How Genes and Genomes Evolve

Chapter 9

BIOL 366

Dr. Matthew Ellis

Learning Objectives for Chapter 9

By the end of this module, you should be able to:

- Describe the various mechanisms that can lead to genetic change within an organism and how these are passed onto its offspring.
- Understand the mechanism of transposon/mobile genetic element movement throughout the genome and how this can affect gene identity and expression.
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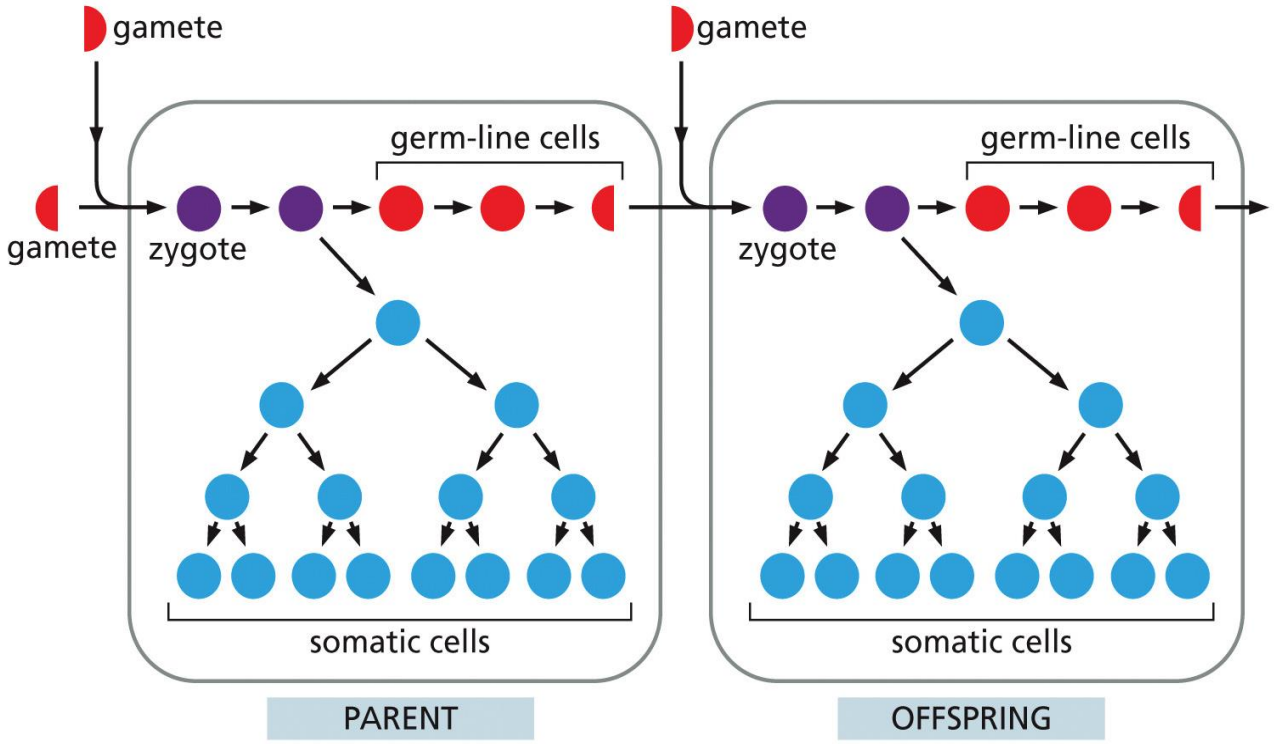
Generating Genetic Variation: Passing on genes to offspring

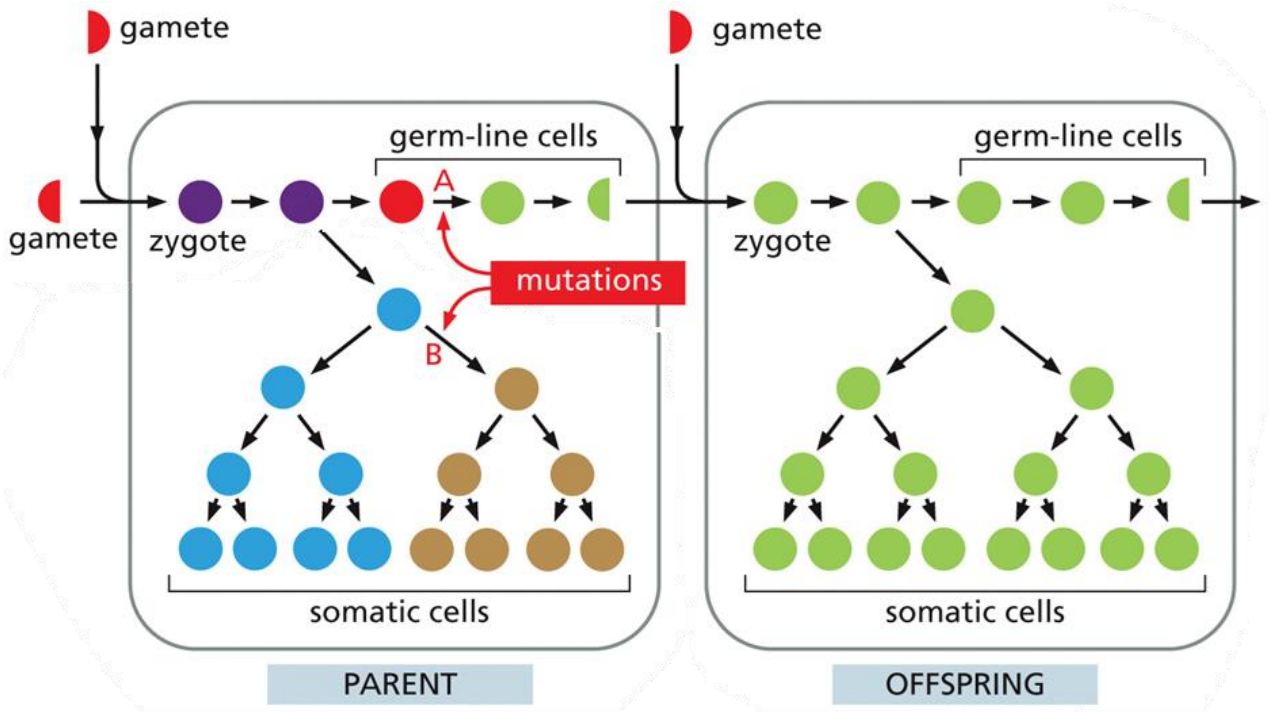
- In sexually reproducing organisms the genetic information is passed on to the next generation only via **germ-line cells/gametes** (e.g., sperm and eggs)

- Two gametes combine to form the fertilized zygote, which gives rise to both germ-line cells and somatic cells
  - Gametes contain only half the number of chromosomes (haploid)
  - **Somatic cells** form the body of the organism but do not contribute their DNA to the next generation



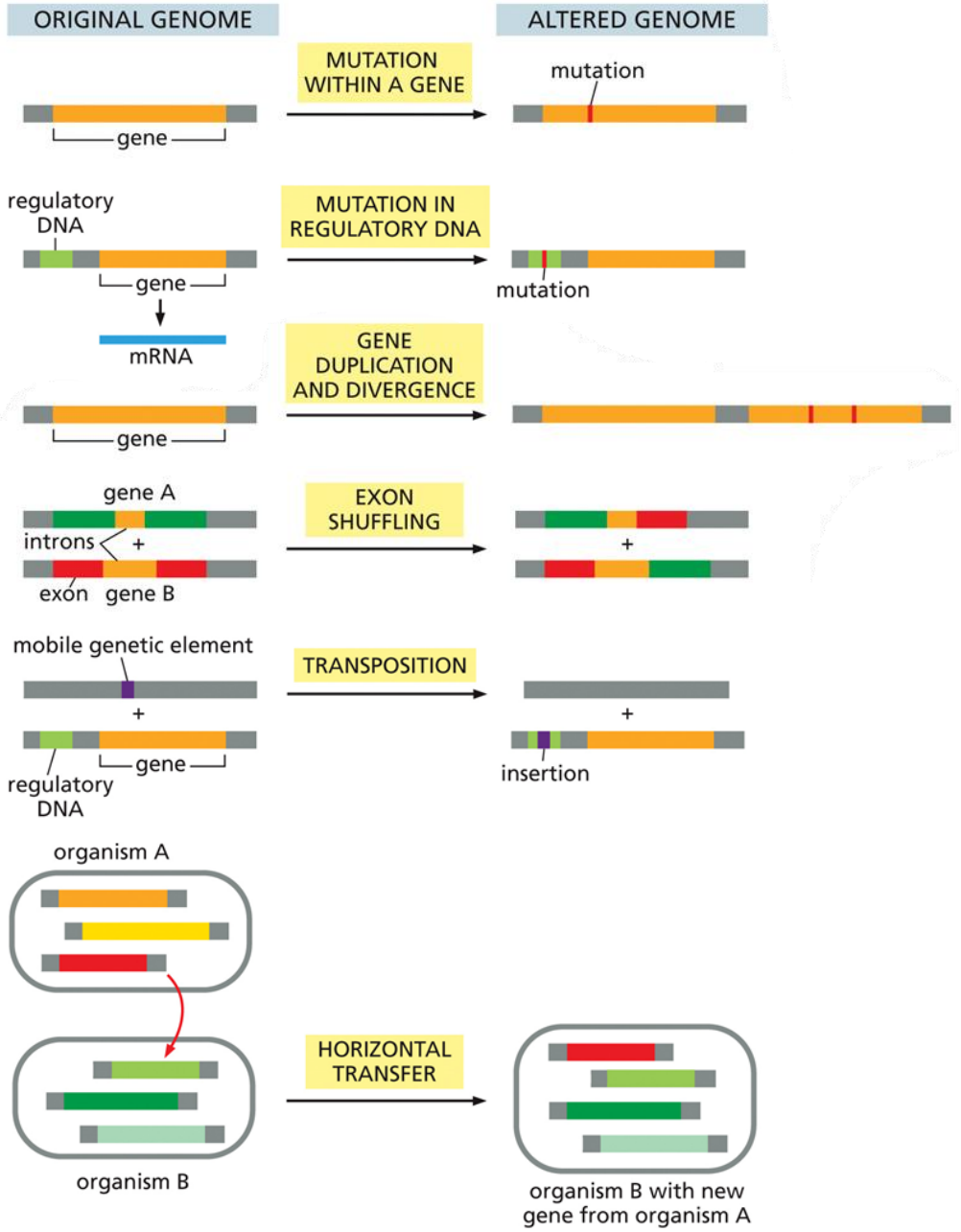
Generating Genetic Variation: The effect of mutations

- Mutations in germ-line cells and somatic cells cause different effects
- A mutation in a *germ-line cell* can be passed on to the next generation (red to green)
- A mutation that arises in a *somatic cell* affects only the progeny of that cell (blue to brown)
  - Will not be passed on to the organism's offspring
  - Mutations in somatic cells are largely responsible for most human cancers

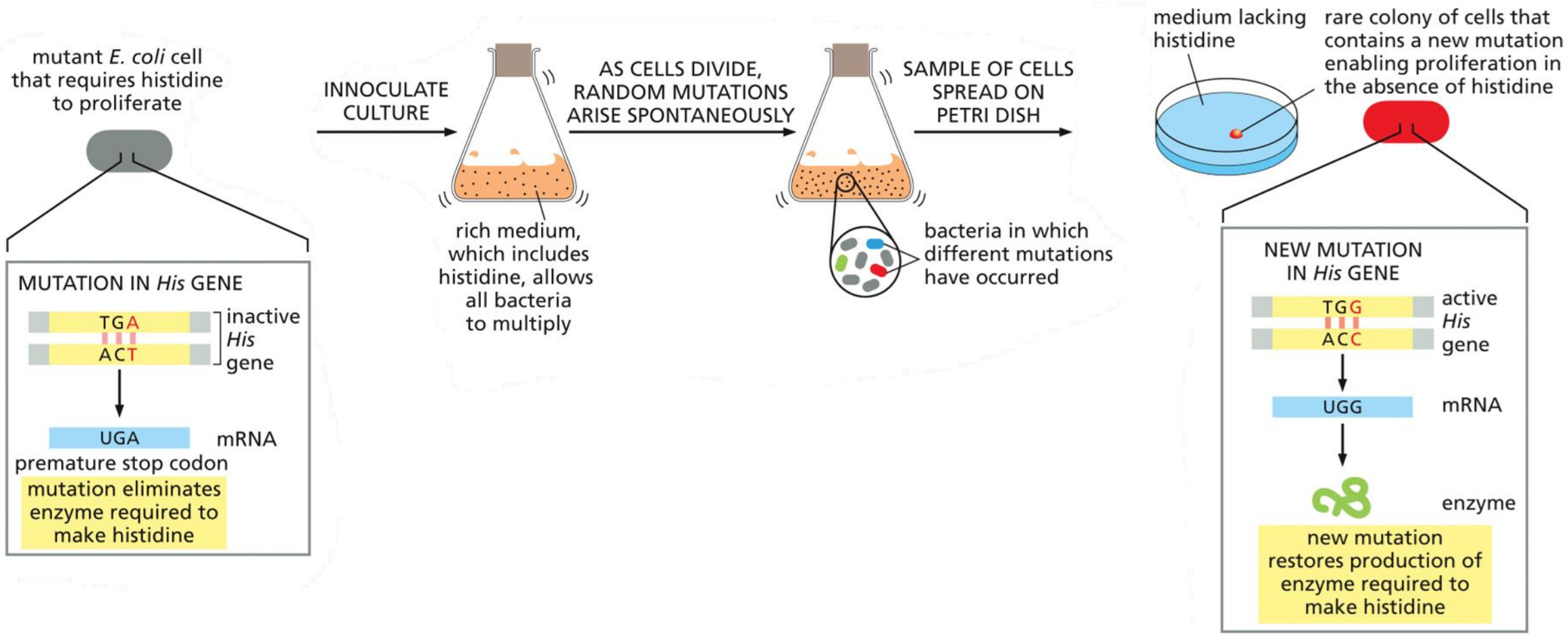


Generating Genetic Variation

Mutations and rearrangements within genes and regulatory sequences can alter both the expression pattern and the gene products produced



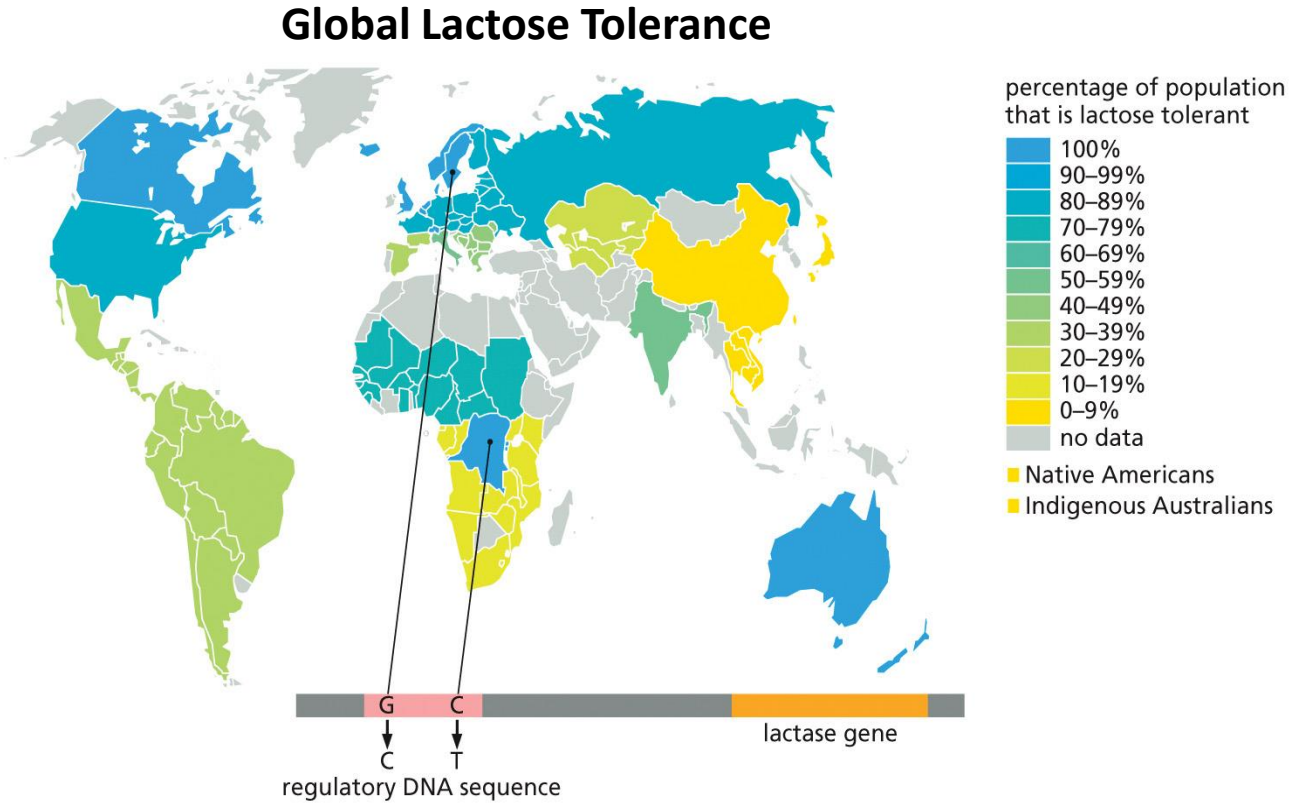
Mutations within a gene can affect protein products



Mutations in regulatory DNA sequences can change the timing and level of gene expression

Example:

- Mutation in the regulatory sequence of the *lactase* gene
  - Produces enzyme to digest lactose found in human milk, whose expression was originally confined to infancy
  - 10,000 years ago farmers in Europe and Africa began to raise cattle
  - Point mutations that allowed continued expression of lactase during adulthood provided *selective advantage* during periods of starvation



Mutations that confer a selective advantage can be propagated in a population and selected for (i.e., *survival of the fittest*)

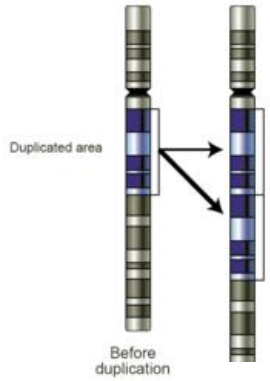
DNA duplications give rise to families of related genes

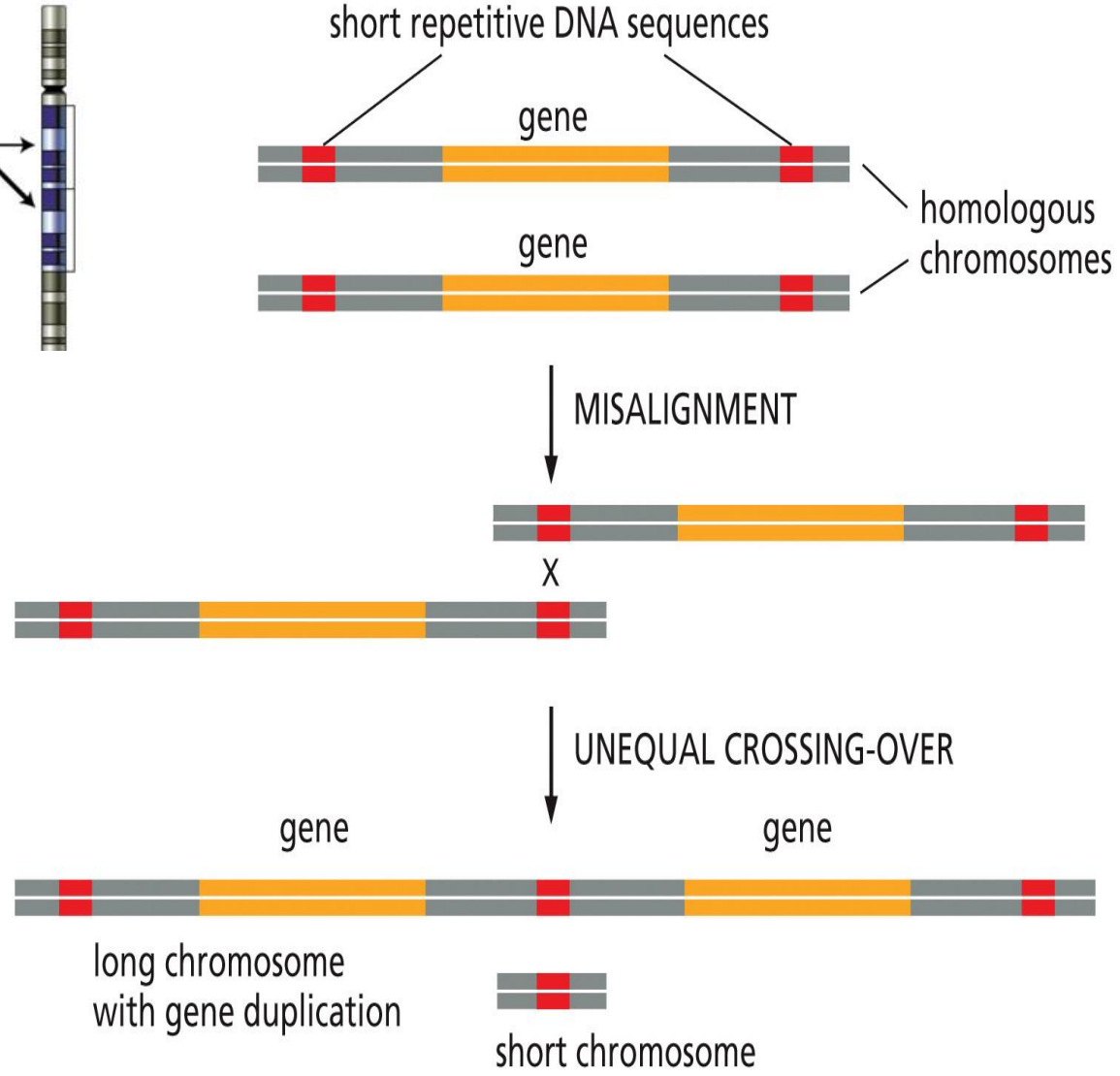
Crossovers between short, repeated DNA sequences in homologous chromosomes can lead to duplication events

Example:

- Two chromosomes undergo homologous recombination repair at short repeated sequences located on both sides of a gene

- If the chromosomes do not line up properly there can be **unequal crossing over**
  - One chromosome gets 2 copies of the gene and the other will get none
  - Were this to occur in the germ line, some progeny will inherit the long chromosome, while others will inherit the short one, increasing genetic diversity



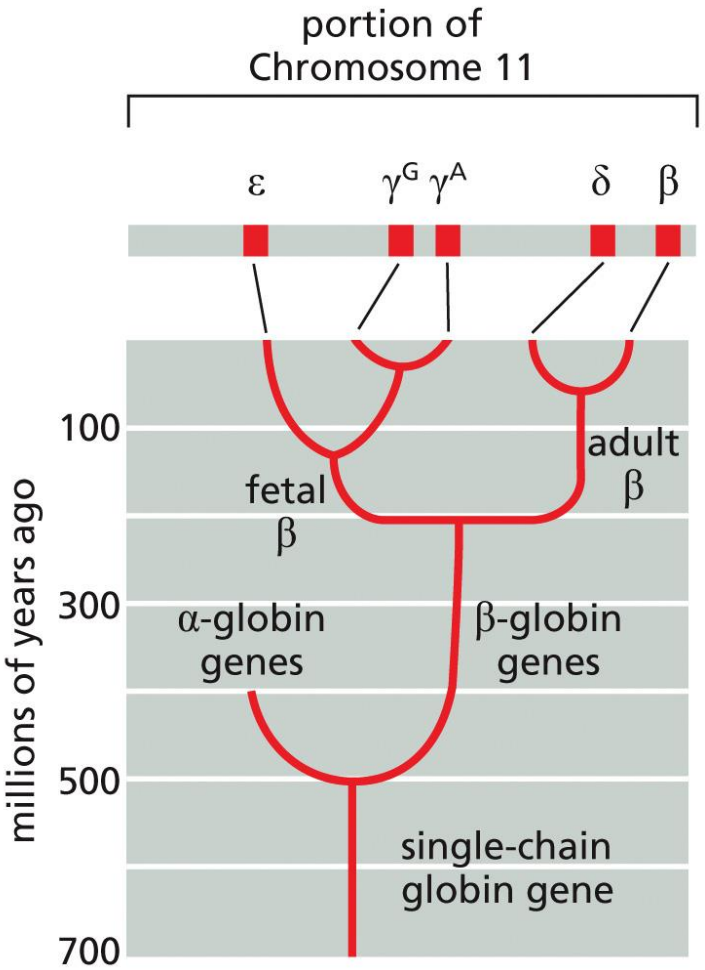


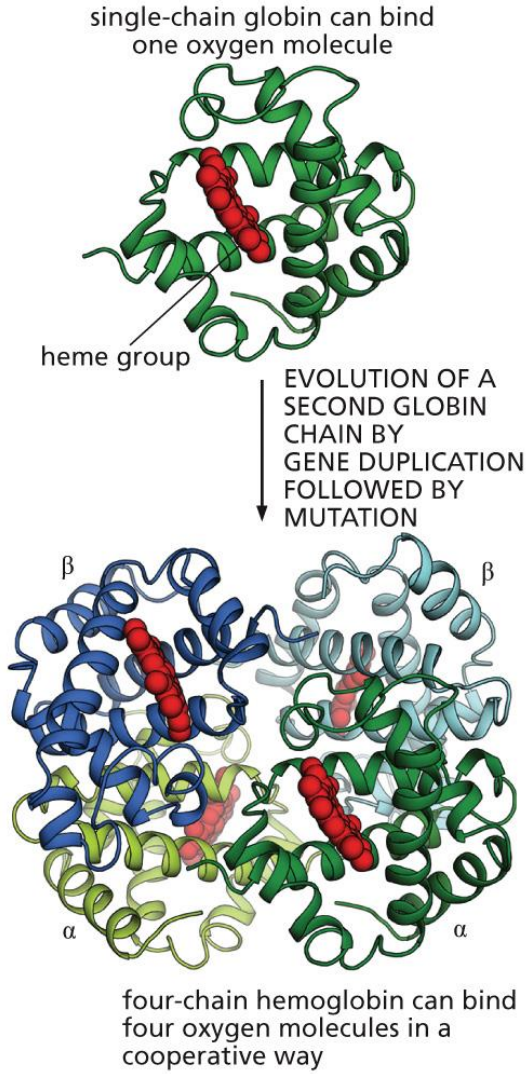
DNA duplications can give rise to families of related genes

- After gene duplication occurs new genes that encode for different functions can form due to continued divergence
  - Each of the two copies of the gene can accumulate different mutations
  - Can lead to differences in the way the protein functions

<u>Example:</u>

- α and β-globin gene families (form hemoglobin; oxygen carrying protein in the blood)
- Leads to more efficient oxygen transport





Whole-genome duplications have shaped the evolutionary history of many species

4N	apple, potato

6N	wheat, kiwi

8N	sugarcane, strawberry

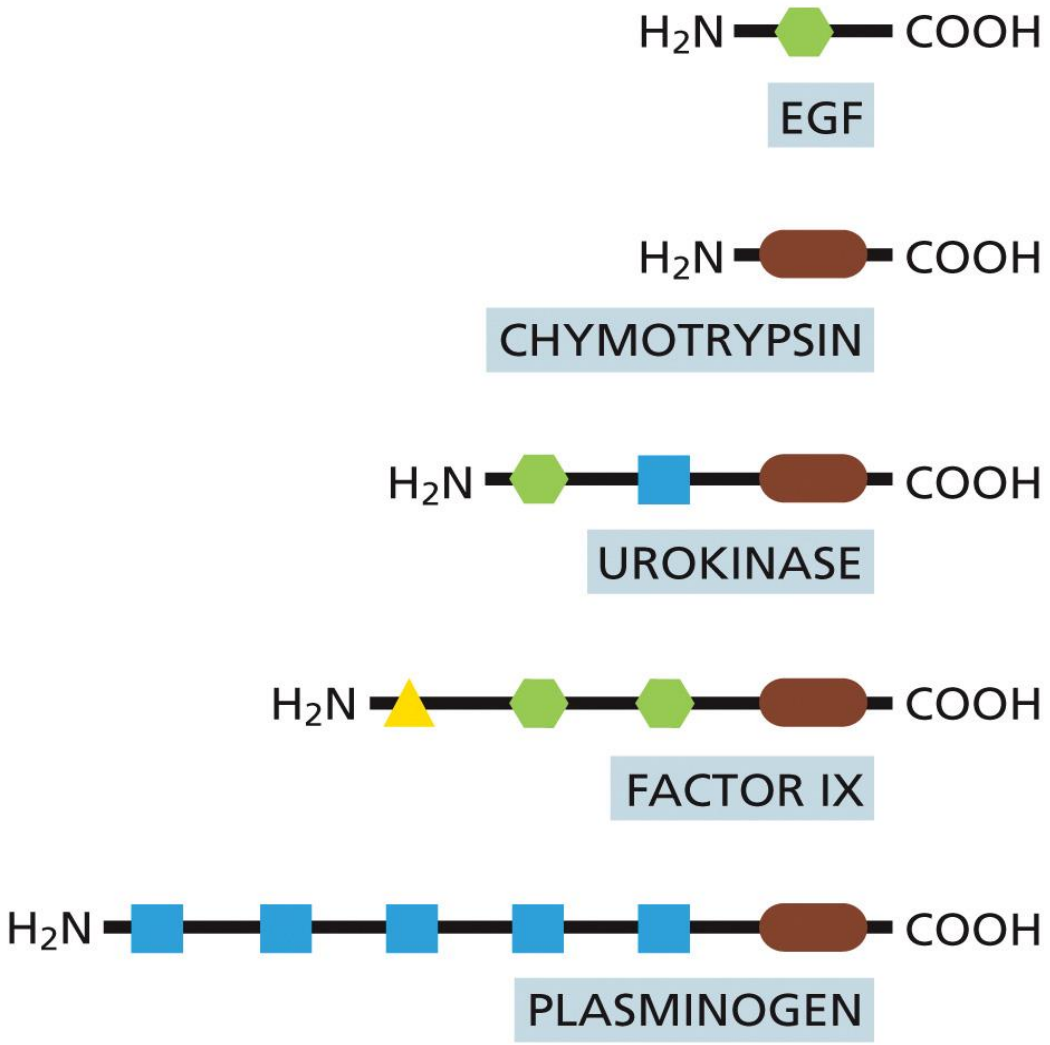
Many crop plants have undergone whole-genome duplications

- Some arose spontaneously while others were intentionally propagated by plant breeders- easier to cultivate, larger fruits, more flavorful, devoid of indigestible seeds, etc.
- N indicates the ploidy of each type of plant: for example, wheat and kiwi are hexaploid—possessing six complete sets of chromosomes (6N)

Novel genes can be created by exon shuffling

Exon shuffling: addition of exons from one gene to another gene

- Recombination that occurs due to intronic splicing that can generate proteins with new combinations of protein domains



Genes can be exchanged between organisms by horizontal gene transfer

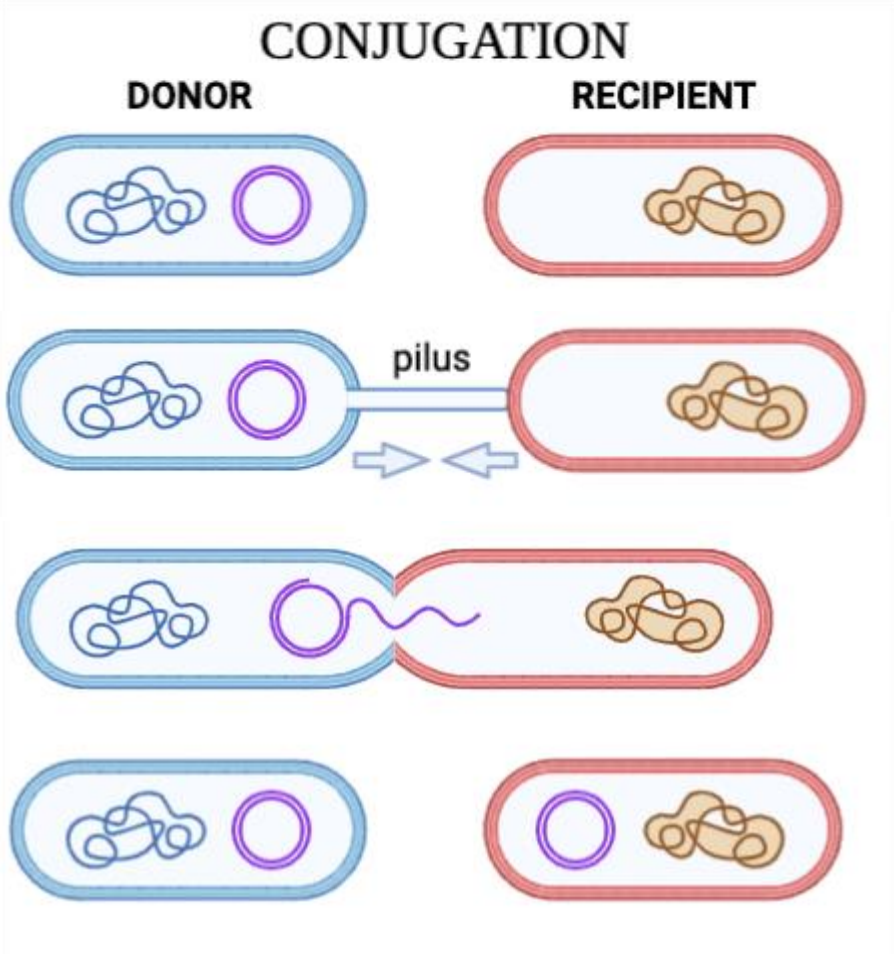
Horizontal gene transfer refers to the movement of genetic material from one species to another

<u>Example:</u>

Bacterial cells can exchange DNA through **conjugation**

- A donor cell captures a recipient cell using a fine appendage called a sex pilus
- DNA moves from the donor cell, through the pilus, into the recipient cell
  - Often this is the mechanism by which antibiotic resistance genes are transferred

- Some simple eukaryotes also undergo horizontal gene transfer



https://study.com/skill/practice/comparing-mechanisms-of-horizontal-acquisition-of-genetic-material-in-prokaryotes-their-effect-on-genetic-variation-questions.html

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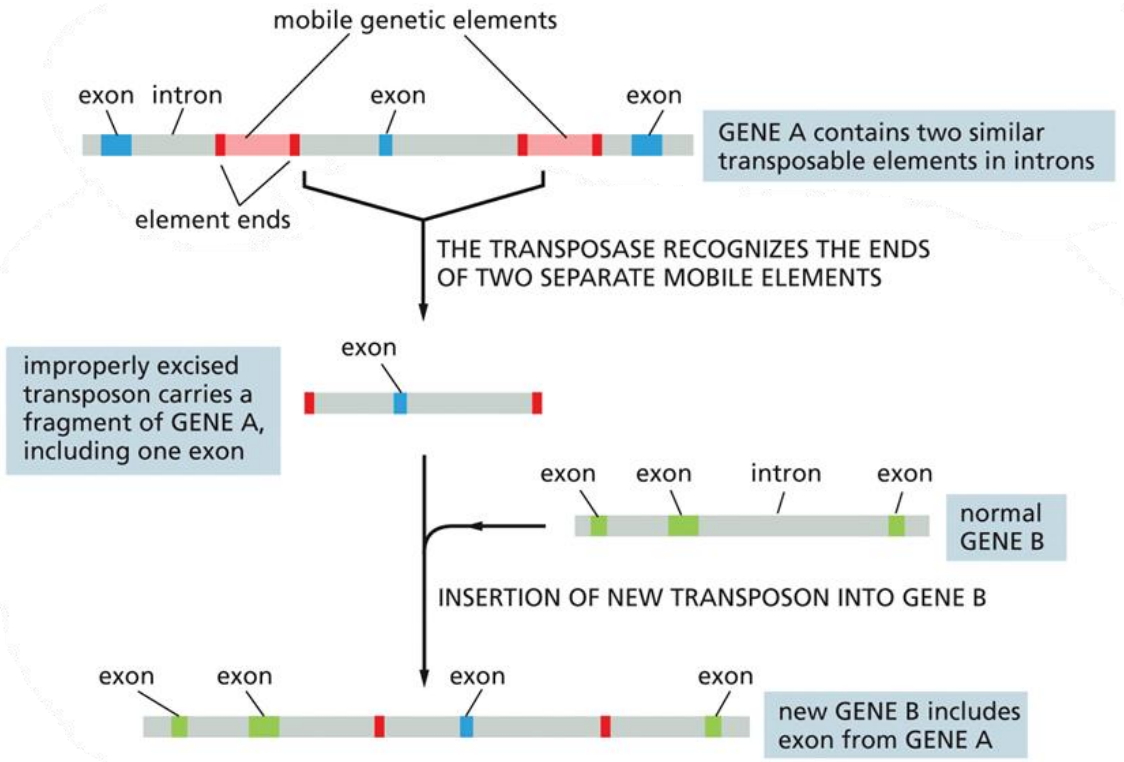
Mobile genetic elements can move exons from one gene to another

- Mobile genetic elements **(transposons/transposable elements):** DNA sequences that can move from one chromosomal location to another “jumping genes”
- **Transposase** is a specialized enzyme that cleaves unique DNA sequences at each end of two mobile elements and mediates insertion into other sequence specific regions in the genome
- Transposons are found in virtually all cells and can insert into any region of the genome, and lead to generation of novel genes or can alter gene regulation



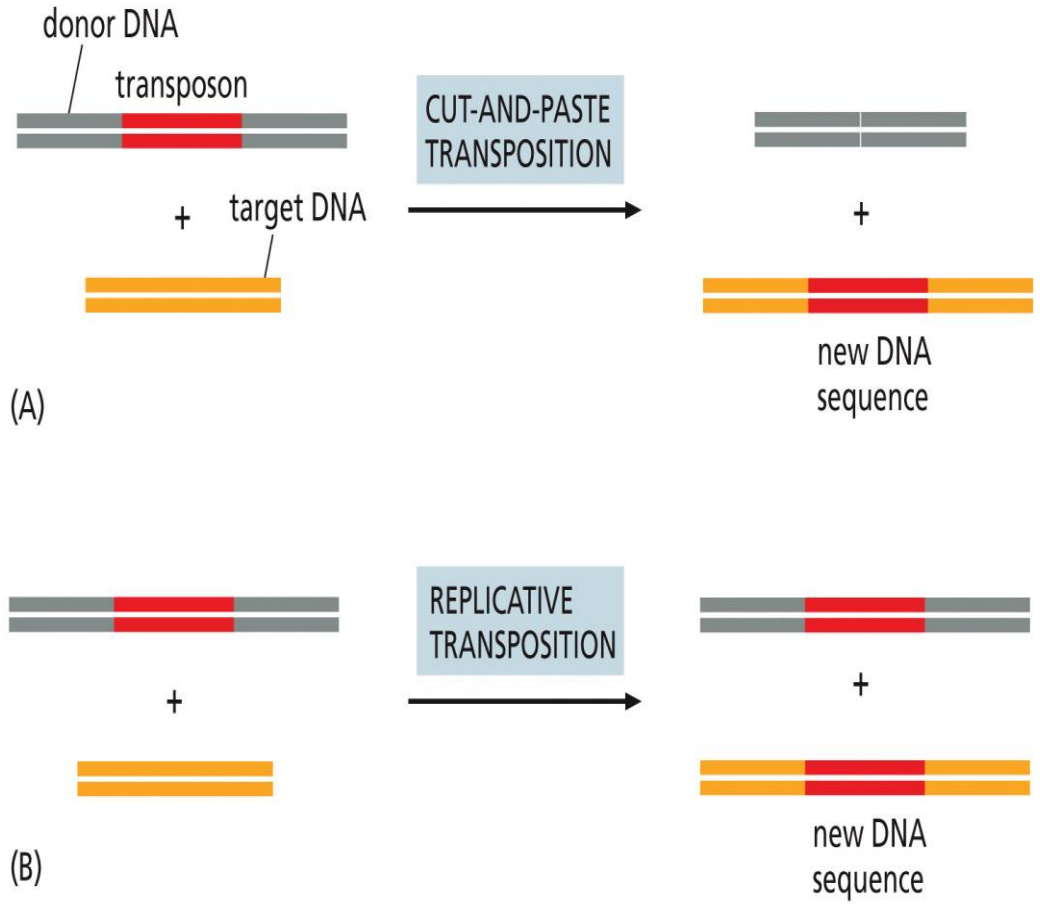
Bacteria utilize DNA-only transposons

Two principal mechanisms:

1. **Cut-and-paste transposition**
   - The transposon gets cut out of the donor DNA and inserted into the target DNA
   - Leaves behind a broken donor DNA molecule, which is then repaired

2. **Replicative transposition**
   - The mobile genetic element is copied by DNA replication, leaving the donor unchanged
   - The target DNA receives a copy of the mobile genetic element

- The donor and target DNAs can be part of the same DNA molecule or reside on different DNA molecules



Eukaryotes also have retrotransposons, which move using an RNA intermediate

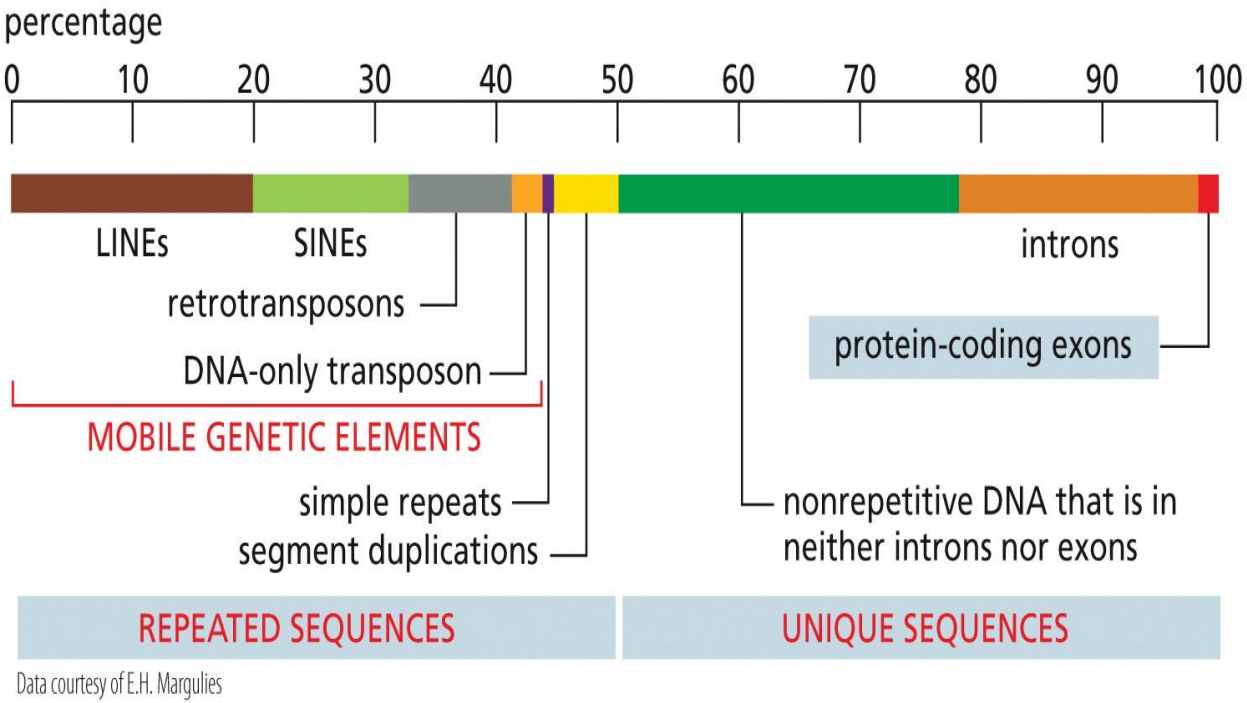
- **Retrotransposons** move throughout the genome utilizing a copy-and-paste mechanism that uses an RNA intermediate

- *Reverse transcriptase* is an enzyme that synthesizes DNA from RNA

1. **Long-interspersed nuclear element (LINEs)**
    - ~20% of human genome
    - Encodes its own reverse transcriptase for transposition

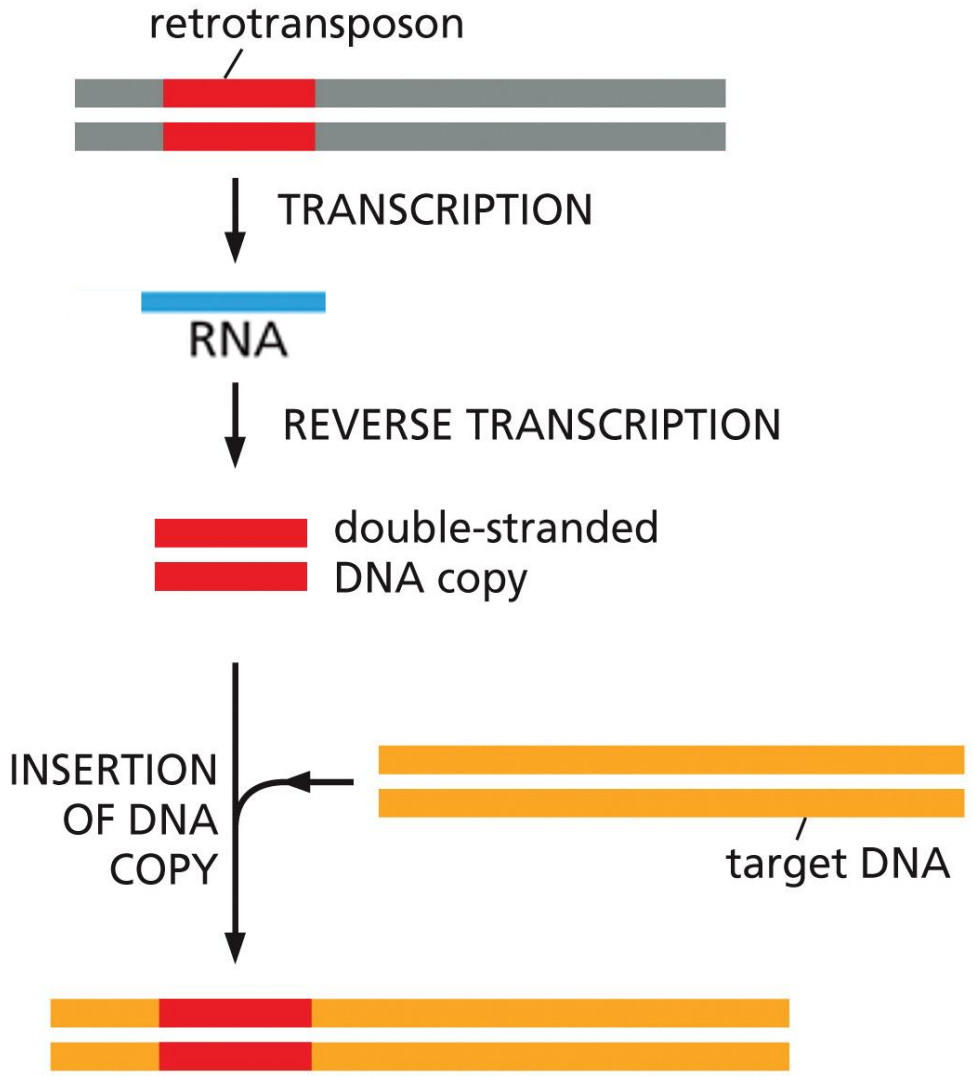
2. **Short interspersed nuclear elements (SINEs)**
    - ~10% of human genome
    - Does not encode for a reverse transcriptase, instead uses the cell's machinery to replicate

Nearly half of human genome is made of repetitive sequences

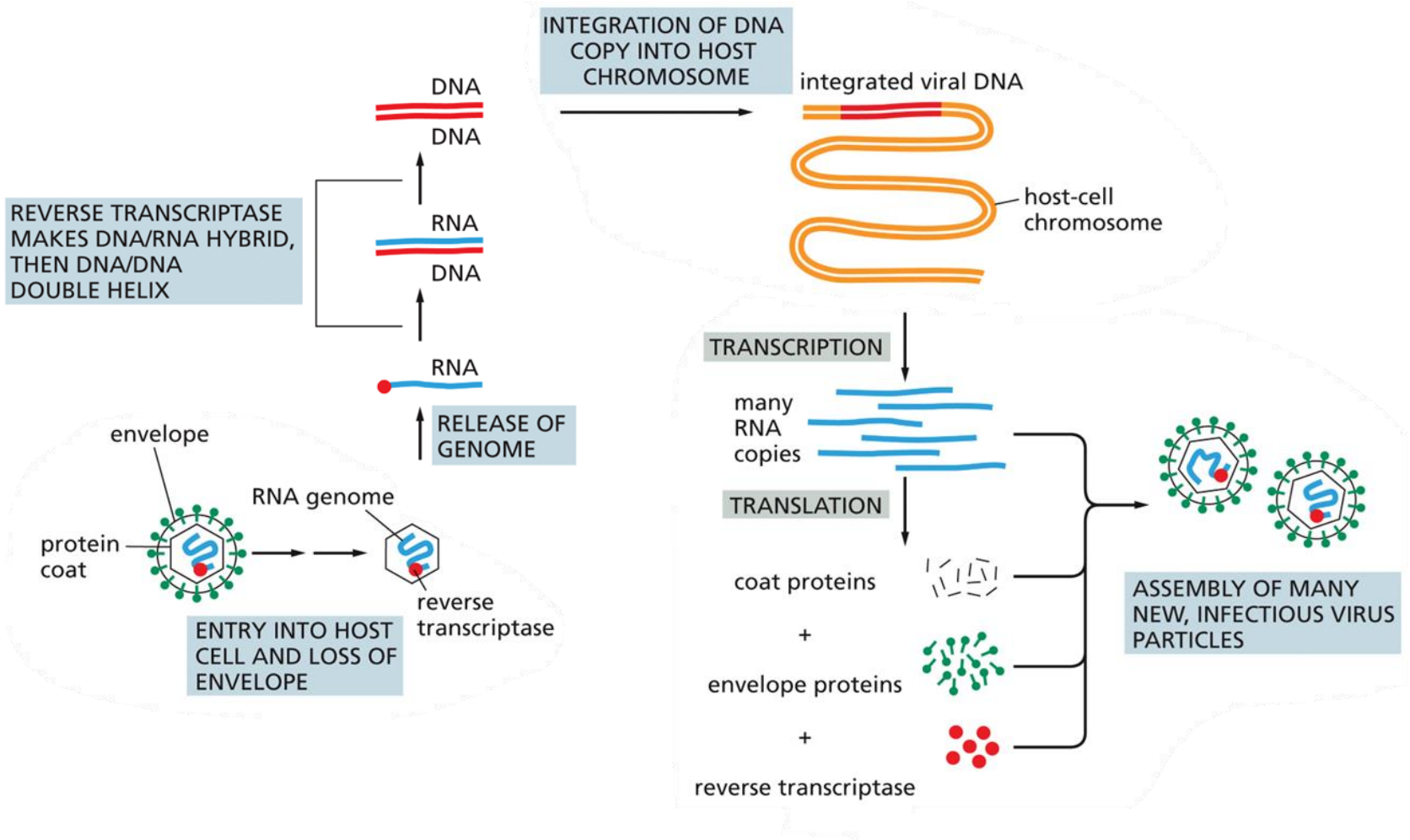


Long-interspersed nuclear elements (LINEs) retrotransposon mechanism

- These transposable elements are transcribed into an RNA intermediate using host cell's RNA polymerase (transcription)
- A double-stranded DNA copy of this RNA is synthesized by the enzyme **reverse transcriptase** (a DNA polymerase that uses RNA as a template)
- The DNA copy is then inserted into the target location, which can be on the same or different DNA molecule
- The donor retrotransposon remains at its original location, so each time it transposes, it duplicates itself.

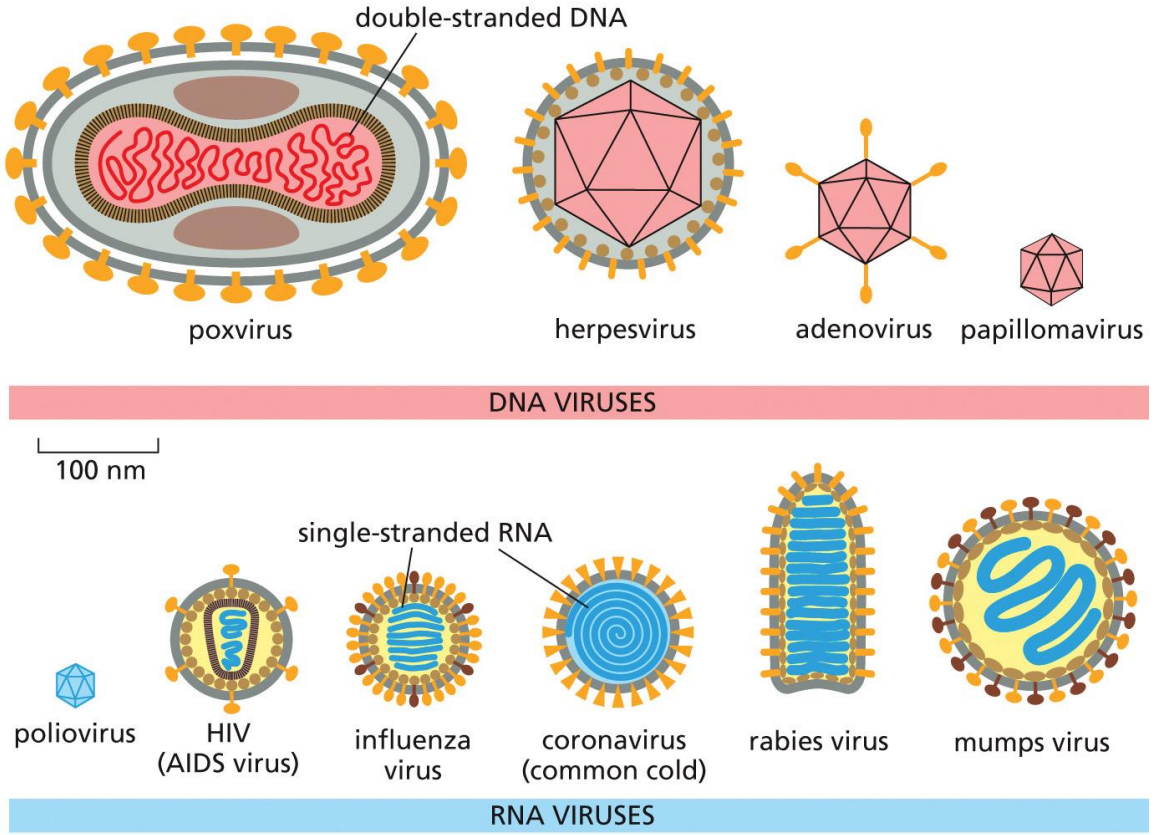


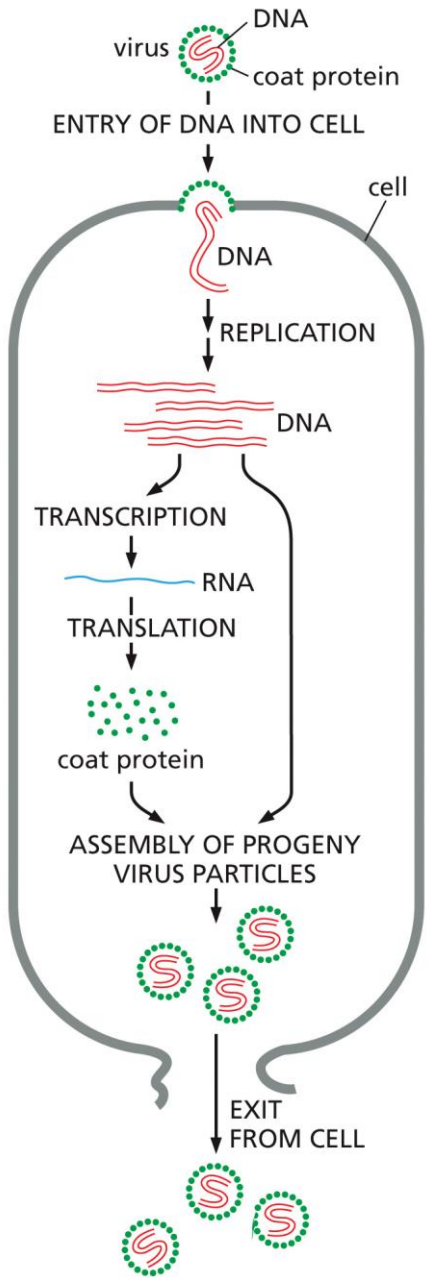
Viruses can integrate into the host genome and hijack the host transcription and translation machinery



*Depending on where a virus integrates it can cause different effects on gene expression
Ex: a strong viral promoter inserted in front of a host gene will turn it on

Viruses can move between cells and organisms





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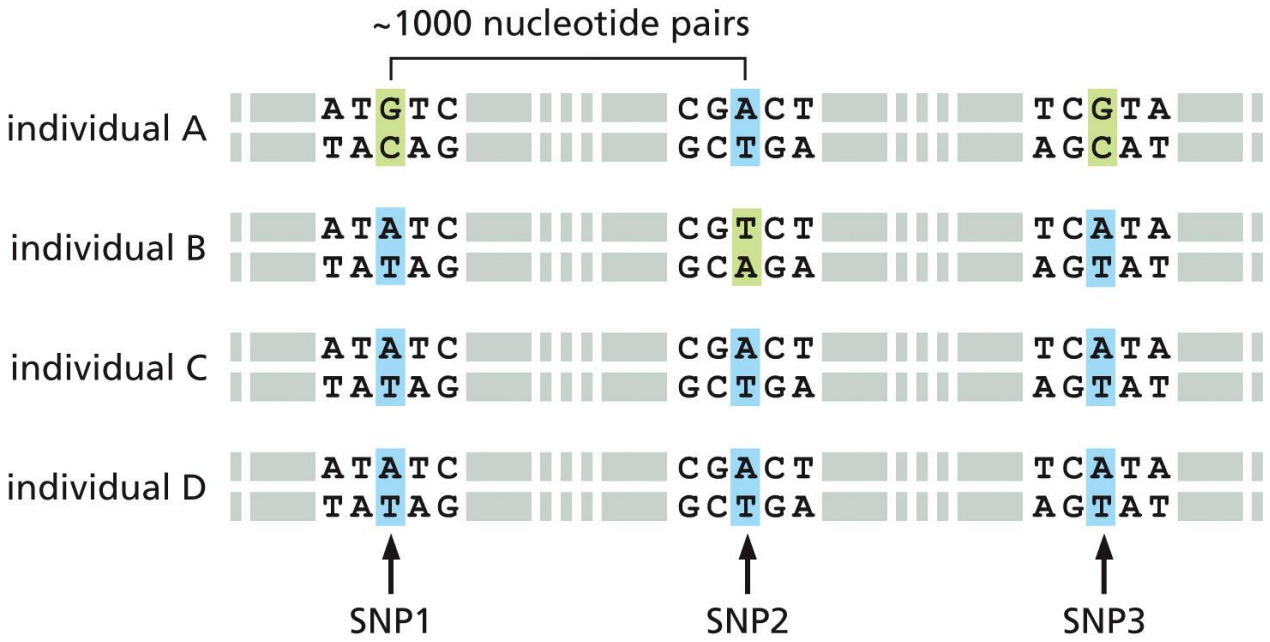
Genome variation contributes to our individuality!

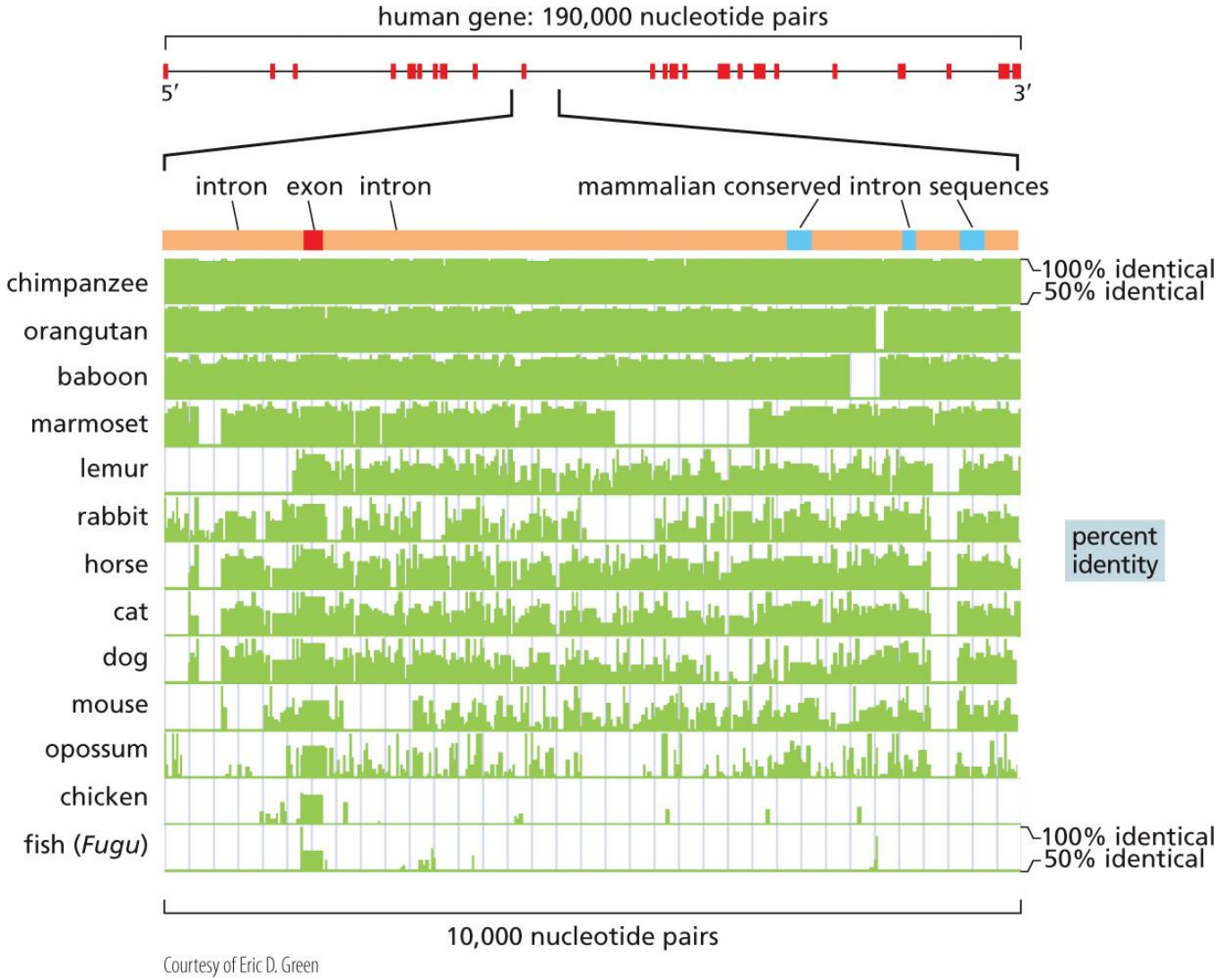
- **Single-nucleotide polymorphisms (SNPs)** are points in the genome that differ by a single nucleotide pair between one portion of the population and another
    - Most-but not all-SNPs in the human genome occur in regions where they do not affect the function of a gene
    - On average two individuals differ in their nucleotide sequences by ~0.1% (1 SNP per every 1000 nucleotide pairs or ~3 million genetic differences as roughly 3 billion nucleotides in the human genome)



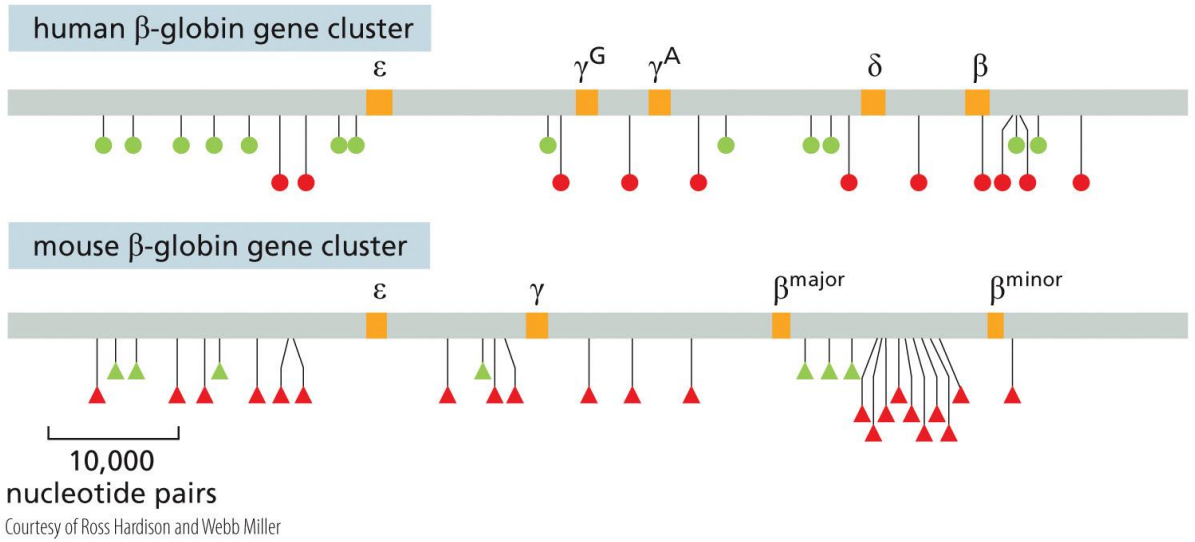
Comparison of nucleotide sequences from many different vertebrates reveals regions of high conservation

- Functionally important genome regions are conserved across species via **purifying selection:** elimination of individuals carrying mutations that interfere with important functions

- *Differences in gene regulation* may help explain how animals with similar genomes can be so different, similar to how our different cell types are so specialized even with the exact same DNA



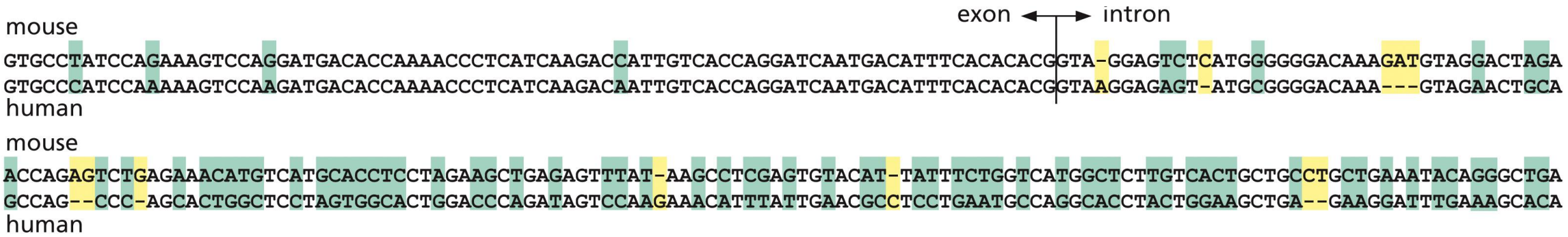
Differences in the positions of mobile genetic elements in the human and mouse genomes reflect the long evolutionary time separating the two species



- Stretch of human Chromosome 11
  - Humans have 5 functional β-globin-like genes vs. 4 in mice
  - *Alu* sequences (*green*) and *L1* sequences (*red*) (SINEs and LINEs, respectively)

  - Note the absence of mobile genetic elements within the globin gene exons; attributed to *purifying selection*

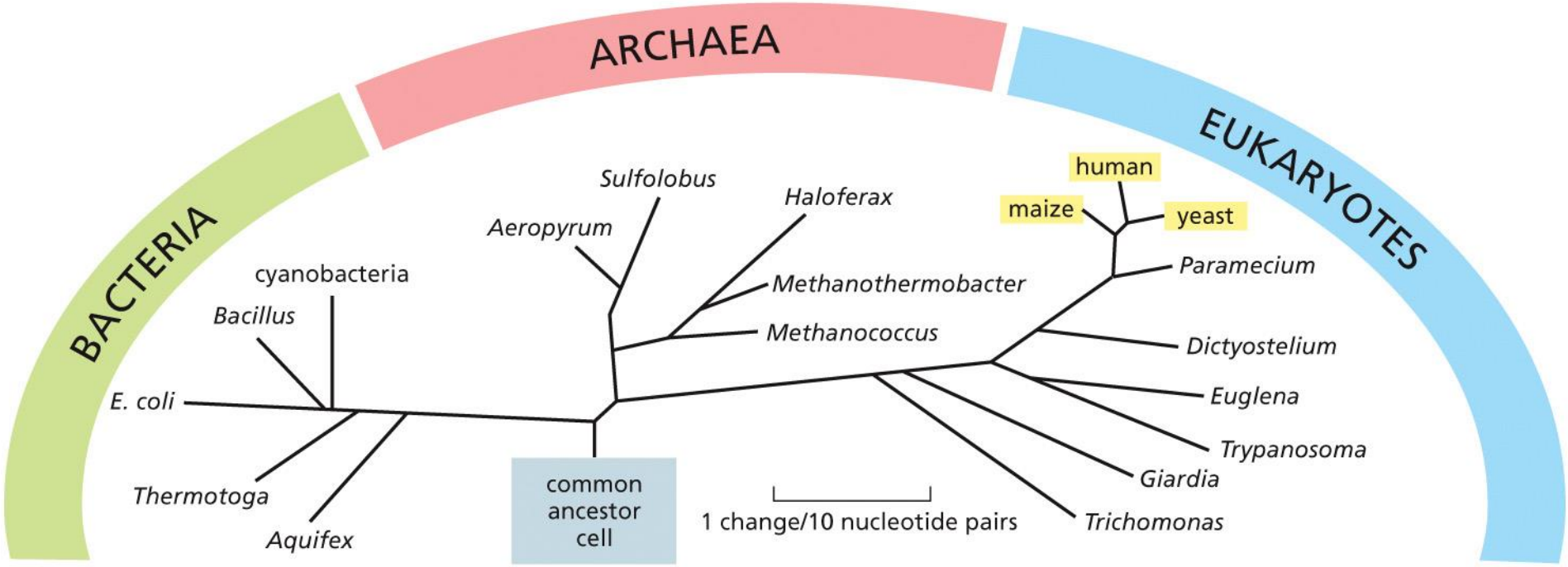
Accumulated mutations have resulted in considerable divergence in the nucleotide sequences of the human and the mouse genomes



- Human and mouse leptin gene sequences
  - Positions where the sequences differ by a single nucleotide substitution are boxed in *green*
  - Positions where they differ by the addition or deletion of nucleotides are boxed in *yellow*

  - Note that the coding sequence of the *exon is much more conserved* than the adjacent intron sequence (rightmost section of top rows as well as second set of rows)

Sequence conservation allows us to trace the most distant evolutionary relationships and build phylogenetic trees



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Reflection/Feedback

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