GENETICS AND EVOLUTION (22ZOOC14)

Mendelian Principles

- Mendel’s experiments
- Mendel’s principle of Inheritance
- Exceptions of Mendel’s principle of Inheritance

Introduction

Genetics is the study of heredity.

Johann Gregor Mendel (1822–1884) - Father of Genetics.Because of Mendel's work, the fundamental principles of heredity were revealed.

He set the framework for genetics **long before chromosomes or genes had been identified**, at a time when **meiosis was not well understood**.

Mendel selected a simple biological system and conducted **methodical, quantitative analyses** using **large sample sizes**.

Genes, carried on chromosomes, are the **basic functional units of heredity** with the capability to be replicated, expressed, or mutated. Today, the postulates put forth by Mendel form the basis of **classical**, or **Mendelian genetics**.

Not all genes are transmitted from parents to offspring according to Mendelian genetics, but Mendel's experiments serve as an excellent starting point for thinking about inheritance.

- Johann Gregor Mendel (1822–1884) was a lifelong learner, teacher, scientist, and man of faith.
- As a young adult, he joined the Augustinian Abbey of St. Thomas in Brno in what is now the Czech Republic.
- Physics, botany, and natural science courses - secondary and university levels.
- In **1856**, he began a decade-long research pursuit involving **inheritance patterns in honeybees and plants**, ultimately settling on pea plants as his primary model system.

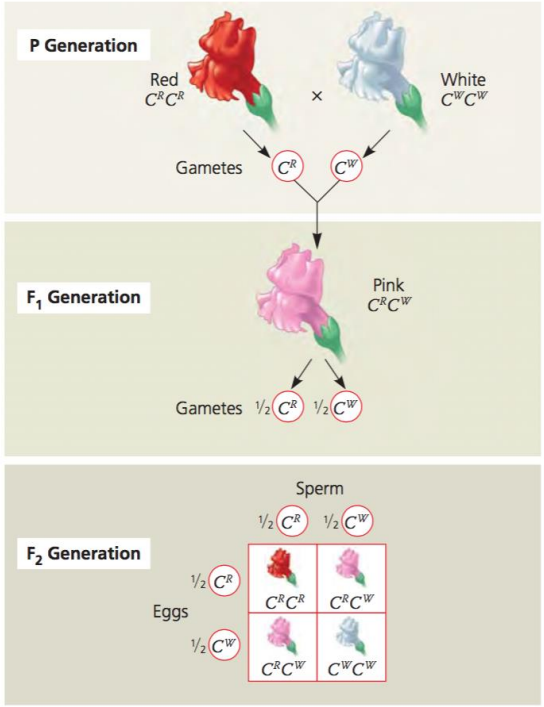
- In **1865**, Mendel presented the results - **30,000 pea plants to the local Natural History Society**. He demonstrated that **traits are transmitted faithfully from parents to offspring independently of other traits** and **in dominant and recessive patterns**.

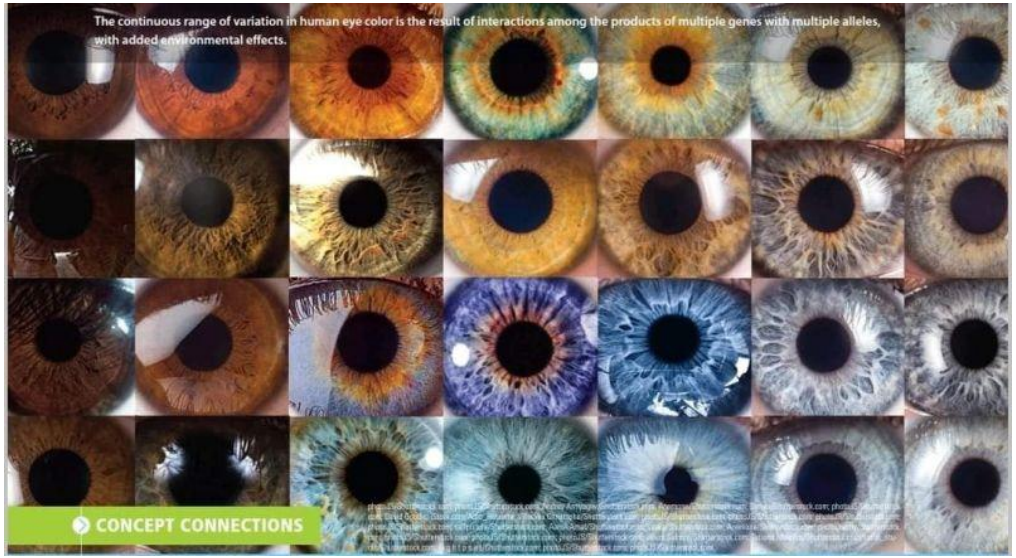
- In **1866,** he published his work, ***Experiments in Plant hybridization*** **in the proceedings of the Natural History Society of Brünn.**

- Mendel's work went **virtually unnoticed by the scientific community** that believed, incorrectly, that the process of inheritance involved a **blending of parental traits** that produced an intermediate physical appearance in offspring - **Continuous variation**.

- Offspring appear to be a **"blend" of their parents' traits**, that exhibit continuous variation.
  - Mendel's work - violet versus white flowers - **Discontinuous variation**.

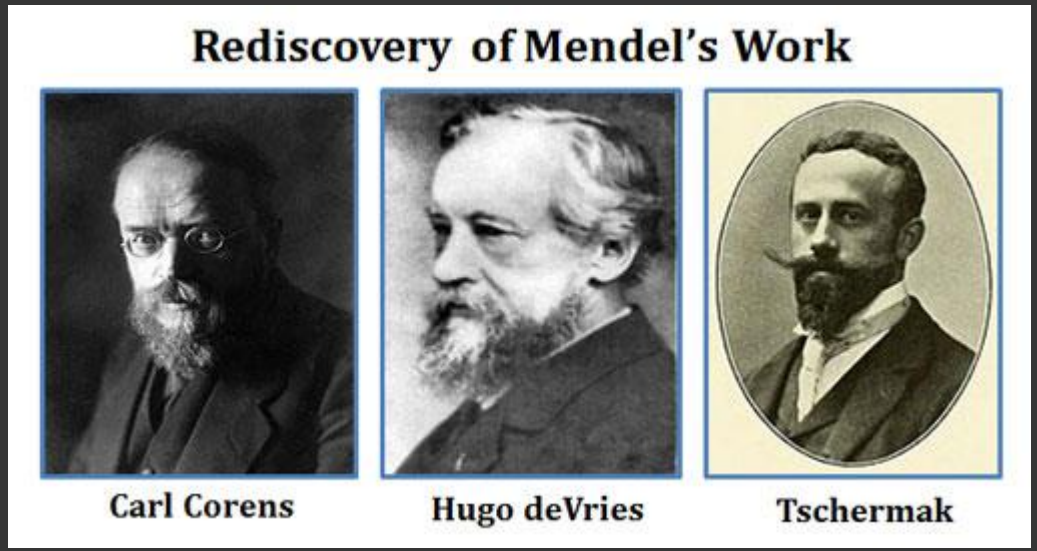
- Observation - Traits were **not blended** in the offspring, nor **absorbed**, but rather that they kept their **distinctness** and could be **passed on**.





- In 1868, Mendel became abbot of the monastery and exchanged his scientific pursuits for his pastoral duties.
- He was not recognized for his extraordinary scientific contributions during his lifetime.
- In fact, it was not until 1900 that his work was rediscovered, reproduced, and revitalized by scientists on the brink of discovering the chromosomal basis of heredity.

- Three Scientists independently rediscovered Mendel's work on the inheritance of characters in plants in In 1900
  - ✓ 34 years after Mendel's published paper in 1866
  - ✓ 16 years after Mendel died in 1884
- Three scientists were
  - ✓ Hugo de Vries of Holland,
  - ✓ Carl Correns of Germany &
  - ✓ Erick von Tschermak of Austria



Mendel's Model System

- Mendel's garden pea, *Pisum sativum* - to study inheritance.
- Naturally self-fertilizes.
- The flower petals remain sealed tightly - result is highly inbred, or "true-breeding," pea plants.
- Plant matures within one season, several generations could be evaluated over a relatively short time.
- So large quantities could be cultivated simultaneously, allowing Mendel to conclude that his results did not come about simply by chance.

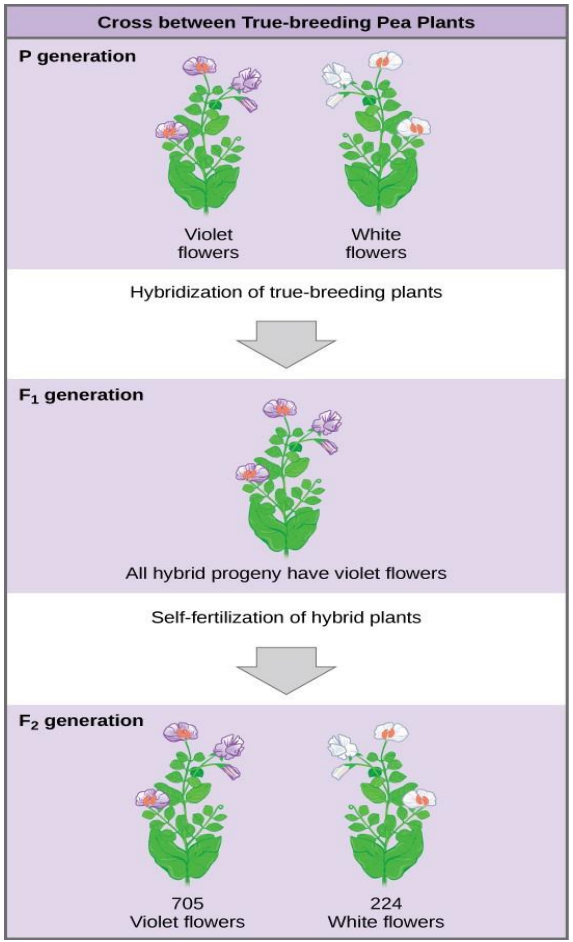
- These are plants that always produce offspring that look like the parent.
- By experimenting with true-breeding pea plants, Mendel avoided the appearance of unexpected traits in offspring that might occur if the plants were not true breeding.

Trait - A variation in the physical appearance of a heritable character.

Traits	Shape of seeds	Colour of seeds	Colour of pods	Shape of pods	Plant height	Position of flowers	Flower colour
Dominant trait	Round 	Yellow 	Green 	Full 	Tall 	At leaf junction 	Purple 
Recessive trait	Wrinkled 	Green 	Yellow 	Flat, constricted 	Short 	At tips of branches 	White 

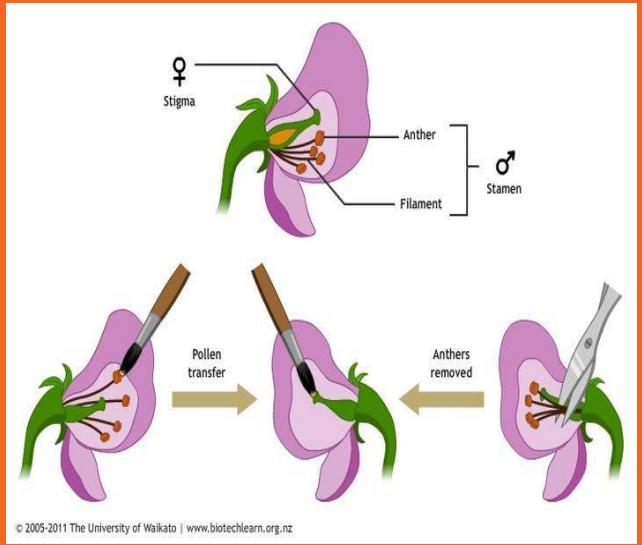
Seven pairs of contrasting traits in pea plant

Mendelian Crosses



- Mendel performed **hybridizations**.
- Mating two true-breeding individuals that have different traits.

Avoided self-fertilizing condition as that were confounding his results



Ratio of characteristics in the P−F1−F2 generations that were the most intriguing and became the basis for Mendel's principles.

Mendel's Principles of Inheritance

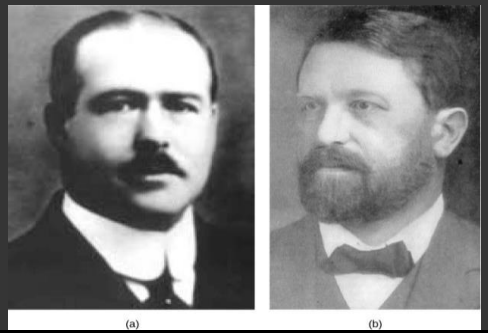
Pea-plant experiments - three principles - basis of inheritance in diploid organisms.

- **Principle of segregation**
- **Principle of dominance**
- **Principle of independent assortment**

Principle of segregation

Individuals have two copies of each trait, and that each parent transmits one of its two copies to its offspring. Eg: White flower trait reappeared in F2 generation.

- The traits that are passed on - result of genes that are inherited on chromosomes during meiosis and fertilization.
- The fact that the genetic factors proposed by Mendel were carried on chromosomes was proposed in **1902** by **Walter and Sutton** and **Theodor Boveri** as the Chromosomal Theory of Inheritance.



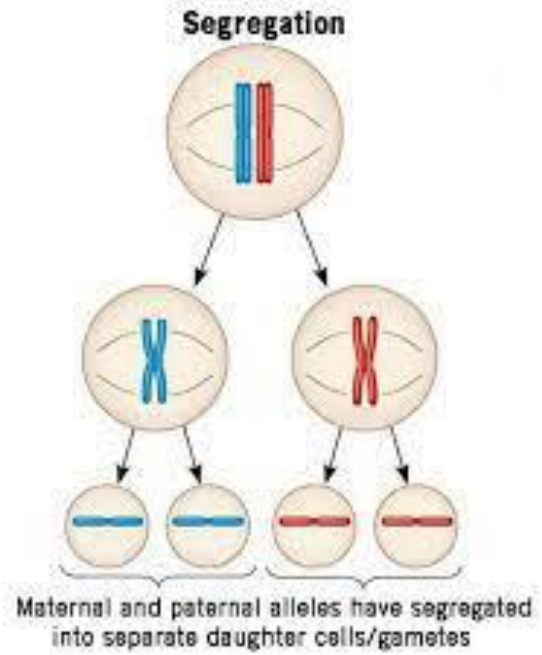
Chromosomes carry the unit of heredity (genes)

- Different versions of genes are called **alleles**.
- Diploid organisms that have two identical alleles of a gene on their two homologous chromosomes are homozygous for that trait.
- Diploid organisms that have two different alleles of a gene on their two homologous chromosomes are heterozygous for that trait.

The **physical basis of the principle of segregation is the first division of meiosis**, in which the homologous chromosomes with their different versions of each gene are segregated into daughter nuclei.

Since each gamete receives only one homolog of each chromosome, it follows that they receive only one allele for each trait.

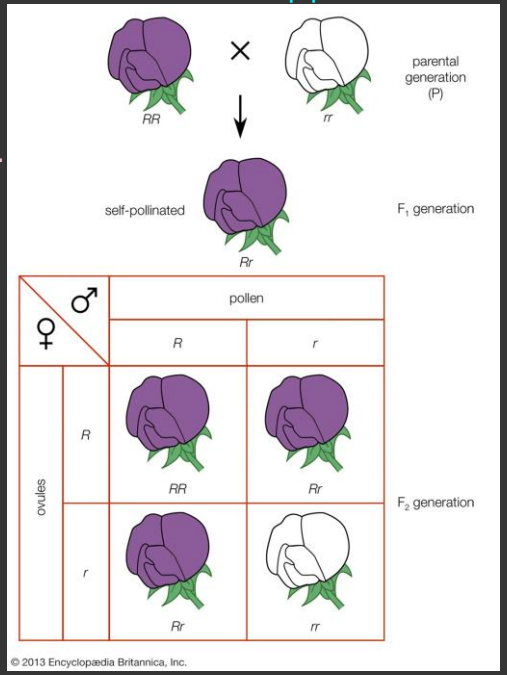
At fertilization, the zygote receives one of each homologous chromosome, and one of each allele, from each parent.



Principle of dominance

- Mendel concluded that the characteristics could be divided into dominant and recessive traits.
- Dominant traits are those that are expressed in a hybridization.
- Recessive traits become latent, or disappear, in the offspring of a hybridization but reappear in the progeny of the hybrid offspring.

Eg: the violet-flower trait is dominant and the white-flower trait is recessive.

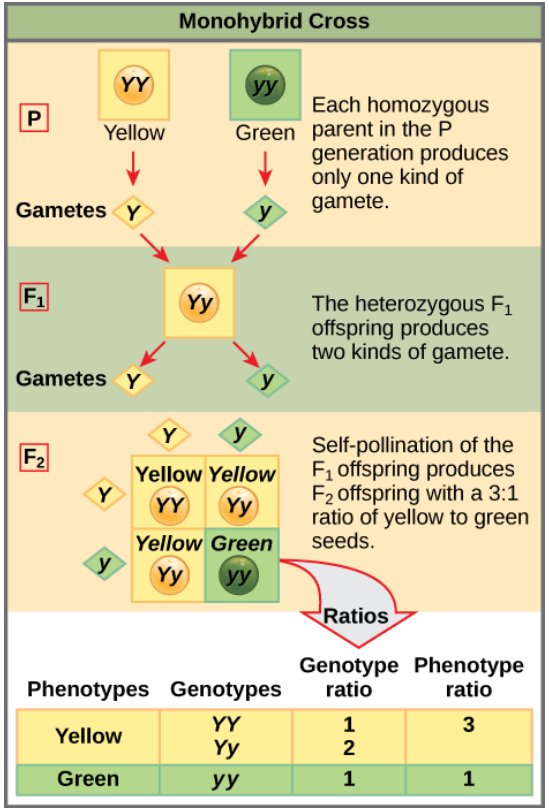


- In Mendel's experiments, the principle of dominance explains **why the F1 heterozygous offspring were identical to one of the parents**, **rather than expressing both alleles.**
- For a gene that is expressed in a dominant and recessive pattern, homozygous dominant and heterozygous organisms will look identical.
- The recessive allele will only be observed in homozygous recessive individuals.

Dominant Traits	Recessive Traits
Achondroplasia	Albinism
Brachydactyly	Cystic fibrosis
Huntington's disease	Duchenne muscular dystrophy
Marfan syndrome	Galactosemia
Neurofibromatosis	Phenylketonuria
Widow's peak	Sickle-cell anemia
Wooly hair	Tay-Sachs disease

Examples of dominant and recessive traits in humans

- Mendel's hybridization experiments demonstrate the difference between **phenotype** and **genotype**.
- **Punnett squares**, devised by the British geneticist **Reginald Punnett**, can be used to predict the possible outcomes of a genetic cross or mating and their expected frequencies.



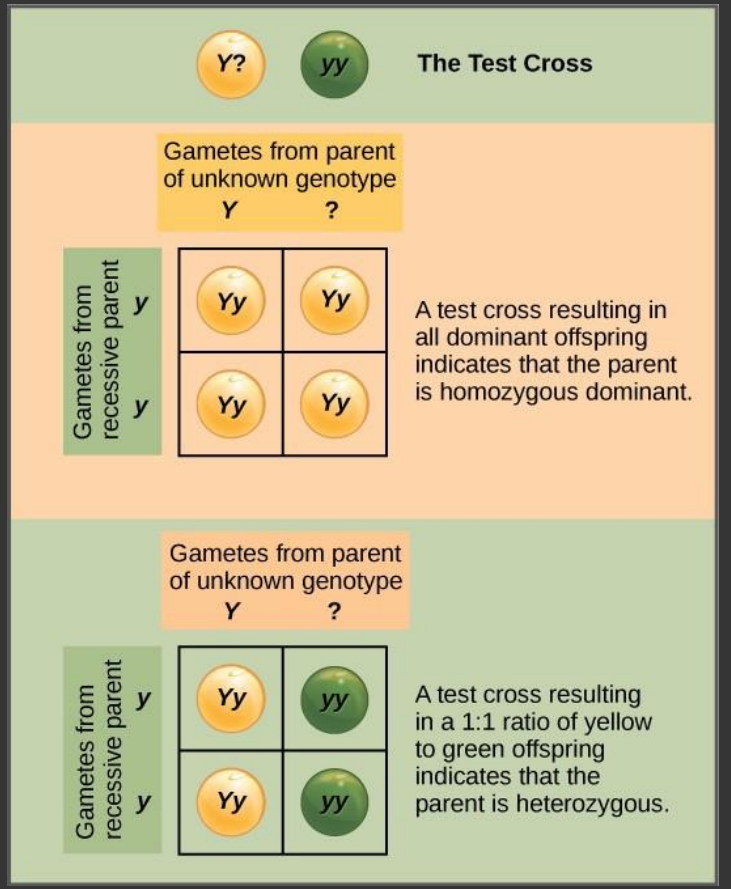
Test Cross to Determine Genotype -

Test cross validates Mendel's postulate that pairs of unit factors segregate equally.

In a test cross, an organism with the dominant phenotype is crossed with an organism that is homozygous recessive for the same characteristic.

If the dominant- expressing organism is a homozygote, then all F1 offspring will be heterozygotes expressing the dominant trait.

If the dominant expressing organism is a heterozygote, the F1 offspring will exhibit a 1:1 ratio of heterozygotes and recessive homozygotes.



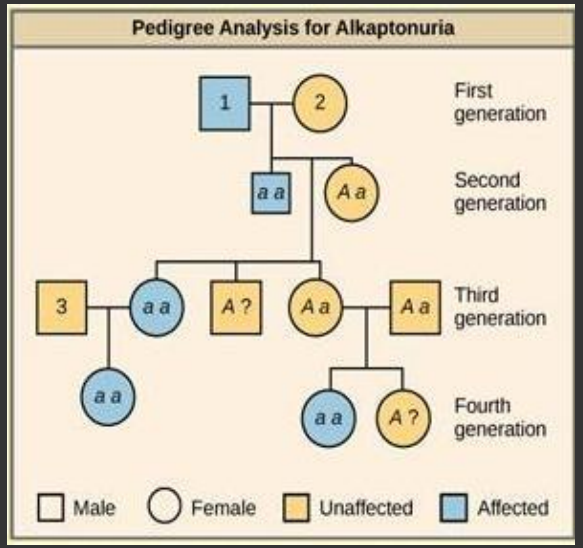
Many **human diseases** are inherited genetically.

A healthy person in a family in which some members **suffer from a recessive genetic disorder** may want to know if he or she has the disease-causing gene and what risk exists of passing the disorder on to his or her offspring.

Doing a test cross in humans is unethical and impractical. Instead, geneticists use pedigree analysis to study the inheritance pattern of human genetic diseases.

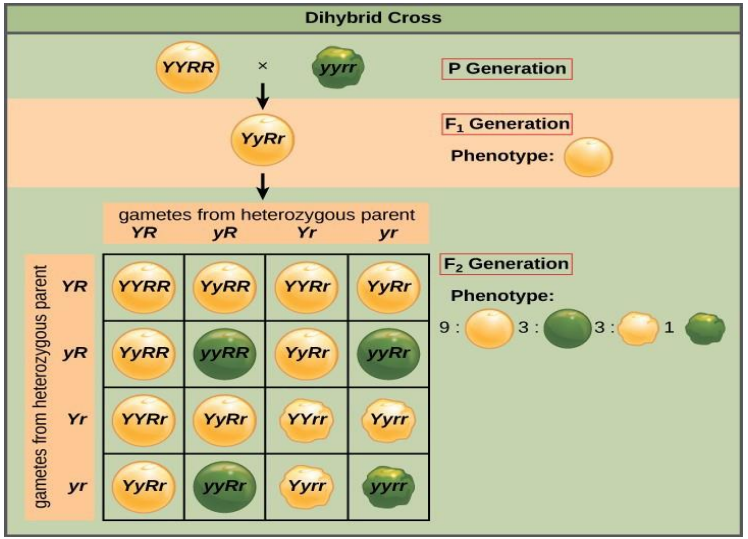
Eg: Recessive genetic disease alkaptonuria cannot properly metabolize two amino acids, phenylalanine and tyrosine.

Affected individuals may have darkened skin and brown urine, and may suffer joint damage and other complications.



Principle of Independent Assortment

“Genes do not influence each other with regard to the sorting of alleles into gametes, and every possible combination of alleles for every gene is equally likely to occur”.



The independent assortment of genes can be illustrated by a **dihybrid cross**, a cross between two true-breeding parents that express different traits for two characteristics.

Mendel’s 3:1 phenotypic ratio of monohybrid cross states that

- F1 hybrids contained two factors for each trait, one dominant and one recessive.
- Factors separated when gametes were formed; a gamete carried one copy of each factor.
- Random fusion of all possible gametes occurred upon fertilization

Test cross:

"It is between an individual with dominant phenotype and individual with recessive phenotype to see **if individual with dominant phenotype is homozygous or heterozygous**".

If tall pea plant is homozygous:

P TT

x tt

Gametes T

only t

F1

If tall pea plant is heterozygous:

P Tt

x tt

Gametes T & t

only t

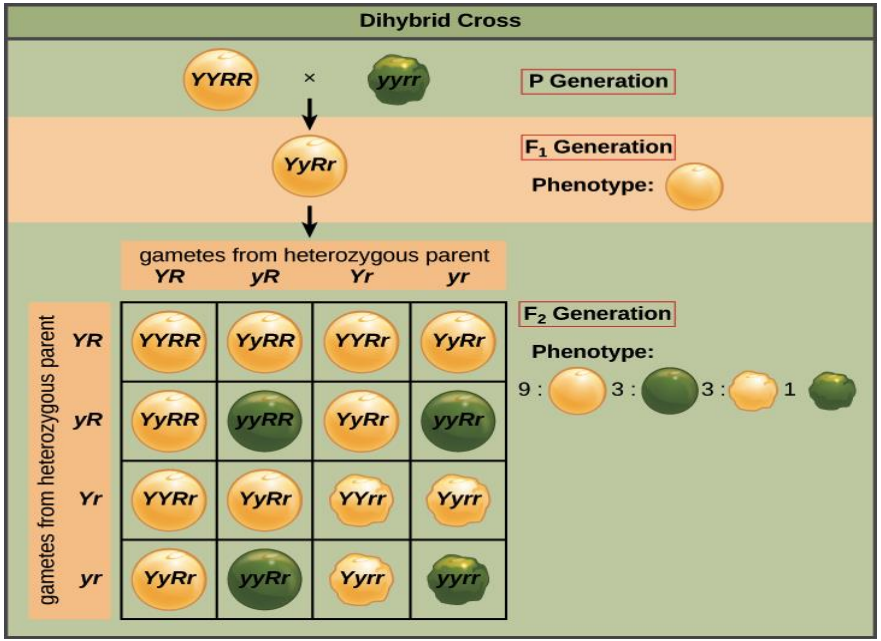
F1 **1 Tt (tall) : 1**

Law of Independent assortment

It was deduced from Mendel's experiment with **dihybrid cross.**

A cross between two independent traits.

"Assortment of genes of one pair is independent of other pair at the time of gametogenesis"



F1 hybrid produce 4 kinds of gametes in equal frequencies.

YR	1 / 4
Yr	1 / 4
yR	1 / 4
Yr	1 / 4

F1 heterozygotes were **self fertilized → (16 combinations)**

Phenotypic ratio 9 : 3 : 3 : 1

So, each pair of contrasting characters behaves independently and bears no association with a particular character.

Alternatives to Dominance and Recessiveness

Since Mendel's experiments with pea plants, other researchers have found that the **principle of dominance does not always hold true**. Instead, several **different patterns of inheritance have been found to exist.**

- Incomplete dominance
- Codominance
- Multiple alleles
- Environmental effects
- X- linked traits
- Human sex-linked disorders
- Lethal alleles
- Linked genes
- Epistasis

→ Don't follow Principle of Independent Assortment

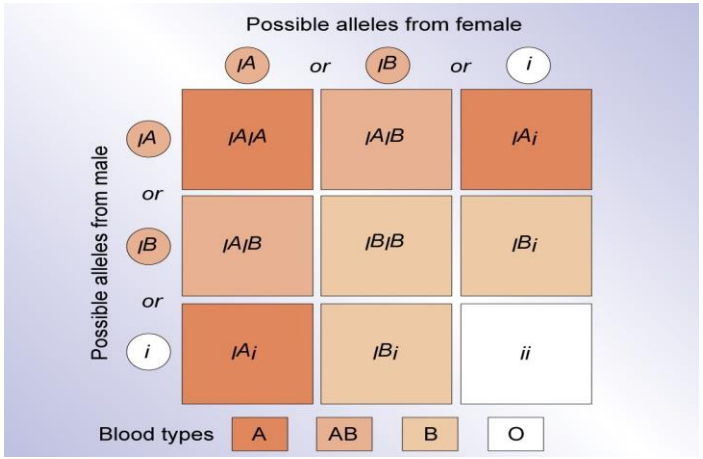
Multiple alleles

According to Mendel's experiment, of all seven pairs of characters selected, there were **only two different forms of a given gene exists** in a species.

When **more than two different forms of a given gene exists in a species**, it is referred as multiple alleles.

- The **no. of different genotypes** possible among **diploid** organisms is a function of the **no. of alleles (n)** that exists for a given gene.

No. of different genotypes = n (n+1) / 2



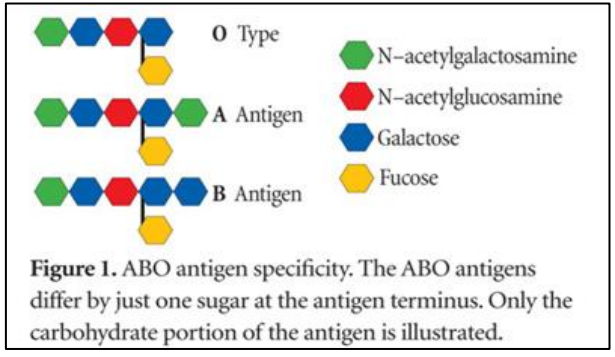
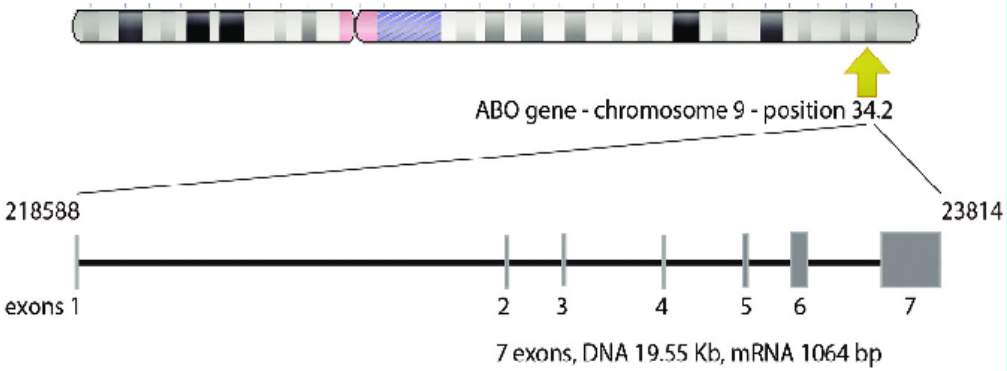
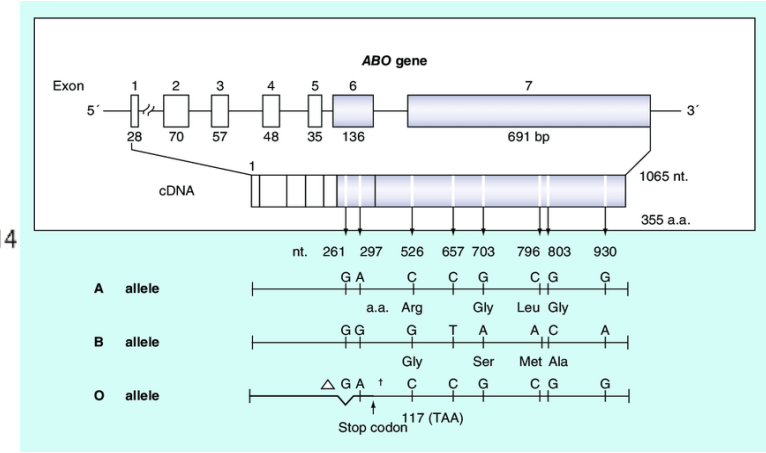


Figure 1. ABO antigen specificity. The ABO antigens differ by just one sugar at the antigen terminus. Only the carbohydrate portion of the antigen is illustrated.

Blood Type	Genotype	
A	$i^A i$ $i^A i^A$	AA AO
B	$i^B i$ $i^B i^B$	BB BO
AB	$i^A i^B$	AB
O	$i\,i$	OO





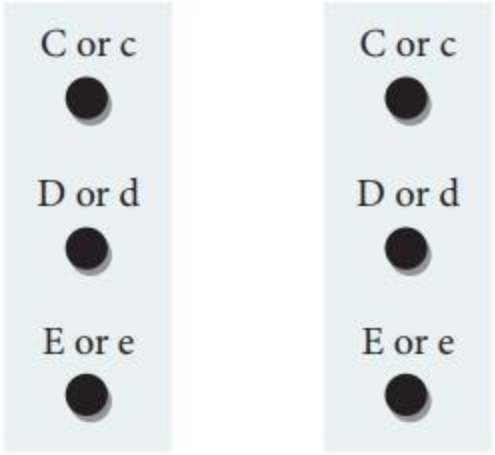


Fig. 4.1 Fischer and Race hypothesis – Rh Blood Type - Homologous Chromosome pair (showing 3 loci and 2 alleles per locus)

Rhesus Blood Group System

- 78 genotypes are possible. Most frequent genotypes are –
  - Cde/cde (33%)
  - Cde/cDe (18%)
  - Cde/cDE (12%)
  - cDE/cde (11%)
  - cde/cde (15%)
  - cdE/cde (1%)
  - Cde/cde (1%)

Rhesus Blood Group System

- For clinical & all practical purposes it is enough to know whether one is Rh POSITIVE or NEGATIVE against anti D sera.
- Rh positive 85%
- Rh negative 15%

Penetrance and expressivity:

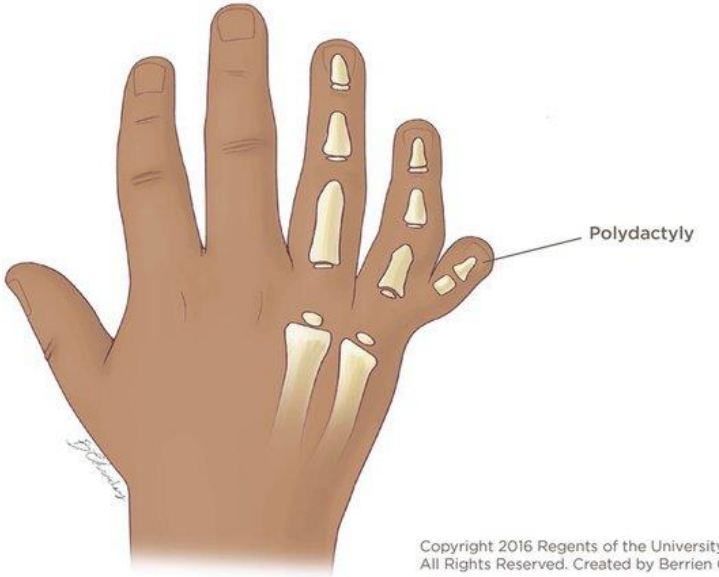
- The percentage of individuals that shown a particular phenotype among those capable of showing it.

- Penetrance is age related and is affected by environmental and behavioral factors such as diet and smoking.

- It is also modified by other genes and epigenetic regulation.

- Penetrance refers to the probability of a gene or trait being expressed.
  In some cases, despite the presence of a dominant allele, a phenotype may not be present.

  Eg: polydactyly in humans (extra fingers and/or toes).



- **Polydactyly - Dominant gene**
- Homozygous recessive - no polydactyly

- Heterozygotes - few are not polydactylus (If 20 %)
  So, genes has a penetrance → 80 %

- Degree of expression of a trait is controlled by a gene.

A particular gene may produce different degrees of expression in different individuals → **Expressivity.**

- A phenotype that is not genetically controlled but looks like a genetically controlled phenotype → **Phenocopy.**

- **An environmentally induced phenotype.**
  Eg: Vit-D resistant rickets

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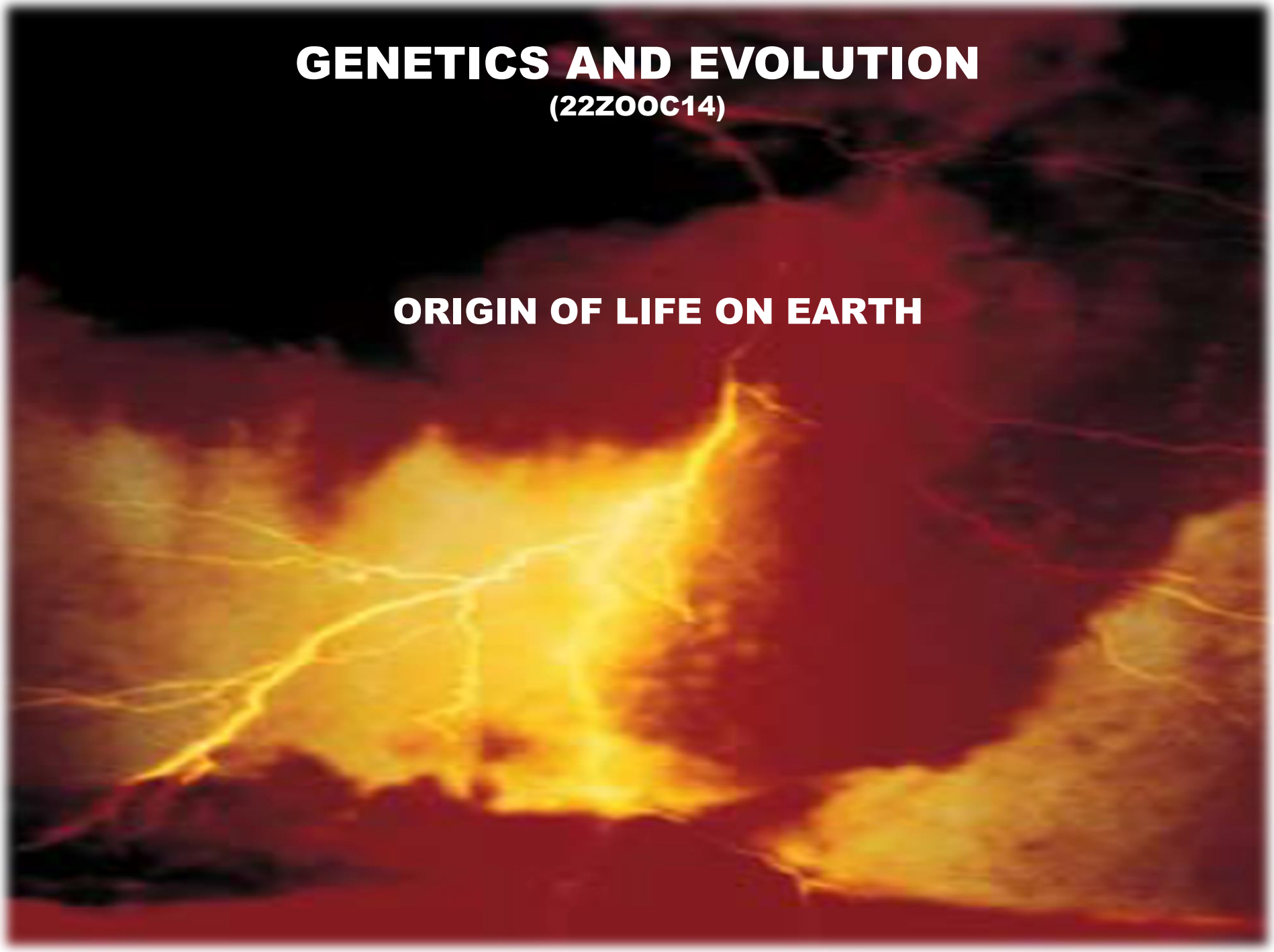
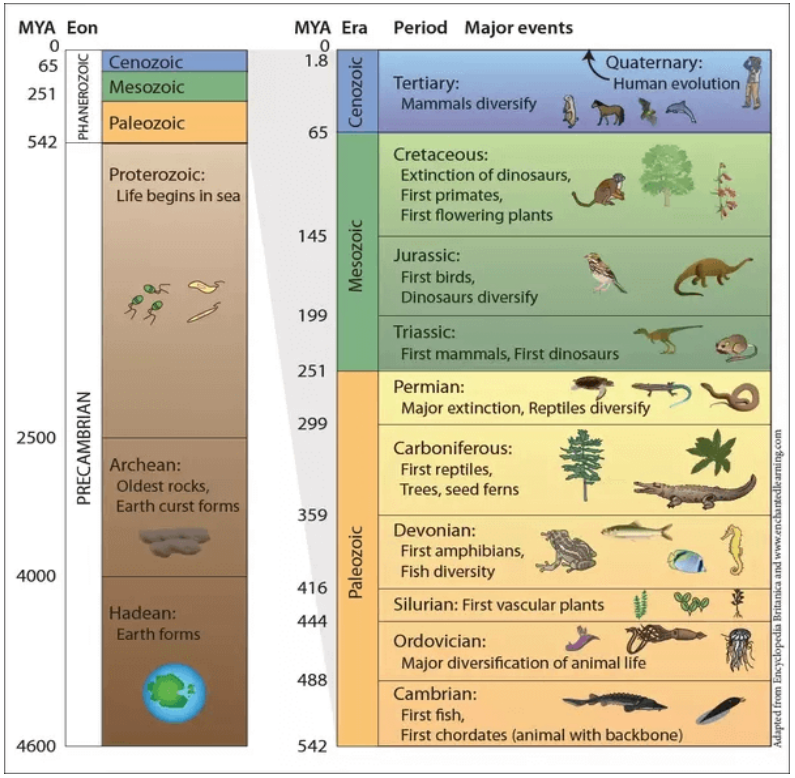


ORIGIN OF LIFE ON EARTH

GEOLOGIC TIME SCALE



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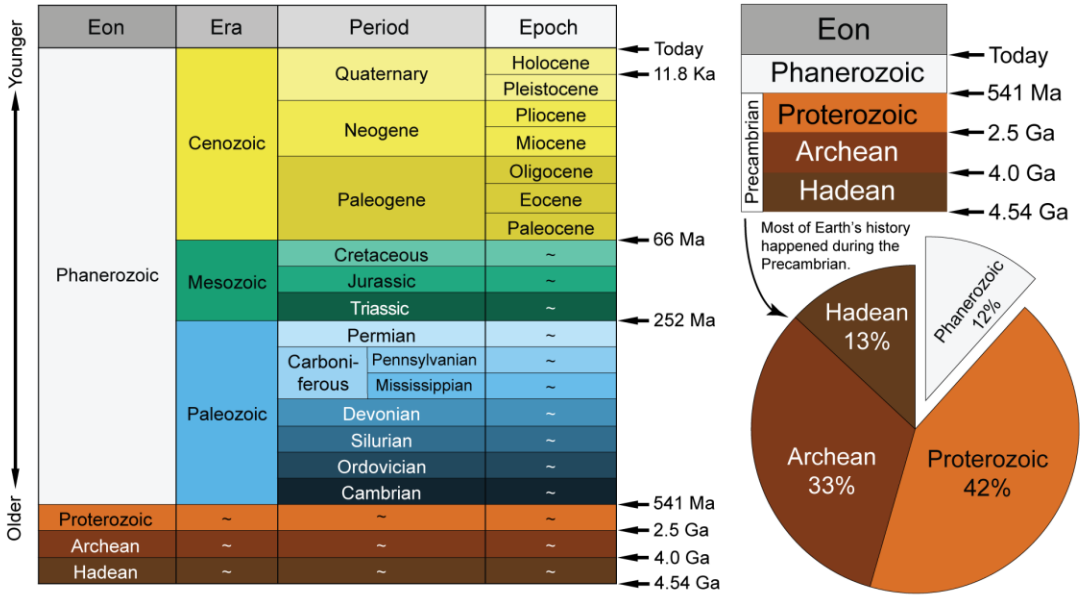


https://earthathome.org/wp-content/uploads/2022/03/Geologic-Time-Scale-2000px.png

MASS EXTINCTIONS

A mass extinction is a sharp spike in the rate of extinction of species caused by a catastrophic event or rapid environmental change. Scientists have been able to identify five mass extinctions in Earth's history, each of which led to a loss of more than 75 percent of animal species.

NATIONAL GEOGRAPHIC

1. ORDOVICIAN-SILURIAN EXTINCTION 440 MILLION YEARS AGO (MA)

86% LOSS

Scientists theorize that there were two main phases to this extinction: a glaciation event and a heating event. Abundant plant life removed carbon dioxide (CO_2) from the air, causing global cooling and glacier formation. This led to a drop in sea levels, reducing habitat. Later came global warming and sea level rising again. Creatures that had adapted to the cooler climate were unable to survive the increased temperature. Since most fauna was marine at the time, 86% of life was lost.

2. LATE DEVONIAN EXTINCTION—365 MA

75% LOSS

About 75% of life died off during this period. One theory suggests that land plants developed deep roots, releasing an abundance of nutrients into the oceans that fed algae. Because of this, algae blooms consumed vast amounts of oxygen (O_2) in the oceans, suffocating many species. Another theory suggests that another global cooling took place, resulting in glaciation and a fall in sea level, leading to habitat loss.

3. PERMIAN-TRIASSIC EXTINCTION—252 MA

96% LOSS

The Permian-Triassic was the deadliest extinction in history: 96% of all life perished. Scientist believe that volcanic activity in Siberia put massive amounts of carbon dioxide, a greenhouse gas, into the atmosphere. Bacteria that thrive on CO_2 began producing methane, another greenhouse gas. Large quantities of both gases warmed the planet and combined with Earth's water, making the ocean and rain acidic, creating a highly toxic environment for life.

4. TRIASSIC-JURASSIC EXTINCTION—201.3 MA

80% LOSS

Some scientists theorize that volcanic eruptions spewed tons of CO_2 into the atmosphere, which trapped heat and acidified the oceans, causing this mass extinction. Other scientists contend that an asteroid or comet impact triggered the extinction. About 80% of life was lost in this extinction, including most of the mammals, making way for the dinosaur's ancestors.

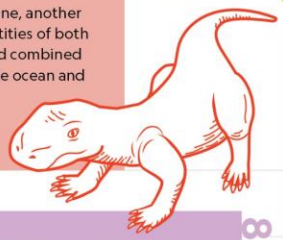
5. CRETACEOUS-PALEOGENE EXTINCTION—66 MA

60-76% LOSS

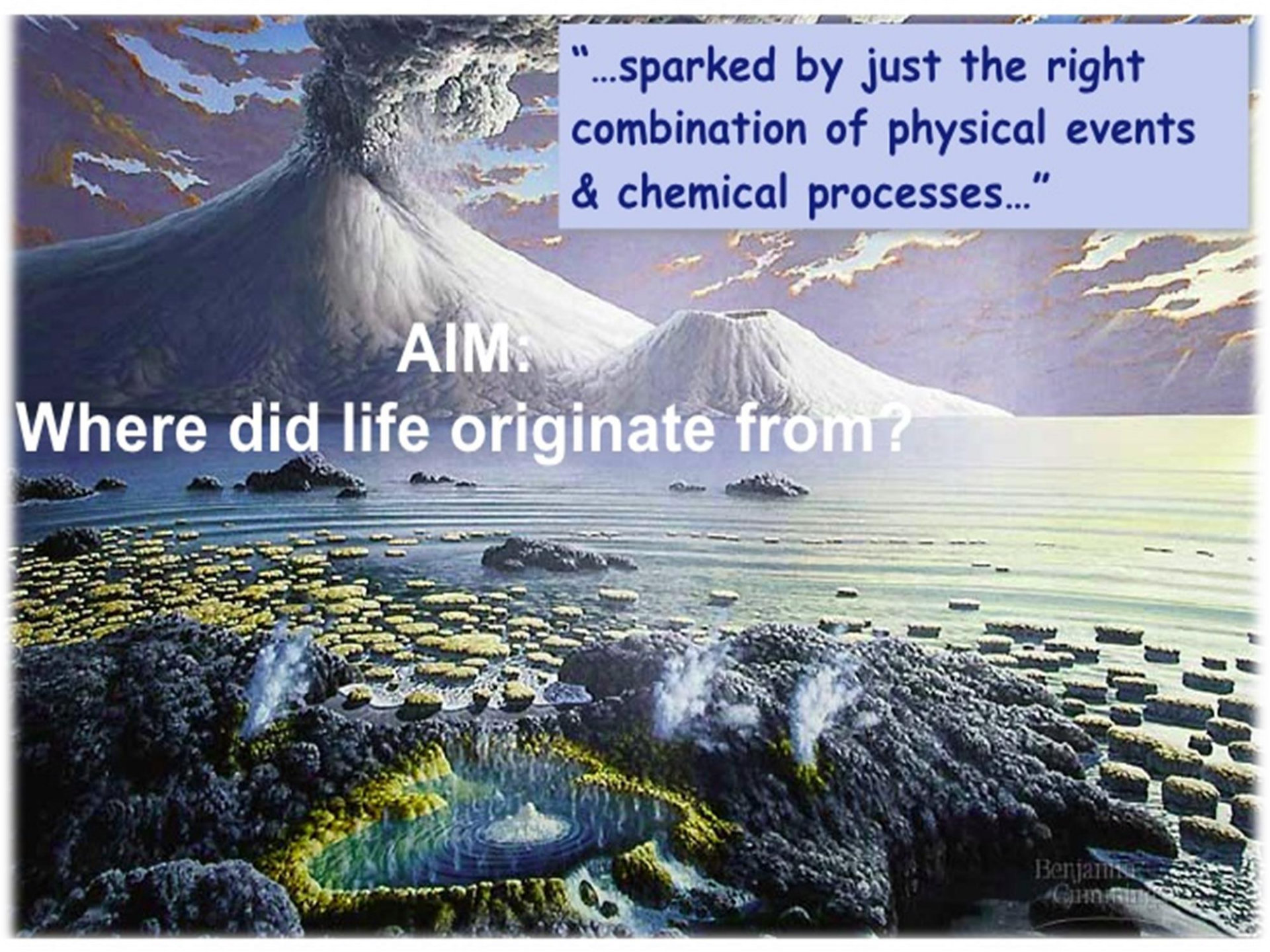
The Cretaceous-Paleogene extinction wiped out the dinosaurs, along with 60–76% of all life on Earth. A widely accepted theory is that an asteroid landed in the Yucatán Peninsula in Mexico and killed the dinosaurs. The impact would have ejected enormous amounts of debris into the atmosphere, causing global temperatures to drop. The impact may have also caused local fires, earthquakes, tsunamis, and acid rain.

6. HOLOCENE EXTINCTION 11,700 YEARS AGO TO PRESENT

The next mass extinction may already be happening. The current extinction rate is at least a thousand times greater than the "normal" extinction rate. A "normal" or background extinction rate is the average rate of extinction based on the longevity of species through time without human influence, determined by the fossil record. Scientists believe that human impact on the environment is the leading cause of extinctions today.



➢ **Extinction – types and role**


"…sparked by just the right combination of physical events & chemical processes…"
AIM:
Where did life originate from?

EARTH -- Hot mass of molten rock (4.6 bya)

↓

Earth cooled

↓

Water vapour **condensed – accumulated** surface in **chemically rich oceans**

↓

Hot smelly soup -- ammonia, formaldehyde, formic acid, cyanide, methane, hydrogen sulphide and organic hydrocarbons

How did organisms evolve from the complex molecules that swirled in the early oceans?

- **Hubble Space Telescope** - the earth itself was formed about **4.6 byrs**.

(NASA named the world's first space-based optical telescope after American astronomer Edwin P. Hubble (1889 -- 1953). Dr. Hubble confirmed an "expanding" universe, which provided the foundation for the big-bang theory.)

- The **oldest** clear evidence of life—**microfossils** (ancient rock) — **3.5 byrs old**. The **combination of physical events and chemical processes**.

The Hubble Space Telescope is named in honor of astronomer Edwin Hubble. Mt. Wilson Observatory near Pasadena, California

Ref: https://www.nasa.gov/

All living things share **key characteristics**.

What Is Life?

Movement
Sensitivity
Death
Complexity

} Inadequate

Cellular organization
Growth
Reproduction
Regulation
Homeostasis

} Inadequate Eg: soap bubble

- A mechanism for the preservation of improvement - **Heredity.**
- **Genetic system** - **adaptation** and **evolution over time**.
  - distinguishing characteristics of living things.

There are many ideas about the **origin of life**.

- Impossible to go back in time;
- No witnesses.

There are, in principle, at least three possibilities:

1. **Special creation:**

   Life-forms may have been put on earth by **supernatural** or **divine forces**.

2. **Extraterrestrial origin (Theory of Panspermia):**

   Life may not have originated on earth at all; instead, life may have **infected earth** from some other planet.

3. **Spontaneous origin:**

   Life may have evolved from **inanimate matter,** as associations among molecules became more and more complex.

Did Life Originate at the Ocean's Edge?

- **Reducing atmosphere hypothesis (Oparin in 1924)**

- In an atmosphere with **amino acids and sugars** react **spontaneously** with the oxygen to form **CO2 and H2O**.

Therefore, the building blocks of life, the **amino acids, would not last long** and the spontaneous formation of complex carbon molecules could not occur.

(Assumption – little oxygen around)

- Our **atmosphere changed** once organisms began to carry out **photosynthesis**, used energy in sunlight to split water molecules and form complex carbon molecules, giving off gaseous oxygen molecules.

The earth's atmosphere is now approximately **21% oxygen**.

Drawback: Rocks and ozone were not discussed / formed in this theory.

Early atmosphere contained principally

- carbon dioxide (CO2) and
- nitrogen gas (N2),

along with significant amounts of

- water vapour (H2O).
- hydrogen gas (H2) and

Hydrogen + (sulfur, nitrogen, and carbon)

Compounds: **hydrogen sulfide (H_2S), ammonia (NH_3), & methane (CH_4).**

↓

Earth's first organisms emerged & lived at very high temperatures
49° to 88°C (120° to 190°F) (from 3.8 – 3.5 bya).

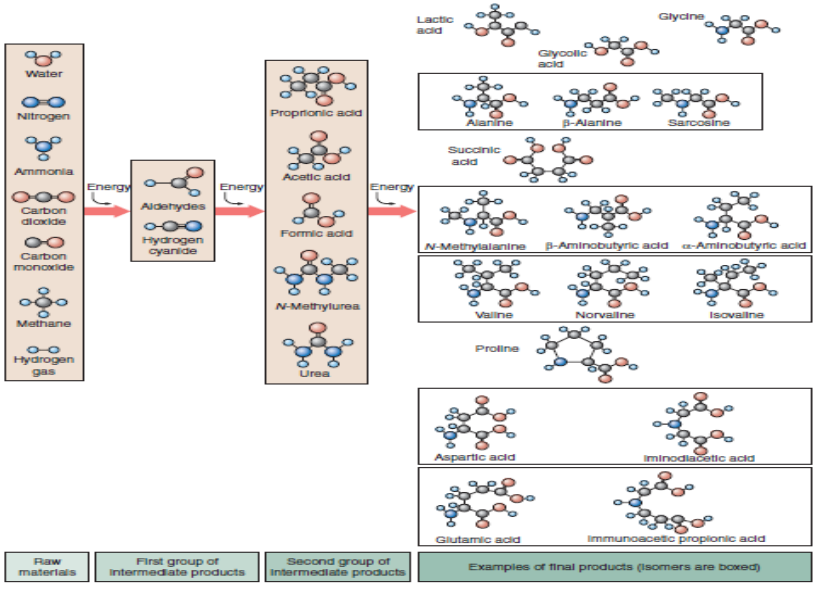
Other suggestions:
Under frozen oceans, Deep in earth's crust, within clay & deep sea vents.

Genomics - the ancestors of today's prokaryotes are most closely related to the bacteria that live on the deep-sea vents.

What kinds of organic molecules might have been produced on the early earth ?

- In 1953 by Stanley L. Miller and Harold C. Urey.
- They attempted to reproduce the conditions at **ocean's edge under a reducing atmosphere.**
- They found that within a week, **15% of the carbon** as
  - methane gas (CH4) as formaldehyde (CH2O) and hydrogen cyanide (HCN).
- Then combined to form simple molecules

  - **formic acid (HCOOH) and**
  - **urea (NH2CONH2)**
  - more complex molecules - **carbon-carbon bonds**, including the **amino acids** (glycine and alanine).



HCN were formed – contributed the formation of complex ring-shaped molecule called **adenine**—one of the **bases** found in DNA and RNA.

The origin of life concerns which organic molecules came first, RNA or proteins ??

Group I: "RNA world"

- "Without a hereditary molecule, other molecules could not have formed consistently".

- **Thomas Cech** at the University of Colorado discovered **ribozymes**, **RNA molecules** that can behave as **enzymes**, catalyzing their own assembly.

- **rRNA** catalyzes the chemical reaction that links **amino acids to form proteins**.

Origin of Genetics: **Dawn of Natural Selection**

RNA is likely the first genetic material

1. Multifunctional
2. Codes information (Self replicating, Inheritance, Natural selection and Evolution)
3. Enzyme functions
4. Transport molecule (tRNA and mRNA)

Group II: "Protein world"

- **Without enzymes** (which are proteins), nothing could replicate at all, heritable or not.
- Nucleotides, the individual units of nucleic acids such as RNA, are too complex to have formed spontaneously.

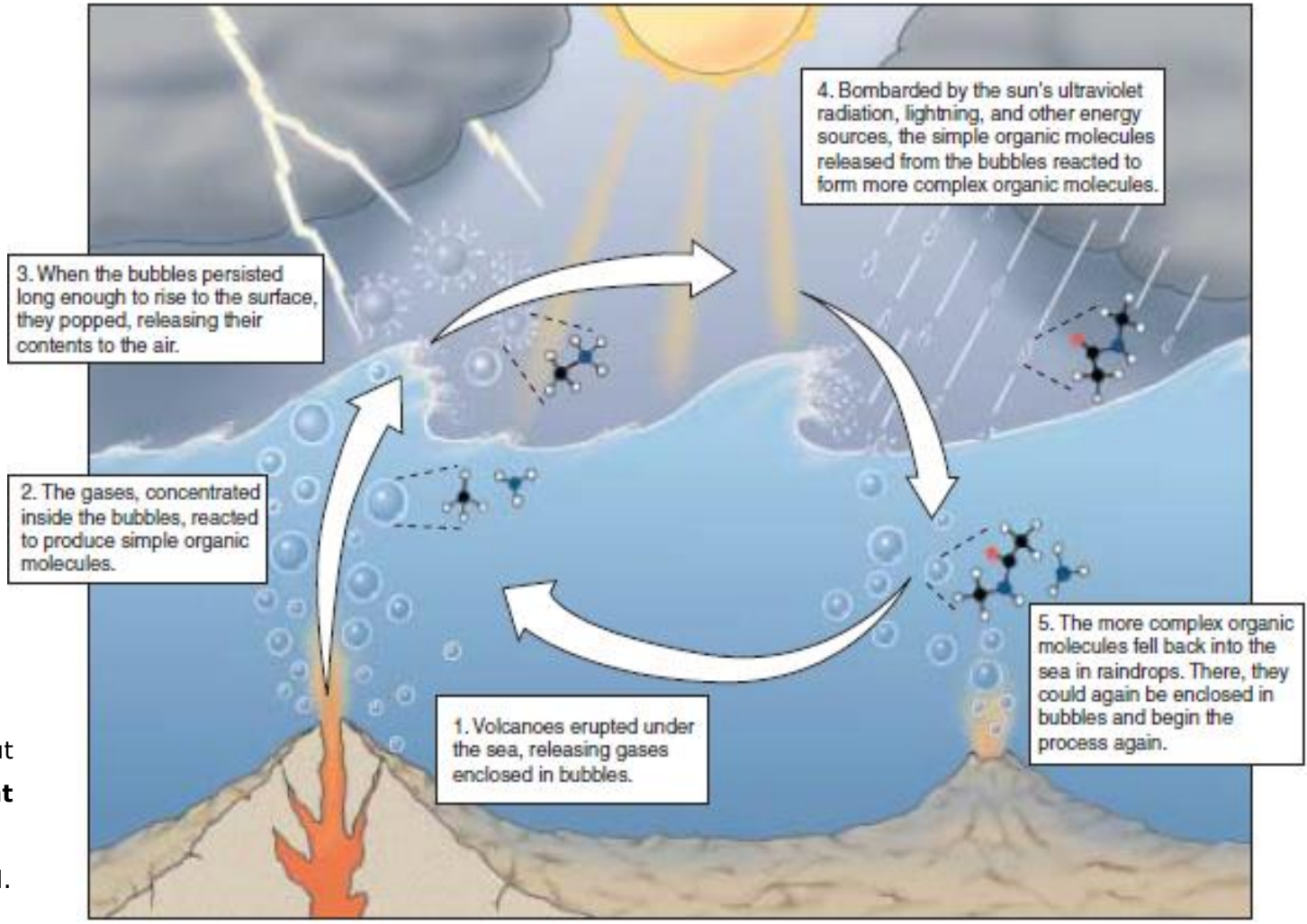
Group III: "Peptide-Nucleic Acid (PNA) World"

- It is an another important and popular theory .
- The **first organic molecules** assumes key roles for both **peptides** and **nucleic acids**.
- Because RNA is so complex and unstable, this theory assumes there must have been a **pre-RNA world** where the **peptide-nucleic acid (PNA)** was the basis for life.
- **PNA** is stable and simple enough to have formed spontaneously, and is also a **self-replicator**.

Current bubble hypothesis:

In **1986** geophysicist, **Louis Lerman** proposed that

"the chemical processes leading to the evolution of life took place within bubbles on the ocean's surface".



Inference:

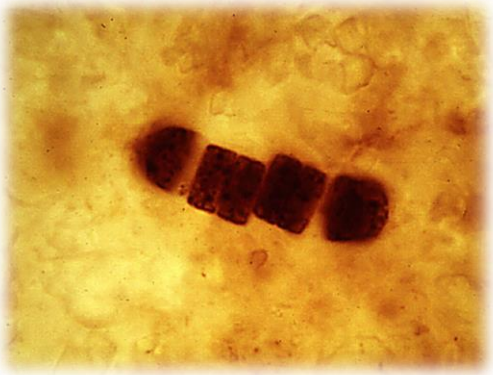
Current hypotheses involve **chemical evolution within bubbles**, but there is **no general agreement** about their composition, or about how the process occurred.

The Earliest Cells (Microfossils):

- The **fossils found in ancient rocks** show an obvious progression from **simple to complex organisms**, (3.5 billion years ago).

- **Life may have been present earlier,**

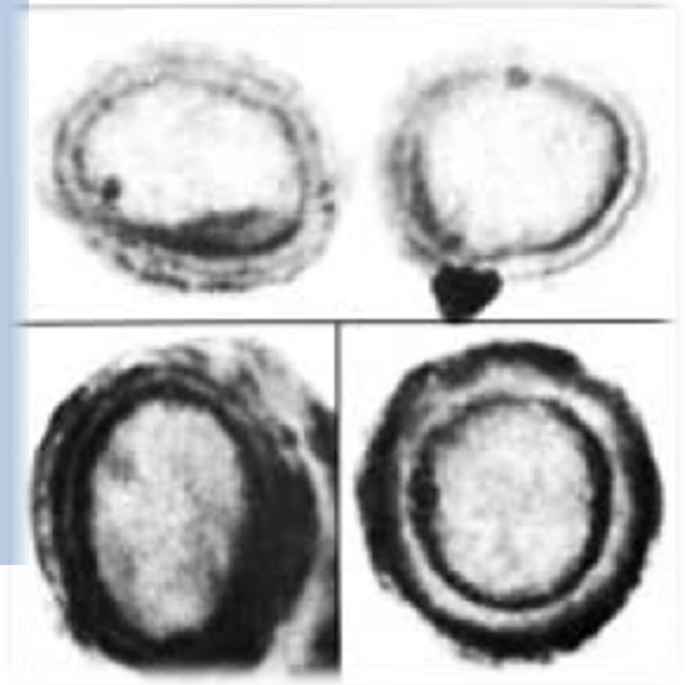
but rocks of such great antiquity are rare, and fossils have not yet been found in them.



Cross-sections of fossil bacteria.

These **microfossils** from the **Bitter Springs formation** of **Australia** are of ancient cyanobacteria.

In this electron micrograph, the cell walls are clearly evident.



Cyanobacteria played an important role in the evolution of Early Earth and the biosphere. They are responsible for the **oxygenation of the atmosphere and oceans** since the **Great Oxidation Event** around 2.4 bya, debatably earlier.

They are also major **primary producers** in **past and present oceans, and the ancestors of the chloroplast**.

Cyanobacteria evolution: Insight from the fossil record.

Demoulin CF[1], Lara YJ[1], Cornet L[2], François C[1], Baurain D[3], Wilmotte A[4], Javaux EJ[1]

Author information

Free Radical Biology & Medicine, 09 May 2019, 140:206-223
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Review Free to read & use

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Shark Bay

See & Do Plan Sleep Play Our Region What's On About Us

Stromatolites

Shark Bay is one of only two places in the world where living marine stromatolites exist. These ancient structures are examples of what life on Earth was like 3.5 billion years ago and are considered living fossils - the earliest record of life on earth.

➢ Stromatolites became more common 2.5 billion years ago, they **gradually changed the Earth's atmosphere** from a carbon dioxide-rich mixture to the present-day **oxygen-rich atmosphere**.

This major change paved the way for the next evolutionary step, the **appearance of life based on the eukaryotic cell (cell with a nucleus).**

Modern stromatolites (columns and mounds) in Shark Bay (Western Australia)

Stromatolites

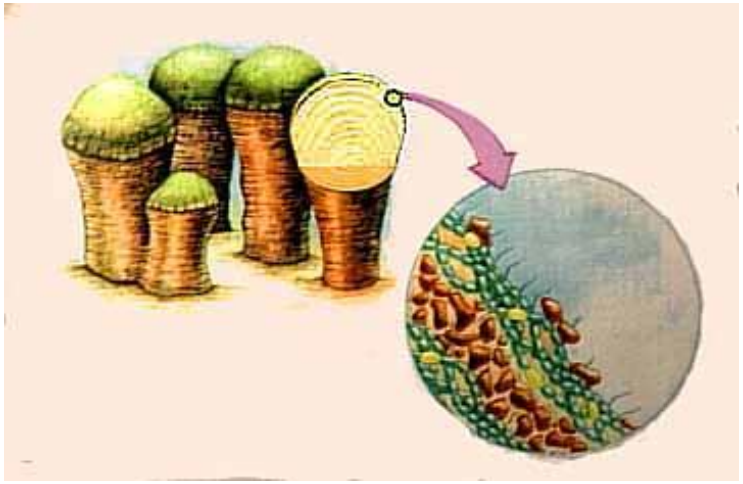
Shark Bay

- Layered mounds, columns, and sheet-like sedimentary rocks.

- They were originally formed by the growth of layer upon **layer of cyanobacteria**, a single-celled photosynthesizing microbe that lives today in a wide range of environments ranging from the shallow shelf to lakes, rivers, and even soils.

- **Cyanobacteria are prokaryotic cells (the simplest form of modern carbon-based life) in that they lack a DNA-packaging nucleus.**

Bacteria, including the photosynthetic cyanobacteria, were the only form of life on Earth for the first 2 billion years that life existed on Earth.



Banded Iron Formation

Ferrous (Fe2+) reduced layer

The bands of finely layered iron-rich sediment from which it is formed. The different bands of colour in the BIF slab are iron oxides and red and brown chert, and golden-brown tiger eye (a type of fibrous quartz).

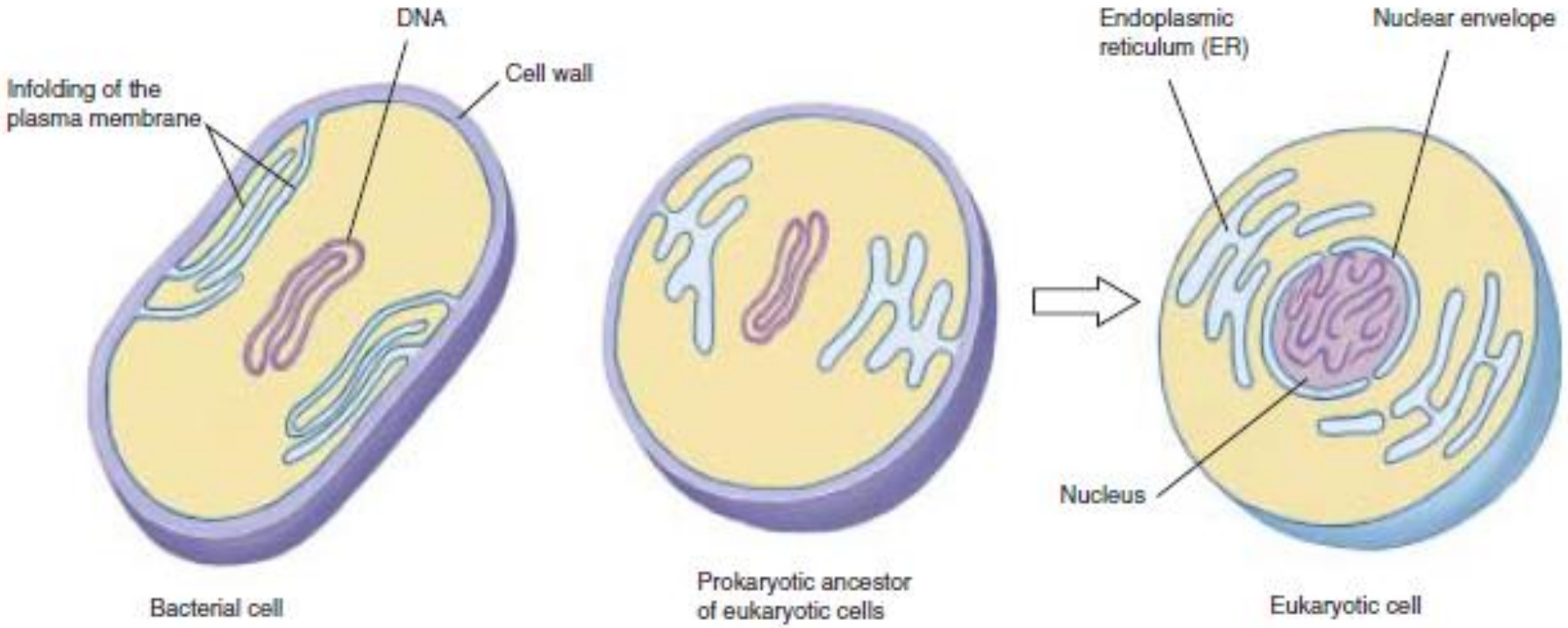
- **They were deposited on the ancient seafloor (2700 to 2400 million years ago) by the action of the earliest life forms on Earth.**
- **Masses of cyanobacteria in the ocean photosynthesised, producing oxygen like many modern plants.**
- **The accumulation of oxygen in seawater caused the iron present to oxidize.**
- **The ocean 'rusted' and iron-rich minerals were deposited to form BIFs.**

Early life continued to produce oxygen that enriched the atmosphere and set scene for the environment we know today, the basis for life on Earth.

Ferric (Fe3+) oxidized layer

ORIGIN OF THE NUCLEUS AND ENDOPLASMIC RETICULUM

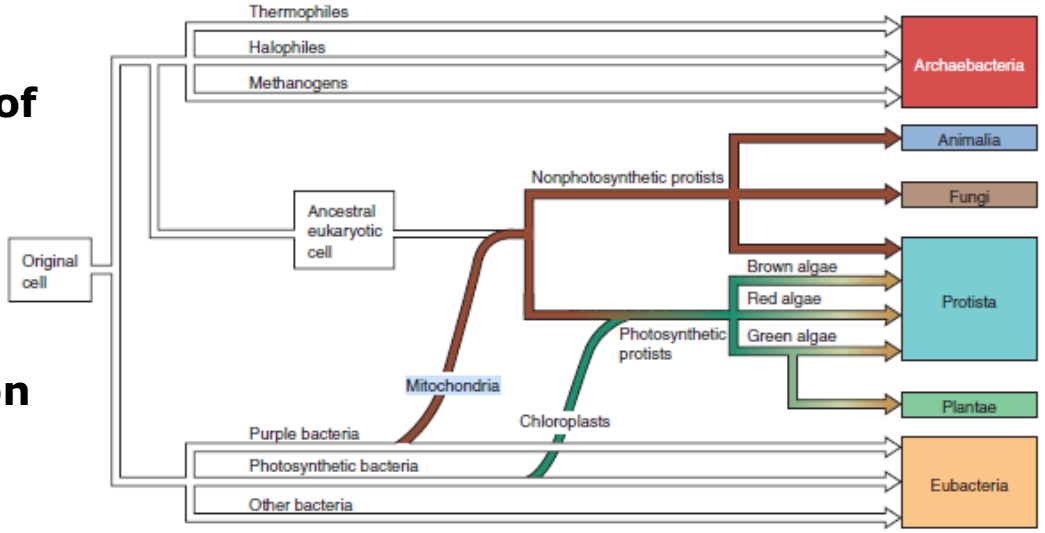
Many bacteria today have infoldings of the plasma membrane

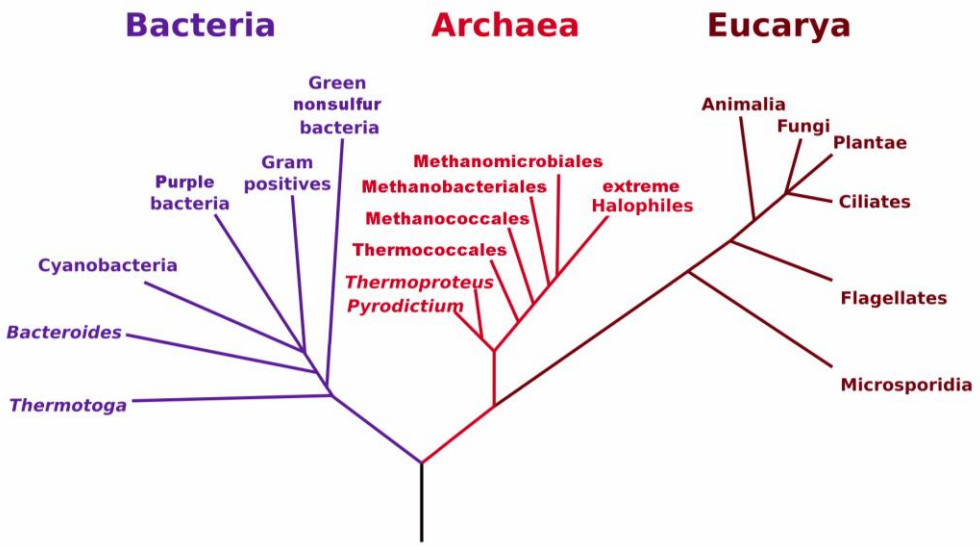


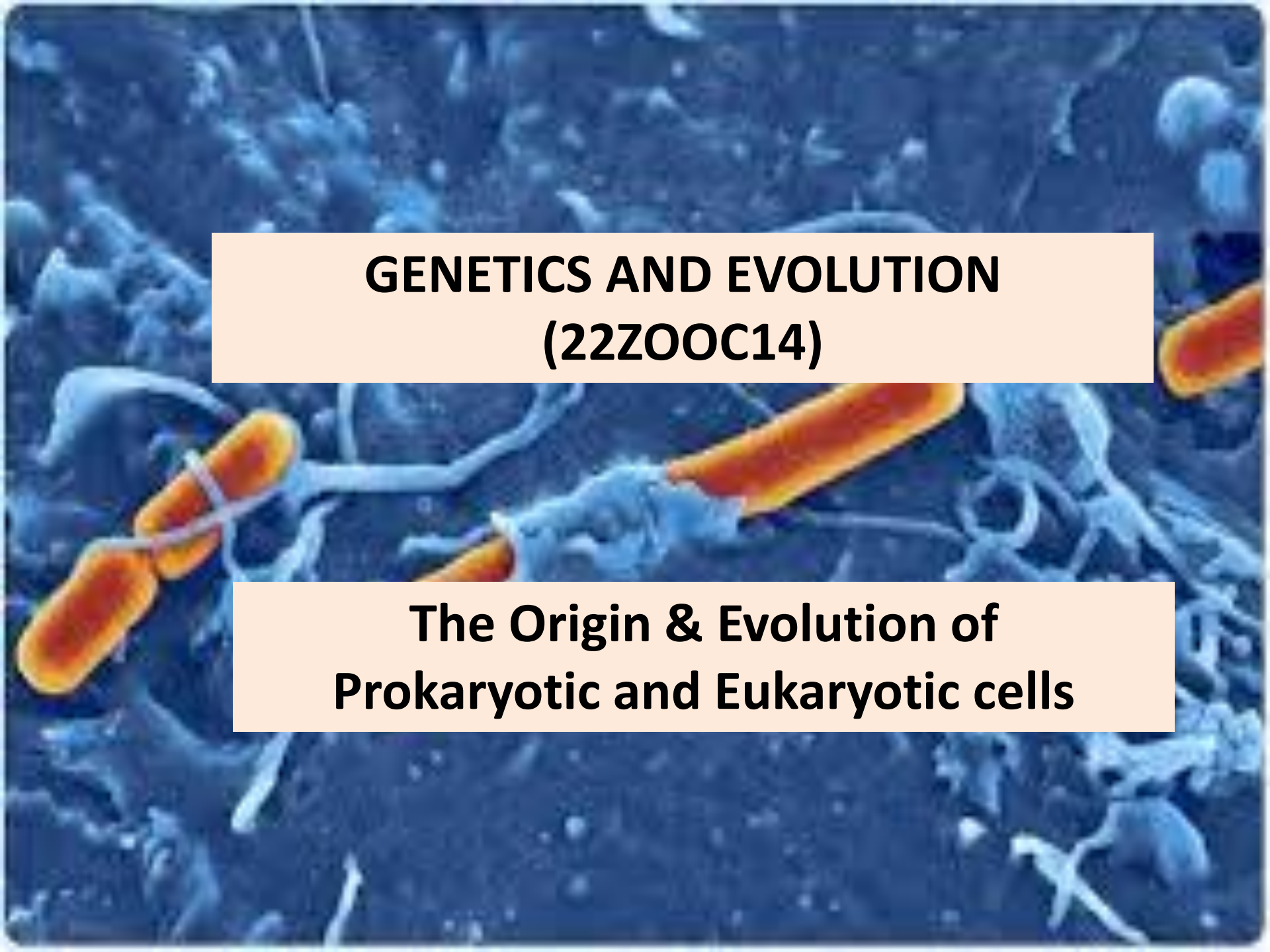
The eukaryotic internal membrane system called the endoplasmic reticulum (ER) and the nuclear envelope may have evolved from such infoldings of the plasma membrane encasing prokaryotic cells that gave rise to eukaryotic cells.

The **tree of life** or **universal tree of life** (UToL) (Doolittle 2009)

A **model** and **research tool** used to **explore the evolution** of **life** and describe the **relationships** between organisms, both **living and extinct –** Charles Darwin's On the Origin of Species (1859).







GENETICS AND EVOLUTION (22ZOOC14)

The Origin & Evolution of Prokaryotic and Eukaryotic cells

Ital. J. Zool., *64*: 107-113 (1997)

On the symbiotic origin of protists, their diversity, and their pivotal role in teaching systematic biology

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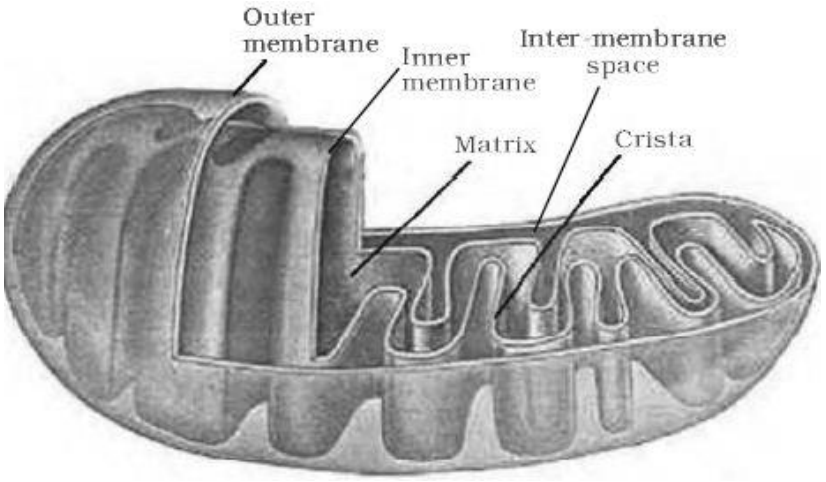
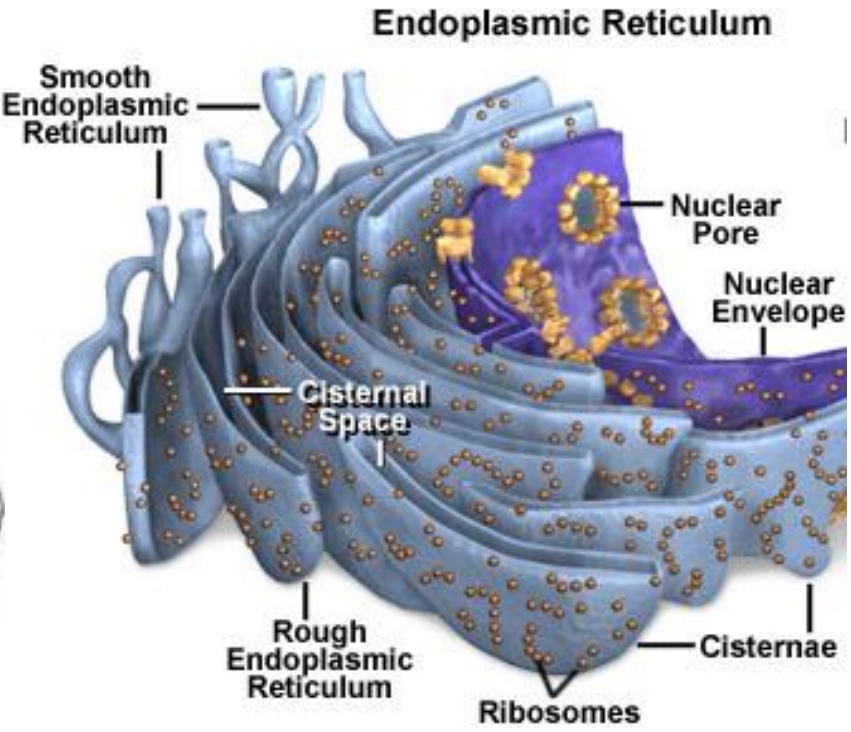
Auf der Morgenstelle 28, D-72076 Tübingen (Germany)

ORGANELLES

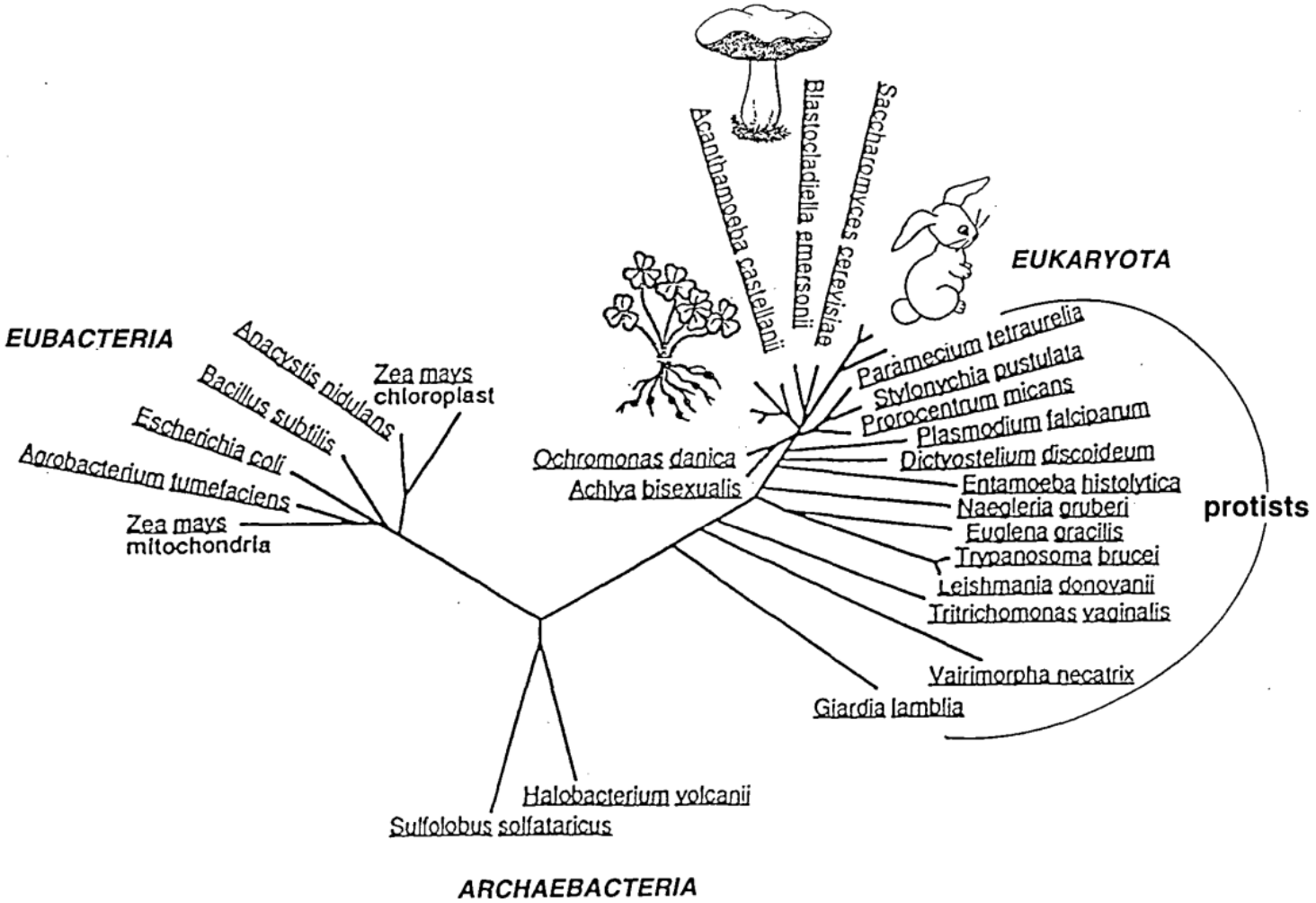
Compartmentalization - specific reaction sequences and pathways

- Transcription in Nucleus,
- Translation in cytoplasm
- Aerobic metabolism in mitochondria
- Photosynthesis in chloroplasts





All multicellular organisms, animals, fungi and plants are derived from unicellular protists which were the first eukaryotes on earth.



MODIFIED SOGIN-TREE SHOWING THE RESULTS OF COMPARATIVE SEQUENCING OF SMALL SUBUNIT RIBOSOMAL RNA (SOGIN, 1989)

A **single-celled eukaryote** whose life-styles in some ways **resemble those of their prokaryotic forebears**, and which are thought to be among the **earliest eukaryotes that are still in existence.**

One single-celled organism, the intestinal parasite ***Giardia lambida***, represents the first line of descent from the ancestral cells that took on eukaryotic features.

(Missing Link between prokaryotes and Eukaryotes)

Current views on the symbiotic origin of **protists (single celled eukaryotes)** and their **organelles.**

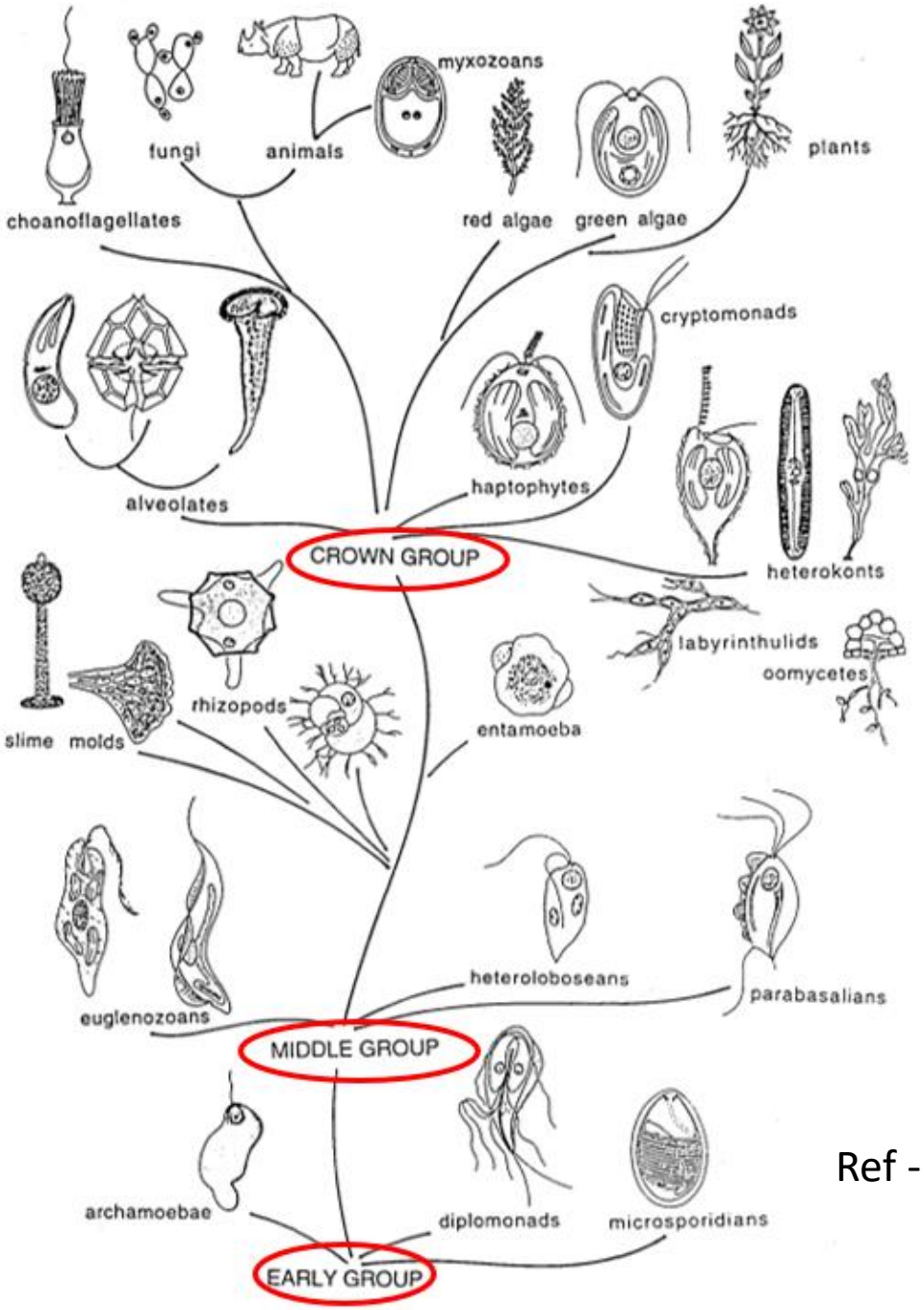
(**nucleus**, **9 + 2 flagellum**, **mitochondria** and **plastids**) are reviewed with particular reference to **extant model organisms** from phototrophic **dinoflagellates** and **cryptomonads** which are examples of "**eukaryotes within eukaryotes**".

- Protists are cayegorized into three major groups.
  - **Early group:** Lack mitochondria primarily and is represented by the Microspora, Giardia, certain amoeboflagellates, trichomonads and polymastigotes.

  But recent discovery of mitochondrial chaperons throws some doubts on this view.

  - **Middle group:** It contains the Euglenozoa (kinetoplastids, euglenoids) and the majority of the former rhizopods.

  - **Crown group:** Due to a rapid radiation, gave rise to
    - the alveolates (apicomplexean sporozoans and dinoflagellates → forming together the sister-group of the ciliates),
    - heterokont algae (brown algae, plus oomycetes and netslime molds, cryptomonads, haptomonads, and
    - three main branches with multicellular organisms - plants and animals and true fungi, the latter being the sister group of the animals).



Ref - The precise branching pattern in all three groups is still largely unsettled

EVOLUTION OF EUKARYOTIC ORGANELLES

The most well supported hypothesis ---

Eukaryotic cells evolved by physically incorporating entire prokaryotic organisms into their cytoplasm

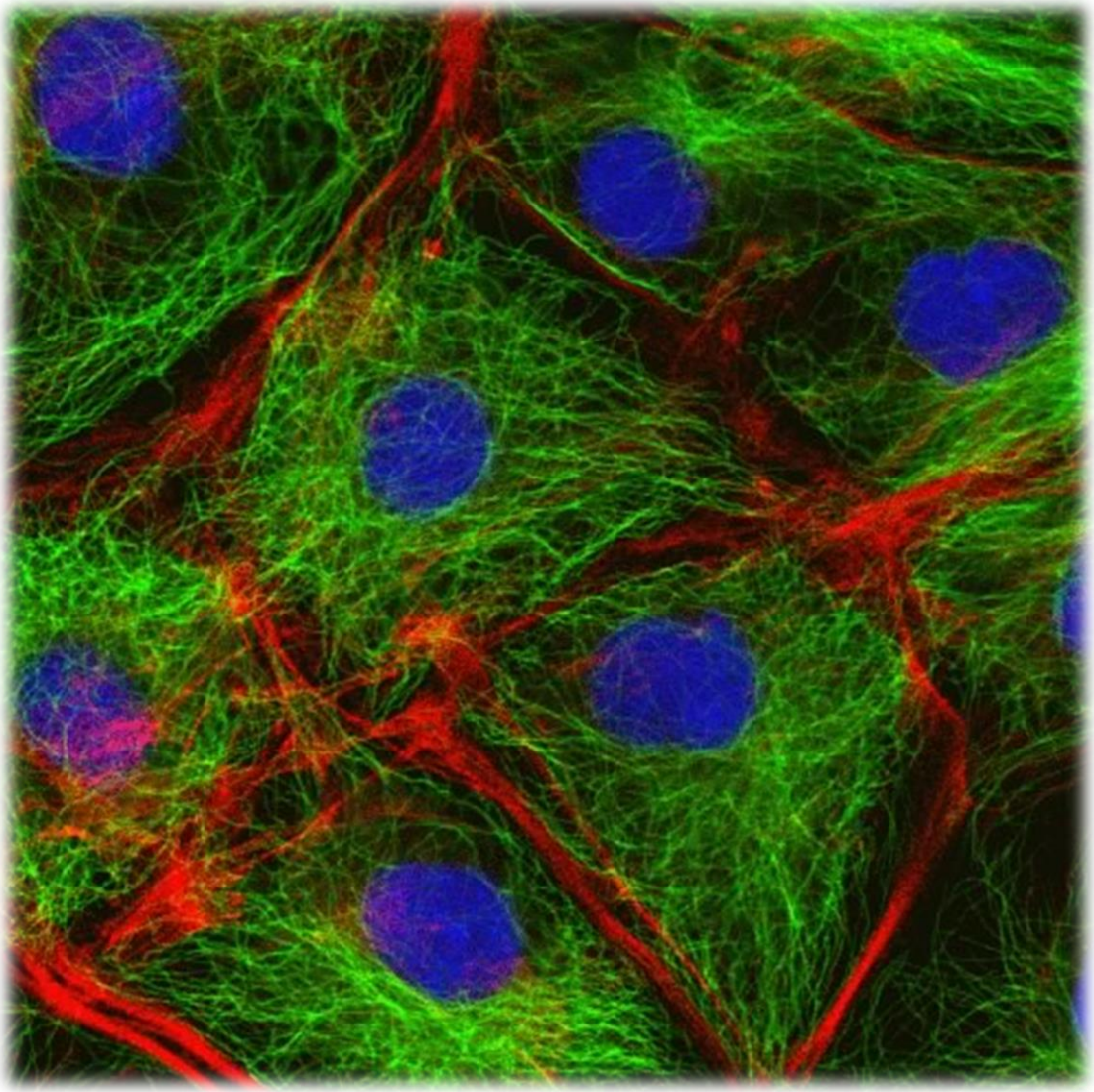
ENDOSYMBIOTIC THEORY

- There are evidences for the Origin of Mitochondria and chloroplasts from prokaryotic endosymbionts – **PRIMARY ENDOSYMBIOSIS .**

- Some eukaryotic algae acquired chloroplasts by **SECONDARY ENDOSYMBIOSIS** from a eukaryotic rather than from prokaryotic cell.

Cytoskeleton of eukaryotic cells:

- **Microfilaments (actin)**
- **Microtubules (tubulin)**

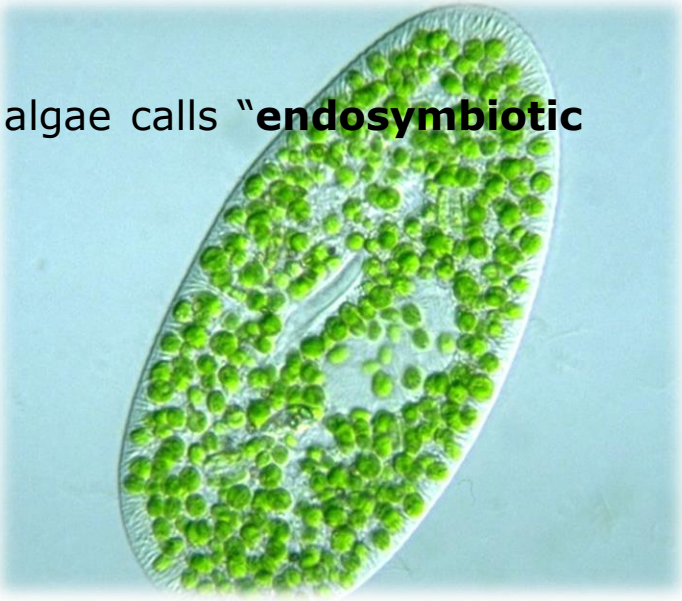


Paramecia are also good landlords – Endosymbiotic relationship and Endosymbionts

Some species of paramecium (*Paramecium bursaria* and *Paramecium chlorelligerum*) allow green algae (*Zoochlorella* or *Chlorella*) living inside its cytoplasm and provide the paramecium cell (the host) with nutrients produced by photosynthesis.

At the same time, paramecium provides the algae with movement and protection, as well as carbon dioxide and nitrogen components which are needed for photosynthesis.

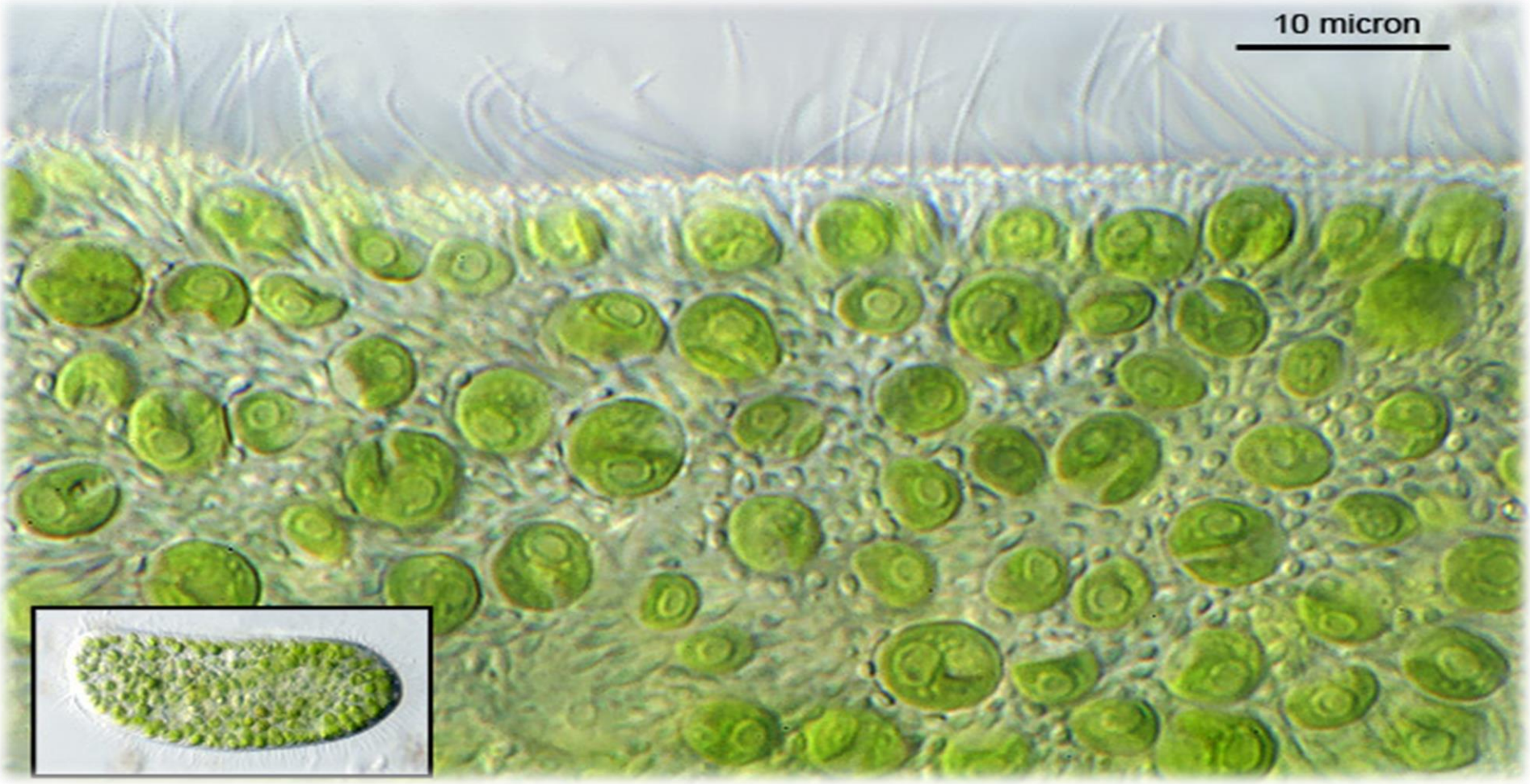
This mutualistic relationship between paramecium and algae calls "**endosymbiotic relationship**".



The cytoplasm of ***Paramecium bursaria*** harbors hundreds of symbiotic algae (Chlorella)

How endosymbiotic relationship start

The endosymbiotic relationship initiates when the *P. bursaria* cell swallows the green algae by phagocytosis. Rather than digests, the host paramecium stores the symbiotic algae in vacuoles as endosymbionts. When the paramecium moves towards areas of greater light intensity, algal photosynthesis supplies each partner with photosynthetic nutrients.

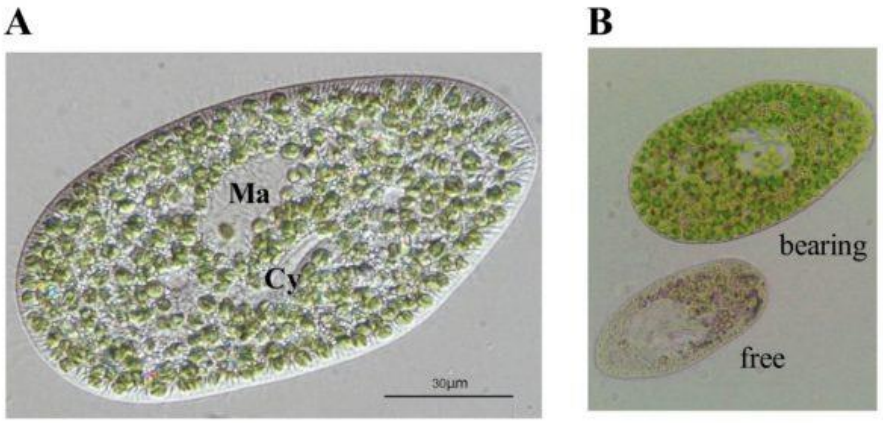


The endosymbiotic relationship between paramecium and algae is facultative, but not obligate mutualism.

P. bursaria and *Zoochlorella* can survive without the others.

However, algae-free *P. bursaria* are rare in nature.

P. bursaria growth is enhanced in cells harboring algal symbionts compared to algae-free cells.



(A) Microscope image of a typical *P. bursaria* cell. "Ma" is macronucleus; "Cy" is cytopharynx.

(B) Microscope images of algae-bearing and algae-free *P. bursaria*.

The benefit of endosymbiotic relationship

Interestingly, endosymbiotic algae also protect their host paramecia from predators. One of the well-studied predators of paramecia, *Didinium nasutum*, tends to keep away from *P. bursaria* hosting endosymbiotic green algae. *D. nasutum* prefers *P. caudatum*, or *P. multi-micronucleatum* which don't have endosymbiotic partners. Scientists hypothesize that the endosymbiotic green algae within *P. bursaria* discourage predation by *D. nasutum* by releasing distasteful metabolites that repel them.

Much like roommates adapting to each other's schedule, the host paramecium and endosymbiotic algae have good communication and are able to synchronize with regards to the timing of cell division and growth. Endosymbiotic algae can even adjust the photosynthesis according to the circadian rhythms in the paramecium host.

Predator-prey Interaction Between *Paramecium caudatum* and *Didinium nasutum* 18

Mitochondrial DNA

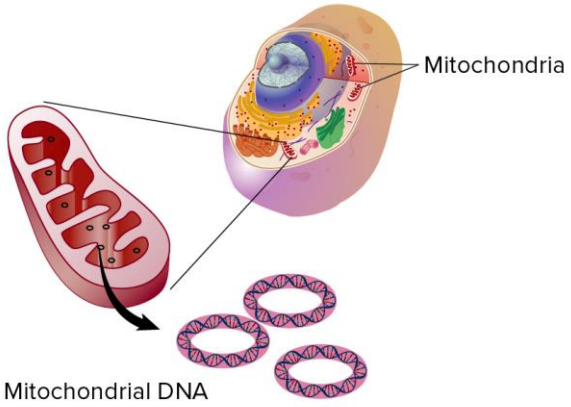
- mtDNA in a single mitochondria **never recombines with nuclear DNA nor with other mitochondria**

- It divides and produces thousands of its **own copies / cell.**

- Only a **single copy of mtDNA in each mitochondria is inherited** from a single parent --

- Egg (maternal only).

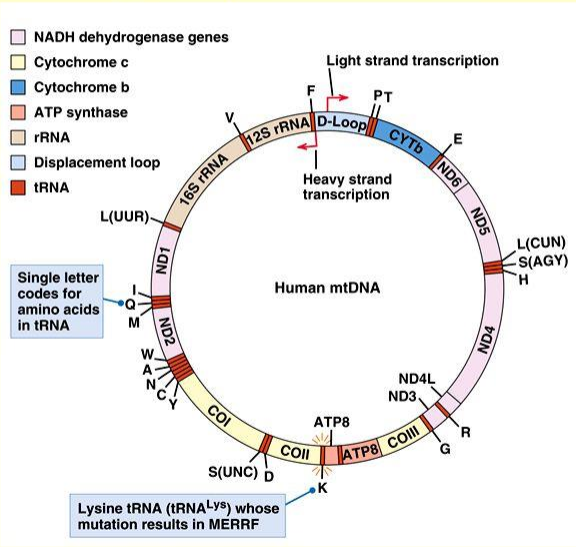
- As a result of **uniparental inheritance** of mitochondria, mutations are the source of difference among the mtDNA copies.



The Human Mitochondrial Genome

2 - 10 copies/mitochondrion

- Circular
- ~ 16 kb (some plants ~100 kb!)
- Crowded (~40 genes)
- 13 genes involved in oxidative phosphorylation + other genes (DNA pol, rDNAs, tRNAs)
- Most proteins in mitochondria are imported from cytoplasm
- 100,000 copies of mitochondrial DNA in ovum



ORIGINS OF INTRONS:

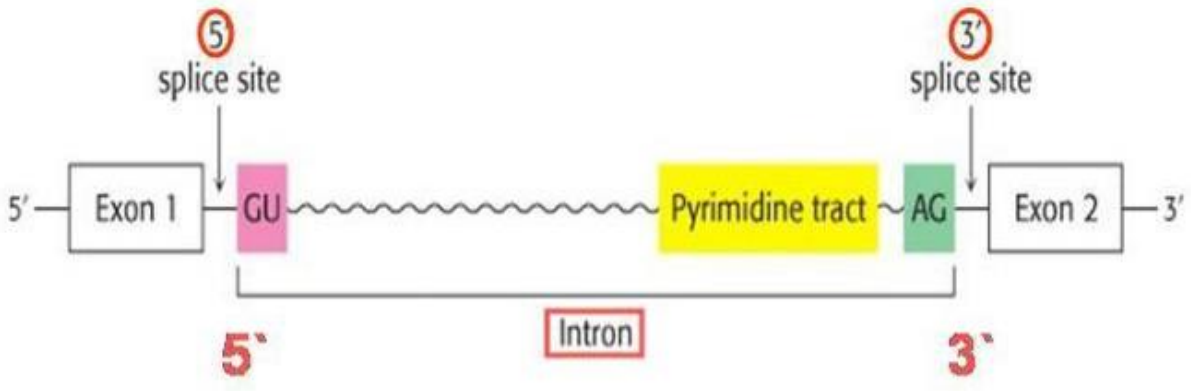
- Self-splicing introns evolved in the **RNA world** and have survived ever since without undergoing a great deal of change.
- Origins of GU-AG introns (found in eukaryotic nuclear pre-mRNA (protein-coding genes)).
- GU-AG rule: (GU-AG rule in pre-mRNA)

RNA intron splicing signals:

They are conserved

✓ An intron starts with dinucleotide GU and ends with AG.

✓ It is called 5' and 3' splice sites respectively.



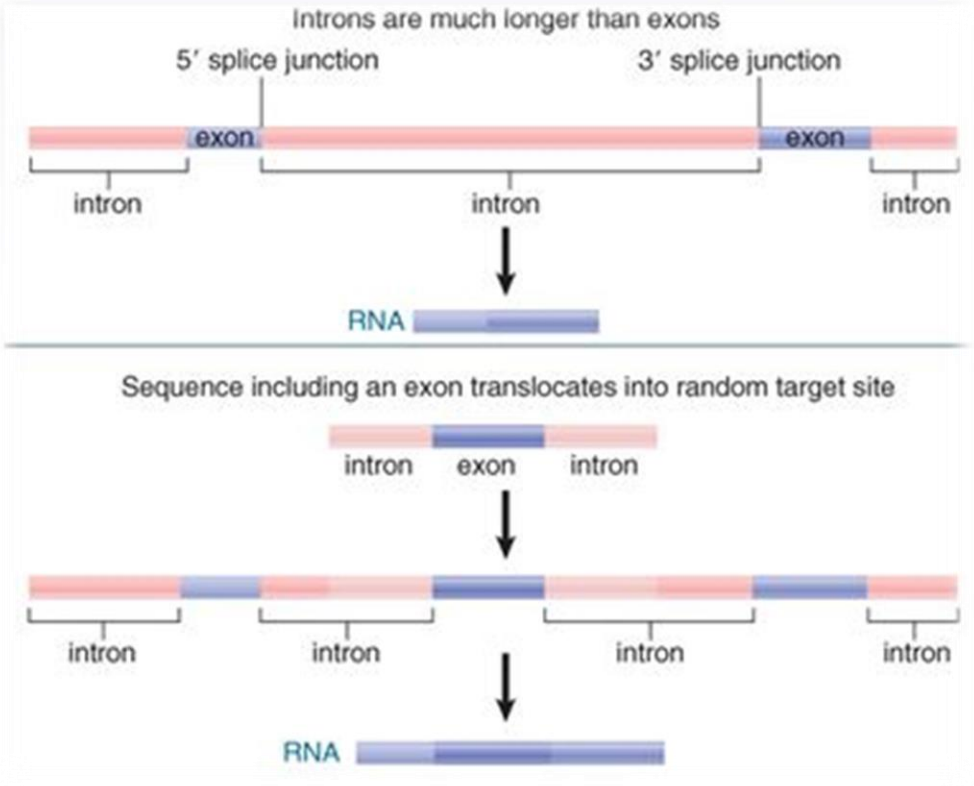
THEORIES OF INTRON ORIGIN: 2 Competing Theories

- 'Introns-late theory' assumes that all spliceosomal introns evolved recently and are gradually accumulating in eukaryotic genomes by insertion of group II introns or transposable elements.

- 'Introns-early theory' extended to the Exon theory of genes (states that introns are very ancient and are gradually being lost from eukaryotic genomes).

Both theories have observational support.

EVOLUTION OF INTERRUPTED GENE



- Interrupted genes that correspond either to proteins or t independently functioning nonprotein-encoding RNAs probably originated in an interrupted form (the **"introns early" hypothesis**).
- **exon shuffling –** The hypothesis that genes have evolved by the recombination of various exons encoding functional protein domains.

Centromere and Kinetochore

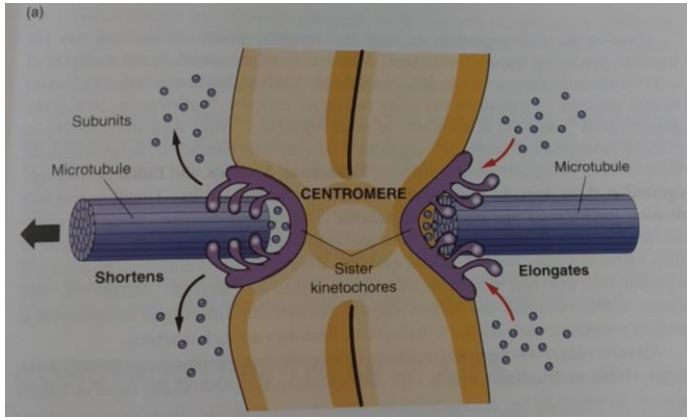
- Kinetochores are essential and universal features of **eukaryotic chromosomes** with a **conserved** set of functions. They are complex protein structures that are normally found at a unique location on each chromosome known as a **centromere** (often visible as a primary constriction in a metaphase chromosome).

- Kinetochores serve to attach chromosomes to **spindle microtubules** in mitosis and meiosis (orderly chromosome segregation). They help to **recruit cohesins** and work in partnership with them to hold sister chromatids together locally in order to generate spindle tension.

- They also carry out the **spindle assembly checkpoint pathway**, which assures that all kinetochores are attached to spindle fibers **before commencing anaphase**.

- The **last eukaryotic common ancestor (LECA)** possessed a complex kinetochore.

A highlight that many present day kinetochores have diverged through rapid sequence evolution, extensive gene loss, duplication, invention and displacement.



Centromere and kinetochore

DOI: 10.13140/RG.2.2.16846.56640

When and how did the centromere of the eukaryotic chromosome arise?

- The symbiotic partnership between an eubacteria inside an archaebacterial host gave rise to the common ancestor of eukaryotes.

- Initially, this partnership was a difficult one, and recurrent symbiont lysis resulted in a constant transfer of genes from the eubacterial genome into the chromosome of the host.

- Comprehensive analyses involving large numbers of genomes have confirmed that the eukaryotic nucleus contains a mix of genes of archaebacterial and eubacterial origins.

- The presence of numerous spliceosomal introns in the same positions from distant eukaryotes strongly suggests that introns arose at the earliest stages of eukaryotic evolution.

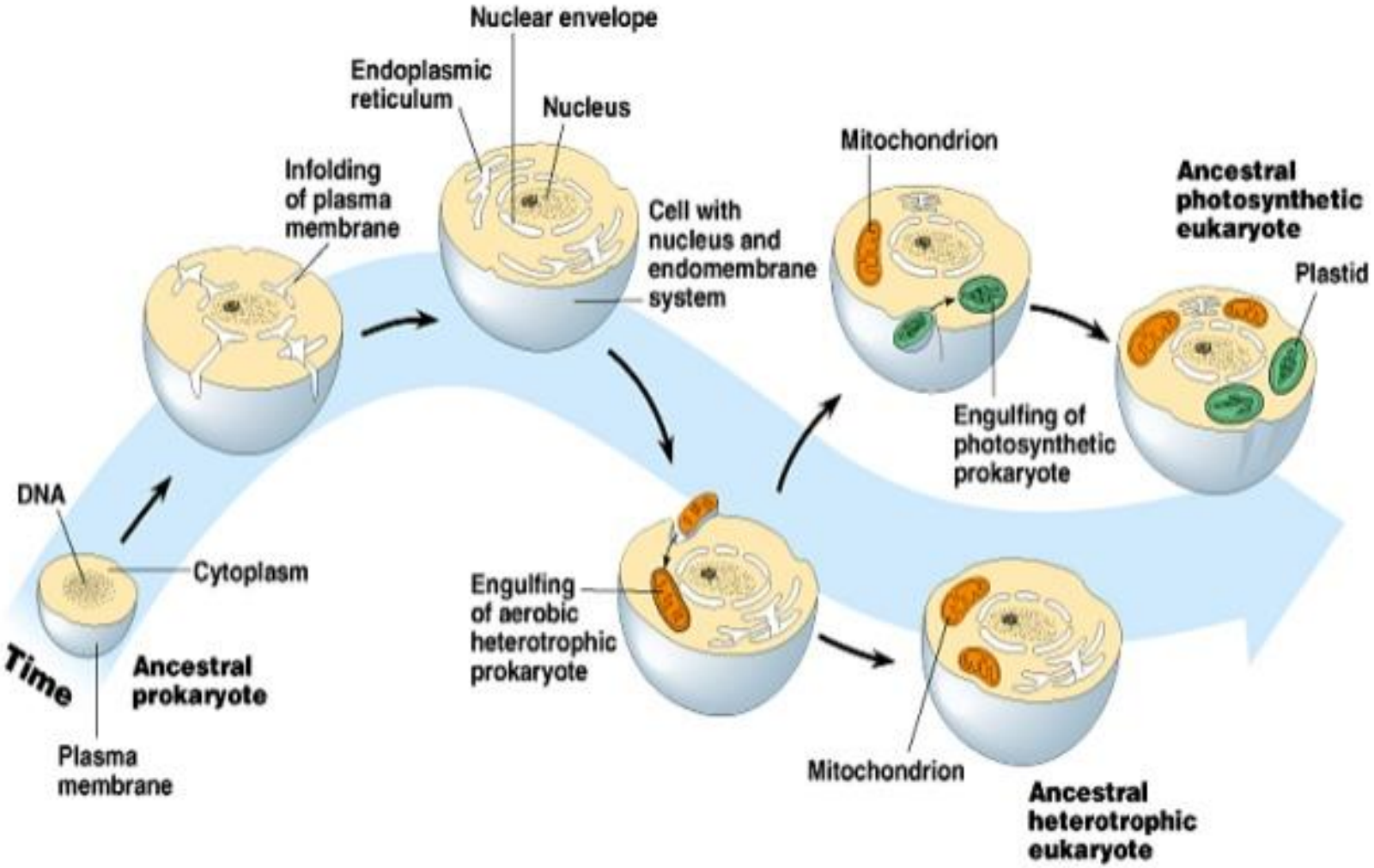
- It is believed that **group II introns**, from the eubacterial endosymbiont, began as retroelements that invaded the genome of the host and subsequently evolved into spliceosomal introns. This catastrophic intron invasion might have been activated as a genomic response to the endosymbiosis.

- During evolution, the **nuclear membrane evolved** to physically separate mRNA processing from translation.

- In prokaryotes, actin-like proteins (underneath the cell membrane) play a dual role, both in chromosome segregation and in cell morphology.

- In the proto-eukaryotic cell (after acquisition of mitochondria), the invention of the nucleus created as well a strong selective pressure to segregate chromosomes using a type of cytoskeleton that did not require interaction with the cell membrane.

But the evolutionary transition to the tubulin-based cytoskeleton, as the principal transport mechanism for chromosome segregation, **also imposed the invention of the centromere to allow chromosome-microtubule interactions**.



- It is recently hypothesized that **centromeres were derived from telomeres during the evolution of the eukaryotic chromosome**.

- The presence of the evolving chromosome within a nuclear compartment, originated the selective pressure that allowed the evolution of a new mechanism of chromosome segregation.

- The phenomenae of unequal exchange and gene conversion led inevitably to the **divergence of the internal telomeric repeats**, giving rise to different sub-telomeric repeats provided the variability that led to the **birth of the centromere**.

The nucleus, the microtubule-based mitotic spindle, the centromere and the telomerase arose within a short evolutionary time: after the endosymbiosis event and before the divergence of the eukaryotic lineages.

Ref: https://doi.org/10.4161/cc.6.23.5047

Genetics and Evolution (22ZOOC14)

Gene structure and regulation in Prokaryotes and Eukaryotes

Overall....

- Why every cell does not express all of its genes ??
- What are the major differences between prokaryotic and eukaryotic gene regulation ??

For a cell to function properly, necessary proteins must be synthesized at the proper time.

All cells control or regulate the synthesis of proteins from information encoded in their DNA.

The process of "turning on" a gene to produce mRNA and protein is called gene expression. Whether in a simple unicellular organism or a complex multicellular organism, each cell controls when its genes are expressed, how much of the protein is made, and when it is time to stop making that protein because it is no longer needed.

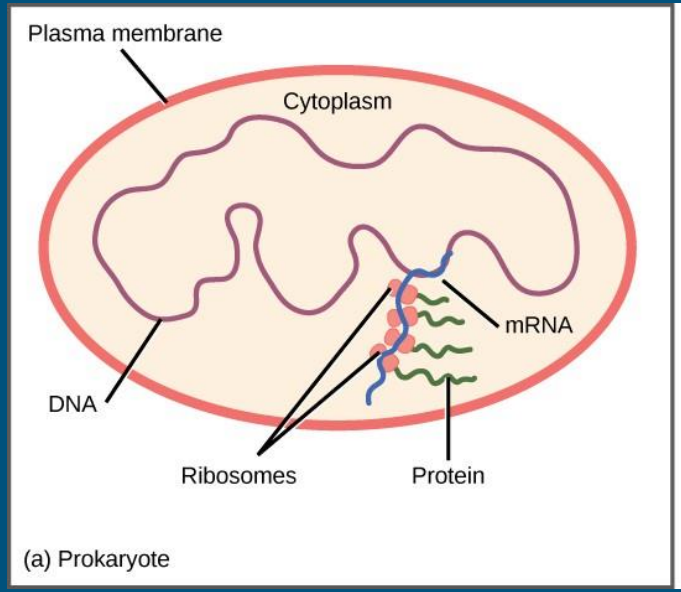
The regulation of gene expression conserves energy and space.

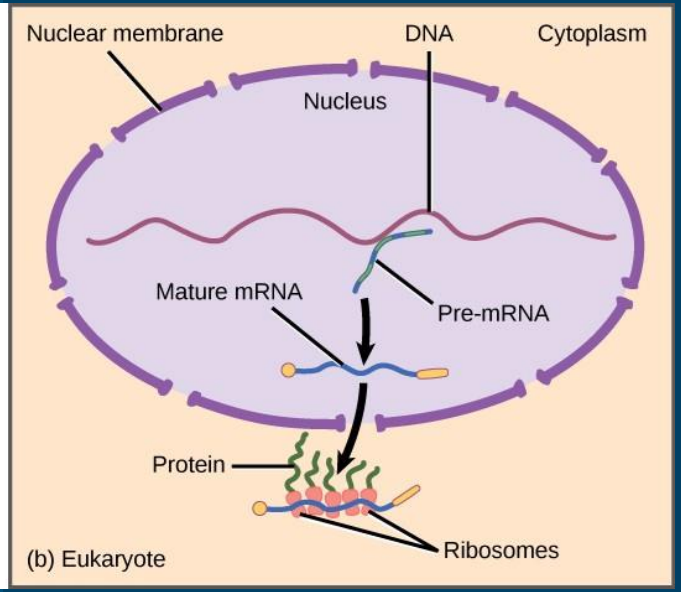
It is more energy efficient to turn on the genes only when they are required.

In addition, only expressing a subset of genes in each cell saves space because DNA must be unwound from its tightly coiled structure to transcribe and translate the DNA.

Cells would have to be enormous if every protein were expressed in every cell all the time.

The control of gene expression is extremely complex. Malfunctions in this process are detrimental to the cell and can lead to the development of many diseases, including cancer.





Prokaryotic transcription and translation occur simultaneously in the cytoplasm, and regulation occurs at the level of transcription.

In eukaryotes, transcription and translation are physically separated, and gene expression is regulated at many different levels.

Prokaryotic Gene Regulation

The DNA of prokaryotes is organized into a circular chromosome that resides in the cell's cytoplasm.

Proteins that are needed for a specific function, or that are involved in the same biochemical pathway, are often encoded together in blocks called operons.

Eg: All five of the genes needed to make the amino acid tryptophan in the bacterium *E. coli* are located next to each other in the *trp* operon.

The genes in an operon are transcribed into a single mRNA molecule.

This allows the genes to be controlled as a unit: either all are expressed, or none is expressed.

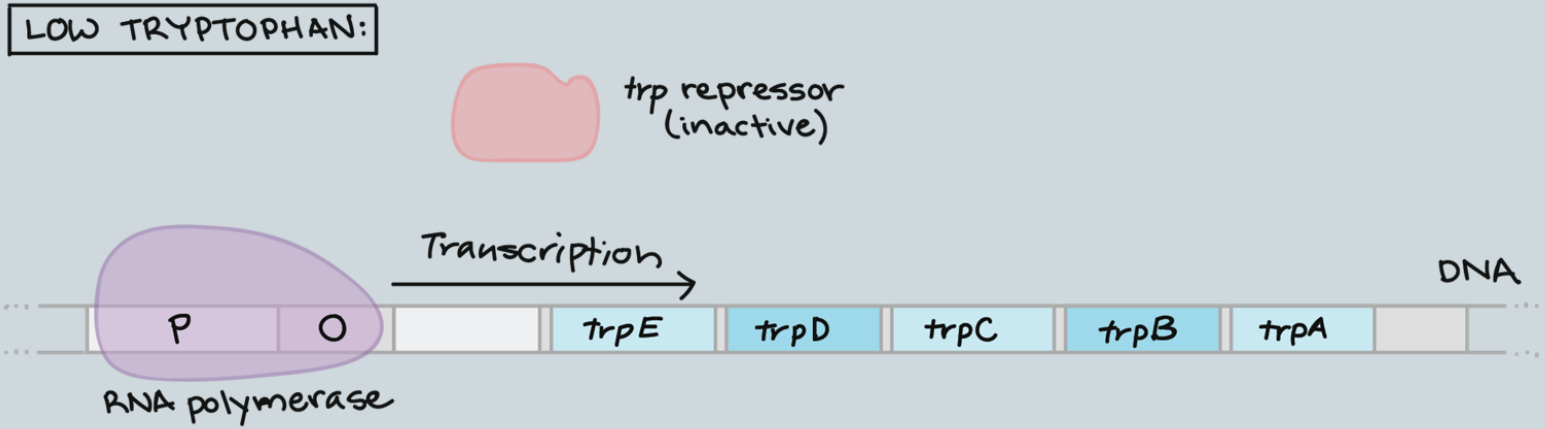
In prokaryotic cells,

there are three types of regulatory molecules that can affect the expression of operons.

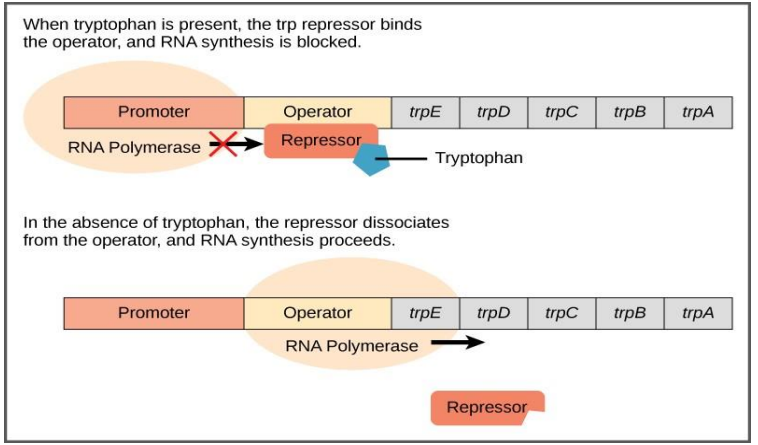
1. **Activators** are proteins that increase the transcription of a gene.
2. **Repressors** are proteins that suppress transcription of a gene.
3. **Inducers** are molecules that bind to repressors and inactivate them.

Each operon needs only one regulatory region, including

- a **promoter**, where RNA polymerase binds, and
- an **operator**, where other regulatory proteins bind.



The *trp* Operon: A Repressor Operon



Like all cells, bacteria need amino acids to survive.

Tryptophan is one amino acid that the bacterium *E. coli* can either ingest from the environment or synthesize.

When *E. coli* needs to synthesize tryptophan, it must express a set of five proteins that are encoded by five genes.

These five genes are located next to each other in the tryptophan (*trp*) operon.

The *lac* Operon: An Inducer Operon

The *lac* operon in *E. coli* has more complex regulation, involving both a repressor and an activator.

E. coli uses glucose for food, but is able to use other sugars, such as lactose, when glucose concentrations are low.

Three proteins are needed to break down lactose; encoded by the three genes of the ***lac* operon**.

When **lactose is not present**, the proteins to digest lactose are not needed.

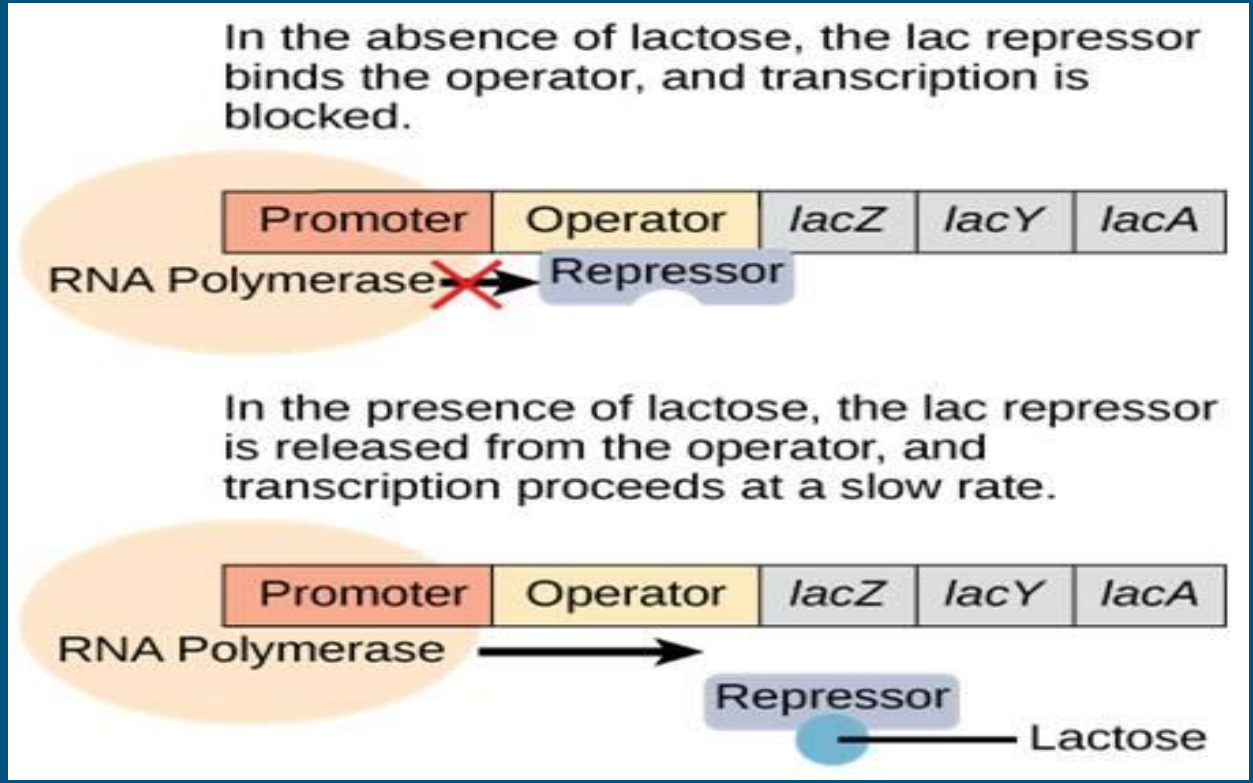
Therefore, a repressor binds to the operator and prevents RNA polymerase from transcribing the operon.

When **lactose is present**, lactose binds to the repressor and removes it from the operator.

RNA polymerase is now free to transcribe the genes necessary to digest lactose

Transcription of the lac operon only occurs when lactose is present.

Lactose binds to the repressor and removes it from the operator.

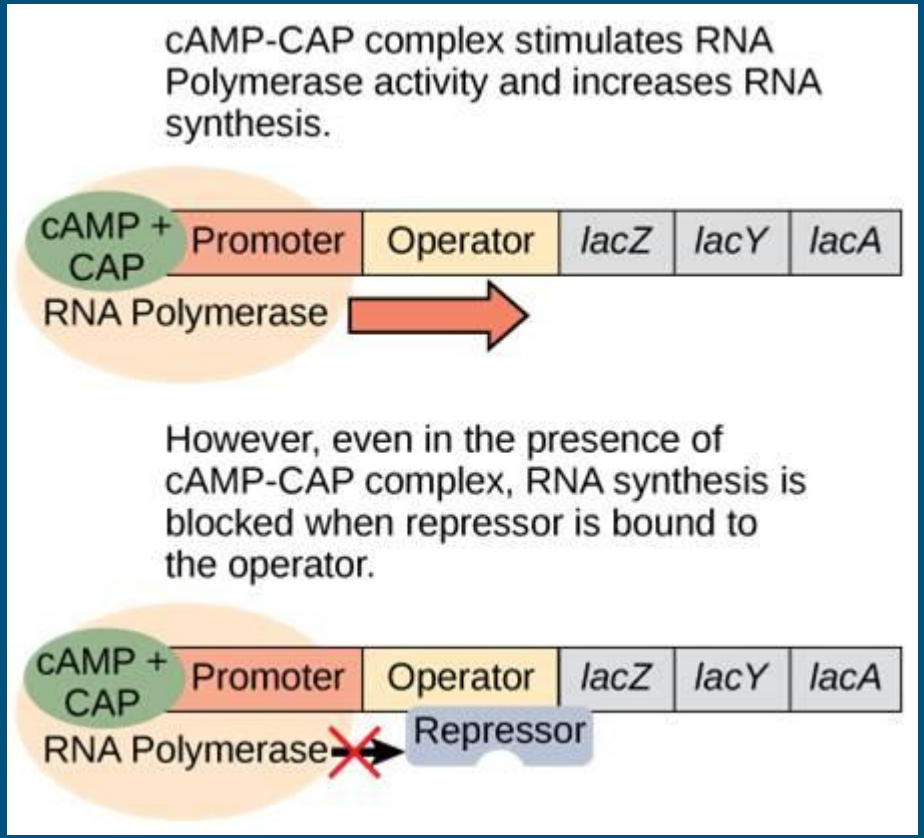


When glucose levels drop,

cyclic AMP (cAMP) begins to accumulate in the cell.

cAMP binds to CAP and the complex binds to the *lac* operon promoter.

This increases the binding ability of RNA polymerase to the promoter and ramps up transcription of the genes.



When there is no glucose, the CAP activator increases transcription of the lac operon. However, if no lactose is present, the operon is not activated.

Eukaryotic Gene Regulation

In eukaryotes, control of gene expression is more complex and can happen at many different levels.

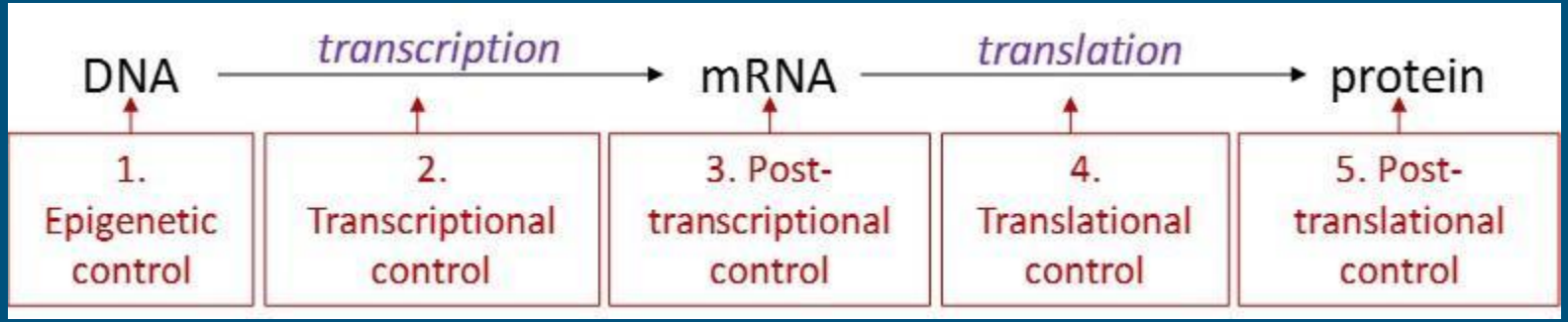
Eukaryotic genes are not organized into operons, so each gene must be regulated independently.

In addition, eukaryotic cells have many more genes than prokaryotic cells.

Regulation of gene expression can happen at any of the stages as DNA is transcribed into mRNA and mRNA is translated into protein.

The regulation is divided into five levels:

1. epigenetic,
2. transcriptional,
3. post-transcriptional,
4. translational, and
5. post-translational regulation



Epigenetic ("around genetics") regulation:

Epigenetics is a relatively new, but growing, field of biology.

Epigenetic control involves changes to genes that do not alter the nucleotide sequence of the DNA and are not permanent.

Instead, these changes alter the chromosomal structure so that genes can be turned on or off.

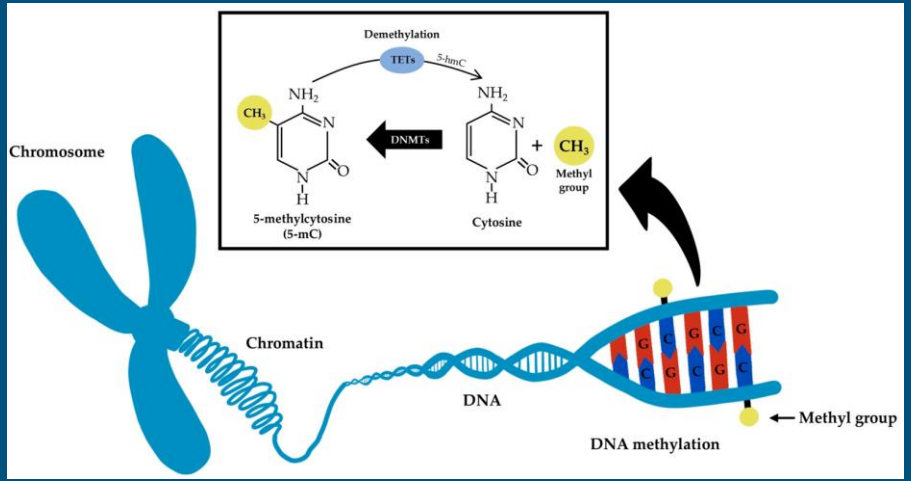
This level of control occurs through heritable chemical modifications of the DNA and/or chromosomal proteins.

Eg: Methylation and histone modification

Addition of methyl groups to the DNA, in a process called methylation.

In general, methylation suppresses transcription. Interestingly, methylation patterns can be passed on as cells divide.

Thus, parents may be able to pass on the tendency of a gene to be expressed in their offspring.



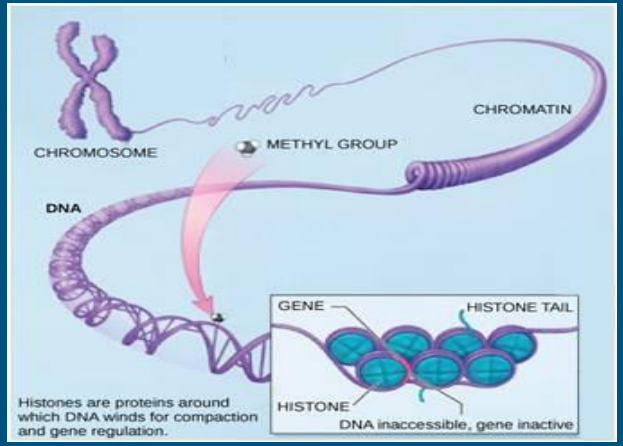
The human genome, consists of over three billion nucleotide pairs.

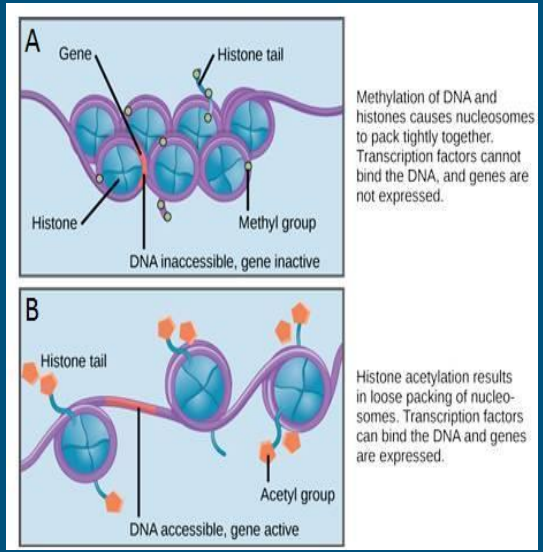
An average chromosome contains 130 million nucleotide pairs, and each body cell contains 46 chromosomes.

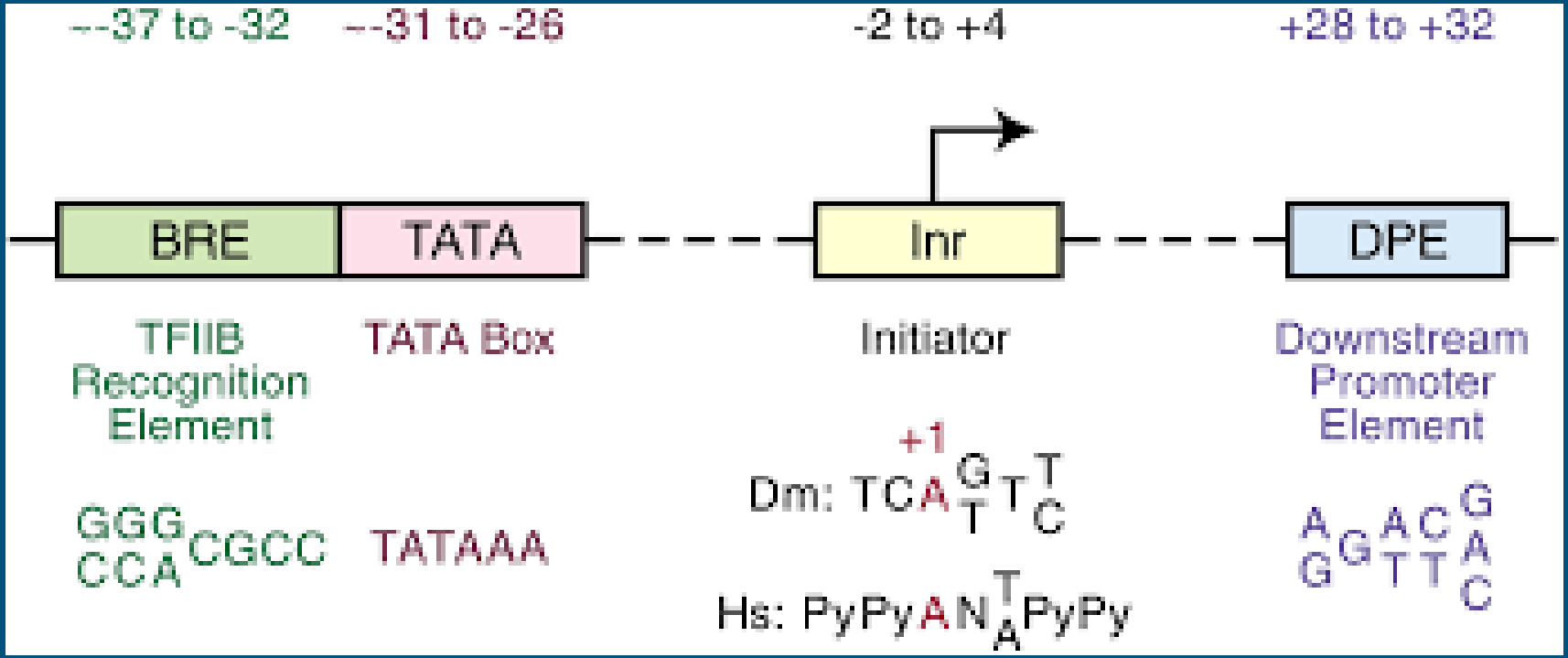
If stretched out linearly, an average human chromosome would be over four centimeters long.

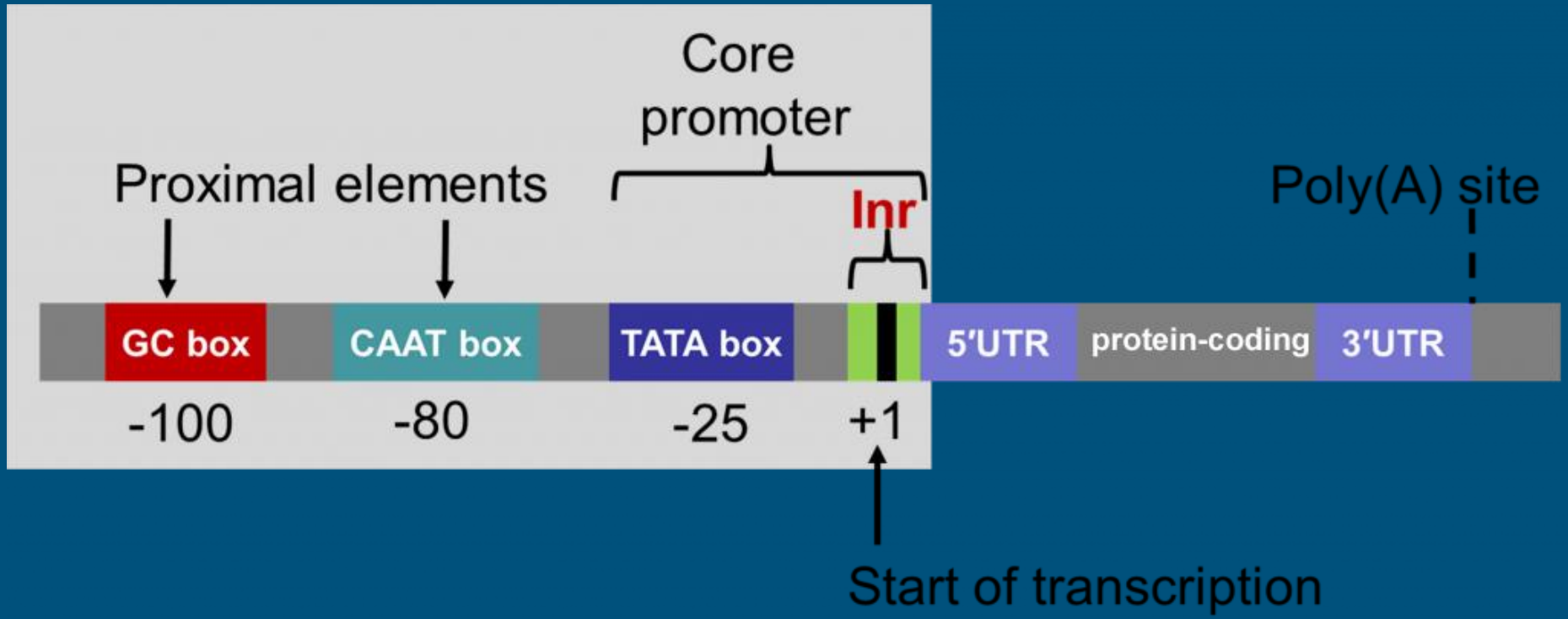
In order to fit all of this DNA into the nucleus of a microscopic cell, the DNA must be tightly wound around proteins.

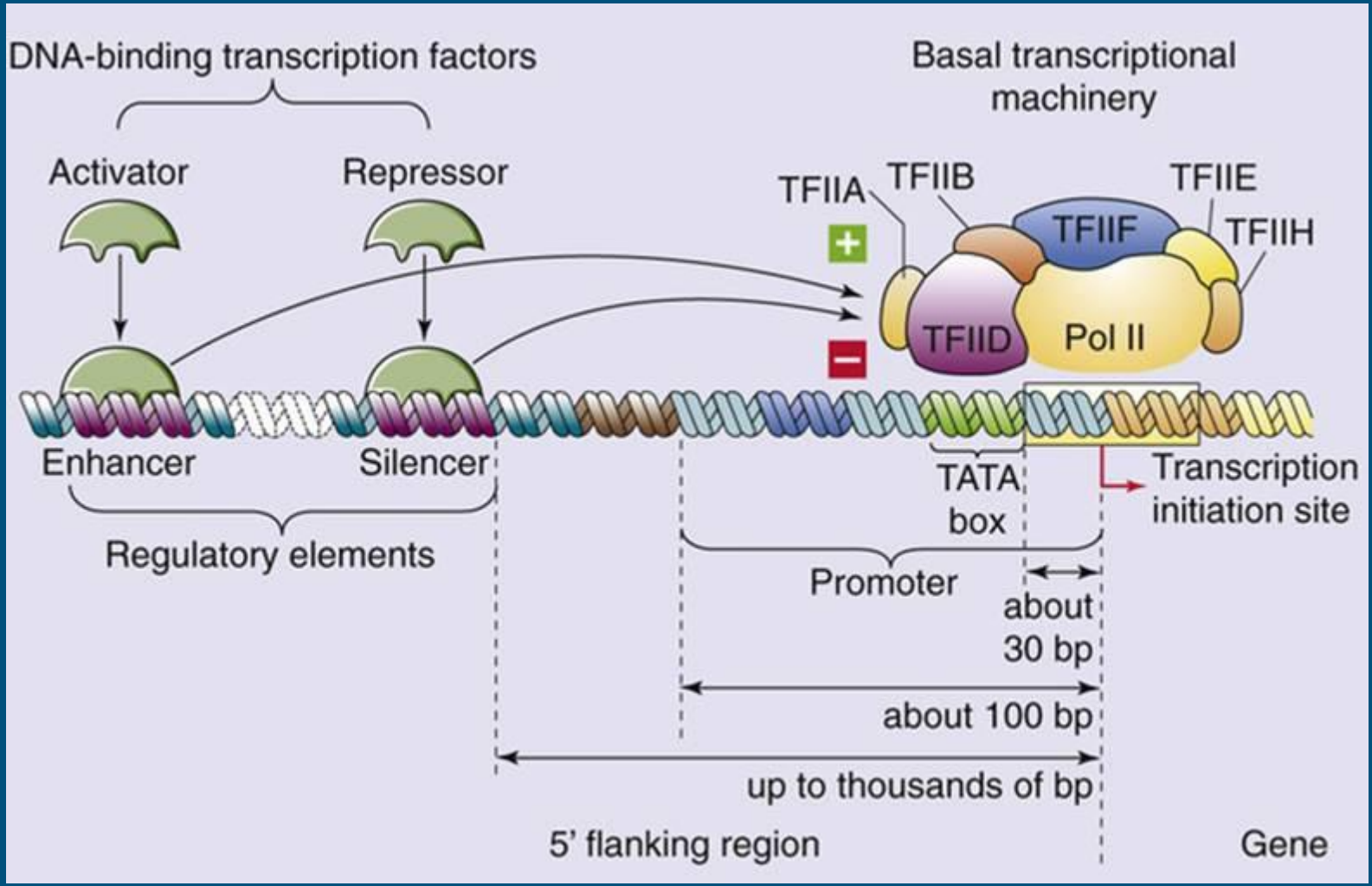
It is also organized so that specific segments can be accessed as needed by a specific cell type.

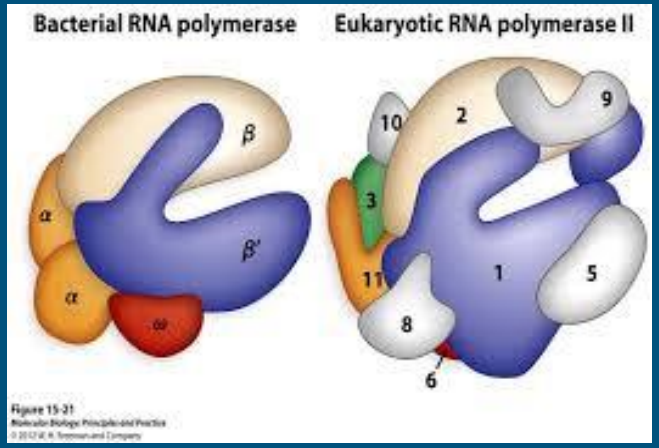




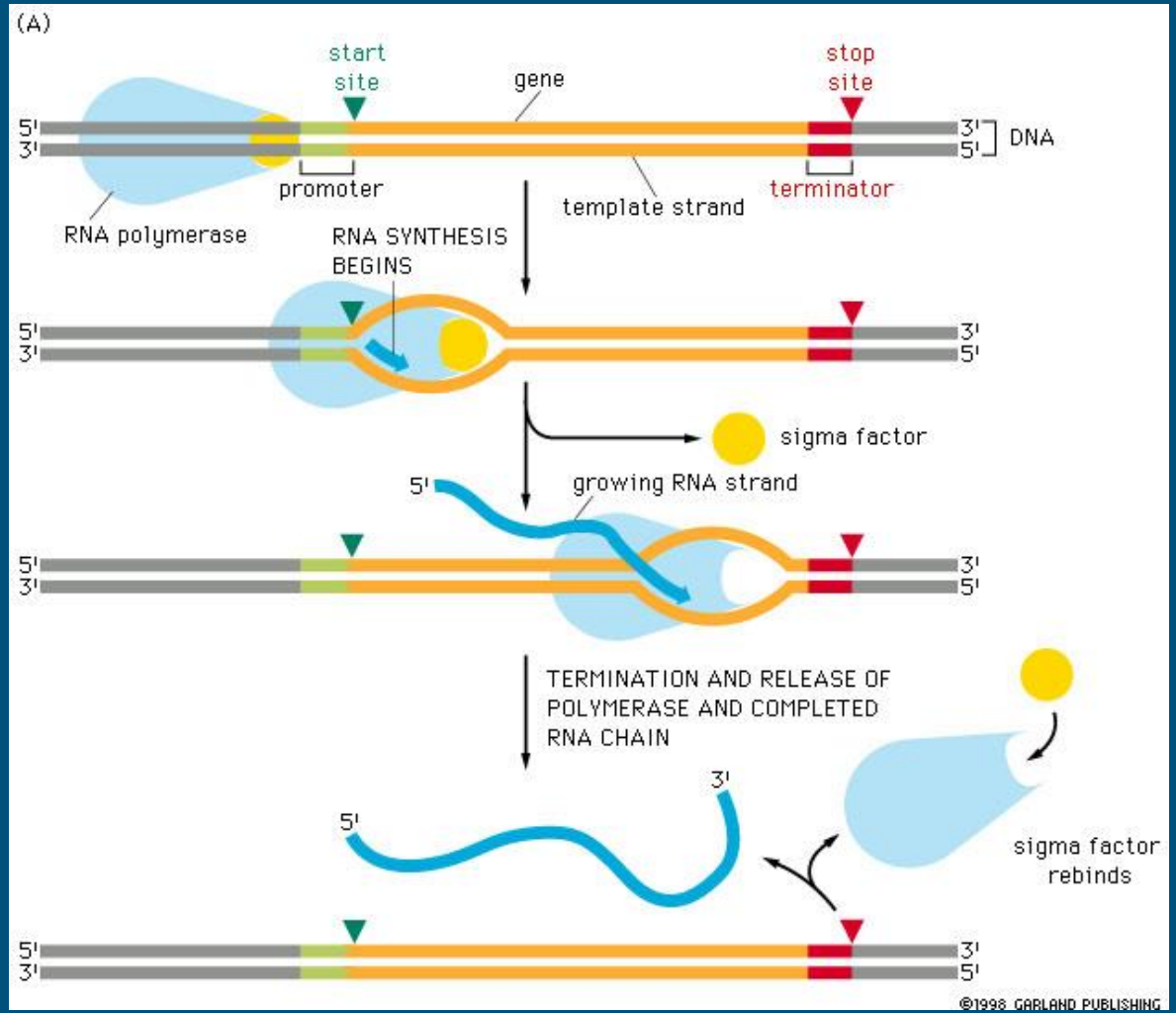

-37 to -32
-31 to -26
-2 to +4
+28 to +32
BRE
TATA
Inr
DPE
TFIIB Recognition Element
TATA Box
Initiator
Downstream Promoter Element
GGG CCA CGCC
TATAAA
+1
Dm: TCA G/T T T/C
Hs: PyPyA N T/A PyPy
A G/G A/T C/T G/A/C

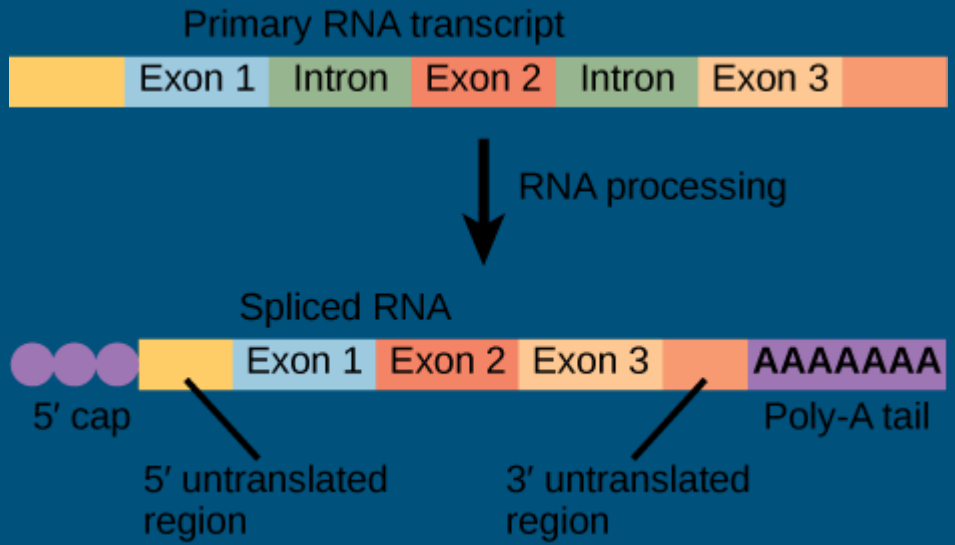

Core
promoter
Proximal elements
Inr
Poly(A) site
GC box
CAAT box
TATA box
5′UTR
protein-coding
3′UTR
-100
-80
-25
+1
Start of transcription


DNA-binding transcription factors
Basal transcriptional machinery
Activator
Repressor
TFIIA
TFIIB
TFIIE
TFIIF
TFIIH
TFIID
Pol II
Enhancer
Silencer
TATA box
Transcription initiation site
Regulatory elements
Promoter
about 30 bp
about 100 bp
up to thousands of bp
5' flanking region
Gene

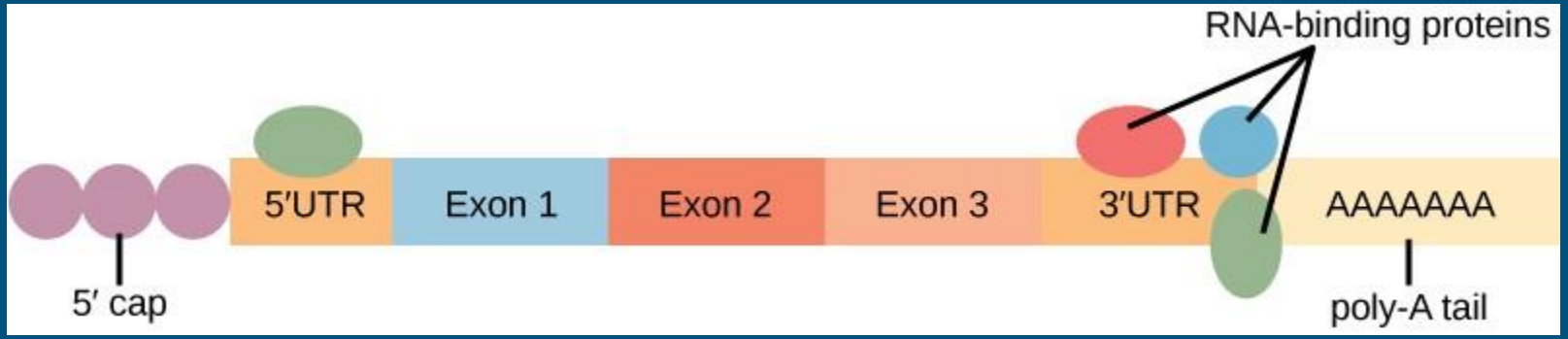


Name	Can be found in	Transcribes
RNA Polymerase I	All Eukaryotes	Large rRNAs
RNA Polymerase II	All Eukaryotes	mRNA, snoRNAs, some snRNAs and miRNAs
RNA Polymerase III	All Eukaryotes	tRNAs, small rRNAs, some snRNAs and miRNAs
RNA Polymerase IV	Plants	some siRNAs
RNA Polymerase V	Plants	RNAs important in heterochromatin formation





Control of RNA Stability



Translational Control of Gene Expression

Translation can also be regulated at the level of binding of the mRNA to the ribosome.

Once the mRNA bound to the ribosome, the speed and level of translation can still be controlled.

An example of translational control occurs in proteins that are destined to end up in an organelle called the endoplasmic reticulum (ER).

The first few amino acids of these proteins are a tag called a signal sequence. As soon as these amino acids are translated, a signal recognition particle (SRP) binds to the signal sequence and stops translation while the mRNA-ribosome complex is shuttled to the ER.

Once they arrive, the SRP is removed and translation resumes.

Post-translational Control of Gene Expression

The activity and/or stability of proteins can also be regulated by adding functional groups, such as methyl, phosphate, or acetyl groups.

Sometimes these modifications can regulate where a protein is found in the cell— in the nucleus, the cytoplasm, or attached to the plasma membrane.

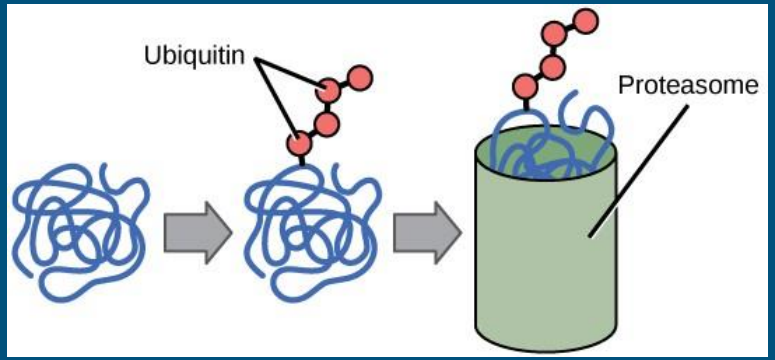
The addition of an ubiquitin group to a protein marks that protein for degradation.

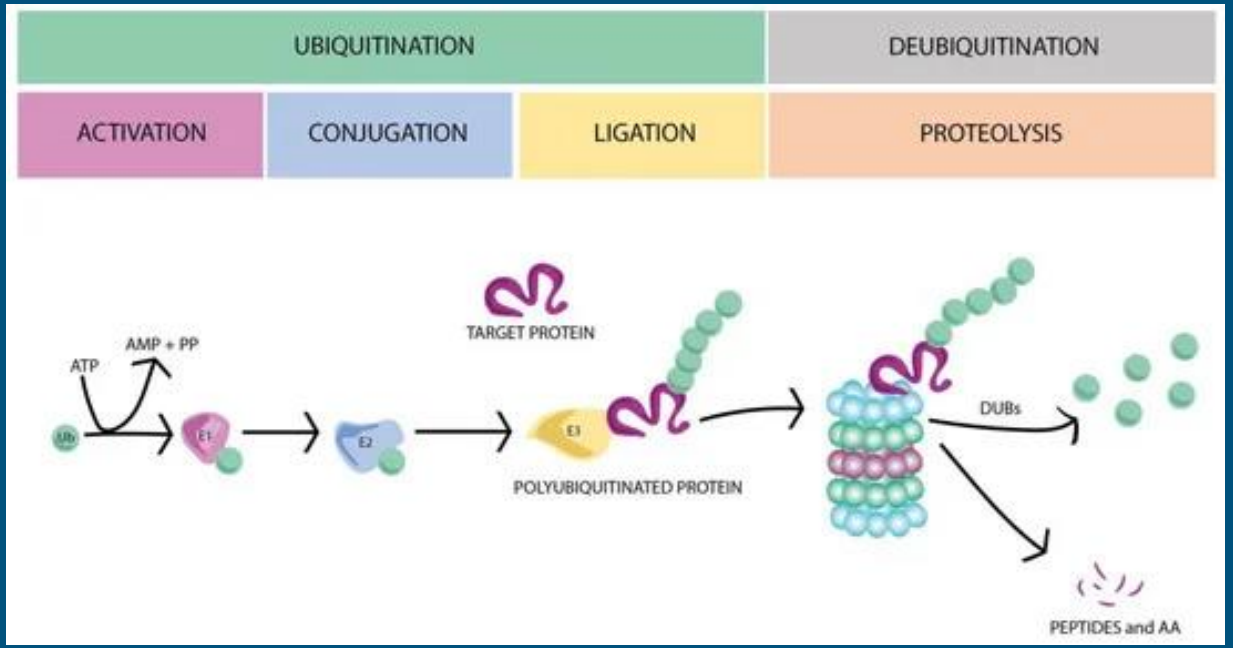
Ubiquitin acts like a flag indicating that the protein's lifespan is complete.

Tagged proteins are moved to a proteasome, an organelle that degrades proteins.

One way to control gene expression, therefore, is to alter the longevity of the protein.

Proteins with ubiquitin tags are marked for degradation within the proteasome.





https://rwu.pressbooks.pub/bio103/chapter/regulation-of-gene-expression/

Genetics and Evolution
(22ZOOC14)

Speciation

- **Species is a group of interbreeding, or potentially interbreeding**, producing fertile organisms.
- **Species** is a group of species reproductively isolated from other species.
- Species have been defined in different ways.
  - Traditionally, species have been defined based on **phenotypic characteristics**.
  - Evolutionary genetics, species have a "defined" a "**shared genetic pool**"

Species arise when a population of organisms splits into genetically distinct groups that **can no longer interbreed** with each other.

Speciation is the development of a new species

A species is defined as a group of organisms that can produce fertile offspring.

Speciation occurs when a population is separated, usually due to a geographical barrier, and natural selection changes the population so much the two groups could no longer interbreed.

Therefore, geographic isolation leads to reproductive isolation.

Reproductive Isolation

It is a feature or mechanism to prevent breeding between species

- **Prezygotic** isolating mechanisms prevent members of different groups from producing hybrid offspring.

- **Postzygotic** isolating mechanisms prevent hybrid offspring from passing on their genes to subsequent generations.

Prezygotic Isolating Mechanisms

- Prezygotic isolating mechanisms **prevent mating** between individuals from different populations of organisms or by preventing the gametes of these individuals to form zygotes.

- These mechanisms include
  - Ecological or Geographical isolation based on habitat preference (different habitats in the same geographical area)
  - Temporal or behavioral factors (e.g., different times sexual maturity or different courtship rituals)
  - Mechanical Anatomical or chemical incompatibilities in reproductive organs or gametes (e.g., failure to mate successfully or to form zygotes)

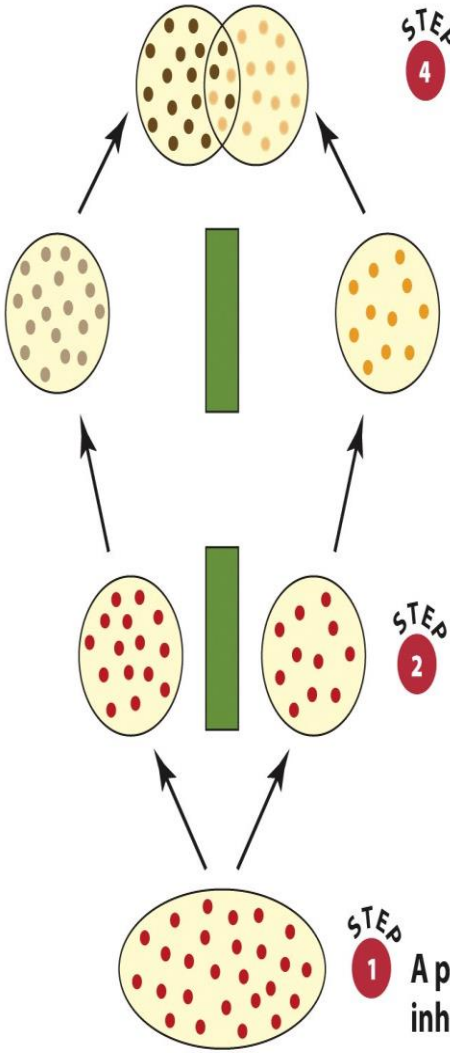
Postzygotic Isolating Mechanisms

- Postzygotic isolating mechanisms operate **after hybrid zygotes have been formed**.

- Mechanisms include
  - Reduction of hybrid viability (e.g., failure to survive or to reach sexual maturity)
  - Impaired hybrid fertility (failure to produce functional gametes)

Modes of speciation

Allopatric



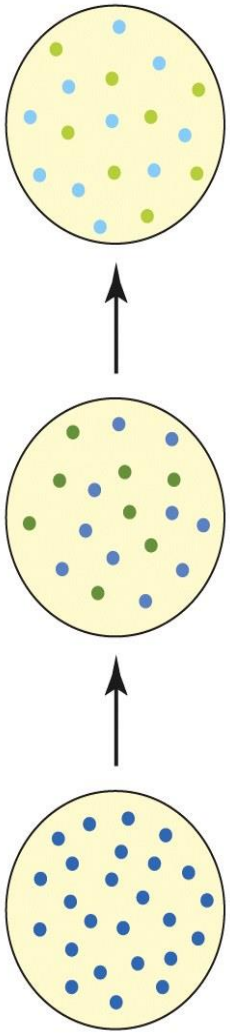
STEP 4 When reunited, the subpopulations cannot exchange genes. Each of them has become a distinct species.

STEP 3 The separated subpopulations change genetically. Reproduct isolating mechanisms evolve.

STEP 2 The population is subdivided by a geographical barrier.

STEP 1 A population of organisms inhabits a territory.

Sympatric



STEP 3 Genetic differentiation leads to reproductive isolation between the subpopulations. Each subpopulation becomes a distinct species.

STEP 2 Subpopulations form in the absence of geographical barriers and begin to differentiate genetically.

STEP 1 A population of organisms inhabits a territory.

Genetic drift

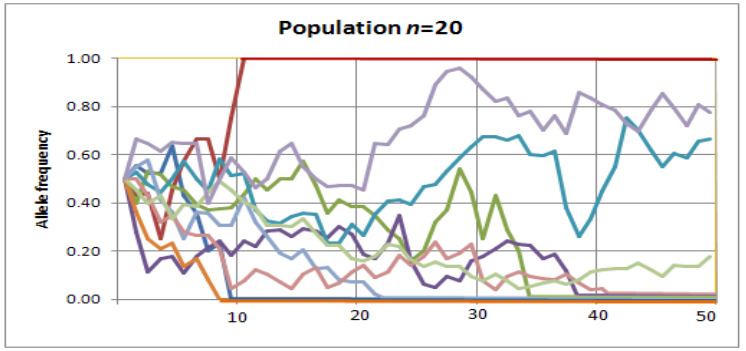
It is a **random process**.

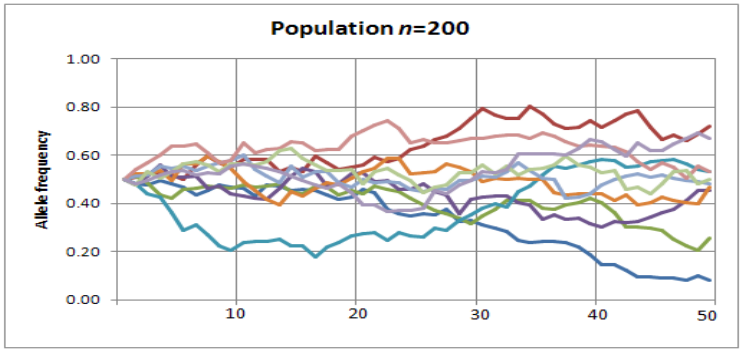
In **small populations**, unpredictable events (disease, predation, abiotic factors etc) can result in the **frequency of alleles varying from one generation to the next**.

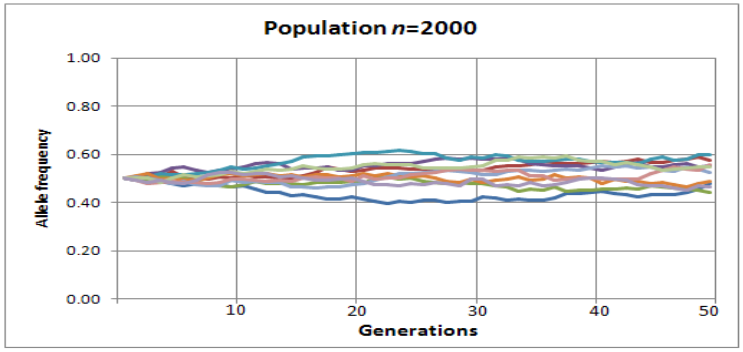
This tends to be **masked** **in larger populations** due to the increased number of individuals).

<u>The effect of population size on genetic drift:</u>

Ten simulations each of **random change** in the frequency distribution of a single hypothetical allele over **50 generations for different sized populations**.

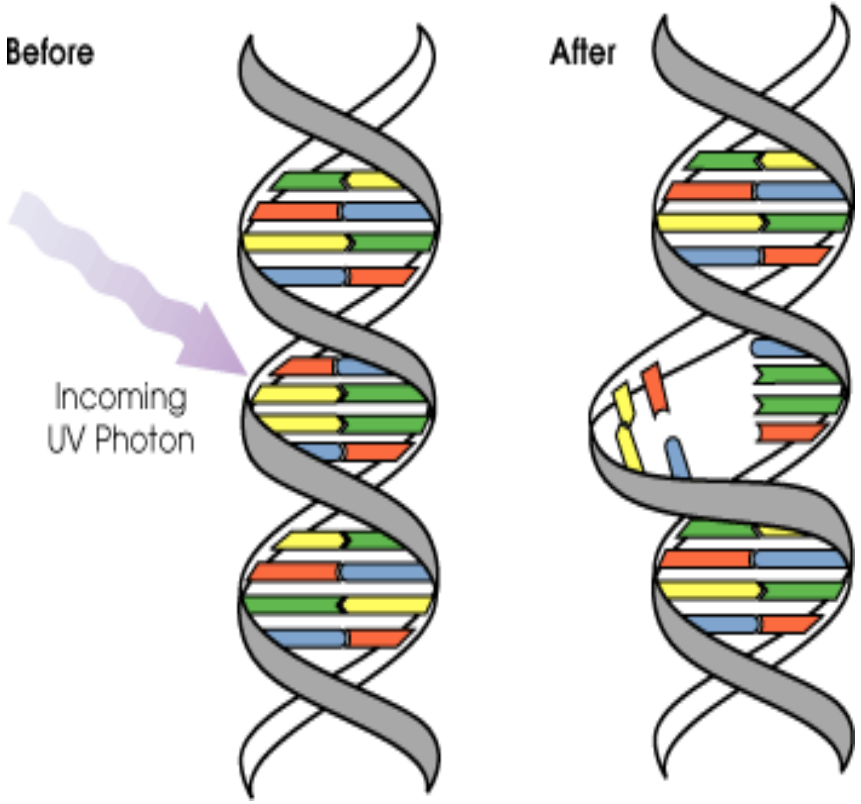






Variation and mutation

The cornerstone of the **theory of natural selection** is the fact that **individuals of a population show variation**.



Variation in inherited traits arises as a result of mutation. **Mutation is the original source of new sequences of DNA.** The new DNA sequences can be **new alleles**.

Mutations to an organisms DNA are usually harmful or they may be neutral.

On rare occasions, a mutation in an individual's DNA can make it better suited to its environment and increase the fitness of an individual, increasing its chances of reproductive success.

Sexual selection

There are two mechanisms, as mentioned by Charles Darwin

Intrasexual selection

Intrasexual selection refers to **selection within the same sex** (usually males). Individuals compete with each other with ritualised displays of strength and stamina to warn off competitors or defend his mate(s) e.g. Red deer.

It is an example of **dominance hierarchy**.

However, it has also been seen in females e.g. Ring Tailed Lemurs.

Intersexual selection

Intersexual selection refers to individuals (usually female) being **very selective** about their **choice of mate**. It is sometimes called ‘mate choice’.

Often bright plumage and showy courtship displays influence a female’s choice. e.g. Peacocks.

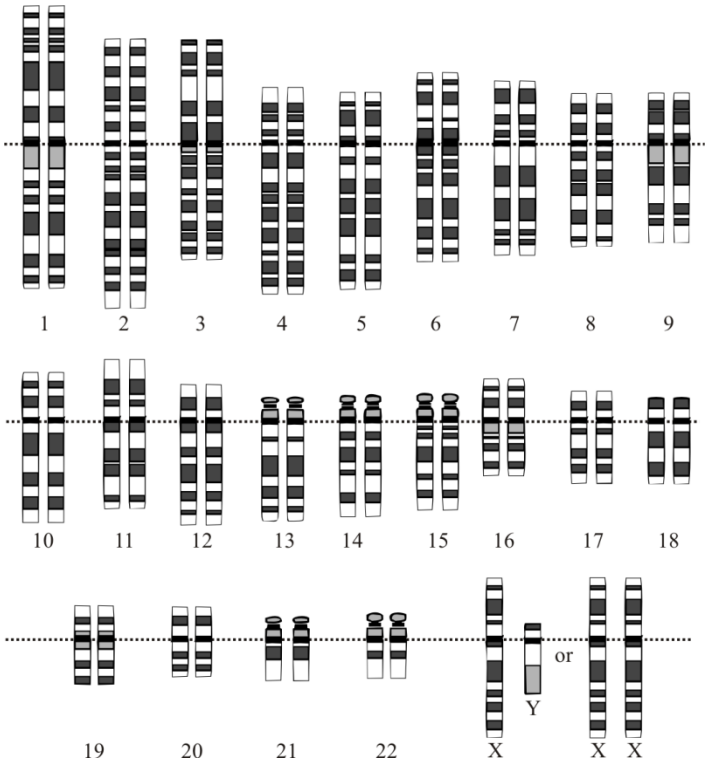
The extreme difference in degree of plumage shown by the male contrasts with the smaller, much less showy, Peahen.

This difference between the sexes, also shown in other forms of adornment in males such as antlers in deer is called **sexual dimorphism**.

Fitness in evolution

Fitness is an indication of an **individuals ability to be successful both at surviving and reproducing**.

It refers to the contribution that is made to the gene pool of the next generation by individual genotypes.



As a result of natural selection we know that there is variation of alleles of genes.

Frequencies of alleles changes through many generations and that there would be an expectation that alleles with the highest fitness would become more common in a population.

Fitness

- **Absolute fitness** - the ratio of frequencies of a particular genotype from one generation to the next.

- **Relative fitness -** the ratio of surviving offspring of one genotype compared with other genotypes.

It is important to remember that the **overall fitness of an individual is affected by its environment**.

The fitness of a phenotype and genotype will differ in different environments.

Eg: If a moth is more successful at producing offspring due to the fact it is more camouflaged from predators, this will increase the **relative fitness** of the moth population. However, if the moth moves to a new environment, this phenotypic adaptive advantage will no longer apply.

Evidence for Evolution:

1. Fossil evidence
2. Biochemical similarities
3. Shared anatomical similarities

1. Fossil evidence provides an **incomplete record of early life**.

It can include any evidence of life, such as imprints and remains of organisms.

This evidence must be interpreted to form an overall picture of how species have changed over time (evolved).

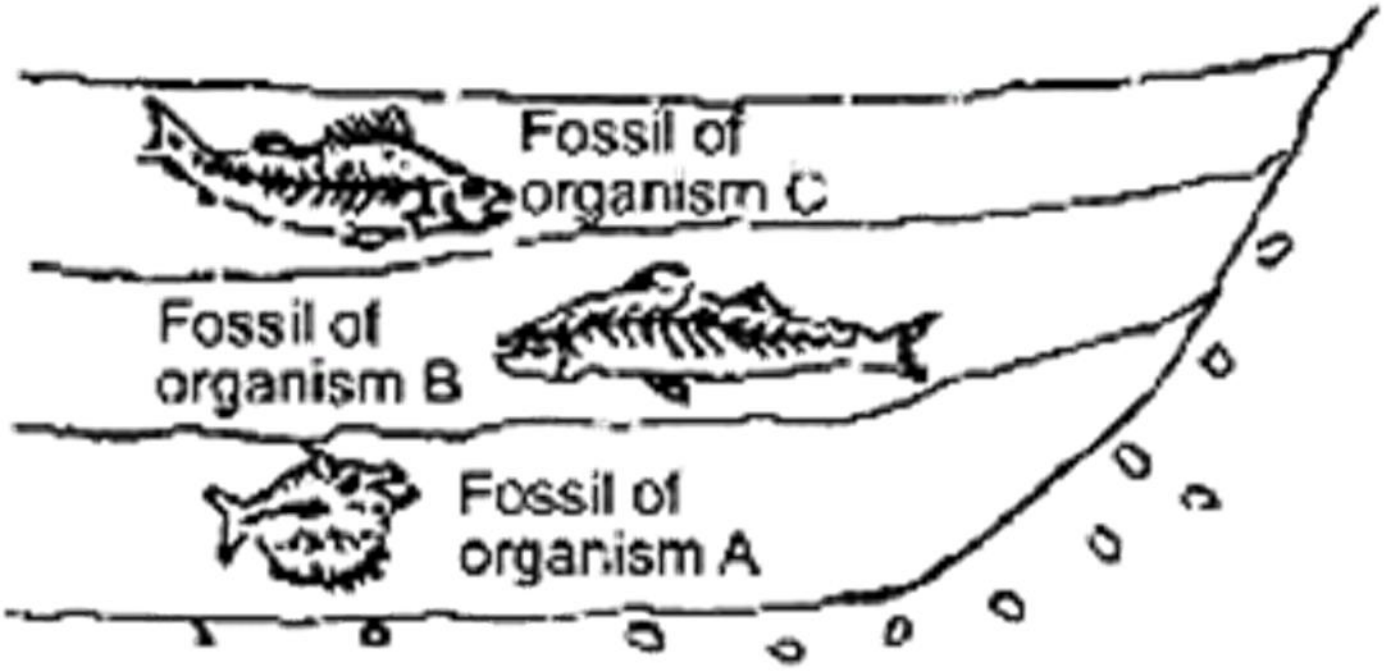
By examining it can be concluded - evolution happens in a simple to complex pattern and life emerged from sea to land.

Fossils **must be dated** to help establish a time frame for the existence of a species.

- There are **two methods of determining the age of fossils**.

- a. **<u>Relative dating:</u>** The exact age of the fossil cannot be determined, only the order of appearance can be found as compared to other fossils found in nearby rocks.

- Fossils occur in layers of sedimentary rock. The fossils near the top will be more recent than fossils in lower layers of rock.



- b. **Radioactive dating**: It gives **a more exact age** using the natural decay of radioactive isotopes in organisms.

2. Biochemical similarities: It include **comparisons of DNA** and the resulting **amino acid sequences** for certain, **shared proteins**.

This is considered one of the **most reliable and objective** types of evidence used **to determine evolutionary relationships**. In general, the fewer differences found between two species, the closer the evolutionary relationship.

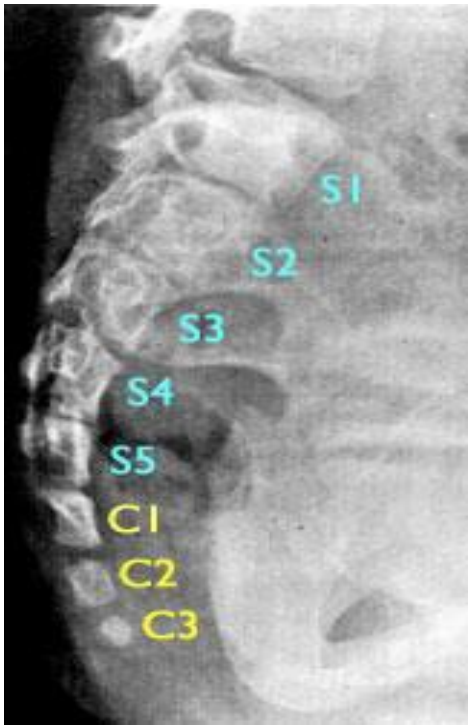
Species	Sequence of Amino Acids in the Same Part of the Hemoglobin Molecules
Human	Lys–Glu–His–Iso
Horse	Arg–Lys–His–Lys
Gorilla	Lys–Glu–His–Lys
Chimpanzee	Lys–Glu–His–Iso
Zebra	Arg–Lys–His–Arg

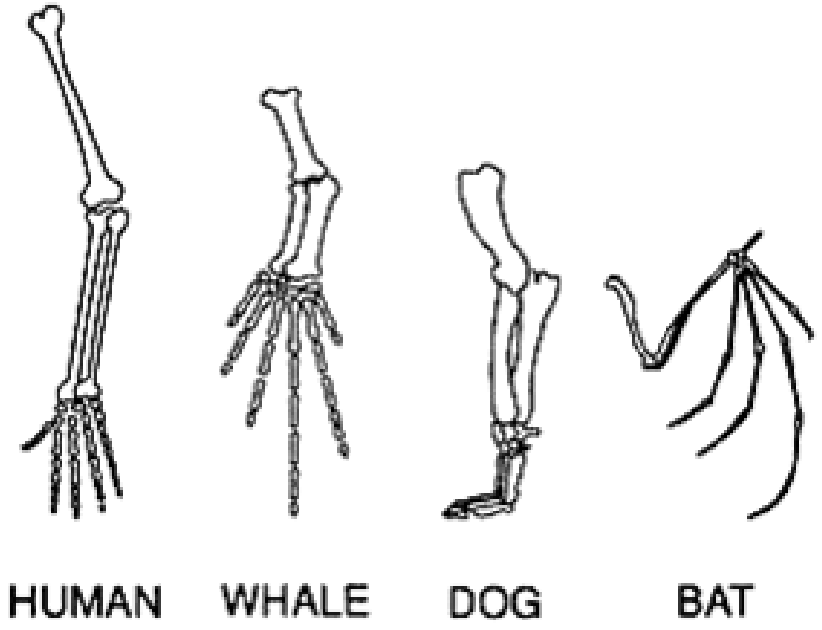
3. **Shared anatomical structures** supports some type of evolutionary relationship.

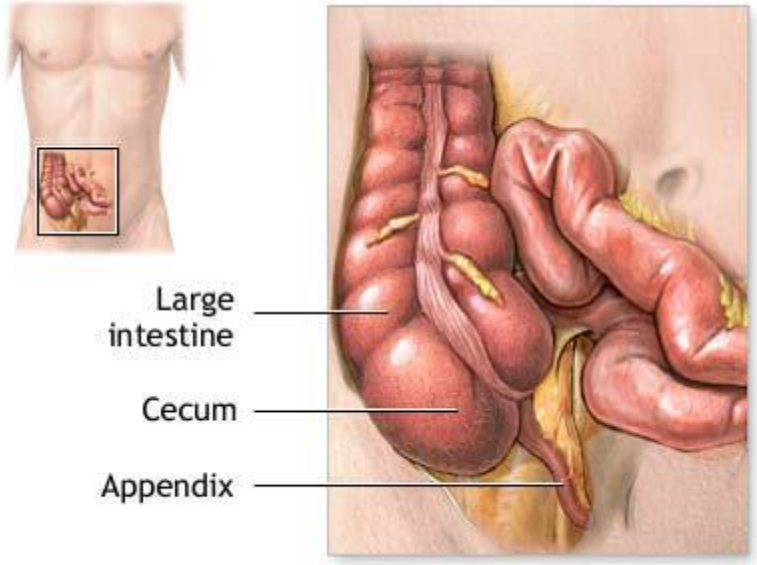
- a. A **similar bone arrangement**, even if the functions are different, supports evolution from a common ancestor.

- b. **Structures that perform the same function** (ex. flying) but are very **different anatomically** (ex. bird wing vs. butterfly wing) supports evolution in similar habitats though not from a recent common ancestor.

- c. **Vestigial structures** (ex. appendix or tail bone in human) are not functional in that organism, but may represent a link to a previous ancestor.


S1
S2
S3
S4
S5
C1
C2
C3


HUMAN
WHALE
DOG
BAT

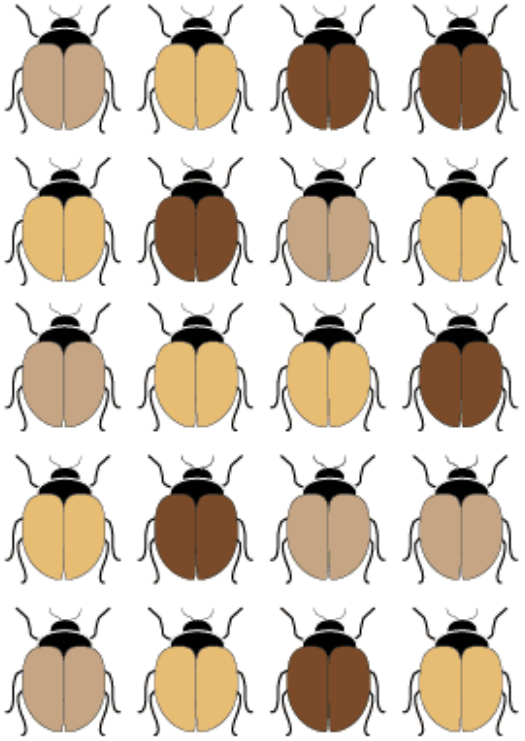

Large intestine
Cecum
Appendix

Mechanisms of Evolution

1. **Individuals don't evolve; populations do**.

The population is the smallest unit of evolution because acquired traits in an individual cannot be passed on (inherited by offspring).

However, different traits already present in a population can be "selected", changing the population.



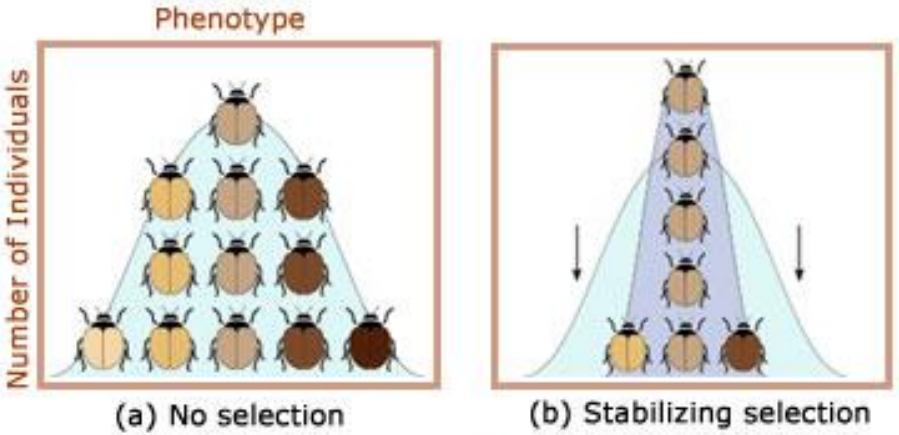
2. **Evolution occurs when the gene pool changes**

(all of the genes of a population).

A change in genotype may lead to a change in phenotype.

Evolution acts on the phenotype.

There are three basic patterns by which natural selection occurs:



i. **Stabilizing selection** favors the "**average**" phenotype in a population.

ii. **Directional selection** favors **ONE of the extreme** ends of the "typical" distribution.

iii. **Disruptive Selection** favors **BOTH of the extreme** ends of the "typical" distribution.