Unit 5

Genetics and Evolution

Genetics and Evolution

- The science of heredity or genetics is the study of two contradictory aspects of nature : **heredity and variation**.
- The process of transmission of characters from one generation to next, either by gametes–sperms and ova–in sexual reproduction or by the asexual reproductive bodies in asexual reproduction, is called **inheritance or heredity*.***
- ***Heredity is the*** cause of similarities between individuals. This is the reason that brothers and sisters with the same parents resemble each other and with their parents.
- **Variation is the cause of differences** between individuals. This is the reason that brothers and sisters who do resemble each other are still unique individuals.
- **The science of genetics** attempts to explain the mechanism and the basis for both similarities and differences between related individuals.
- It also tries to explain the phenomenon of **evolution and cytodifferentiation.**
- The **heredity and variations** play an important role in the formation of new **species (speciation**).

Genetics and Evolution

The **basic components of genetics** are

- ***DNA*** - ***genes***
- ***RNA*** ***-chromosomes and genetic inheritance***

- DNA molecules
  - hold all the genetic information for almost all organisms.
  - provide cells with the information they need to perform tasks that allow an organism to grow, survive and reproduce.

- A gene

- is one particular section of a DNA molecule that tells a cell to perform one specific task.
- a hereditary unit that occupies a specific position (locus) within the genome or chromosome;
- a unit that has one or more specific effects upon the phenotype of the organism;
- a unit that can mutate to various allelic forms; and
- a unit that recombines with other such units.
- **Heredity** is what makes offspring look like their parents. During reproduction, DNA is replicated and passed from a parent to their offspring.

Genetics and Evolution

- This **inheritance of genetic material** by offspring influences the appearance and behavior of the offspring.
- The environment that an organism lives in can also influence how genes are expressed.
- Genes (and, thus, the traits they code for) are passed from parent to offspring.
- From generation to generation, well-understood molecular mechanisms reshuffle, duplicate, and alter genes in a way that produces **genetic variation**.
- This **variation** is the raw material for **evolution**.

Basic Principles of Mendelian genetics and patterns of

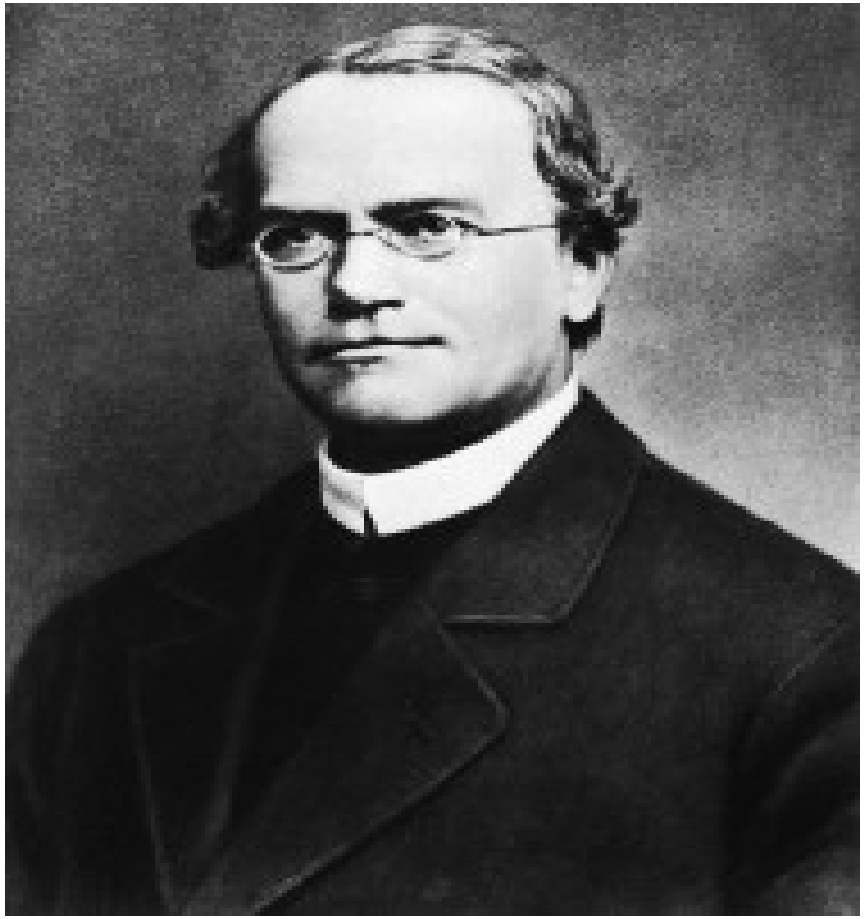
- It is wondering why you are the only one in your family with your grandfather's nose?
- The way in which traits are passed from one generation to the next, and sometimes skip generations was first explained by Gregor Mendel.
- By experimenting with pea plant breeding, Mendel developed **three principles of inheritance** that described the **transmission of genetic traits**, before anyone knew genes existed.
- Mendel's insight greatly expanded the understanding of genetic inheritance, and led to the development of new experimental methods.

Gregor Mendel
(1822-1884)

Responsible for the Laws governing Inheritance of Traits

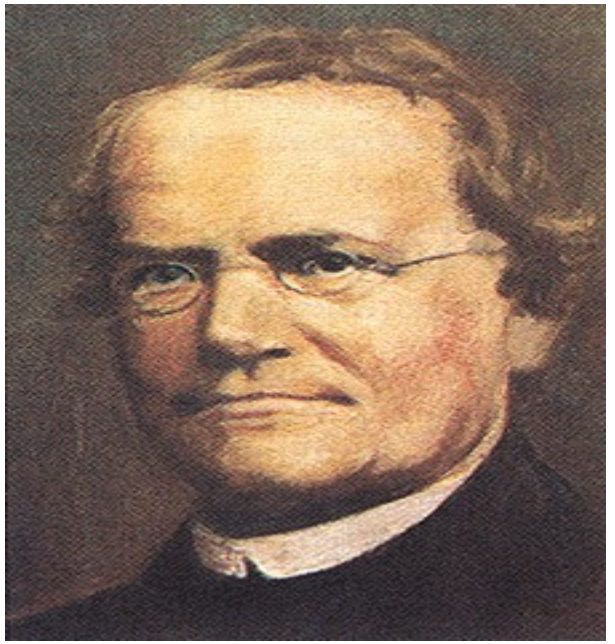


- 1822 - Brunn in Austria, now, Brno (Czecholovakia)
- Financial and Health Problem
- Unable to continue studies
- Joined St.Augustinian Monastery
- 1847 – Priest
- 1851 – Higher studies at Vienna
- University
- 1854- Science teacher



Gregor Johann Mendel

- Between 1856 and 1863, Mendel cultivated and tested some 28,000 pea plants
- He found that the plants' offspring retained traits of the parents
  - Austrian monk
  - Developed the laws of inheritance
  - Presented his findings in the Natural History Society of Brunn – 1865
  - Paper entitled, "Experiments in Plant Hybridization" – 1866
  - German language.



- **Mendel's work was not recognized until the turn of the 20th century**
- **1900 - Carl Correns, Hugo deVries, and Erich von Tschermak rediscover and confirm**
- **Called the "Father of Genetics"**



(a) Gregor Mendel



(b) Carl Correns



(c) Hugo de Vries



(d) Eric von Tschermak

Why peas, *Pisum sativum*?

- Can be grown in a small area
- Produce lots of offspring
- Produce pure plants when allowed to self-pollinate
- several generations
- Can be artificially cross-pollinated
- Bisexual.
- Many traits known.
- easy to grow



Basic Principles of Mendelian genetics

- Traits are passed down in families in different patterns.
- Pedigrees can illustrate these patterns by following the history of specific characteristics, or phenotypes, as they appear in a family.
- The inheritance pattern in which traits of generation I has passed down through the family tree is considered dominant, because it is observable in every generation.
- Thus, every individual who carries the genetic code for this characteristic will show evidence of the characteristic.
- A characteristic may disappear in one generation, only to reappear in a subsequent one.
- This pattern of inheritance, in which the parents do not show the phenotype but some of the children do, is considered recessive.

Mendel's Laws

1. Law of Dominance
2. Law of Segregation
3. Law of Independent assortment

Law of Dominance

States that on crossing homozygous organisms for single pair of contrasting characters, only one characters make its appearance in F_1 generation and is name as Dominant character.

Results of Monohybrid Crosses

- Inheritable factors or genes are responsible for all heritable characteristics
- Phenotype is based on Genotype
- Each trait is based on two genes, one from the mother and the other from the father
- True-breeding individuals are homozygous (both alleles) are the same

Law of Dominance

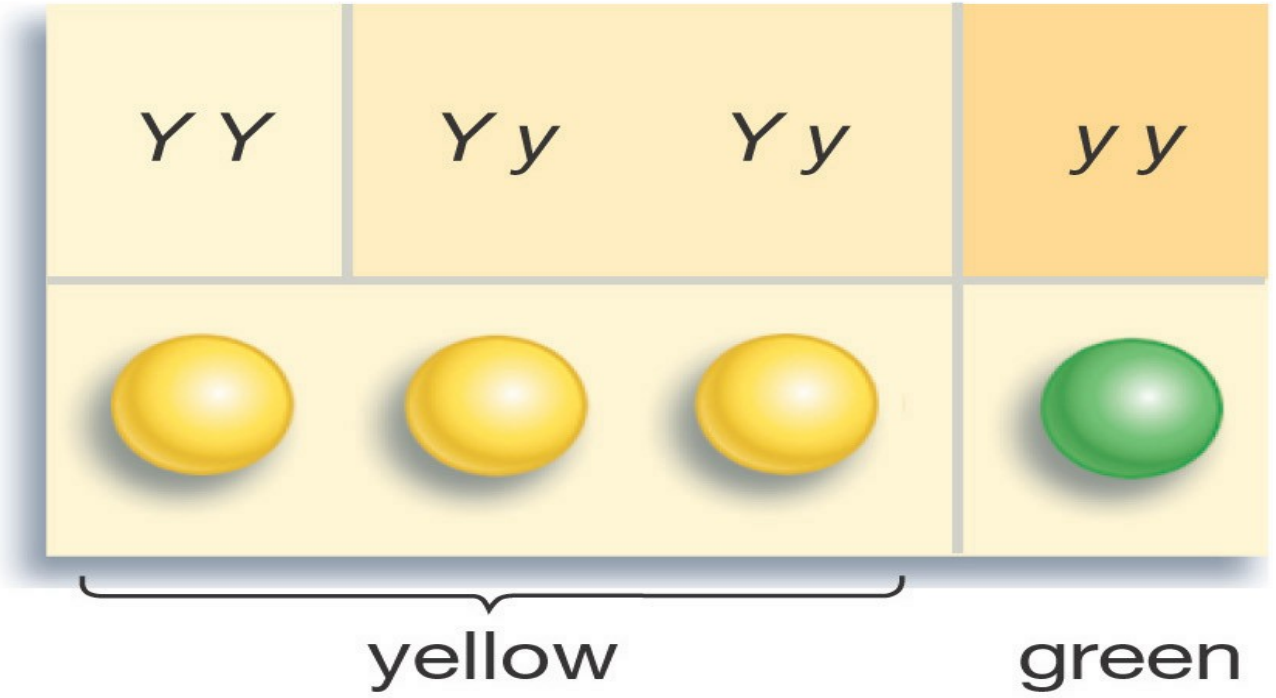


- *In a cross of parents that are pure for contrasting traits, only one form of the trait will appear in the next generation*

- *All the offspring will be heterozygous and express only the dominant trait.*

- *RR x rr yields all Rr (round seeds)*

Law of Dominance



Three genotypes yield . . .

two phenotypes.

	Character	Dominant	Recessive
Drosophila	Body Colour	Gray	Black
	Eye colour	Red	White
Man	Colour of hair	Dark	Light
	Form of hair	Curly	Straight
	Colour of eye	Brown	Blue
	Lips	Broad and thicken	Thin
	Blood group	A,B,AB	O

Law of Segregation

Two allele of a gene remain separate and do not contaminate each other in F_1 or hybrid.

- OR

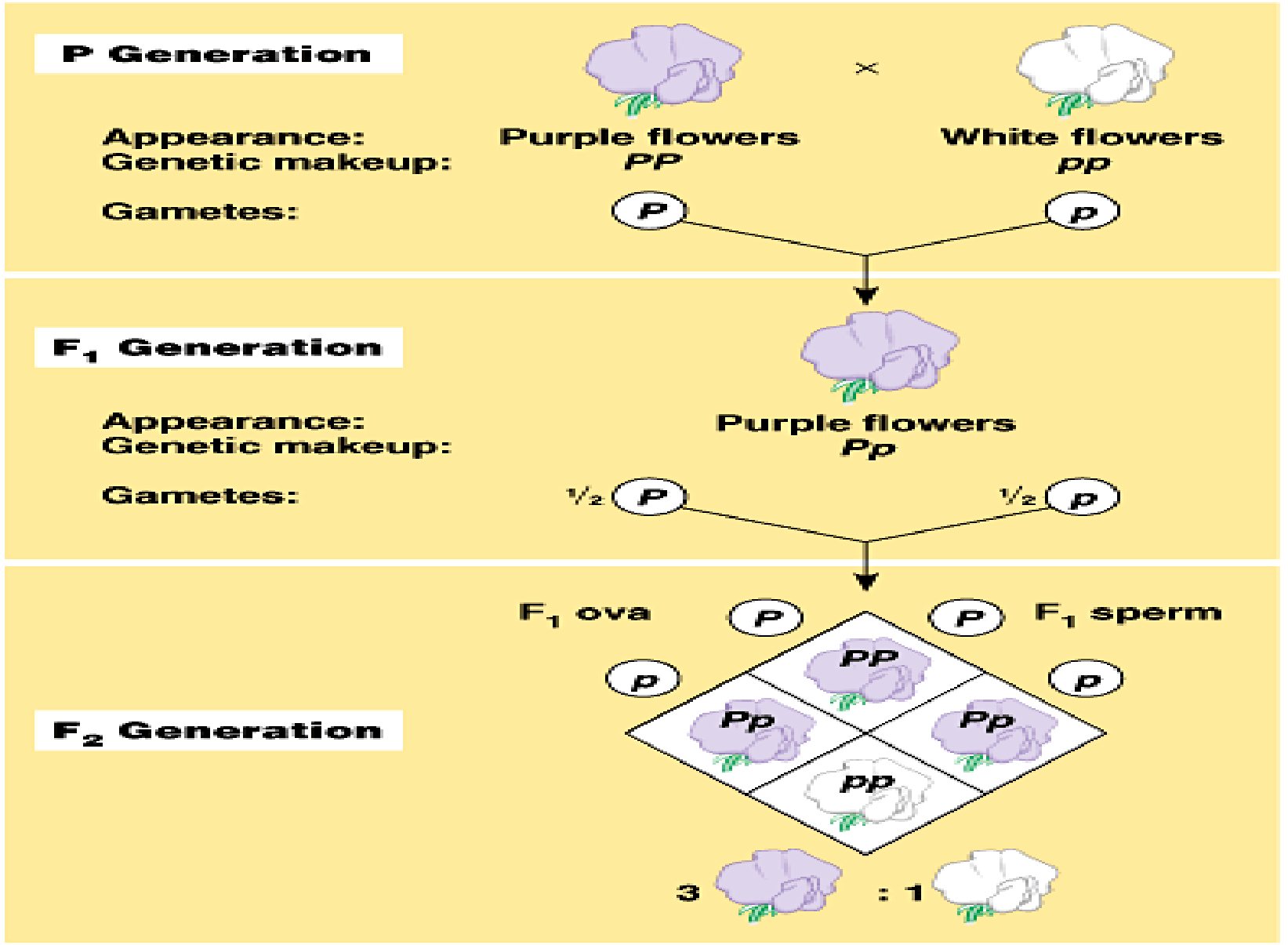
The two alleles for each trait separate during gamete formation.

Law of Segregation

- During the formation of gametes (eggs or sperm), the two alleles responsible for a trait separate from each other.

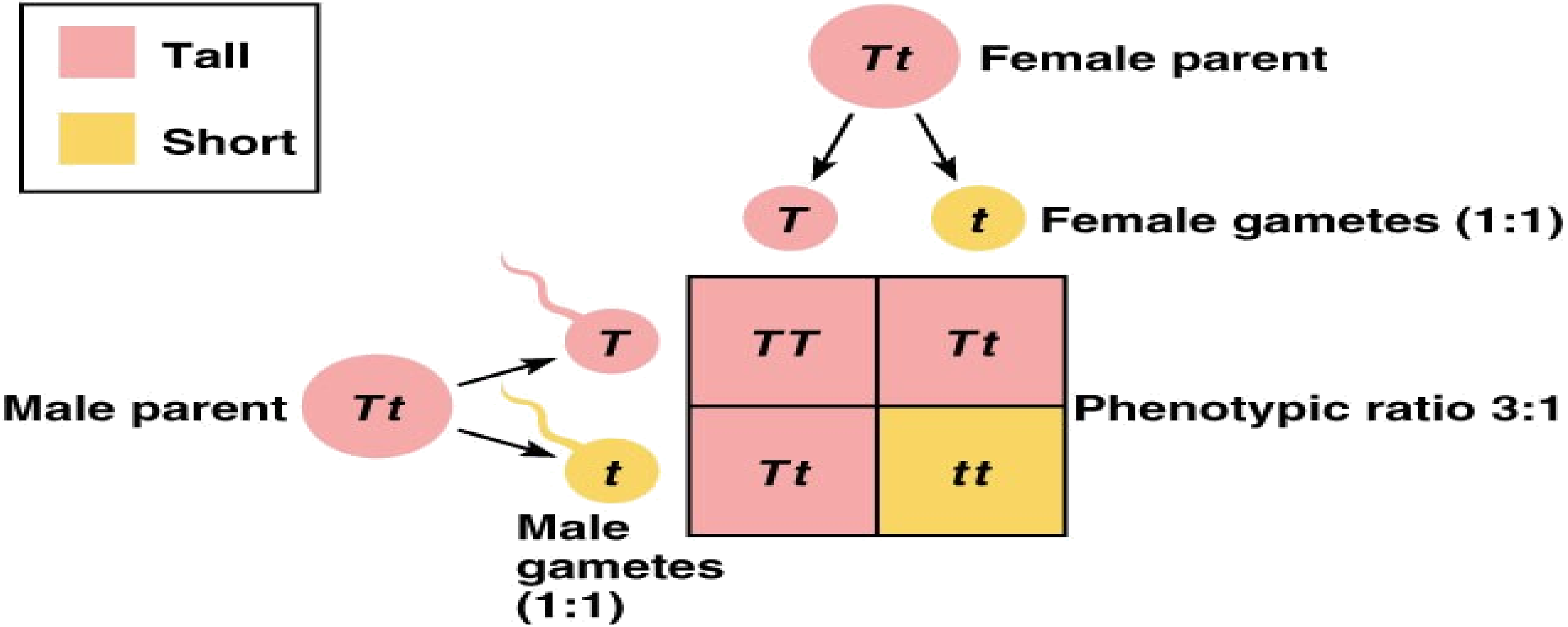
- Alleles for a trait are then "recombined" at fertilization, producing the genotype for the traits of the offspring.

Law of segregation



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Applying the Law of Segregation



Law of Independent Assortment

- Alleles for ***different*** traits are distributed to sex cells (& offspring) independently of one another.

- This law can be illustrated using ***dihybrid crosses***.

Dihybrid Cross

- **Traits: Seed shape & Seed color**
- **Alleles: R round**
  r wrinkled
  Y yellow
  y green
-

RrYy x RrYy

RY Ry rY ry

RY Ry rY ry

All possible gamete combinations

Dihybrid Cross

	RY	Ry	rY	ry
RY	RRYY	RRYy	RrYY	RrYy
Ry	RRYy	RRyy	RrYy	Rryy
rY	RrYY	RrYy	rrYY	rrYy
ry	RrYy	Rryy	rrYy	rryy

Round/Yellow: 9
Round/green: 3
wrinkled/Yellow: 3
wrinkled/green: 1

9:3:3:1

Trihybrid and Multihybrid Crosses

Mendel extended his observations to trihybrid crosses involving three pairs of contrasting characters. The characters he considered were: seed shape – smooth (*S*) vs. wrinkled (s); colour of cotyledons – yellow (*Y*) vs. green (*y*); and flower colour – violet (V) vs. white (*v*).

P: Smooth Yellow Violet × Wrinkled Green White

SSYYVV *ssyyvv*

Gametes: *SYV* *syv*

F_1: Smooth Yellow Violet × Self

SsYyVv

Gametes: SYV, SyV, SYv, Syv

sYV, syV, sYv, syv

F_2: 27 Smooth Yellow Violet: 9 Smooth Yellow White:
9 Smooth Green Violet: 9 Wrinkled Yellow Violet:
3 Smooth Green White: 3 Wrinkled Yellow White:
3 Wrinkled Green Violet: 1 Wrinkled Green White.

Use the branching or forked line method and find

A)Possible gametes in F1(No, type and proportion)
B)Possible genotypes in F2 (No, type and proportion)
C)Possible phenotypes in F2 (No, type and proportion)

Summary of Mendel's laws

LAW	PARENT CROSS	OFFSPRING
DOMINANCE	TT x tt tall x short	100% Tt tall
SEGREGATION	Tt x Tt tall x tall	75% tall 25% short
INDEPENDENT ASSORTMENT	RrGg x RrGg round & green x round & green	9/16 round seeds & green pods 3/16 round seeds & yellow pods 3/16 wrinkled seeds & green pods 1/16 wrinkled seeds & yellow pods

Basic Principles of Mendelian genetics

- Mendelian inheritance is a type of biological inheritance that follows the principles originally proposed by Gregor Mendel in 1865 and 1866.
- It was re-discovered in 1900 and popularized by William Bateson. These principles were initially controversial and even rejected by some scholars.
- When Mendel's theories were integrated with the Boveri–Sutton chromosome theory of inheritance by Thomas Hunt Morgan in 1915, they became the core of classical genetics.
- Ronald Fisher combined these ideas with the theory of natural selection in his 1930 book.
- The genetic theory of natural selection, putting evolution onto a mathematical footing and forming the basis for population genetics within the modern evolutionary synthesis.

REASONS FOR MENDEL'S SUCCESS

An important aspect of Mendel's success can be traced to his decision to start his crosses only with plants he demonstrated were **true-breeding.**

- Mendel studied the inheritance of only one pair of contrasting characters at a time He only measured **discrete (binary) characteristics**, such as color, shape, and position of the seeds, rather than quantitatively variable characteristics.
- He expressed his results numerically and subjected them to **statistical analysis.**
- His success was his ability for an accurate and incisive analysis of the reasons for failure of earlier workers
- Mendel carried out his experiments with great care and elaborateness
- His knowledge of mathematics was a definite asset for the interpretation of his findings
- Mendel was able to formulate **appropriate hypotheses** on the basis of the explanation he offered for his experimental

REASONS FOR MENDEL'S SUCCESS

Mendel's conclusions

Mendel's conclusions/deductions

1) Characters are unitary they are discrete (purple vs. white, tall vs. dwarf).

2) Genetic characteristics have alternate forms, each inherited from one of two parents. Today, we call these *alleles*.

3) One allele is dominant over the other. The phenotype reflects the dominant allele.

4) Gametes are created by random segregation. Heterozygotic individuals produce gametes with an equal frequency of the two alleles.

5) Different traits have independent assortment. In modern terms, genes are unlinked

Comparing genes and chromosomes

Chromosomes	Mendel's gene
1. Chromosomes occur in pair and known as homologous chromosomes	Mendel's genes occur in pairs
2. Paired chromosomes separate during meiosis. Each sex cell receives only one chromosome of the pair.	Mendel's paired genes separate. Each sex cell receives only one gene of the pair.
3. Pairs of chromosomes pairs separate independently of other chromosomal pairs during meiosis.	Mendel's gene pairs separate independently of other gene pairs.
4. Individual chromosomes return to their original paired number in the fertilized egg.	Mendel's individual genes become paired genes in the offsprings.

ABO blood groups and Rh Factors

- Early experiments with human blood transfusion often resulted in the death of the patient for unknown reasons.

- In 1901, it was discovered that there were three blood types, A, B, and O, and that mixing blood from different types caused an immune response that resulted in clumping.
- Type AB is rare and was discovered later.

ABO Blood Type:

- An individual's red blood cells will contain proteins of type A, or B, or both, or neither.

- The body produces antibodies that will attack any foreign type.

- Alleles of types $\mathbf{I_A}$ and $\mathbf{I_B}$ are dominant over type **i**.

ABO blood groups and Rh Factors

Types of blood

Universal donor

Universal acceptor

Genotype	Blood Type	Antigens	Antibodies	Donate To	Receive From
i i	Type O	None	Anti-A and Anti-B	A, B, AB, O	O
I_A I_A	Type A	A	Anti-B	A, AB	A, O
I_A i	Type A	A	Anti-B	A, AB	A, O
I_B I_B	Type B	B	Anti-A	B, AB	B, O
I_B i	Type B	B	Anti-A	B, AB	B, O
I_A I_B	Type AB	A and B	None	AB	A, B, AB, O

Blood groups (phenotype)	Antigen in red blood cells	Antibodies in plasma	Can give blood to groups	Can receive blood from group	Genotype
O	None	Anti-A, Anti-B,	O, A, B, AB	O	ii
A	A	Anti-B	A, AB	O, A	I^AI^A or I^Ai
B	B	Anti-A	B, AB	O, B	I^BI^B or I^Bi
AB	A and B	None	AB	O, A, B, AB	I^AI^B

ABO Blood Type

Rh Factor (D antigen)

- The Rh factor, the second most important blood group system after the ABO blood group system, was first discovered in Rhesus monkeys.
- The Rh factor is inherited independently from the ABO blood type.
- Genotypes for the Rh factor are **+/+**, **+/-**, and **-/-**.
- People who are +/+ or +/- possess the Rh(D) antigen and test as Rh positive.
- People who are -/- do not possess the Rh(D) antigen and test as Rh negative.

ABO Blood Type

Genotype	Phenotype	Blood Proteins	Blood Antibodies	Donate To	Receive From
+/+	Rh+	Rh(D) proteins	None	Rh+	Rh+ or Rh-
+/-	Rh+	Rh(D) proteins	None	Rh+	Rh+ or Rh-
-/-	Rh-	None	Anti-Rh(D)	Rh+ or Rh-	Rh-

Rh Sensitization

Rh Sensitization

- One interesting medical scenario involves an Rh negative mother who carries an Rh positive baby.
- The baby of an Rh positive father and an Rh negative mother can be +/- or -/-)
- If the baby is +/-, the first pregnancy causes Rh sensitization in the mother, because she is exposed to foreign proteins and builds up antibodies against them.
- Future pregnancies can be increasingly difficult, as the mother's antibodies attack the baby.

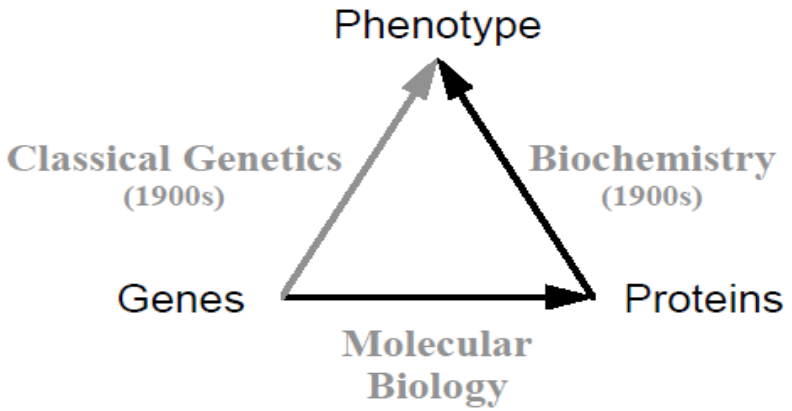
Possible blood phenotypes when Rh and ABO blood systems are considered simultaneously

ABO blood types	Rh antigen	Blood phenotype
A	Rh^+ Rh^-	A^+ A^-
B	Rh^+ Rh^-	B^+ B^-
AB	Rh^+ Rh^-	AB^+ AB^-
O	Rh^+ Rh^-	O^+ O^-

Hemolytic Disease of the Newborn/Erythr-oblastosis fetalis.

Hemolytic Disease of the Newborn/Erythroblastosis fetalis.

- **This condition results from incompatibility** between maternal and fetal blood groups.
- Rh +ve fetus may produce antibodies in Rh –ve mother.
- In **Rh incompatibility, the first child** often escapes.
- But in the second pregnancy, the Rh antibodies will pass from mother to the fetus.
- They would start destroying the fetal red cells even before birth.
- **Sometimes the child is born with severe** hemolytic disease, often referred to as **erythroblastosis fetalis.**



Molecular genetics and inheritance

DNA, Gene, Chromosomes and Cell division

- DNA (deoxyribonucleic acid)-
  - it is the molecule that holds the genetic information for a cell and an organism.
  - contains a code that can be used by a cell to express certain genes.
- Specific sections of a DNA molecule provide the information to build specific proteins which can then be used by a cell to express the desired gene.
- DNA comes in the form of a long, linear molecule referred to as a strand.
- Each strand of DNA is bonded to a second strand of DNA to form a DNA double helix.
- In eukaryotic cells, DNA is found in the nucleus as a tightly coiled double helix.
- DNA molecules are replicated during cell division.
- When a cell divides, the two new cells contain all the same DNA that the original cell had.
- In sexual reproduction with two parents, half of the DNA of the offspring is provided by each of the parents.
- The genetic material of a child is made from 50% of their mother's DNA and 50% their father's DNA.

Structure of DNA and chromosome

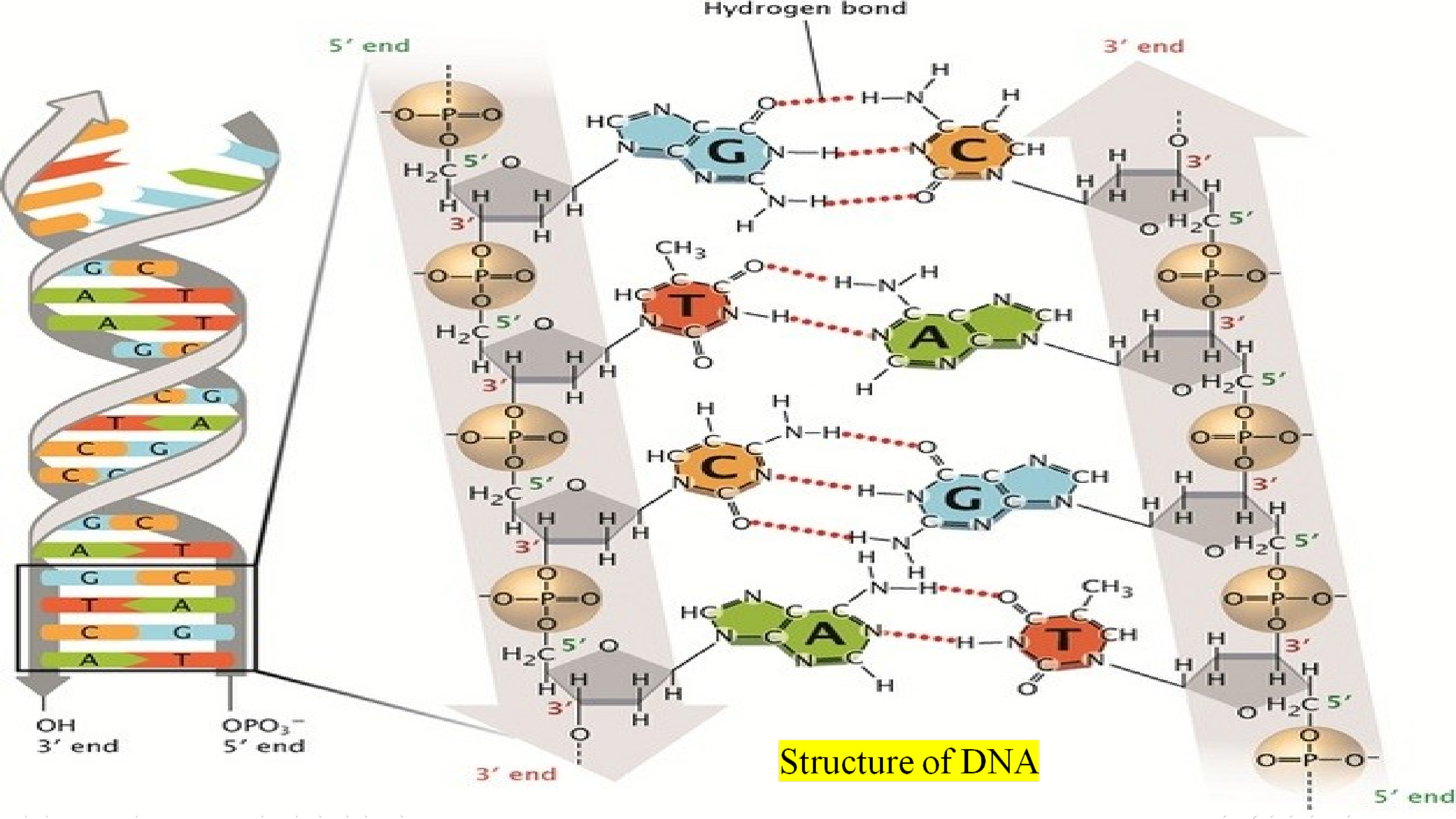
- **Chromosome** is a thread- like structure that is made up of DNA (deoxyribonucleic acid) and histone (a set of globular (3^0) proteins).
- While DNA is the molecule that stores genetic information whereas histone is the core of a chromosome around which chromosome's DNA wrapped is the loosely organized form of chromosome throughout the nucleus in loops when the cell is not dividing is called **Chromatin**.
- Individual chromosomes are not easily distinguished unless condensed.
- On the condensed organization of chromosome, genes are inactive while uncondensed or loose organization allows the genes to be active.
- As a cell prepares for cell division, the chromatin loops will have duplicated themselves and become compacted or "condensed" to form a chromosome that is visible under light microscope.

Structure of DNA and chromosome

- DNA-is made up two strands of polynucleotides joined together and twisted into a double helix and the strands are anti-parallel to each other.
- The basic unit of DNA strand is a nucleotide (monomers of DNA). There are four types of nucleotides:-
  - Adenine (A) – containing nucleotide
  - Guanine (G) – containing nucleotide
  - Cytosine (C) - containing nucleotide
  - Thymine (T) – containing nucleotide (in DNA, or Uracil (U) nucleotide in RNA)
- All nucleotides have:
  - a phosphate group,
  - a pentose sugar (deoxyribose in DNA and ribose sugar in RNA) and
  - one of four nitrogen bases- adenine, cytosine guanine and either thymine (DNA) or uracil (RNA).

Structure of DNA and chromosome

- Bonds between the sugar in one nucleotide and the phosphate group in the next hold the nucleotides together.
- The base does not take in this linking of the nucleotides in a strand. That is the reason why we say ‘sugar- phosphate’ backbone.
- The nucleotides in one strand are paired with the nucleotides in the other strand according to the base pairing- rule.
  - Adenine – Thymine(uracil in RNA)
  - Cytosine – Guanine
- DNA is a very stable molecule at normal temperature.
- The hydrogen bonds hold the two strands together in position through the bases.
- The stability of the DNA molecule is important in ensuring the genetic code in the DNA molecule – does not become corrupted.



Structure of DNA

DNA replication and cell division

- The ability of DNA to make copy of itself (DNA-replication) is the basis for reproduction and inheritance.
- DNA molecule exists within chromosome (in the nucleus) and is surrounded by a 'soup' of free DNA nucleotides (which is to build new DNA molecules).
- DNA structure is double helix and must replicate **semi-conservatively.**

 Each formed new DNA molecule contains one strand from the original (old) DNA and one new strand DNA molecules.

- Both new DNA molecules formed are identical to each other and to the original molecule.
- **Several Enzymes** are involved in this process and the main stages are:

DNA replication and cell division

1. DNA helicase enzyme - break H-bonds to reveal two single strands and unwind (open) the helix DNA

2. DNA polymerase follows the helicase enzyme along each single-stranded region, which acts as a template for the synthesis of a new strand.

3. DNA polymerase assembles free DNA nucleotides into new strands alongside each of the template strands. The base sequence in each of these new strands is complementary to its template strand because of base- pairing rule, A-T, C-G.

4. Two-identical DNA molecules to each other and the original one is resulted. Each contains one strand from the original (old) and one newly synthesized.

Summary of DNA Replication

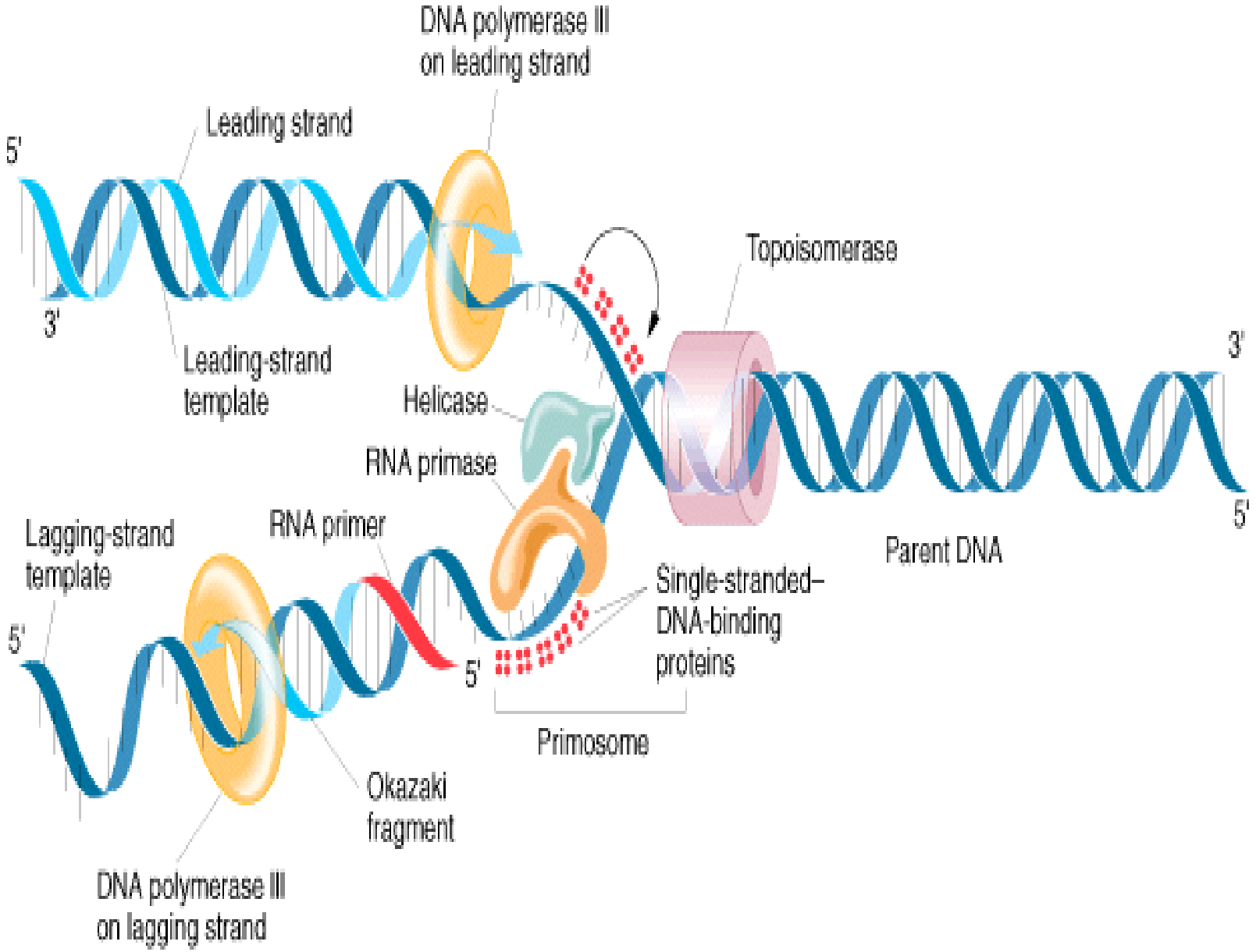
1.Origin of replication is identified. Then unwinding of parental DNA to form a replication fork.

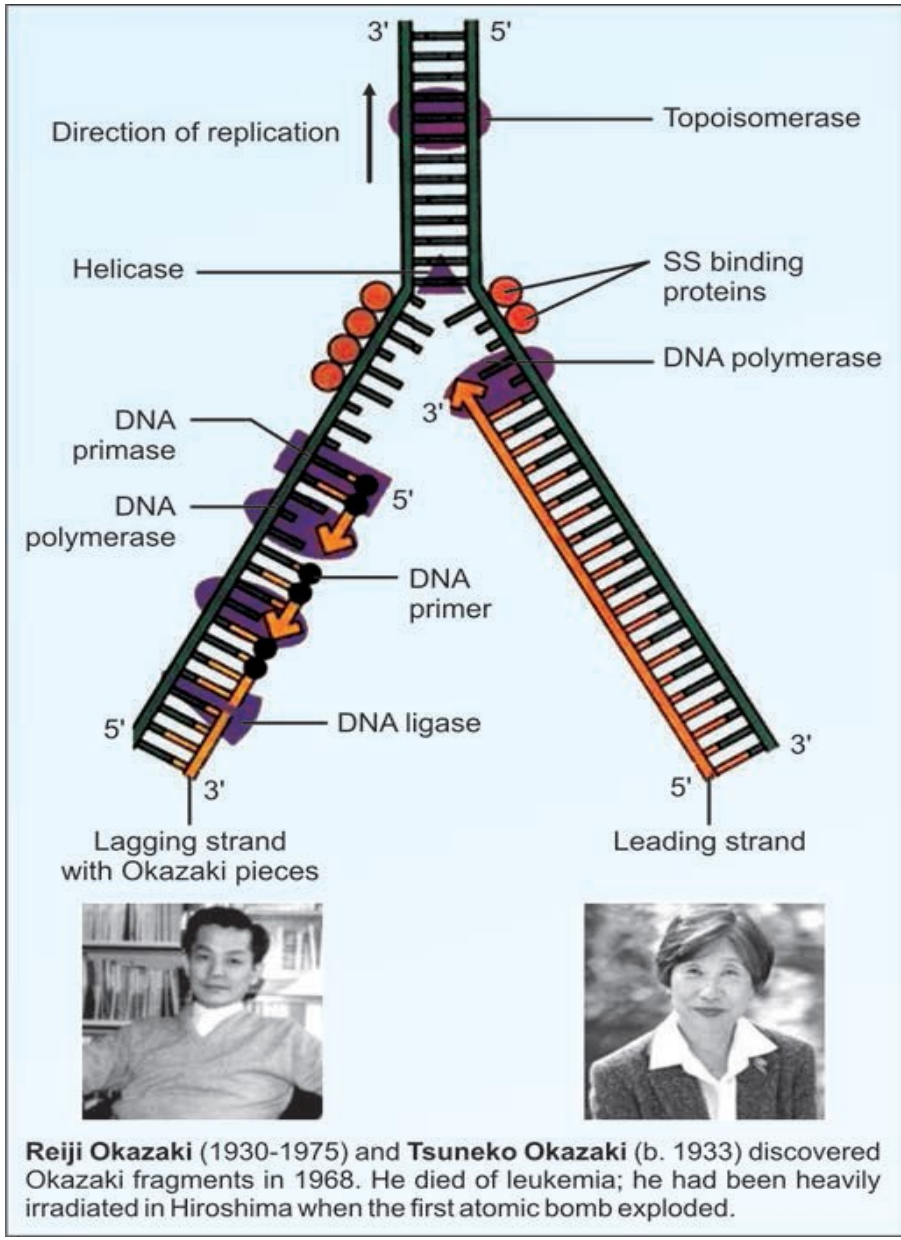
2. RNA primer complementary to the DNA template is synthesized by RNA primase.

3. DNA synthesis is continuous in the leading strand (towards replication fork) by DNA polymerase.

4. DNA synthesis is discontinuous in the lagging strand (away from the fork), as Okazaki fragments.

5. Elongation: In both strands, the synthesis is from 5' to 3' direction.
6. Then the RNA pieces are removed; the gaps filled by deoxynucleotides by DNAP and the pieces are ligated by DNA ligase.
7. Proof reading is done by the DNA polymerase.
8. Finally organised into chromatin.
9. Main enzymes involved in replication are:
DNA polymerases; Helicases; Topoisomerases; DNA primase; Single strand binding proteins; and DNA ligase.





Reiji Okazaki (1930-1975) and **Tsuneko Okazaki** (b. 1933) discovered Okazaki fragments in 1968. He died of leukemia; he had been heavily irradiated in Hiroshima when the first atomic bomb exploded.

Cell division

- Cell division is the process by which a parent cell divides into two or more daughter cells.
- Cell division usually occurs as part of a larger cell cycle.
- In eukaryotes, there are two distinct types of cell division:
  - a vegetative division, whereby each daughter cell is genetically identical to the parent cell **(mitosis),** and
  - a reproductive cell division, whereby the number of chromosomes in the daughter cells is reduced by half to produce haploid gametes (**meiosis).**
- Meiosis results in four haploid daughter cells by undergoing one round of DNA replication followed by two divisions.
- Homologous chromosomes are separated in the **first division**, and **sister chromatids** are separated in the **second division.**
- Both of these cell division cycles are used in the process of sexual reproduction at some point in their life cycle.
- Both are believed to be present in the last eukaryotic common ancestor.

Cell division

- **Prokaryotes (bacteria)** undergo a vegetative cell division known as binary fission, where their genetic material is segregated equally into two daughter cells.
- While binary fission may be the means of division by most prokaryotes, there are alternative manners of division, such as budding, that have been observed.
- All cell divisions, regardless of organism, are preceded by a single round of DNA replication.
- For simple unicellular microorganisms such as amoeba, one cell division is equivalent to reproduction; an entire new organism is created.
- On a larger scale, mitotic cell division can create progeny from multicellular organisms, such as plants that grow from cuttings.

Cell division

- Mitotic cell division enables sexually reproducing organisms to develop from the one-celled zygote, which itself was produced by meiotic cell division from gametes.
- After growth, cell division by mitosis allows for continual construction and repair of the organism.
- The human body experiences about 10 quadrillion cell divisions in a lifetime
- The primary concern of cell division is the maintenance of the original cell's genome.
- Before division can occur, the genomic information that is stored in chromosomes must be replicated, and the duplicated genome must be separated cleanly between cells.
- A great deal of cellular infrastructure is involved in keeping genomic information consistent between generations.

Protein synthesis

- Code for protein synthesis is specified by DNA and has to be sent to ribosome. DNA is a huge molecule and remains in nucleus to assemble amino acids in the correct sequence to form protein.
- **Events during protein synthesis**
  - Transcription – DNA code for protein is rewritten in a molecule of messenger RNA (MRNA)
  - MRNA travels from nucleus to ribosome
  - Free amino acids are transported from cytoplasm to ribosome by transfer RNA (tRNA) molecules
  - Ribosome read mRNA code and assembles amino acids presented by tRNA into a protein by a process = translation
  - Translation – is process in which mRNA code is converted into a sequence of amino acids



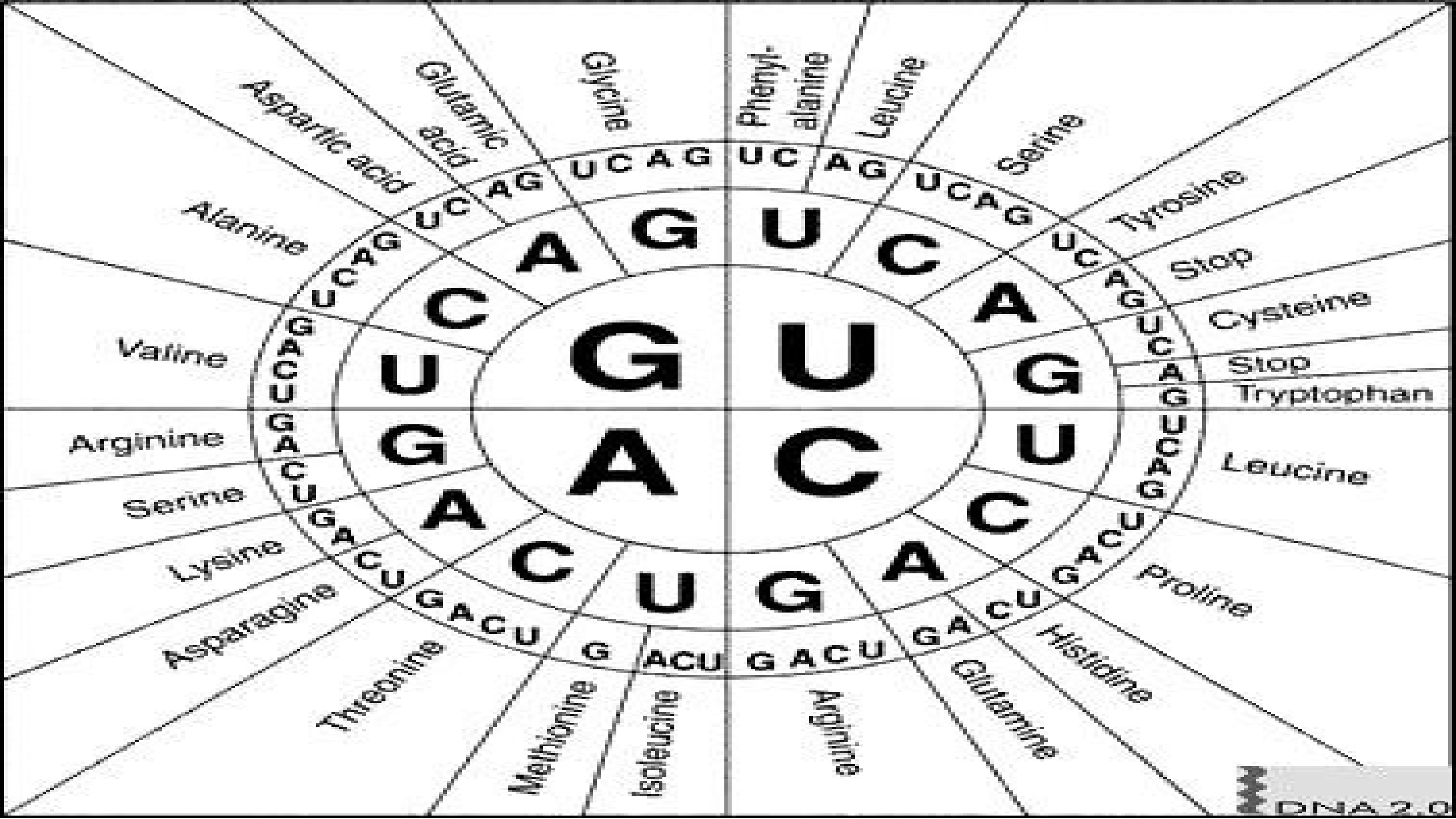
Genetic code

- The genetic code is held in the DNA molecule.
- It is because of the nucleotides sequence in the DNA molecule made a gene that codes for protein to that each amino acid in the protein is coded for by a triplet /sequence of three bases/.
- Hence, a gene is a sequence of base triplets in the DNA molecule that carries a code for a protein.

1. Since there are 4 bases, there is $4^3 = 64$ possible triplet codes /amino acids/ but only 20 amino acids are used to make all different proteins.
    - None of them is spare or redundant.

Genetic code

- 2. Only one strands of a DNA molecule carries the code for proteins (It is called sense strand or coding stand) and the other strand is non-coding or antisense strand.
- 3. Most amino acids have more than one code, only methionine and tryptophan have one code.
  - Arginine has six codes
- 4. Three triplets (TAA, TAG, and TGA) do not code for amino acids and they are called stop codons. stop codes signify the end of the coding sequence
- 5. Degenerate code – the DNA code is non-overlapping codes i.e., each triplet is distinct from all other triplets.
- 6. The genetic code is also a universal codei.e. the triplet code TAT in the DNA code for amino acid tyrosine in human, redwood tree, bacterium or in any organism
  - E.g. ACC – threonine, GGG-glycine


G U A C
U C A G
Glycine
Phenyl-alanine
Leucine
Serine
Tyrosine
Stop
Cysteine
Stop
Tryptophan
Leucine
Proline
Histidine
Glutamine
Arginine
Isoleucine
Methionine
Threonine
Asparagine
Lysine
Serine
Arginine
Valine
Alanine
Aspartic acid
Glutamic acid
DNA 2.0

Genetic code

- N.B. It is not just the base that matter but the sequence, e.g.ATT codes of different amino acid from TTA
- The enzyme DNA-dependent RNA polymerase binds with a section of DNA next to the gene to be transcribed. Transcription factors activate the enzyme.
- The enzymes “unwind” and separate the strands of a DNA molecule.
- The polymerase assembles free RNA nucleotides on the complementary strand (antisense strand). mRNA (transcribed RNA) leaves the DNA; the strands of DNA rejoin & re-coil.
- N.B. The mRNA molecule now contains the code for the protein that was held in the DNA of the genes

Genetic code

- Translation of mRNA code into a protein depends on the interaction within a ribosome between mRNA and tRNA. All tRNA molecules have same basic structure.
- The ''cloverleaf'' configuration of the molecules has at one end a triplet of bases called anticodon. The anticodon is a triplet on tRNA.
- The other end of tRNA molecule has an attachment site for the amino acid that is specified by the mRNA codon.
- Ribosomes are made from ribosomal RNA and proteins organized into large & small subunits.
- Within ribosome, there are three sites called A, P and E sites.

Events during translation of proteins

- The first two codons of the mRNA enter the ribosome and tRNA molecules (with amino acid attached) that have complementary anticodons to the 1st two codons of the mRNA bind to those codons.
- A peptide bond forms between amino acid carried by two tRNA molecules and dipeptide is transferred to the tRNA in the A-site.
- The Ribosome moves along the mRNA by one codon bringing the 3rd codon into the ribosome.
- At the same time the free tRNA exits the ribosome from the E-site and tRNA with the dipeptide moves into the P-site.
- A tRNA with complementary anticodon binds with the third codon and brings its amino acid into position next to the 2nd.
- A peptide bond is formed between 2nd and 3rd amino acid. The whole process is repeated until a ''stop'' codon is in position and translation is stopped.

- Note: - TGA, TAA and TAG – are stop codons.
- N.B. The translation of the mRNA code into a protein molecule requires energy that comes from hydrolysis of GTP /guanoisne triphosphate/ not ATP
- Though it is similar to eukaryotes, there are some differences like: Prokaryotic cell have no nucleus. Prokaryotic mRNA does not need post-transcriptional process because it only contains exons (coding genes) not introns (non-coding genes).
- Transcription and translation are coupled - mRNA translated at one end of ribosome while it is start transcribing from DNA at other end.
- Transamination: - is a process of amino group of an amino acid is removed and transferred to a keto acid, then it becomes a different amino acid/keto acid
- E.g. pyruvic acid (keto acid) → alanine (amino acid) - by transamination.
- Note. Not all amino acids are produced by transamination only those we obtain from our food. They are called essential amino acid.

Control of Gene expression

- Genes are switched on by "transcription factors"- are proteins that bind to a regulatory sequence of DNA near to the gene they influence. They operate in the following way:
- The transcription factors bind to a promoter sequence of DNA near the gene to be activated.
- RNA polymerase binds to the DNA/ transcription factor complex.
- The RNA polymerase activated and moves away from the DNA along the gene.
- The RNA polymerase transcribes the antisense strand of the DNA and the gene is now being expressed.

Control of Gene expression

- Like transcription factors that promote gene expression, there are factors to repress (switched off) gene action. Short interfering RNA (siRNA)
- are unusual-very short
- only 21-23 nucleotides
- double stranded
- They don’t act on the gene itself, but interfere/silence the mRNA once it has been transcribed from DNA.
- Is post transcriptional interference
- If mRNA is prevented from translating its Codons into amino acid then the protein for which the gene codes for cannot be built (silenced).

Mutations

- **Mutation** is the alteration of the nucleotide sequence of the genome of an organism, virus, or extrachromosomal DNA
- Mutation could occur spontaneously (accidentally) during duplication. They are rare events

Mutations

- Mutation could occur spontaneously (accidentally) during duplication.
- They are rare events.
- Each cell contains $6x10^9$/billion/ base pairs

occurrence $\frac{1}{50 \times 10^6}$

- Each new cell has an average 120 mutations. But
- Most mutations detected & repaired
- 95% of our DNA is non-coding, most mutation unlikely to affect coding gens.
- Rate of mutations can be increased by:-
- Carcinogenic - chemical in tobacco smoke
- High energy radiation- ultraviolet radiation, x- ray

Mutations

- There are many different ways that DNA can be changed, resulting in different types of mutiation as described below:
- Point mutations- changes that involves a single base. There are several types of point mutations, some of these are:
- Substitution- one base is replaced by other base
- Addition-a base is missed out during DNA replication.
- Deletions – an extra base is added.
- Chromosomal Mutations- situation where part /segment/ of a chromosome sequence of DNA is disturbed/ a chromosome is missed/added. Types of chromosomal mutations:

- Euploidy/polyploidy/-is a condition where an organism/ a cell has one complete set of extra chromosomes/an exact multiple of a complete set.
- Variation in number of chromosomes sets (one set of chromosome). E.g – Triploid ,Tetraploid
- Aneuploidy – is a condition where an organism/a cell lost from or added to one or more chromosomes to/the normal set of chromosome
- 2 n+1 = trisomy/47 chromosomes, e.g. Down's syndrome
- 2n-1 = monosomics – turner syndrome/45chr.
- 2n+1+1 = double trisomy
- 2n-2 – nullisomic, organisms that loss one homologous chromosome.
- Gidu syndrome = mental retardation – caused by part of deletion of chromosome number 5 of man.

Consequence of gene-mutations

- Mutations on body cell (non- sex cell) cannot be inherited but may result in one of the following:
- harmless
- damage the cell
- kill the cell
- Make cell cancerous- which might kill the person.
- Mutations in different genes will obviously produce different effects but two types of gene are important in regulating cell division and preventing the formation of a tumor.
- Proto-oncogenes-when proto-oncogenes mutate,they often become active oncogenes-which stimulate cell to divide in an uncontrolled manner.
- Tumor-suppressor genes- recognize this uncontrolled cell division and act to suppress cell division.
- If these genes mutate and become inactive, a tumor will form as uncontrolled cell division continues.
- **Tumor**-is a mass of cells created when cell replication gets out of control and cause cancer.

Manipulation of DNA Genetic engineering

- Genetic engineering is a process by which the genome of an organism is altered, usually by having extra (foreign) gene from different organisms, and the organism is termed genetically modified organism (GMO), transgenic organism or genetically engineered organism.
- Genetic engineering is early done on Bactria, to produce useful products like:
- Insulin, antibiotics, washing enzyme
- Enzyme for food processing industry
- Human growth hormone
- Vaccines-Hepatitis B
- Bovine somatotrophin, high yield milk and muscle development in cattle.

Manipulation of DNA Genetic engineering

- Plants are also genetically modified:
- To absorb more CO_2 – global warming
- Disease (drought) resistant
- Improved yield
- To produce specific product e.g. golden rice produce beta-carotene (in vitamin 'A') prevent night blindness.
- Moreover, animals are genetically modified for the following purposes
- high yield of protein, high growth rate
- Specific products eg. insulin,
- Detection of water pollution = glofish glow in the dark b/c they have had bioluminescent gene from jelly fish.
- Genetically modified salmon and Tilapia fish grow bigger and faster.

Manipulation of DNA Genetic engineering

- Some other potential applications of genetic engineering are:
- Disease could be prevented by detecting defected genes.
- To treat infectious diseases by implanting antiviral proteins (antibodies).
- To produce plants and animals that have high growth rate and reduced susceptibility to disease – reduce use of fertilizers and pesticides.
- Animals and plants can be 'tailor made' to show desirable characteristics

Introduction to Evolution

- Evolution is the gradual change of organisms on the earth over long periods, with new forms replacing old ones.
- As evolution has progressed new species are arising, the biodiversity of the planet increasing, larger and more complex organisms replaced the smaller and some species has get extinct.

5.4.1 Theories on the origin of life on Earth

- Evolution can also be the change in genetic composition of a population over successive generations which may be caused by meiosis, hybridization, natural selection or mutation.
- This leads to a sequence of events by which the population diverges from other populations of the same species and may lead to the origin of a new species.
- Theories about the origin of life on Earth.
- The theory of evolution describes how the various forms of life on earth (including humans) emerged and developed.
- There are five main theories of the origin of life on Earth:
  - special creationism
  - spontaneous generation
  - eternity of life
  - cosmozoan theory
  - biochemical origin

a) Special creationism

- Special creation theory states that the different forms of life on earth were created by a Supreme Being/ God/ at once with six days.
- Special creation is always linked to religion and mainly focused on spiritual matters that cannot be seen, touched or measured effectively. There are two views on the origin of life:
- **Types of creationism theory**
- **Gap creation** – discusses a large gap between the formation of the earth and the creation of all the animals and plants.
- The gap could be billions or millions of year.
- Progressive creation – accepts the Big Bangs as the origin of the universe. It accepts the fossil record of a series of creation for all of the organisms but it does not accept these as part of a continuing process (each is seen as unique creation).

- **Evolutionary creationism** (Theistic evolution) – This view of evolution maintains that God ‘invented’ evolution and takes some form of an active part in the ongoing process of evolution.
- **Intelligent design** – a theory states that life developed (formed) due to a combination of natural forces and the intervention of a supernatural being.
- **b) Spontaneous generation theory**
- Suggests that life can evolve 'spontaneously' from non-living objects. E.g. people believed that rotting meat turned into flies.
- **c) Eternity of life**
- The theory of eternity of life states that the universe has always existed and that there has always been life in the universe. There is no beginning and no end to life on earth and so life is neither created nor generated from non-living matters

- **d) Cosmozoan Theory**
- States that life on the earth originally came from elsewhere in the universe (possibly from another planet). E.g. Meteorites brought bacterial spores, germs to the earth.
- This theory did not gain any significant support because it lacks evidence and it is strongly linked to the ‘eternity of life’ theory of the origin life.
- **e) Biochemical theory**
- It suggests that life on earth originated as a result of a number of biochemical reactions producing organic molecules, which combined (associated) to form cells.
- This theory is also called abiogenesis; states life originates from chemical inanimate (abiotic substances).
- The two scientists (biologists) who developed the theory of abiogenesis (origin of life from chemicals) were:- Aleksander Oparin (1924) and John Haldane (1929). They both suggested:

- The primitive atmosphere of the earth was a reducing atmosphere – no free oxygen (no oxygen gas).
- There was an appropriate supply of energy, such as lightening or ultraviolet light (Uv – rays).
- This would provide the energy for reactions that would synthesize a wide range of organic compounds (amino acids, sugars and fatty acids).

- Oparin suggested that the atmosphere of the primitive earth contained NH_3, H_2, water vapour (H_2O (g)) and CH_4. He suggested that at high temperature (above boiling point), these gases could have undergone a series of chemical reactions and might have formed colloidal aggregates or coacervates (collections of droplets that composed of molecules of different types). Oparin considered that these coacervates were able to absorb and assimilate organic compounds (amino acids, DNA and others) and leading to the first true cells.
- J. Haldane – proposed that the chemical reactions of these gases were occurred in the primitive sea by solar energy and the sea became a ‘hot dilute soup’ of organic monomers and small polymers.

- In 1953, Stanley Miller tested the biochemical theory. He applied electrical sparks repeatedly through a mixture of gases (NH_3, water, H_2 and CH_4) that were represent the primitive atmosphere of the earth (no oxygen) connected a flask of heated water.
- Miller leaving the equipment for longer periods of time, a larger variety and more complex organic molecules were formed including:
- amino acids - to form proteins
- pentose sugar - to form nucleic acids
- hexose sugar - need for respiration and to form starch and cellulose.
- Hydrogen cyanide (HCN)- starting point for synthesizes of nitrogen containing bases in nucleotides.

- This experiment gave strong evidence to support the Oparin-Haldane hypothesis.
- Origin of Autotrophy: The first life (cell) appeared on earth – about 4 billion years ago and they were:- anaerobes, heterotrophs and prokaryotes. They would have fed on organic chemicals (soup) which formed spontaneously in the waters of the earth. The three consecutive lines of evolution of organisms on earth leading to:
- Archaebacteria - are first appeared organisms. They are prokaryotes including thermophilic (high temp. loving), sulphobacteria, methanobacteria and halophilic (salt loving) bacteria.
- They can survive under harsh (extreme) environment.
- Eubacteria – are true (ordinary) bacteria, ordinary bacteria and cyanobacteria (blue – green algae)
- Prokaryotes – evolved into protista, fungi, plants, animals (nearly all are aerobes).

- Later (3.5 billion years ago) – the first autotrophs (photosynthetic organisms) like cyanobacteria (blue-green algae) came in existence.
- With the evolution of chlorophyll molecules, oxygen (O_2) was released as a by-product of photosynthesis and this changes the earth's atmosphere from the reducing to an atmosphere containing oxygen.
- The fossil record shows that cyanobacteria had been producing oxygen by photosynthesis from about 305 million years ago but the levels of oxygen in atmosphere did not rise for almost 1 billion years ago.
- Because oxygen was absorbed by the vast amount iron in the earth it rusted.

5.4.2 Theories of Evolution

- There have been many theories of evolution that have explained:- how does evolution happen? And what drives the population to become a new species?
- **A. Lamarck Theory of evolution**
- In the 19th century (1809), Lamarck published a paper entitled Philosophic ‘Zoologique’ in which he described the two-part mechanism by which change was gradually introduced into the species and passed down through the generations. This theory is also called ‘theory of transformation’ or Lamarckism. The two parts of Lamarck theory are:
- Use and disuse
- Inheritance acquired traits

- Use and disuse – Lamarck suggested that a structure or process in organism that can be used continuous will become enlarged or more developed but any structure that is not.
- Example, According Lamarck, giraffe had short neck but they stretched their neck to reach high branches, developed an elongated neck use theory.
- The wings of penguins would have become smaller than those of other birds because penguins do not use their wings to fly disuse theory.
- Inheritance of Acquired traits – Lamarck believed that traits changed (acquired during an organism's lifetime could be passed on to its offsprings.
- Example: - Giraffes that had acquired long necks would have offspring with long necks.
- Note: - Nowadays, Lamarck's theories are not accepted because the environmental changes that were believed by Lamarck have brought about the changes in the phenotypes (Physical appearance) of the organisms have no effect on their gametes and hence their heredity.

B. Charles Darwin and Natural Selection

- In 1858, both Charles Darwin and Alfred Wallace jointly published a scientific paper that proposed species were modified by natural selection. Darwin visited five of the Galapagos Islands, made drawings, and collected species.
- In particular, Darwin studied the finches found on the different islands and noted there were many similarities between them but have some obvious differences.
- Darwin concluded that an “ancestral finch” had colonized the Islands from mainland and been able to adapt to the different conditions on the islands and evolve into different species.
- E.g. He suggested that some finches had evolved into insect eaters (pointed peak), other in to seedeaters (crushing peak). Darwin summarized his observations in two main ideas:
- all species tend to produce more offsprings than can possibly survive (Fecundity)
- There is a variation among the offsprings.

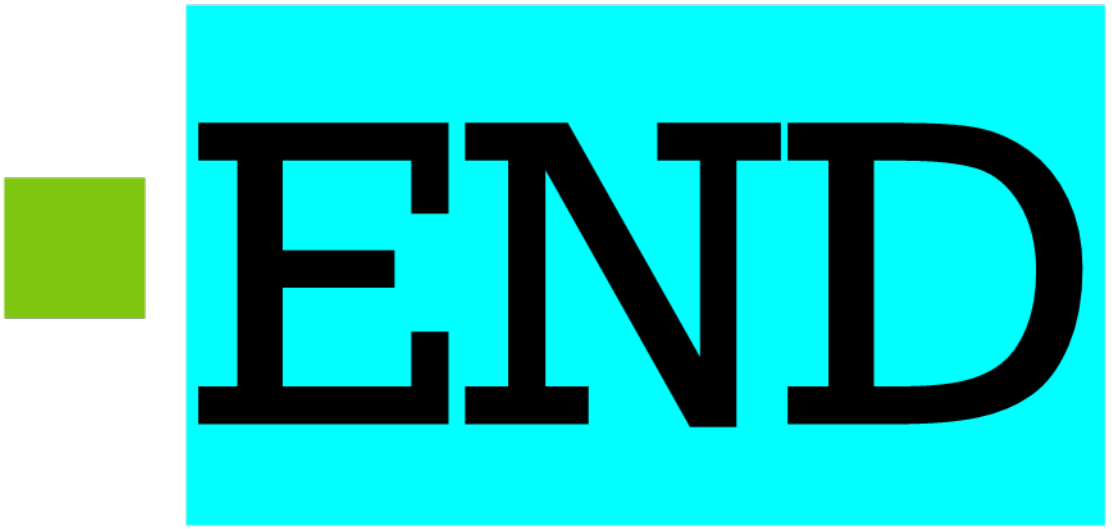
- From these observation Darwin deduced (concluded) that:
  - There will be a “struggle for existence” between members of a species because they are over – reproduced and resources are limited.
  - Some members of a species will be better adapted than others to the environment because there is a variation in the offspring.
- Darwin proposed: - Those members of a species, which are best adapted to their environment, will survive and reproduce in greater number than other less adapted (died out).

C. Neo – Darwinism Theory

- Charles Darwin knew very little about genetics and did not propose how a variation in the population was passed to the next generation.
- Now day's genes and gene action are the driving force of evolution in the theory of Natural selection.
- A Gene pool is all the alleles in the population. It might be evolve a population into a new species.
- Suppose an allele determines a feature that gives an organism an advantage in its environment. The following will happen
  - Those individuals with the advantageous allele of a gene will survive to reproduce in greater number than other types
  - Advantageous alleles pass to their offsprings in greater numbers than other genes (alleles).
  - The frequency of the advantageous allele will be higher in the next generation of a population.

- Mutations are important in introducing variation into population. Any mutation could produce an allele which:
- Increase in frequency if they are beneficial in their effect, may increase slowly, stable or decrease if they are neutral and decrease and could disappear if they are harmful (disadvantages) in their effects.
- Neo-Darwinism is a modification of Darwin’s original theory that takes into account: - genetics and ethology(behavioral patterns can also be advantageous or not).
- E.g. Young geese ‘imprint’ upon the first moving object after they are hatched.

END



Work/Study hard for successful Achievements in your life/ and make your dreams come true.