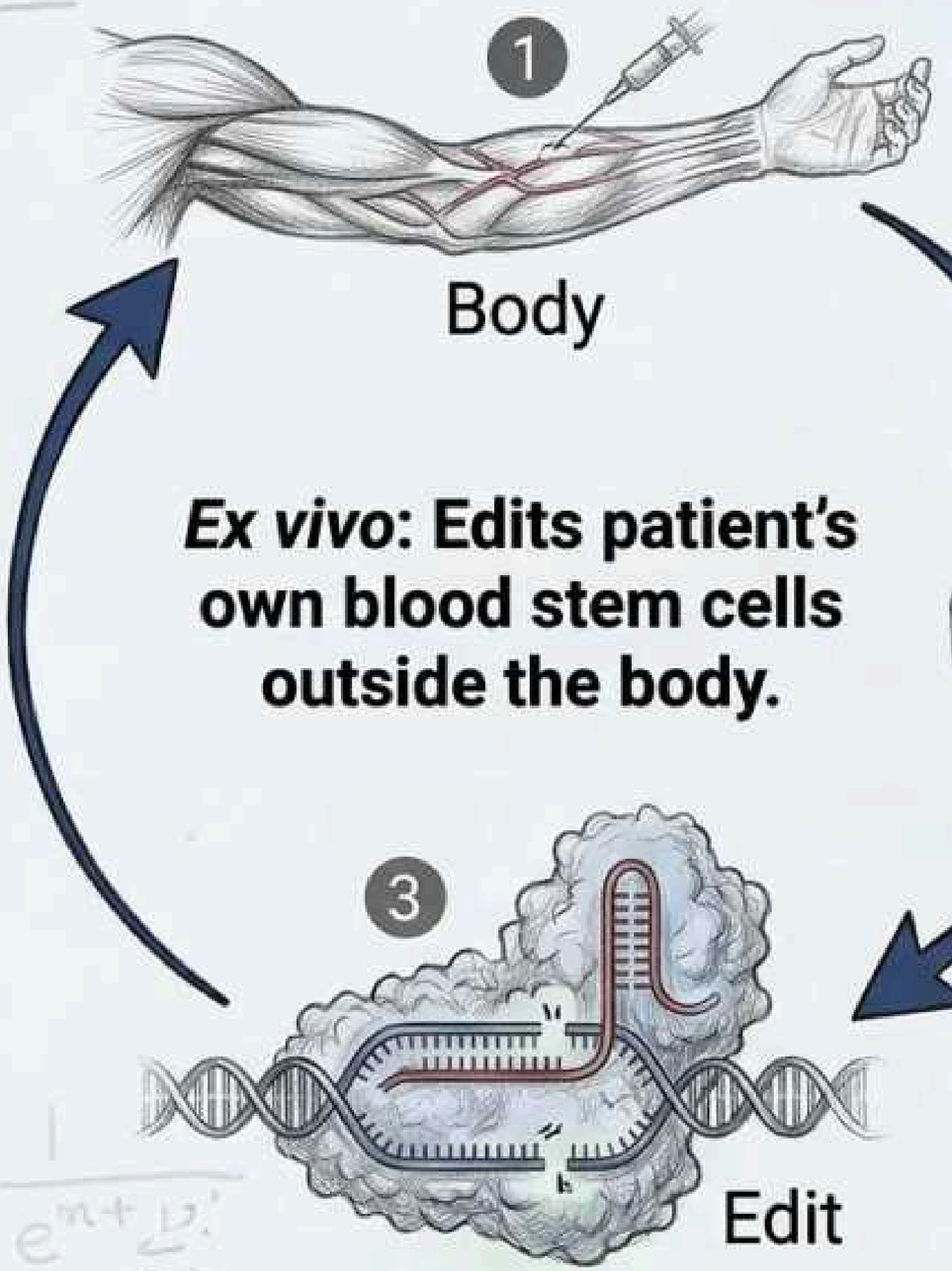


Short RNA-like molecules can change reading or silence harmful genes.

¶6.6

CRISPR: A guide molecule brings a cutting enzyme to a precise genome spot, changing DNA directly.

Milestone: December 2023: First CRISPR treatment approved in the US for sickle cell disease.



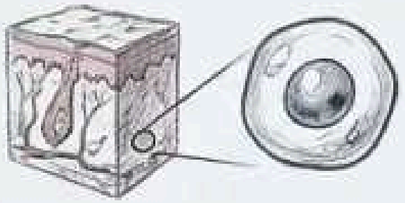
***Ex vivo*: Edits patient's own blood stem cells outside the body.**

Caveat: Complex, expensive, long-term effects still under study.

¶6.7

Somatic Cell Editing

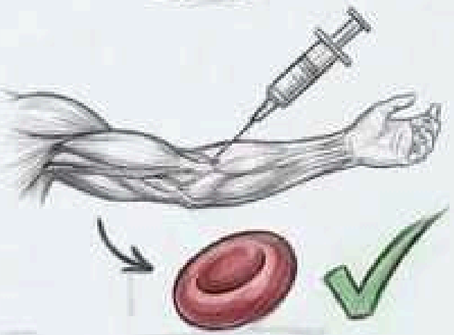
- **Target:** Patient's body cells.



- **Scope:** Affects only that person.



- **Status:** Approved for specific clinical use (e.g., sickle cell).



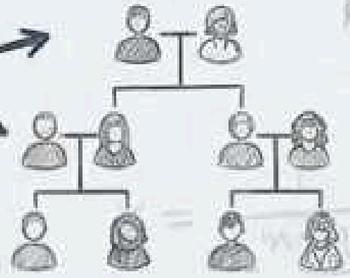
Crucial Boundary

Germline Editing

- **Target:** Eggs, sperm, or embryos.

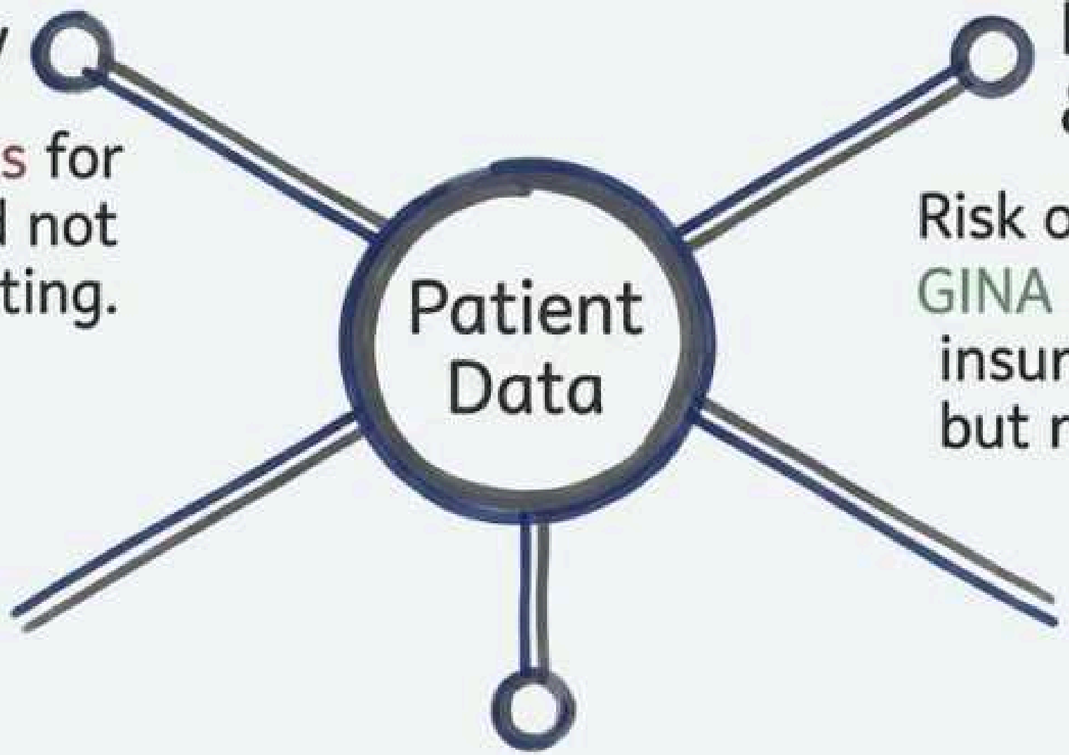


- **Scope:** Inherited by future generations.



- **Status:** 2018 edited babies widely condemned; scientist imprisoned in China. Not permitted clinically. Broad agreement against use.





Family

Results reveal risks for relatives who did not ask for testing.

Employment & Insurance

Risk of genetic discrimination. GINA 2008 (US) protects health insurance and employment, but not all insurance types.

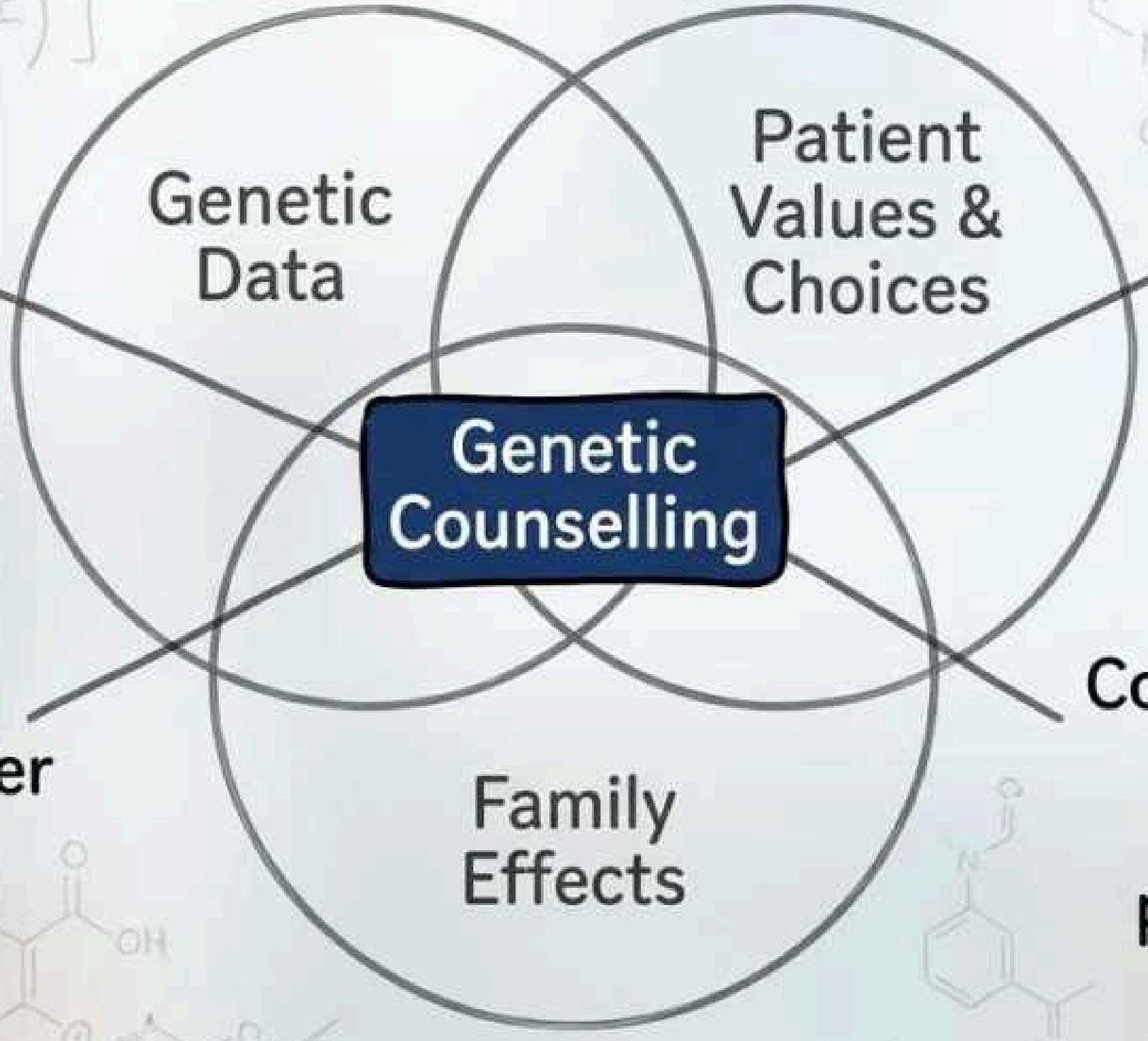
Direct-to-Consumer

Commercial DNA test kits.

Raises questions of accuracy, privacy, and interpretation without professional support.

¶ 6.9

Help people understand complex information.



Support decisions on whether to be tested.

Interpret predictive/carrier results.

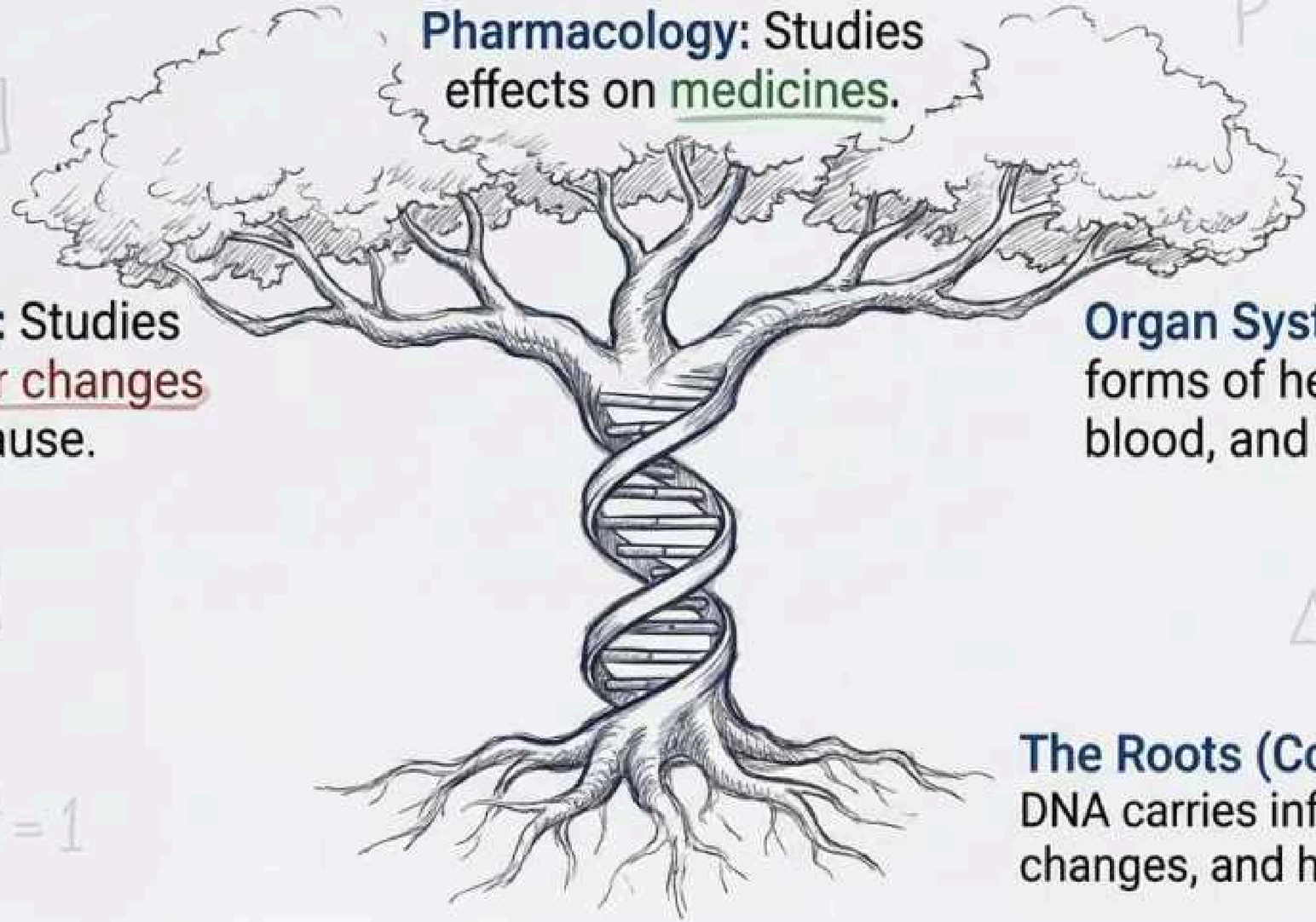
Cope with worrying findings while respecting personal values.

¶ 6.10

Genetics will touch almost every Medical School course that follows.

Pharmacology: Studies effects on medicines.

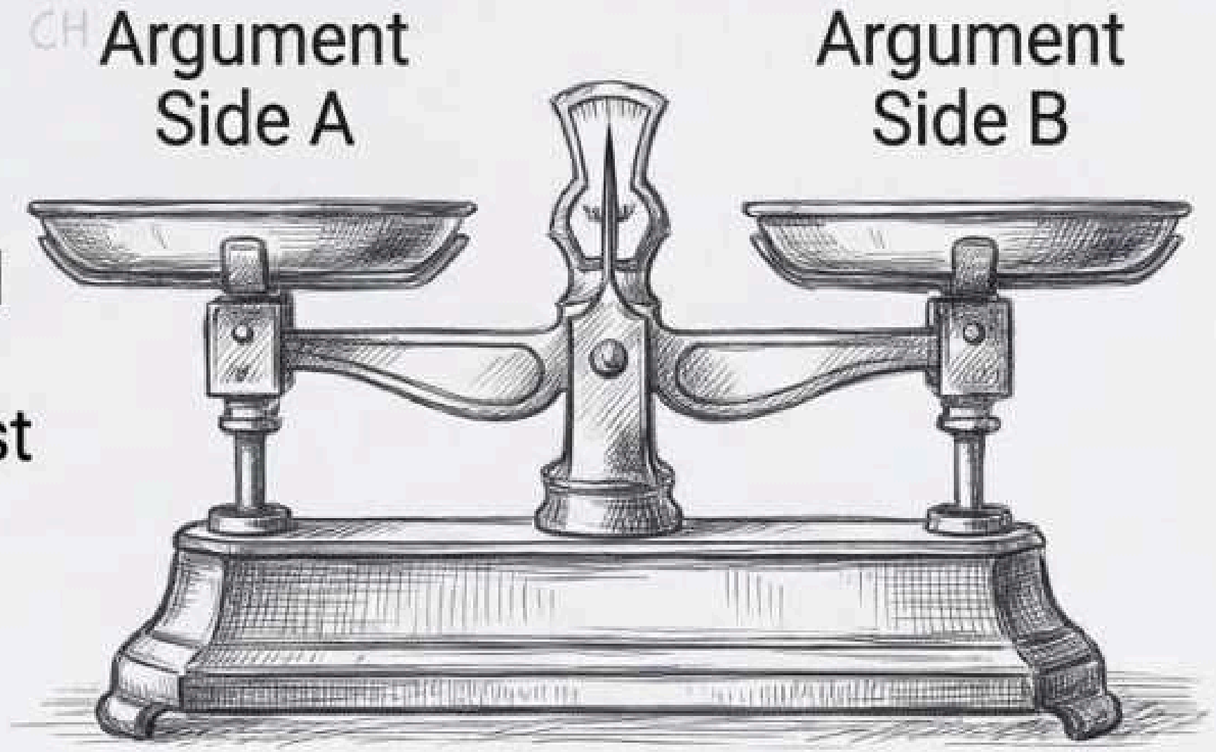
Pathology: Studies the cellular changes variants cause.



Organ Systems: Inherited forms of heart, kidney, blood, and nerve disease.

The Roots (Core Ideas): DNA carries information, how it changes, and how it is inherited.

The Challenge:
Choose one ethical from this lecture. Set out the strongest arguments on two sides.



The Reflection:
Where do you come down, and why?

Check your understanding of the facts against the Course Book.

Thank you for taking Medicine 503 B.

The next course is Medicine 504 B,
Cellular and Molecular Immunology.



References & Further Reading

- MIT Catalog, HST.160 Genetics in Modern Medicine
- MedlinePlus Genetics, Help Me Understand Genetics
- National Human Genome Research Institute
- National Society of Genetic Counselors
- Grokipedia: Human genome, Mendelian inheritance, Knudson hypothesis
- Thompson and Thompson, Genetics and Genomics in Medicine
- Anderson Academy: BIO 210 B Genetics
- Anderson Academy: MED 500 B How to Use the Medical School Courses