Summer of Science: Evolution

A Journey from Genes to Ecosystems

Introduction

Your Teachers

- Alexandros Kourtidis

- Maria Madrid

- Loran Heymans(Ned)

- Bram Janssens(Ned)

- Filip Vandelook(Ned)

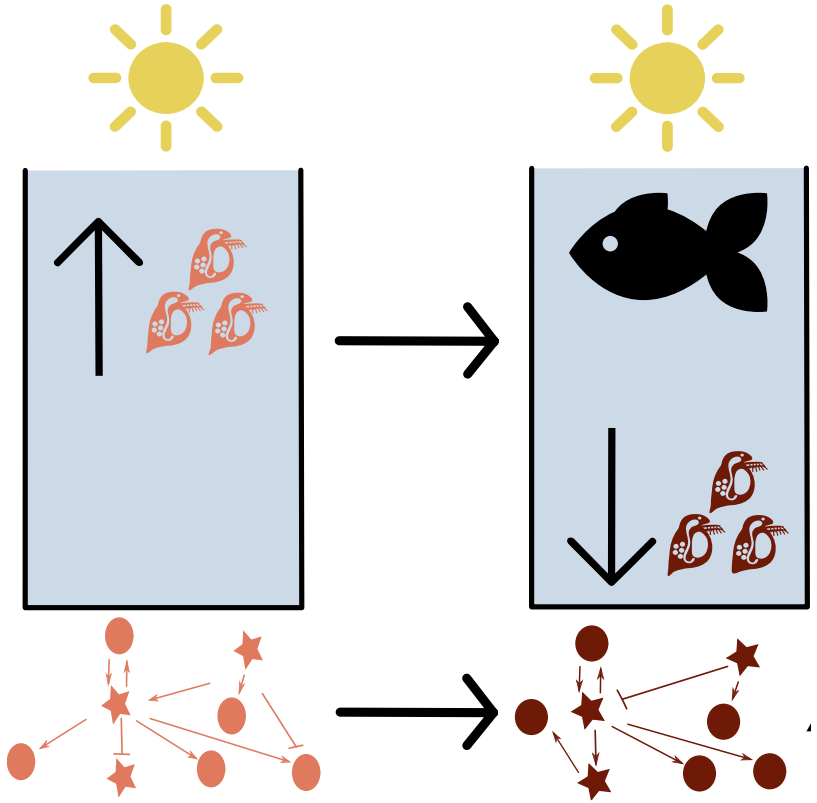
- Kristina Yefimak(Ned)

- Aaron Kiggen(Ned)

- Zorimar Pacheco

Your Teachers

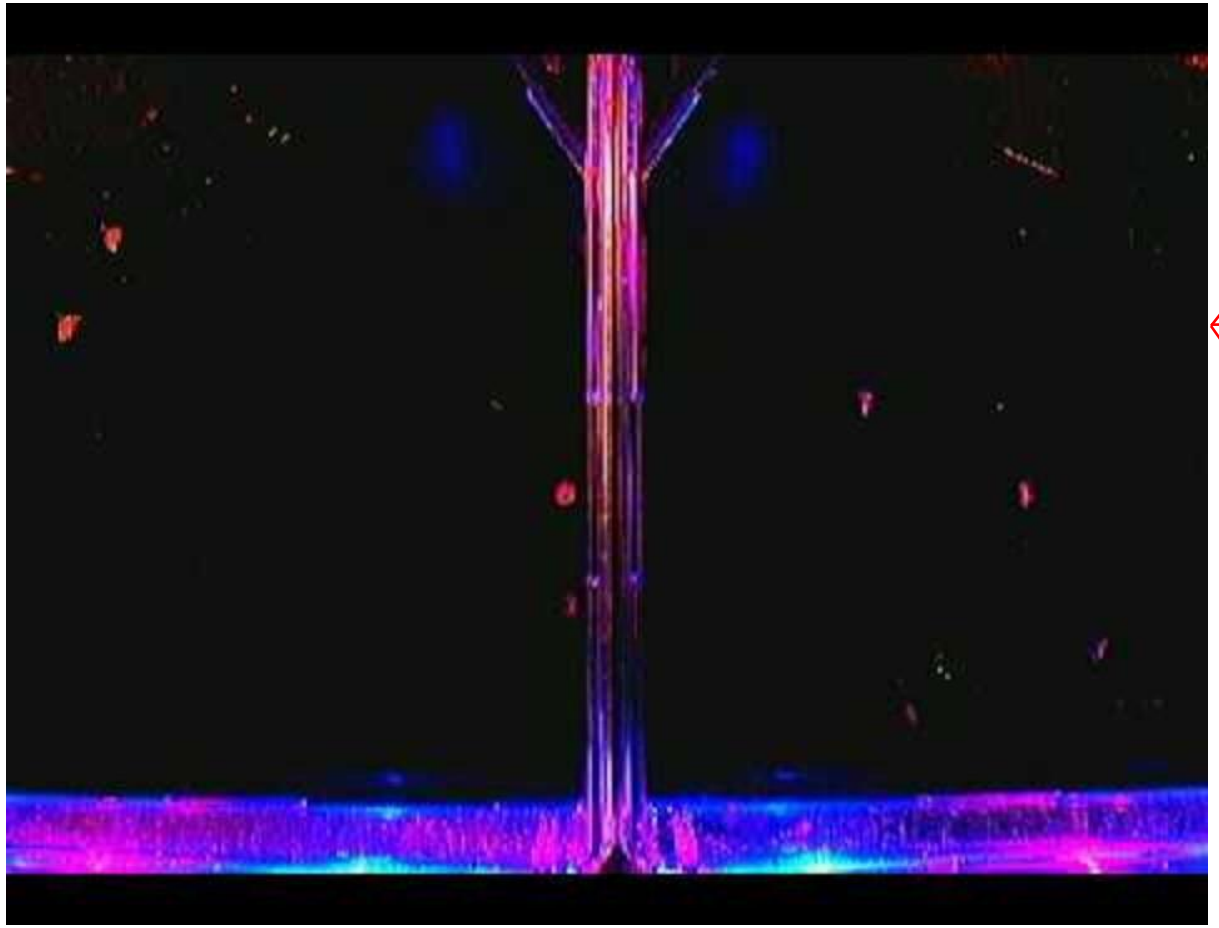
- Aaron Kiggen(Ned)



1. What are the **mechanisms** behind this plastic response?
   - *Day 2*
2. How is **variation** in this behavioural response achieved?
   - *Day 1 & 2*
3. How does it **evolve**?
   - *All the days!*

Your Teachers

- Aaron Kiggen(Ned)



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2. How is **variation** in this behavioural response achieved?
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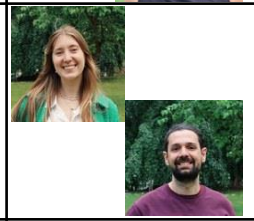
3. How does it **evolve**?
   - *All the days!*

The Schedule

= theory session

= practical session

DAY 1	• **Evolutionary theory** • **Central dogma** • **Variation**		**/**	
DAY 2				 
DAY 3				 
DAY 4				 
DAY 5		 		 

Learning goals

DAY 1	• **Think like a researcher** • **Grasp the core principles of evolutionary history** • **Molecular basis of inheritance** • **How to map genotypic variation to phenotypic variation**
DAY 2	
DAY 3	
DAY 4	
DAY 5	

Exciting?

Day 1:

Learning goals

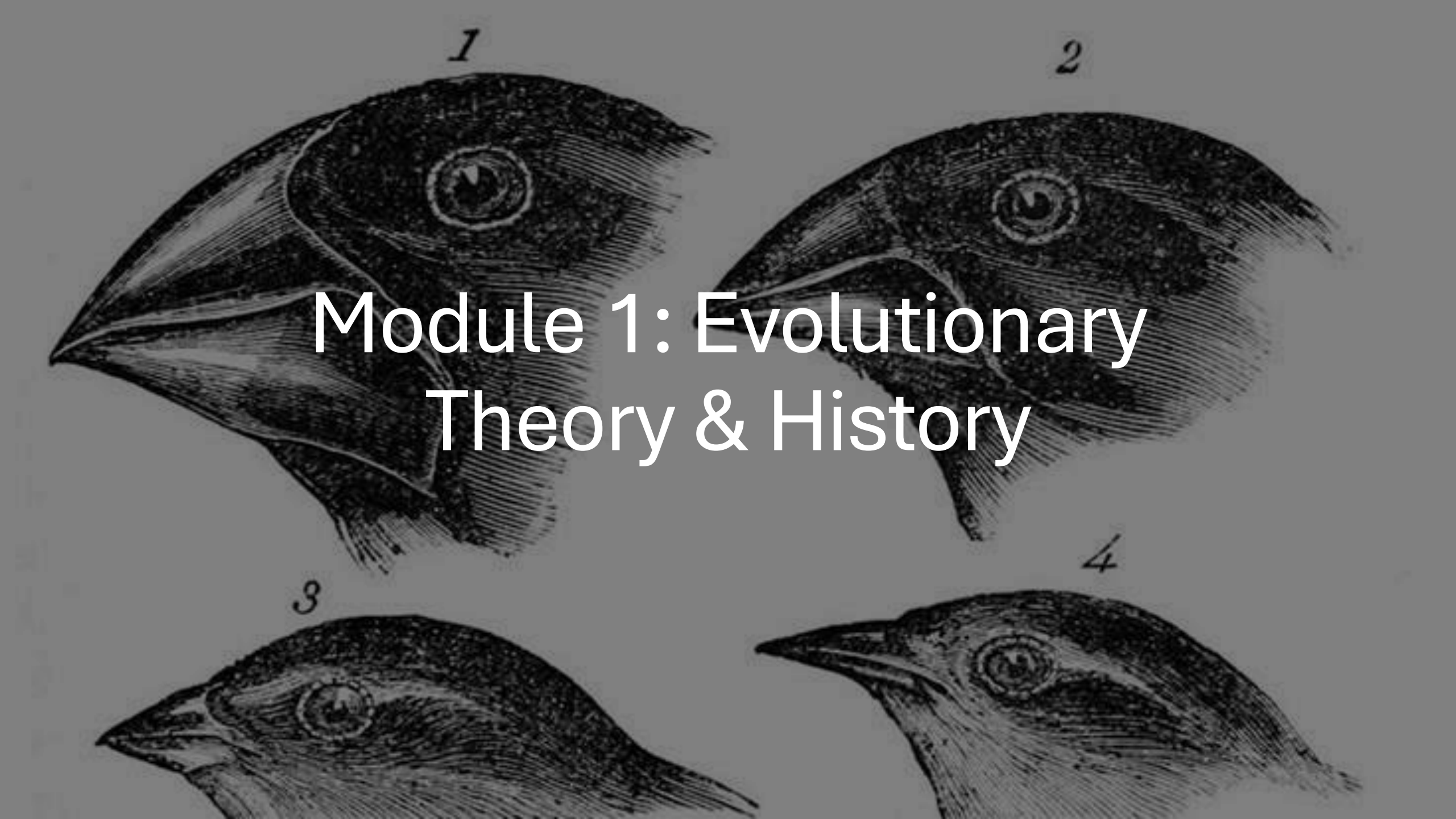
Exciting?

DAY 1	• **Think like a researcher** • **Grasp the core principles of evolutionary history** • **Molecular basis of inheritance** • **How to map genotypic variation to phenotypic variation**

→ Ask questions! About anything really!

→ Think a step ahead and discuss! How could the things you learn here relate to your specific interests?

→ ....



Module 1: Evolutionary Theory & History

The scientific method

The scientific method

?

The scientific method

Step 1: Make an observation that leads you to ask a question

Turles have shells but ….

How are they made?
Why did turtles evolve shells?
Are they all the same?
…

The scientific method

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Turles have shells but ....

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The scientific method

Step 2: Make a hypothesis - a testable explanation, or reasonable prediction of what will happen.

Turles have shells but ….

How are they made?
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Hypothesis:

Turtles have evolved shells to protect them against predators and environmental threats.

The scientific method

Step 3: Test that hypothesis with an experiment that can be repeated.

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Turles have shells but ....

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...

Land turtles are slow and vulnerable to predation

Sea turtles are fast and can swim away from predators

Hypothesis:

Turtles have evolved shells to protect them against predators and environmental threats.

Measure **shell size** and **predation risk** across 356 species in the turtle order

The scientific method

Step 4: Analyze the results.
Step 5: Report the conclusions.

Across 356 species in the turtle order those who experience **more predation** risk have **thicker and stronger shells**

Turles have shells but ….

How are they made?
Why did turtles evolve shells?
Are they all the same?
…

Hypothesis:

Turtles have evolved shells to protect them against predators and environmental threats.

Land turtles are slow and vulnerable to predation

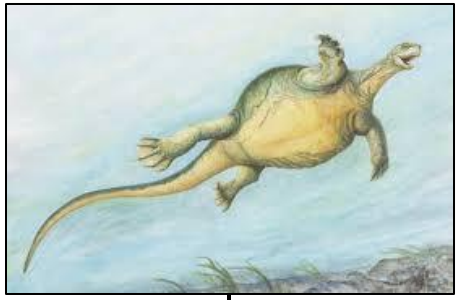
➔ Have thick shells to protect themselves

Sea turtles are fast and can swim away from predators

➔ Have lighter shells to swim faster

The scientific method

Across 356 species in the turtle order those who experience more predation risk have thicker andstronger shells



Step 6: Use the conclusion to make new hypotheses.

Turles have shells but ….

How are they made?
Why did turtles evolve shells?
Are they all the same?
…

Hypothesis:

Turtles have evolved shells to protect them against predators and environmental threats.

Hypothesis:

Turtles have evolved shells that protect them against predators and environmental threats which then evolved to be thicker or thinner depending on the predation risk.

The scientific method

Scientific Theory

Evolution: fact and theory

Fact:

Turtles have **evolved** protective shells.

Theory:

This evolution of the turtle shell can be explained by the theory of **natural selection**—those ancestral reptiles had slight protective **advantages against predators** and were **more likely to survive and reproduce**. Over many generations, this led to the evolution of fully enclosed shells.

Evolution: fact and theory

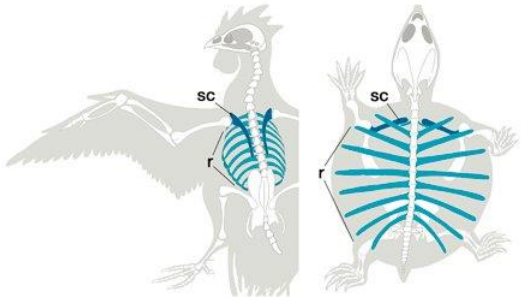
Theory:

This evolution of the turtle shell can be explained by the theory of **natural selection**—those ancestral reptiles had slight protective **advantages against predators** and were **more likely to survive and reproduce**. Over many generations, this led to the evolution of fully enclosed shells.

It this really the whole story? Well no, new observations can cause new questions and updates on the theory. That is why it called a theory and not a fact.

Evolution: fact and theory

Step 1: Make an observation that leads you to ask a question



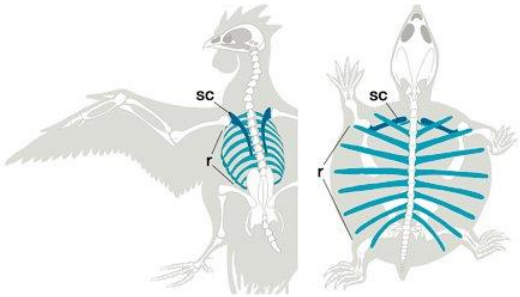
Developmental Biology

Odontochelys

Odontochelys and the current turtle carapace grew from vertebra, which contradicts the prevailing hypothesis that the shell was formed by bony deposits fusing together.

Evolution: fact and theory

Step 1: Make an observation that leads you to ask a question



Developmental Biology

Odontochelys

So broad shoulders and ribs seemed to play an important role in shell evolution… Why?

Evolution: fact and theory

Step 1: Make an observation that leads you to ask a question

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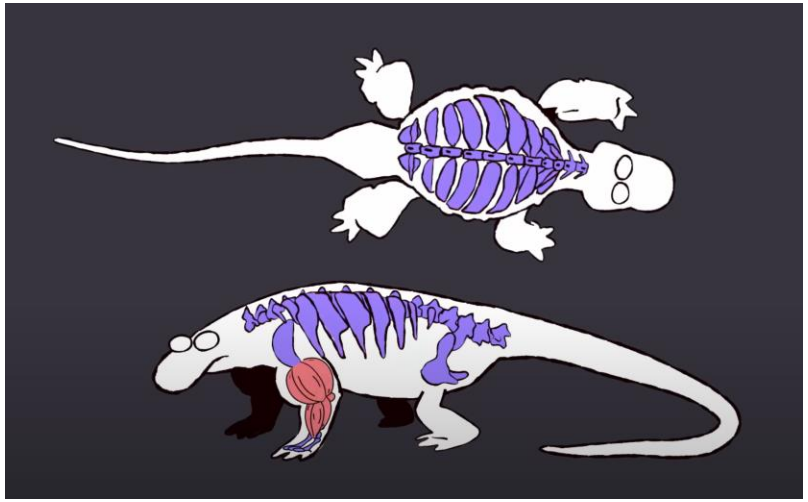


Eunotosaurus, a turtle without a shell? Looked a lot like odontochelys and had the same big ribs…

But it lived on land? And why did it need its big ribs in the first place?

Evolution: fact and theory

Evolution: fact and theory

Ancestor had small powerful legs to dig
=> ribs broad and fused together as an anchor for digging

These adaptations made them really slow…
=> needed more protection and water became a more attractive habitat

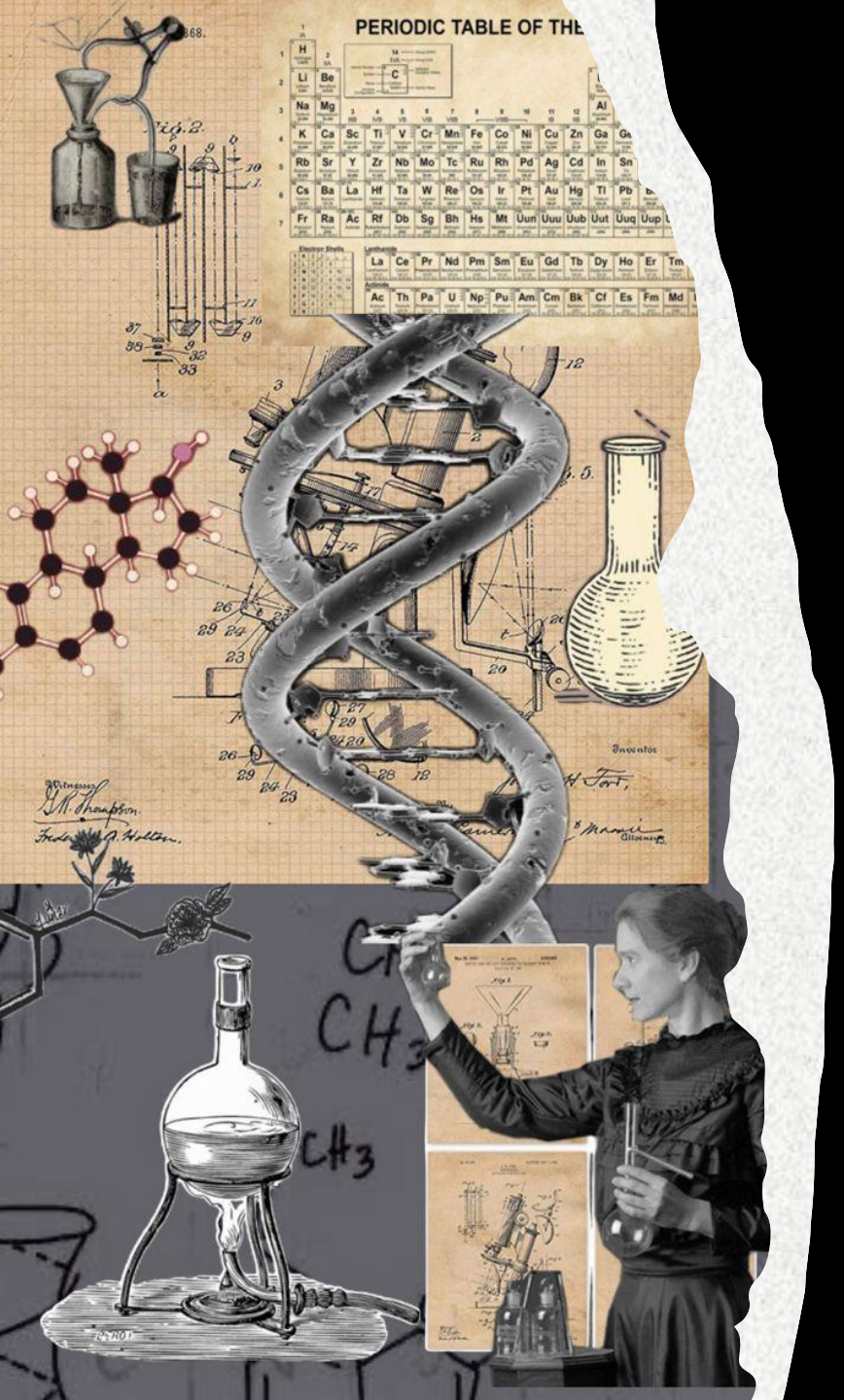
Odontochelys shows us where the shell came from i.e. these rib adaptations made for digging
=> eventually forming the entire shell for protection

Why did turtles evolve shells?

Hypothesis:

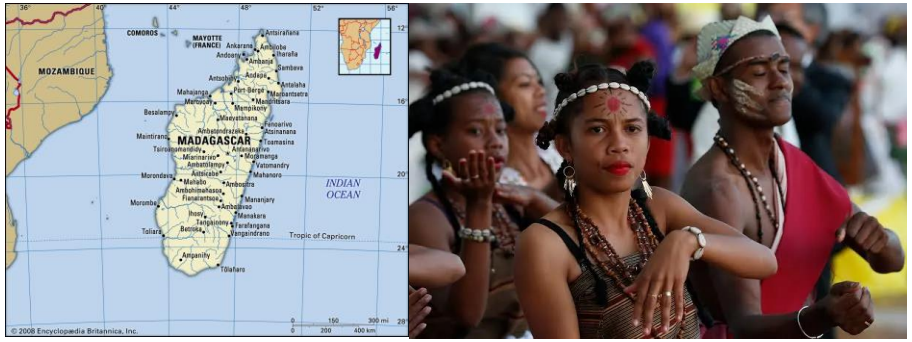
Over time the evolutionary purpose of the turtle's "shell" changed from digging to protection!

Evolution is a fact but how it exactly works is the theory! If this was also a fact, I would not have a job 😝



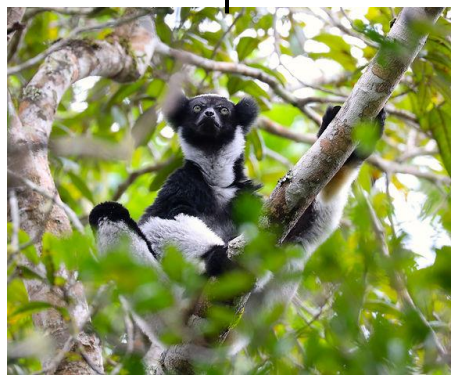
The history behind the theory

The Evolution of Evolution: Pre-Darwin (<1800)



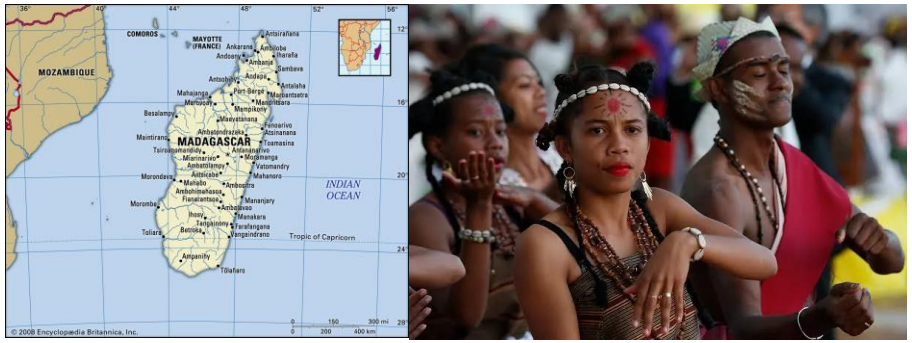
Malagasi people from Madagascar

Hmmm… Common ancestor…
Forms changing depending on the environment….
Looks like something I know….



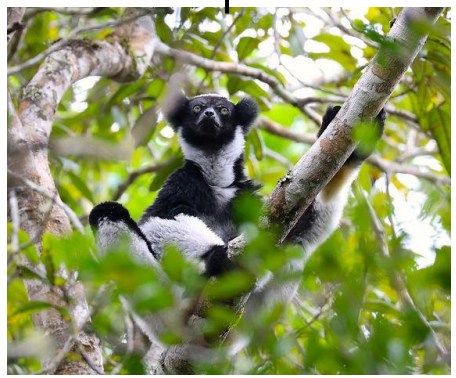
Indri (Lemur native to Madagascar)

The Evolution of Evolution: Pre-Darwin (<1800)



Malagasi people from Madagascar

Why?



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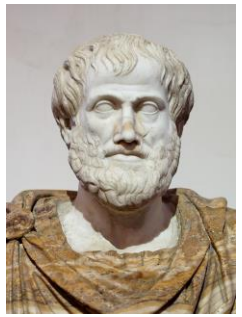
Why?

The Evolution of Evolution: Pre-Darwin (<1800)

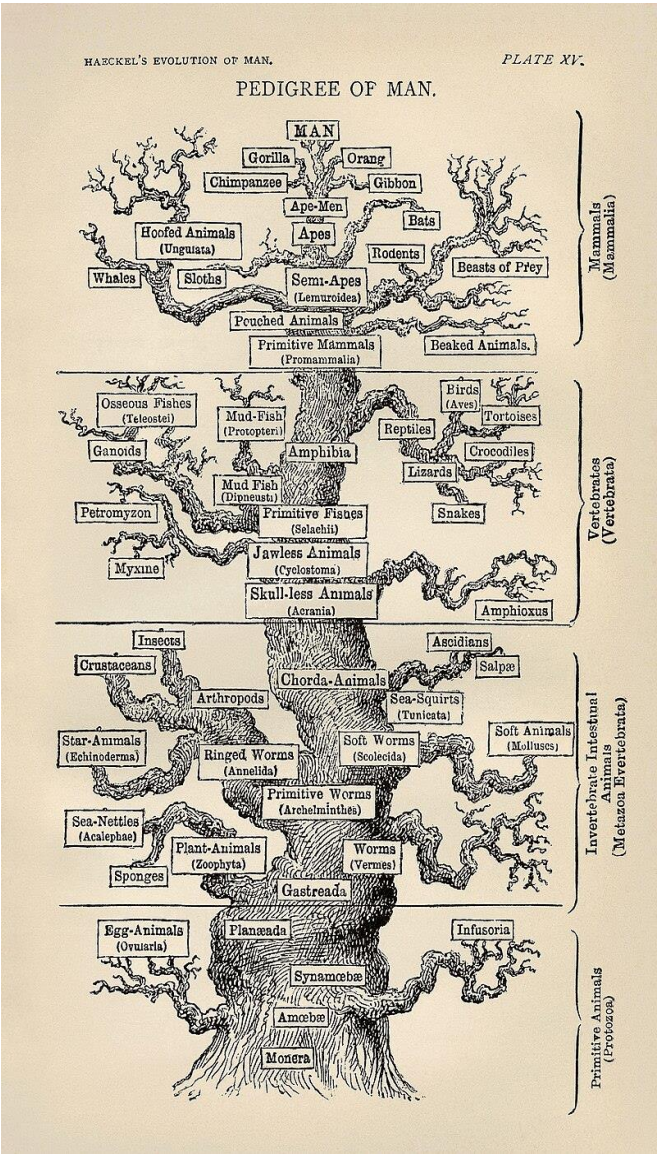
Greek essentialism & 17th century taxonomists



Aristotle

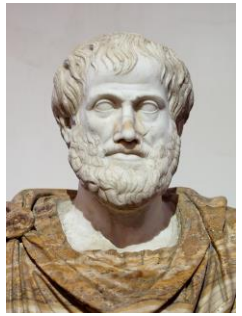
Aristotle:

- Ranking beings based on "complexity of structure and function", scala naturae ➔ **species are unique forms that can not change**
  - Essentialism
  - Nature strived to be perfect so change would mean imperfection
- Living organisms have a "final cause" i.e. their form perfectly suits their function ➔ **purposeful design in nature**
- Medieval European scholars were guided by both Aristotle and the Bible, and they believed that nature reflected God's perfect design



The Evolution of Evolution: Pre-Darwin (<1800)

Greek essentialism & 17th century taxonomists



Aristotle

Linnaeus

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- Medieval European scholars were guided by both Aristotle and the Bible, and they believed that nature reflected God’s perfect design

Linnaeus:

- naturalists tried to understand this divine plan by searching for a rational pattern in the bewildering array of species.
- Linneaus was convinced that he could organize all of life into a single artificial system based on traits of his choosing, one that would be his first step towards comprehending God’s design in nature.
- Got criticized for grouping humans and apes together as primates.

The Evolution of Evolution: Pre-Darwin (<1800)

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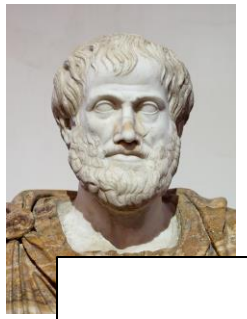

Ari

Aristotle:

- Ranking beings based on “complexity of structure and function” ➔ species unique forms that can not change
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Species were created separately with traits perfectly adjusted to their “function”…
= designed with purpose

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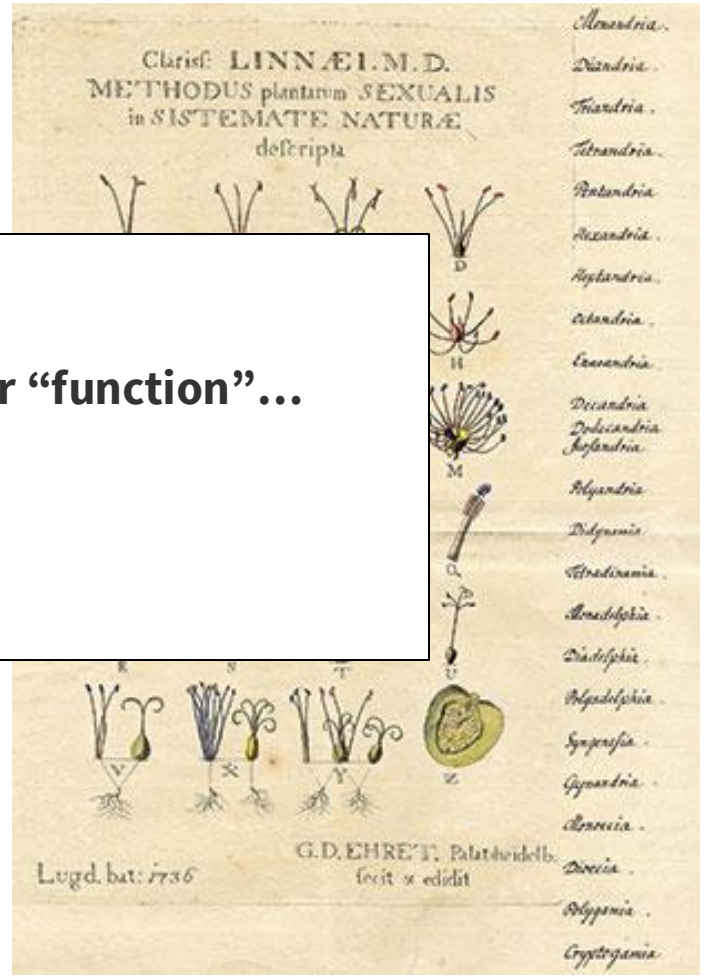

The Evolution of Evolution: Pre-Darwin (<1800)

If species were created separately and perfectly adjusted to their "function"…

Step 1: Make an observation that leads you to ask a question

Buffon

- Then why do European and North American temperate forests appear similar but host **entirely different bird and mammal species?**

The Evolution of Evolution: Pre-Darwin (<1800)

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- Then why do European and North American temperate forests appear similar but host **entirely different bird and mammal species?**
  - If species were individually created solely for their function in a given environment, you'd expect similar climates to be home to the same species—yet that's not what's observed.

Species might migrate and when they do can "improve" or "degenerate" according to their new environment but are limited by a "mould"…

The Evolution of Evolution: Pre-Darwin (<1800)

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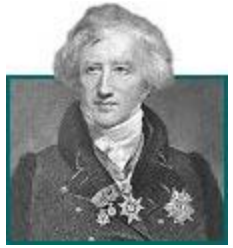

Cuvier

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- Then why do certain species such as mammoths go **extinct** if they were **perfectly adjusted**?

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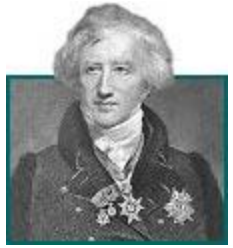


Cuvier

- Then why do certain species such as mammoths go extinct if they were perfectly adjusted?

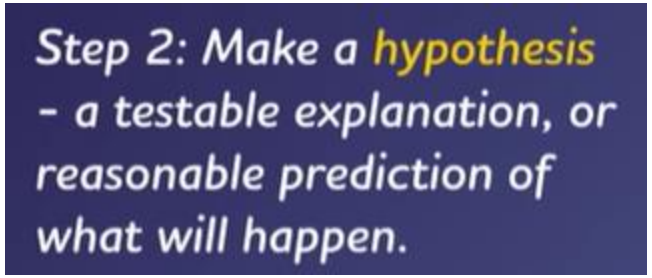
Any future theory should be able to explain as to why species can go extinct.

➔ "catastrophes" ?

Observation: Life might not have been fixed since creation. ➔ needs a theory to fit this data

The Evolution of Evolution: Darwin (1800)

If species were created separately and perfectly adjusted to their "function"…

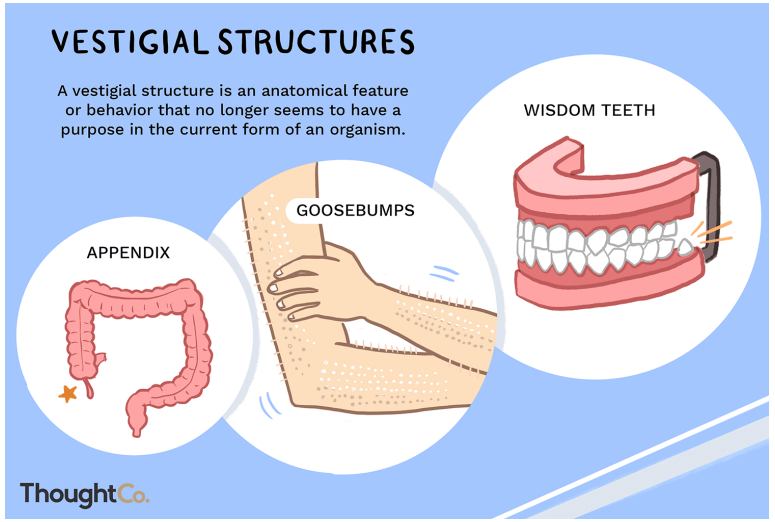


The Evolution of Evolution: Darwin (1800)

If species were created separately and perfectly adjusted to their "function"…

Step 2: Make a hypothesis - a testable explanation, or reasonable prediction of what will happen.

Lamarck



Observations:

- Animals in similar ecosystems exhibit similar traits
- Ever expanding fossil record
- Artificial breeding of dogs and other animals
- Vestigial structures (like our tailbone)

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Observations:

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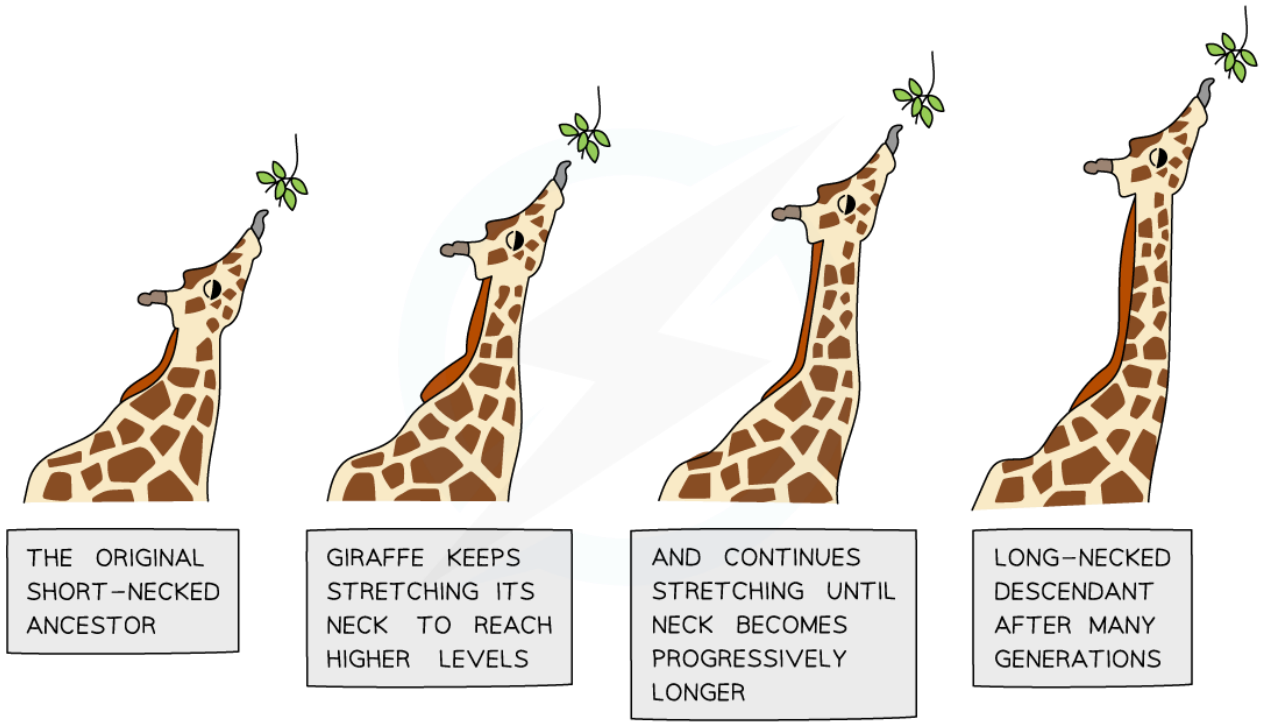
- Vestigial structures (like our tailbone)

Hypothesis: Change through use and disuse



The Evolution of Evolution: Darwin (1800)

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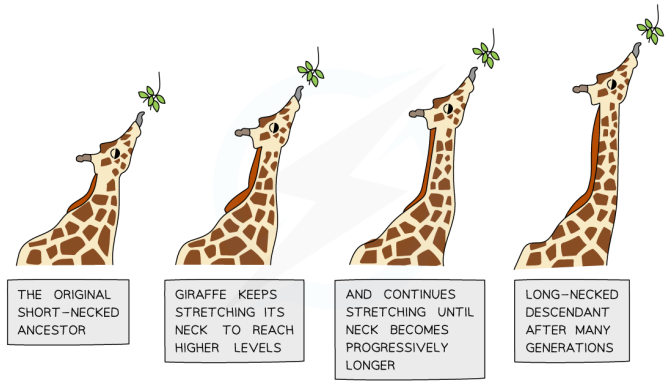
Lamarck

You might think this is a silly idea but given the evidence at the time it could fit and was a very brave idea!

Remember the dominant view was still God's perfect design and Lamarcks views were shunned by scientists at the time and Lamarck died in 1829 in poverty and obscurity.



Think for yourself:

- What current observations are not in line with this theory? What can it not explain?
- How would you test/disprove this theory?

The Evolution of Evolution: Darwin (1800)

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Lamarck

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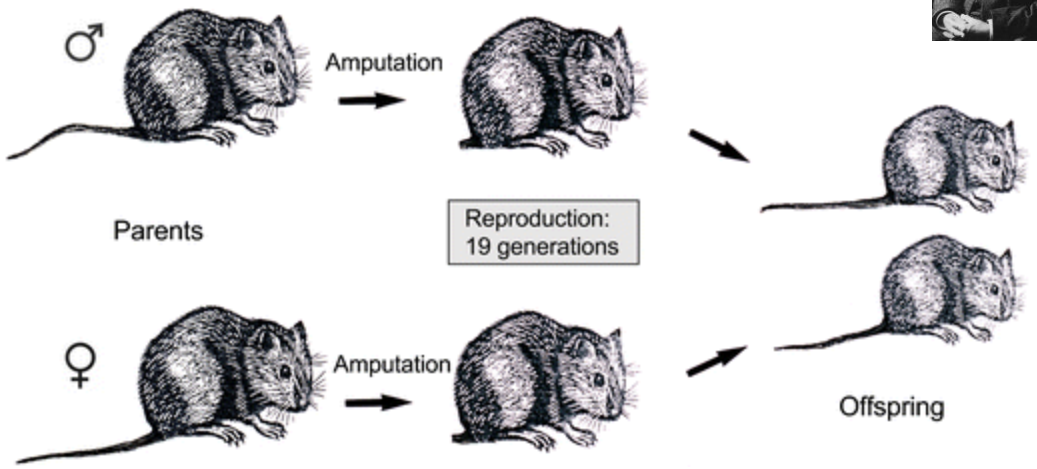
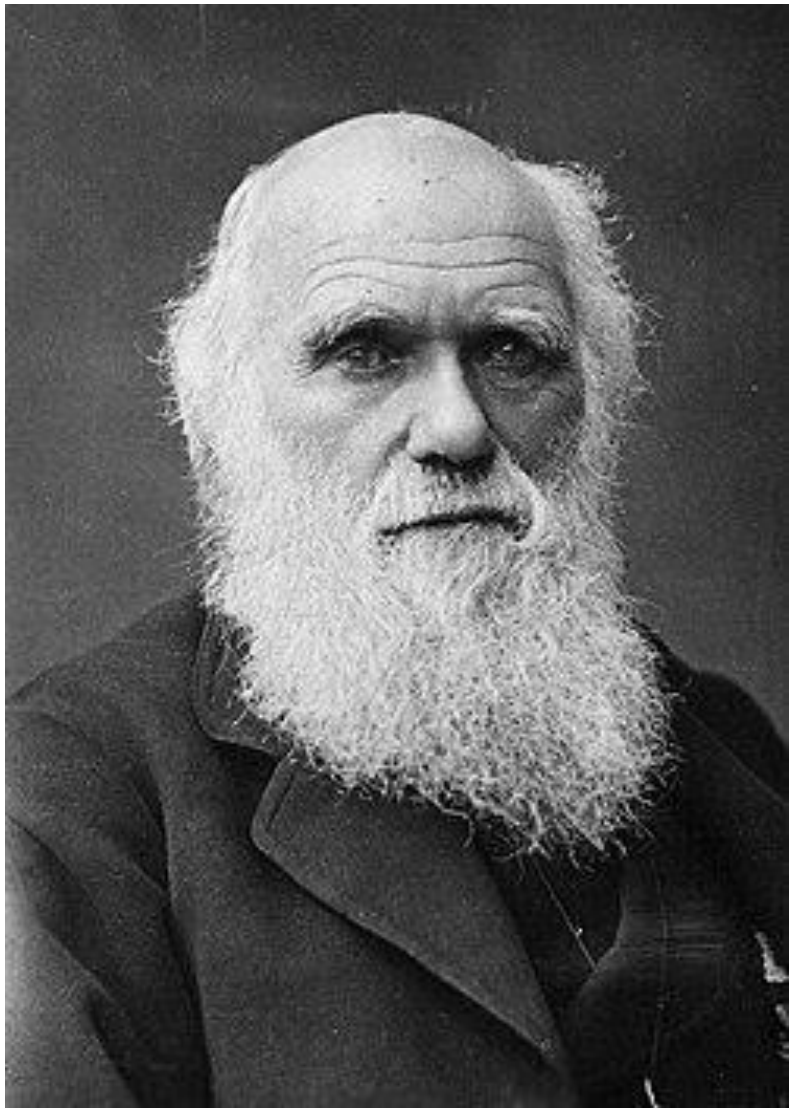
August Weismann



Lamarck would have his revenge although through mechanisms he could never have envisioned…

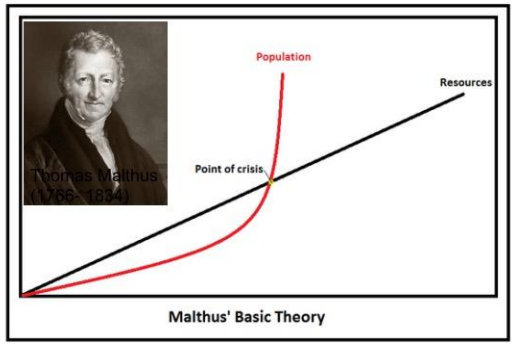
The Evolution of Evolution: Darwin / Wallace



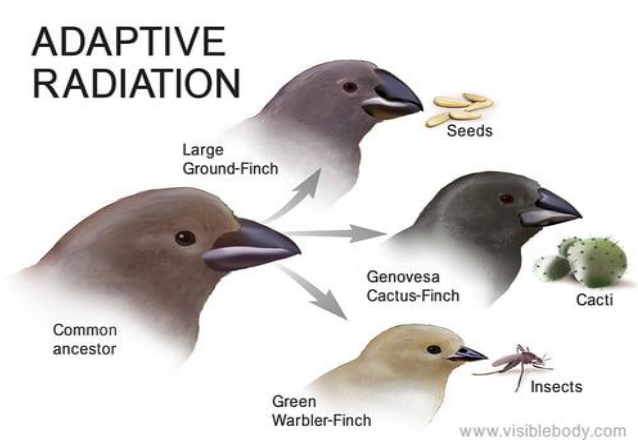

BBC NEWS

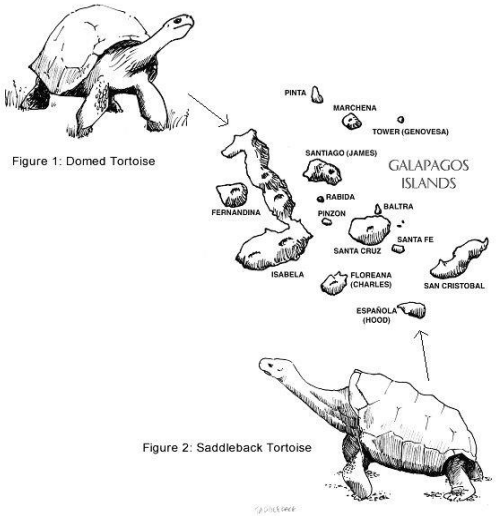
It is possible to look through the history of biology from the ancient Greeks onwards and discover anticipations of almost all of Charles Darwin's key ideas:

SELECTION:



ADAPTATION:





The four postulates of natural selection

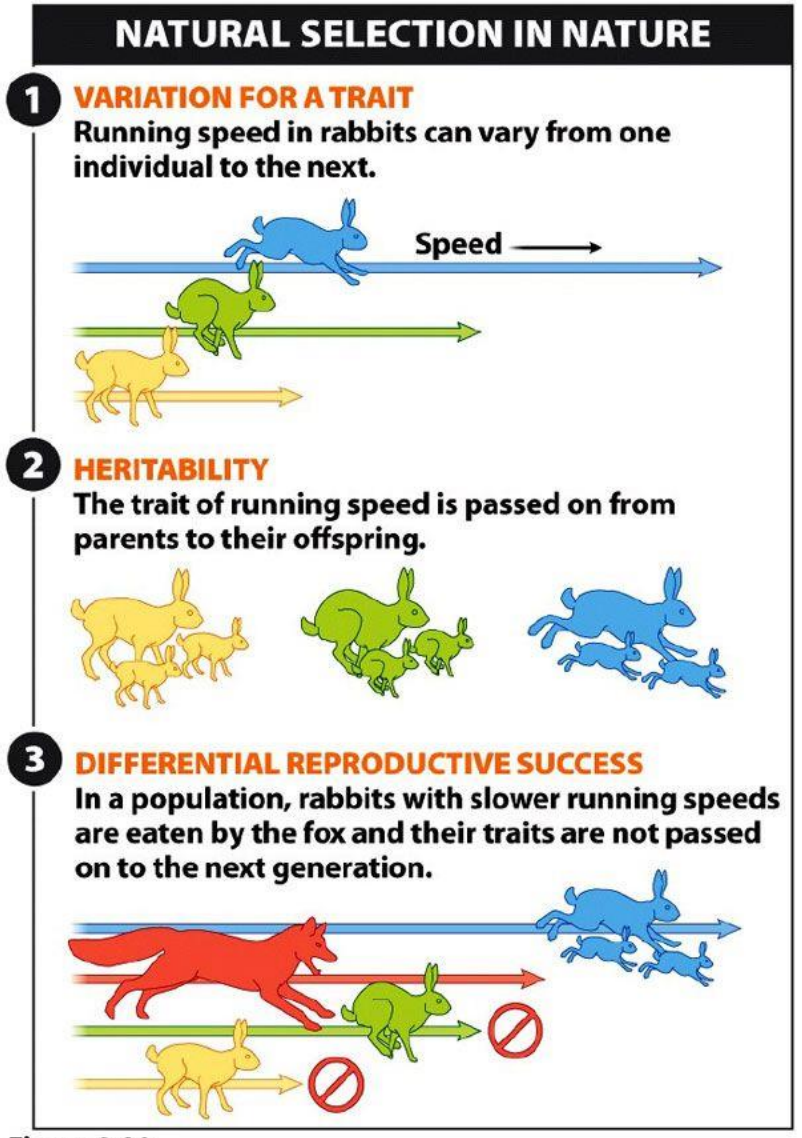
How can evolution through natural selection happen?

(1) individuals vary in traits (=**variation**)

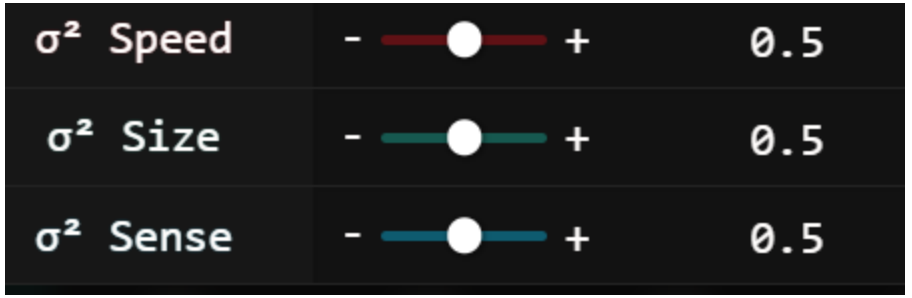
(2) traits are passed down from parents to their offspring (=**heritability**)

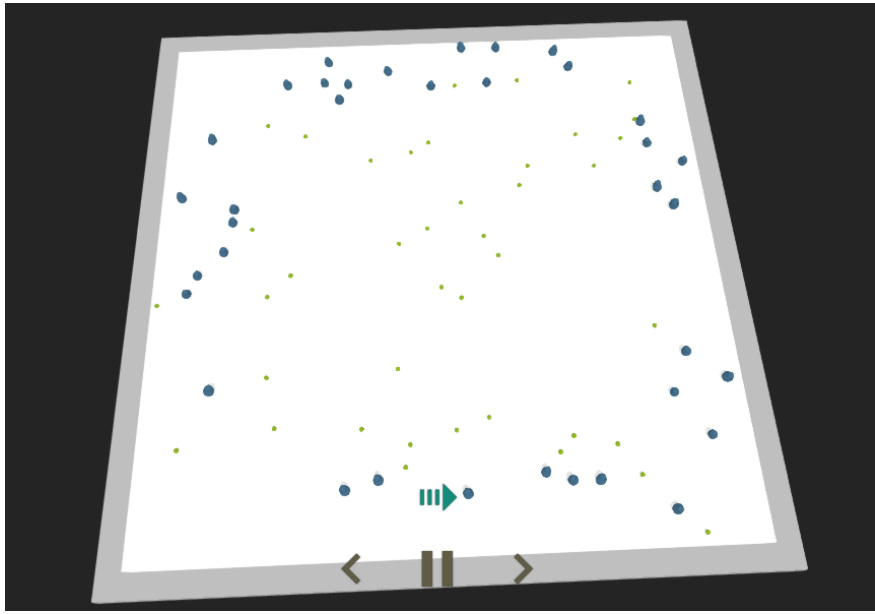
(3) individuals with favorable traits leave more descendants (=**fitness**)

(4) over generations this leads to adaptation and speciation (=**evolution**)



The four postulates of natural selection





$$\text{Energy Lost Per Move} \sim \text{Sight} + (\text{Size})^3(\text{Speed})^2$$

[Evolution Simulator | MinuteLabs.io]

The Evolution of Evolution: Darwin / Wallace

The Evolution of Evolution: Darwin / Wallace

What was still **missing** in this theory?

=> Imagine you are a scientist in 19th century can you come up with counter-arguments?

=> what puzzle pieces are still missing?

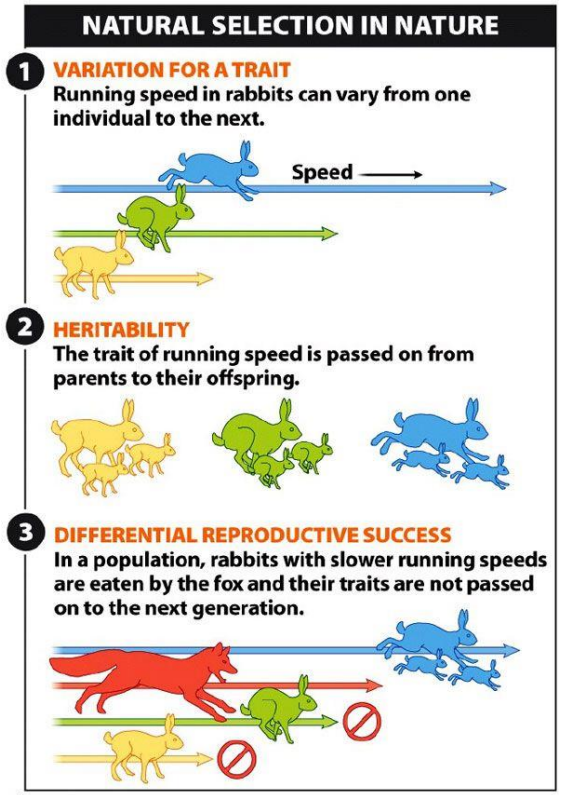


Figure 8-20
What Is Life? A Guide to Biology

The four postulates of natural selection

How can evolution through natural selection happen?

(1) & (2) Lack of a Mechanism for Inheritance

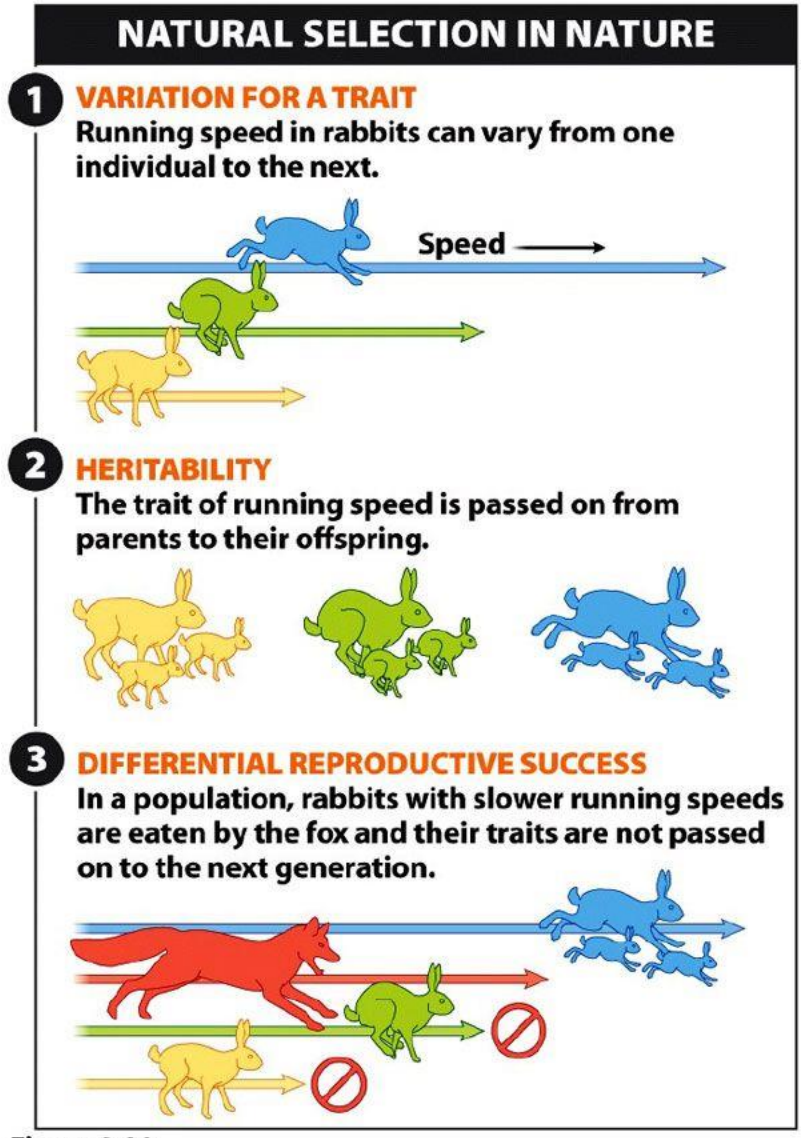
- Darwin didn't know how traits were passed down.
- He proposed "blending inheritance", which actually dilutes variation over generations — contradicting his own theory.

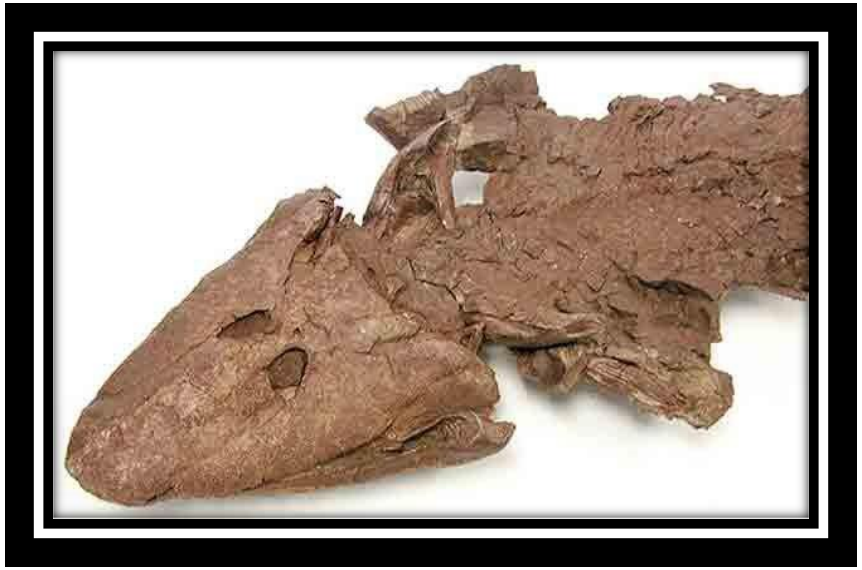
Remember: DNA, genes, etc was not known at the time.

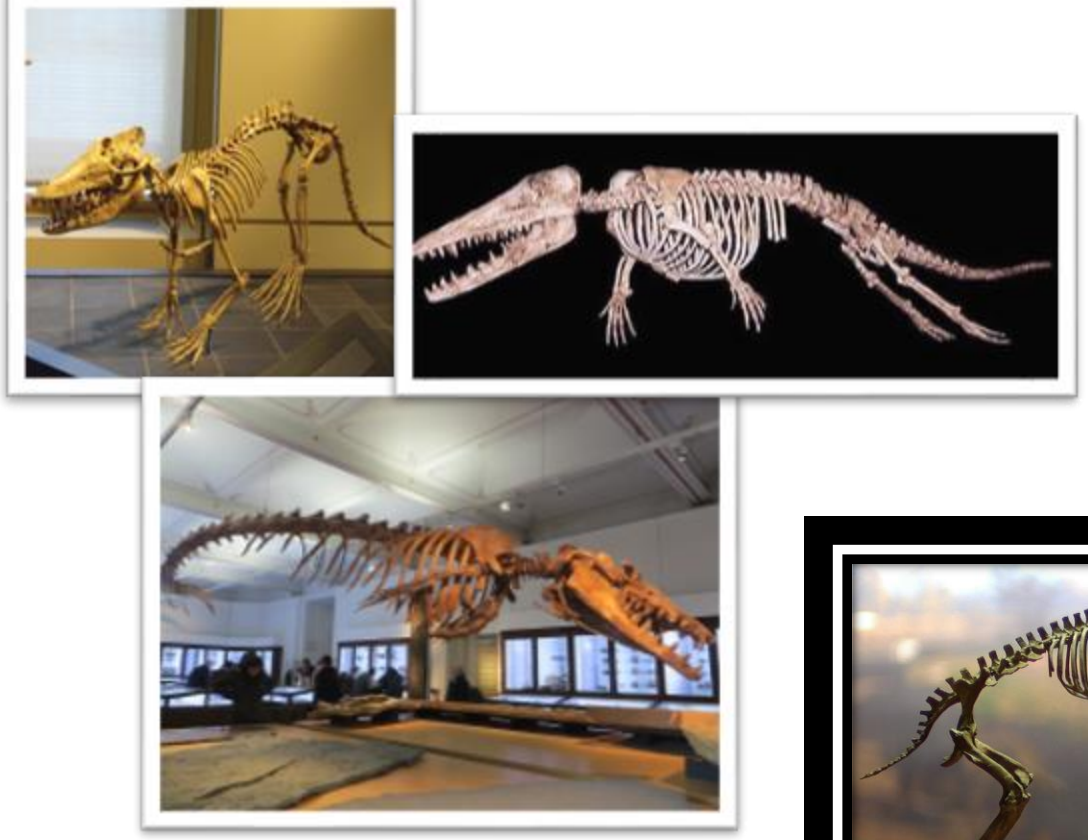
(4) Gaps in the Fossil Record

- Critics said if evolution were gradual, the fossil record should show many transitional forms — but many were missing at the time.
- Geologist Adam Sedgwick and others pointed out the "abrupt appearance" of new forms (e.g., Cambrian explosion**).**

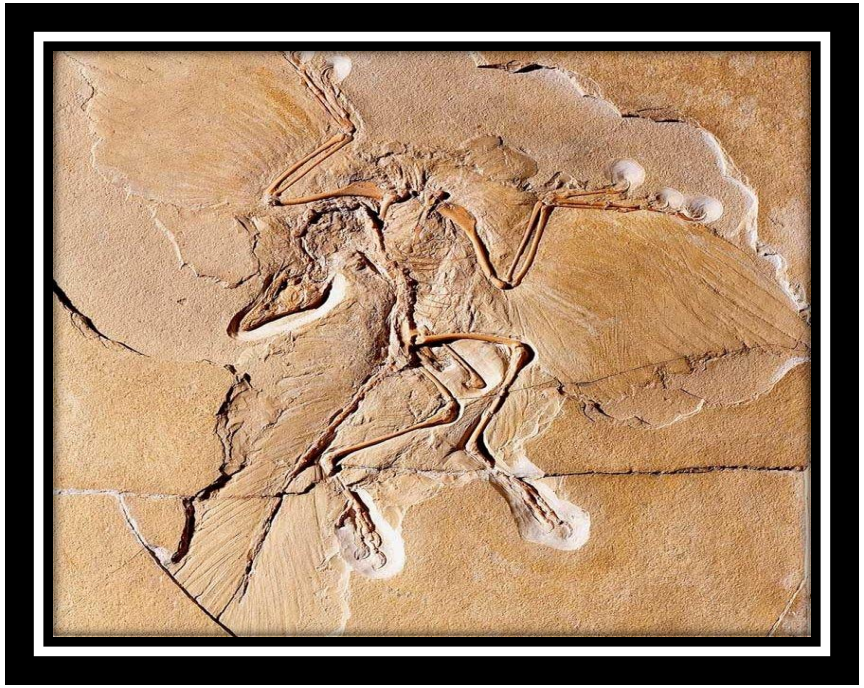
Remember: it's estimated that less than 1% of the current known fossil species were identified at the time.



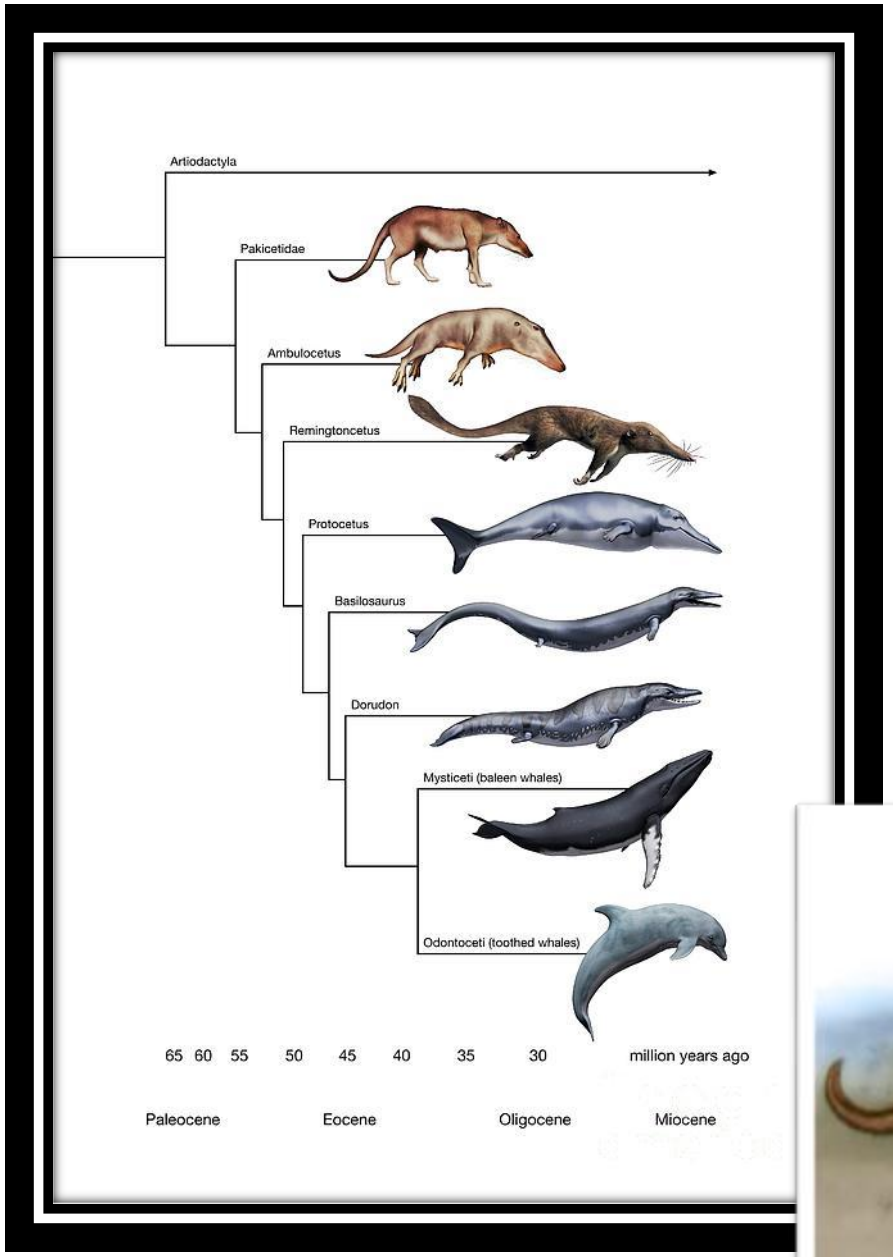


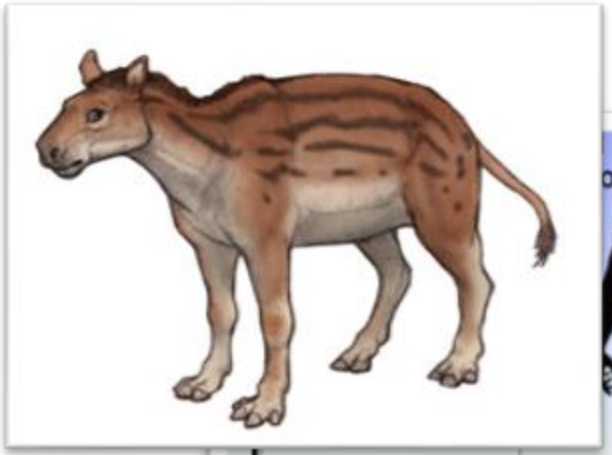


Hyracotherium



Cynodont


Artiodactyla
Pakicetidae
Ambulocetus
Remingtoncetus
Protocetus
Basilosaurus
Dorudon
Mysticeti (baleen whales)
Odontoceti (toothed whales)
65 60 55 50 45 40 35 30 million years ago
Paleocene
Eocene
Oligocene
Miocene



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DIMENSIONS IN METERS
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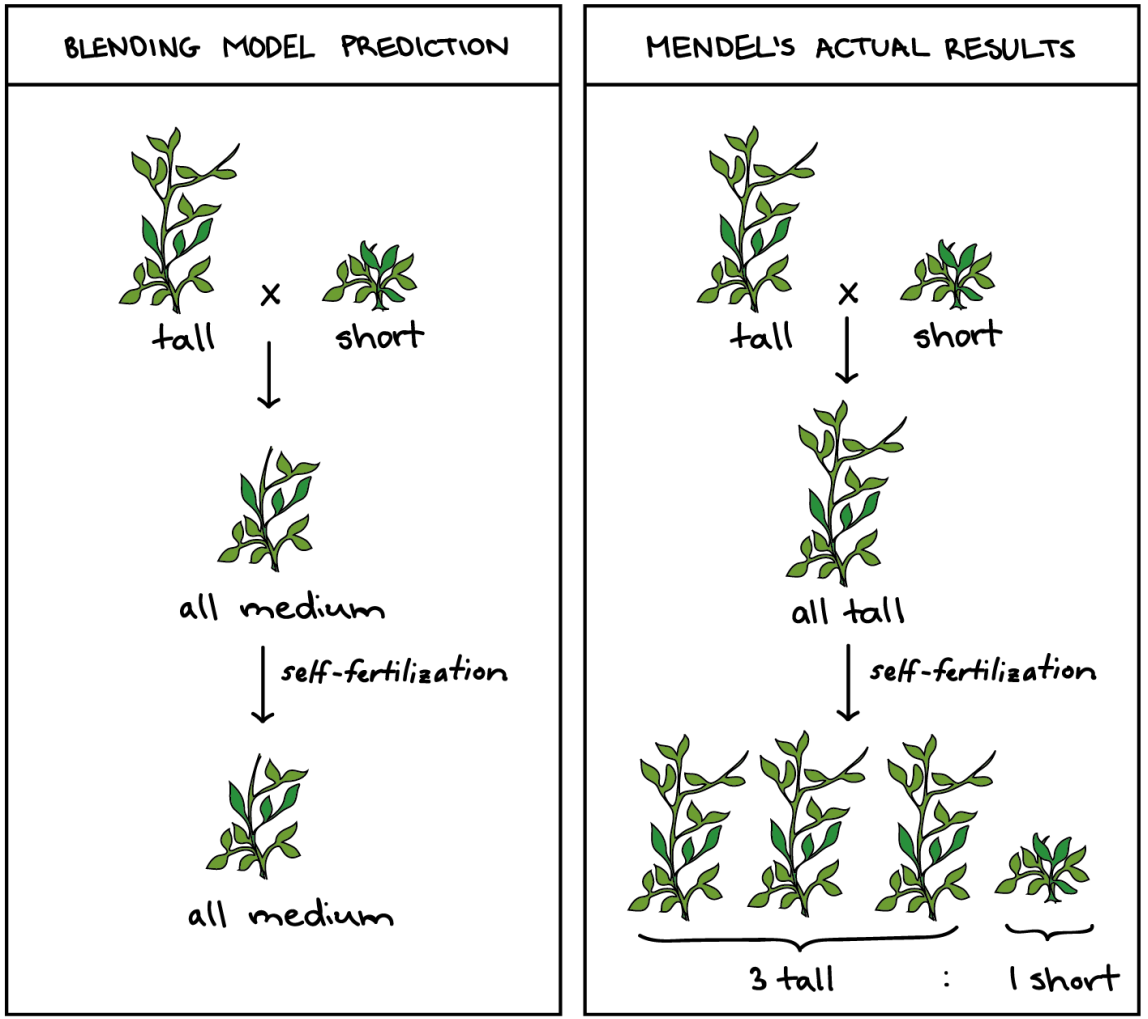
The Evolution of Evolution: Modern Synthesis

3:1

The Evolution of Evolution: Modern Synthesis

Mendel



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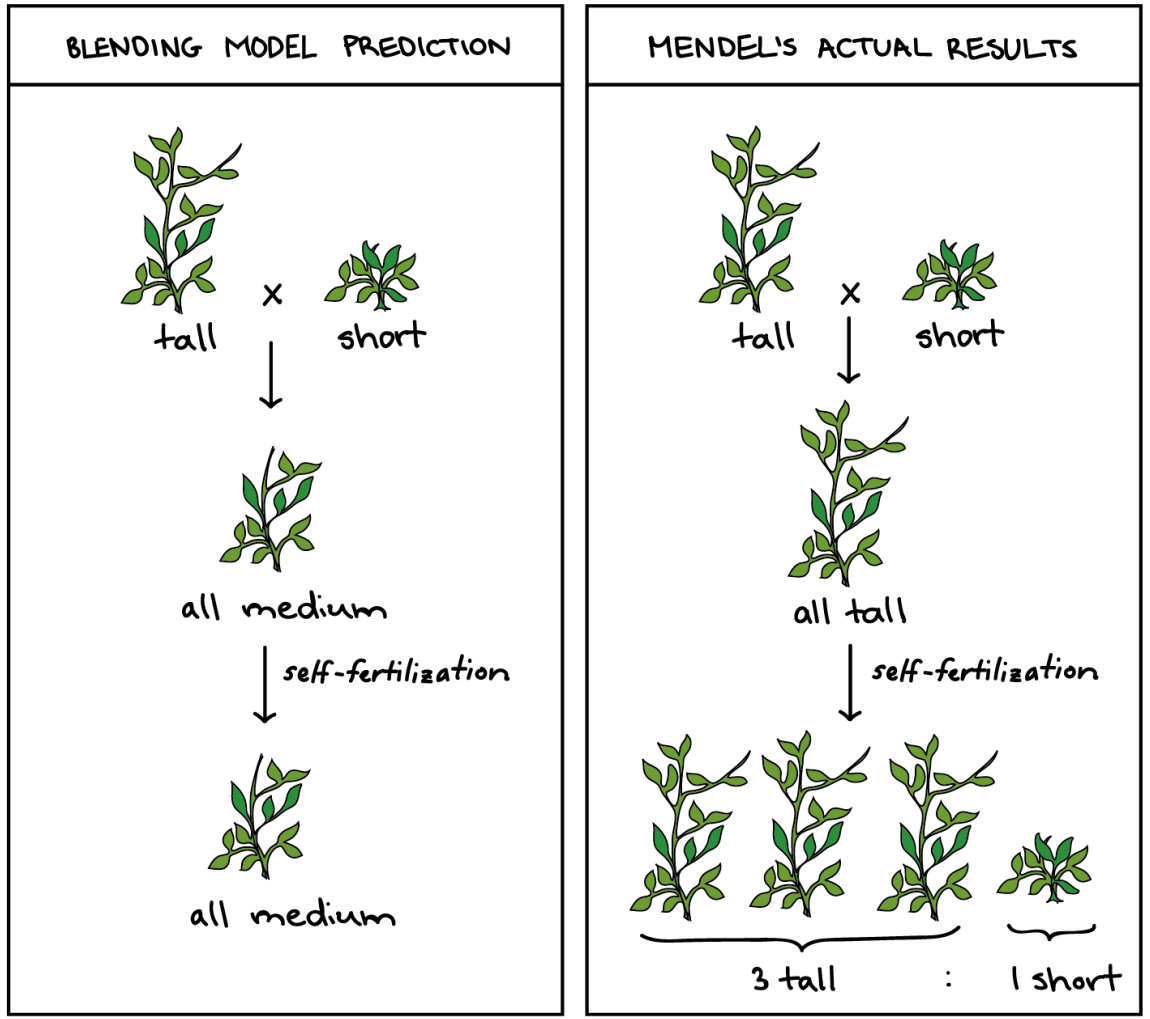
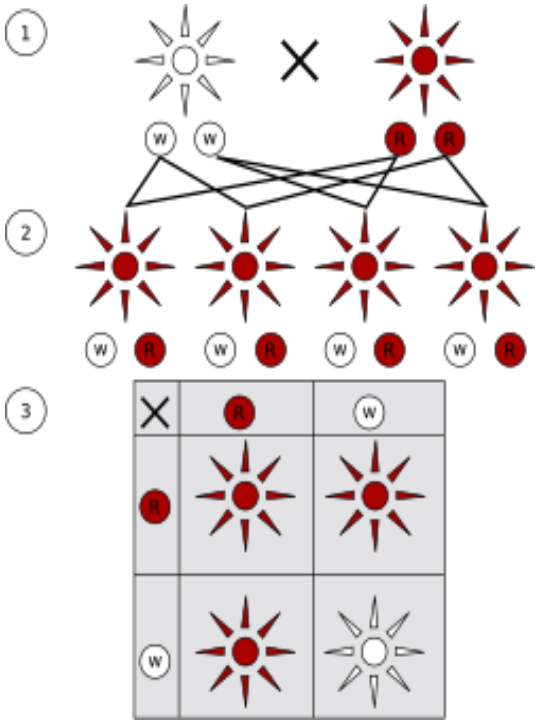
The Evolution of Evolution: Modern Synthesis

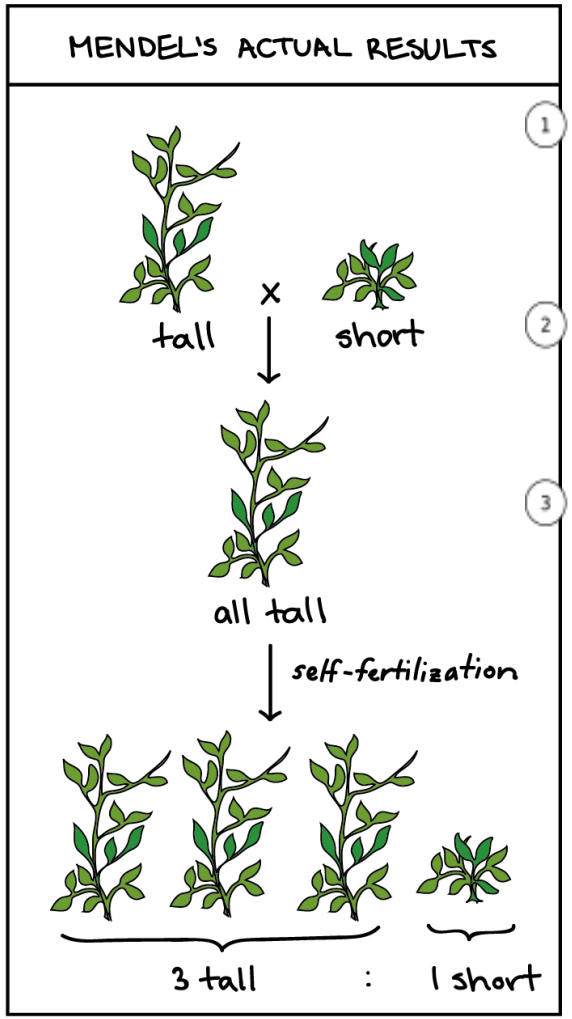
Mendel

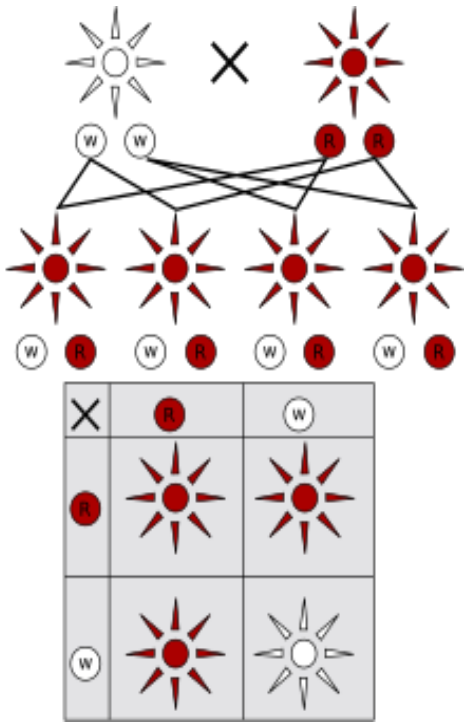


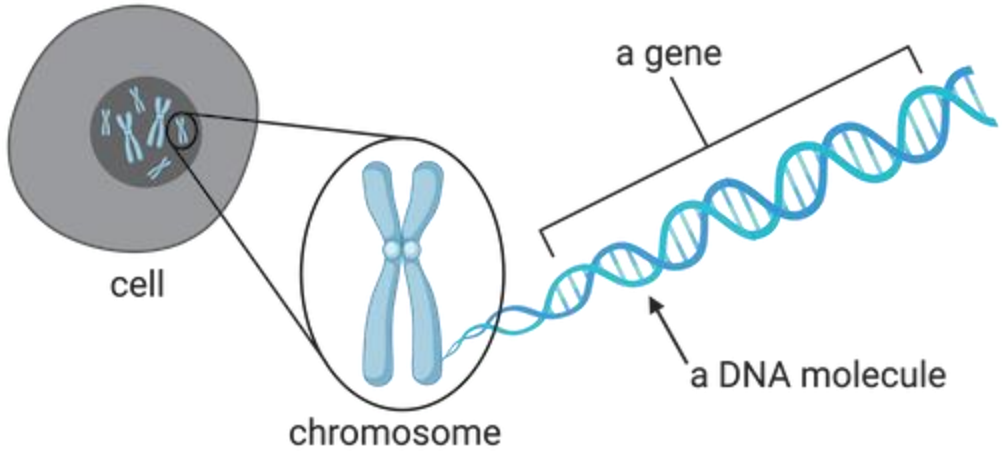


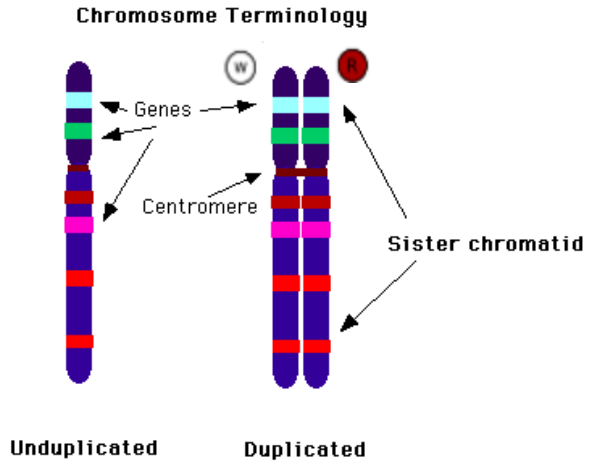
The Evolution of Evolution: Modern Synthesis

Mendel



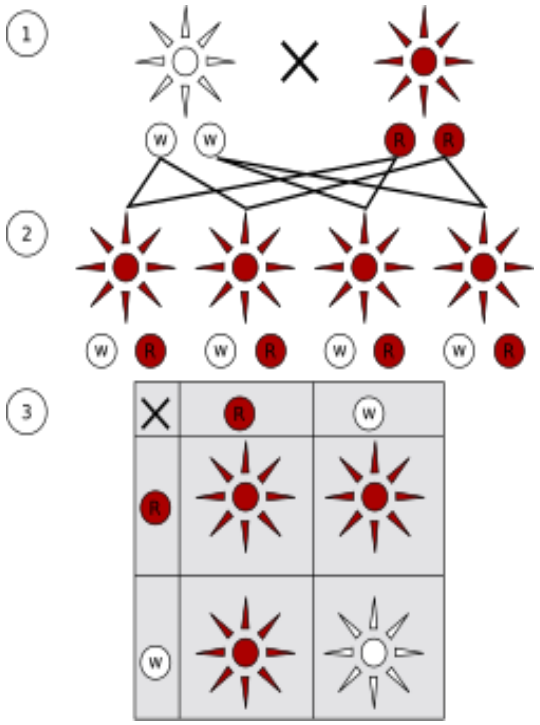






The Evolution of Evolution: Modern Synthesis

Mendel



Mendel's laws:

1. Law of Segregation

- **Each individual has two alleles** (gene copies) for each trait, one from each parent.
- These alleles **segregate (separate)** during gamete (sperm or egg) formation.
- Each gamete gets **only one allele**.
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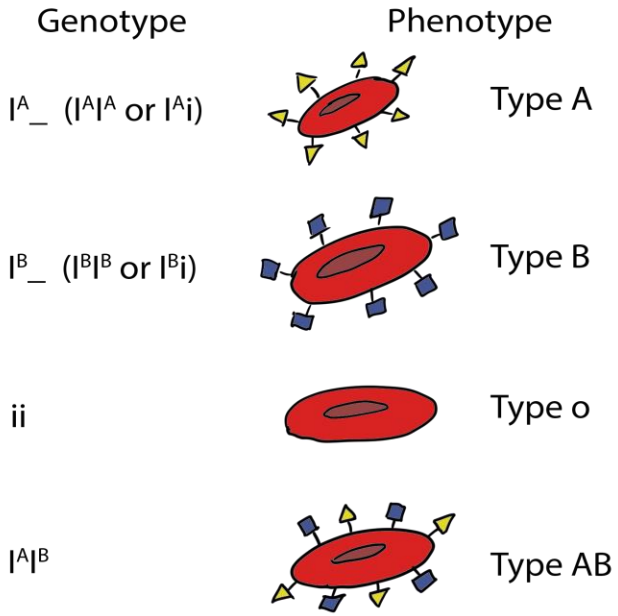
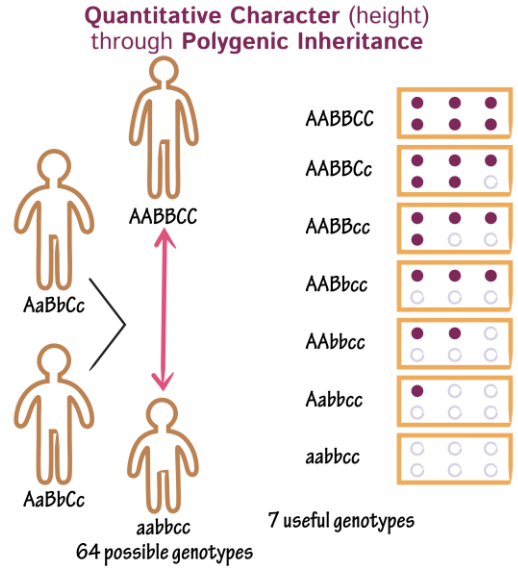
BIOLOGY MEANS EXCEPTIONS

Incomplete dominance



Co-dominance



Polygenic traits

And many many many more….

The Evolution of Evolution: Modern Synthesis

Thomas
Hunt Morgan

Where does variation come from then and where is it located?

The Evolution of Evolution: Modern Synthesis

Where does variation come from then and where is it located?

Thomas
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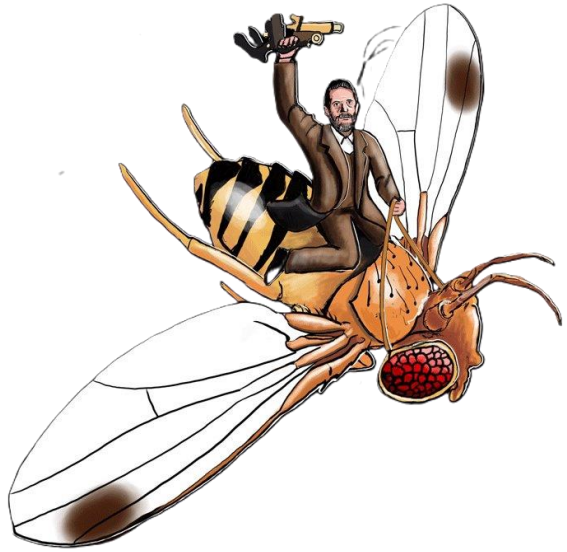

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Cross	Outcome	
	Expected Phenotypes	Observed Phenotypes
P_1 Red ♀ × P_1 White ♂	F_1 = All Red	F_1 = All Red*
F_1 Red ♀ × F_1 Red ♂	75% Red ♀ and ♂ 25% White ♀ and ♂	50% Red ♀ 25% Red ♂ 25% White ♂

The Evolution of Evolution: Modern Synthesis

Thomas
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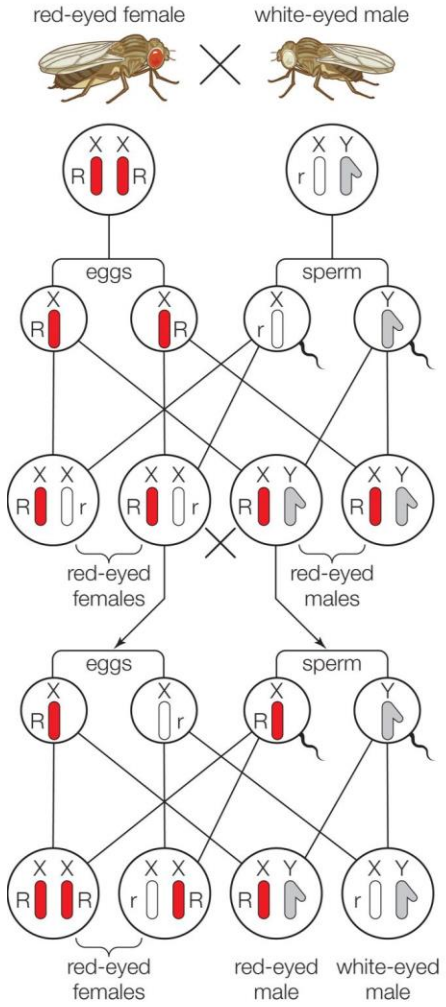

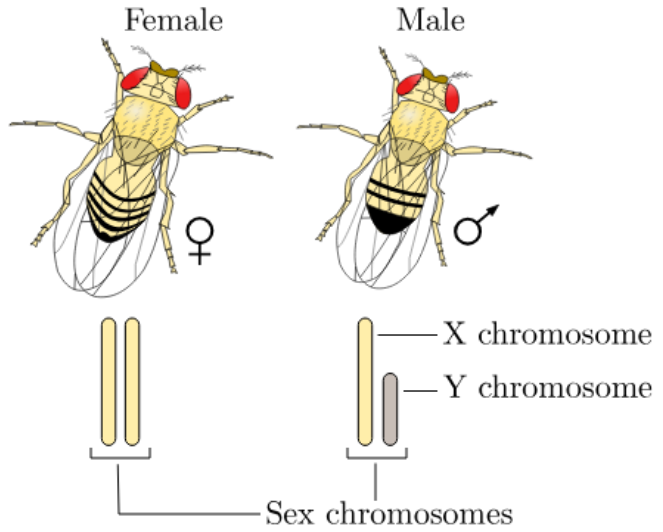

The Evolution of Evolution: Modern Synthesis

Thomas Hunt Morgan

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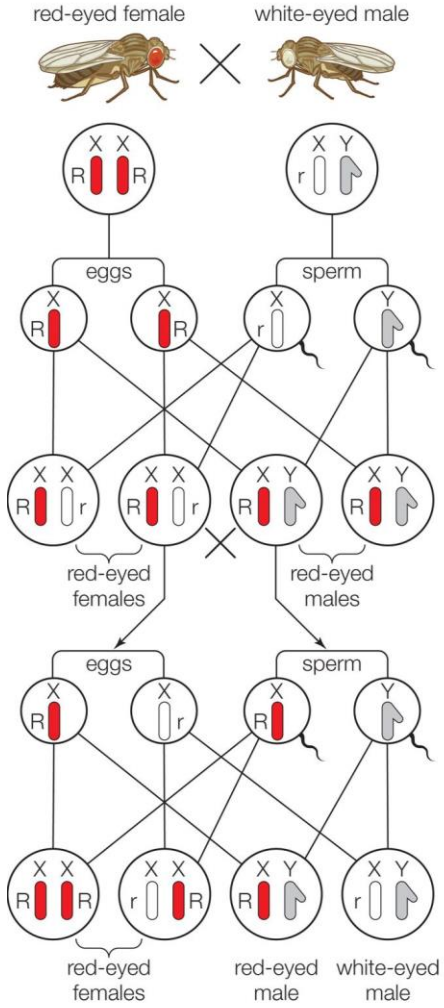
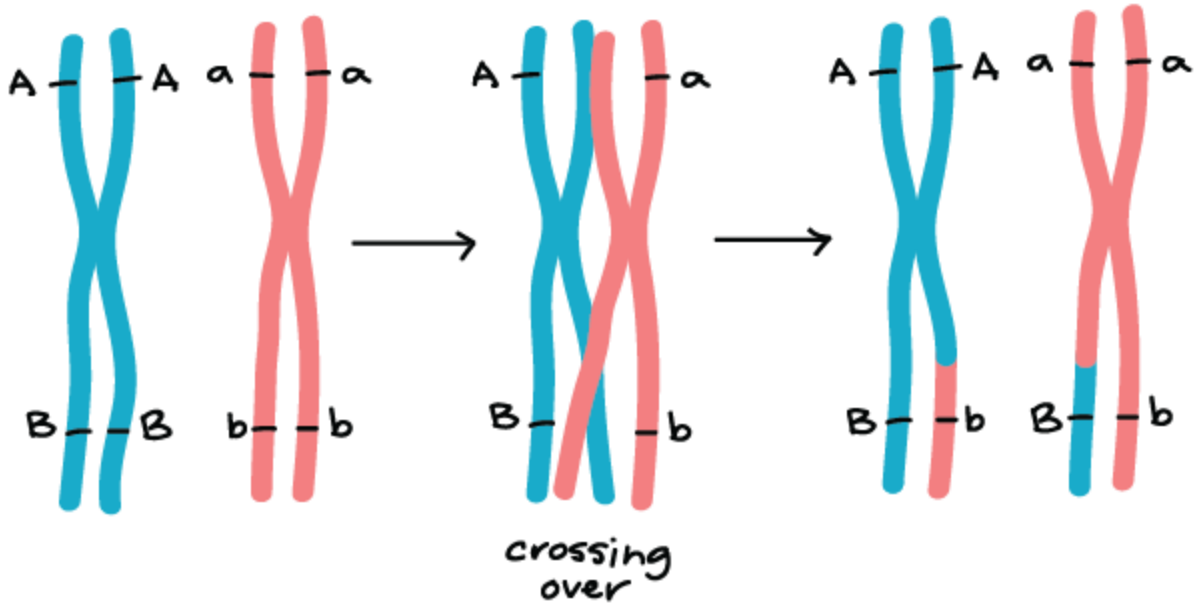


- **Mutations** in specific genes can cause variation in a trait
  - First source of variation that was discovered
- **Genes Are on Chromosomes**
  - Before Morgan, Mendel's "factors" (genes) were abstract.
  - Morgan showed that genes are physically located on chromosomes, using inheritance patterns in fruit flies.
- **Sex-Linked Inheritance**
  - Morgan’s white-eye mutation was the first documented sex-linked trait.
  - The gene for eye color was found to reside on the X chromosome.

The Evolution of Evolution: Modern Synthesis

Thomas
Hunt Morgan

Another exception to Mendelian genetics:



The Evolution of Evolution: Modern Synthesis

Thomas
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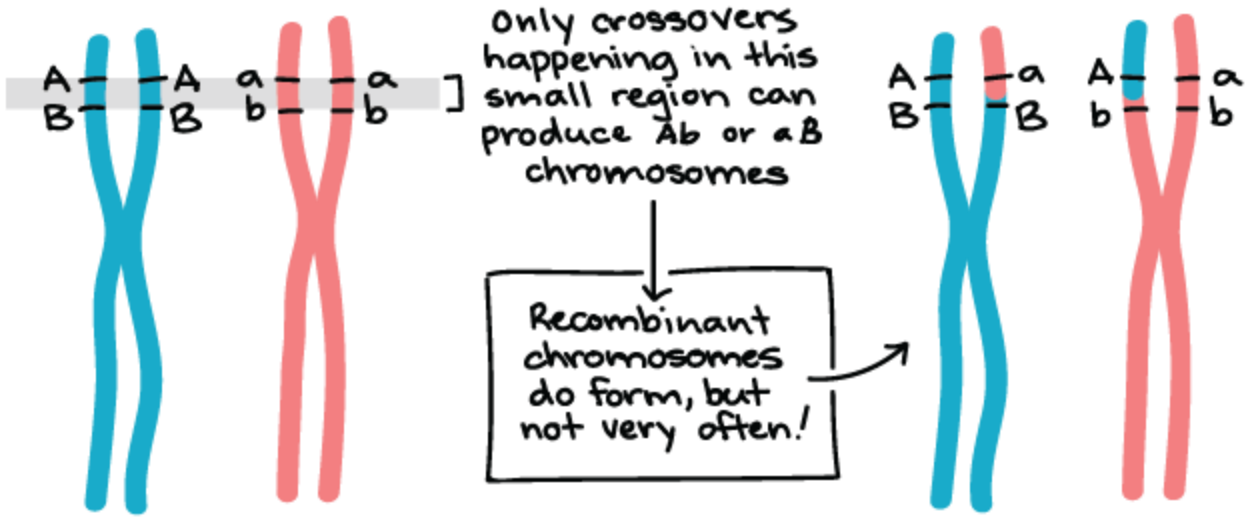

The Evolution of Evolution: Modern Synthesis

Thomas
Hunt Morgan

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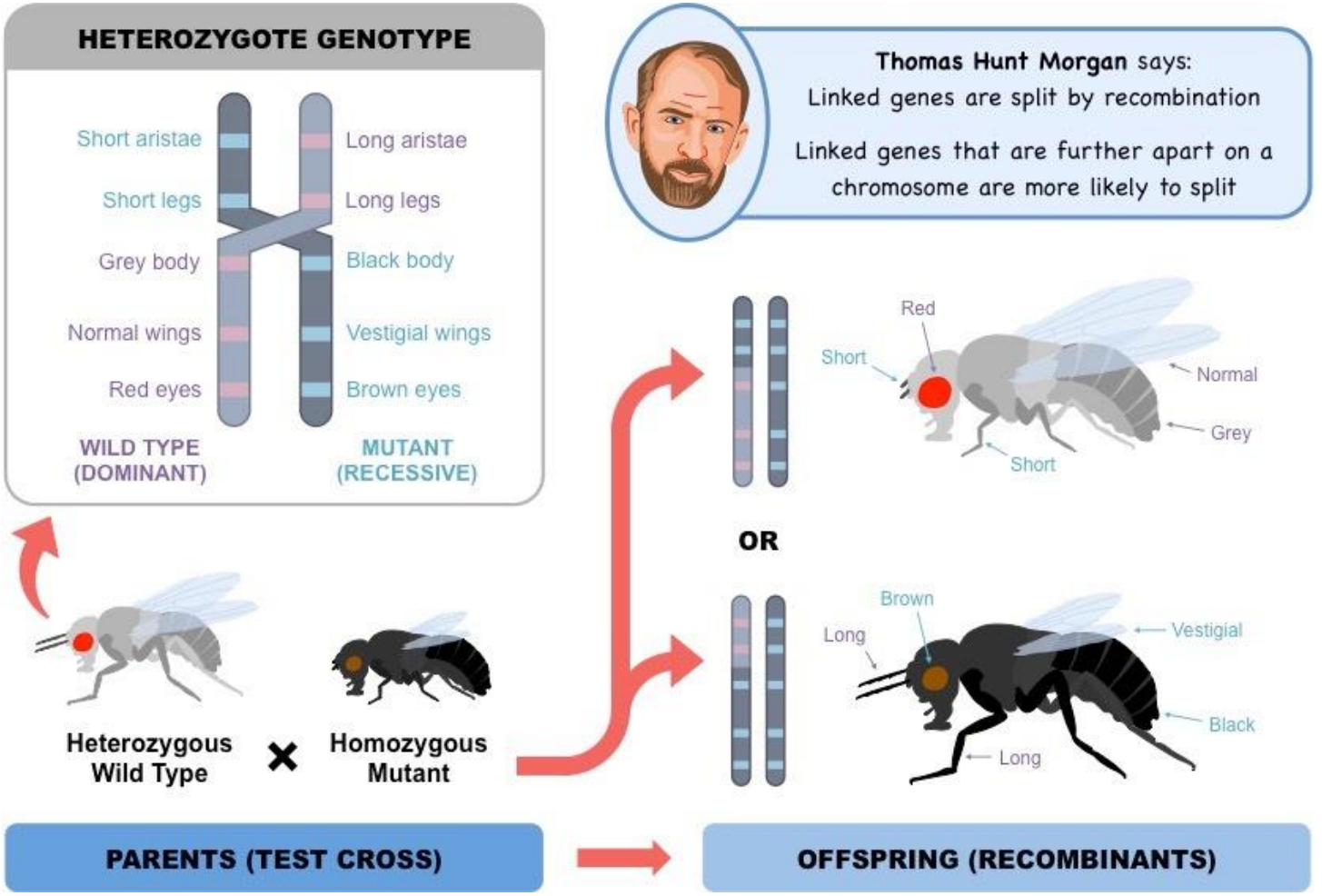

The Evolution of Evolution: Modern Synthesis

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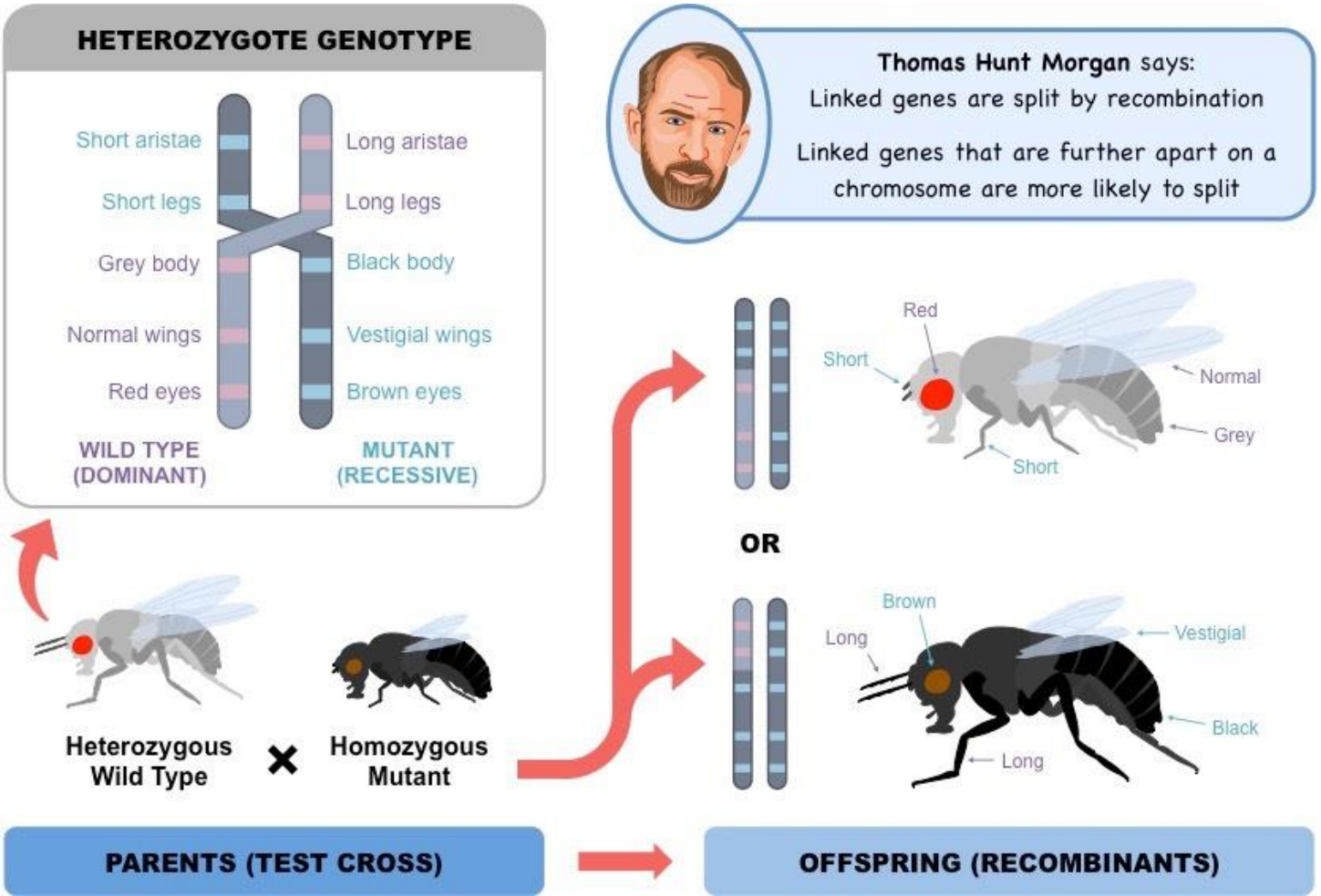

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The Evolution of Evolution: Modern Synthesis

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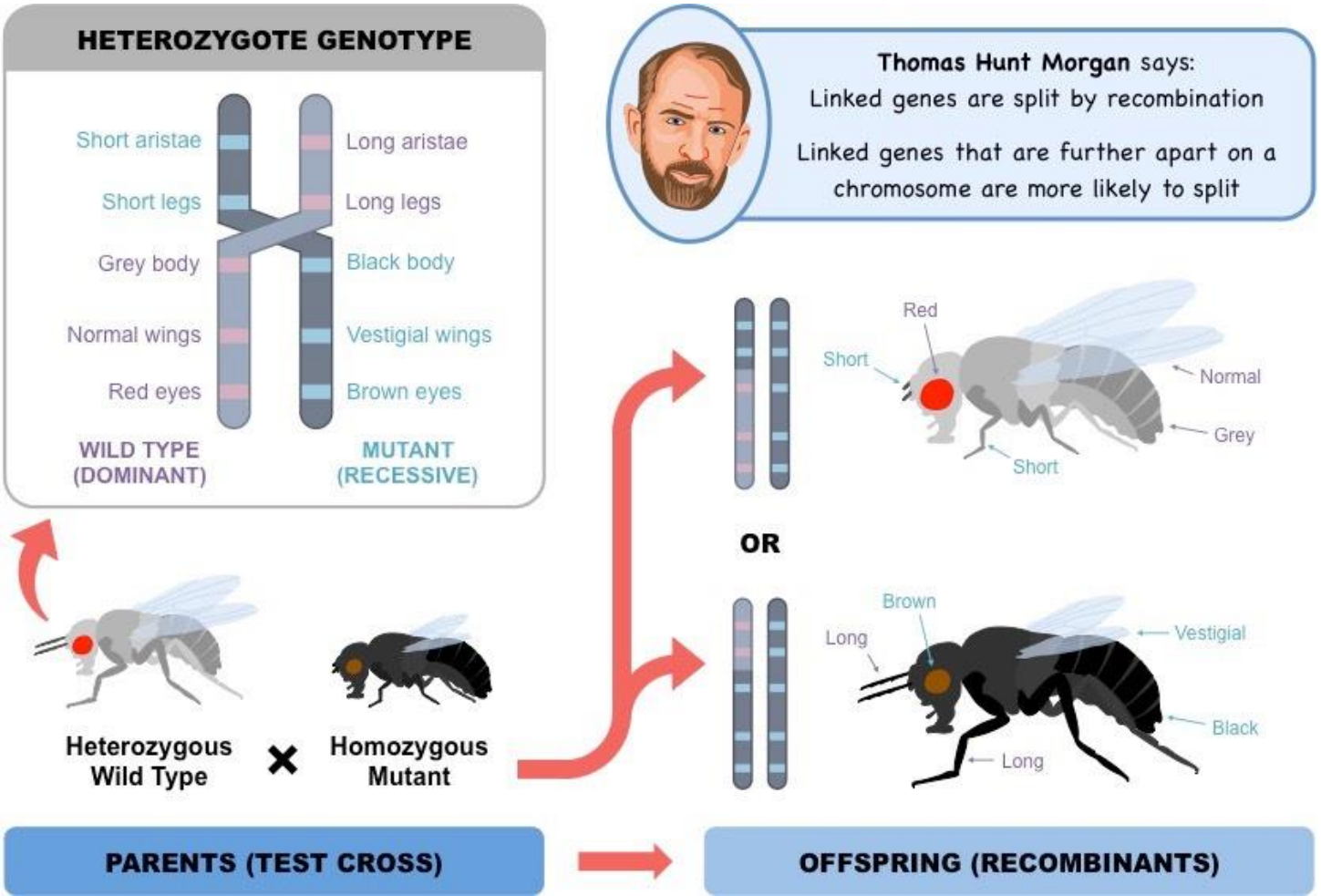

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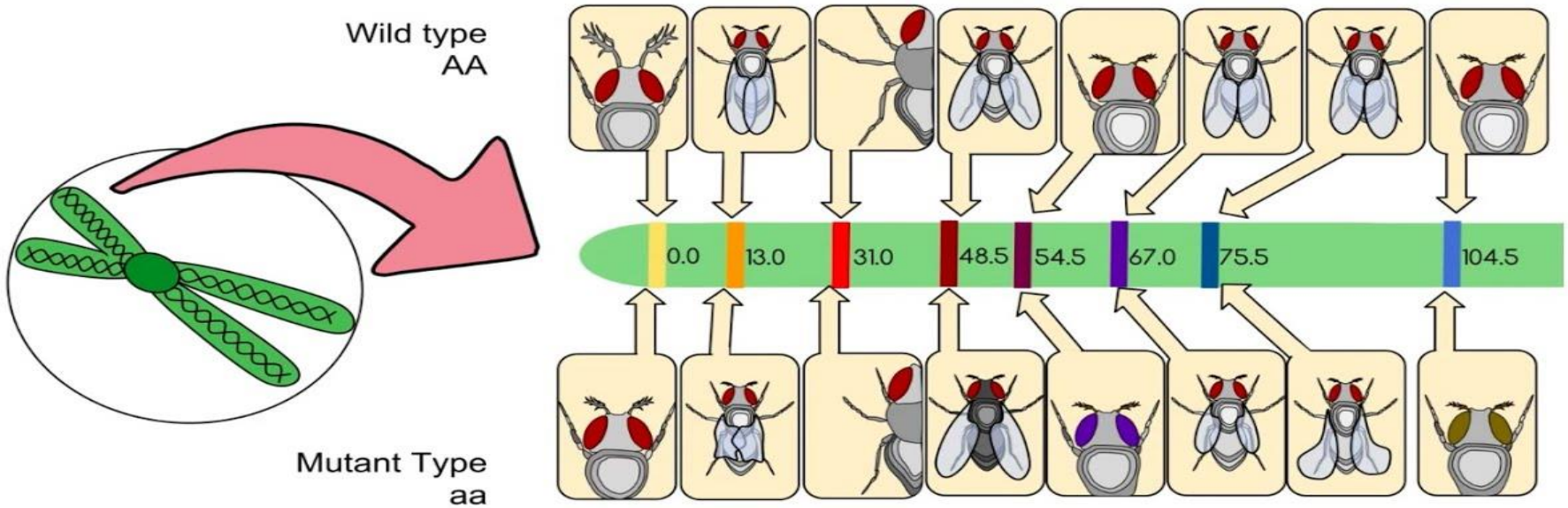
- **Genetic Linkage:**
  - Occurs when **genes** are located **close together** on the same chromosome.
  - These genes are likely to be **inherited together**, because they do not assort independently.
  - So: Linkage violates the assumption of independent assortment.

The Evolution of Evolution: Modern Synthesis

The first genetic map!

Sturtevant and his colleagues were able to map many of the fruit fly genes in this way



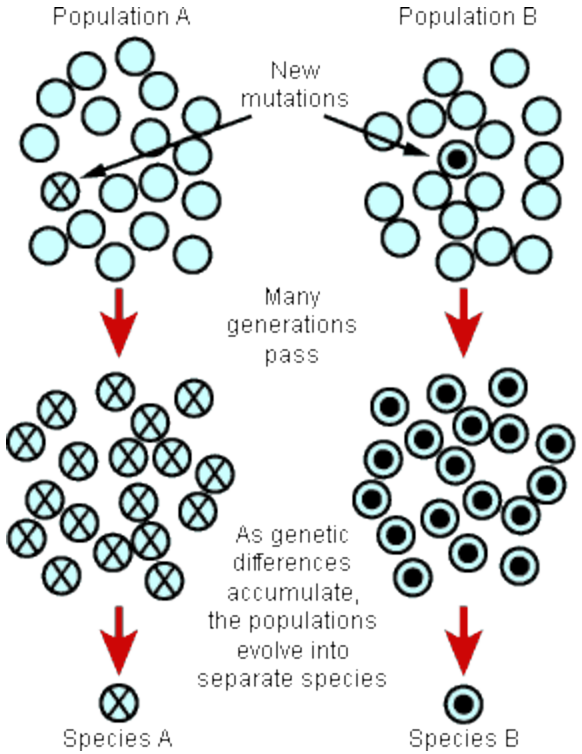
The Evolution of Evolution: Modern Synthesis

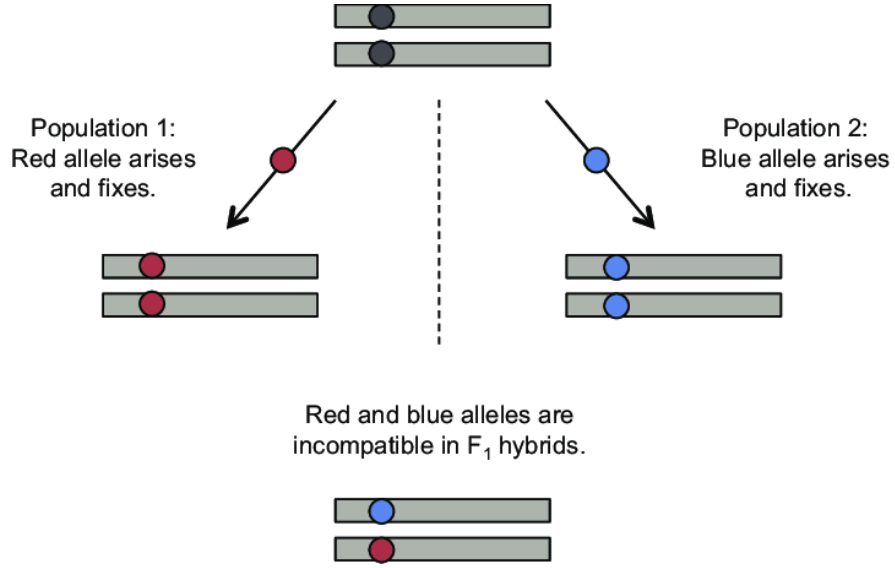
Dobzhansky, student of Morgan

First population genetic assay!

At the time, most biologists assumed that all of the members of any given species had practically identical genes. How do we then define a species?

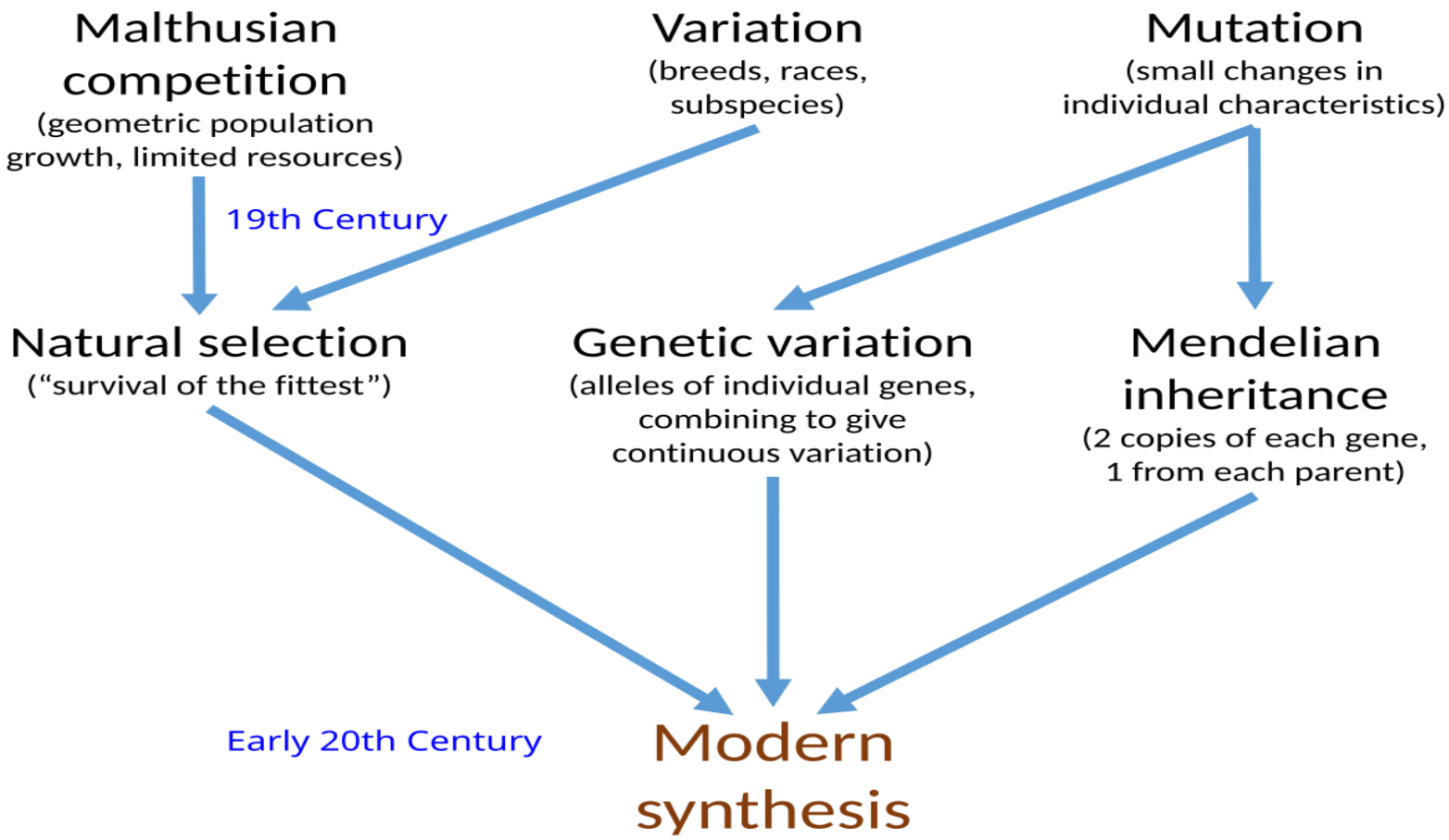
Genetics and the Origin of Species





The Evolution of Evolution: Modern Synthesis

The basics…



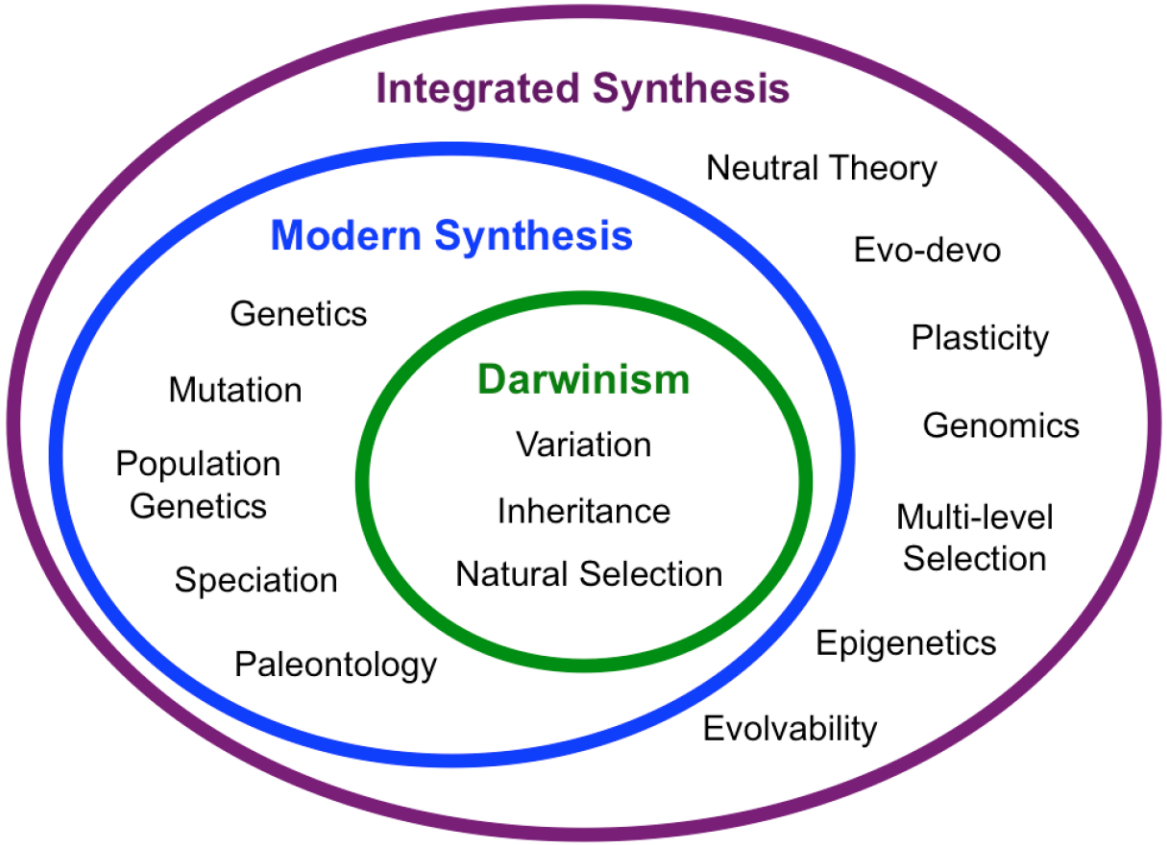
The Evolution of Evolution: Modern Synthesis, The Extended Version

Modern synthesis focuses on genes:

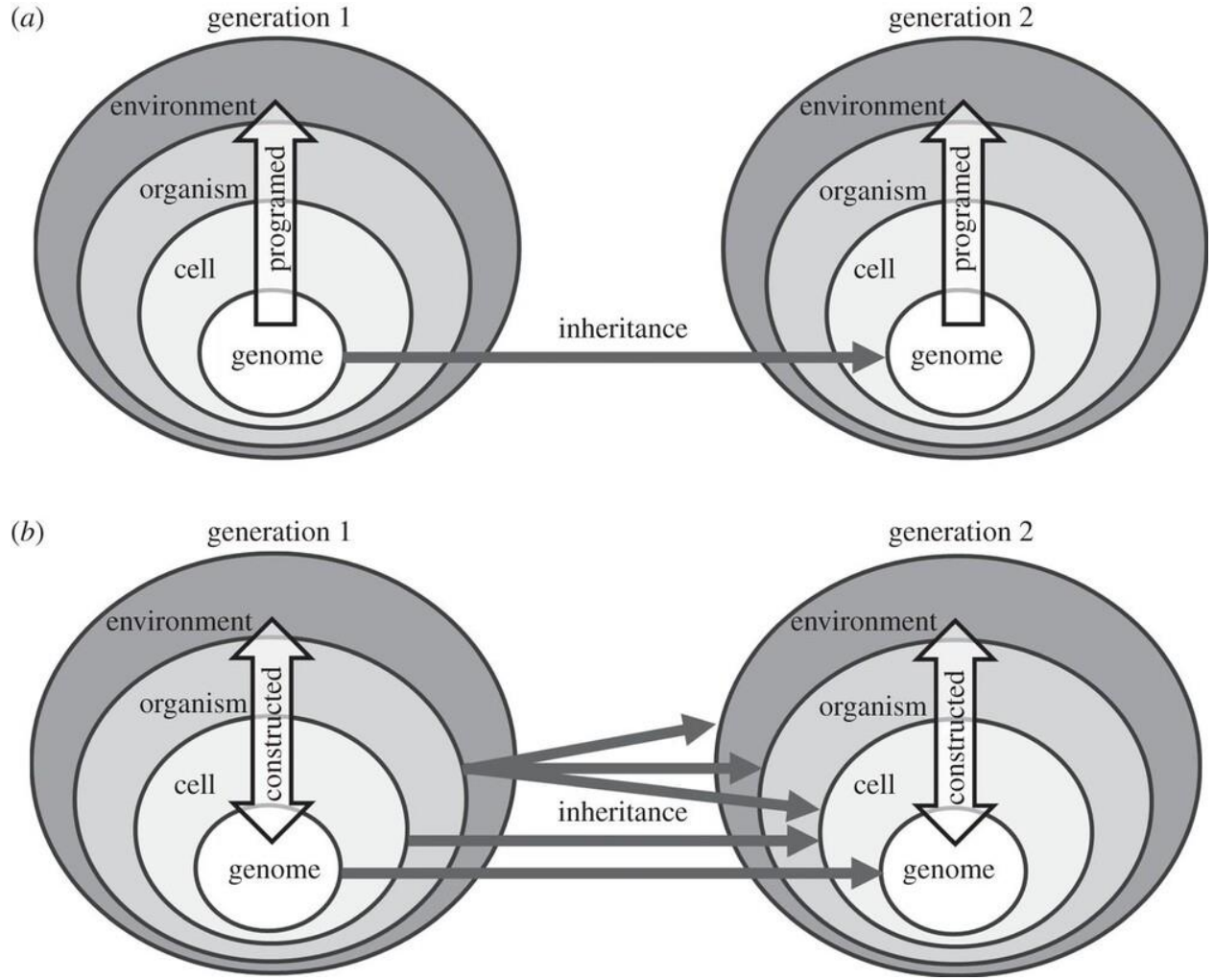
- new variation arises through random genetic mutation
- inheritance occurs through DNA
- natural selection of genes is the sole cause of adaptation

The field of evolutionary biology has evolved, incorporating many new theoretical and empirical findings ➔ extended version

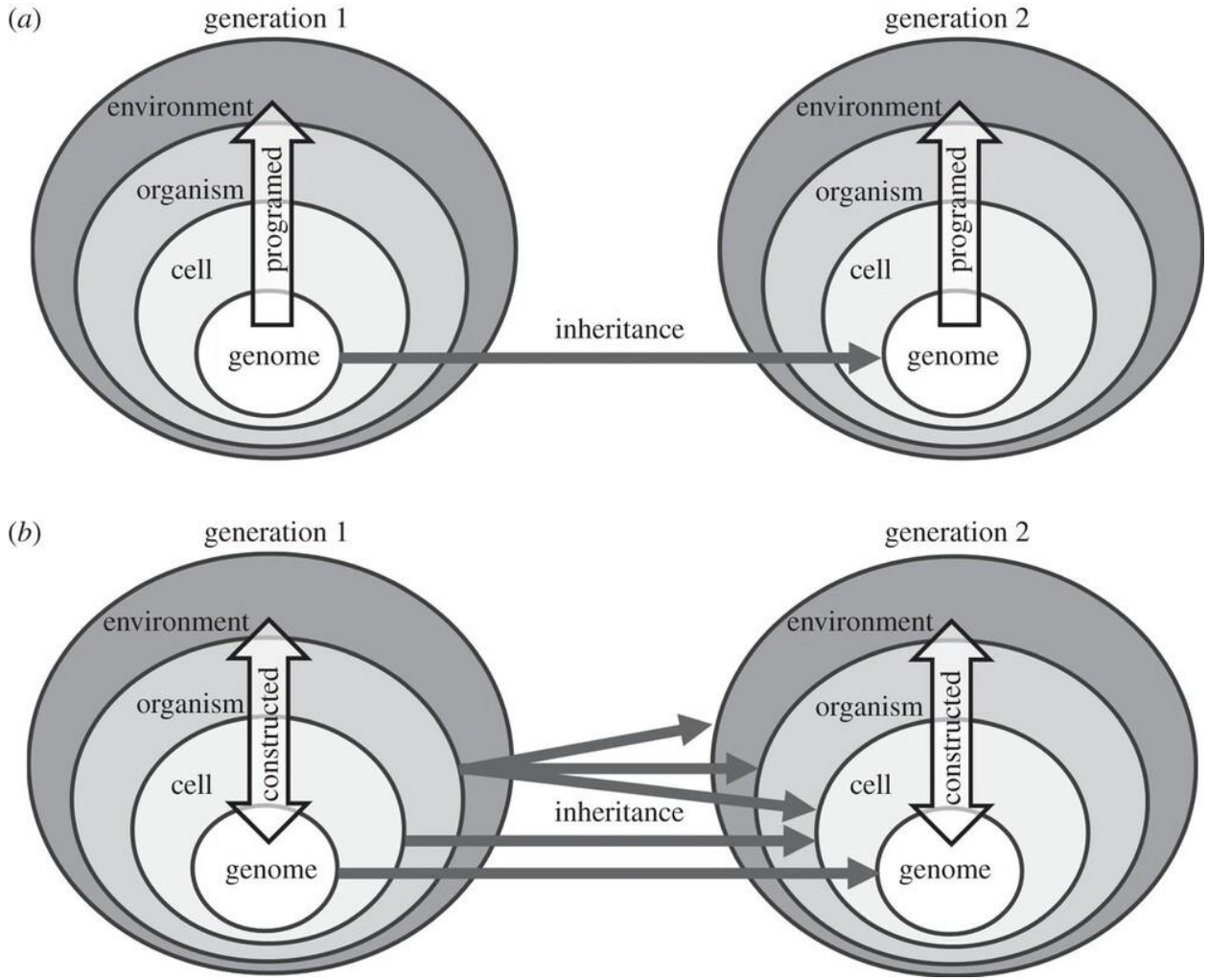
The Evolution of Evolution: Modern Synthesis, The Extended Version

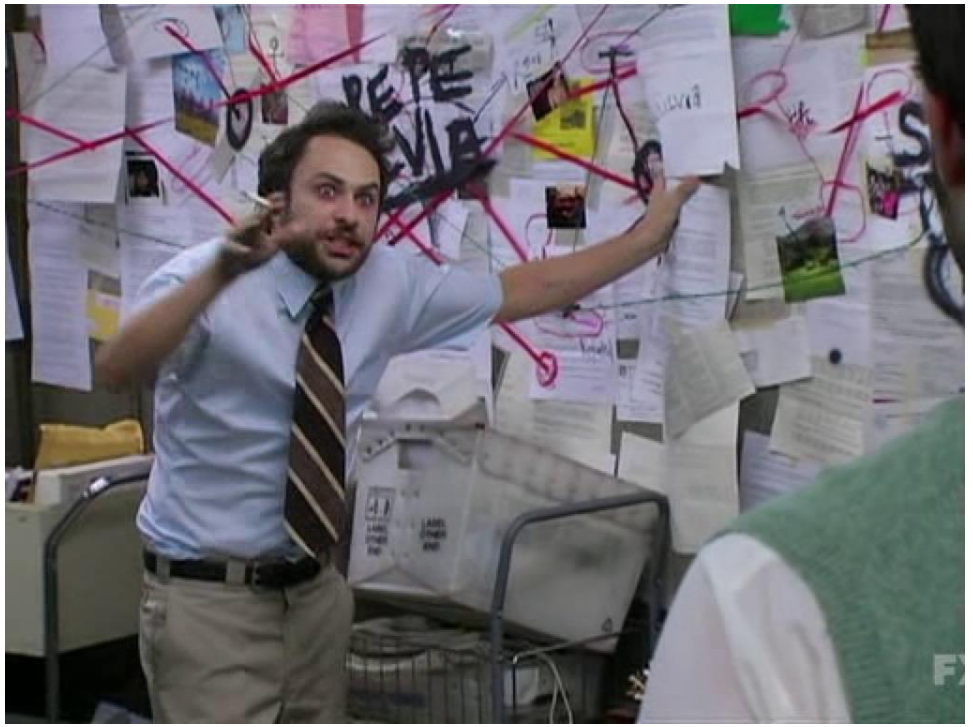


The Evolution of Evolution: Modern Synthesis, The Extended Version



The Evolution of Evolution: Modern Synthesis, The Extended Version





Module 2: Central Dogma: DNA → RNA → Protein

Chapter 15 : DNA & RNA

RNA
- phosphate - sugar (ribose) - base [Adenine, Uracil / Guanine, Cytosine]
- sequence of bases in RNA is determined by the sequence of bases in DNA.
- can move into cytoplasm // DNA = nucleus.

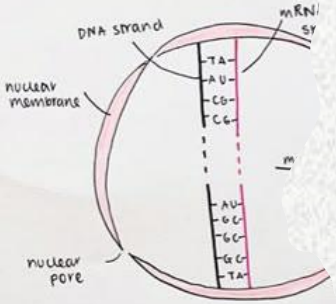
RNA
- ribose
- AU CG
- single strand

PROTEIN

Initiation

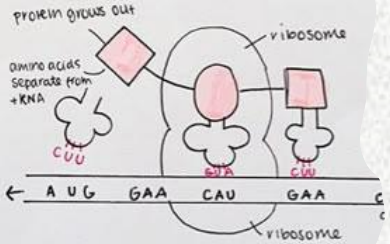
1. DNA double helix unwinds at the site of the gene that c

Transcription



2. RNA bases move across nuclear membrane from cytoplasm
3. RNA polymerase binds RNA base together to form mRNA.
   each mRNA has complementary bases to the DNA from which it was transcribed. 3 bases = codon
   Start, stop & amino acid producing
4. every gene has 1 start, lots ↑, 1 stop.

Translation



5. mRNA moves into cytoplasm → into ribosome & binds with rRNA
6. cytoplasm contains tRNA molecules that carry: an anticodon & amino acid.
7. tRNA molecules are attracted to the mRNA. each anticodon on tRNA is complementary to a codon on the mRNA.
8. 1st tRNA will be attracted to mRNA after start codon & brings in an amino acid.
9. amino acids detach from tRNA & bind together to form a polypeptide c
10. tRNA molecules leave ribosome without amino acids & pull mRNA stran
11. tRNA continues to bind w mRNA until stop codon @ - mRNA sequence is co
12. Protein now folds into correct shape.

activity is: To Isolate DNA From Plant Tissue

1. salt + washing up liquid → water
2. cut onion into small cubes
3. onion → beaker w salt + detergent
4. beaker → water bath @ 60° for 15m.
5. beaker → ice bath for 5m.
6. blend for 3s on high speed.
7. filter → coffee filter paper
8. syringe → filtrate → boiling tube.
9. add protease enzyme & mix
10. ice cold ethanol + side of boiling tube → form layer.
11. small glass rod → twist, strands of DNA should attach

A REPLICATION

...ucture is a double helix or ...ted ladder.

...quence of DNA bases ...de for specific proteins

...eins are key for cell ...nction and life

...ication means "a copy"

It occurs during S phase of mitosis cell cycle ← parent cell

D ← Daughter cells

A - phosphates
B - 5 carbon Sugar
C - Nitrogenous bases
D - Weak (H) bond

Step 1 Enzyme helicase - unwinds and unzips DNA by breaking the weak hydrogen bond.

Step 2 Enzyme DNA Polymerase ...matches the complementary base

Step 3 Enzyme DNA ligase repairs the sugar phosphate back bone.

PROTEIN SYNTHESIS

TRANSCRI

Produces an RNA c

mRNA - messenger → ta... to the riboso...

rRNA - ribosomal → ma...

tRNA - transfer → brings ... to the ribosome

...DNA polymera... of the recip... Thymine ...

...once comp... travel... to th...

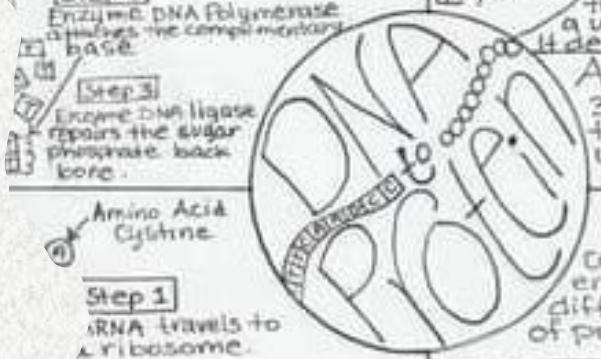
CODO

3 sequence... that togeth... a unit of "ge... It determines ...

ANTI-C

3 sequence... that togeth... unit of ge... transfer ...

DNA to PROTEIN



Amino Acid Cystine

Step 1 ...RNA travels to ...ribosome.

Step 2 Ribosome "reads" the mRNA codons, and "calls" a corresponding tRNA to come to the ribosome.

20 different amino acids

mRNA

Step 3 The tRNA that comes to the ribosome bringing the matching amino acid. As the tRNA's ...come they detach from ...heir amino acid.

...ep 4 ...no Acids attach to ... other with peptide ...s forming a poly- ...de → protein

...ATION

CELLS

Cells respon... environment... different types of protein.

All cells have the same DNA → But di... expres...

Advantages → inju...

Disadvantages → ca...

PROTEINS

1. Structural ... of cell ... materials
2. Funct... of horm... en...

substitutions "point mutations"	deletions
ACC AAC one base is substituted for another base.	ABC DGT ACGGT a base is deleted
May or may not change amino acid	will likely change all ...ino acids after the dele...

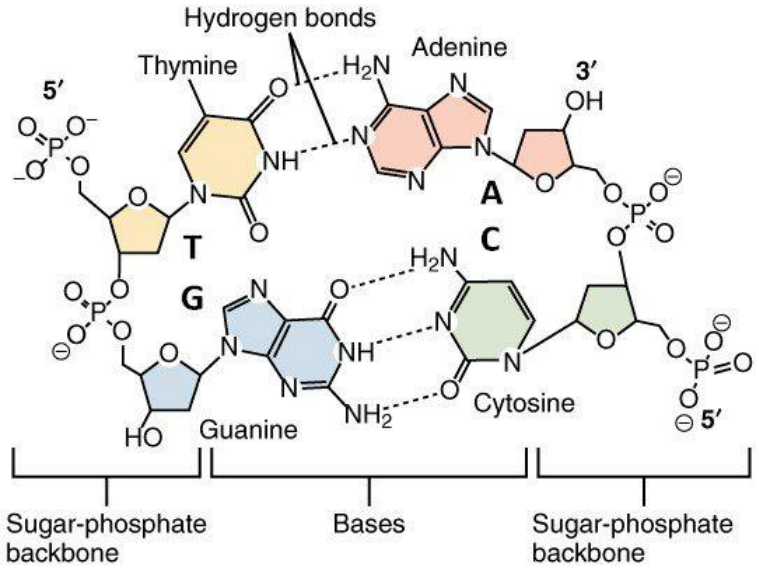
MUTATION

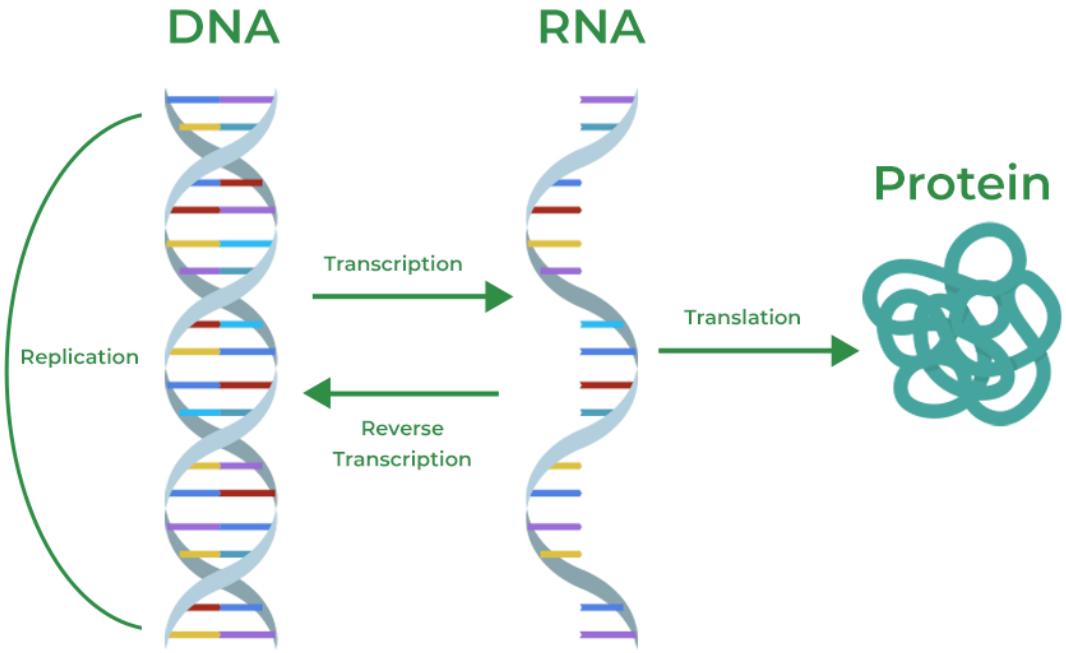
Building blocks of life

Watson & Crick

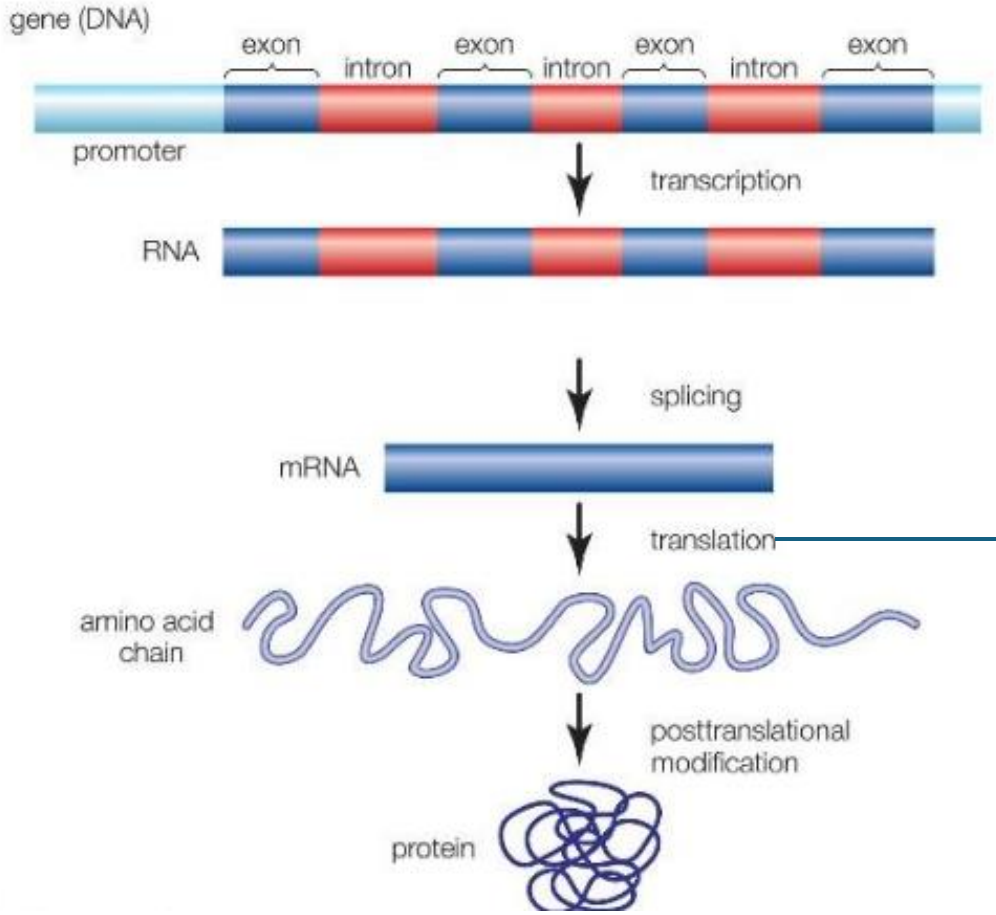
Rosalind Franklin

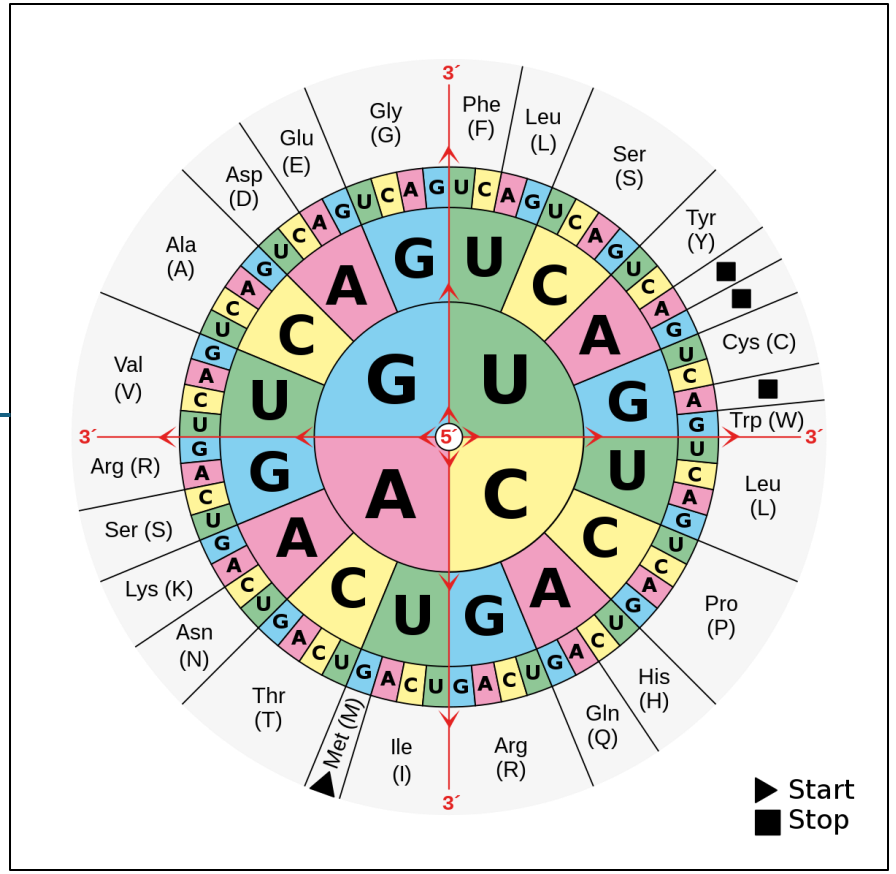
While evolutionary biologists were fashioning the Modern Synthesis, geneticists around the world searched furiously for the molecules that carried genetic information.



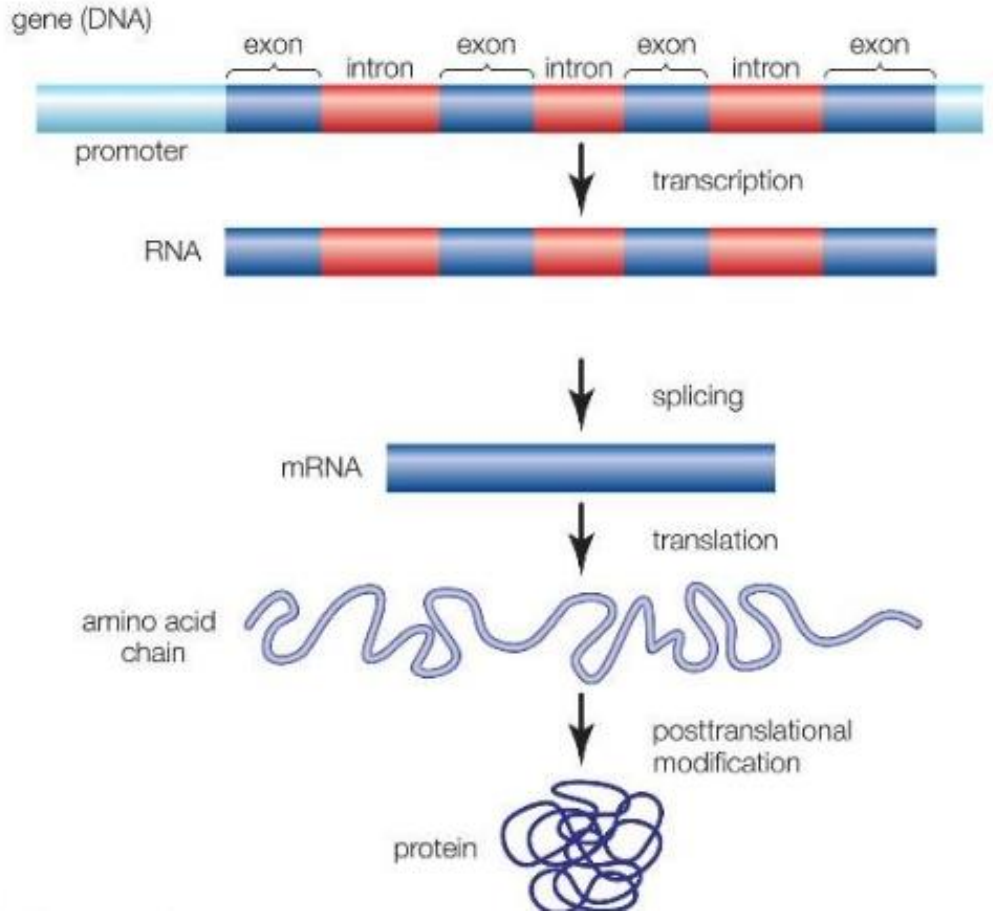


Building blocks of life





Building blocks of life

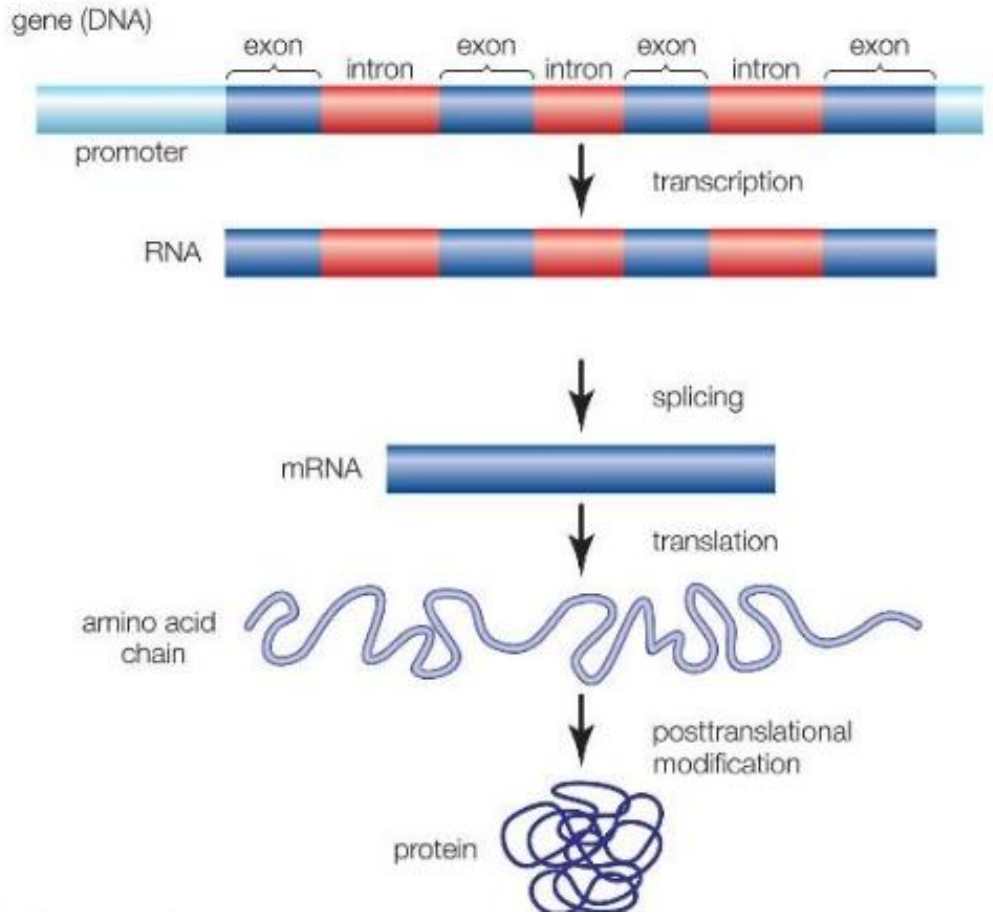


- **How many genes are in the human genome?**
  - A: Less than 5,000
  - B: Between 5,000 and 10,000 genes
  - C:Between 20,000 and 25,000 genes
  - D: Between 50,000 and 100,000 genes
  - E: More than100,000 genes

- **What percentage of the human genome is made up of protein-coding DNA?**
  - A: Between 1-2%
  - B: Around 10%
  - C: Between 10-20%
  - D: More than 20%

- **What percentage of genes are shared between all humans?**
  - A: Less than 70%
  - B: Around 70-80%
  - C: Around 80-90%
  - D: More than 90%

Building blocks of life

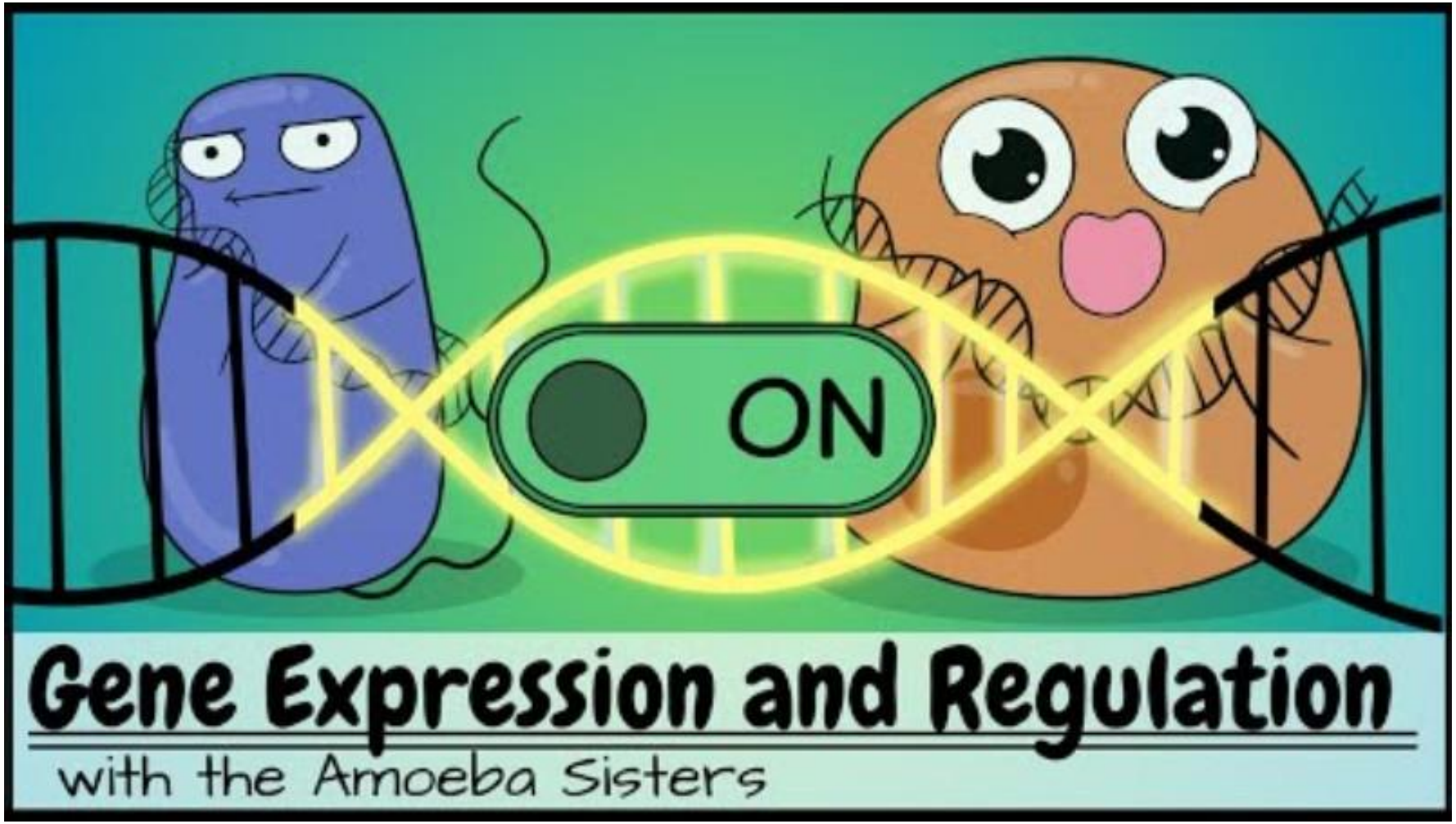


- **How many genes are in the human genome?**
  - A: Less than 5,000
  - B: Between 5,000 and 10,000 genes
  - **C:Between 20,000 and 25,000 genes**
  - D: Between 50,000 and 100,000 genes
  - E: More than100,000 genes
- **What percentage of the human genome is made up of protein-coding DNA?**
  - **A: Between 1-2%**
  - B: Around 10%
  - C: Between 10-20%
  - D: More than 20%
- **What percentage of genes are shared between all humans?**
  - A: Less than 70%
  - B: Around 70-80%
  - C: Around 80-90%
  - **D: More than 90%**

99,4%. This 0.6% difference is responsible for the diversity in physical traits, disease susceptibility, etc

➔ How you can build a human and have big differences between them with just 20,000 genes and “little” variation between these genes is something you will learn in the next days!

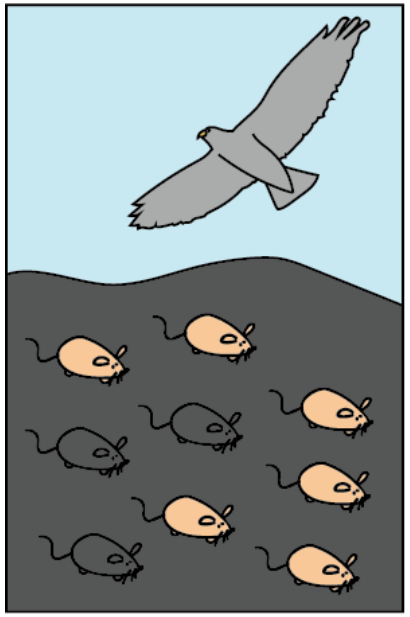
Building blocks of life



Module 3: Sources of Phenotypic Variation

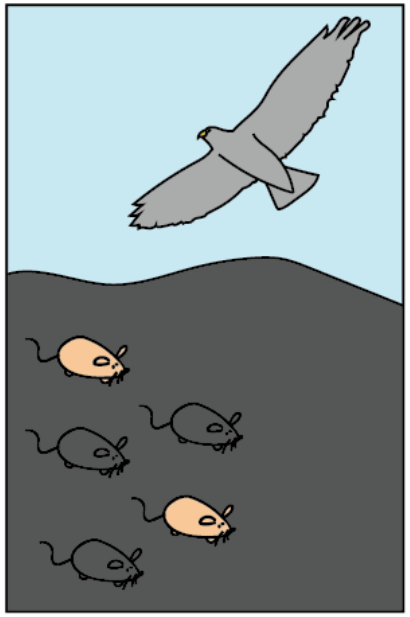
How can variation occur?

?



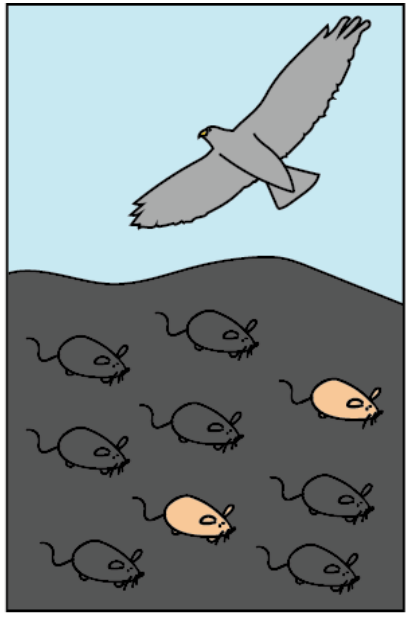
A population of mice has moved into a new area where the rocks are very dark. Due to natural genetic variation, some mice are black, while others are tan.

Some mice are eaten by birds



Tan mice are more visible to predatory birds than black mice. Thus, tan mice are eaten at higher frequency than black mice. Only the surviving mice reach reproductive age and leave offspring.

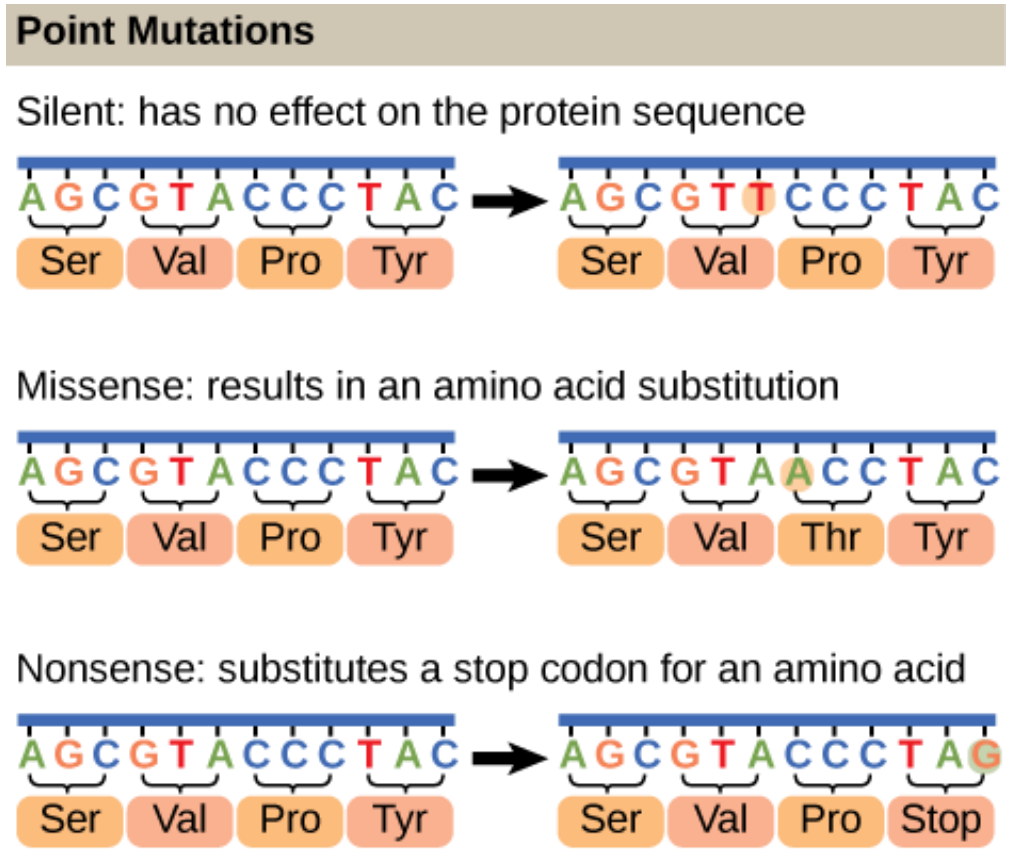
Mice reproduce, giving next generation



Because black mice had a higher chance of leaving offspring than tan mice, the next generation contains a higher fraction of black mice than the previous generation.

Genetic mutations and structural variation

- **Mutations and indels**

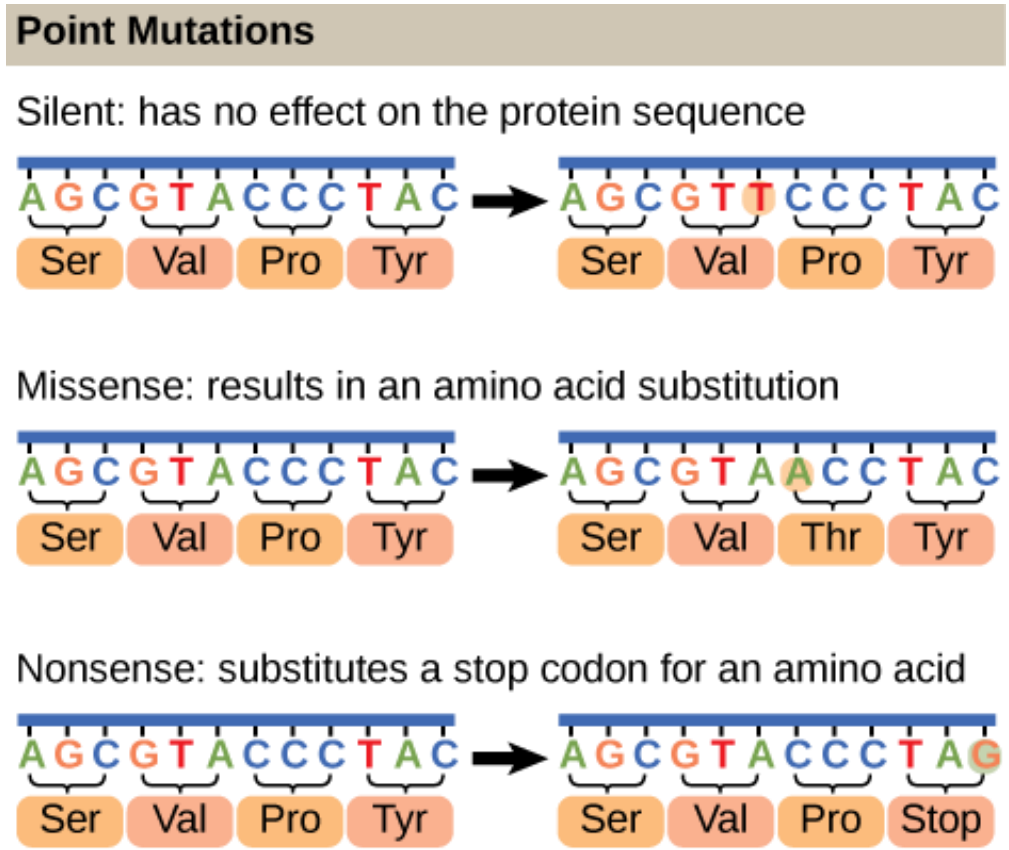


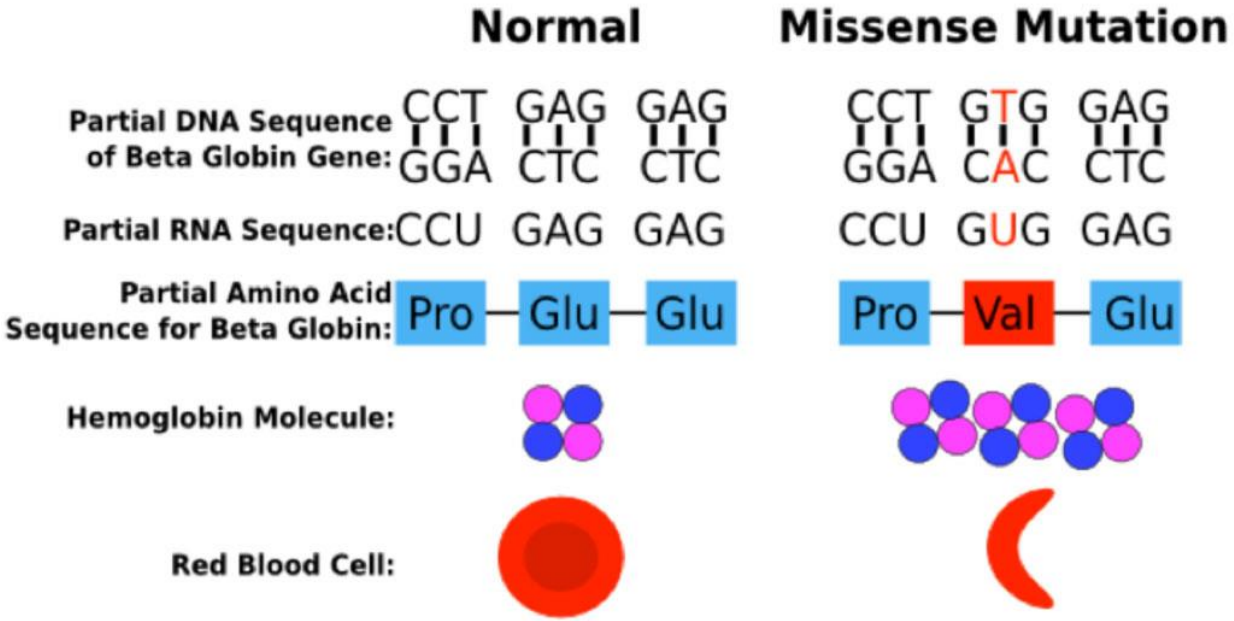
Important definition in genetics:

A single nucleotide polymorphism (**SNP**), pronounced "snip," is a variation at a single position in a DNA sequence among individuals

Genetic mutations and structural variation

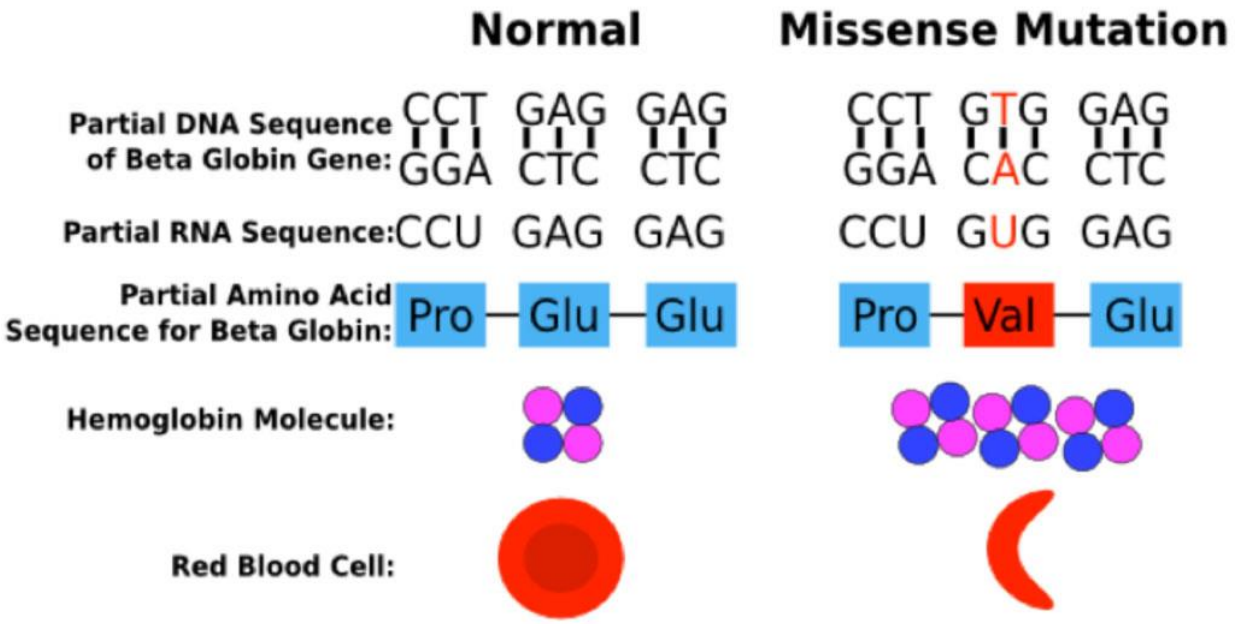
- **Mutations and indels**

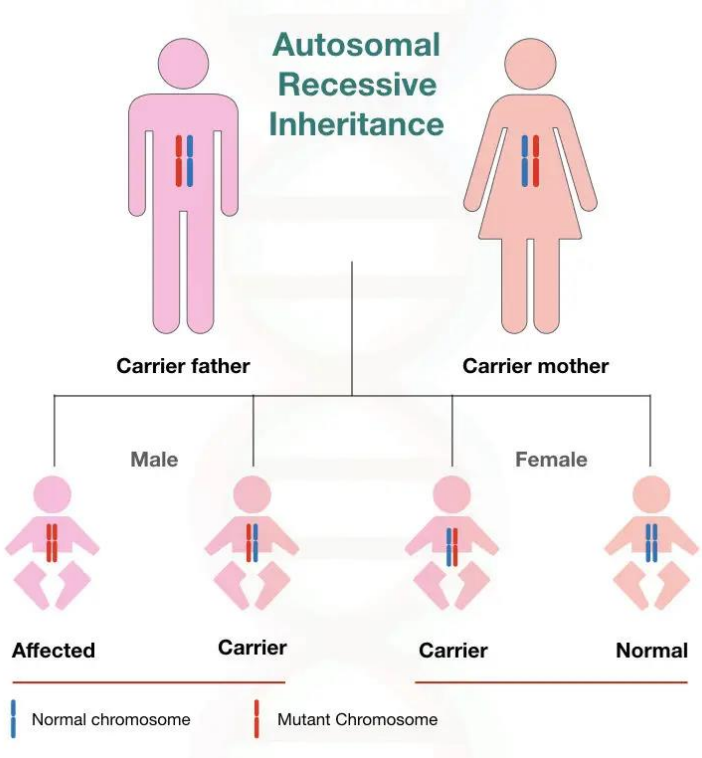




Genetic mutations and structural variation

- **Mutations and indels**

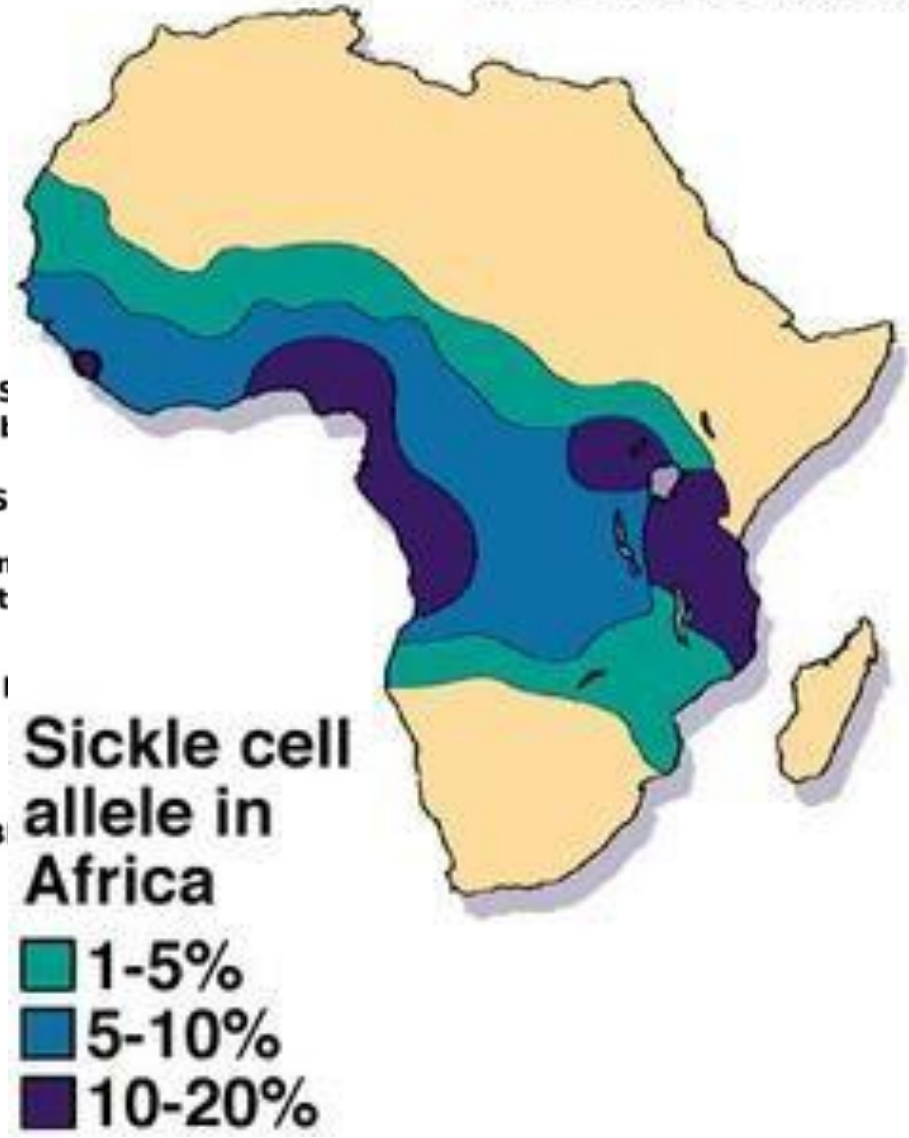


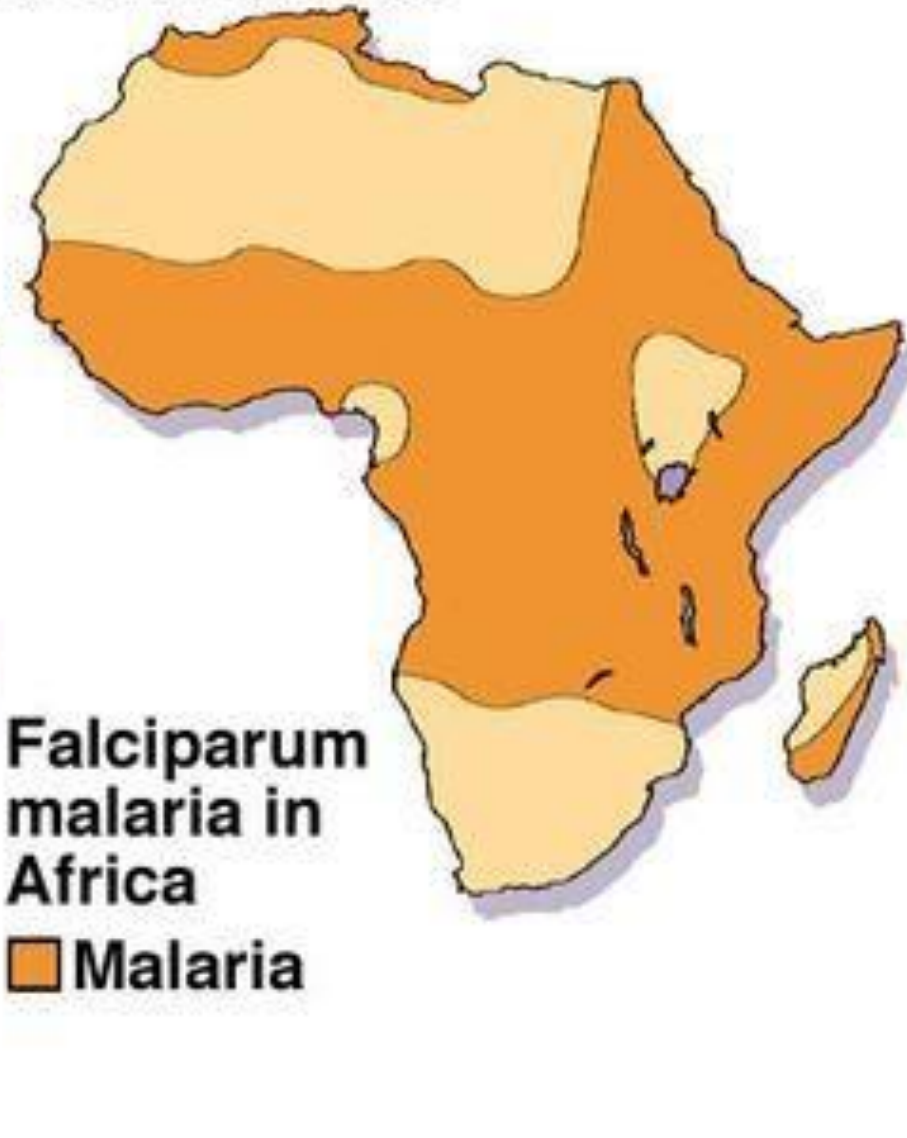


- Sickle cell is co-dominant trait

G

Correlation between Sickle Cell Allele and Malaria





Partial DNA S
of Beta Glob

Partial RNA S

Partial Ar
Sequence for Bet

Hemoglobin

Red B

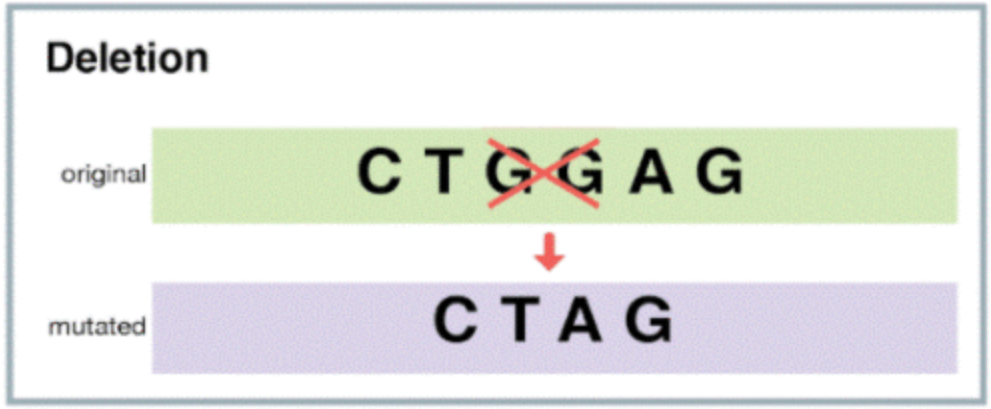
nother

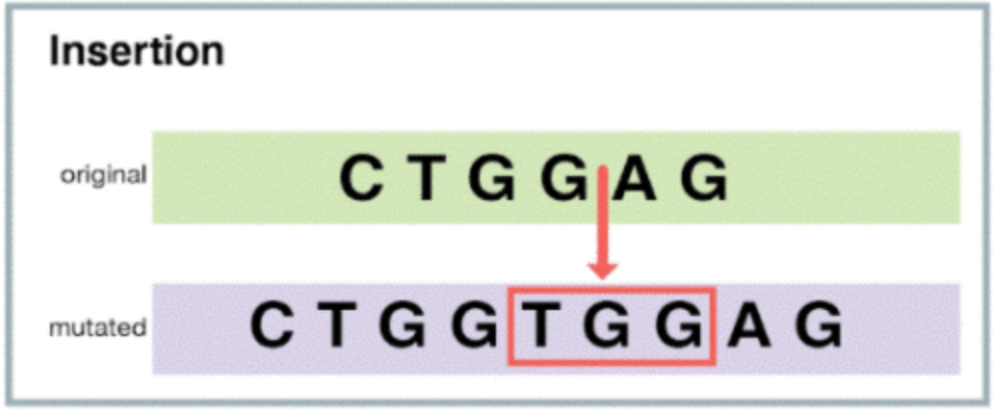
nale

Normal

Genetic mutations and structural variation

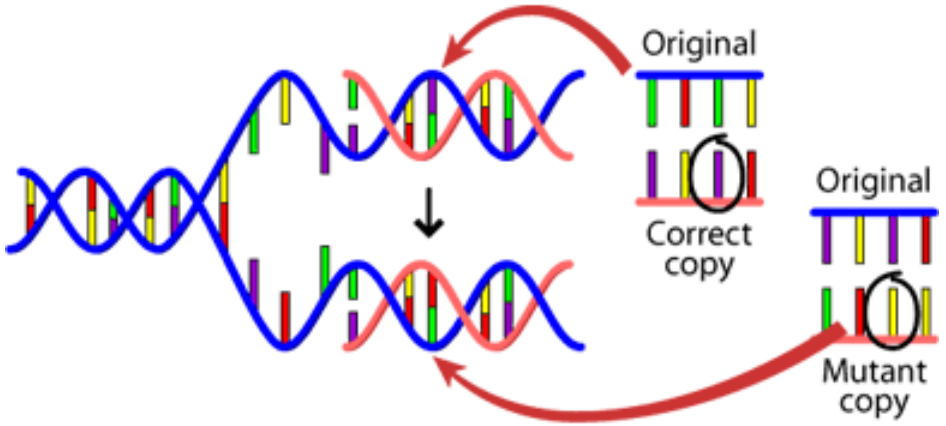
- **Mutations and indels**





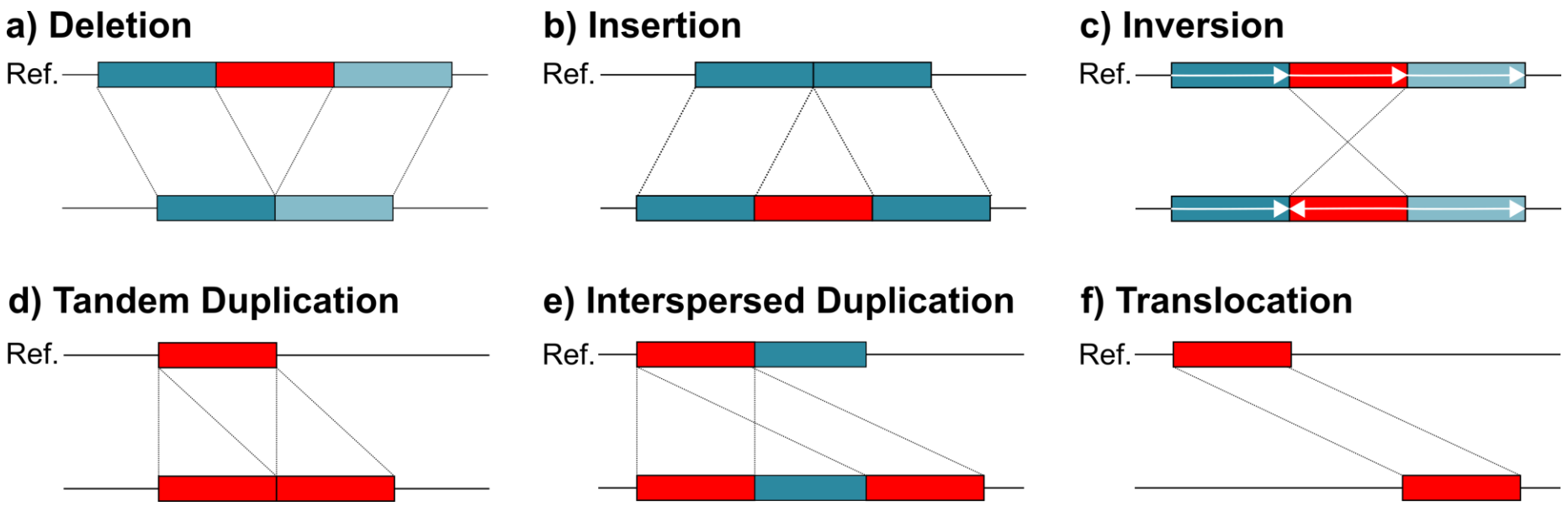
Genetic mutations and structural variation

- **Cause**
  - **1. DNA fails to copy accurately**
    - Most of the mutations that we think matter to evolution are “naturally-occurring.”
    - For example, when a cell divides, it makes a copy of its DNA — and sometimes the copy is not quite perfect. That small difference from the original DNA sequence is a mutation.
  - **2. External influences can create mutations**
    - Mutations can also be caused by exposure to specific chemicals or radiation.
    - These agents cause the DNA to break down.
    - This is not necessarily unnatural — even in the most isolated and pristine environments, DNA breaks down.
    - Nevertheless, when the cell repairs the DNA, it might not do a perfect job of the repair. So the cell would end up with DNA slightly different than the original DNA and hence, a mutation.



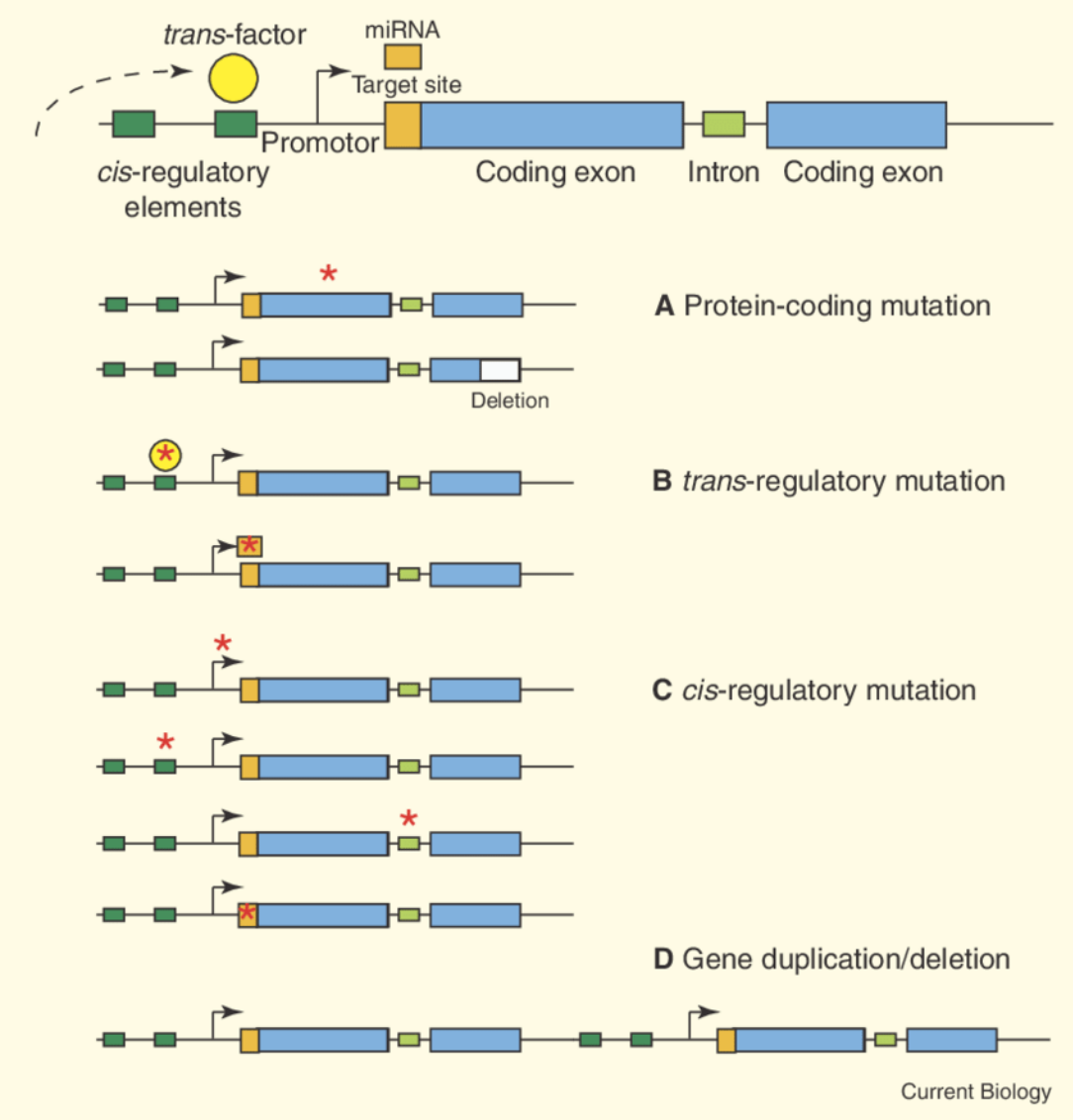
Genetic mutations and structural variation

Structural variation

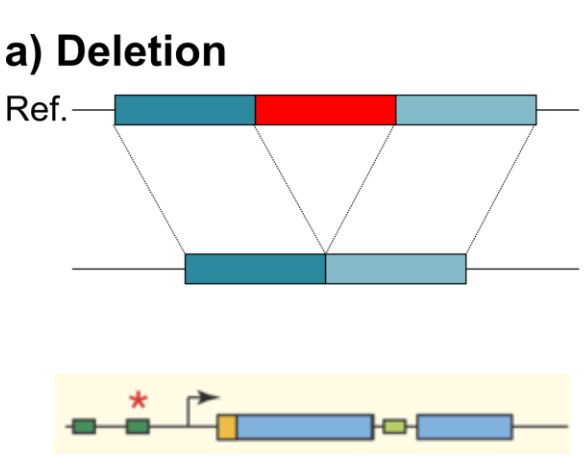


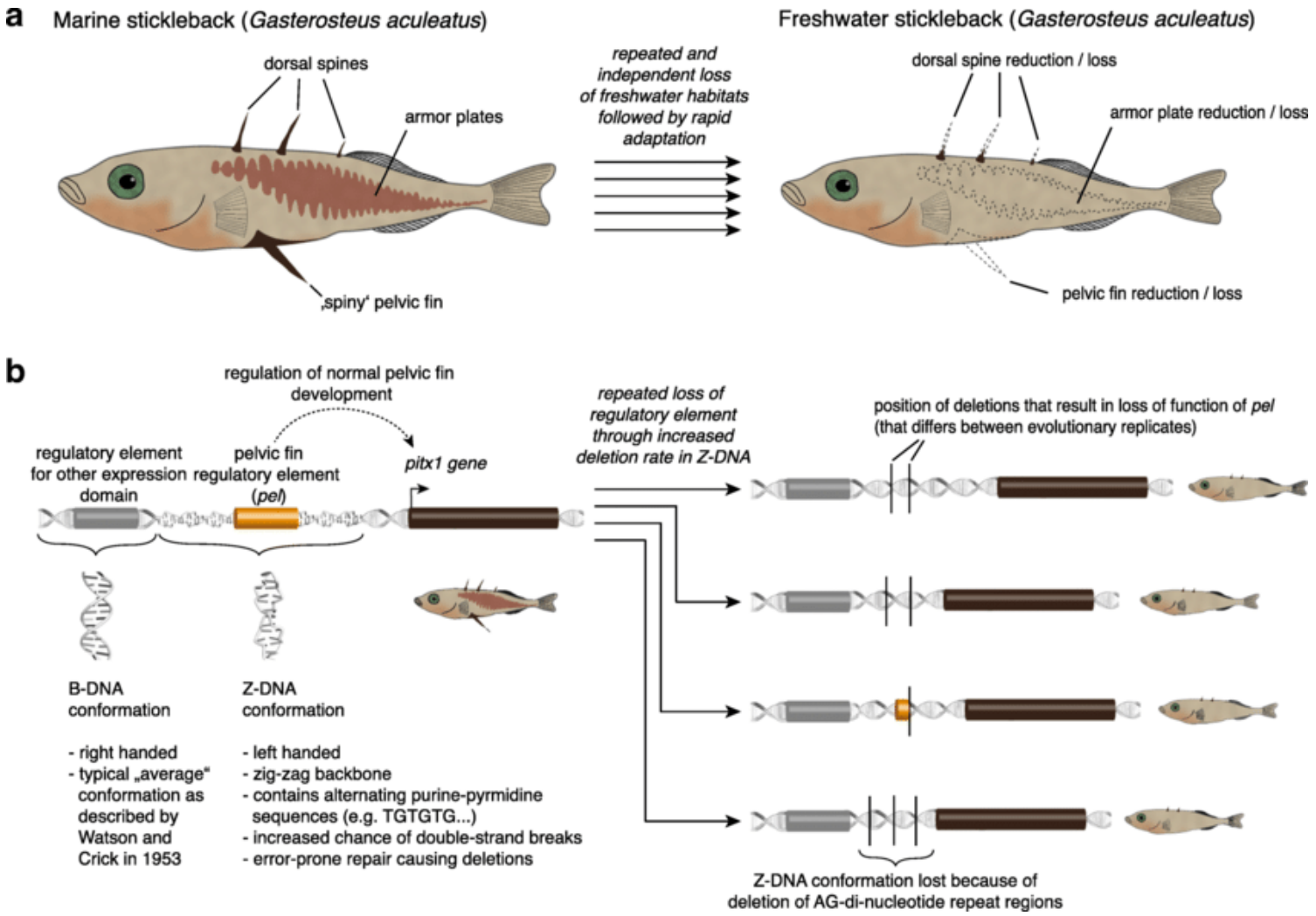
Cause: errors during recombination or DNA replication, transposons, ...

Regulatory-Sequence Variation



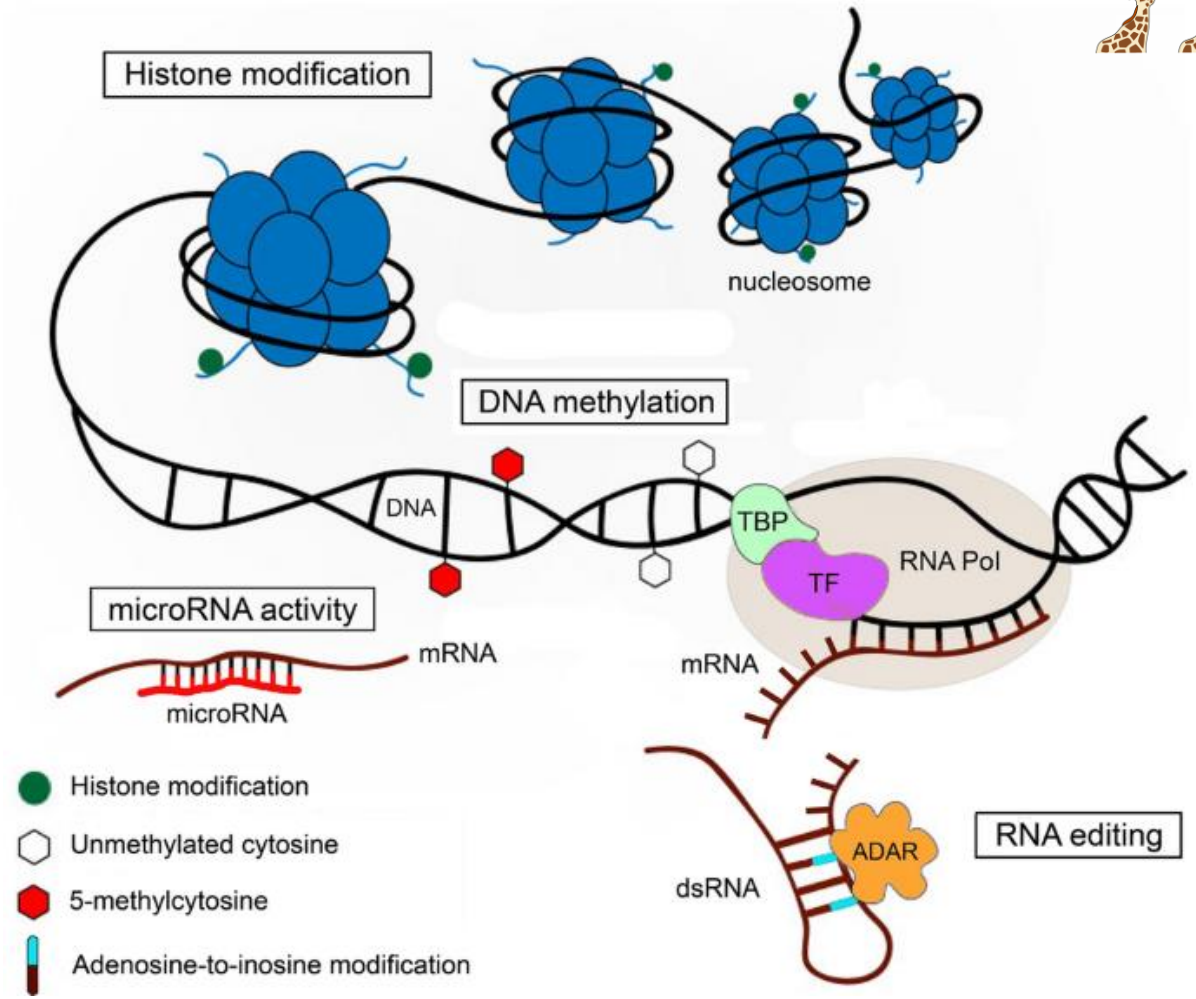
Genetic mutations and structural variation





Epigenetic Variation

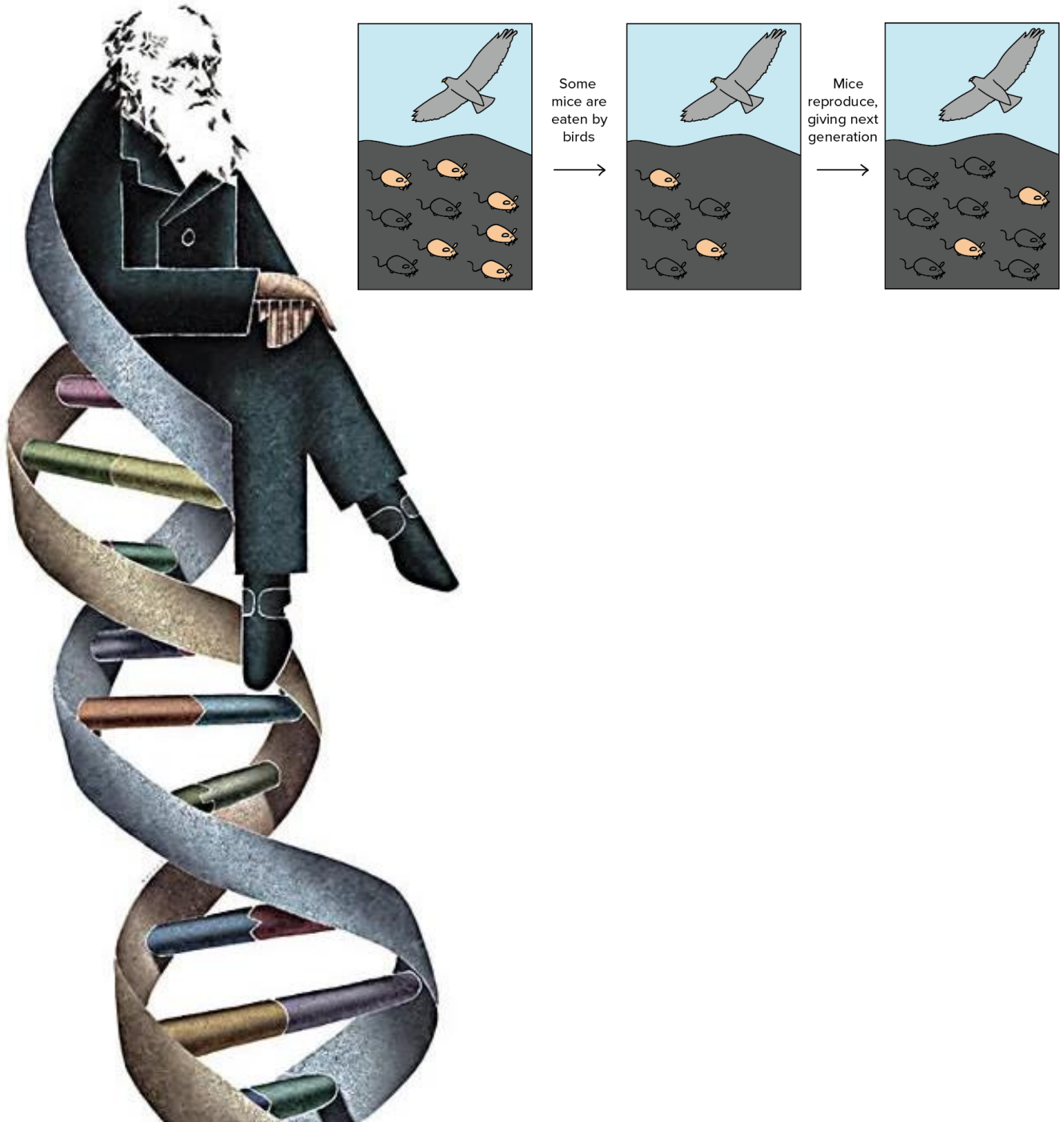
- **heritable changes in gene expression or cellular function that do not involve changes to the underlying DNA sequence.**
- https://www.youtube.com/watch?v=AvB0q3mg4sQ



Heritable variation arises from multiple molecular mechanisms

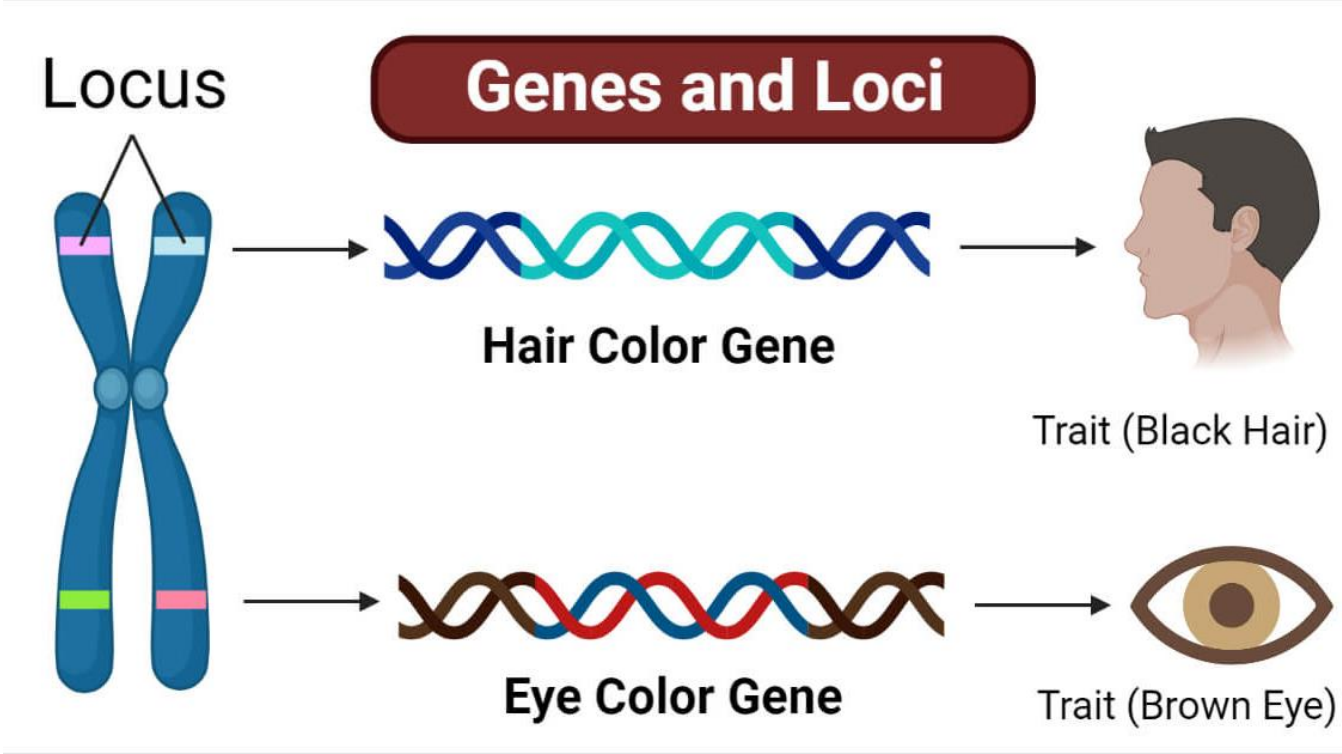
—sequence changes, structural rearrangements, regulatory shifts, and epigenetic marks—

all of which feed into evolutionary change.

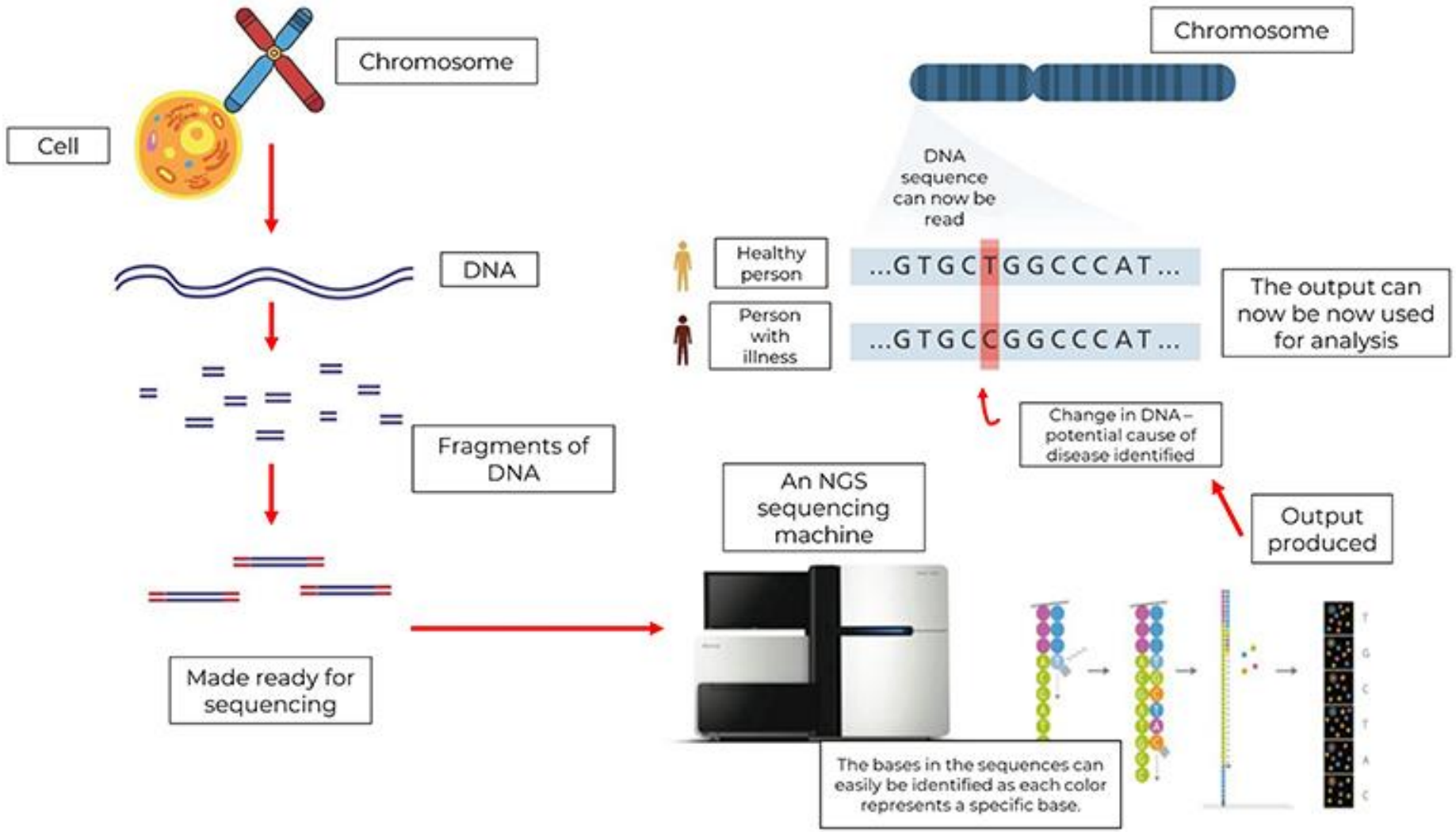


Module 4: From Genes to Traits: Methods and Case-studies

How do researchers study this?
What tools do we use to connect genes and their regulation to their phenotype?



NGS: Next-Generation-Seqencing



NGS: Next-Generation-Seqencing

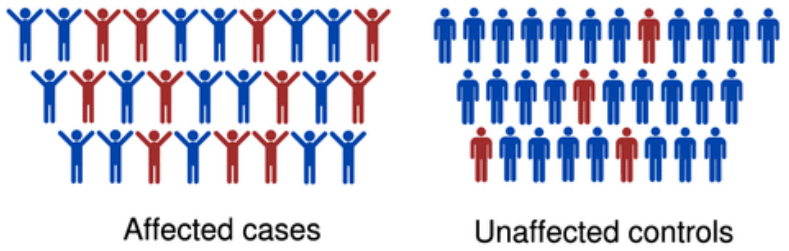
https://youtu.be/WKAUtJQ69n8

Genome-wide association study (GWAS)

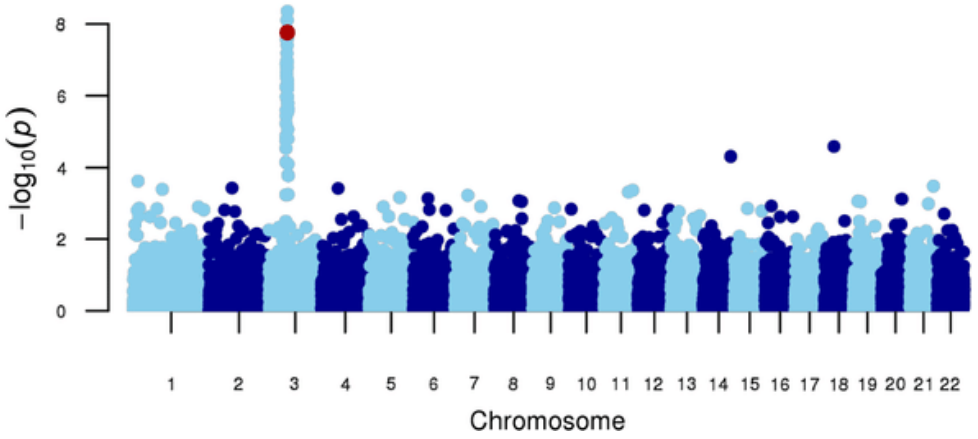
Sequence Variation

ATGCCAGTGTTTCAAGATGCTTGGCCAGCTGGACGAGGGCGATGAC
ATGCCAGTGTTTCAAGATG**T**TTGGCCAGCTGGACGAGGGCGATGAC

Disease



GWAS



- Genetic variation
- Phenotypic variation
- Linking: what location in the genome is associated with phenotypic variation

The Journal of Nutrition
Available online 7 June 2025
In Press, Corrected Proof What's this?

Genomics, Proteomics, and Metabolomics

A Genome-Wide Association Study of Taste Liking in the Danish Population

Background

Taste liking, a complex trait, plays an important role in food choice and eating behavior, thereby influencing risk of diet-related diseases.

Objectives

This study aimed to identify novel loci that could explain differences in liking of 5 basic tastes, fat sensation, and 2 oral sensations, represented by several food items.

Methods

Liking scores were derived using a newly developed taste liking questionnaire (TasteLQ), validated in the Danish population. We conducted a genome-wide association study (GWAS) of liking of 6 modalities (sweet, salty, sour, bitter-astringency, umami, and pungency) and 9 factors representing modality subgroups among 6,437 Danish adults. As a secondary analysis, GWASs of 44 single food items from TasteLQ were also undertaken.

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Genomics, Proteomics, and Metabolomics

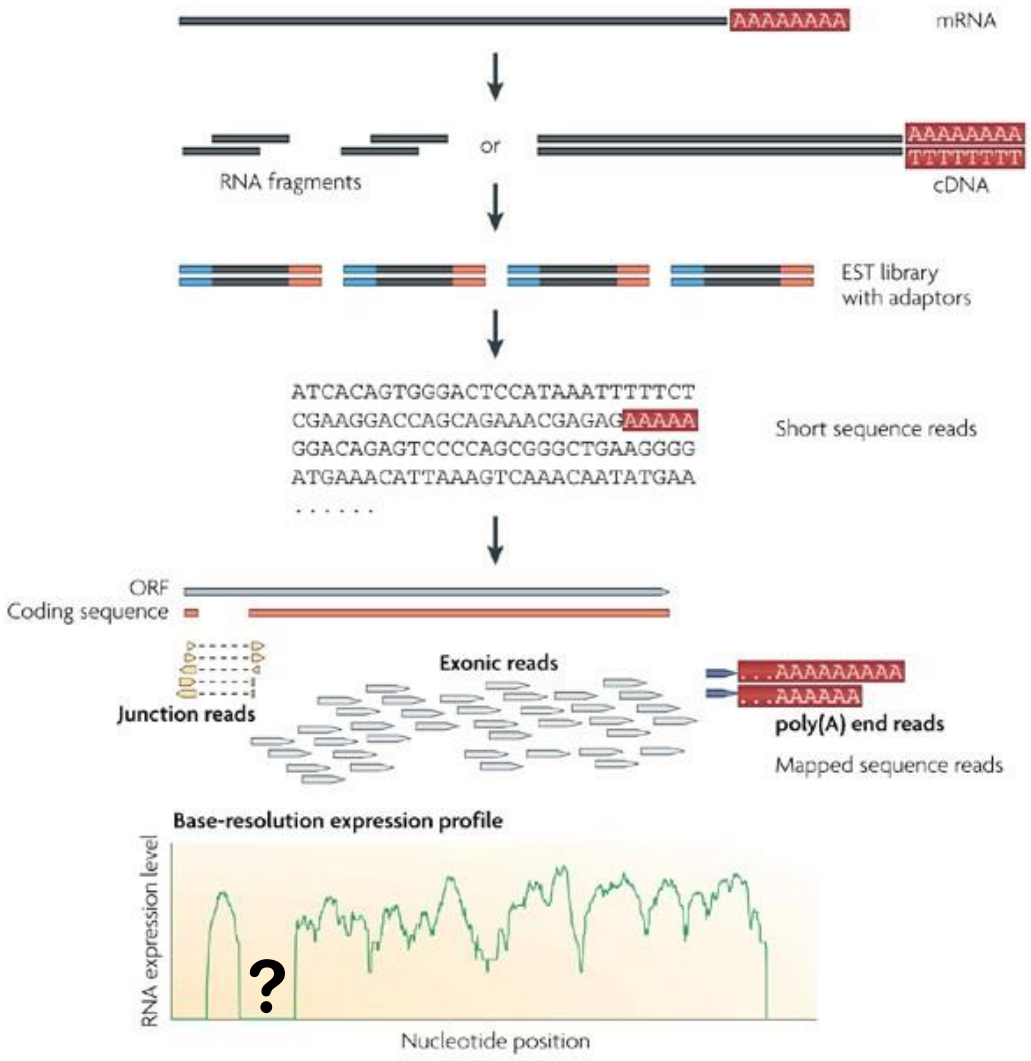
A Genome-Wide Association Study of Taste Liking in the Danish Population

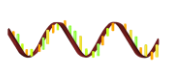
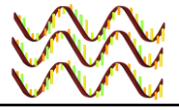
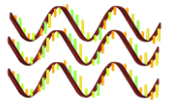
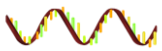
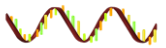
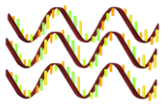
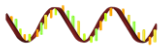
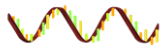
The TAS2R38 gene plays a significant role in bitter taste perception.

Variations in this gene, specifically single nucleotide polymorphisms (SNPs), can lead to differences in how strongly individuals perceive bitterness.

These variations can result in individuals being classified as "tasters" or "non-tasters" of bitter compounds, which may influence food preferences and dietary choices.

Functional genomics: RNAseq

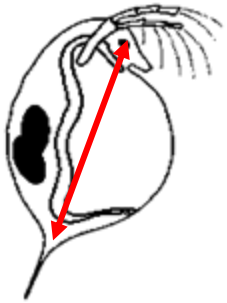


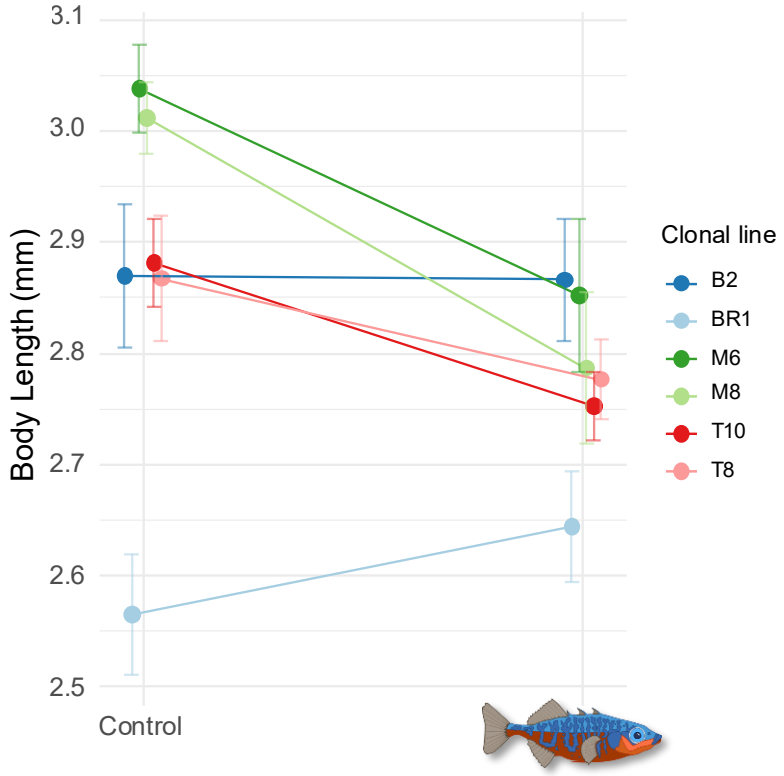
	CONTROL	TREATMENT	
GENE A			
GENE B			
GENE C			
GENE D			

Results

1

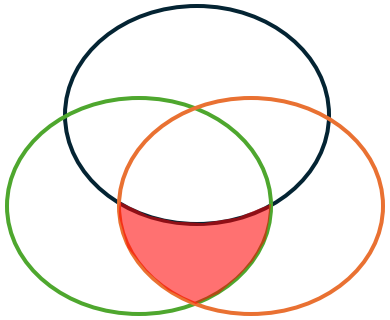
△ phenotype





Results

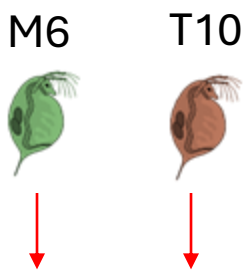
3 △ gene expression



plastic response genes

26 DEGs with 13 having consistent expression profile

Ex.:



- **lipid catabolic process / vitellogenin: ↓**
  - *algae-rich diet ➔ egg formation precursor lipids ↑ ➔ use in fecundity instead of catabolism ➔ fecundity ↑*
    - M & T ➔ fecundity ↑
  - *Neonates smaller ➔ yolk per egg ↓ ➔ vitellogenin↓*
    - M & T ➔ size neonates↓
- **Cuticle production and formation: ↑M & ↓*T***
  - *↑M: growth rate ↑ ➔ moulting ↑ ➔* cuticle ↑
  - *↓T: growth rate ↓ ➔ moulting ↓ ➔* cuticle ↓
- **transmembrane transporter ↓ / haemoglobin ↓ / …**

Up to you

- Search for research using these techniques on your favorite animal/plant or a historical figure in the slides you found interesting and I will help explain it

- We can watch a video of a scientist explaining their research and do some exercises
  - Dog GWAS
    - https://www.biointeractive.org/classroom-resources/dog-dna-human-diseases
  - Maize domestication
    - https://www.biointeractive.org/classroom-resources/popped-secret-mysterious-origin-corn
  - Mice fur color
    - https://www.biointeractive.org/classroom-resources/allele-and-phenotype-frequencies-rock-pocket-mouse-populations

- We can play around with the natural selection simulation

- If you've had enough, we can also relax…

You can also ask questions if you have any☺²