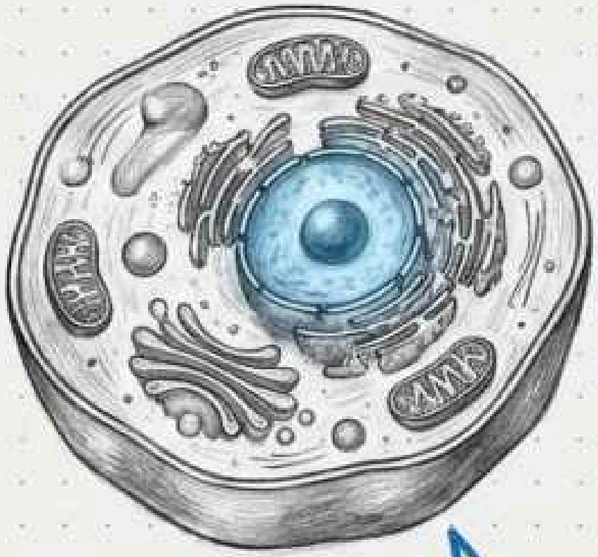
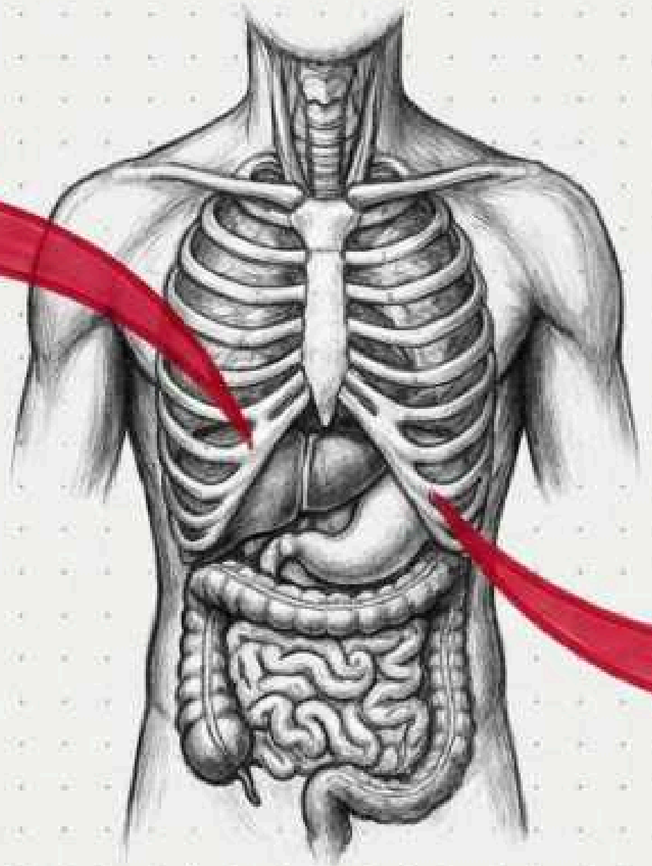
The Universal Instruction Manual

¶ 1.1



Contains complete instructions for building a human being.



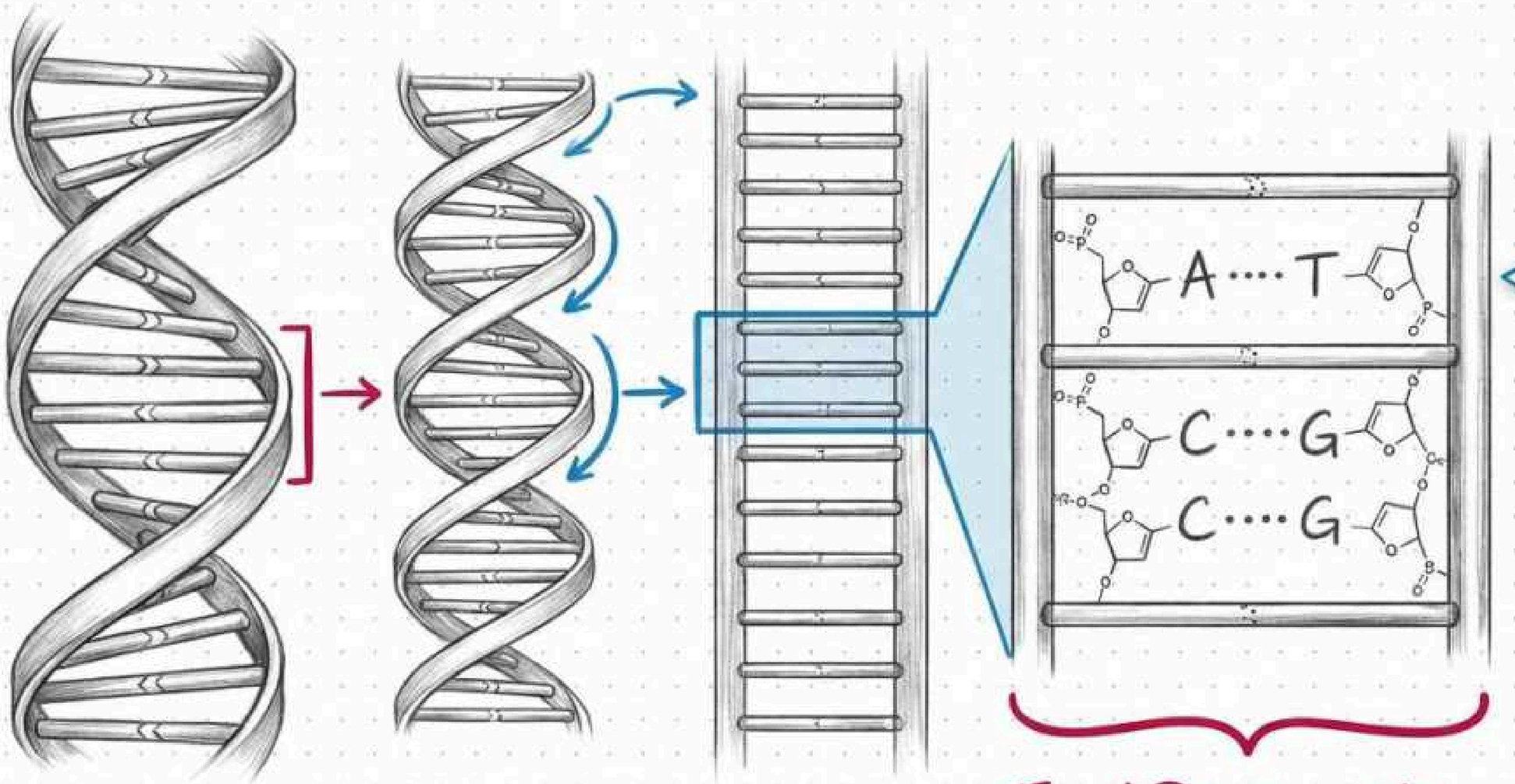
Exception: Mature red blood cells lack a nucleus.



These instructions are written in DNA (Deoxyribonucleic Acid) – the foundation of medical genetics.

Nucleotides: The Four Building Blocks

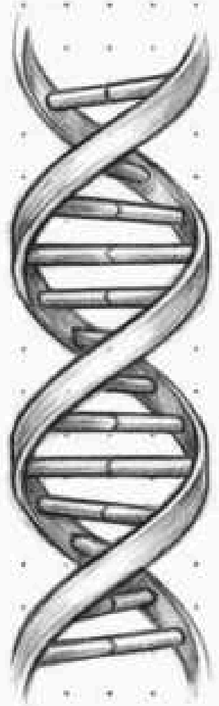
¶ 1.2



Because A always pairs with T, and C with G, each each strand acts as a perfect template when a cell divides.

Fixed Pairing = Mirror Templates

¶ 1.3



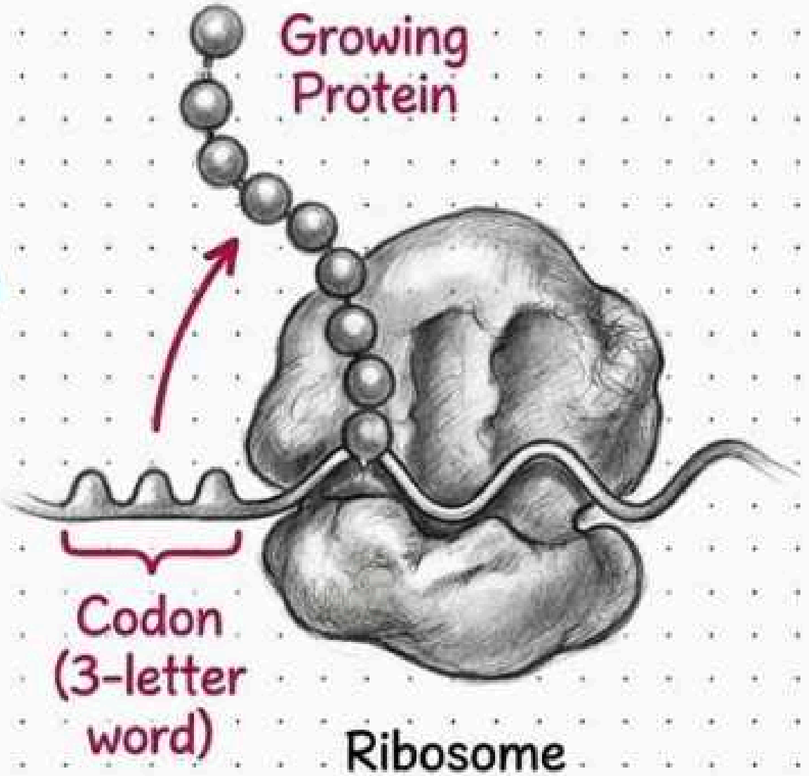
Gene
(instructions
for a product)

Transcription

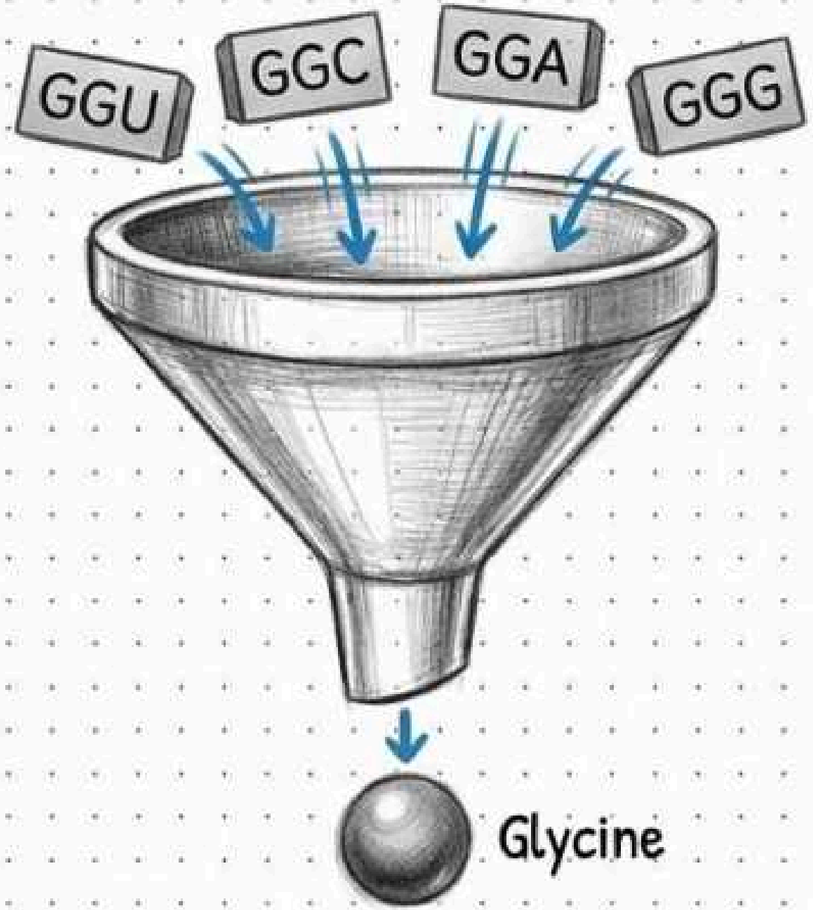


RNA: The Messenger

Translation



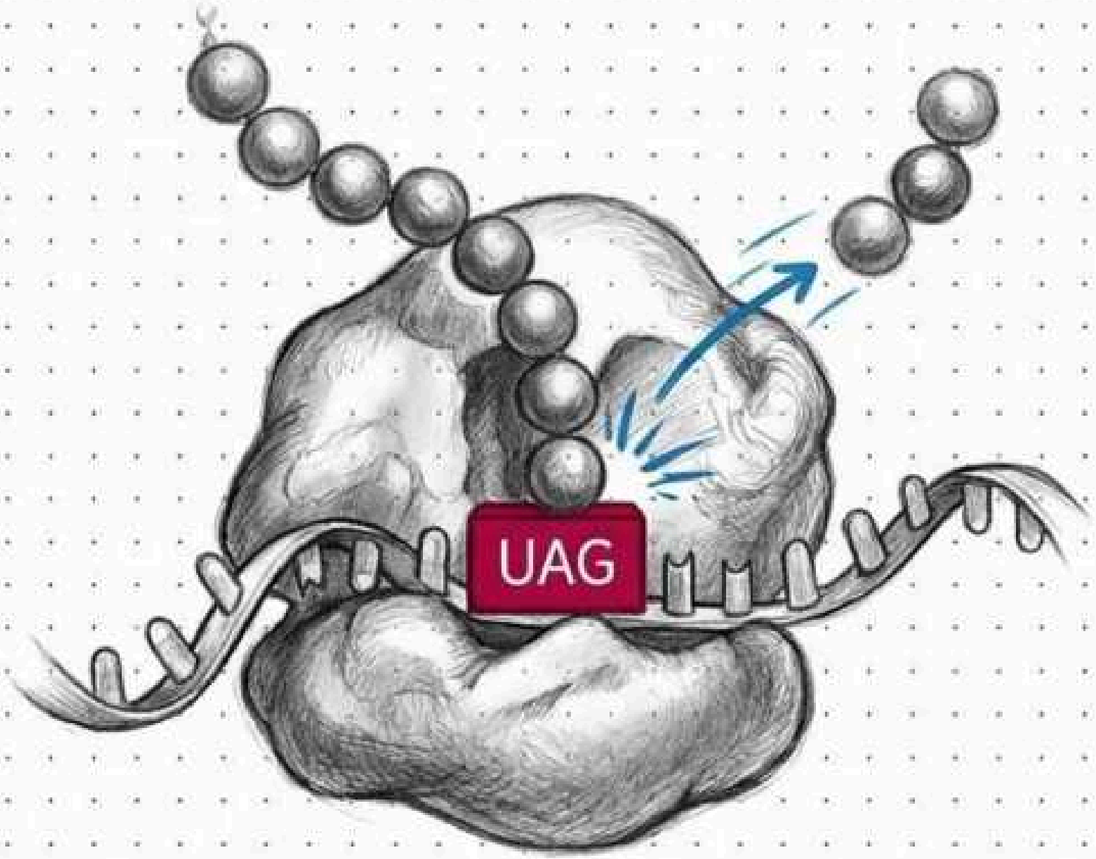
Redundancy / Safety



Built-in Safety: Multiple codons specify the same amino acid. Some DNA changes make zero difference.

Termination / Danger

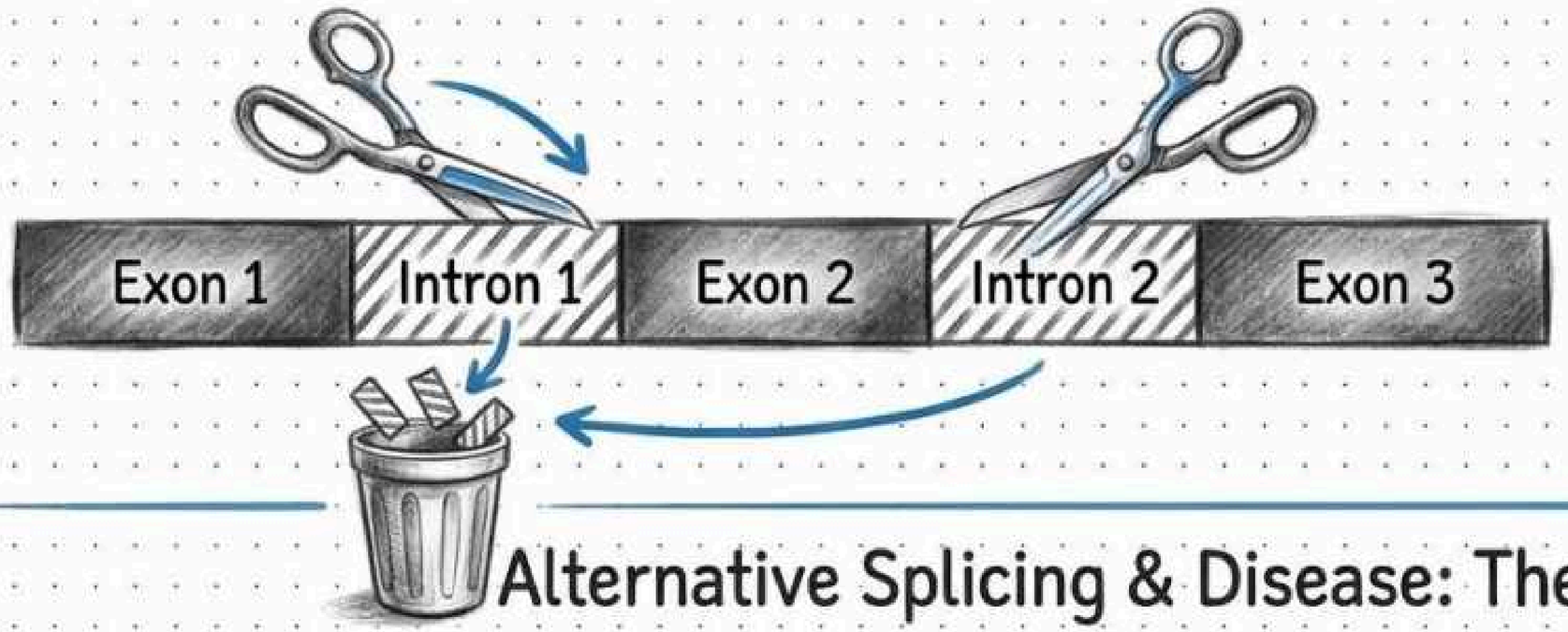
¶ 1.4



Premature Stop Signal = Shortened, useless protein (Key mechanism in genetic disease).

Gene Splicing: The Assembly Line

¶ 1.5



Exons = Coding segments

Introns = Non-coding segments

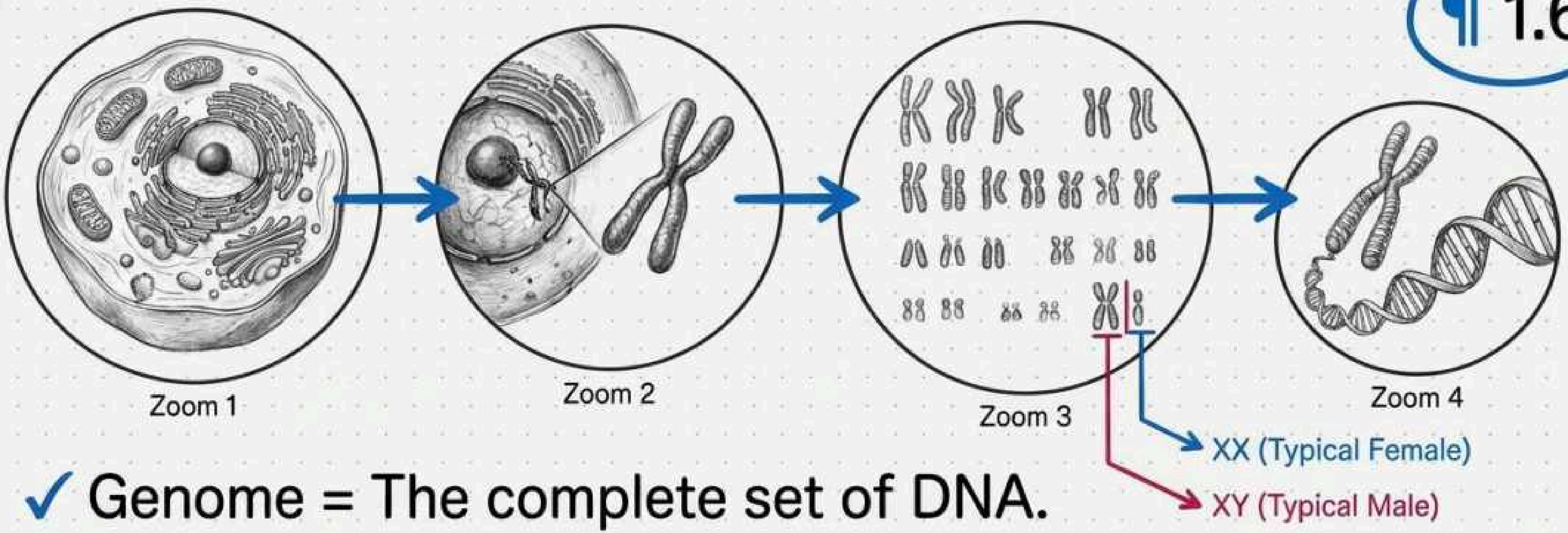
Alternative Splicing & Disease: The Math

[Exon 1] + [Exon 2] + [Exon 3] = Protein A

[Exon 1] + [Exon 3] = Protein B (Related Variant)

Errors in this splicing process are a major cause of genetic disease.

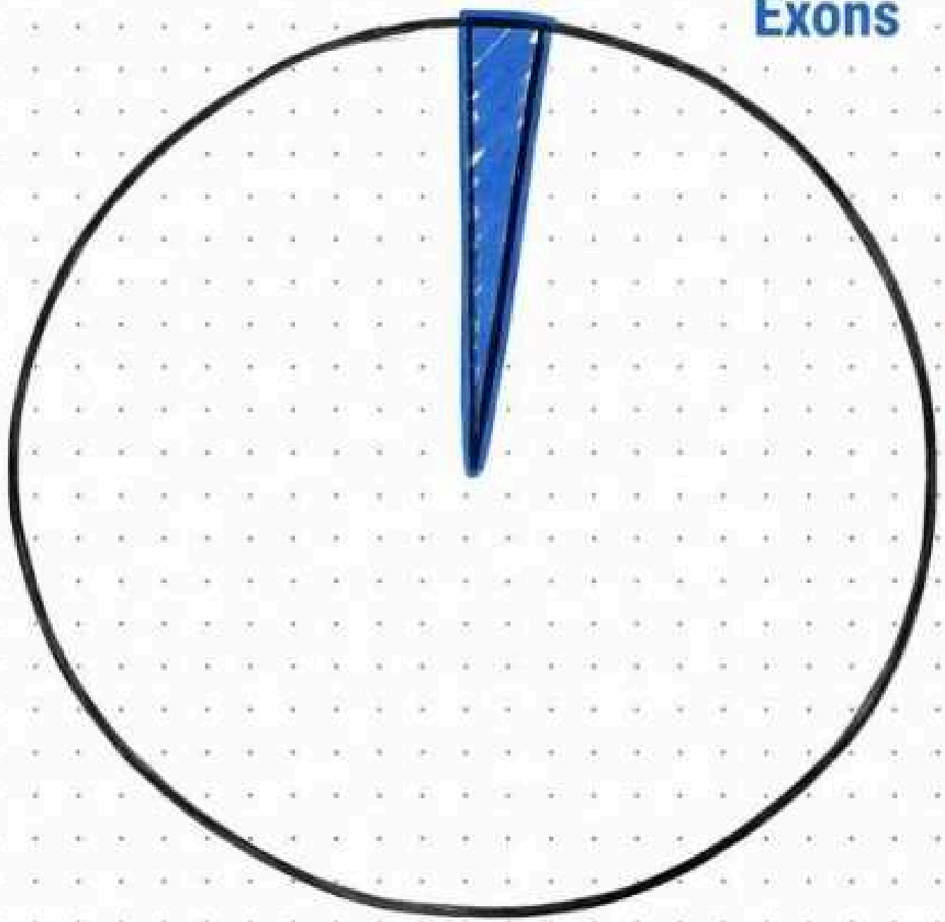
¶ 1.6



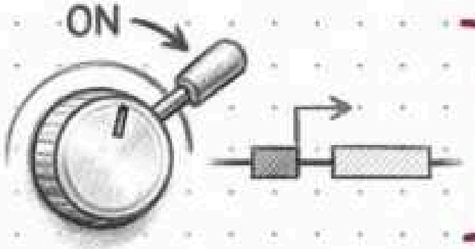
- ✓ Genome = The complete set of DNA.
- ✓ 3 Billion Base Pairs.
- ✓ 23 Pairs of Chromosomes (One set from each parent).
- ✓ 22 Autosomes + 1 pair Sex Chromosomes.
- ✓ ~20,000 Protein-coding Genes.

JUNK DNA

¶ 1.7

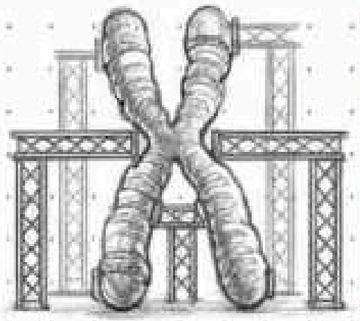


1



Control switches

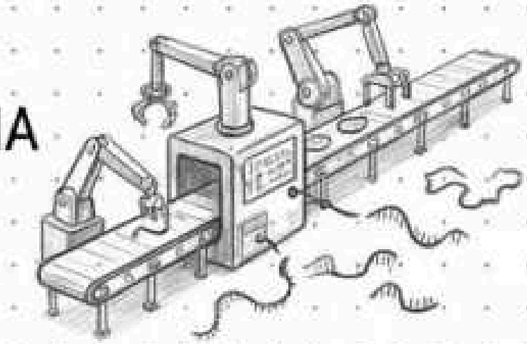
2



Chromosome architecture

3

Non-coding RNA manufacturing

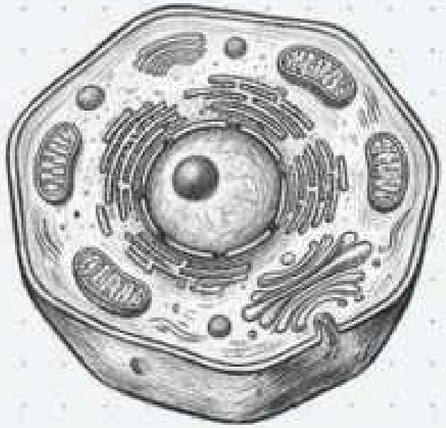
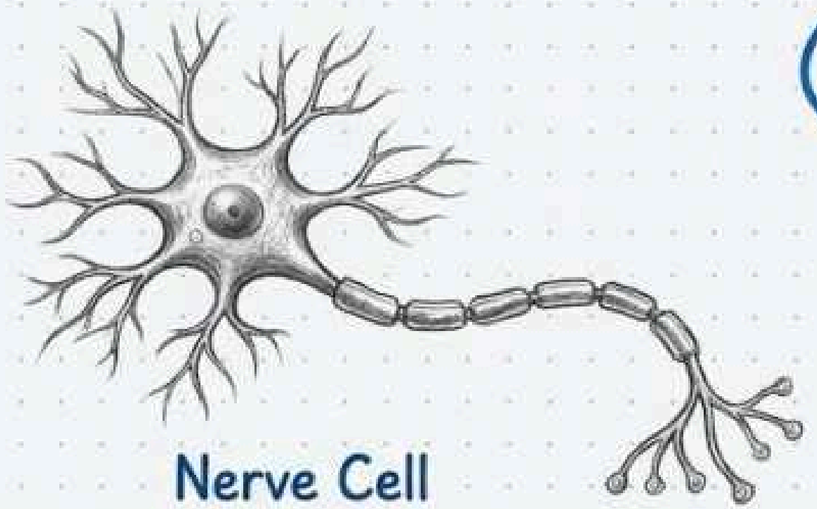


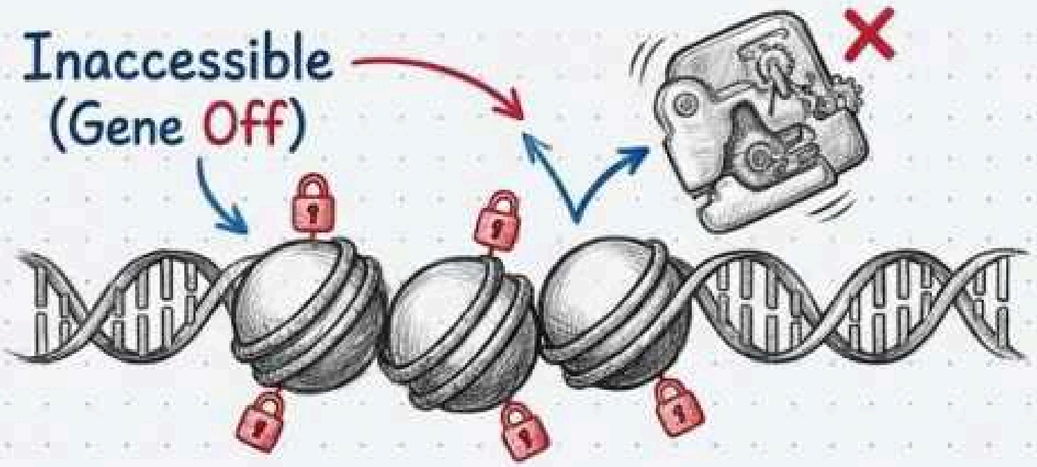
4

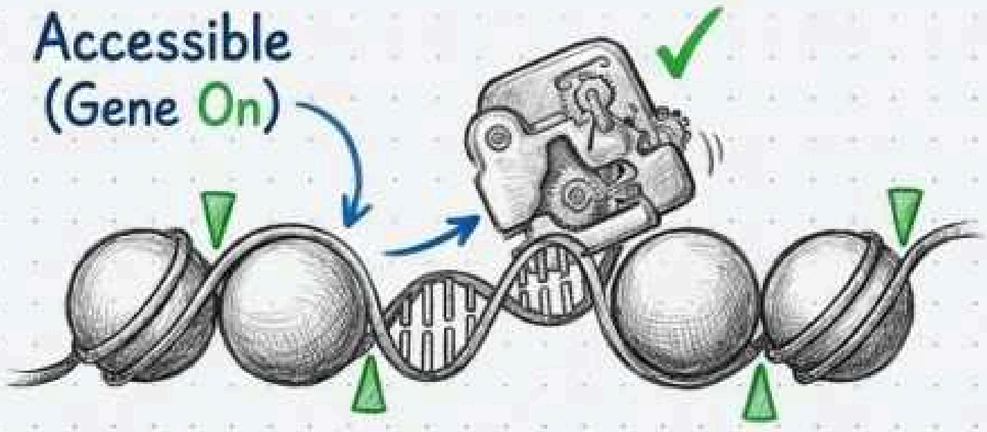


Unknown functions
(Researchers continuously finding new roles).

¶ 1.8

	 Liver Cell	 Nerve Cell
[Genome]	100% Identical	100% Identical
[Active Transcription Factors]	Set A (Hepatic)	Set B (Neural)



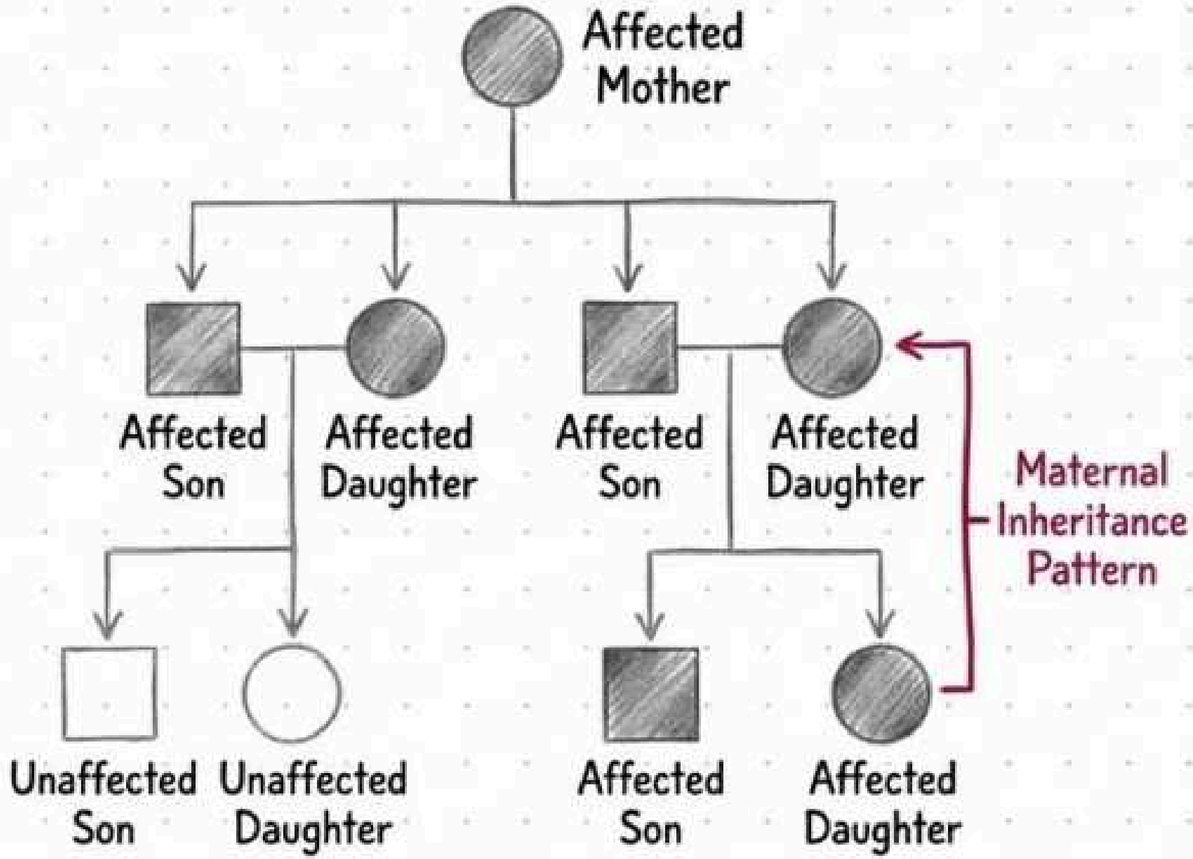


Epigenetics: Chemical marks on DNA and packaging proteins influenced by development and environment.

¶ 1.9

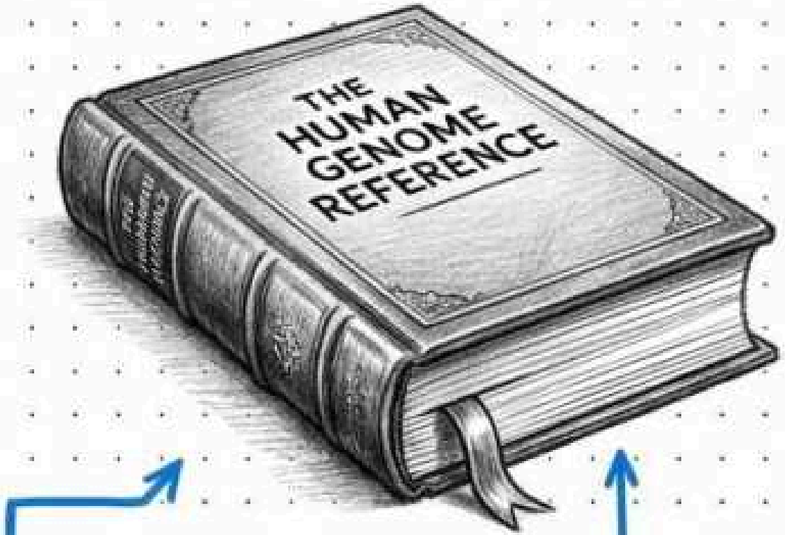
Nuclear DNA vs. Mitochondrial DNA

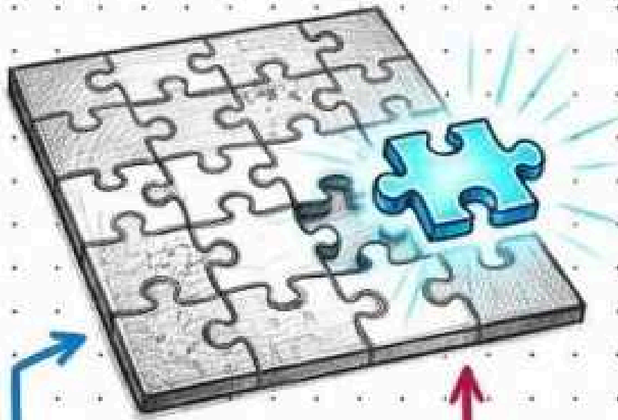
	Nucleus	Mitochondria
Location:	Nucleus	Mitochondria 
Shape:	Linear Chromosomes	Small Circular DNA 
Gene Count:	~20,000	37 (Mostly energy production)
Inheritance:	Biparental	Strict Maternal



Mitochondrial DNA (mtDNA) has a distinctive pattern of inheritance (Detailed in Lecture 3).

¶ 1.10





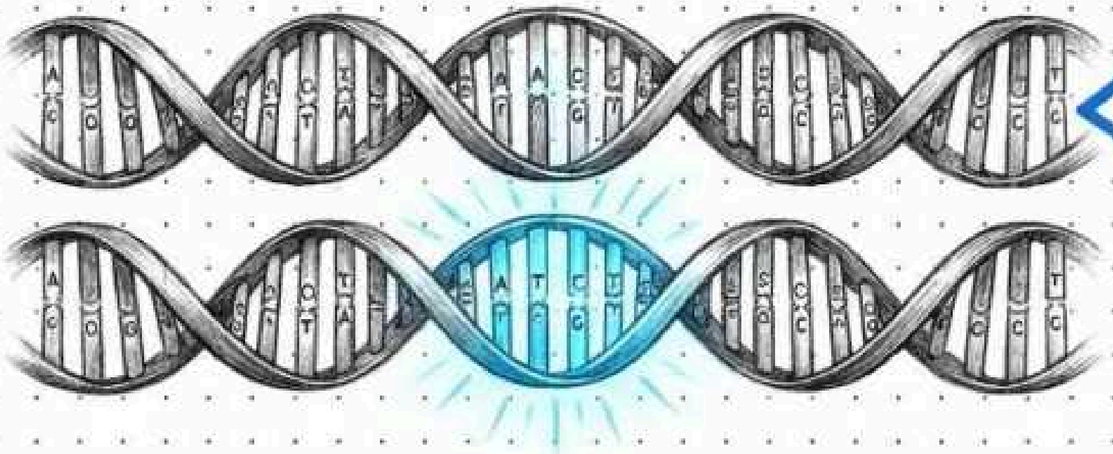
2003

2022

Human Genome Project Complete:
The First Reference Sequence.

Doctors and researchers still use
the reference sequence today.

A Fully Complete Sequence:
Filling the last stubborn gaps.

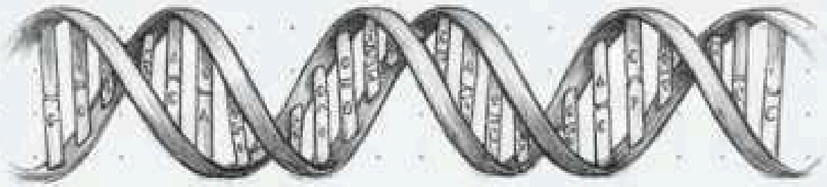


Each person's genome differs from the reference at millions of positions.
(Most are completely harmless).

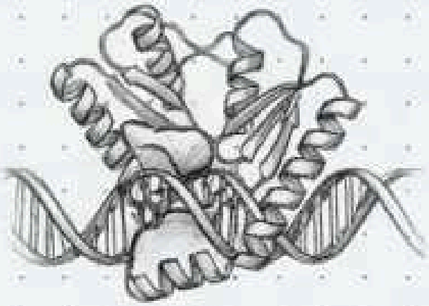
¶ 1.11

The Core Question: If every cell carries the same genome, how can the body contain so many different kinds of cells?

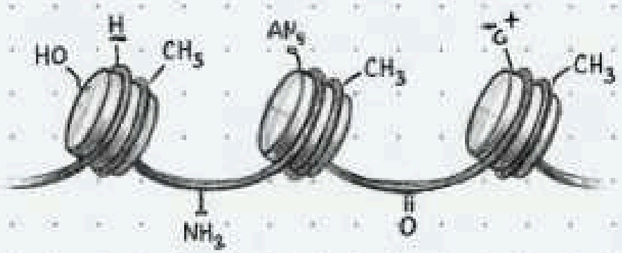
[**Universal Genome**]



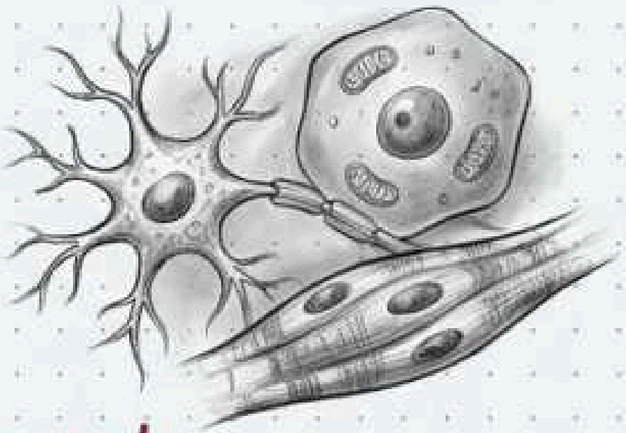
\+ [**Specific Transcription Factors**]



\+ [**Epigenetic Marks**]



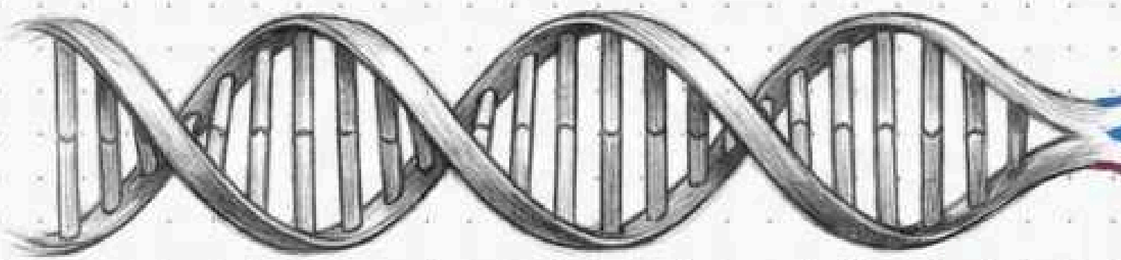
= [**UNIQUE CELLULAR IDENTITY**]



Answer: It's not just what genes you have; it's how they are expressed.

Looking Ahead: Lecture 2

¶ 1.12



How DNA Changes
(And why most changes are completely harmless).



The Rare Exceptions
(Why a few specific changes cause disease).



End of Lecture 1.