Sanjay Khare's

GENETICS

For NEET, Medical
& Biotech Students

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Preface

This book "**Genetics**" is primarily designed to the students preparin g for various Medical Entrance Exams. The book has been written to cover all the important points which cover the entire syllabus of Unit-Genetics for NEET.

During preparation of this book I have tried to bear in mind the needs of students to enable them to understand the subject in an easy manner and they can revise quickly the whole subject in a short time.

Needless to say, many of my brilliant students who have secured hundred percent marks in Biolog y in PMT exams, and most of them are now well estab lished doctors, gave me feedback over the preceding years of my teaching concerning the study material, style and its relevancy.

Dr. Rashmi Khare, my younger sister and my student, who herself got 100% marks in PMT Biology and is alumna of King George Me dical College, now a we ll established professional has revised the original Genetics book written by me in a very simple language covering all the aspects of NEET.

It is a pleasure to acknowledge the debt I owe to all my selected students including my sister Dr. Rashmi for inspiring and encouraging me to writing this book so that maximum students can get perfection in the subject after reading the book.

I am thankful to M s Pratibha and Ms. Ruma Mishra to helping me in manuscript writing. Many persons ha ve worked for many days together for the preparation of this book. I offer my appreciation and wish to all of them who helped and invested their valuable time in designing the concept of this book.

Finally I must admit that my family members specially my youngest sister Trapti Khare, Director Sunshine and my brother Kuldeep Khare showed immense patience and tolerance throughout the preparation of all the books written by me in many ways.

Suggestions from learned teachers and curious students for further improvements of this book will be highly appreciated and incorporated in the subsequent editions.

Suggestions can be mailed at publications@sunshineeducation.org

Dated: 01st January, 2018 Sanjay Khare

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7. **Cytology**

GENETIC MATERIAL

1. The chemical analysis of the nucleus shows that it contains nucleic acids.
2. DNA was discovered by a Swiss biochemist **Friedrich Miescher** in 1868 and he named it **'nuclein'** (CPMT 1986, 99, 2004).
3. The term **'nucleic acid'** was introduced by **Altmann** in 1889.
4. Nucleic acids are macromolecules and polymers of nucleotides.
5. The link between generatio ns is provided by nucleic acids (CPMT 2010).
6. There are two kinds of nucleic acids namely **deoxyribonucleic acid** (DNA) and **ribonucleic acid** (RNA).

DEOXYRIBONUCLEIC ACID

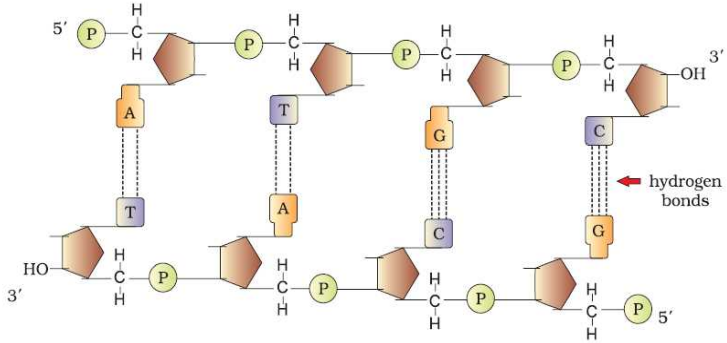
1. DNA is the major store of genetic inf ormation and the hereditary information is carried in DNA (CPMT 1977).
2. The first undoubted evidence about DNA being the genetic material came from genetic transformation experiments on Streptococcus (Diplococcus) pneumoniae (CPMT 1989).
3. **Frederick Griffith** (1928) noticed transformation of **rough** (a virulent) strain of *Streptococcus pneumoniae* into **smooth (**virulent) strain.
4. **Oswald Avery, MacLeod and McCarty** (1944) demonstrated that the transforming substance is DNA (CBSE 1993).
5. Further evidence supporting DNA as the genetic material was provided by **Alfred D. Hershey** and **Martha Chase** in 1952.
6. In their experiment, ^{32}P was used to label viral DNA and ^{35}S was used to label the protein coat of virus (AIIMS 1992).
7. The Hershey and Chase experiments showed that DNA an d not protein is the genetic material of th.e **T_2 bacteriophage**.
8. Lederberg and Tatum demonstrated the transfer of DNA as genetic material from F+ (male) to f- (female) in E coli.

CHEMISTRY AND STRUCTURE

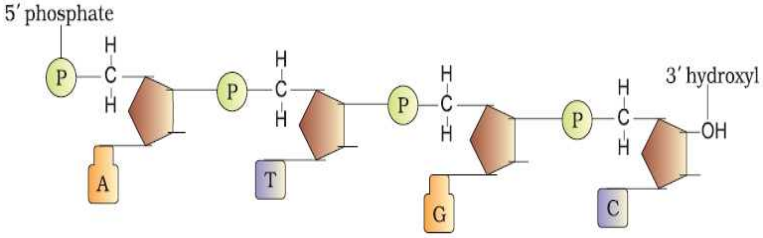
1. Chemical studies on the bases of DNA were performed by **Erwin Chargaff** in 1950.
2. According to Chargaff the percentage of **adenine** (A) is equal to the percentage of **thymine** (T) and the percentage of **guanine** (G) is equal to the percentage of **cytosine** (C).

3. The percentage of A+G equals 50% and the percentage of T+C equals 50%. These relationships are called '**Chargaff's rules**'.
4. Building blocks of the nucleic acids are **nucleotides** (CPMT 1978).
5. A nucleotide consists of nitrogenous base, pentose sugar and phosphate (CPMT 1988; AMU 1986, CBSE 2008).
6. Nitrogenous bases in DNA are pyrimidines and purines.
7. Pyrimidines have a single heterocyclic ring whereas purines have two fused rings. (CPMT 1998).
8. Pyrimidine base of DNA are represented by thymine and cytosine and purine bases by adenine and guanine (BHU 1985, CBSE 2009).
9. Between adenine and thymine there are **two** and between guanine and cytosine **three** weak non -covalent hydrogen bonds are present. Distance between H Bonds is 2.83-2.90Å. (AFMC 1997)
10. A **nucleoside** consists of nitrogenous base and pentose sugar (nucleotide minus phosphate) (BHU 2005).
11. DNA contains a 5 -carbon sugar (pent ose sugar) deoxyribose (CPMT 1997, 2008).
12. **Rosalind Franklin,** a student of **M.H.F. Wilkins** at King's College in London, Studied the structure of DNA using X-rays.
13. The X-ray diffraction pattern of DNA produced by Rosalind Franklin showed that DNA is a helix.
14. In 1953, **James Watson and Francis Crick** proposed the three -dimensional structure of DNA based on X -ray diffraction photographs of DNA fibres taken by Rosalind Franklin and M.H.F. Wilkins.
15. For discovering the structure of DNA, **Nobel Prize was awarded to Watson**, **Crick and Wilkins** in the year 1962 (CPMT 1999, 2007).
16. The Watson and Crick model shows that DNA is a **double helix** with sugar phosphate backbones on the outside and paired bases on the inside.
17. The diameter of DNA molecule is 20Å (CPMT 1999; BHU 2007).
18. The pitch (a complete turn) of DNA has a length of about 34 Å.
19. Adjacent bases are separated by 3.4 Å along the helix axis and related by a rotation of 36 degrees.
20. There are about 10 base pai rs in each turn of DNA double helix (BHU 1998).
21. The carbon atoms of the pentose sugar involved in **phosphodiester bond** formation in DNA are C_3 and C_5.
22. The two strands of DNA are **antiparallel and complementary.**

23. If one strand of the double stranded DNA has the nucleotide sequence GTA GAA, its complementary strand will be CAT CTT.
24. DNA is found in nucleus and cytoplasm (CPMT 19 98). The contribution of cytoplasmic DNA in the cell`s total DNA is about 1-5%. (DPMT 1986)
25. **Extranuclear or cytoplasmic** DNA is found in mitoch ondria and chloroplasts (BHU 1988).
26. **DNA is always double stranded** (CPMT 1980, CBSE 2010), DNA is rarely single stranded.
27. A single stranded DNA has been reported by **Robert Sinsheimer** in ϕX 174 virus.(BHU 1997)
28. **Cistron** is the functional unit of a DNA molecule.
29. RL model of DNA was given by Sashisekharan.
30. ¢R, SB, and Fd also have single stranded DNA.



Double stranded polynucleotide chain

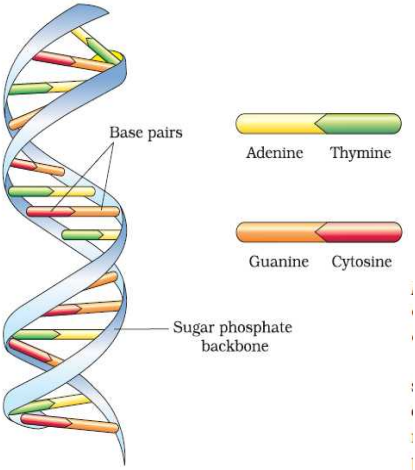


A Polynucleotide chain

FORMS OF DNA

1. Under different conditions of isolation, purification and crystal lization, several forms of DNA have been recognized. They are A -, B -, C -, D - and Z-DNA.

2. **Watson and Crick's** analysis was based on X -ray studies of the **B-DNA.**
3. A -, B -, C - and D-DNA are **right-handed** helices whereas Z-DNA is **left handed** helix with 12 base pairs per turn and assumes zigzag conformation; hence its name



DNA double helix Polynucleotide chain

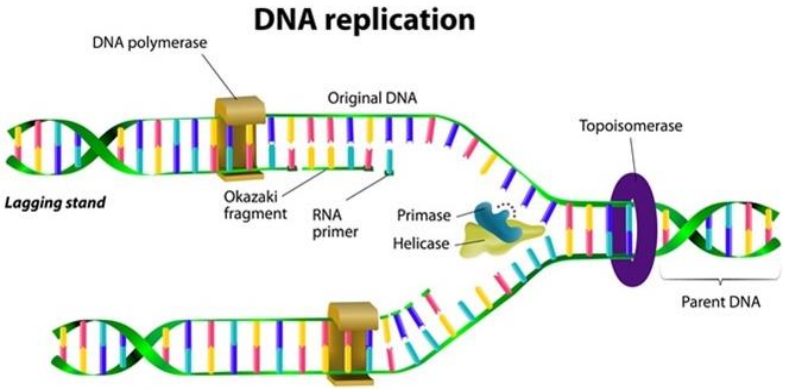
Characters of Different Forms of DNA

Helix Type	Base pairs per turn	Rotation per bp	Vertical rise per	Helical diameter
A	11	+ 32.7 Right handed	2.56 Å	23Å
B	10	+ 36.0 Right handed	3.38 Å	19Å
C	9.33	+ 38.6 Right handed	3.32 Å	19Å
D	8	+ 45.0 Right handed	3.03 Å	-
Z	12	+ 30.0 Left handed	3.71 Å	18Å

DNA REPLICATION

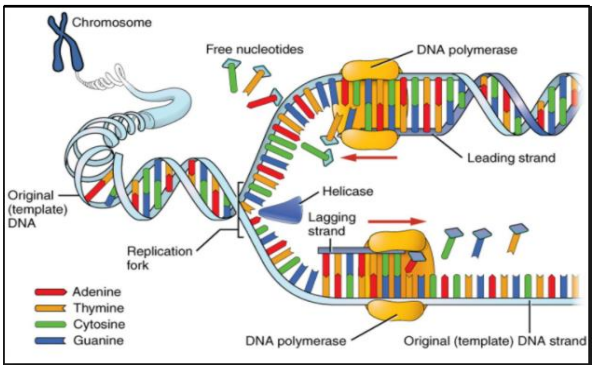
1. Duplication of DNA takes place by **replication** (CPMT 2002; AFMC 1996, CBSE 2011).
2. DNA replication takes place during **S-phase** of interphase during cell cycle.
3. DNA replication is **semi-conservative** (CPMT 1985; CBSE 1989; Orissa JEE 1995, CET, 2013). Each daughter molecule contains an old and a new strand.

4. **Matthew Meselson** and **Franklin Stahl** in 1958 provided strong evidence for semi -conservative mode of DNA replication in *Escherichia coli* (CBSE 1992).
5. Meselson and Stahl first grew *E. coli* in a medium containing ^{15}N so that only heavy DNA molecules were present in the cells.
6. Then they switched the bacteria to a medium containing ^{14}N. After one generation, only hybrid DNA molecules were in the cells.
7. After two generations, half of DNA molecules were light (^{14}N ^{14}N) and half were hybrid (^{14}N ^{15}N). These were e xactly the results to be expected if DNA replication is **semi-conservative** (MPPMT 1993).
8. Semi-conservative mode of replication of **eukaryotic chromosomes** was presented in 1957 by **Herbert Taylor, Philip Woods and Walter Hughes** (BHU 1997, 2007).
9. Taylor, Woods and Hughes (1957) experimented with root tips of the broad bean *Vicia faba.* labelled DNA with 3H-thymidine, a radioactive precursor of DNA and performing **auto radiography.**
10. Nobel Prizes to **Severo Ochoa** (Spain) and **Arthur Kornberg** (USA) were awarded in 1959 for artificial synthesis of RNA and DNA (BHU 1982, AIIMS 2007).
11. Nobel Prizes to Ochoa and Kornberg were given for enzymes associated with **synthesis of RNA** and **DNA** (DPMT 1986).
12. In 1970, **Har Govind Khorana** (together with 12 associates) reported the total synthesis of the gene for an alanine tRNA from yeast.
13. Har Govind Khorana is credited for the synthesis of gene in the laboratory.



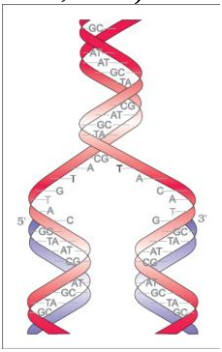
Mechanism of DNA Replication

1. **DNA replication requires the following steps:**
   (i) **Unwinding:** The old strands that make up the parent DNA molecule are unwound and "unzipped" (i.e., the weak hydrogen bonds between the paired bases are broken). This step is carried by a special enzyme called **helicase**.
   (ii) **Complementary base pairing:** New complementary nucleotides, always present in th e nucleus, are positioned by the process of complementary base pairing.
   (iii) **Joining:** The complementary nucleotides join to form new strands. Each daughter DNA molecule contains an old strand and a new strand.



2. Unwinding a DNA double helix requires **helicases**, single-stranded DNA binding proteins and **topoisomerases**.
3. DNA helicases are proteins that bind to single -stranded DNA and travel along it, driven by energy derived from the hydrolysis of ATP.
4. The helicase that functions at the replica tion fork forms a tight complex with DNA **primase,** producing a structure called **primosome.**
5. **The leading strand** serves as a template for continuous synthesis; **in the lagging strand** discontinuous synthesis occurs resulting in **Okazaki fragments** (discovered by **Reiji Okazaki** in 1968).
6. Single-stranded DNA binding protein s maintain the unwound DNA in a single stranded state until **DNA polymerase III** can synthesize the next Okazaki fragment.
7. **DNA polymerase I** removes the **RNA primers** and fills the resulting gaps and **DNA ligase** joins the fragments.

8. **Topoisomerase** introduces transie nt breaks in one of the two DNA strands ahead of the replication fork, thereby allowing the helix to unwind locally without having to rotate the entire DNA molecule.
9. **DNA polymera se III** catalyses DNA sy nthesis in a 5' to 3' direction only.
10. DNA polymerase helps in joining monomers of DNA (BHU 1997).
11. Protein helping in opening of DNA doubl e helix in front of replication fork is **DNA gyrase** (CBSE 1994, BHU 1997, AIIMS 2016).
12. **Helicase** breaks the hydrogen bo nds between complementary pairs during DNA replication (MPPMT 1993, DELHI PMT 2008).
13. Ligase joints short segments of DNA together (BHU 1985; MPPMT 1993, 2011).



14. While proposing the double helical structure for DNA, Watson and Crick had immediately proposed a scheme for replication of DNA. The scheme suggested that the two strands would separate and act as template for the synthesis of new complementary strands.
After the completion of replication, each DN A molecule would have one parental and one newly synthesised strand. This scheme was termed as **semiconservative** DNA replication

Watson - Crick Model for semiconservative DNA replication.

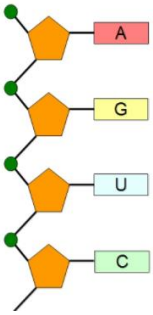
ENZYMES AFFECTING DNA

Enzyme	Action	Function in the cell
Restriction Endonuclease	Cuts DNA at specific base sequences	Destroy foreign DNA
DNA ligase	Links DNA molecules	Completes DNA replication process
DNA polymerase I	Attaches nucleotides to the growing DNA molecule, removes RNA primers	Fills gaps in DNA, primarily for DNA repair and removes primers
DNA polymerase III	Attaches nucleotides to the growing DNA molecule, proofreads each inserted nucleotide	Replicates DNA

DNA gyrase (Topoisomerase II)	Increases the twisting pattern of DNA, promoting super coiling	Maintains compact structure of DNA
DNA helicase	Binds to DNA near replicating fork	Promotes DNA strand separation
Primase	Makes short RNA chains using a DNA template	Needed to make primer to be used by DNA polymerase
Topoisomerase I	Relaxes supercoiled DNA	Helps to maintain the proper level of supercoiling
DNase	Degrades DNA to nucleotides	Destroys DNA
DNA methylase	Places methyl groups on DNA bases, thus inhibiting restrictionendonuclease action	Modifies cellular DNA, so that it is not affected its own restriction endonuclease

RIBONUCLEIC ACID

Structure of RNA:



1. RNA is widely distributed in the cell (CPMT 1997, 2008, 2014) and is **synthesized by DNA in nucleus**.
2. RNA is composed of **ribo-nucleotides.**
3. The pentose sugar in RNA is **ribose** (CPMT 1991; JIPMER 1998). Purine bases of RNA are represented by **adenine** and **guanine**, the pyrimidine bases are **uracil** and **cytosine**.
4. The thymine in DNA is replaced by uracil in RNA (CPMT 2006, 15).
5. **Fraenkel Conrat** established that RNA is the genetic material in some viruses.
6. RNA is single stranded, but **double stranded RNA** is present in **rheovirus** (AIIMS 2004) and **wound tumour virus.**
7. RNA was the first amino acid to appear on earth in origin of life.
8. In **retroviruses,** the genetic material is RNA.
9. **Rice dwarf virus**es have **dsRNA**.
10. There are three major classes of RNA, each with specific functions in protein synthesis:

MESSENGER RNA (mRNA)

1. Messenger RNA is produced by DNA; the process is called **transcription**
2. The m-RNA is formed from DNA in nucleus (NCERT. 1983) and play an important role in the synthesis of proteins (AFMC 1985).
3. Messenger RNA encodes the amino acid sequence of a protein in their nucleotide base sequence.
4. A triplet of nitrogenous bases spec ifying an amino acid in m -RNA is called **codon**.
5. m-RNA is **least abundant** but synthesised in maximum amount.
6. m-RNA was reported by **Jacob and Monad.**
7. Synthesis of mRNA is maximum during **cleavage**
8. m-RNA is synthesised on **Sensestrand (Masterstrand)** of DNA only.

TRANSFER RNA (t-RNA)

1. t-RNA is also k nown as **soluble RNA (sRNA);** it is soluble in 1 molar solution of sodium chloride.
2. t-RNA identifies amino acids in the cytop lasm and transports them to the ribosome.
3. **Holley (1965)** reported the sequence of an **alanine t-RNA** from yeast & proposed **two-dimensional structure of tRNA (clover leaf model).**
4. **Anticodon** is a **three-base sequence** in a tRNA molecule that forms complementary base pairs with a codon of mRNA.
5. All transfer RNA posses the sequence CCA at the ir 3' ends; the amino acid is attached to the terminal A residue.
6. **Loop II of tRNA** bears anticodon to recognise codon on mRNA durin protein synthesis.
7. t-RNA was reported by **Hoagland**.
8. It has some unusual bases like **Ψ(Pseudouridine), DHU (Dihydroxyuridine)** and a **high GC ratio.**
9. t-RNA is also known as **Supernant tRNA.**
10. **t-RNA or Adoptive** RNA is smallest RNA and constitute about **10-15%** of total cellular RNA.
11. Synthesis of tRNA is maximum at the end of cleavage.

RIBOSOMAL RNA (r-RNA)

1. Ribosomal RNA is found in ribosomes of ce lls and is also called **insoluble RNA**.

2. The main function of rRNA is to attrac t and provide large surface for spreading of m -RNA over ribosomes during translocation process of protein synthesis.
3. r-RNA was reported by **Kuntz.**
4. **Synthesis of mRNA is maximum during gastrulation.**
5. r-RNA is synthsised in Nucleolus.
6. r-RNA is the largest RNA and constitutes **about 80% of total cellular RNA.**

ADDITIONAL POINTS

1. Sex cells (gametes) generally **possess half** the amount of DNA as the body cells (CPMT 1986, 2008).
2. **Circular DNA** is present in prokaryotes and mitochondria and chloroplasts of eukaryotic cells (MPPMT 1998, CBSE 2012).
3. A DNA strand is directly involved in the synthesis of all like another DNA molecule, tRNA, mRNA and rRNA molecules (AIIMS 1998).
4. The best arrangement of the compounds in order of increasing molecular weight is tRNA, mRNA, rRNA and DNA (CBSE 2013).

GENETIC CODE

1. The **genetic code** is the sequence of **nucleotide bases in m -RNA** which encloses the information for the synthesis of protein molecules.
2. The relationship between the sequence of amino acids in polypeptide with base sequence of DNA or m-RNA is genetic code.
3. Genetic code **determines the sequence** of amino acids in a protein.
4. **The first work on genetic code** was carried out by **George Gamow.** He proposed the **diamond code in 1954.**
5. The genetic studies by **Crick** (1961) on a virus established that genetic code is a **triplet**. **Codon** is represented by **three consecutive nucleotides.**
6. A triplet would code for a given amino acid as l ong as three bases are present in a particular sequence (AIIMS 1985, 2012).
7. **Marshall Nirenberg (1961)** attempted to find out the spelling of a codon; he showed that uracil (UUU) codes for **phenylalanine.**
8. Later in a cell -free system, **Marshall Nirenberg** and **Philip Leder** (1964) were able to show that **GUU codes for** the amino acid **valine**.
9. The spellings of further codons were discovered by **R. Holley, H. Khorana** and **M. Nirenberg.** They have been awarded the **Nobel Prize in 1968** for researches in **genetic code** (CPMT 1998, 2009).

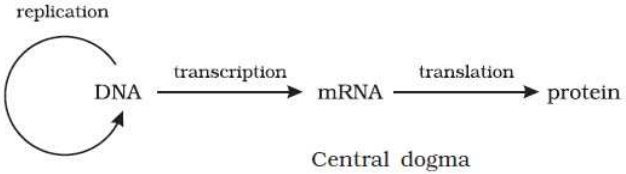
CODING DICTIONARY

1. A table of all the code words or cod ons that specify amino acids is termed as **coding dictionary.**
2. Coding dictionary contains 4^3 or 4 x 4 x 4 = **64 possible triplet codons**.
3. Usually the codons are read only in the 5' to 3' direction.
4. Of the 64 codons, 61 codes for amino acids and the termed as **sense codons.**
5. Three triplets **UAA (ochre), UAG (amber) and UGA (opal)** do not code for an amino acid and the called **non-sense codons** (AIIMS 1992, CBSE 1977).
6. The function of non -sense codons is to terminate the message for gene controlled protein synthesis (AIIMS 1985).
7. Non-sense codons are also called as **chain termination codons.**
8. AUG which code for **methionine (Met)** is used as **start signal** in protein synthesis (AIIMS 1986). It is most common chain **initiation codon** (CBSE 1994, 99).
9. In prokaryotes, the codons **GUG and UUG** are also employed in rare instances as **start codons**.
10. The genetic code is **degenerate**. This means that most amino acids have more than one codon; **leucine, serine and arginine** have **six different codons.**
11. Genetic code is **nonoverlapping** code. No signal base takes part in the formation of more than one codon.
12. The genetic code is **commaless** as there are no intermediary (spacers) between codons.
13. The genetic code has been found to be **universal,** *i.e.* the same for all organism.

PROTEIN SYNTHESIS

1. Proteins are **polypeptide chains** formed by polymerization of **amino acids.**
2. The components linked in long chain to form a protein are amino acids.
3. Bonds established between adjacent amino acids during the formation of proteins are **covalent peptide bonds.**
4. The **carboxyl group** of one amino acids is linked to the **amino group** of a second amino acid with the elimination of one molecule of water.
5. Only **20 amino acids** are biologically important in the participation of protein synthesis.

CENTRAL DOGMA



Central dogma

1. Central dogma was originally formulated by **Crick** stating that th e biological information flows in the pattern: DNA→RNA→Protein.
2. Flow of information from DNA to mR NA and then to protein is known as **central dogma** (AFMC 1986).
3. Central dogma of modern biology is: **DNA→RNA→Proteins** (BHU 1985).
4. RNA plays an important role in the synthesis of proteins (AFMC 1985).
5. The exception of central dogma is the **reverse transcription** reported by **H.M. Temin** and **D. Baltimore** (1970) independently in the RNA - containing **Rous Sarcoma virus (RSV).**
6. Temin and Baltimore were awarded with Nobel Prize in 1975 for the discovery of the enzyme **'reverse transcriptase'** or **RNA-dependent DNA polymerase** (CBSE 1994).
7. Central Dogma Reverse is also called **Teminism.**
8. Commoner (1968) suggested a circular flow of information DNA → RNA → Protein → RNA → DNA.

MECHANISM OF PROTEIN SYNTHESIS

1. The protein synthesis takes place in following steps:
   (i) Transcription, (ii) Activation of amino acids, (iii) Attachment of activated amino acids with tRNA and (iv) Translation.

Transcription

1. The mode by which DNA passes its genetic information to mRNA is known as **transcription** (AIIMS 1981; BHU 1982; MPPMT 19 84, 94; DPMT 1986).
2. Transcription, which takes place in the nuc leus of eukaryotic cell, is the first step required for protein synthesis.
3. The molecules tRNA and rRNA are also transcribed from DNA templates.
4. Wherever A, T, G or C is present in th e DNA template, U, A, C or G is incorporated into the mRNA molecule.

Process of Transcription in Bacteria

5. A segment of the DNA helix unwinds and unzips and complementary RNA nucleotides pair with DNA nucleot ides of the strand that is to transcribed.
6. Transcription begins when **RNA polymerage** attaches to a region of DNA called a **promoter.**
7. A promoter defines the start of a gene, the direction of transcription and the strand to be copied.
8. Finally RNA poly merase comes to a **terminator** sequence at the end of the gene being transcribed.
9. The terminator causes RNA polymera se to stop transcription and to release the mRNA molecule, now called an **RNA transcript.**
10. Transcription copy of mRNA from DNA- AGT CCT TGG AAT will be UCA GGA ACC UUA (DPMT 1983).
11. Base sequence of DNA strand is CAT TAG CAT CAT GAC. The base sequence on RNA strand will be GUA AUC GUA GUA CUG (CPMT 1984).
12. The process of inhibition of RNA synthesis on a DNA template is called **repression.**
13. RNA synthesis (transcription) is inhibited by **Actinomycin D** (BHU 1986).

Activation of Amino Acids

1. Amino acids in the cytoplasm are **inactive**. They cannot take part directly in protein synthesis.
2. The activation of amino acids is done by ATP resulting in the formation of **aminoacy1 adenylate** and **pyrophosphate**.

Attachment of Activated Amino Acid with tRNA

1. Activated amino acids (aminoacyl adenyl ate) get attached with their specific tRNA molecules with the help of the enzyme aminoacyl transfer RNA synthesis. The product formed is known as **Aminoacyl transfer RNA complex.**

TRANSLATION

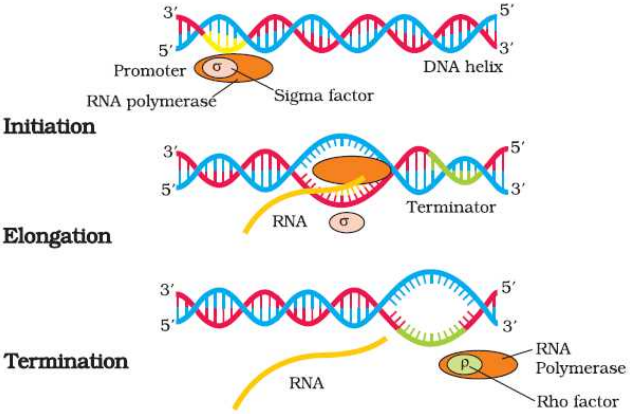
1. The synthesis of a chain of polypeptide by mRNA is know as **translation.**
2. Translation is the process when protein sy nthesis takes place at the site of **ribosomes** (CPMT 1979).

3. Protein synthesis is associated with, or ce ntres of protein synthesis are ribosomes (CPMT 1974, AFMC 1982,84; BHU 1985).
4. Translation involves three steps:**initiation, elongation and termination.**

A. Initiation of translation

(i) A small ribosomal subunit attaches to the mRNA near initiation codon (AUG).

(ii) The first or initiator tRNA (with f -met) pairs with this codon, a large ribosomal subunit joins to the small unit and translation begins.



B. Chain elongation

(i) Each ribosome cont ains two sites, the **P** (for polypeptide) site and the **A** (for amino acid) site.

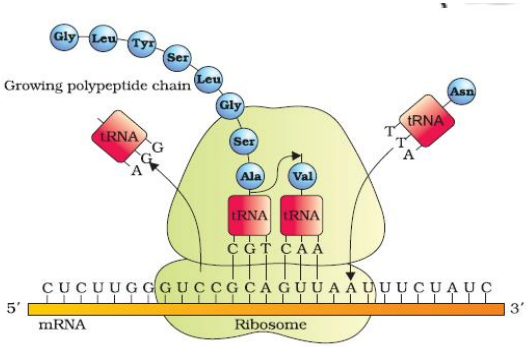
(ii) A tRNA with attached polypeptide is at the P site and a tRNA amino acid complex just arrives at the A site.

(iii) The polypeptide is transferred and attached by a peptide bond to the newly arrived amino acid.

(iv) An enzymes **peptidyl transferase** is needed to bring about the transfer. Now the tRNA molecule at the P site leaves.

(v) **Translocation** occurs when the mRNA along with peptide -bearing **tRNA moves from the A site to the empty P site.**

(vi) Since the ribosome has moved forward three nucleotides, there is a new codon now located at the empty A site.



Translation

C. Chain termination

(i) **Termination** of polypeptide synthesis occurs at a **stop codon (UAA, UGA and UAG**), which does not code for an amino acid.

(ii) The arrival of stop codon in A site results in the completion of protein synthesis and enzymatic cleavage of last tRNA from polypeptide.

5. Poly ribosomes are aggregation of several ribosomes held together by a string of mRNA (DPMT 1984; BHU 1985, 88).

6. Protein synthesis is **inhibited** by antibiotics such as **puromycin, streptomycin, chloramphenicol** and **cycloheximide**.

GENES AND CHROMOSOMES

CHROMOSOME

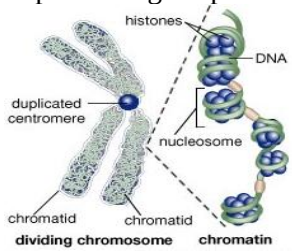
1. **The chromosome theory** of inheritance was postulated by **Sutton** and **Boveri** independently in **1902.**
2. The chr omosome theory states that genes are located on the chromosome.
3. Chromosomes can be counted best at Metaphase.
4. Chromosomes have **affinity** for **basic dyes**.

CHROMOSOMES IN EUKARYOTIC ORGANISMS

1. Chromosomes were discovered by **Hofmeister** in the pollen mother cells of *Tradescantia* in 1948 (CPMT 1984, 92).
2. **E. Strasburger** (1875) observed chromosomes during cell division.
3. **W. Waldeyer** (1888) introduced the term **'chromosome'** (AFMC 1999).

Chromosome Structure

1. A chromosome consists of chro monema, chromomere, c entromere, secondary constriction, telomere, satellite, etc.
2. The filaments constituting a chromosome are known as **chromonemata.**
3. **Chromomeres** are linearly arranged bead-like and compact segments or granules found on leptotene and zygotene chromosomes.
4. **Centromere** is also called as **kinetochore or primary constriction**.
5. Centromere is a specialized chromomere to which the spindle fibres are attached during cell division.
6. Centromere is concerned with anaphase movement of chromosomes to poles.
7. **Levan, Fredga and Sandberg** (1964) have proposed a nomenclature for centromeric position on chromosome.
8. Four morphologic types of chromosomes according to the position of the centromere are: **metacentric, submetacentric, acrocentric and telocentric.**
9. **Metacentric** chromosomes have equal arms (BHU 1996) and are **'V'** shaped during anaphase.

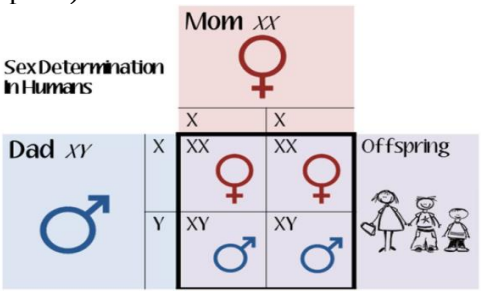


10. **Submetacentric** chromosomes have unequal arms and are **'L'** shaped.
11. **Acrocentric** chromosomes have subterminal centromere with a very small and very large arm, j shaped.
12. **Telocentric** chromosomes have strictly centromere and with only one arm.
13. **Telomere** is the term applied to each **end of a chromosome** (JIPMER 1986; AMU 1987).
14. **Secondary constrictions** are additional constrictions on the chromosome. They are constant in their position.
15. **SAT-chromosome** (Heitz 1931) is a chromosome with secondary constriction.
16. The chromosome with secondary constriction associated with the formation of the nucleolus is called **'nucleolar SAT chromosome'**.
17. Chromosome size is normally measured at mitotic metaphase and can be as small as 0.25 μm in fungi and as long as 30 μm in some plants like *Trillium*.
18. **Isochromosomes** are formed from Telocentric chromosomes.

19. In human 13, 14, 15, 21 and 22 pair of chromosomes h ave nucleolus organiser region.

Sex Chromosomes and Autosomes

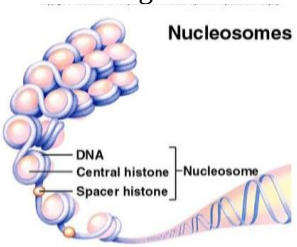
1. **Autosomes** are the chromosomes other than sex chromosomes (BHU 1981).
2. **Allosomes** (**heterochromosomes or heterosomes)** are related with sex determination (BHU 1986; MPPMT 1986; CPMT 1995).
3. **Genome** is complete set of ge nes present in a haploid set of chromosomes contributed by a parent(BHU 1991; KMC ManipaI1995).
4. In chromosomes, the material controlling heredity is DNA (CPMT 1976).
5. According to chromosome theory of inheritance, genes are arranged in the chromosomes in a linear fashion.
6. Chromosome contains almost equal amounts of **DNA (45%)** and **histones** (**45%)** (BHU 1998), a variable amount of nonhistone chromosomal proteins (also called **hertone**), and a small amount of chromosomal RNA.
7. A very small amount of lipids, polysaccharides and metal ions like calcium are reported from chromosome.
8. Two types of proteins in chromosomes of eukaryotes are: **basic protein histone, acidic protein- hertone.**
9. Histone protein regulates nucleic acid activity.
10. Male honey bee has 16 chromosomes but worker and queen honey bee have 2n = 32 (diploid).



Nucleosomes

1. The chromosome (chromatin) is formed of a series of repeating units called **nucleosomes.**
2. The term '**nucleosome**' was given by **Oudet** (1975).

3. Each nucleosome is formed of a chain of DNA having **140 base pairs** making **1 ¾ turns** and **twist around a histone octamer.**
4. The core of nucleosome consists of 4 his tones namely **H_2A, H_2B, H_3** and **H_4** (BHU 1984). Another histone namely **H_1** is associated with linker region.



The C Value

1. The total amount of DNA in the haploid genome is characteristic of each living species.
2. The amount of DNA in a prokaryotic chromosome or the haploid (N) amount of DNA in a eukaryotic cell is called the **C value**.

3. There is not a direct relationship between the C value and the structural or organizational complexity of the organism.

GIANT CHROMOSOMES

1. **Polytene chromosomes** were discovered by **Balbiani** (1881) in the salivary gland cells of *Chironomus* larva (AIIMS 1983; CBSE 1995,99).
2. **Kolle**r gave the term **Polytene.**
3. Polytene chromosomes are also found in the larva of other **dipteran** insects like fruitfly *(Drosophila)* and mosquito.
4. All polytene chr omosomes of *Drosophila* appear to be attached together by a **chromocentre** (CBSE 1995).
5. **Puffs** in the salivary gland chromosome of *Drosophila* represent the **site of RNA synthesis.**
6. **Puffs** are made up of **Balbiani rings.**
7. **Lampbrush chromosomes** are **much larger** than the polytene chromosomes.
8. **Polytene chromosomes** are **somatic chromosomes** whereas **lampbrush chromosomes** are **germinal chromosomes.**
9. **Lampbrush chromosomes** are found mainly in Oocytes of amphibians and **sperm and oocytes** of **Drosophila**

GENE

1. **Gene** is the hereditary unit and is made of **polynucleotides** (DNA).
2. Genes are composed of DNA/RNA in prokaryotes (viruses) and DNA only in, eukaryotes (MPPMT 1996).

3. Gene is a piece of DNA having about 500 – 2000 base pairs.
4. Biochemical nature of a gene was established by **Avery, MacLeod** and **McCarty** (1944) based on transformation experiments in bacteria *Diplococcus pneumoniae.*
5. The ultrastructure of gene was given by **Benzer** (1957). According to him, genes include **cistron**, recon and **muton** (CPMT 1987).
6. **Cistron** is the **functional unit** of DNA molecule; it codes *for* particular gene product.
7. **Recon** is the **cross over unit** or it is a segment of DNA, which participates in crossing over.
8. **Muton** is the **basic unit of gene mutation** (AFMC 1993), represented by a mutational site (a nucleotide pair).
9. **H.G. Khorana** (together with twelve associates) reported the total synthesis of the gene for an **alanine tRNA from yeast** (1970).
10. Gene pool is the sum total of genes in a population.
11. **Plasma genes** are the genes present in the cytoplasm.
12. **Oncogenes** are concerned with the production of cancer (MPPMT 1993).
13. Sum total of genes at any time in a unit of evolution is termed as Gene Pool. (CPMT 1993)

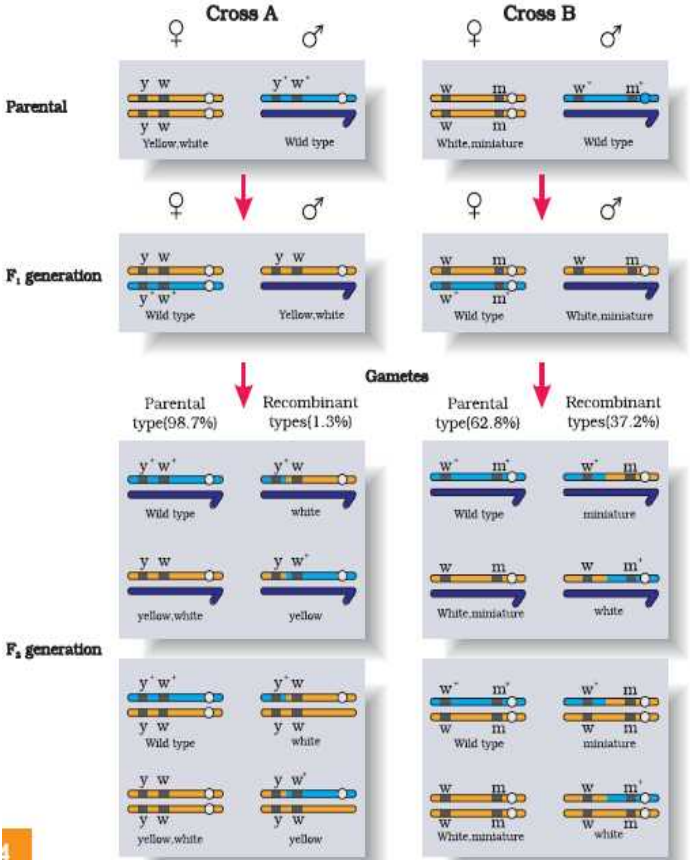
TRANSPOSON

1. **Barbara McClintock** (1902-92) discovered" controlling elements" that could move from one location to another on the chromosome.
2. Today, McClintock's controlling elements are called movable genetic elements, **transposons, or "jumping genes".**
3. Barbara McClintock (1983) got the **Nobal Prize** for the discovery of 'jumping genes' in maize (AIIMS 1984).
4. Two transposable controlling elements discovered by McClintock are dissociation (Ds.) and activator (Ac).
5. The term 'transposon' was introduced by **R. W. Hedges** and **A.E. Jacob** in 1974(CBSE 1997).
6. Transposon is a DNA segment or genetic element moving from one chromosome to another (CPMT 1992).
7. In bacterial chromosomes (plasmid), transposons carry genes for resistance to antibiotic **ampicillin**.

LINKAGE AND CROSSING OVER

LINKAGE

1. The tendency of gene s to remain together during the process of inheritance is called linkage.



2. Bateson, Saunders and Punnett (1905) working with *Lathyrus odoratus* (sweet pea) were the first to discover linkage, an exception to the law of independent assortment (BHU 1991).
   Linkage: Results of two dihybrid crosses conducted by Morgan. Cross A shows crossing between gene y and w; Cross B shows crossing between genes w and m.Here dominant wild type alleles are represented with (+) sign in superscript.
   Note: The strength of linkage between y and w is higher than w and m.

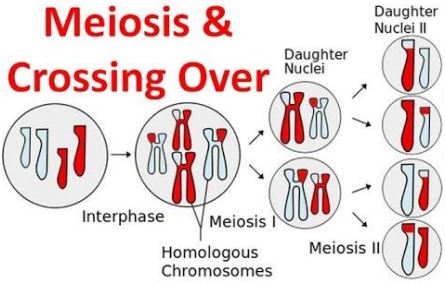
3. The first attempt to show linkage in plants was done in *Lathyrus odoratus* (BHU 1984, CBSE 1997).

4. **An exception to Mendel's law is linkage** (CPMT 1980 CBSE 1999).
5. If two alleles A and B come from the same parent (AAB B x aabb) then they will enter gametes and transmit together; this is called **coupling**.
6. When the same alleles (A and B) come from different parents (AAbb x aaBB) then they will enter into different gametes; this is known as **repulsion**.
7. **Morgan** (1910) discover ed that 'coupling and repulsion' are two aspects of the same phenomenon 'linkage'.

LINKAGE GROUPS

1. All the linked genes of a chromosome form a linkage group.
2. An individual has innumerable characters and fewer chromosomes; hence each chromosome should carry a number of genes.
3. The genes situated on the same chromosome should be inherited together and hence cannot show independent assortment.
4. The genes located on the same chromosome remain linked together and should form one linkage group.
5. The number of linkage groups corresponds to the haploid number of chromosomes (DPMT 1983).
6. Bacterial and blue-green algal cells contain only one linkage group.

CROSSING OVER



1. The term 'crossing over' was introduced by Morgan and Cattell (1912) for the process of separation of linked genes. "
2. The transference of genes from one chromosome to another during synapsis is termed as 'crossing over' (CPMT 1991,94).
3. Crossing over takes place at 4-strand stage (DPMT 1983;MPPMT 1985) between pachytene and diplotene.
4. Continuous variation is attributed to crossing over (CPMT 1983).
5. Crossing over results in the recombination of linked genes(CPMT1991).

6. The probability of crossing over between two linked genes is directly proportional to the distance between them.
7. There is a greater probability for a cross over to occur between two genes farther apart than two genes nearer each other.
8. When two genes are situated very close to each other in a chromosome, hardly any crossing over is detected (AIIMS 1980).

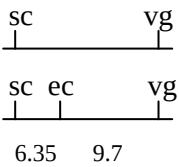
MAPPING OF CHROMOSOMES

1. The distance between two genes in a chromosome is measured in cross over units which represent the percentage of crossing over between them.
2. Map unit is a number t hat corresponds to a recombination (crossing over) frequency of one per cent.
3. If the percentage of recombination is 10 between two genes, the distance between the genes is 10 map units (DPMT 1986).
4. Recombination frequency is used to constructed linkage map.
5. Unit Morgan refers to linkage map.
6. The evidence that crossing over occurs in 4 -strand stage comes from 2:2:2:2 arrangements of *Neurospora* ascospores.

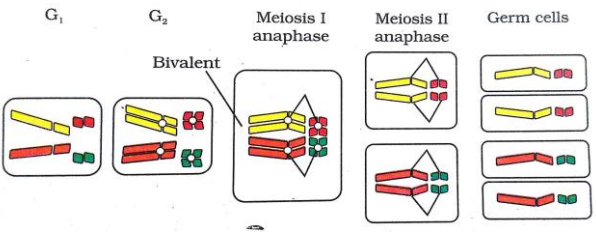
HOW TO CHROSOME MAP

(1) Most distant?? sc & vg
total map distance = 13.6)

(2) The intermediate gene is *ec*. It is closer to sc. So we locate it like this.



The genetic map is incomplete !



Meiosis and germ cell formation in a cell with four chromosomes. Can you see how chromosomes segregate when germ cells.

MUTATION

1. Mutation may be defined as a sudden inheritable genetic change, altering the genetic message of a cell.

2. Mutations are mainly responsible for variation in organisms (CPMT 1977).
3. Mutations a re called the fountainhead of evolution because they add new variations.
4. Sudden hereditary change in an organism which breeds true during reproduction is termed mutation (MPPMT 1988).
5. Mutations were first seen in the 18th century by Seth Wright, a New England farmer in a male Ancon sheep.
6. The term 'mutation' was first used by the Dutch Botanist Hugo de vries (1901) in connection with the studies on *Oenothera lamarckiana* (Evening primrose) (BHU 1989; CPMT 1992,97).
7. Darwin called these 'sports' and Bateson c alled these 'discontinuous or Saltatory variations'.
8. Mutation theory of evolution was proposed by de Vries (MPPMT 1987).
9. Mutations are rarely useful (CPMT 1980; JIPMER 1986).
10. Neutral or even harmful mutations are not lost from the gene pool because they a re recessive and survive in heterozygous individuals (AIIMS 1981).

Kinds of Mutations

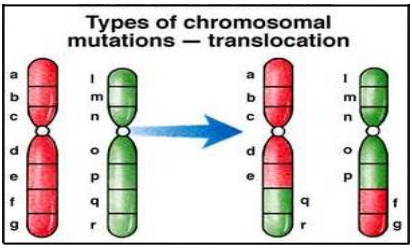
1. Depending on the kind of cell in which mutation occurs, mutations may be somatic or germinal.
2. Malignant or cancerous growth is a kind of mutation, which is local ized and not transmitted to offspring. This can be called somatic mutation.
3. Germinal mutation occurs in germ cells mostly during gametogenesis. To be evolutionary successful, a mutation must occur in germplasm DNA (CPMT 1982).
4. The mutation which occurs automatically without any apparent external reason is called spontaneous mutation.
5. Seed coat colour in pea suddenly changes from gray to white. This is an example of spontaneous mutation (AFMC 1986).
6. According to their direction, mutation may be forward or backward (reverse).
7. According to phenotypic expression, mutation may be dominant or

recessive. Majority of mutations are recessive (CPMT 1997).

8. A pleiotropic mutation influences many characters (CBSE 1998).
9. **Amatto** (19.50) classified mutations into three types:
   (i) Chromosomal mutation (chromosomal aberration)
   (ii) Genomatic mutation (change in chromosome number)
   (iii) Gene mutation (point mutation)
10. Haploids are better for the study of mutations (both recessive and dominant) be cause they possess only one allele for a gene (CPMT 1982).

CHROMOSOMAL MUTATIONS

1. The change in chromosome morphology or structure is called chromosomal aberration (CPMT 1998). Four types of chromosomal aberrations are:
   1. **Deletion or deficiency:** The loss of some genes due to the breaking of a fragment.
   2. **Duplication:** The inclusion of extra parts of the chromosome; some genes may represent twice.
   3. **Inversion:** A piece of chromosome is removed and rejoined in reverse orientation.
   4. **Translocation:** Mutual exchange of the chromosome segments between nonhomologous chromosomes.



GENOMATIC MUTAIONS

1. Numerical changes in chromosomes or variations in the chromosome number (heteroploidy) can be mainly two types:
   (i) Aneuploidy
   (ii) Euploidy

Aneuploidy

1. Aneuploidy is the term applied for the chromosomal mutations involving the loss (hypoploidy) or the addition (hyperploidy) of one or more

chromosomes.

2. When the chromosome number is different from a multiple of basic chromosome number, it is referred to as aneuploidy (AMU 1986).
   1) **Monosomics (2n - 1):** If the haploid number of the chromosome in a plant is 12 then the number of chromosomes in monosomic is 23 (BHU 1983).
      A flower was fertilized by pollen grains having the constitution n-1. If the ovules are normal, monosomic seeds will be produced.
   2) **Nullisomics (2n - 2):** Nullisomy is the term used for the condition when an organism has a complete set of chromosomes except one homologous pair.
   3) **Trisomics (2n+1):** A trisomic individual has one extra chromosomes (BHU 1985; CSSE 1998).
3. Trisomic individual has a chromosome number of 2n + 1 (CPMT 1985). Blakeslee and Belling (1924) reported 12 possible trisomies in Jimson weed, *Datura stramonium.*
   Datura is a classical example of a trisomic (BOO 1984).
   4) Tetrasomic (2n + 2): A tetrasomic individual has a particular chromosome represented in four doses.
4. Aneuploidy arises due to non-disjunction of chromosomes in a cell.
5. Organisms having mor e than 200 chromosomes are Amoeba, Ophioglossum and Geomatrid moth.

Euploidy

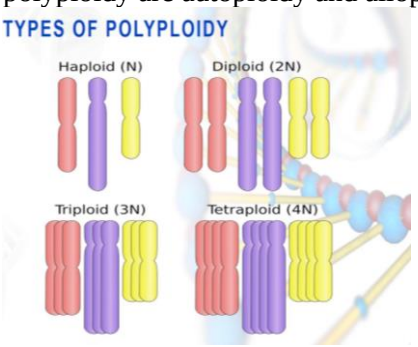
1. When chromosome sets are present in multiples of n, the condition is called as euploidy (MPPMT 1986).
2. Euploidy is the term applied for those mutations which involve addition of complete set of chromosome.
   1. **Monoploid (Haploid) :** With onl y one set of chromosomes (n) or with a single genome. Genome refers to haploid set of chromosomes or total number of genes present on a haploid set of chromosomes.
3. Monoploid phase is common in sexually reproducing algae, fungi and in all bryophytes.
4. Monoploidy is rare in animals (on e exception, male honeybee) but represented by gametes.
5. Bacteria are haploid.

2. Diploid (2n)

1. The common chromosome number in the somatic cells of plants and animals.

3. Polyploids

1. Euploids with three or more complete sets of chromosomes are called polyploids.
2. Polyploidy is rather common in the plant kingdom but rare in animals.
3. Polyploidy is common in Ferns.
4. The term 'polyploid' was introduced by Strasburger in 1910.
5. The tapetal nuclei in anthers of many angiosperm flowers are mostly polyploid.
6. An example oftriploid tissue is lily endosperm (CPMT 1983) and endosperm of wheat (CPMT 1978,96).
7. On the basis of the source of chromosomes, two main kinds of polyploidy are autoploidy and allopolypldidy.



(i) Autopolypoloids:

A polyploid species with genomes derived from the same original species is an autopolyploid, e.g., AAAA.

8. A utopolyploids are obtained by crossing a tetraploid with diploid.
9. Triploid watermelons are nearly seedless, created by crossing the normal diploid with tetraploid.
10. Helianthus tuberosus (AAAABB) is an example of autoallopolyploidy.

(ii) Allopolyploids:

A polyploid species with genomes derived from two different species is an allopolyploid, e.g., AABB..

11. A classical example of allopolyploidy is *Raphanobrassica.*
12. The Russian cytologist Karpechenko (1928) synthesized a new genus *Raphanobrassica* from the crosses between the radish (Raphanus sativus, 2n = 18) and the cabbage (Brassica oleracea, 2n = 18).
13. By doubling the F, sterile hybr id (2n = 18), allotetraploid or amphi-diploid *Raphanobrassica* (2n = 36) was obtained.
14. The first man-made cereal Triticale is an artificial allopolyploid. It has been developed from a cross between wheat(Triticum) and rye (Secale) (CBSE 1989; BOO 1995; AIIMS 1995).

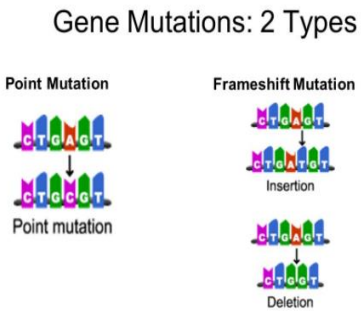
(iii) Induction of Polyploidy:

15. In natural population, polyploidy may arise as the result of interference with cytokinesis once chromosomes replication has occurred.

16. Application of the alkaloid colchicine derived from the autumn crocus *(Colchicum autumnale)* induces polyploid (CBSE 1995).
17. The chemical usually applied to induce polyploidy in plants is colchicine (BHU 1987, 92).
18. Colchicine interferes in organization of spindle fibres (BHU 1983, 86).

GENE MUTATIONS

1. A stable change in a gene is termed point mutation (BHU 1999).
2. A sudden change in the action of a gene is mutation (AFMC 1979).
3. Mutagens are agents inducing mutations (DPMT 1996).
4. Gene mutation occurs in DNA (CPMT 1976; BOO 1985,2001).
5. Gene mutation is caused by a change in the sequence of nitrogenous bases in DNA (CPMT 1978).
6. The first scientific study of true mutation was done by T.H. Morgan.
7. The first mutation that appeared in the laboratory of Morgan in 1909 was that of white-eye *Drosophila.*
8. The mutation which appears in response to an external factor or substance is called induced mutation.
9. Mutagenic effect of X -rays was discovered by H. J. Muller (1927) in *Drosophila* (CPMT 1985; BHU 1984; MPPMT 1996).
10. H.J. Muller had received Nobel Prize in 1946 for production of mutations by X-ray irradiations (CBSE 1996).
11. L.J. Stadler (1928) reported mutations by X-rays in barley and maize.
12. X-rays can produce mutations in plants (CPMT 1986; BHU 1992).
13. UV rays cause non -ionizing radiation and it has many specific biological effects (AMU 1989).
14. UV rays prove lethal due to inactivation of proteins, pigments and nucleic acids (AMU 1989).
15. The mutational changes have been more frequent at the time when living cells first evolved; less ozone was present thus permitting more radiation to penetrate the earth's atmosphere (AIIMS 1982, BHU 2001).
16. Among different crops, mutational s tudies have been more intensively done on wheat (MPPMT 1989).
17. Nitrous acid induces mutations by oxidative deamination.
18. Coal tar is a potential carcinogen (BHU 1089).



19. Nitrogen mustard is a mutagenic agent which can produce mutations in both replicating and nonreplicating DNA.
20. 5-bromouracil is a base analog (MPPMT 1990, CBSE 2000).
21. Streptomycin is the most commonly used antibiotic for inducing mutations in many cytoplasmic genes (DPMT 1986).

MOLECULAR BASIS OF MUTATION

1. The gene mutation is caused by a change in the sequence of nitrogen bases in DNA.(CPMT 1999)
2. Mutation can take place in the following ways:
   (i) **Deletion mutation:** Gene mutation, which is caused due to loss for deletion of a single nucleotide in triplet codon of a gene.
   (ii) **Insertion mutations:** Gene mutation, which occurs due to addition of one or more extra nucleotides to a gene.
   (iii) **Substitution mutation:** Gene mutation in which a nitrogen base of triplet codon of DNA is replaced by another nitrogen base or some other derivatives of nitrogen base.
   Substitution mutation may be ofthe following kinds:
   (i) **Transition:** Replacement of a purine by a purine or pyrimidine by a pyrimidine. Transitions occur due to tautomerization, deamination or base analogs.
   (ii) **Transversion:** Replacement of a purine by pyrimidine or vi ce versa (BHU 1986). Replacement of a pyrimidine nucleotide by a purine nucleotide is called transversion (AMU 1989).
3. Tautomeric A pairs with C, G pairs with T, C pairs with T and T pairs with G.

KINDS OF MUTATIONS

1. **Frameshift mutation**
   - If the mutation involves loss or addition of a single base or a segment of DNA, then the entire reading frame will change from the site or mutation. (BHU 1999)
   - Mutations resulting from changes reading the set of nitrogen bases due to addition or removal of a nitrogen ba se is called frameshift mutation.

- Acridine dyes (acridine orange, acridine yellow and proflavin) act by permitting base additions or deletions. This results in frameshift mutation or gibberish mutation. (AIIMS 2000)

2. **Mis-sense mutation:** The replacement of one amino acid by another.
3. **Non-sense mutation:** The conversion of a normal codon into any of the non-sense codon (UAA, UAG or UGA).
4. **Silent mutation:** Mutations which do not cause any change in the protein are referred to as silent mutations. Thi s happens when the mutation take place in the third base (wobble position).

VARIATIONS

1. All dissimilarities of characters between members of the same species are called variation.
2. The heritable variations form the 'raw material' for evolution.

KINDS AND CLASSIFICATION

1. Variations may be morphological, physiological or psychological.
2. Morphological variations may relate to the shape, size, colour and structure of body or its parts.
3. Physiological variations deal with the functional capacities an d psychological to mental attributes. .

Somatogenic and Blastogenic Variations

1. Somatogenic, somatic or phenotypic variation is restricted to the body parts. These variations are nonheritable and hence useless *for* evolution.
2. Blastogenic vari ations are germinal, i.e., due to differences in the genotypes of variants (CPMT 2000).
3. Blastogenic variations *form* the basis of evolution (CBSE1999).

Continuous and Discontinuous Variations

1. Continuous variations are small and graded; these inclu de variation in colour, shape, size, weight and structure of body parts. These variations can be somatogenic or blastogenic.
2. Discontinuous variations are discrete and distinct variations. These arise by sudden genic changes, i.e., mutations.
3. Discontinuous variations are called qualitative (substantive) when these relate to colour, *form,* size and other characteristics of body and its parts.
4. Ancon sheep of Seth Wright (1791) is a famous example of substantive discontinous variations.

5. Quantitative (meristic) variations are called positive when the number of parts in varient is more than normal and negative when the number is less than normal.(CBSE 1993)
6. More than five digits in hands or feet, and more than twelve pairs of ribs in certain people, six or more arms in starfishes, and more sepals and petals in flowers are common example of positive meristic variations.
7. A child is having only one kidney since birth. This variation is known as negative meristic (CPMT 1983).

Determinate and Indeterminate Variations

1. Determinate variations are adaptive and selective variations evolutionary lines, developing progressively generation after generation. These are also called orthogenic variations. (CPMT 1994)
2. **Indeterminate** variations are of no particular evolutionary line and of no specific limit of development. These are caused by mutation and appear suddenly.

EXTRANUCLEAR GENES AND CYTOPLASMIC INHERITANCE

1. Some characters are controlled by cytoplasmic genes or plasma genes.
2. The sum total of plasma genes or extrachromosomal hereditary determinants in a cell *form* 'plasmon'.
3. The organelles responsible *for* cytoplasmic inheritance among eukaryotes are chloroplasts and mitrochondria. (AIIMS 1997)
4. Cytoplasmic genes were first observed in *Mirabilis jalapa* with variegated leaf" colouration by Correns in 1909.
5. Cytoplasmic male sterility is passed down maternally. (CBSE 1997)

KAPPA PARTICLES IN PARAMECIUM

1. Sonneborn (1943) gave the name *'Kappa'* to the extrachromosomal particles present in the cytoplasm of *Paramecium aurelia.*
2. Paramecia with kappa particles are called killers. They secrete a poison namely paramecin which can kill the sensith.e paramecia.

NEUROSPORA GENETICS

1. The *Neurospora* (pink bread mold), a fungus of class Ascomycetes, has been extensively used as an experimental material in the study of genetics.
2. Dodge is known as father of neurospora genetics.
3. *Neurospora* is an excellent genetic material (like *Drosophila)* because Neurospora can easily be grown in a defined media in the laboratory. It

is a haploid organism and recessive genes can express themselves. It has a short life cycle, only of 10 days.

4. *Neurospora* is known as *'Drosophila* of the plant kingdom'. (BHU 1987)
5. Like other fungi *Neurospora* is a saprophyte.
6. *Neurospora* can easily be grown on a minimal medium containing sugar, a *few* simple salts and biotin.
7. The asexual reproduction occurs through the asexual spores called conidia whereas the sexual reproduction occurs through ascospores.
8. During sexual reproduction, the hyphae of + and - strain mycelia come into contact.
9. The haploid nuclei *from* the two hyphae *fuse* and *form* a diploid zygote.
10. Soon after zygote formation, the diploid nucleus undergoes meiosis followed by mitotic division to *form* eight haploid nuclei.
11. Meiosis takes place in *Neurospora* during ascospore development.
12. Lindgren (1993) reported that crossing over in *Neurospora* takes place at four-strand stage. (CBSE 1999,2000)
13. The evidence that crossing over occurs at fo ur-strand stage and not at two strand of chromosomes comes from 2:2:2:2 arrangement of ascospores in *Neurospora.*
14. If the crossing over had occurred at two-strand stage in *Neurospora,* the ascospores would be arranged in 4 : 4 position.
15. Tetrad analysis is d one in the ascospores of *Neurospora* to study the process of crossing over and evidence for the occurrence of first - division and second division segregations.
16. The wild -type (+) *Neurospora* produces pink conidia and a mutant strain called albino (al) produces white conidia.
17. The sequence of ascospores, + + + + al al al al, results from the segregation of alleles at first meiotic division.
18. The sequence of as cop ores, + + al al + + al al, results from the second-division segregation (a crossover occurs between two of the homologous chromatids during prophase I).
19. In *Neurospora,* 8 ascospores are formed 2A, 2a, 2A, 2a; it shows second-generation division. (CBSE 1998)
20. aaaa AAAA arrangement of ascospores in *Neurospora* does not represent second-division segregation.(CBSE 1993)
21. Chief advantage of linear arrangement of ascospores in Neurospora is that in genetic studies it allows ready observation of mutant phenotype.

NUTRITIONAL MUTANTS

1. Technique for detecting and screening the nutritional mutants in *Neurospora crassa* was developed by Beadle and Tatum in 1941.
2. Beadle and Tatum proposed their 'one gene one enzyme hypothesis' on the basis of biochemical genetics in *Neurospora crassa.(BHU* 1995)
3. According to one gene one enzyme hypothesis, a metabolic reaction is usually completed in several intermediate steps and each step requires a specific enzyme synthesized by a specific gene. (CPMT 1995)
4. In the case any of these genes gets mutated, the reaction cannot proceed to completion due to the absence of a specific enzyme.
5. Beadle and Tatum (1941) induced nutritional mutation in *Neurospora crassa* and *N. sitophila* by X-ray irradiation of conidia.
6. The wild strain of *Neurospora* which can grow on a simple minimal medium is known as prototroph.
7. A mut ant who has lost its ability to synthesize one or more essential compounds is called auxotroph (DPMT 1982,CPMT 1999).
8. A mutant of *Neurospora* which fails to grow on synthetic medium unless supplemented with a certain nutrient is called auxotroph.

GENE EXPRESSION

1. Gene expression results in the formation of a polypeptide by joining amino acids in a predetermined order.
2. The genetic information present in the gene is not always used; many genes remain silent, express only when their products are needed.
3. There are some genes, which are expressed always, as their products are constantly needed for cellular activity. These genes are called as 'house- keeping genes'.

GENE EXPRESSION IN PROKARYOTES

1. The expression of bacterial genes is usually controlled to ach ieve maximum cellular economy.
2. The genes will be turned on or off as per requirement. The gene can be turned on by an inducer and turned off by a repressor. (CSSE 1992)

OPERON CONCEPT

1. French microbiologists Francois Jacob and Jacques Monod proposed 'Operon Model' to explain the regulation of gene expression in the bacterium *Escherichia coli* in 1961.
2. In bacteria, different genes concerned with the same traits ae often

found clustered together in a group known as operon.

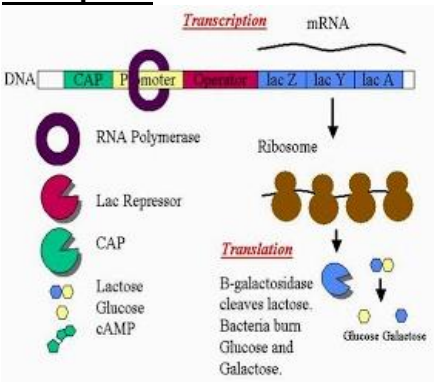
3. In an operon, the component genes are transcribed as a unit and thereby under a joint control.
4. The correct sequence of genes within an operon are regulator, promoter, operator and structural genes. (CPMT 1987)

   Regulator z ene: A gene that codes for a repressor protein molecule. The repressor molecule binds to the operator and prevents RNA polymerase, an enzyme needed for mRNA synthesis, from binding to the promoter. **Promoter:** A short sequence of DNA where RNA polymerase first attaches when a gene is to be transcribed.

   Operator: A short sequence of DNA where the repressor binds, preventing RNA polymerase from attaching to the promoter. This i s called the on/off switch of transcription.

   Structural Z enes: One to several genes for a metabolic pathway that are transcribed as aunit.
5. In operon model, RNA polymerase binds to promoter gene. (BHU 97)
6. A gene which synthesises a repressor protein is regulator gene.
7. In operon model, regulator gene stops the transcription ofthe structural genes.
8. Two bacterial operons in *E. coli* are Lac operon and Trp operon.

Lac Operon

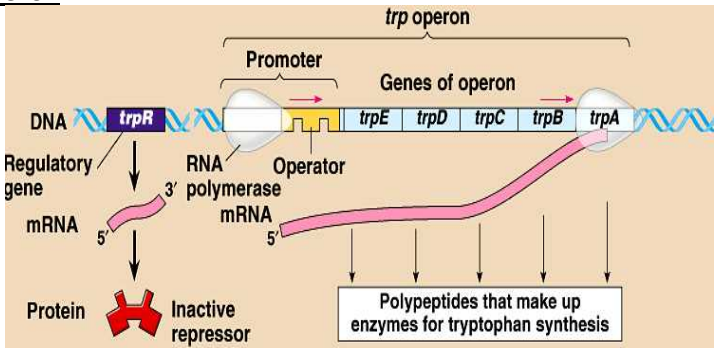


9. Lac operon consists of a regulator gene (lac I), promoter gene (lac P), operator gene (lac 0) and three structural genes namely lac Z, lac Y and lac A.
10. The structural genes lac Z, Y and A code for three enzym es called β galactosidase, pemlease and transacetylase catalysing a catabolic pathway.
11. β-galactosidase breaks down the disaccharide lactose to glucose and galactose.
12. The permease facilitates the entry of lactose into the cell.
13. Transacetylase has an accessory function in lactose metabolism.
14. Under normal conditions (absence of lactose) a repressor protein produced by lac I gene gets attached to operator switching off the

structural genes Z, Y and A.

15. Lactose on introduction (presence o f lactose) modifies the shape of repressor (forming inactive repressor -lactose. complex) making it incapable to get attached on operator.
16. The promoter is able to bind to RNA polymerase, and it carries out transcription of structural genes and three enzymes are produced.
17. Lactose works an inducer turning switch on (CBSE 1994).
18. An environmental agent that triggers transcription from an operon is an inducer. (CBSE 1995)
19. The lac operon is an example of inducible operon. (CBSE 1995)

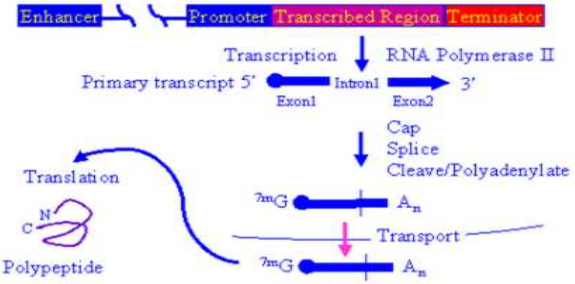
Trp Operon



1. Trp operon consists of a regulator gene (trp R), promoter gene (trp P), operator gene (trpO) and five structural genes namely trp E, D, C, B and A.
2. Structural genes code for enzymes catalysing the synthesis of the amino acid tryptophan and constitute an anabolic pathway.
3. Under normal conditions the structural genes are on, the apo-repressor produced by the regulator gene R is unable to attach to the operator.
4. Tryptophan on introduction works as 'co -repressor'. It combines with apo- repressor to form the functional repressor.
5. The functional repressor now binds to the operator, preventing the transcription of the operon. The switch is turned off.
6. The trp operon is an example of repressible operon.
7. Thus the ope ron model can explain both the induction and repression phenomena seen in prokaryotes.

GENE EXPRESSION IN EUKARYOTES

1. In eukaryotes, functionally related genes may not be clustered together constituting an operon.
2. The most popular model is known as 'Britten -Davidson model' or Gene-battery model' proposed by Britten and Davidson in 1969.



3. Gene-battery model assumes the presence of four classes of sequences:
   (i) **Producer gene**: A producer gene is comparable to structural ge ne of prokaryotic operon.
   (ii) **Receptor site** : A receptor site comparable to operator gene of bacterial operon and one such receptor site is assumed to be present adjacent to each producer gene.
   (iii) **Integrator gene**: Integrator gene is comparable to regulator gene and is responsible for synthesis of an activator RNA. The activator RNA mayor may not give rise to proteins before it activates the receptor.
   (iv) **Sensor site**: A sensor site regulates the activity of integrator gene. Activator gene can be transcribed only when th e sensor site is activated. The sensor sites are recognized by agents, which change the patterns of gene expression like hormones and proteins.

SPLIT GENES

1. The regulation of gene expression in eukaryotes is more complex than in prokaryotes.
2. Control of gene expression occurs at four levels in eukaryotes.
3. In the nucleus, there is transcriptional and post-transcriptional control.
4. In the cytoplasm, there is translational and post-translational control.
5. The eukaryotic gene is split into exons and introns.
6. The regions of a gene which become part of mRNA and code for

different regions of protein are referred as exons.

7. The regions of a gene which do not form the part of mRNA and are removed from primary mRNA are called as introns. (CBSE 1992)
8. During transcription, the primary mRNA product contains regions corresponding to both exons and introns.
9. The primary mRNA transcript is processed before it becomes mature mRNA transcript.
10. During splicing and processing, the intronic parts of mRNA are removed whereas the exonic portions are spliced together.
11. The identical primary mRNA transcripts can be processed differently in two cells giving rise to different proteins.
12. The mature mRNA thus formed will be transported across the nuclear membrane into the cytoplasm.
13. The mature mRNA then associate with ribosomes and are translated into proteins.
14. Split genes are made of exons and introns coding sequences are called exons.
15. The concept of split gene is that the coding sequences in many eukaryotic genes are often separated by non-coding sequences. (CBSE 1995)

SOME MISCELLANEOUS POINTS

1. E.H. Fisher & E.J. Krebs of USA got Nobel Prize in 1992 for their discoveries concerning reversible protein phosphorylation as a biological regulation mechanism.
2. Bacteria may become resistant to drug by developing mutant resistant strains. The genes for drug resistant are present either on the bacterial chromosome or R plasmid.
3. For producing test tube baby, ovum of mother is obtained, and fertilized by her husband's sperm in vitro, where it is allowed to develop upto 32 celled Morula stage thereafter it is implanted back into the uterus for development.
4. Exonuclease is an enzyme, which attacks at the free end of a polynucleotide where as endonuclease attacks at some point within the polynucleotide chain.
5. The enzyme tyrosinase brings about the synthesis of melanin. Lack of the enzyme results in the absence of Melanin pigment so that albinos have white skin and hair.
6. Animals have DNA as genetic material. All the cells of Transgenic

animal's body develop from transgenic zygote hence transgenic animals are those which have foreign DNA in all its cells.

7. Done marrow cells of our body are diploid.
8. In sickle cell anaemia, at the position 6 of p chain of haemoglobin, Glutamic acid is re placed by valine. This leads to the formation of abnormal Hb and is a classical example of point mutation.
9. The change of the lighter coloured variety of peppered moth (Diston betularia) to its darker variety (Biston Carbonaria) is due to mutation in a single gene.
10. Somatostatin was the first hormone produced using genetic engineered E. coli by Boyer and his colleagues at California in 1977.
11. Crea, Kraszewski and others (1979) raised strain of genetically engineered E.Coli capable of producing human insulin.
12. After crossing two plants, the progeneis are found to be male sterile. This phenomenon is found to be maternally inherited and is due to some genes, which are present in Mitochondra.
13. Polyamines like spermidine and cadaverine (instead of Histone) are associated with DNA packaging in prokaryotes.
14. The term 'Hot Spots' was used by Benzer for the sites which are more mutable than other sites studies in 1978 revealed than 5 -melhyl cytosine residue occur at the position of each hot spot for spontaneous point mutation.
15. In the beginning of S phase, DNA replication occurs. DNA Replication can occur in diffuse/less tightly coiled euchromatin.
16. The term genetic mutation covers somatic mutation as well as germinal mutation (occurring during reproduction).
17. Skin colour in human being is believed to be controlled by at least three pairs of genes Aa, Bb, Cc located in different chromosomes and inherited separately.
18. Industrial melanism refers to the occurrence, of dark (melanic) forms (of insects) in region with high industrial pollution - where surfaces on \vhich the insects rest are darkened by soot and where atmospheric SO_2 level are high enough to prevent the growth of lichens. The mutant dark forms of insects would not be easily seen and avoid being preyed ; thus the population of dark forms of insect would increase- natural selection being in their favour as compared to lighter forms which would be easily sighted and eaten.
19. In prokaryotes, the nucleoid consists of ds circular molecule of DNA without histone protein.

20. Plasmids replicate autonomously. These carry a signal situated at their replication origin which determines how many copies are to be made and this number can be artificially increased for cloning a given gene.
21. Interferons (INFs) are a group of three vertebrate glycoproteins (α,β,γ). Out of these, to (α and β) be produced within virally infected cells. Interferon induces, among adjacent cells, an antiviral state by inducing synthesis of the enzyme, which inhibits the viral production cycle.
22. The 3 -D structure of tRNA deduced from X -ray diffraction analysis shows that it look like a twisted "L".
23. Homeotic Genes are control genes, which either by getting expressed orby remaining silent during development influence the differentiation of organs. These have been found i n insects, one nematodes and some plants. A DNA sequence called Homeobox, present in the genes, is involved in specification of organs. A mutation that causes a body part to develop in a appropriate position in an organism, is called homeotic mutation e.g. in Drosophila, such mutation may cause legs to develop on the head in place of antennae.
24. Telomeres seal the ends of chromosomes rendering them non-sticky.
25. Ribozymes, catalytically active RNA molecules discovered in 1980, are self splicing introns indicat ing there possible role as Intermediates in the evolution of biological systems iI-om abiotic substances.
26. Genes and Cistrons words are some times used synonymously because-many genes are monocistronic.
27. Large circular plasmid of 1700-kilo base has been rep orted in RHIZOBIUM MELILOTI. So, maximum number of bases in plasmids discovered so far is 1700 kilo base. (CBSE-2001)
28. Probability of having a son in one delivery is ½ (50%) and of daughter is also ½ (50%).
29. About 100 types of restriction enzymes are avail able. Each restriction enzyme cleaves a molecule only at a particular nucleotide sequence.
30. Genetic Drift is most likely to occur in small, isolated populations. In these, even a chance change in gene frequency is perpetuated because the gene pool is small.
31. Forthcoming generation are less adaptive than their parental generation due to natural selection (BHU 2001).
32. In his experiment, Mendel obtained wrinkled pea. The wrinkling was due to deposition of sugar instead of starch. This happened due to absence of starch-branching enzymes.

33. In round seeds starch-branching enzymes (SBE-I) is found but is absent from wrinkled seeds. ,
34. Monoclonal antibodies are identical molecules specific *for* one type of antigen these are obtained by injecting the target antigen int o a rat or mouse. Sometimes later, the spleen cells producing antigens are fused with myeloma cells to produce monoclonal antibodies.
35. Foetal sex can be determined by examining cells from the Amniotic fluid by looking *for* Barr bodies.
36. Pure line breed conti nue to breed true and are formed in homozygous individuals that are identical.
37. Most of the phenot)pic characters are controlled by genes present in the chromosomes but some character are expressed by factors present in cytoplasm. These factors lying in the cytoplasm are called plasmagenes. They also transmit character from generation to generation. It was first described by Correns. e.g. (i) Kappa particles in paramecium (ii) Shell - coiling in Snail (Dextral - towards right and Sinistral- towards left) (iii)Plastid inheritance in Mirabiles (iv) Pigments inheritance in Ephestia.
38. Barr bodies are studied in interphase nuclei of human female.
39. The number of base substitutions possible in amino acid codons is - 549 (There are 61 codon or amino acids. Each codon has three bases, each of which can undergo transiti on and transverse substitution. 61 x $3^2 = 549$).
40. M-RNA is genetic RNA. Non-genetic RNA is tRNA and rRNA.
41. Unwindase or helicase takes part in separation of two strands. In prokaryotes, helicase or unwindase is assisted by Gyrose in this function.
42. Wobble hypothsis establishes economy in tRNA molecules.
43. Wobble hypothesis was proposed by Crick (1968).
44. Wobble hypothesis proposed that the first two nucleotides of a triplet code specify an amino acid.
45. Cosmid is f ragment of DNA, about 40,000 base pairs in length, inserted in bacteria *for* replication along with bacterial DNA to produce copies *for* gene library.
46. Linkage was initially discovered by Bateson & Punnet but they thought of the phenomena of coupling and repulsion. Morgan found linkage to be a fundamental *feature.*
47. Parthenogenesis is development of a single gamete, generally female.

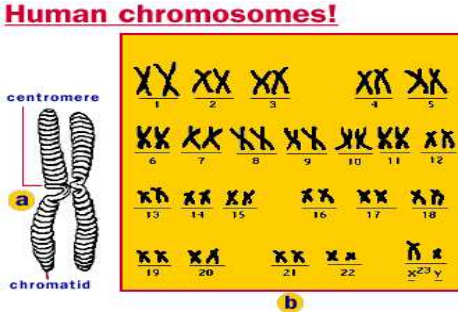
In higher organism parthenogenesis is successful only if the new organism develops from diploid female egg.

48. Planosome are sup ernumerary chromosome formed due to non - disjunction at the time of meiosis.
49. Complete nuclotide sequencing has been carried out of the whole human genome with the help of automated DNA sequencing machines on *June* 26, 2000. The genome contains 3.2 billion nu cleotides however the total number of genes and their function have yet to be determined.
50. The number of base pairs in genome of Oro sop hila is 132million.
51. 99.8% of ON A is similar is all individuals, only 0.2% is different causing variations.
52. m-chromosome are minute but functional chromosomes (0.5μm or less) which occur in some Bryophytes and insects.

HUMAN GENETICS

HUMAN CHROMOSOMES

1. Human chromosome preparations were first made by **Flamming** in 1882.
2. First determination of the chromosome number was made by **Winiwarter** in 1912. He reported 2n = 48.
3. The correct human chromosome number 2n = 46 (BHU 1990, 93; CBSE 1989) was discovered by **Tjio** and **Levan** in 1956 (CBSE 1994).
4. An **autosome** is a chromosome other than sex chromosomes (CPMT 1999). The term 'autosome' was introduced by **Montgomery** in 1904.
5. Sex chromosomes X and Y were discoverd by Stevens and Wilson (BHU 1994).
6. **Sex chromosomes** are also called as **'heterosomes'**. The term 'sex chromosome' was introduced by **Wilson** in 1906.
7. The total number of chromosomes in human beings is 46 (MPPMT 1996). There are 22 pairs of autosomes and two sex chromosomes (CPMT 1997).
8. The sex chromosome constitution of male is XY and of female is XX.
9. **Karyotype** is the chromosomal complement of an organism. It is made by the arrangement o f metaphase chromosome pairs in a standard sequence.
10. The short arm of a chromosome is called as 'p' arm and the longer arm as 'q' arm (BHU 1999).

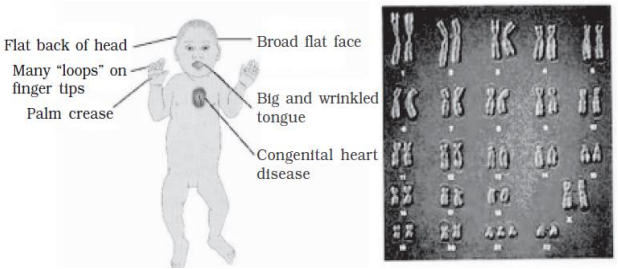


11. Trisomy is a condition in which the chromosome number is 2n + 1 (CBSE 2000).
12. Human being share banding pattern of chromosomes with Primates.
13. There are seven groups of autosomal chromosomes

according to the Dever (colorado) convention (1960) which is based on size of chromosomes and position of centromere.

TRISOMY IN HUMAN BEINGS

Down's Syndrome



A representative figure showing an individual inflicted with Down's syndrome and the corresponding chromosomes of the individual

1. It was discovered by the British physician **Langdon Down** in 1866, also called **mongolism** (CPMT 1983).
2. Down syndrome is caused by mutations that involve entire chromosome rather than a single gene.
3. Down's syndrome is due to the **trisomy of 21st autosomes** (MPPMT 1992, 95). A Down's syndrome will be 45A + XX/XY (CBSE 1991,97).
4. Down's syndrome is characterized by **mental retardation**(IQ below 40).
5. Down's syndrome is due to one extra chromosome in 21st pair (AFMC 1995; CPMT.1998;CBSE 1992).
6. **Nondisjunction** during oogensis is the cause of Down's syndrome (CBSE 1991,98).
7. Translocation of a po rtion of chromosome 21 on autosome 14 also results in Down's syndrome (BHU 1990).

8. In a translocation Down's syndrome chromosome number is 2n = 46.

Edward's Syndrome

1. It was discovered by **Edward** in 1960 and is an abnormality due to **trisomy 18** (MPPMT 1994, AIlMS 1999).
2. A child suffering from Edward's syndrome dies generally around 3 -4 months of age, but may be delayed for nearly two years.

Patau's Syndrome

1. It was discovered by Patau in 1960 and is an autosomal abnormality due to trisomy-13.

DELETIONS IN HUMAN BEINGS

1. Deletion is the loss of a segment of the genetic material from a chromosome.
2. The size of the deletion can vary from a single nucleotide to sections containing a number of genes.

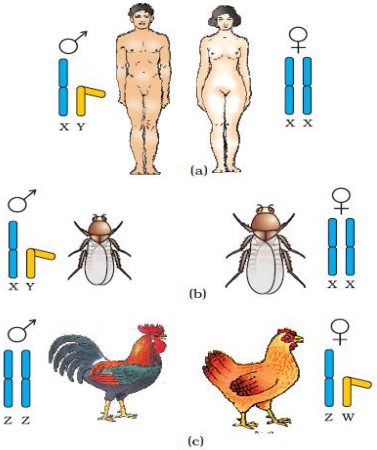
Cat cry Syndrome (Cri du chat syndrome)

1. Cat cry syndrome was discovered by **Lejeune** in 1963 and is due to the deletion of a large part of the small arm of one of the **5th autosomes**.
2. Affected child during infancy has a characteristic high-pitched cry of a kitten.
3. This syndrome is associated with malformation of the larynx.

Chronic Myeloid Leukemia (CML)

1. Patients of CML carry **'Philadelphia chromosome'** (AIIMS 1994) which is one of the 22 autosornes that has lost most of the distal part of its longer arm.
2. CML is characterized by an excess of granular leucocytes in the blood. With the increase in the number of leucocy1es, there is a reduction in the nuober of RBCs resulting in severe anaemia.

CHROMOSOMAL BASIS OF SEX DETERMINATION



Determination of sex by chromosomal differences: (a,b) Both in humans and in *Drosophila*, the female has a pair of XX chromosomes (homogametic) and the male XY (heterogametic) composition; (c) In many birds, female has a pair of dissimilar chromosomes ZW and male two similar ZZ chromosomes

1. **'Chromosomal theor: of sex determination'** was proposed by an American **McClung** (1902) based on chromosome study of **grasshopper.**
2. In man, sex is determined at fertilization by the nature of the sperm that fertilizes the egg. Y -bearing sperms produce male zygotes. X -bearing sperms produce female zygotes.
3. Wilson (1909) proposed the term Y chromosome for odd chrpmpspme.
4. In man, the female individuals are **homogametic** with chromosomes constitution 44A + XX (CPMT 1989, 92).
5. The male individuals are **heterogametic** with the chromosome constitution 44A + XY.
6. Males produce two types of spermatozoa, 50% with X -chromosome and 50% with Y -chromosome in addition to 22 autosomes.
7. Since X-carrying and Y -carrying sperms are produ ced in 1:1 (CPMT 1989, 93; MPPMT 1993).
8. **Y is the masculinization chromosome** ; it's small arm carries a gene designated SRY (for sex-determining region Y).
9. **X is the feminization chromosome,** It carries genes for femaleness.

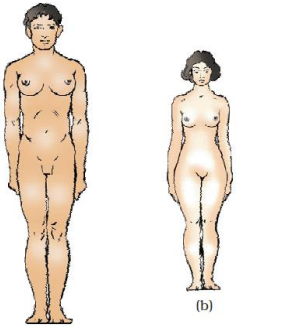
GYNANDROMORPH

1. Gynandromorph is an individual of a dioecious (unisexual) species, which is a sexual mosaic.
2. It is typically male in certain portions (sectors) of the body and female in others (CPMT 1997).
3. The cases of gynandromorphism have been reported in man, *Drosophila*, silkworms, bees, butterflies, beetles, etc.
4. **Gynandromorphs** are half male and half female (CPMT 1996). 13. A fruitfully exhibiting both male and female traits is gynandromorph (CBSE 1996).
5. Gynandromorphism in certain files is the resul t of misdivision of chromosomes whereby one of the X chromosomes gets lost (CPMT 1995).
6. The term **'gynandromorph'** was introduced by **Goldschmidt** in 1915.
7. An individual with half body male and half body female is known as gynandromorph (CBSE).
8. Gynandromorphs appear in Drosophila because of X chromosomes.

HUMAN SEX ANOMALIES

1. Human sex abnormality arises due to nondisjunction of sex chromosomes during meiosis. Four important abnormalities are Turner's syndrome, Klinefelter's syndrome, XYY syn drome (criminal syndrome) and poly-X females.

Turner's Syndrome



(a) Tall stature with feminised character

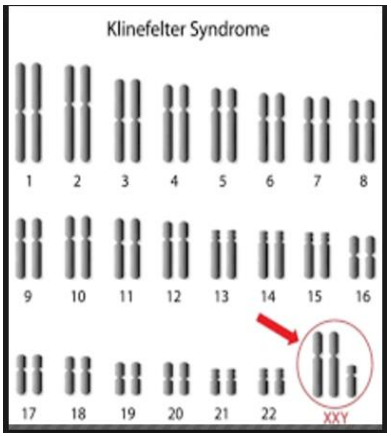
(b) Short stature and underdeveloped feminine character

1. The chromosome number 2n = 45, the chromosomal formula is 44A + XO (BHU 1983, 89; CPMT 1991; MPPMT 1992; CBSE 1992, 93).

2. Turner's syndrome is a sterile female. The important character istics are short stature, **'webbing of the neck'** (AIIMS 1993), irregular hairline, no mens - truation, no breast development and lack of egg production.

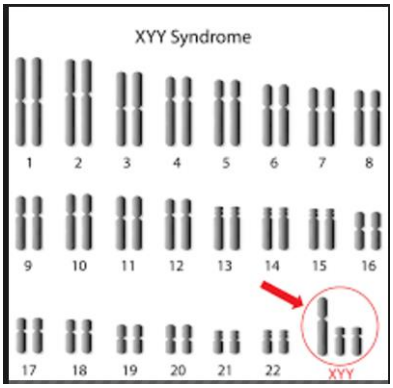
Diagrammatic representation of genetic disorders d ue to sex chromosome composition in humans : (a) K linefelter Syndrome; (b) Turner's Syndrome

Klinefelter's Syndrome



1. The chromosomal number 2n = 47; chromosomal formula is 44A + XXY (CPMT 1994,96,97; CBSE 1995; AIIMS 1996; MPPMT 1998).
2. Klinefelter's syndrome is a male in general appear ance; testes are Underdeveloped, enlarged breast (gynecomastia), mentally defective and abnormally tall.
3. The karyotypes of extreme Klinefelter's syndrome are 44A + XXXY (DPMT 1996), 44A + XXXXY, 44A + XXXYY. etc.
4. Mental retardation increases with incr eased X complement (CBSE 1998).

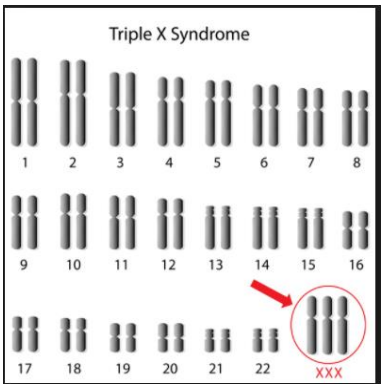
XYY Syndrome (Criminal syndrome)



1. XYY males were first reported by Sandberg in 1961; the chromosomal number 2n = 47, chromosomal formula is 44A + XYY.
2. XYY male usually overproduces male hormones.
3. There is a high f requency of this syndrome among criminals, so called 'criminal syndrome'.

Poly-X Females

1. The chromosome number 2n = 4 7: chromosoma l formula is 44A + XXX. This is called as 'triplo-x-females'.



SEX CHROMATIN

1. Sex chromatin body was discovered by Canadian geneticists Murray L. Barr and E.G. Bertram (19-9) in interphase nerve cell of female cats.
2. In human females, Barr body can be easily demonstrated in cells of buccal epithelium and hair roots.
3. Barr body is a highly condensed structure: it lies against the nuclear membrane of the interphase cells of females (BHU 1993; MPPMT 1998).
4. According to British geneticist **Mary Lyon**, (1961) one of the two X chromosomes of a normal female becomes heterochromatic and appears as Barr body.
5. This inactivation of one of the two X-chromosomes of a normal female (CBSE 1995) is dosage compensation or 'Lyon's hypothesis'.
6. Turner's syndrome is sex chromatin negative and Klinefelter's syndrome is sex chromatin positive (DPMT 1983; CPMT 1993, 97).
7. Drumsticks are the se x chromatin present in the neutrophil (polymorphonuclear leucocyte) of 3 to 5% cells in females. Drumsticks are absent in the neutrophils of males. (AIIMS 1999).
8. **Y- chromosome** can be identified as bright spot by staining cells with **acridine dyes** and viewing them under the ultraviolet microscope.

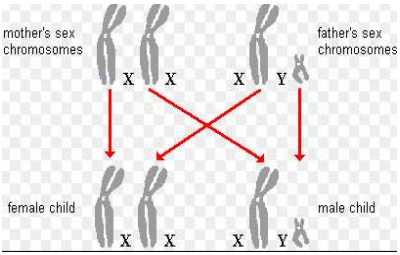
AMNIOCENTESIS

1. The technique which makes use of amniotic fluid for the detection of prenatal disorders is called as **amniocentesis** (MPPMT 1995, CBSE 1998).
2. Foetal sex can be determined by examining cells from the am niotic fluid by looking for Barr bodies and sex chromosomes (CBSE 1997).

3. Amniocentesis is the process to determine any hereditary disease in the embryo (CBSE 1997).

INHERITANCE RELATED TO SEX

1. The genes located exclusively on X -chromosome are called **sex-linked** or **X linked** genes (CPMT 1987,99).
2. The genes that occur only on the Y -chromosome are called **holandric** genes.



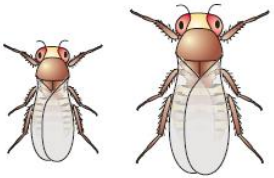
3. The genes present on homologous regions in sex chromosomes are called **XY-linked** genes.
4. A man carrying a sex linked gene on his Y chromo some will transmit this gene to all his male offsprings.

SEX-LINKED INHERITANCE

1. The recessive X-linked genes have characteristic **criss-cross** inheritance, *i.e.,* male transmits his X -linked recessive genes to his grandson through his daughter (MPP~ 1993).
2. Sex linked characters are mostly recessive.
3. Criss – Cross inheritance was discovered by Morgan.
4. Criss – Cross inheritance in Drosophila led to the discovery of sex – linked inheritance.

Drosophila

1. First X-linked gene was discovered by **T.H. Morgan** (1910) for white-eye mutation.



Drosophila melanogaster
(a) Male (b) Female

2. In *Drosophila* **red eye** colour, **white-eye** colour and **gray body** colour are examples of X-linked genes (AIIMS 1989).
3. A cross between red -eyed female and white-eyed male fly produce red -eyed females and males in F_1, In subsequent cross in F_2, all females red - eyed while 50% of males are white-eyed.
4. A cross between white-eyed female and red-eyed male fly produces red-eyed females and white-eyes males in F_1, In subsequent cross in F_2 50%

of females and 50% of males are white-eyed.

5. A female is homozygous or heterozygous and male is always hemizygous (only one allele is present).

Humans

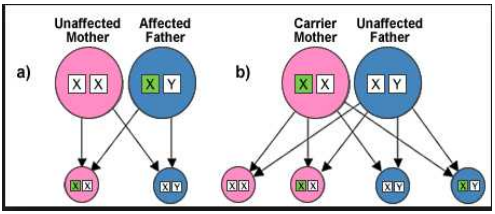
6. A number of well -known diseases an d traits are cau sed by X -linked recessive genes (CPMT 1996, BHU 1998).
7. The most important and common X-linked diseases are red-green colour blindness, haemophilia, Duchenne muscular dystrophy and fragile X syndrome.
8. Certain X-linked diseases such as defect ive dentine which causes the teeth to wear rapidly are dominant character. This character is expressed more frequently in females.

Colour Blindness

1. Colour blindness in man is a sex-linked abnormality (CPMT 1982,95; AIIMS 1997) caused by a recessive X-linked gene.
2. A very rare autosomal gene causes total colour blindness (CPMT 1992).

Haemophilia

1. Haemophilia is also called 'bleeder's disease'. It was first studied by John Cotto in 1803.



2. Like colour blindness, haemophilia is a well-known disorder which is sex linked (CPMT 1985,95. BOO 1996; MPPMT 1998). It is a recessive condition.
3. In a patient of haemophilia, blood clotting is deficient (CBSE 1992; Orissa JEE 1993; AFMC 1998) because of lack of the necessary substrate, thromboplastin (CPMT 1991).

HOLANDRIC OR Y-LINKED INHERITANCE

1. Holandric genes are those that occur on the Y-chromosome only (CBSE 1994; AIIMS 1997); they are not expressed in females.
2. Hairy ears **(hypertrichosis)** in man is inherited through genes on Ychromosome.

3. Y -linked holandric genes are transmitted directly from father to son.

XY-LINKED INHERITANCE

1. The genes which occur in homologous sections of X and Y - chromosomes are called XY -linked genes and they have inheritance like the autosomal genes.
2. Examples of XY -linked genes are those *for* the inheritance of **xeroderma pigmentosa** (a skin disease characterized by the pigment patches and cancerous growth on the body) and nephritis (a kidney disease).

SEX-INFLUENCED OR SEX-CONTROLLED GENES

1. Inheritance of pattern baldness in man is an example of sex-influenced character (EAMCET 1987). This trait is dominant in males and recessive in females (CBSE 1992).
2. Pattern baldness is more common in males than in females.

SEX LIMITED GENES

1. In sex-limited genes, the phenotypic expression is determined by the presence or absence of one of the sex hormones. Their phenotypic expression is limited to sex or the other.
2. Many secondary characters like mammary gland development and milk production in female are sex limited.

BLOOD GROUP INHERITANCE

1. **Landsteiner** in 1900 discovered that agglutination or clumping of RBCs takes place by antigen-antibody reaction.
2. **Landsteiner** recognized three kinds of blood types or blood groups: group A (with antigen A) group B (with antigen B) and group O (without antigens) (BHU 1991; CPMT 1999).
3. The fourth and most rare AB blood group was discovered in 1902 by two of **Landsteiner's** students, **Von Decastello** and **Sturli**.
4. Antigens A and B were thought as proteins. Recent chemical investigations have shown that A and B antigens are not proteins, but are **mucopolysaccharides** (sugar + amino acids) of 3,00,000 molecular weights.

5. Characteristics of human blood groups are as follows:

Blood Group	Antigen in RBCs	Antibodies in plasma	Can donate to groups	Can receive from groups
A	A	anti-B	A,AB	O, A
B	B	anti-A	B,AB	O,B
AB	A & B	None	AB	O,A,B,AB
a	None	anti-A anti-B	O,A,B,AB	O

6. As a rule, a person is given transfusions only from a person of the same blood group. But in emergency, some combinations can be made.
7. Agglutination of the red blood cells in blood vessels of he recipient will cause obstruction of the capillaries usually resulting in death.
8. RBCs of the person s of **'O' blood group** have no antigens, so their blood can be donated to persons of all blood groups (A, B, AB and O). Persons or 'O' blood group are called **'universal donors'** (CPMT 1979,87,93; BHU 1997*).*
9. RBCs of the persons of **'AB' blood group** have both antigens A and B. but their plasma is without antibodies A and B (MPPMT 1996,98). Such persons can receive blood of all other persons; they are 'universal recipients' (CPMT 1971,86,92; BHU 1979, AIIMS 1999).
10. Inheritance of A, B, AB and O blood types in man are discovered by Bernstein.
11. A set of three multiple alleles (CBSE 1990) on the autosome is responsible for the four blood types.
12. The gene controlling blood types has been labelled as I (standing for **isohemagglutinin**) or L (after **Landsteiner**).
13. The genes exist in three different allelic forms L^A, L^B and L^O.
14. L^A gene produces antigen A and L^B produces antigen B. L^O produces no antigens (AIIMS 1998; AFMC 2001).
15. L^A and L^B are both dominant to L^O gene (CBSE 1998; CPMT 2000).

Phenotypes and Genotypes of Blood Groups

Phenotype	Genotype
A	L^AL^A, L^AL^O
B	L^BL^B, L^BL^O
AB	L^AL^B
O	L^OL^O

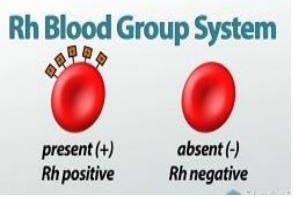
16. If a human mother has blood group O, the foetus in the womb would not die even if the blood group of the foetus is A or B.
17. In a wrong blood transfusion, RBCs of donor agglutinate by the

antibodies of the recipient.

18. Study of antigen–antibody reactions in blood groups is called Serology.

Rh FACTOR

1. Rh factor was first of all reported in RBCs of *Macaca rhesus* (rhesus monkey) (CPMl 1987, 88,91) by **Landsteiner** and **Wiener** in 1 940 (AFMC 1992; MPPMT 1998).
2. Person with Rh factor is called Rh^+ and without Rh factor is Rh.
3. About 85% of white people are Rh positive and 15% Rh negative.
4. Percentage of Rh -positive people in India is 97% of the population (AFMC 2000).
5. Rh factor is foun d only in man and rhesus monkey; it is not reported from other animals.
6. Rh factor is dominant character in heredity (MPPMT 1988).
7. The disease **erythroblastosis foetalis** in human embryo is caused due to disadjustment of Rh factor (CPMT 1992).
8. Erythroblastosis foetalis can occur when father is Rh positive and mother is Rh negative (CPMT 1980,90; BHU 1995; CBSE 1990,93; AIIMS 1996).



9. An Rh negative women can be sensitized when she bears an Rh^+ child, and future Rh^+ children may have erythroblastosis foetalis (also called **haemolytic disease of the newborn** , HDN).
10. In developing foetus, erythroblastosis foetalis is caused by haemolysis (BHU 1997; AFMC 1997; AIIMS 2000).
11. Erythroblastosis foetalis is the death of foetus due to homolytic jaundice and anemia.

DIAGNOSTIC TECHNIQUES - BLOOD TESTS

1. A large number of techniques and instruments are available these days for the diagnosis and treatment of diseases. **Imaging techniques** are those that provide a pictorial or graphic representation of the internal parts of human body.
2. Human body contains a variety of fluids, of which blood is the most important one (CBSE 1999).
3. Blood is a fluid connective tissue and is pumped by the heart into blood vessels to reach all the body parts and back to the heart.

4. In order to assess the well being of the human body, the blood is subjected to a number of diagnostic tests like haemoglobin estimation, blood glucose, total leucocyte count (TLC), differential leucocyte count (DLC), erythrocyte sedimentation rate (ES R), lipid profile, blood urea, ELISA test and Widal test.

1. **Haemog1obin estimation:**
   (a) Haemoglobin (Hb) is the red -coloured pigment found in RBCs. Being an oxygen-carrier; it is vital for body functioning.
   (b) Normal range of Db
   In men = 15.5 1: 2.5 g/dl
   In women = 14.0 :t 2.5 g/dl
   In children (upto 12 years) = 11.01: 1.5 g/dl
   (c) Less Hb content than the normal range results in anaemia.
   (d) Hb estimation is commonly carried out by Sahil's Acid Haematin method.

2. **Blood Sugar Estimation:**
   (a) Abnormalities in blood suga r levels reflect disturbances in carbohydrates metabolism.
   (b) For estimation, the blood sample is collected in a tube containing a fluoride oxalate mixture.
   (c) Normal blood sugar range
   Blood sugar fasting = *90* - 120 mg/dl
   Blood sugar Post Prandial = upto 180 mg/dl
   (Post Prandial means after meals/breakfast)
   (d) Higher values of blood sugar fasting (hyperdycemia) indicate diabetes mellitus.

3. **Total leucocyte Count (TLC):**
   (a) Leucocyte or WBC count helps us to determine the status of infection in the body; the number of WBCs in blood increases beyond the normal value in case of infection; in same cases, TLC may even decrease.
   (b) Normal Range of TLC:
   (i) In new born = 18 ± 8 x 103/cμ.mm
   (ii) Infants. 1 yr = 12 ± 6 x 103/cμ.mm
   (iii) Children. 4-10 yrs. 11 ± 5 x $10^3$1cμ.mm
   (iv) Adults = 7.5 ± 3.5 x 10^3/cμ.mm
   (c) Increase in TLC (leucocytosis) occurs during acute infection, haemorrhage, burns and leukaemia.

(d) Decrease in TLC (leucopenia) is seen in measles, typhoid fever and agranulocytosis

4. **Differential Leucocyte Count (DLC):**
   (a) As leucocytes or WBCs are of different kinds (monocytes, lymphocytes, neutrophils basophils and easinophils), DLC is used to detect the number of different kinds of WBCs.
   (b) For this count, a blood film is prepared, stained with either Giemsa or Le ishman stain and the percentage of various kinds of WBCs calculated.
   (c) Normal DLC values:

(i) Monocytes	2 - 10%	(ii) Lymphocytes	20 - 45%
(iii) Neutrophils	40 - 75%	(iv) Basophils	0 - 1%
(v) Eosinophils	1 - 6%		

   (d) Increased eosinophil count indicates allergy.

5. **Erythrocytic Sedimentation Rate (ESR):**
   (a) Sedimentation rate of RBCs when uncoagulated blood is allowed to stand vertical for one hour is called ESR. The sediment at the bottom is measured under standard conditions.
   (b) ESR can be estimated either by Wi ntrobe method or Westergen method.
   (c) Normal ESR Values

	Wintrobe method	**Westergen method**
Men	0-9 mm in Ist hour	0-5 mm in Ist hour
Women	0-20 mm in Ist hour	0-7 mm in Ist hour

   (d) Rise in ESR is an indicator of diseases; it is high in pregnancy, tuberculosis, etc.

6. **Lipid Profile:**
   (a) In the blood, lipids being insoluble in water are transported in small particles bound to proteins, as lipoproteins.
   (b) There are five main types of lipoproteins – chylomicrons, very low density lipoproteins (VLDL), intermediate densi ty lipoproteins (IDL), low density lipoproteins (LDL), and high density lipoproteins (HDL).
   (c) Estimation of lipids is related to evaluation of coronary heart disease due to atherosclerosis. (In atherosclerosis, the lumen of arteries is reduced or narrowed du e to the deposition of cholestrol, and as result blood flow is reduced).
   (d) Important lipid profile tests include (i) Total lipids (ii) Serum total

cholestrol, (iii) Serum HDL cholestrol, and (iv) Total cholesterol /HDL – cholestrol ratio.

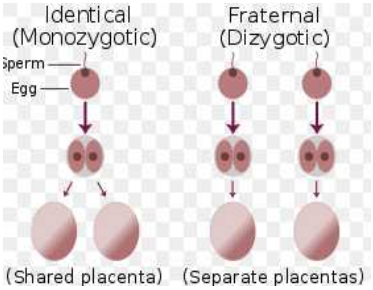
7. **Blood Urea Estimatation:**
   (a) Blood urea estimation is one of the kidney function tests.
   (b) Normal blood urea level is 30-40 ml/dl in adults.
   (c) Increase in blood urea is indicative of kidney disease.
   (d) Blood urea decreases during normal pregnancy.
8. **ELISA TEST** (For AIDS and other diseases):
   (a) The Elisa (Enzyme – linked immuno sorbent assay) test is used to detect various antigens or antibodies.
   (b) The test is used for diagnosing a number of diseases – AIDS (caused by HIV infection), hepatitis, STD (sexually transmitted diseases) and thyroid disorder.
9. **Widal Test** (For Typhoid):
   (a) It is a serological test used for the detection of typhoid (enteric fever).
   (b) The antibody formation induced by bacteria during infection, and the antibody level is tested after two weeks of infection by using commercially available antigen "H" or "O", and is measured as titre.
   (c) A titre of more than 1 : 20 of 'O' is significant, and rise in titres on repetition of test after one week will confirm the occurrence of typhoid.
10. **Principle involved in Sahil's Method for Hb estimation:**
    Haemoglobin is converted into acid haematin by hydrochloric acid. The brown colour of the compound, thus, developed is matched against a brown glass standard in a comparator (haemoglobinometer). The reading is in gram percent.
11. At room temperature glycolysis in the RBCs reduces the blood glucose at a rate of 5% every hour. Sodium fluoride is added to inhibit glycolysis.
12. **Rouleaux**: During ESR, RBCs sediment to the bottom while sedimenting, RBCs stack over each other, and such a stack is called rouleaux.
13. LDL and HDL are related to transport of cholesterol. LDL is a harmful lipoprotein, being related to coronary heart diseases, while HDL is the useful one as it removes cholesterol from plasma and cells and transports it to liver for processing or removal.

14. Higher levels of cholesterol (beyond 250 mg/dl) are associated with hardening of blood vessels (atherosclersis), kidney disease (nephrites), diabetes mellitus, obstruction jaundice and myxoedema.

MULTIPLE BIRTHS

1. Normally a women gives birth to only one young at a time; sometimes more than one child is born to a woman at the same time. These are called the cases of multiple births.
2. If there are two births, they are called twins.

Monozygotic Twins:



1. They are also called identical twins, the two blastomeres resulting from the first cleavage of zygotic completely separate from each other and develop into independent embryos.
2. Nature of identical twins is mono - zygotic. They are produced when on e sperm fertilizes an ovum and first two blastomeres separate from each other and become independent.
3. Monozygotic twins have same genetype and sex.

Dizygotic Twins:

1. These are also called fraternal or nonidentical twins, formed by simultaneous fertilization of two different ova by two different sperms.
2. When two eggs are fertilized and im planted, they produce dizygoticdichorionic twins.
3. Dizygotic twins vary genetically and may be of same or different sexes.

Conjoined Twins:

1. About 1 out of each 500 pairs of twins born will be joined together wholly or in part.
2. In conjoined twins, the separation of the twins is incomplete and these are born attached and remain so even after.
3. Conjoined twins are also called **'Siamese twins'.**

Freemartins:

1. Dizygotic twins are common in cattle like cow, sheep, goat etc. Sometimes the placentae of the two dizygotic twins fuse forming blood

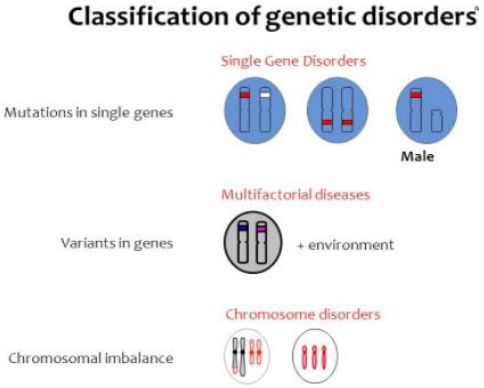
vascular connections between two developing foetuses.

2. The male hormone produced by the male twins suppresses differentiation of the female internal sex organs.
3. Such a sterile female with undeveloped ovaries, oviducts, uteri, etc. is called free-martin.
3. Freemartins are common in cattle (cow, sheep, goat and pig).
4. In freemartin conditions, female is sterile and male is normal.

GENETIC DISEASES IN MAN (Single Gene Disorders)

Albinism:

1. Albinism is a genetic trait and a hereditary disease.
2. Albinism is an autosomal recessive mutation, and reported from both white and black races.
3. A person suffering from albinism cannot synthesize melanin which provides black colouration to skin and hair.
4. Albinism is due to tyrosinase deficiency. The enzyme tyrosinase normally converts the amino acid tyrosine to melanin through an intermediate product DOPA (dihydroxy phenyl alanine).
5. Gene for albinism is epistatic to the genes for heavy pigmentation.



Sickle-cell Anaemia:

1. Sickle-cell anaemia is a genetic disease reported from negros.
2. Sickle cell anaemia is due to a molecular gene mutation of gene Hb which produces the β chain of adult haemoglobin.
3. A mutated gene Hb^S produces sickle-cell haemoglobin. The sixth amino acid in β chain of normal haemoglobin is glutamic acid. In sickle -cell haemoglobin this amino acid is replaced by valine.

A

4. The children homozygous (Hb^SHb^S) produce rigid chains. When oxygen level of the blood drops below a certain level, RBCs undergo sickling. Such cells do not transport oxygen e fficiently; they are removed by spleen causing severe anaemia.
5. Heterozygous people (Hb^AHb^S) generally do not have anaemia, they are said to have „sickle-cell trait'.

Thalassemia:

1. This is a human anaemia due to an autosomal mutant gene and when this gene is present in double dose, the disease is severe thalassemia major with death occurring in childhood.
2. The person suffering from thalassemia major are unable to produce β chain. Their haemoglobin contains δ chains like that of foetus. It is unable to carry out normal oxygen transporting function.

Alkaptonuria:

1. Alkaptonuria was discovered by Archibald Garrod in 1902.
2. Garrod was known as 'Father of Physiological Genetics' or 'Father of Biochemical Genetics'.
3. Patients of alkaptonuria excrete large amounts of homogentisic acid in urine. Such urine turns black upon exposure to light.
4. Alkaptonuria was the first of the recessive human traits discovered.

Phenylketonuria (PKU):

1. PKU results when there is a deficiency of the liver enzyme that converts phenylalanine into tyrosine.
2. PKU pat ients who are homozygous recessive lack the enzym e phenylalanine hydroxylase.
3. There is a high level of phenylalanine in their blood and tissue fluids.

Gaucher's Disease

4. Gaucher's disease is a genetic disease associated with abnormal fat metabolism.
5. It is caused by the absence of the enz yme glucocerebrosidase required for proper processing of lipids.

Galactosemia:

1. Galactosemia in man is inherited as an autosomal recessive, and the affected person is unable to convert galactose to glucose.
2. This disease is due to the deficiency of the enzyme galactose phosphate uridyl transferase (GTP).

Taste Blindness of PTC:

1. It is a genetic trait, not a disease, discovered by Fox in 1932.
2. PTC (phenyl thiocarbamide) is a compound of nitrogen, carbon and sulphur with sour taste.
3. The genotypes TT and Tt are tasters of PTC, while tt are non-tasters or tasteblind persons.

EUGENICS, EUTHENICS, EUPHENICS AND GENETIC ENGINEERING

1. The term eugenics (Gr. *eugenes,* well born) was coined by an English scientist Sir Francis Galton in 1883 (CPMT 1994).
2. Galton is called 'The Father of Eugenics' as this branch has been started by him.
3. Eugenics is the branch of science which deals with improvement of human race genetically (CPMT 198 4; CBSE 1990; Orissa JEE 1992; BHU1999).
4. Eugenics can be divided into two types: negative eugenics and positive eugenics.
5. Under negative eugenics, people with inferior and undesirable (dysgenic) traits are prevented from reproducing.
6. Sterilization of men i s done by vasectomy involving an operation in which" was deferens or sperm duct is blocked.
7. Sterilization of women is done by tubectomy. This involves an operation in which fallopian tube is blocked (DPMT 1996; CPMT 1998).

Euthenics:

1. Improvement of th e human race by improving the nurture (environmental conditions), i.e. by subjecting them to better nutrition, better unpolluted ecological conditions, better education and sufficient amount of medical facilities is known as euthenics (CBSE 1990; MPPMT 1995).
2. Eugenics is the science of being well-born and euthenics is the science of learning to live well.

Euphenics:

1. The symptomatic treatment of g enetic disease of man is called euphenics.
2. Euphenics deals with the control of several inherited human diseases, especially errors of metabolism.
3. Branch dealing with genetic engineering is euphenics (CPMT 1986).

Intelligence Quotient (I.Q):

1. The ratio between actual (chronological) age and mental age multiplied with 100 is known as I.Q.

$$IQ = \frac{\text{Mental Age}}{\text{Actual Age}} \times 100$$

2. Mental competence in relation to chronological age in man is called IQ (BHU 1997, CPMT 1998).
3. If the mental age of a 6-year-old child is 4, his mental age when he will be 15 years old should be 10 (CPMT 1987).
4. Different levels of IQ are:
   Idiot 0-24, Imbecile 25-49, Moron 50-69, Dull 70-79, Ordinary 80-89, Average 90-109, Superior 110-119, Most superior 120-139 and Genius 140 or more (CPMT 1987, 97).

GENETIC ENGINEERING:

1. Genetic engineering is the la test branch in applied genetics dealing the laboratory alteration of genes.
2. It is also known as biotechnology, recombination DNA technology or gene splicing.
3. Genetic engineering aims at adding, removing or repairing a part of genetic material.
4. Plasmids are rings of DNA occurring in bacteria additional to main genome (EAMCET 1996).
5. Plasmids are without vital genes but carry genes for sexuality, antibiotic resistance, etc. (MPPMT 1998). Bacteria can survive without plasmids.
6. Episome is a plasmid which ex ists in two states within the bacterial cell, either independently in the cytoplasm or following insertion, as an integral part of the host's chromosome.
7. Restriction endonuclease is used to cut the plasmid as well as the foreign DNA molecules at specific points (CBSE 1995; BHU 1995).
8. Two enzymes used in genetic engineering are restriction endonuclease and ligases.
9. Transduction is the transfer of bacterial genetic material from one bacterium to another using a phage as the vector (CPMT 1996).
10. Sexduction is the process in which a fragment of genetic material from one bacterium is carried with the sex-factor F to a second bacterium.
11. Genetic engineering permits clonhtg.

12. Genetic engineering, a field of biotechnology, is defined as a technique for artificially and deliberately modifying DNA or genes to suit human needs. In other words, it is a means of manipulating genes.
13. Genetic engineering is often referred to as "recombinant DNA technology".
14. Works of Stanley Cohen and Herbert Boyer in the mid 1970s invent ed and laid the foundation of genetic engineering.
15. Genetic engineering involves transfer of specific and desirable genes *from* one organism to another through the use of restriction enzymes (restriction endonucleases), gene cloning and appropriate vectors (like viruses and plasmids).
16. The technique of genetic engineering is being used in various ways:
    (a) To understand molecular events in biological processes,
    (b) To manufacture pharmaceutically important compounds,
    (c) To produce genetically modified organisms (GMOs or Transgenics), and
    (d) To remove genes responsible *for* 'causing hereditary diseases and replace them with normal genes (gene therapy).
17. Cloning refers to the production of an exact copy or copies of a single parent.
18. In nature, cloning occurs in bacteria (like E. coli) and protists (such as *Amoeba proteus*) which reproduce asexually by fission. In cloning, the organism or progeny produced is genetically identical to the parent organism.
19. Artificially, cloning has been used to produce "Dolly". Dolly is the clone of its mother sheep (the single parent).
20. Cloning may be cell cloning, gene cloning or organismal cloning.
21. Restriction enzymes are endonucleases which cut DNA at specific sites (acting as molecular scissors) into fragments containing identifiable genes.
22. Other enzymes used in recombinant DNA (or r-DNA) technology are DNA ligase which joins pieces of DNA molecules and DNA polymerase which synthesizes a new strand of DNA complementary to an existing DNA or RNA template.
23. Gene transfer can be mediated by two methods
    (a) Indirect methods through vectors or carries (like plasmids, viruses and *Agrobacterium)*
    (b) Direct vectorless metho d (through electroporation, chemical - mediated genetic transformation, microinjection and particle gun).

24. The bacterium *Agrobacteium tumefaciens,* is known as the "natural genetic engineer of plants." It contains a large Ti-plasmid, which can be used as a gene vector *for* delivering useful foreign genes into target plant tissue and cells.
25. The techniques of genetic engineering have permitted the production of a large number of genetically modified organisms (transgenics) with beneficial features.
26. Transgenic plants that are resistant to disease s, pests, herbicides, salinity and drought, or possess increased shelf -life of *fruits* (slow ripening) have been successfully produced.
27. Genetic engineering of microbes has been used to produce useful compounds like human insulin and penicillin.
28. Microbes have been engineered to reduce environmental pollution (bioremediation) *for* producing useful compounds such as enzymes and vitamins on a mass scale, and *for* a number of other purposes.
29. Gene transfer and cloning in animals, though more difficult than that in plants, has been used *for* 'producing vaccines *for* influenza, measles,mumps, rabies, rubella and now the genetic disorders in humans may even become curable.
30. Although the potentialities of using the technology are enormous and fantastic, they have triggered public concern raising ethical questions and sociological consequences.
31. **Genetic engineering-** Genetic engineering is a technique for artificially and deliberately modifying DNA or genes to suit human needs. OR It is the manipulation/engineering of genes and involves combining DNA *from* two different organisms to produce a recombinant DNA.
32. **Cloning-** Cloning refers to the production of an exact copy or copies of a single parent.
33. **Transgenic organism-** An organism in which a foreign gene is inserted (i.e., a genetically modified organism) is called a transgenic organism.
34. **Gene therapy-** The removal of defective genes responsible *for* causing hereditary diseases and their replacement with normal genes is called gene therapy.
35. **Totipotency-** The ability of an individual plant or animal cell or repeatedly divide and grow into a complete plant or animal in a defined nutrient medium under laboratory conditions is called totipotency.
36. **Cell cloning -** Obtaining a group of genetically identical cells by growing or culturing of a single cell is called cell cloning.

37. **Gene cloning-** Gene cloning is a technique to obtain iden tical copies (clone) of a particular DNA molecular (gene). Recombinant DNA technology is used.
38. **Restriction enzyme -** Restriction enzymes are endonucleases, which cut DNA at specific sites into fragments containing desirable genes. These enzymes, thus, act as molecular scissors.
39. **Restriction endonuclease -** Restriction endonuclease is an enzyme used in genetic engineering to cut DNA molecular at specific sites into fragments containing desirable genes.
40. **Plasmid-** In addition to the normal chromosom e in bacteria, the bacterial cytoplasm contains a circular DNA. This self-duplicating ring of additional DNA in the bacterial cytoplasm is called plasmid.
41. **Bacteriophage-** Viruses that infect bacteria are called bacteriophages.
42. **DNA ligase-** It is an enzyme used in recombinant DNA technology to join DNA molecules.
43. **DNA polymerage -** DNA polym erase is an enzyme which allows synthesis of a new stand of DNA molecule complementary to an already existing DNA or RNA template.
44. **Electroporation-** It is a method of vectorless transfer in which temporary holes are produced in the plasma membrane of the cell to allow entry of foreign DNA.
45. **Ti-Plasmid-** Ti-Plasmid is a tumour -inducing plasmid present in the bacterium, *Agrobaterium tumefaciens*. This plasmid acts as a vector for passing tumour-causing gene into the genome of the host plant.
46. **Microinjection-** It is a method of vectorless gene transfer in which foreign genes are introduced into plant or animal cells usoing micropipettes (glass needles).
47. **Particle gun -** In particle gun method of gene transfer, tungsten particles coated with foreign DNA are bombarded into target cells.
48. **'nif' genes-** These are nitrogen-fixing genes, which can be transferred into plants that are not able to fix atmospheric nitrogen, so as to reduce the dependency of plants on nitrogenous fertilizers.
49. **Hybridoma-** There are cells obtained from fusion of human cells and cells producing antibodies. Hybridoma technology is used in the production of monoclonal antibodies.
50. **Boiethics-** The application of genetic enginee ring and production of genetically modified organisms (trans genics) have led to a debate on the social, economical and environmental implications of this technology, and is referred to as bioethics.

51. **Vector-** In genetic engineering, a DNA molecule usually a plasmid or viral DNA, which is used to transfer genes into cells is called a vector.
52. Direct method of gene transfer is one in which vector is not used fortransferring gene. Gene transfer can be brought about b y any of the following methods: Electroporation, microinjection, particle gun or chemical-mediated genetic transfer. In direct method, gene transfer is brought through vectors or carries like plasmids, virus es, crown gall bacterium *(Agrobacterium tumefaciens).*
53. **Cell Cloning and Organismal Cloning:** Cloning of a single cell under experimental conditions on a suitable medium due to the property of totipotency, to produce a complete organism is called cell cloning. This is very successful in plant cells. Cell cloning is not possible to produce a fully differentiated organism in case of animals. However as has been done in case of "Dolly", a cloned sheep produced in the laboratory, it is possible to remove a somatic cell from the mother's udder and introduced it into the mothers enucleated egg. This egg can then be implanted into another female's uterus where it grows into a full -grown baby, as in "Dolly". Such a cloning is called organismal cloning.
54. ***Agrobacterium,* a pathogenic bacterium** , contains a tumour -inducing plasmid (Ti-plasmid). This bacterium passes on its Ti plasmid into the genome of the host plant on infection. As a result, galls are formed on the host plant. For this reason, *Agrobacterium,* is describe d as the natural genetic engineer of plants.
55. *Agrobacterium tumefaciens* is a pathogenic bacterium. It causes formation of tumours called crown galls in almost all dicotyledonous plants. The bacterium contains tumour -inducing plasmid called Ti - plasmid. On infection, the tumour -causing gene is passed into the genome of the host plant, resulting in the formation of galls. Thus, gene transfer occurs naturally without man's involvement. Now, however, Ti-plasmids are being used as gene vectors for delivering useful foreign genes into desired plants cells.
56. **Gene therapy** is the techniques of removal of defective genes responsible for causing hereditary diseases and replacing them with normal genes, so that the genetic disorders could be cured permanently. This has been made possible due to recombinant DNA technology technique. Through still its infancy, gene therapy in future may become a possibility for mankind.

57. **The various steps, in a sequential manner, in r-DNA technology are as follows**:
    (i) Cell culture of cells with the required DNA sequences is obtained through tissue culture technique.
    (ii) The enzyme restriction endonuclease is used to cut specific DNA at two places; the cut DNA piece is called restriction fragment.
    (iii) The same restriction endonuclease is u sed to cut a matching DNA sequence from a plasmid.
    (iv) The enzyme DNA ligase is used to join the restriction fragment (foreign DNA) with the cut DNA segment of the plasmid. The plasmid DNA containing foreign DNA is called recombinant DNA.
    (v) The recombinant plasm ids containing recombinant DNA are now inserted into a host cell (i.e., *E. coli)* by any of the gene transfer techniques like electroporation.
    (vi) Host bacterium divides to give multiple copies of recombinant DNA. The recombinant DNA is often preserved fo r future use in a DNA or gene library (also called genomic DNA library).

58. The technique of genetic engineering and the transgenic plants produced have been used in crop improvement in following ways:
    (a) **Producing herbicide resistance-** In soyabean, cotton and corn.
    (b) **Producing insect resistance** - In cotton, com, rice brinjal and tomato.
    (c) **Producing viral resistance-** In potato, tomato, tobacco and rice.
    (d) **Producing resistance against fungi-** In tomato and potato
    (e) **Producing vitamin-rich crops-** Vitamin A rich in rice.
    (f) **Increasing shelf -life of fruits (low ripening)** - In t omato(Flavr Slavr transgenic tomato have show fruit ripening).
    (g)Transferring 'nif' genes to enable non -leguminous plants to fix atmospheric nitrogen, there by reducing their depe ndence on Artificial fertilizers.

59. Gene Library: With the discovery of restriction enzymes and the development of methodologies to produce large quantities of DNA fragments containing genes through cloning into bacterial, yeast, plant or animal cells, it be came possible to develop a gene library of an organism. The gene library would contain almost all the genes of an organism.

    Significance

    (i) Genes of interest can be selected from the gene library and can be

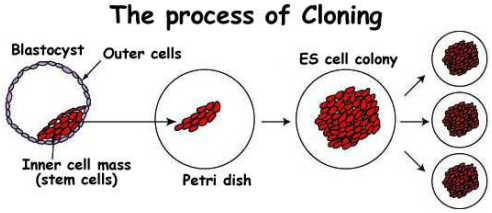
used for improving the characteristics of an organism.

(ii) The desired gene can be isolated in large quantities by growing the particular clone in large cultures. The isolated genes can be characterized by using techniques of genetic engineering.

(iii) Gene library in future may be used for curing hereditary diseases in man.

Cloning:

1. Clone is a population of cells or individuals, which are genetically identical (MPPMT 1998).
2. There are two types of cloning, gene cloning at molecular level and cloning of organisms. .
3. Cloning is meant for preservation of the genotype o f the organism (AFMC 1997).
4. Simian cloning (cloning of monkeys) was carried out by Don Wolf (USA) in 1996 *from* an eight-cell embryo.
5. Ian Wilmut of Roslin Institute, Edinburgh, and U.K. has produced as clone of adult lamb named "Dolly" (Feb 1997).



6. The fus ed cell developed into an embryo, which was planted into the uterus of another sheep, which acted as surrogate mother.
7. The Scottish scientists who cloned Dolly have now (July 1997) produced Molly and Polly, two lambs cloned with human gene for blood clotting Factor IX.
8. The milk of "Molly" and "Polly" contains Factor IX that can be extracted for use in treating human haemophilia.
9. **DNA fingerprinting:** Using DNA fragment lengths, resulting *from* restriction enzyme cleavage to identify particular individuals.
10. **DNA Probe:** A known sequence of DNA that is used to find complementary DNA strands.
11. **Gene therapy**: The use of bio-engineered cells or other bio-technology techniques to treat human genetic disorders.
12. Har Govind Khorana is associated with genetic engineering. He synthesized 'gene' artificially in a test tube (1969).

13. First protoplast fusion was done by Harries and Watkins of Oxford using somatic cells of mouse and man.
14. Hybridization by protoplast fusion is called parasexual hybridization.
15. Recent genetic analys is of Dolly's DNA has shown that she is "chimera", not a perfect clone.
16. Dolly has two genetic mothers as confirmed by the analysis of her mitochondria by Eric Schon and Ian Wilmut in 1999.

DNA FINGERPRINTING

1. The technique of DNA Fingerprinting was initia lly developed by the Alec Jeffreys.

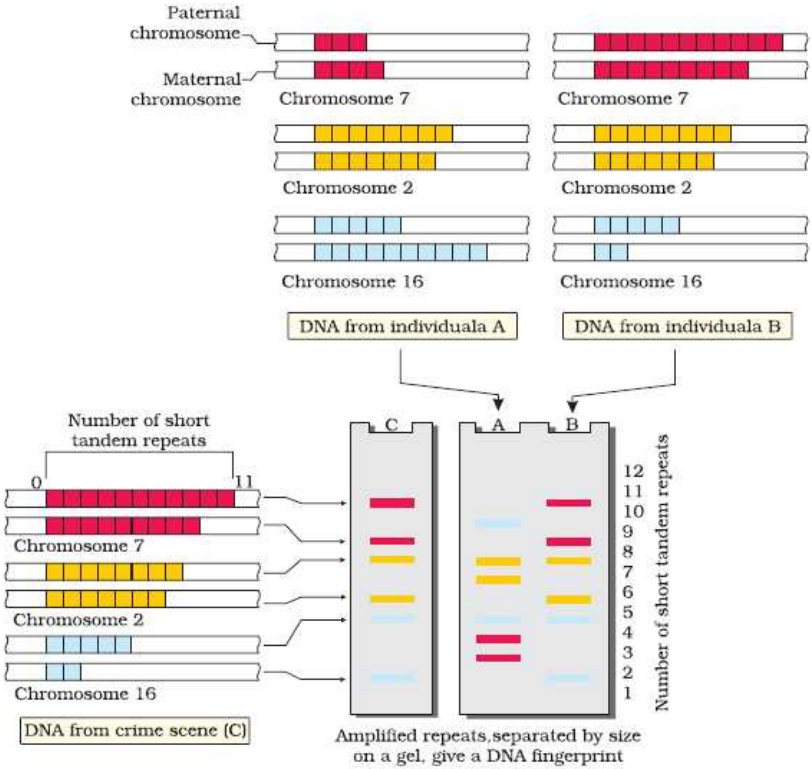


Figure *Schematic representation of DNA fingerprinting: Few representative chromosomes have been shown to contain different copy number of VNTR. For the sake of understanding different colour schemes have been used to trace the origin of each band in the gel. The two alleles (paternal and maternal) of a chromosome also contain different copy numbers of VNTR. It is clear that the banding pattern of DNA from crime scene matches with individual B, and not with A.*

2. He used a satellite DNA as probe that shows very high degree of polymorphism. It was called as **Variable Number of Tandem Repeats** (VNTR).
3. The technique, as used earlier, involved Southern blot hybridisation using radiolabelled VNTR as a probe. It included
   (i) isolation of DNA,
   (ii) digestion of DNA by restriction endonucleases,
   (iii) separation of DNA fragments by electrophoresis,
   (iv) transferring (blotting) of separated DNA fragments to synthetic membrances, such as nitrocellulose or nylon,
   (v) hybridisation using labled VNTR probe, and
   (vi) defection of hybridised DNA fragments by autoradiography. A schematic representation of DNA fingerprinting is shown in below given Figure.
4. The VNTR belongs to a class of satellite DNA r eferred to as minisatellite.
5. A small DNA sequence is arranged tandemly in many copy numbers. The copy number varies from chromosome to chromosome in an individual.
6. The numbers of repeat show very high degree of polymorphism. As a result the size of VNTR varies in size from 0.1 to 20 kb. Consequently, after hybridisation with VNTR probe, the autoradiogram gives many bands of differing sizes.
7. These bands give a characteristi c pattern for an individual DNA . It differs from individual to individual in population except in the case of monozygotic (identical) twins.
8. The sensitivity of the technique has been increased by use of polymerase chain reaction (PCR). Consequently, DNA from a single cell is enough to perform DNA fingerprinting analysis. In addition to application in forensic science, it has much wider application, such as in determining population and genetic diversities. Currently many different probes are used to generate DNA fingerprints.
9. DNA fingerprinting, also known as DNA typing or DNA profiling , is a technique of ascertaining relationships and identifying particular individuals using characteristics of their DNA.
10. The method of hybridization of DNA fragments with probe is called southern blotting, after the name of the inventor, E.M. Southern.
11. DNA fingerprinting is effectively used in forensic science for (i)

solving cases of paternity mix up and (ii) for identifying criminals such as murderers and rapists.

12. Blood, semen, hair bulb or any other body cells are used in DNA finger printing.
13. Just as the finger prints of an individual are very specific and no two humans (except monozygotic twins) possess the same fingerprint. Similarly, the DNA of every individual has distinctive characteristics and differences exist in the sequence of nucleot ides of different individuals.
14. Monozygotic (or identical) twins are formed from the division of the same zygote. Genetically, the two individuals, so formed, are identical in the c haracteristics of their DNA. Hence, the fingerprints are also identical.
15. A probe is a single-stranded nucleotide sequence that will hybridize or pair with a certain piece of DNA. The probe can be easily located as it is radioactive.
16. Polymerase Chain Reaction (PCR) is a technique whereby many copies of a piece of DNA or even a single gene can be made in the laboratory. Making many copies of DNA is called amplification, and PCR is used for it.

HUMAN GENOME PROJECT

In the preceding sections you have learnt that it is the sequences o f bases in DNA that determines the genetic information of a given organism. In other words, genetic make -up of an organism or an individual lies in the DNA sequences. If two individuals differ, then their DNA sequences should also be different, at least a t some places. These assumptions led to the quest of finding out the complete DNA sequence of human genome. With the establishment of genetic engineering techniques where it was possible to isolate and clone any piece of DNA and availability of simple and fast techniques for determining DNA sequences, a very ambitious project of sequencing human genome was launched in the year 1990.

Human Genome Project (HGP) was called a mega project. You can imagine the magnitude and the requirements for the project if w e simply define the aims of the project as follows:

Human genome is said to have approximately 3×10^9 bp, and if the cost of sequencing required in US $ 3 per bp (the estimated cost in the beginning), the total estimated cost of the project would be app roximately 9 billion US dollars. Further, if the obtained sequences were to be stored in typed form

in books, and if each page of the book contained 1000 letters and each book contained 1000 pages, then 3300 such books would be required to stored the information of DNA sequence from a single human cell. The enormous amount of data expected to be generated also necessitated the use of high speed computational devices for data storage and retrieval, and analysis. HGP was closely associated with the rapid deve lopment of a new area in biology called as **Bioinformatics.**

Goals of HGP

Some of the important goals of HGP were as follows :

(i) Identify all the approximately 20,000-25,000 genes in human DNA;

(ii) Determine the sequences of the 3 billion chemical bas e pairs that makeup human DNA;

(iii) Store this information in database;

(iv) Improve tools for data analysis;

(v) Transfer related technologies to other sectors, such as industries;

(vi) Address the ethical, legal and social issues (ELSI) that may ari se from the project.

The Human Genome Project was a 13 -year project coordinated by the U.S. Department of Energy and the National Institute of Health. During the early years of the HGP, the Wellcome Trust (U.K.) became a major partner; additional contribut ions came from Japan, France, Germany, China and others. The project was completed in 2003. Knowledge about the effects of DNA variations among individuals can lead to revolutionary new ways to diagnose, treat and someday prevent the thousands of disorders that affect human beings. Besides providing clues to understanding human biology, learning about non -human organisms DNA sequences can lead to an understanding of their natural capabilities that can be applied toward solving challenges in health care, agr iculture, energy production, environmental remediation. Many non -human model organisms, such as bacteria, yeast, *caenorhabditis elegans* (a free living non -pathogenic nematode), *drosophila* (the fruit fly), plants (rice and *Arabidopsis),* etc., have also been sequenced.

Methodologies : The methods involved two major approaches. One approach focused on identifying all the genes that expressed as RNA (referred to as **Expressed Sequence Tags** (ESTs). The other took the blind approach of simply sequencing the who le set of genome that contained all the coding and non-coding sequence, and later assigning different regions

in the sequence with functions (a term referred to as **sequence Annotation).** For sequencing, the total DNA from a cell is isolated and converted into random fragments of relatively smaller sizes (recall DNA is very long polymer, and there are technical limitations in sequencing very long pieces of DNA) and cloned in suitable host using specialised vectors. The cloning resulted into amplification o f each piece of DNA fragment so that it subsequently could be sequenced with ease. The commonly used hosts were bacteria and yeast, and the vectors were called as **BAC** (bacterial artificial chromosomes), and **YAC** (yeast artificial chromosomes).

The fragme nts were sequenced using automated DNA sequencers that worked on the principle of a method developed by Frederick Sanger. (Remember, Sanger is also credited for developing method for determination of ami no acid sequences in proteins). These sequences were then arranged based on some overlapping regions present in them. This required generation of overlapping fragments of sequencing. Alignment of these sequences was humanly not possible. Therefore, specialised computer based programs were developed (Figure 6.5).

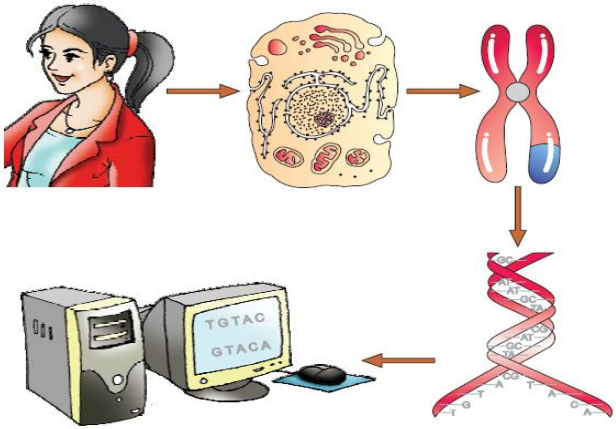


Figure 6.15 *A representative diagram of human genome project.*

These sequences were subsequently annotated and were assigned to each chromosome. The sequence of chromosome 1 was completed only in May 2006 (this was the last of the 24 human chromoso mes – 22 autosomes and X and Y – to be sequenced). Another challenging task was assigning the genetic and physical maps on the genome. This was generated using information on polymorphism of restriction endonuclease recognition sites,

and some repetitive DNA sequences known as microsatellites (one of the applications of polymorphism in repetitive DNA sequences shall be explained in next section of DNA fingerprinting).

6.9.1 Salient Features of Human Genome

Some of the salient observations drawn fro m human genome project are as follows:

(i) The human genome contains 3164.7 million nucleotide bases.
(ii) The average gene consists of 3000 bases, but sizes vary greatly, with the largest known human gene being dystrophin at 2.4 million bases.
(iii) The total number of genes is estimated at 30,000 -much lower than previous estimates of 80,000 to 1,40,000 genes. Almost all (99.9 per cent) nucleotide bases are exactly the same in all people.
(iv) The functions are unknown for over 50 per cent of discovered genes.
(v) Less than 2 per cent of genome codes for proteins.
(vi) Repeated sequences make up very large portion of the human genome.
(vii) Repetitive sequences are stretches of DNA sequences that are repeated many times, sometimes hundred to thousand times. They are thought to have not direct coding functions, but they shed light on chromosome structure, dynamics and evolution.
(viii) Chromosome I has most genes (2968), and the Y has the fewest (231).
(x) Scientists have identified about 1.4 million locations where single -base DNA differences (**SNPs – single nucleotide polymorphism,** pronounced as 'snips') occur in humans. This information promises to revolutionise the processes of finding chromosomal locations for disease-associated sequences and tracing human history.

6.9.2 Application and Future Challenges

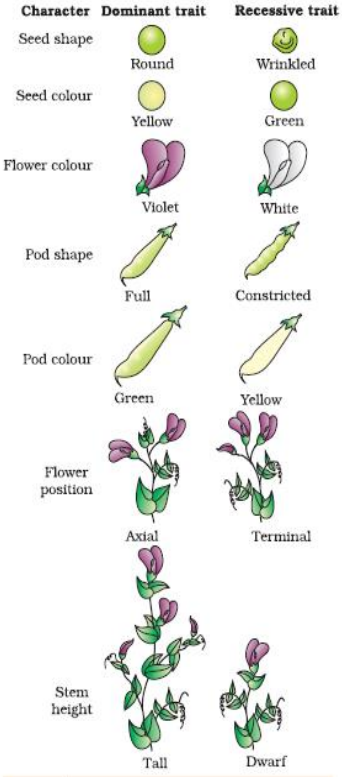
Deriving meaningful knowledge from the DNA sequences will define research thorough the coming decades leading to our understanding of biological systems. This enormous task will requ ire the expertise and creativity of ten thousands of scientists from varied discipline in both the public and private sectors worldwide. One of the greatest impacts of having the HG sequence may well be enabling a radically new approach to biological research. In the past, researches studied one or a few genes at a time. With whole –genome sequences and new high -throughput technologies, we can approach questions systematically and on a much broader scale. They can study all the genes in a genome, for example, all

the transcripts in a particular tissue or organ or tumor, or how tens of thousands of genes and proteins work together in interconnected networks to orchestrate the chemistry of life.

HEREDITY AND VARIATION

1. Heredity characters are transferred fr om parents to offspring through gametes.
2. The study of inheritance from one generation to another is known as ‘genetics’.

MENDAL’S LAWS OF INHERITANCE



3. Genetics is the science of heredity and variation (AFMC 1987).
4. The term 'genetics' was introduced by B ateson in 1905 (AMU 1988; CPMT 1994).
5. Gene is the unit of heredity and is equivalent to factor of Mendel. The term 'gene' was coined by Johannsen in 1909.
6. The various forms of a gene are called alleles (allelomorphs).
7. Alleles are alternate forms of a gene or a pair of contrasting characters.
8. The term 'allele' was coined by Johannsen in 1909.
9. An individual with two identical members of a pair of genetic factors is called homozygote.
10. Individuals having different alleles on homologous chromosomes *are* called heterozygotes.
11. Bateson and Saunders (1902) introduced the terms 'homozygote' and heteroztgote.
12. The genetic complement of the organism is known as 'genotype’.
13. Genotype is the type of hereditary properties of an individual.
14. Phenotype is the external appearanc e produced by the reaction of the organism of a given genotype.

15. The terms 'genotype' and 'phenotype' were introduced by Johannsen in 1909.
16. Offspring obtained from a cross between individuals having contrasting characters in the pure state is called hybrid.
17. Generation of hybrids obtained from a cross between two genetically different parents is termed first filial or F_1 generation.
18. Generation of individuals which is obtained from breeding of hybrids is called second filial generation or F_2 generation.

Seven pairs of contrasting traits in pea plant studied by Mendel

1. Gregor Johann Mendel (1822 -1884) is known as the 'Father of 'genetics'.
2. The first scientific study leading to formulations of laws of inheritance was carried out by Mendel (CPMT 1981,86; BHU 2001).
3. The first great geneticist was Mendel (CPMT 1984; JIPMER 1986; CBSE 1991).
4. Mendel was an Austrian monk who worked in 19th century (CPMT 1980; MPPMT 1999; JIPMER 2001).
5. Mendel was born in 1822 in Silisian, a village in Heizendorf, Austria (CPMT 1980, 93).
6. The first laws of heredity were given by Mendel (CPMT 1996).
7. Mendel work is related to principals of inheritance.
8. Before proposing his theory, Mendel knew nothing about chromosomes and genes (BHU 1983; AIIMS 2000).
9. Mendel had conducted his hybri disation experiments in garden pea *(Pisum sativum)* (CPMT 1988,90; BHU 1990; AFMC 1995).
10. Mendel is famous for his work on *Pisum* (CPMT 1980; CBSE 2000).
11. On the basis of his experiments, Mendel was able to explain fundamentals of inheritance.
12. Mendel derive d two laws or principles. The two important laws of heredity proposed by Mendel relate to segregation (purity of gametes) and independent assortment.
13. Mendel published his work in 1866 in a paper entitled 'Experiments in Plant Hybridisation', but other sci entists did not take Mendel's work seriously.
14. The year 1900 is highly significant for geneticists due to the rediscovery of Mendelism (BHU 1984; CPMT 1995).
15. Mendle`s work was published in journal proceeding of Brunn Natural History Society.

16. Mendal's law were rediscovered simultaneously by three great scientists namely Hugo de Vries (Holland), Erich von Tschermak (Austria) and Cart Correns (Germany) (BHU1989, 2001).

REASONS FOR THE SUCCESS OF MENDAL IN HIS EXPERIMENTS

1. Mendel selected peas for many reas ons. Pea plant is easy to grow and interbreed. The pea plant is self-fertilising in nature.
2. Mendel chose pea plants because they were having contrasting characters.
3. The number of characters studied by Mendel in pea plant was seven.
4. The number of chromosomes in *Pisum sativum* is 14 (2n).
5. Mendel restricted his experiments to one or few pairs of contrasting traits in each experiment.
6. Mendel also kept accurate quantitative records, a necessity in genetic experiments.
7. Self-fertilisation in pea can be prevented b y removing anthers (emasculation) before pollen grains mature (EAMCET 1996; CPMT 2000).

	AB	Ab	aB	ab
AB	AABB	AABb	AaBB	AaBb
Ab	AABb	AAbb	AaBb	Aabb
aB	AaBB	AaBb	aaBB	aaBb
ab	AaBb	Aabb	aaBb	aabb

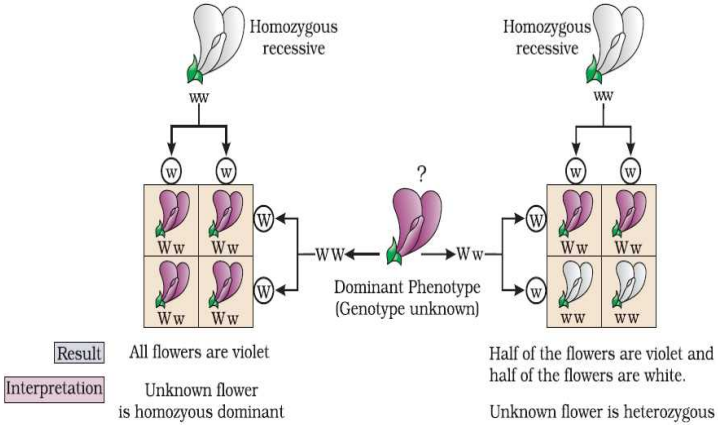
MONOHYBRID CROSS

1. Monohybrid cross is the most simple cross -performed by Mendel. It is the cross -made to study inheritance of a single character (trait). Phenotypic ratio 3 : 1 & Genotypic ratio 1 : 2 : 1 (CPMT 1998).
2. Mendel crossed two varieties of *Pisum sativum.* tall and dwarf. In the first generation (F_1), all plants were tall. Now he allowed F_1 plants to self-pollinate. The next generation (F_2) resulted in the production of tall and dwarf in the ratio of 3 : 1. So their phenotypic ratio was 3 : 1. But they had a genotypic ratio 1 : 2: 1 (1TT, 2Tt and 1tt). The reciprocal crosses gave similar results.

3. Tall plants produced in the F_2 generation may be of either TT or Tt genotype.

BACK CROSS AND TEST CROSS:

1. Back cross is a cross between F, hybrid and either parent.
2. Back cross to the recessive parent is known as test cross.
3. A cross of Fl with the recessive parent is known as test cross (Orissa JEE 1997; CPMT 2000; BHU 2001; JIPMER 2001).
4. A cross -used to ascertain whether a dominant phenotype is homozygous or heterozygous is termed test cross (CBSE 1991).
5. Tt x tt is a test cross (JIPMER 1984).
6. When tall homozygous is crossed with dwarf (test cross), all progenies will be tall.
7. Mendel proposed the existence of particulate unit factor for each trait. He suggested that these factors serve as basic units of heredity.
8. Factors are passed unchanged from generation to generation.
9. Back cross is also called Outcross.



Diagrammatic representation of a test cross

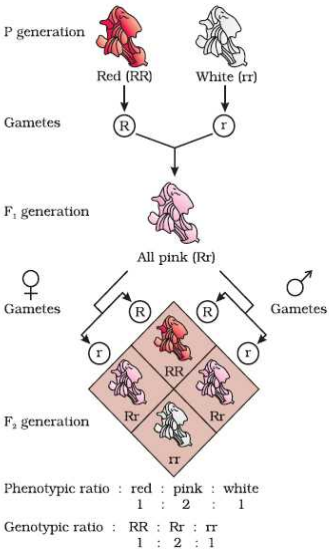
DOMINANCE-RECESSIVENESS

1. When two unlike unit factors are present in one individual, one unit factor is dominant to the other, which is said to be recessive.
2. When an allele fails to express itself in the presence of the other, the former is said to be recessive (CBSE 1991,99).
3. Mendel observed red flower in F_1 when he crossed red and white because of dominance (AFMC 1984; CBSE 1997).

4. A red flowered pea plant is crossed with the white flowered pea plant. F_1 is red. White flower colour is recessive (CPMT 1995).
5. Mendel formulated his first law, the law of segregation, with the help of monohybrid cross (BHU 1998).
6. Law of Dominance is a phenomenon rather than a law.

LAW OF SEGREGATION:

1. The hereditary characters are determined by particulate units or factors. These units occur in pairs in an individual, but in the formation of germ cells they become segregated so that only one memb er of a pair is transmitted through anyone gamete. This law is also called law of purity of gametes.



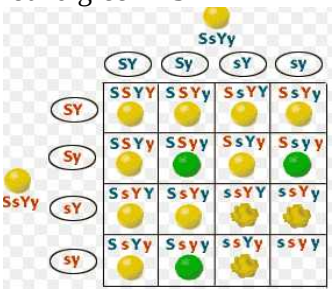
Result of monohybri d cross in the plant Snapdragon , where one allele is incompletely dominant over the other allele

1. The law of segregation of germinal units was formulated by Mendel.
2. Mendel's first law is known as law of segregation.
3. When 75% of the off springs have dominant character, the parents are both hybrids.

DIHYBRID CROSS

1. A cross between two individuals for studying inh eritance of two characters is known as dihybrid cross.

2. Mendel crossed a pea plant with round yellow seeds (RRYY) with plant having wrinkled green seeds (rryy). In F_1 generation all the plants produced only round yellow seeds (Rr Yy). This means round is dominant over wrinkled and yellow is dominant over green. The selfing of these plants produced four types of combinations in F_2. They are:
   Round yellow – 9 Wrinkled yellow – 3
   Round green – 3 Wrinkled green – 1



3. Thus the phenotypic ratio of F_2 in dihybrid cross is 9 : 3 : 3 : 1 (CPMT 1993; Karnataka CET 1993; CMEET 1996; MPPMT 1998; AMUI999).
4. Genotypic ratio of dihybrid cross is 1: 2 : 2 : 4 : 1 : 2 : 1 : 2 : 1 (EAMCET 1988; CPMT 1999).
5. Mendel's second law, the law *of* independent assortment is based on F_2 ratio of dihybrid cross (RPMT 1996; BHU 2000).

Laws of Independent Assortment:

1. The members of one pair of factors (alleles) segregate independently of the members of other pairs at the time of gametic formation (pair Rr segregates independently of pair Yy).
2. In the dihybrid cross, it has been found that colour of seeds is independent of shape of the seed. At the time of the gamete formation factor for found (R) or wrinkled (r) assort independently of yellow (Y) or green (y). It produces *four* types of gametes with two parental and two recombinations, *i.e.,* RY, Ry, rY and ry. Their fusion with opposite gamete results in the production of phenotypes in the ratio 9 : 3 : 3 : 1 in F_2 generation.
3. All seven characters or genes controlling seven traits in pea studied by Mendel were actually located on four chromosomes (CBSE 1997; BHU 1994).
4. Independent assortment is not applicable for the genes located on the same chromosome (linked genes).
5. In his experiments, Mendel di d not recognize linkage phenomenon because characters studied were located on different chromosomes, or the distance separating the syntenic loci was sufficiently great so that the genes so that the genes were inherited as though they were on separate chromosomes.

6. Genes that are present on the same chromosomes are called syntenic.
7. Mendelian recombinations were mainly due to independent assortment.
8. Linkage is an exception to Mendelian principles.
9. TtRr genotype represents a true dihybrid condition (CBSE 1991).

Test Cross: Two Characters:

1. Test cross is also applicable to dihybrid cross.
2. A cross between F_1 dihybrid and double recessive parent is an example of test cross.
3. When a tall red plant is crossed with dwarf white plant, all the plants of F_1 generation are found to be tall red. When a test cross is made the ratio will be 1:1:1:1(CPMT1986;BHU 2000; AIIMS 1998; AMU 1997).
4. A cross between F_1 heterozygous for two factors and recessive plant, recessive for both factors gives a phenotypic ratio of 1 : 1 : 1 : 1.

Trihybrid Cross:

1. Trihybrid cross is a cross involving three pairs of contrasting characters.
2. A trihybrid cross -involving height, seed shape and seed colour can be represented by TTRRYY x ttrryy.
3. A trihybrid in F_1 (TtRrYy) will produce eight types of gametes as TRY, TRy, TrY, Try, tRY, tRy. trY and try.
4. The F_2 phenotypic number in a trihybrid cross is $(2)^3 = 8$ and genotypjc number is $(3)^3 = 27$.

Miscellaneous Points:

1. Mendel quantitatively analysed his data. This proce dure different from those of his predecessor and contributed most to his success (CPMT 1988).
2. Johannsen (1903) introduced the term pure line which refers to series of self breeding generations having homozygous genotypes (BHU 1998).
3. Gibberellic acid is a compound extracted from the fungus *Gibberella fujikuroi.* If treated with gibberellic acid a dwarf plant can be made to grow to the normal height.
4. A dwarf pea plant is treated with gibberellic acid. It grows as a pure tall pea plant. If the treated plant i s crossed with a tall plant, then the phenotypic ratio of F_1 is likely to be all-tall (AIIMS 1980, 93)
5. Phenocopy refers to nonhereditary phenotypic modification caused by special environmental conditions. The term Phenocopy was introduced

by Goldschmidt in 1935 (CBSE 1998).

6. One genotype may produce two different phenotypes due to the difference in environment.

NON-MENDELIAN INHERITANCE

1. All patterns of inheritance could not be explained exclusively on the basis of Mendel's original principles alone and complexities were observed by later workers.
2. Some important examples of non -Mendelian inheritance are given below:

Incomplete Dominance:

1. In the snapdragon, *Antirrhinum majus* (CBSE 1991) or the four o'clock plant, *Mirabilis jalapa* (RPMT 1990), a c ross between varieties with red and white flower produces all pink in F_1 progeny (CPMT 1999).
2. The F_1 when selfed gives F_2 progenies which are red, pink and white in the ratio of 1 : 2 : 1.
3. The phenotypic and the genotypic ratios of F_2 are the same in the case of incomplete dominance (BHU 1995, 98).
4. In monohybrid crosses, absence of complete dominance is indicated by the F_1 plants that are intermediate and thus can be further confirmed if the phenotypic ratio in F_2 is 1: 2 : 1.
5. Due to incomplete dominance, a cross between blue and white Andalusian fowl results in 50% blue and 50% white fowls (BHU 1997).

Codominance:

1. In this inheritance, the heterozygote exhibits a mixture of the phenotypic characters of both homozygotes, instead of a single intermediate expression.
2. In shorthorn cattle, genes for red (r_1) and white (r_2) coat occur. Crosses between red (r_1r_1) and white (r_2r_2) produce (r_1r_2) roan.
3. Roan coat is due to a mixture of red hairs and white hairs, indicating the expression of both genes r_1 and r_2.
4. The human being of AB blood group is showing the phenotypic effect of both I^A and I^B codominant genes (CBSE 2001).

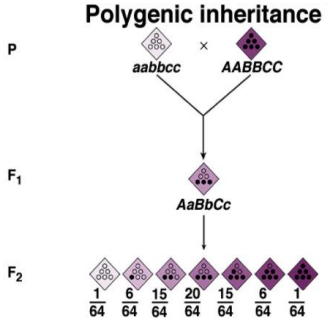
Multiple Alleles:

1. When any of three or more genes occupy the same locus in a given pair of homologous chromosomes they are said to constitute a series of multiple alleles (CBSE 2001).

2. Two examples of multiple alleles are: Coat colours in rabbits and A, B, O blood groups in man (JIPMER 2001, CBSE 2001).
3. If the number of alleles in a series is n, the number of genotypes will be n/2 (n+1). The number of genotypes of the coat colour in rabbit will be ten and blood groups in man is six.

Polygenic Inheritance:

1. A character whose expression is brought out by n umber of genes is called polygenic inhderitance or quantitative inheritance.
2. The expression is due to cumulative effect of the genes.



3. The important examples of polygenic inheritance are kernel colour in wheat and human skin colour.
4. Kolreuter (1760) is called the father of Polygenic (Quantitative) inheritance.
5. A Polygene is a gene that accounts for only a partial expression of a quantitative trait.
6. Only dominant polygene contribute to the expression of trait.
7. Number of phenotypes in polygenic inheritance are calculated as (2+n) +1, where n is the number of pairs of polygenes controlling the trait.

Kernel Colour in Wheat:

1. Two pairs of genes control the pigment production in the kernel of wheat.
2. The crosses of AABB (dark red) with aabb (white) produced intermediate red in F_1 generation.
3. The phenotypic ratio of 15 : 1 (red : white) is obtained in the F_2 generation.

Human Skin Colour:

1. Human skin colour is controlled by polygenic effect (BHU 1983; AFMC 1998; CBSE 2001) by at least three separate genes.
2. Each gene contributes to a unit of darkness due to incomplete dominance.
3. Skin colour is determined by cumulative genes, This hypothesis was designed by Davenport and Davenport in 1910.
4. Davenport designated five phenotypic classes controlled by two genes A and B (like Kernal colour in wheat).

5. Hughes (1944) recognized seven phenotypic classes, designating genes A, Band C.
6. A person with AaBbCc *(i.e.*. heterozygous for all three genes) will have an intermediate colour, mulatto. The number of possible allele combinations in the gametes is eight (ABC, ABc, AbC, aBC, abC, aBc, Abc, abc) for such person.
7. So total of64 phenotypic combinations are possible when two persons of similar genic combinations marry.
1. Other two examples of polygenic inheritance in man are: human height and eye colour.

Complementary Genes:

1. Bateson (1905) crossed two white coloured varieties of sweet pea, *Lathyrus odo ratus* (CCpp x ccPP), all the progeny (CcPp) were with purple flowers.
2. As one gene (C) complements the expression of the other (P), these genes are known as complementary genes.
3. A cross of purple CCPP and white ccpp gives all purple (CcPp) F 1 hybrid. Selfing the hybrid gives purple and white flowered progeny in the phenotypic ratio of 9 : 7 (instead of the normal 9: 3: 3: 1) (DPMT 1980; AMU 1989,93; CPMT 1998).
4. The F_2 digenetic ratio 9 : 7 is explained on the basis of complementary gene interaction.

Supplementary Factors:

1. Two pairs of supplementary genes interact in such a way that one dominant will produce its effect whether the other is present or not, while the second can produce its effect only in the presence of the first.
2. The F_2 digenetic ratio 9:3: 4 is explained on the basis of supplementary factors.

Collaborative Genes:

1. Each of the two dominant non -allelic genes shows independent expression but when present together, they interact to produce a new trait.
2. Modifier or collaborative supplementary genes in poultry give F_2 ratio walnut 9 : rose 3 : pea 3 : single 1.

Epistasis:

1. When one gene pair hides the effect of the other pair, the phenomenon is referred to as epistasis (AIIMS 1996; BHU 2000).

2. Epistasis implies one pair of genes can completely mask the expression of another pair of genes (AIIMS 1985; JIPMER 2001).
3. The gene that is masked is hypostatic.

Dominant Epistasis (12 : 3 : 1):

1. Among dogs, the colours of coat depend upon the action of two genes I and B.
2. The allele I prevents the expression of colour by B gene by producing white phenotype, iiBB and iiBb express black colour and iibb brown.

Recessive Epistasis:

1. Epistatic gene suppresses the expression of nonallelic gene only when it is in homozygous recessive condition.
2. Recessive epistasis produces a dihybrid ratio 9 : 3 : 4 sim ilar to supplementary factors.

Duplicate Genes (15: 1):

1. Two or more independent genes usually found on different chromosomes produce same or nearly similar phenotypes in dominant state.
   Example: Fruit shape in Shepherd's purse, *Capsella.*

Interaction of Genes

	Type of gene	Dihybrid ratio
1	Complementary genes	9 : 7
2	Supplementary factors	9 : 3 : 4
3	Collaborative genes	9 : 3 : 3 : 1
4	Dominant epistasis	12 : 3 : 1
5	Recessive epistasis	9 : 3 : 4
6	Duplicate genes	15 : 1

Lethal Gene:

1. A gene whose phenotypic effect kills the bearer. Death from different lethal genes may occur at any time from fertilization of egg to advanced age.
2. Lethal genes may be dominant, incompletely dominant or recessive.
3. Yellow Lethal in Mice: Cuenot (1904 -05) reported an incompletely dominant allele Y for yellow coat in mice.
4. Albinism in corn is due to lethal gene (BHU 1983).
5. Lethal gene disturbs phenotypic ratio from 3:1 to 0:2:1 or 2:1

Pleiotropy:

1. The ability of gene to have multiple effects is called 'pleiotropy' (CPMT 1999).
2. In the garden pea, the same gene co ntrols both flower and seed coat colour.
3. A famous example of pleiotropy is the hereditar)" disease called sickle-cell anaemia.
4. A person suffering from sickle -cell anaemia dies due to blocking of capillaries and tissue destruction.

VARIATION AND HEREDITY

- Bateson – Father of animal genetics.
- Morgan – Father of Experimental genetics
- Mendel Simply described his results and drew certain conclusions Correns gave these conclusions a shape of Laws.
- To find out results of different crosses following formulae are useful.
  a) Number of squares in Punnett's checker board = n^2 where n is the number of gametes.
  b) Type of Phenotypes and genotypes in F_1 are always one in any cross.
  c) In Incomplete dominance ratio of 1 : 2 : 1 is genotypically as well as phenotypically same.
- Various conclusions for F_2 generation can be obtained by following table

Number of trait n	Type of cross	Type of gametes $(2)^n$	Number of Zygotic combinations $(4)^n$	Phenotypes &Phenotypic Ratio $(2)^n$	Genotypes & Genotypic Ratio $(3)^n$
1.	Monohybrid cross	$(2)^1 = 2$	$(4)^1 = 4$	$(2)^1 = 2$ (3 : 1)	$(3)^1 = 3$ (1 : 2 : 1)
2.	Dihybrid cross	$(2)^1 = 4$	$(4)^2 = 16$	$(2)^2 = 4$ (9 : 3 : 3 : 1)	$(3)^2 = 9$ (1 : 2 : 1 : 2 : 4 : 2 : 1 : 2 : 1)
3.	Trihybrid cross	$(2)^3 = 8$	$(4)^3 = 64$	$(2)^3 = 8$ (27 : 9 : 9 : 9 : 3 : 3 : 3 : 1)	$(3)^3 = 27$ (1:2:1:2:4:2:1:2:1: 2:4:2:4:8:4:2:4:2: 1:2:1:2:4:2:1:2:1)

- In a Dihybrid cross, if one trait is showing incomplete dominance and other trait follows law of dominance then the ratio is 3 : 6 : 3 : 1 : 2: 1.
- When both the traits in a dihybrid cross show incomplete dominance the ratio will be 1 : 2 : 1 : 2 : 4 : 2 : 1 : 2 : 1.

Genes and Chromosomes: Emerson and East studied the cob length in maize.

1. A change in gene pool with an environmental gradient is called *clin.*
2. Gene is definable only by operational terms.
3. Gene can be defined as a specific length of double stranded DNA having about 500-2000 base pairs.
4. Concept of gene was given by *Sutton.*
5. Gene is a stable discrete unit of information.
6. Gene controls heredity and protein synthesis.
7. Movement of genes as a result of mating and gene exchange within population is called **gene flow**.
8. Chromosomes are passed from parents to offsprings.
9. Superiority of hybrid over its parents is called hybrid vigour (**Heterosis**) it is maximum in Maize. It was given by **schull.**
10. Inheritance shown by Mendel in pea is monogenic inheritance.
11. Height and colour show both monogenic and polygenic inheritance.
12. Gene of Red flower was considered by Mendel as monogenic though it is a polygene.
13. A gene which can occur in more than two al ternative forms present on the some locus is called **multiple allele**.
14. When heredity trait is controlled by one gene that has two alternative forms, it is called (monogenic trait) ***Qualitative trait.***
15. When a heredity trait is controlled by more than one non-allelic gene, it is called *Polygenic trait* or **continuous metric trait** or **Quantitative trait**.
16. Geometrid moth, Adder's Tongue Fern and Amoeba are polyploid.
17. In plants **maximum number** of chromosomes 2n = 1262 are found in ***Ophioglossum reticulatum***.
18. Symptoms repre senting a particular disease due to chromosomal abnormalities are referred to as ***syndrome.***
19. C-bands occur in chromosome regions during chromosome staining these contain **Heterochromatin.**
20. According to the **Denver** (Colorado) convention (1960) the autosomal chromosomes are divided into seven groups(A,B,C,D,E,F,G)according to the size of chromosomes and position of centromere.
21. **Acrocentric chromosomes** are placed in group D.
22. Denver system has maximum number of chromsomess in group C.

23. In the Denver system sex chromosomes are placed in no group.
24. On the basis of centromere, Drosophila has four types of chromosomes, *Trillium* has only metacentric chromosomes and man has three types of chromosomes.
25. In human karyotype both D and G group has SAT chromosomes.
26. *Chiasmata* are formed by exchange between non -sister chromatids of homologous chromosomes. These were reported by ***Janssens*.**
27. Unit of distance between genes in a chromosome is **centimorgan.**
28. Term centimorgan is used in eukaryotic genetics and map unit or morgan in a microbial genetics.
29. **Karyotype** is the pictorial or photographic representation of all autosomes and **allosomes** of a somatic diploid cell at mitotic metaphage stage.
30. **WBC** are taken to make karyotype.
31. When karyotype is represented diagrammatically and arranged homologous pairs in order of decreasing length. it is called *Idiogram.*
32. Chromosomes related with sex determination are known as **Heterosomes or Allosomes or Idiosomes.**
33. Concept of gene mapping was first suggested by *sturtvant.*
34. The chromosomal theory of heredity me ans that genes are located on the chromosomes.
35. To make a karyotype, chromosomes are photographed during mitosis.
36. There is a single DNA duplex, which is highly coiled and folded in chromosomes.
37. **Karyotype** means arranging chromosomes along with their homologous pairs.
38. The maximum number of chromosomes in a flowering plant is 2n= 265 in *Poa litorosa.*
39. The minimum number of chromosome in an angiospermic plant is (2n = 4) in ***Haplopappus gracilis***.
40. New characteristics can be produced in organisms by subjecting them to sublethal radiations and selection.
41. In haploids both recessive and dominant mutations express themselves because there is one allele for each trait.
42. In human males, even some recessive sex linked alleles are able to express their effect because there is only one X chromosome.
43. Drosophila is used in the study of genetics because-
    a) Single mating produces over 100 offspring's.

b) Generation period is 10-20 days.
c) Chromosome complement is simple with three pairs of autosomes and XY sex chromosomes where Y is hooked.

44. Phenomenon of linked genes being inherited together for two or more generations is known as *complete linkage.*
45. One way of determining sex-linked inheritance is son resembles mother and daughter resembles father.
46. A child receives genes from his father to the extent of 50%.
47. Sex of human child is due to Y – chromosomes.
48. Maize has 10 pairs of chromosomes. It would have 10 linkage groups.
49. Exchange of chromosome segments between maternal and paternal chromatids during meiosis is called **crossing over.**
50. Crossing over brings about recombination of genes.
51. The crossing over between homologous chromosomes never exceeds beyond 50%.
52. Number of linkage group in Pea are 7, in maize 10 and in Ascaris is 1.
53. The numbers of linkage groups are equal to number of haploid s ets of chromosomes.
54. If an organism has 2n = 40 chromosomes the linkage groups will be 20.
55. In XX – XY type of organisms, number of linkage groups will be one more in male than in the female as in male X and Y both are different.
56. Number of linkage groups in human female is 23 and in human male is 24.
57. In linkage two pairs of traits in coupling phase linkage will be AABB x aabb. (CBSE 1993)
58. According to chromosome theory of linkage of **Morgan and Castle** (1912)-
    a) Genes lie in a linear order in the chromosome.
    b) Strength of linkage between two successive genes is inversely proportional to the distance between two genes.
    c) Linked genes are arranged in **CIS or TRANS** Manner
    d) CIS arrangement  brings coupling that is parental combinations.
    e) Trans arrangement  brings recombination's that is repulsion.
59. Traits controlled by genes located on X -chromosomes are called sex linked.
60. When body of an organism shows both genetically male and female tissue. It is called Gynanadromorph.

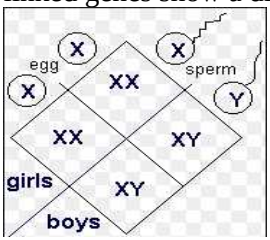
61. Gynandromorphism is developed due to non -disjunction of X -chromosomesorAneuploidy
62. Male or female gynanders with patches of tissues of other sex are called sex pie bolds or chimera.
63. First chromosomal map was prepared for X -chromosomes in *Drosophila* by ***Sturtevent***.
64. Chromosomal maps show linear sequence and relative distance between genes. First chromosome map of a plant was of maize, prepared by *Emerson.*
65. Crossing ov er is nil between genes located near Telomere and centromere.
66. In Bugs, Cockroach and round worms the male possess one chromosome less than the female (XO male).
67. An animal where the male has half the number of chromosomes as compared to the female is *Honeybee.*
68. Females are heterogametic amongst Birds.
69. Sex is determined by environment in Bonellia.
70. Morgan (1910) proved that genes are located on chromosomes.

Sex determination:

1. XX-XY is the type of sex determination in which Y determines sex of maleness.
2. XX-XO type, in which O determines sex of maleness as in bugs, cockroach, grasshopper, round worm.
3. ZZ-ZW type, W determines sex of femaleness as in Birds, reptiles, fish and silkworm.
4. ZO-ZZ type, O determines sex of femaleness and it is opposite to XX -XO type found in Butterflies, moth, pigeon and duck.
5. Haplo-diploidy: males haploid & females d iploid in ants, bees and wasps.Males develop by parthenogenic development of unfertilised egg.
6. Sex determination in plants is X-Y, XO and Z-W type.
7. Nutritional theory of sex determination was given by *Schenk.*
8. Genic Balance Theory of sex determination by Bridges 1925.
9. Quantitative theory of sex determination by *Goldschmid.*
10. Harmonal theory of sex determination by Lillie.
11. Theory of metabolic differentiation by Riddle.
12. A par allelism of behaviour between chromosomes and Mendelian factors was suggested by **Sutton and Boveri (1902)**.
13. When two genes are located very close to each other on the same

chromosome hardly any crossing over takes place.

14. 'Col' Plasmids in Bacteria produce bacteriocidal and bacteriostatic chemicals.
15. Chromosomes are vehicles of heredity. It was stated by **Sutton**.
16. A dihybrid test cross ratio for two linked genes in a hybrid is 1 : 1.
17. The coupling and repulsion theory of Bateson and Punnet t later on modified linkage and crossing over by Morgan. Two completely linked genes show a dihybrid ratio of 3 : 1.



18. An organism heterozygous for two pairs of genes (AaBb) undergoes meiosis. The possible genotypic combinations in gametes are AB : Ab : aB : ab.
19. The unit Morgan or centimorgan or morganoid is related to linkage map, chromosome map and genetic map.

<u>In Neurospora Genetics</u>:

1. In Neurospora ascus, 8 ascospores show 2A, 4A, 2a arrangement it suggests II division segregation.
2. In the ascus of *Neurospora cross* the number of recombinants is directly proportional to the distance of gene from its centromere.
3. If in Neurospora, the progenies of the cross between an orange colonial strain and Albino spreading strain segregated into 1:1:1:1 ratio of albino spreading: orange colonial: Normal : Albino colonial, then the inference is that the traits are controlled by 4 genes.
4. A clone is a collection of genetically identical individuals produced by vegetative propagation.
5. Crossing over occurs at four str anded stage this was proved by the observation that usually two gametes resulting from meiosis are recombinants.
6. Assuming no linkage and no crossing over, segregation of Mendelian factors during meiosis occurs at Anaphase I.
7. In *Neurospora crossa* (pink brea d mould) Genotype Aa is used for a study of segregation of 8 ascaspores in ascus, aaaa AAAA ordered arrangement does not represent second division segregation (1993).
8. In Neurospora and Sordaria (ascomy cetes) the ascospores in the ascus are arranged as either 8 or 4 ascospores but always in a linear order.
9. Tetrad analysis started by *Dodge* in Neurospora is helpful in knowing recombination frequencies.

10. In haploid organisms for one gene pair after fertilization two contrasting forms of a hybrid trait segr egate during zygotic meiosis to give a ratio of 1 : 1.
11. Same haploid organism for two gene pairs will show a ratio of other than 9 : 3 : 3 : 1 is 1 : 1 : 1 : 1.
12. A Diploid with only one allele (man is for XY chromosomes) is known as *Hemizygous.*
13. Sons of a man have 0% chances to inherit a rare X-linked disorder from their father.
14. Drosophila has four pairs of chromosomes, male drosophila has 5 linkage groups while female has 4 linkage groups.
15. Bateson and Punnett discovered that in the cases involving linked genes the assortment of genes during germ cell formation is not random in violation of Mendel`s second law.
16. Occasional separation of two genes on the same chromosomes by a recombinant event is a case of incomplete linkage.
17. The pre sent concept of linkage and crossing over is based on the presumption that genes are arranged in a chromosome in linear fashion.
18. Infrared rays do not result in any mutational change.
19. A wild type strain of microorganism which is capable of growth on a defined minimal essential medium (MEM) is termed as *prototroph.*
20. Evidences that crossing over occurs at four stranded stage and not at two stranded stage of chromosomes by the studies of Lindegren (1933) and Dodge (1929) fr om 2 : 2 :2 : 2 arrangement of ascospores in Neurospora.
21. According to Riddles theory of metab olic differentiation males have high metabolic ratio over females.
22. **Giemsa** is related with chromosomes banding.
23. **Quinacrine** mustard stains region rich in A-T Bases.
24. A haemophilic man marries a normal homozygous women produce 0% haemophilic son and daughter.
25. Haematoxylin, a red nuclear stain is obtained from the heart wood of *Haematoxylon campechianum.*
26. Carmine dye is obtained from cochneal insects.
27. Some persons experience phenyl thio carbamide as bitter. Others find it tasteless. It is a heredity character. T stands for dominant allele and t for recessive one.

28. A normal woman whose father was haemophilic marries a normal man. She will produce some sons haemophilic.
29. A colour blind woman X „X „ marries a normal man the progeny shall be carrier normal daughters and colour blind sons.
30. Haemophilia hardly occurs in females as they have two X-chromosomes.
31. **Acromegaly** is a Harmonal disease.
32. Night blindness in man is not a wholly genetic trait.
33. A recessive gene expresses only in homozygous condition. Sometimes, a single recessive gene can express itself when it is located on X-chromosome of male.
34. Baldness is more common in man than in women because genes of baldness are autosomal but influenced by androgens.
35. When sex is determined before fertilization it is called **progamic.**
36. Sex linked genes are generally located on X-chromosomes determination of sex of child depends upon nature of sperm.
37. Sex is determined soon after fertilization.
38. When a man with hairy ears marries a normal woman the percentage of their sons would be 100% to have hairy ear.
39. A colour blind daughter will be born when both parents are colour blind or mother is carrier and father is colour blind.
40. If both parents are normal the male child may be colour blind if woman`s father was colour blind.
41. Two dominant non -allelic genes are exactly 50 map units apart. It means that linkage is Transtype.(CBSE 1993)
42. Disease caused by pleiotropic genes is **a syndrome.**

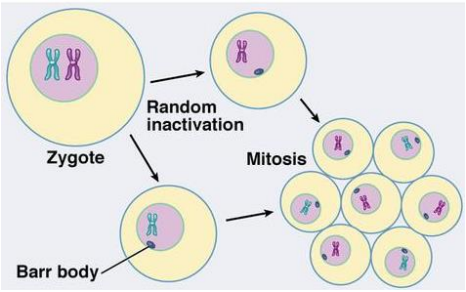
Colour Blindness:

1. Simple tests for colour blindness are conducted by using colour cards called **Ishihara cards.**
2. **Congenital colour blindness** is called **Dichromatism** or **Daltonism** as it was reported in Dalton.
3. Red colour blindness is called **Protanopia**.
4. Green colour blindness is called **Deutaranopia.**
5. Blue colour blindness is called **Tritanopia.**

Barr Body:

1. Barr and Bertram (1949) found a small dark stained chromatin body adhered to the nuclear membrane of nerve cells of female cat.

2. Lyon (1962) suggested that one X chromosomes of maternal side become coiled and inert and heteropycnotic and forms a barr body.
3. Number of borr bodies is always one less than the total number of X chromosomes in female. Normal male has no barr body.
4. Barr bodies can be present in both sexes but X spot are found only in males.
5. Neutrophils are examined for drumsticks for female sex determination.
6. Sex of fo etus is determined by examining cells from amniotic fluid looking for barr bodies.



Induced Mutation:

1. Muller is called the father of actinobiology: effect of radiations to induce mutations.
2. Muller was first to use X-rays to induce mutations in fruitfly.
3. Altenberg used UV rays and gamma rays as mutagens.
4. Stadler used X -rays to induce mutations in Barley and Maize. Auerbach and Robson used mustard gas as first chemical mutagen.
5. Gamma rays generated by cobalt^{60} in *Gamma Garden* are used in plants to induce mutations.
6. Sudden change in pH and temperature cause breaking of sugar - PO_4 bonds.

Hybrids:

Fairchild Mule (Sweet william (Centauria) x Carnation (Dianthus) plant by Fairchild.

Mule – Male donkey x female Horse
Hinny (Pony) Male horse x Female donkey
Zebronky (Zebra x donkey)
Liger (Male lion x Female tiger)
Tigon (Male Tiger x Lioness)

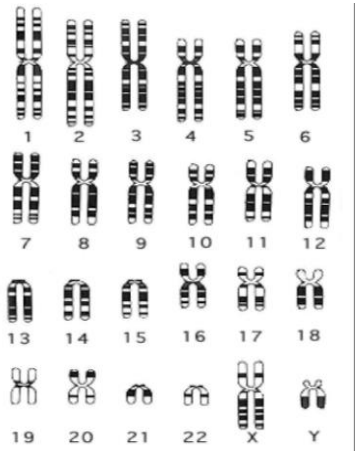
Palindrome DNA: It is a DNA with two fold rotational symmetry. DNA dublex have some sites where the sequence of 3 – 10 Nucleotides is the same but oppo site in the two strands. These areas transcribe same type of RNA and widely used in genetic engineering. It is like-

Repetitive DNA or DNA Finger Print or Satellite DNA: It is found in Eukaryotes only.

- It consists of a sequence of one or few nitrogen base s repeated 16 -64 times in tandem.
- Such areas are found near centromere and have unique and constant sequence specific for every species/organisms.

CHROMOSOME MAPPING:

1. Variation in recombination frequency is governed by the distance between the genes. Clos er the two genes lesser is the chance of cross over.
2. A chromosome map is a graphic representation of a linkage group in the form of a line, which shows the sequence of genes and the relative distances between genes.
3. A chromosome map is also called a **Linkage Map or Genetic Map**.
4. Alfred H. Sturtevant suggested that the frequency of recombination can be used to suggest the relative distance between the genes on the chromosomes and a map of linked gene can be produced.
5. Method of preparing chromosome maps of a sp ecies is called chromosome mapping.
6. Chromosome mapping is based on two genetic principles-
   a) The genes are arranged in a linear order in the chromosome.
   b) The frequency of crossing over between two genes is directly proportional to the distance between them in the chromosomes.
7. Frequency of crossing over is used in preparing chromosome maps due to this chromosome maps are also called **Cross Over Maps.**
8. A frequency of one percent crossing over between two genes is taken to represent one unit of map distance between these two genes.
9. Unit to distance in chromosome map is **Morgan.**
10. Frequencies of crossing over or recombination between three genes: yellow body (y) white eye (w) and miniature wing (m) are as follows:
    a) Yellow body-White eye - 1.5%
    b) White eye-Miniature wing - 34.5%
    c) Yellow body-Miniature wing - 36.1%



11. Sturtevant suggested that if the genes are linearly arranged the distances between them should be additive.
12. According to the above conclusion the sequence of three genes should be (y,w,m).
13. 1 per cent recombination frequency is taken as equivalent to one map unit distance and referred to as centimorgan *(CM)*.
14. The map distance between y and m is 36.1 percent units which is the sum of the map distances 1.5 + 34.5 = 36.
15. In most real case s map distances do no add up perfectly because of double cross over.
16. Morgan and Sturtevant got recombination frequencies-
    a) 18 between genes b and Vg b) 9 between b and cn
    b) 9.5 between cn and Vg

CHROMOSOME MAPPING

1. Sequence of genes on the chromosome will be b, cn, Vg.
2. In offspring we can observe frequency of recombinants and not the frequency of crossing over.
3. Frequency of crossing over will be slightly higher than the observed frequency of recombinants.
4. Degree of intensity with which two genes are linked together is called as **Linkage Value**
5. Chromosome maps were first prepared for Drosophila and Maize.
6. First chromosome map was prepared for X-chromosome in *Drosophila* by **Sturtevant**
7. Chromosome maps of certain viruses and for the circular DNA of some bacteria have also been prepared.
   One map unit corresponds to the physical length of the chromosome in which one cross over occurs **in every 50 meiosis**.
8. If the percentage of crossing over between two genes is 10 the distance between these two genes will be 10 morgan.
9. Process by which frequency of crossing over is increased is called **Coincidence.**

10. Interference is decrease in frequency of crossing over because chiasma at one point disfavours formation of chiasma at second point.

- **Importance of Chromosome Maps**:

1. Gives **exact locations** of genes in a chromosome.
2. Indicates which genes lie in which chromosomes.
3. Indicates the strength of linkage and changes of crossing over betwee n two genes.
4. Help in predicting the results of dihybrid and trihybrid crosses.
5. Chromosome maps confirm that
   a) Genes have linear arrangement.
   b) Genes have definite positions called **loci.**
   c) Specific genes are located in specific chromosomes.
6. Helping predicting loss of a chromosome segment.
7. Assists in judging the site on a chromosome for genetic manipulation by surgery.

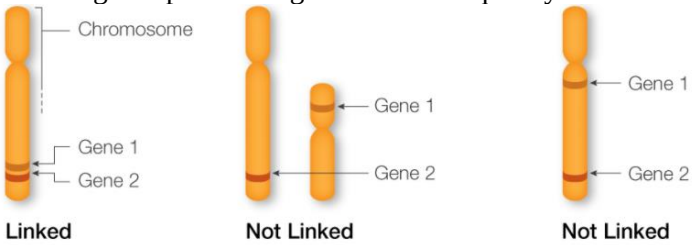
LINKAGE:

1. Tendency of the genes present in the same chromosome to stay together in hereditary transmission is known as **linkage**.
2. The genes located in the same chromosome are called **linked genes**.
3. Genes present on different chromosomes are termed **unlinked genes.**
4. The red eye colour and gray body colour in *drosophila melanogaster* lie in the same chromosomes and therefore go together in the gamete and the nest generation.
5. Linkage is an exception to Mendel's law.
6. The first attempt to show linkage in plants was done in *lathyrus odoratus*.
7. A pair of genes is linked if their recombination frequency in test cross is lower than 50%.
8. Unlinked genes assort independe ntly give a dihybrid ratio of 9:3:3:1 and show a test cross ratio of 1:1:1:1.
9. **Bateson sounder and Punnet** (1905) working with *lathyrus odoratus* (sweet pea) were the first to discover linkage an exception to the low of independent assortment.
10. **Morgan and cast le** formulated the **chromosomes theory of linkage** according to which-
    (a) Genes lie in linear order.
    (b) Linked genes are located in the same chromosomes.
    (c) Genes tend to stay in parental combination except for crossingover.

(d) The distance between the linked genes in the chromosomes determines the strength of linkage.

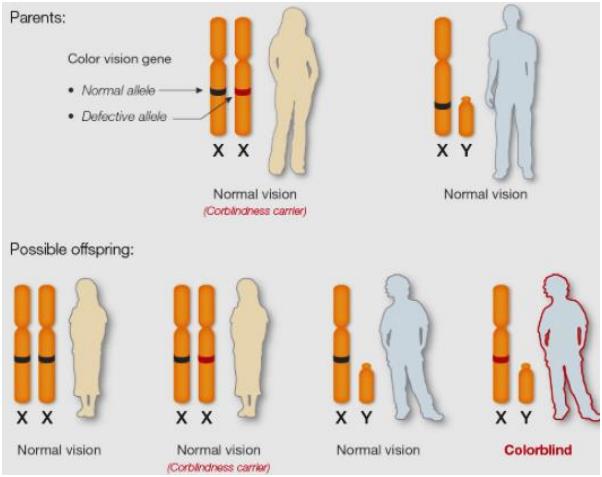
11. If two alleles A and B come from the same parent (AABB x aabb) than they will enter gametes and transmit together this is called **coupling.**
12. When the same alleles (A and B) come from different parents (AAbb x aaBB) then they will enter in to different gametes this is known as **repulsion**.
13. Morgan (1910) discovered that **coupling** and **repulsion** are two aspects of the same phenomenon **linkage.**
14. Closely located genes have no chance of separation by crossing over and are always transmitted together to the same gamete and the same offspring. This phenomenon is called **complete linkage.**
15. When female *drosophila* homozygous for red eyes and normal wings (a wild type) is crossed with a double recessive male *drosophila* homozygous for purple eyes and vestigial wings.
16. F_1 flies are all heterozygous red-eyes and normal winged.
17. When F_1 dihybrid female fly is test crossed with a double recessive male having purple eyes and vestigial wings the F_2 flies are of two types red-eyed, normal winged and purple eyed, vestigial winged in the ratio of 1:1.
18. No recombinant types are formed because of complete linkage and no crossing over.
19. Complete linkage is found in male *Drosophila*.
20. Distantly located genes have a chance of separation by crossing over and of going in to different gametes and off springs. This is known as **incomplete linkage.**
21. **Bateson** and **Punnet** (1906) noted the separation of linked genes for the first time in *Lathyrus odoratus*.
22. They crossed a double dominant sweet pea plant homozygous for blue flowers and long pollen grains (BBLL) with a double recessive plant homozygous for red flower and round pollen grains (bbll).
23. F_1 plants were heterozygous for blue flowers and long pollen grains.
24. When F_1 dihybrid plant was test crossed with a double recessive parent (bbll) the progenies show unexpected phenotypic ratio of 7:1:1:7 instead of 1:1:1:1.
25. A far higher frequency of parental forms (87.4%) than that of recombinants (12.6%) shows that genes do not assort independently but pass together because they are linked.

26. In sweet pea linkage is incomplete because few recombination are obtained.
27. Genes having complete linkage show 0% frequency for recombination.



LINKAGE GROUPS:

1. All the genes in a particular chromosomes form a linkage group.
2. An i ndividual has innumerable characters and fewer chromosomes hence each chromosome should carry a number of genes.
3. The genes situated on the same chromosome should be inherited together and hence could not show independent assortment.
4. The genes located on th e same chromosomes remain linked together and should form one linkage group.
5. A pair of homologous chromosomes are said to form a single linkage group.



6. Number of linkage groups in a species corresponds to its haploid number of chromosomes. This principle is known as the **Limitation of linkage groups**.

7. Size of the linkage group depends upon the size of the chromosome.
8. Human X chromosome carries over 102 genes where as Y chromosome bears only over 10 genes.
9. *Drosophila melanogaster* has 4 linkage groups (3 large 1 small).
10. Garden pea has 7 linkage groups and 7 pairs of chromosomes.
11. In XX -XY type of organisms number of linkage groups will be one more in male than in the female as in male X and Y both are different.
12. Number of linkage group in human female is 23 an d in human male is 24.
13. Number of linkage groups in female *Drosophila* is 3 and in male *Drosophila* is 4.
14. Number of linkage groups in Maiz is 10, in ascaris is 1& in Barley is 7.
15. The number of linkage group corresponds to the haploid number of chromosomes.
16. Bacterial and blue-green algal cells contain only one linkage group.

Genes Linkage group

- **Factors Affecting Linkage:**

  Besides the distance between the genes some physiological and environmental factors affecting the linkage of genes are

  (a) **Age-** The chances of crossing over lessen with advancing age, ultimately **increasing** the strength of **linkage.**

  (b) **Temperature-** Rise in temperature increase the chances of chiasm formation hence, **decreasing** the strength of **linkage**.

  (c) **X-Rays-** Exposure to X-rays reduces the strength of linkage.

- **Significance:**

1. Linkage helps in keeping parental, racial and specific traits together.
2. Helps in maintaining valuable traits of newly developed variety.
3. Linkage disallows to combine all the traits in a single variety.
4. It is an important evidence for the location of genes in the chromosomes.

- **Chargaff's Rules:**

1. Chemical studies on the **bases of DNA** were performed by **Erwin Chargaff in 1950**.
2. Chargaff formulated important generalizations about DNA structure, which are called **chargaff's rule** in his honour. They are as follows.

   (a) The amount of purines and pyrimidines are equal A+G = T+C.

(b) The amount of Adenine is always equal to that of Thymine and the amount of Guanine is always equal to that of Cytosine (A=T and G=C).

(c) The base ratio $\frac{A+T}{G+C}$ may very from one species to another species but is constant for a given species.

(d) The **base ratio** is rarely equal to one and varies between **0.4** and **1.9.**

(e) The deoxyribose sugar and phosphate components occur in equal proportions.

DENATURATION OR MELTING:

1. When DNA molecule is heated above the physiological temperature (above 100^0C) the two **helics separate** and the phenomenon is called **Melting or Denaturation**.
2. **Denaturation** depends upon the **G + C** contents of the molecule.
3. If the G + C contents are high, the melting temperature (Tm) for DNA is also high due to more energy requirement to break GC pair than AT.
4. When the denatured **DNA is cooled slowly** the two strands re-unite and the phenomenon is called as **Annealing or Renaturation.**
5. This property of DNA enables the synthesis of hybrid DNA molecules.

WILD AND MUTANT ALLELES:

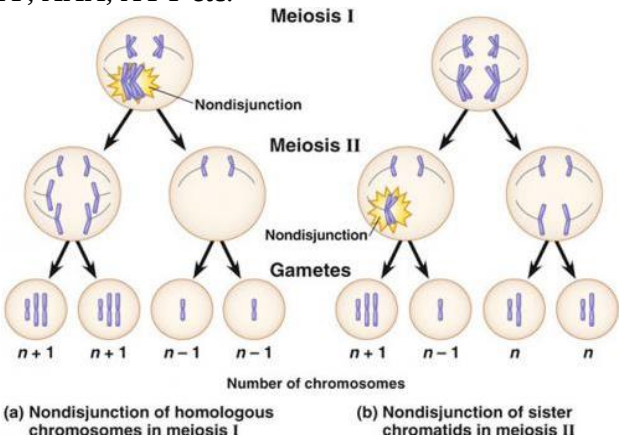
1. **Wild allele** is the one which was originally present and is even most widespread in the population.
2. **Wild allele** is usually **dominant.**
3. Change in the wild allele gives rise to **Mutant allele.**
4. **Mutant allele** is generally **recessive** and **less common**.
5. There may be several mutant alleles of the same wild allele.
6. The symbol used for such an allele system is represented by small letter for the most recessive one and using super scripts for others for example- w (white eye) w^i, w^{bl}. w^w, w^c, w^e, w^h.
7. Sign of + is used for the wild type as w^+ (red eye) in Drosophila.
8. Term **wild type** refers to a strain, organism or gene that **is predominant in the wild population**.

THEORY OF PROBABILITY:

1. The probability of occurrence of one out of two alternate events is **50% or ½**
2. The **probability** of occurrence of two **independent events** is the product of their individual probabilities that is **½ x ½ = ¼ or 25%.**

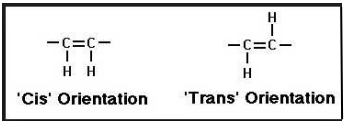
NONDISJUNCTION:

1. Failure of chromosomes to separate during anaphase is called **nondisjunction.**
2. **Nondisjunction** was discovered by **Bridge (1916)** when he found that an occasional white eye of female had a chromosome complement of XXY.
3. Here **male is XO**.
4. Failure of synapsed homologous chromosomes to separa te during anaphase I is called **Meiotic Nondisjunction.**
5. Failure of Daughter chromosomes to separate during mitotic anaphase is called **Mitotic Nondisjunction.**
6. Initial nondisjunction is called **Primary Nondisjunction**.
7. Primary nondisjunction remains in the progeny.
8. The presence of nonseparated chromosomes in the progeny due to previous nondisjunction is called **Secondary Nondisjunction.**
9. **Bridges** obtained a number of abnormal flies in the progeny of XXY like XXY, XXX, XYY etc.



CIS- TRANS ARRANGEMENT:

1. **Cis-arrangement** is the occurrence of two linked dominant genes or alleles on the same chromosome and the recessive ones on its homologue.
2. **In trans -arrangement** the linked genes are both dominant and recessive.
3. At 50% map units the linked alleles show regular crossing over so that Cis-arrangement is changed to trans-type and vice versa.

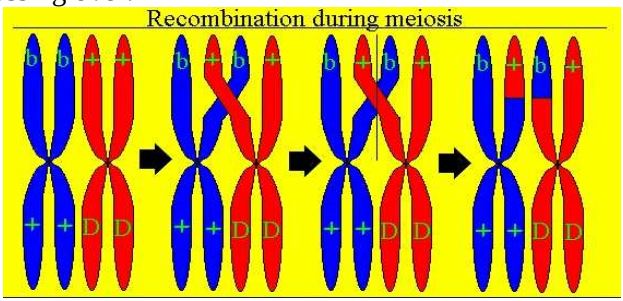


Y-SPOT (Y-CHROMATIN):

1. **Quinacrine** stained nucleus of a normal male shows a **bright flourescent band on the long arm of Y-chromosome**.
2. A single spot indicates normal male two spots in dicate super male (XYY).

RECOMBINATIONS:

1. Recombinations are a type of genetic variations which appear due to **reshuffling** of genes in linkage groups resulting in change of genotype.
2. Recombinations can occur due to
   a) Independent assortment or
   b) Random fertilization
   c) Crossing over.



3. Independent assortment occurs at the time of **meiosis or gametogenesis**.
4. Independent assortment can produce 2^2 recombinations in case of two pairs of chromosomes 2^7 types of recombinations in case of seven pairs of chromosomes and 2^{23} or 86 million types of recombinations in case of 23 pairs of chromosomes.
5. According to the **random fertilization** any of the possible recombinations in male gamete can pair with any of the possible recombinations in female gamete.
6. Chance of chromosome recombination multiplies as fallows:
   (a) $2^2 \times 2^2$ in case of two pairs of chromosomes
   (b) $2^7 \times 2^7$ in case of seven pairs of chromosomes

(c) 2^{23} x 2^{23} (8.6 million x 8.6 million or 7 x 10^{12}) in case of 23 pairs of chromosome.

7. Crossing over establishes new linkages due t o exchange of segments between nonsister chromatids.

CRYPTOGRAM:

Cryptogram is the symbolic representation of various traits of viruses.

1. It was proposed by **Gibbs et al (1966)** and adopted by **ICNV**.
2. A cryptogram has four pairs
   c) Nucleic acid type, D or R and number of strands 1 or 2
   d) Molecular weight of nucleic acid in millions/percentage of nucleic acid of total particle.
   e) Outline of particle like S or E or X (complex)/outline of nucleocapsid like S.
   f) Host/Vector like Seed plants (S) invertebrate (I) Vertebrate (V) bacterium (B) Fungus (Fu) dipera (Di) Coleoptera (cl) aphid (Af) nil (zero or 0) unknown (*)
3. **Influenza virus** (*Myxovirus influenza*) R/1, 2-3/10, S/E, V/0
4. **POX virus** (*poxvirus variolae*) D/2, 160/5 – 7.5, X /*, V/0
5. **T_4** D/2. 130/40, X/X
6. **Poliovirus** (*Poliovirus primus*) R/1. 2.5/30, S/S, V0

CANCER:

1. **Cancer** is uncontrolled growth of cells often forming **tumours** and invading other tissues.
2. **Tumour** is **cellular lump.**
3. If tumour remians restricted to the area of formation it is called **benign or non cancerous or non malignant tumour.**
4. A tumour which invades neighbouring organs by sending secondaries is known as **malignant or cancerous tumour.**
5. The secondary growth or invasion is known as **Metastasis.**
6. Rous (1910) found that *Raus Sercoma Virus* causes cancer.
7. In human beings cancer is caused by
   a) chromosomal translocations
   b) Extra-activation of certain genes
   c) Jumping genes or transposons
   d) Amplification or depletion of certain genes
8. Fragment C-abl from chromosome 9 when translocated to chromosome 22 causes chronic **myelogenous leukemia.**

9. Translocation of segment C -myc from 8 to 14 chromosomes causes **Burkitt's lymphoma.**
10. Extra activation of **Proto-oncogenes** in to cellular oncogenes also causes cancer.
11. **Jumping genes** may cause gene changes or take strong promoters for proto-oncogenes.
12. Till now no human cancer is known definitely to be due to any virus.
13. Factors which alter the activity of certain genes resulting in to cancer are
    a) Heat b) Radiations c) Tobacco d) Irritation e) Tar, Azo dyes f) Aromatic amines g) Urathene h) Metals like Beryllium, chromium, Arsenic, Nickel i) Asbestos, mine dust j) Certain viruses, excess secretion of certain harmones, excess of antibiotics and certain vitamins.
14. **Oncogenes** were discovered by **Bishop and Varmus.**

HARDY-WEINBER LAW:

1. In populatio n genetics the **gene frequency** can be studied through **Hardy-Weinberg Law** or **Hardy-Weinberg equation.**
2. According to Hardy-Weinberg law
   $P + q = 1$ $(P + q)^2 = 1^2 = 1$ $P^2 + 2Pq + q^2 = 1$
3. $1-\sqrt{q^2} = P$ where **P is dominant trait** and **q is recessive trait** that appears only in homozygous (q^2) state.
4. **Phenyl thiocarbamide (PTC)** is a water soluble compound which even in dilute solution is bitter to test for most persons **(tasters TT or Tt).**
5. Bitterness is not felt by some persons called **non taster (tt).**
6. Most persons can roll t heir tongue **(RR, Rr)** while a few are unable to do it **(rr).**
7. **Ear lobes** are **free** in most cases **(dominant)** and fused in some (recessive).
8. The trait of **dimpled cheeks** is **dominant** while that of normal cheeks is recessive.

QUESTIONS

1. Which one fro m those given below is the period of Mendel's hybridisation experiments? [NEET 2017]
   (a) 1856 - 1863 (b) 1840 -1850 (c) 1857 - 1869 (d) 1870 - 1877
2. Among the following characters, which one was not considered by Mendel in his experiments on pea? [NEET 2017]
   (a) Stem - Tall or Dwarf (b) Trichomes -Glandular or Non -glandular
   (c) Seed - Green or Yellow (d) Pod -Inflated or Constricted
3. The genotypes of a husband and wife are I^AI^B and I^Ai. Among the blood types of their children, how many different geno types and phenotypes are possible? [NEET 2017]
   (a) 3 genotypes; 3 phenotypes (b) 3 genotypes; 4 phenotypes
   (c) 4 genotypes; 3 phenotypes (d) 4 genotypes; 4 phenotypes
4. Thalassemia and sickle -cell anaemia are caused due to a problem in globin molecule synthesis. Select the correct statement. [NEFT 2017]
   (a) Both are due to a qualitative defect in global chain synthesis
   (b) Both are due to a quantitative defect in globin chain synthesis
   (c) Thalassemia is due to less synthesis of globin molecules
   (d) Sickle -cells anaemia is due to a quantitative problem of globin molecules
5. A disease caused by an autosomal primary non-disjunction is [NEFT 2017]
   (a) down's syndrome (b) klinefelter's syndrome
   (c) turner's syndrome (d) sickle-cell anemia
6. Which of the following most appropriately describes haemophilia? [NEFT 2016,Phase I]
   (a) X-linked recessive gene disorder
   (b) Chromosomal disorder
   (c) Dominant gene disorder
   (d) Recessive gene disorder
7. A cell at telophase stage is observed by a student in a plant brought from the field. He tells his teacher that this cell is not like other cells at telophase stage. There is no formation of cell plate and thus the cell is containing more number of chromosomes as compared to other dividing cells. This would result in [NEFT 2016, Phase I]
   (a) polyploidy (b) somaclonal variation
   (c) polyteny (d) aneuploidy

8. Match the terms in column I with their description in column II and choose the correct option. [NEET 2016, Phase I]

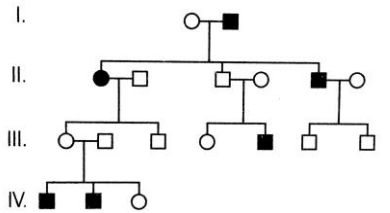
	Column I		Column 11
A.	Dominance	1.	Many genes govern a single character
B.	Codominance	2.	In heterozygous organism only one allele expresses itself
C.	Pleiotropy	3.	In heterozygous organism both alleles express themselves fully
D.	Polygenic inheritance	4.	single gene influences many characters

Code

	A	B	C	D		A	B	C	D
(a)	2	3	4	1	(b)	4	1	2	3
(c)	4	3	1	2	(d)	2	1	4	3

9. In a test cross involving F_1 dihybrid flies, more parental -type offspring were produced than the recombinant type offspring. This indicates [NEET 2016, Phase I]
   (a) chromosomes failed to separate during meiosis
   (b) the two genes are linked and present on the same chromosome
   (c) both of the characters are controlled by more than one gene
   (d) the two genes are located on two different chromosomes
10. Pick out the correct statements. [NFFT 2016, Phase I]
    I. Haemophilia is a sex-linked recessive disease.
    II. Down's syndrome is due to aneuploidy.
    III. Phenylketonuria is an autosomal recessive gene disorder.
    IV. Sickle-cell anaemia is an X – linked recessive gene disorder.
    (a) II and IV are correct (b) I, III and IV are correct
    (c) I, II and III are correct (d) I and IV are correct
11. A tall true breeding garden pea plant is crossed with a dwarf true breeding garden pea plant. When the F_1 plants were selfed the resulting genotypes were in the ratio of [NEFT 2016, Phase I]
    (a) 1 : 2 : 1:: Tall heterozygous: Tall homozygous : Dwarf
    (b) 3 : 1 :: Tall : Dwarf
    (c) 3 : 1 :: Dwarf : Tall
    (d) 1 : 2 : 1 :: Tall homozygous : Tall heterozygous : Dwarf

12. If a colourblind man marries a woman who is homozygous for normal colour vision, the probability of their son being colour blind is [NEFT 2016, Phase II]
(a) 0 (b) 0.5 (c) 0.75 (d) 1
13. A true breeding plant is [NEET 2016, Phase II]
(a) one that is able to breed on its own
(b) produced due to cross-pollination among unrelated plants
(c) near homozygous and produces offspring of its own kind
(d) always homozygous recessive in its genetic constitution
14. The mechanism that causes a gene to move from one linkage group to another is called [NEET 2016, Phase II]
(a) inversion (b) duplication (c) translocation (d) crossing over
15. A gene showing codominance has [CBSE AIPMT 2015]
(a) one allele dominant on the other
(b)alleles tightly linked on the same chromosome
(c) alleles that are recessive to each other
(d) Both alleles independently expressed in the heterozygote
16. The term "linkage" was coined by [CBSE AIPMT 2015]
(a) TH Morgan (b) T Boveri (c) G Mendel (d) W Sutton
17. In the following human pedigree, the filled symbols represent the affected individuals. Identify the type of given pedigree.[CBSE AIPMT 2015]



(a) Autosomal dominant (b) X-linked recessive
(c) Autosomal recessive (d) X-linked dominant
18. A colourblind man marries a woman with normal sight who has no history of colour blindness in her family. What is the probability of their grandson being colour blind? [CBSE AIPMT 2015]
(a) 0.5 (b) 1 (c) Nil (d) 0.25
19. In his classic experiments on pea plants, Mendel did not use [CBSE AIPMT 2015]
(a) seed colour (b) pod length
(c) seed shape (d) flower position

20. A pleiotropic gene [CBSE AIPMT 2015]
(a) is expressed only in primitive plants
(b) is a gene evolved during Pliocene
(c) controls a trait only in combination with another gene
(d) controls multiple traits in an individual

21. A man whose father was colour blind marries a woman, who had a colour blind mother and normal father. What percentage of male children of this couple. will be colour blind? [CBSE AIPMT 2014]
(a) 25% (b) 0% (c) 50% (d) 75%

22. A human female with Turner's syndrome [CBSE AIPMT 2014]
(a) has 45 chromosomes with XO
(b) has one additional X-chromosome
(c) exhibits male characters
(d) is able to produce children with normal husband

23. Fruit colour in squash is an example of [CBSE AIPMT 2014]
(a) recessive epistasis (b) dominant epistasis
(c) complementary genes (d) inhibitory genes

24. Which of the following statements is not true of two genes that show 50%recombination frequency? [NEET 2013]
(a) The genes may be on different chromosomes
(b) The genes are tightly linked
(c) The genes show independent assortment
(d) If the genes are present on the same chromosome, they undergo more than one crossovers in every meiosis

25. If two persons with 'AB' blood group marry and have sufficiently large number of children, these children could be classified as 'A' blood group: 'AB' blood group: 'B' blood group in 1 : 2 : 1ratio. Modern technique of protein electrophoresis reveals presence of both 'A' and 'B' type proteins in 'AB' blood group individuals. This is an example of [NEET 2013]
(a) codominance (b) incomplete dominance
(c) partial dominance (d) complete dominance

26. If both parents are carriers for thalassaemia, which is an autosomal recessive disorder, what are the chances of pregnancy resulting in an affected child? [NEET 2013]
(a) No chance (b) 50% (c) 25% (d) 100%

27. Which Mendelian idea is depicted by a cross in which the F1 generation resembles both the parents? [NEET 2013]

(a) Incomplete dominance (b) Law of dominance
(c) Inheritance of one gene (d) Codominance

28. The incorrect statement with regard to haemophilia is [NEET 2013]
(a) it is a sex-linked disease
(b) it is a recessive disease
(c) it is a dominant disease
(d) a single protein involved in the clotting of blood is affected

29. A normal -visioned man whose father was colour blind, marries a woman whose father was also colour blind. They have their first child as a daughter. What are the chances that this child would be colour blind? [CBSE AIPMT 2012]
(a) 100% (b) 0% (c) 25% (d) 50%

30. Fz -generation in a Mende lian cross showed that both genotypic and phenotypic ratios are same as 1 : 2 : 1. It represents a case of
(a) codominance [CBSE AIPMT 2012]
(b) dihybrid cross
(c) monohybrid cross with complete dominance
(d)monohybrid cross with incomplete dominance

31. Which one of the following conditions correctly describes the manner of determining the sex in the given example? [CBSE AIPMT 2011]
(a) XO type of sex chromosomes determine male sex in grasshopper
(b) XO condition in humans as found in Turner syndrome , determines female sex
(c) Homozygous sex chromosomes (XX) produce male in Orosophila
(d) Homozygous sex chromosomes(ZZ) determine female sex in birds

32. Select the correct statement from the ones given below with respect to dihybrid cross.[CBSE AIPMT 2010]
(a) Tightly linked genes on the same chromos ome show higher recombinations
(b) Genes far apart on the same chromosome show very few recombinations
(c) Genes loosely linked on the same chromosome show similar recombinations as the tightly linked ones
(d)Tightly linked genes on the same chromosome show very few recombinations

33. ABO blood groups in humans are controlled by the gene I. -It has three alleles - IA, IB and i. Since there are three different alleles, six different genotypes are possible. How many phenotypes can occur? [CBSE AIPMT 2010]

(a) Three (b) One (c) Four (d) Two

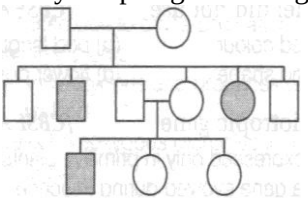
34. Which one of the following symbols and its representation, used in human pedigree analysis is correct? [CBSE AIPMT 2010]
(a) 0 = 0 = Mating between relatives (b) 0 = Unaffected male
(c) D = Unaffected female (d) . = Male affected

35. Which one of the following cannot be explained on the basis of Mendel's Law of Dominance? [CBSE AIPMT 2010]
(a)The discrete unit controlling a particular character is called a factor
(b) Out of one pair of factors one is dominant, and the other recessive
(c) Alleles do not show any blending and both the characters recover as such in F_2-generation
(d) Factors occur in pairs

36. The genotype of a plant showing the dominant phenot ype can be determined by [CBSE AIPMT 2010]
(a) test cross (b) dihybrid cross (c) pedigree analysis (d) back cross

37. Select the incorrect statement from the following [CBSE AIPMT 2009]
(a) linkage is an exception to the principle of independent assortment in heredity
(b) galactosemia is an inborn error of metabolism
(c) small population size results in random genetic drift in a population
(d) baldness is a sex limited trait

38. Study the pedigree chart given below.



What does it show?[CBSE AIPMT 2009]
(a) Inheritance of a sex-linked inborn error of metabolism like phenylketonuria
(b) Inheritance of a condition like phenylketonuria as an autosomal recessive trait.
(c) The pedigree chart is wrong as this is not possible.
(d) Inheritance of a recessive sex-linked disease like haemophilia.

39. Point mutation involves [CBSE AIPMT 2009]
(a) insertion (b) change in single base pair (c) duplication (d) deletion

40. Which one of the following condition in humans is correctly matched with its chromosomal abnormality/linkage? [CBSE AIPMT 2008]
(a) Klinefelter's syndrome- 44 autosomes + XX'
(b) Colour blindness - Y-linked
(c) Erythroblastosis foetalis- x-linked
(d) Down syndrome- 44 autosomes + XO

41. In pea plants, yellow seeds are dominant to green. If a heterozygous yellow seeded plant is crossed with a green seeded plant, what ratio of yellow and green seeded plants would you expect in I-generation? [CBSE AIPMT 2007]
(a) 50:50 (b) 9: 1 (c) 1:3 (d) 3: 1

42. A common test to find the genotype of a hybrid is by [CBSE AIPMT 2007]
(a) crossing of one F_2 progeny with male parent
(b) crossing of one F_2 progeny with female parent
(c) studying the sexual behaviour of F_1-progenies
(d) crossing of one F_1 progeny with male parent

43. Two genes R and Y are located very close on the chromosomal linkage map of maize plant. When RRYY and rryy genotypes are hybridised, then F_2 segregation will show [CBSE AIPMT 2007]
(a) higher number of the recombinant types
(b) segregation in the expected 9 : 3 : 3 : 1 ratio
(c) segregation in 3 : 1 ratio
(d) higher number of the parental types

44. A human male produces sperms with the genotypes AB, Ab, aB and ab pertaining to two diallelic characters in equal proportions. What is the corresponding genotype of this person? [CBSE AIPMT 2007]
(a) AaBb (b) AaBB (c) AABb (d) AABB

45. Test cross involves [CBSE AIPMT 2006]
(a) crossing between two genotypes with recessive trait
(b) crossing between two F_1 hybrids
(c) crossing the F_1 hybrid with a double recessive genotype
(d) crossing between two genotypes with dominant trait

46. In which mode of inheritance do you expect more maternal influence among the offspring? [CBSE AIPMT 2006]
(a) Autosomal (b) Cytoplasmic (c) Y-linked (d) X-linked

47. How many different kinds of gamete s will be produced by a plan t having the genotype AABbCC? [CBSE AIPMT 2006]
(a) Three (b) Four (c) Nine (d) Two

48. In Mendel's experiments with garden pea, round seed shape (RR) was dominant over wrinkled seeds (rr), yellow cotyledon (YY) was dominant over green cotyledon (yy). What are the expected phenotypes in the F_2 -generation of the cross RRYY x rryy?[CBSE AIPMT 2006]
(a) Only round seeds with green cotyledons

(b) Only wrinkled seeds with yellow cotyledons
(c) Only wrinkled seeds with green cotyledons
(d) Round seeds with yellow cotyledons and wrinkled seeds with yellow cotyledons

49. Phenotype of an organism is the result of [CBSE AIPMT 2006]
(a) mutations and linkages
(b) cytoplasmic effects and nutrition
(c)environmental changes and sexual dimorphism
(d) genotype and environmental interactions

50. In order to find out the different types of gametes produced by a pea plant having the genotype AaBb, it should be crossed to a plant with the genotype [CBSE AIPMT 2005]
(a) aaBB (b) AaBb (c) AABB (d) aabb

51. Haemophilia is more commonly seen in human males than in human females because [CBSE AIPMT 2005]
(a) this disease is due to an X-linked dominant mutation
(b) a greater proportion of girls die in infancy
(c) this disease is due to an X-linked recessive mutation
(d) this disease is due to a Y-linked recessive mutation

52. A woman with normal vision, but whose father was colour blind, marries a colour blind man. Suppose that the fourth child of this couple was a boy. This boy [CBSE AIPMT 2005]
(a) must have normal colour vision
(b) will be partially colour blind since he is heterozygous for the colour blind mutant allele
(c) must be colour blind
(d) may be colour blind or may be of normal vision

53. A man and a woman, who d o not show any apparent signs of a certain inherited disease, have seven children (2 daughters and5 sons). Three of the sons suffer from the given disease but none of the daughters are affected. Which of the following mode of inheritance do you suggest for this disease? [CBSE AIPMT 2005]
(a) Autosomal dominant (b) Sex-linked dominant
(c) Sex-limited recessive (d) Sex-linked recessive

54. A normal woman whose father was colour blind is married to a normal man. The sons would be [CBSE AIPMT 2004]
(a) 75% colour blind (b) 50% colour blind
(c) all normal (d) all colour blind

55. In a plant, red fruit (R) is dominant over yellow fruit (r) and tallness (T) is dominant over shortness (t), If a plant with RRTt genotype is crossed with a plant that is rrtt [CBSE AIPMT 2004]
(a) 25% will be tall with red fruit
(b) 50% will be tall with red fruit
(c) 75% will be tall with red fruit
(d) all of the offspring will tall with red fruit

56. A male human is heterozygous for autosomal genes A and B and is also hemizygous for haemophilic gene h. What proportion of his sperms will be abh? [CBSE AIPMT 2004]
(a) $\frac{1}{8}$ (b) $\frac{1}{32}$ (c) $\frac{1}{16}$ (d) $\frac{1}{4}$

57. The recessive genes located on X-chromosome I humans are always [CBSE AIPMT 2004]
(a) lethal (b) sublethal (c) expressed in males (d) expressed in females

58. Extranuclear inheritance is a consequence of presence of genes in [CBSE AIPMT 2004]
(a) mitochondria and chloroplasts
(b) endoplasmic reticulum and mitochondria
(c) ribosomes and chloroplast
(d) Iysosomes and ribosomes

59. Lack of independent assortment of two genes A and B in f ruit fly Drosophila is due to [CBSE AIPMT 2004]
(a) repulsion (b) recombination (c) linkage (d) crossing over

60. In a mutational e vent, when adenine is replaced by guanine, it is the case of [CBSE AIPMT 2004]
(a) frameshift mutation (b) transcription
(c) transition (d) transversion

61. One of the parents of a cross has mutation in its mitochondria. In that cross, that parent is tak en as a male. During segregation of F_2 - progenies that mutation is found in [CBSE AIPMT 2004]
(a) one-third of the progenies (b) none of the progenies
(c) all of the progenies (d) fifty per cent of the progenies

62. Pattern baldness, moustaches and beard in human males are examples of [CBSE AIPMT 2003]
(a) sex differentiating traits (b) sex determining traits
(c) sex linked traits (d) sex limited traits

63. Two crosses between the same pair 01 genotypes or phenotypes in which the sources of the gametes are reversed in one cross, is known as

[CBSE AIPMT 2003]
(a) dihybrid cross (b) reverse cross (c) test cross (d)reciprocal cross

64. The linkage map of X -chromosome of fruit fly has 66 units, with yellow body gene (y) at one end and bobbed hair (b) gene at the other end. The recombination frequency between these two genes (y and b) should be [CBSE AIPMT 2003]
(a) s 50% (b) 100% (c) 66% (d) > 50%

65. Down's syndrome is caused by an extra copy of chromosome number 21. What percentage of offspring produced by an affected mother and a normal father would be affected by this disorder? [CBSE AIPMT 2003]
(a) 50% (b) 25% (c) 100% (d) 75%

66. The genes controlling the seven pea characters studied by Mendel are now known to be located on how many different chromosomes? [CBSE AIPMT 2003]
(a) Five (b) Four (c) Seven (d) Six

67. Which one of the following traits of garden pea studied by Mendel was a recessive feature? [CBSE AIPMT 2003]
(a) Green pod colour (b) Round seed shape
(c) Axial flower position (d) Green seed colour

68. Which of the following discoveries resulted in a Nobel Prize? [CBSE AIPMT 2003]
(a) Recombination of linked genes
(b) Genetic engineering
(c) X-rays induce sex-linked recessive lethal mutations
(d) Cytoplasmic inheritance

69. When a cluster of genes show linkage behaviour they [CBSE AIPMT 2003]
(a) do not show independent assortment (b) induce cell division
(c) do not show a chromosome map (d) show recombination during meiosis

70. In Drosophila, the sex is determined by [CBSE AIPMT 2003]
(a) the ratio of pairs of X-chromosomes to the pairs of autosomes
(b) whether the egg is fertilised or develops parthenogenetically
(c) the ratio of number of X-chromosomes to the set of autosomes
(d) X and Y-chromosomes

71. Genes for cytoplasmic male sterility in plants are generally located in [CBSE AIPMT 2003]
(a) nuclear genome (b) cytosol

(c)chloroplast genome (d) mitochondrial genome

72. In recent years, DNA sequences (nucleotide sequence) of mtDNA and Y -chromosomes were considered for the study of human evolution, because [CBSE AIPMT 2003]
(a) their structure is known in greater detail
(b) they can be studied from the samples of fossil remains
(c) they are small and therefore, easy to study
(d) they are uniparental in origin and do not take part in recombination

73. Genetic map is one that [CBSE AIPMT 2003]
(a) shows the stages during the cell division
(b) shows the distribution of various species in a region
(c) establishes sites of the genes on a chromosome
(d) establishes the various stages in gene evolution

74. There are three genes a, b, c, percentage of crossing over between a and b is 20%, b and c is 28% and a and c is 8%. What is the sequence of genes on chromosome? [CBSE AIPMT 2002]
(a) b, a, c (b) a, b, c (c) a, c, b (d) None of these

75. Which of the following is the example of sex-linked disease? [CBSE AIPMT 2002]
(a) AIDS (b) Colour blindness (c) Syphilis (d) Gonorrhoea

76. Pleiotropic gene is [CBSE AIPMT 2002]
(a) haemophilia (b) thalassemia
(c) sickle-cell anaemia (d) colour blindness

77. Change in the sequence of nucleotide in DNA is called as [CBSE AIPMT 2002]
(a) mutagen (b) mutation (c) recombination (d) translation

78. A plant of F_1-generation has genotype 'AABbCC'. On selfing of this plant, the phenotypic ratio in F_2 -generation will be [CBSE AIPMT 2002]
(a) 3 : 1 (b) 1 : 1 (c) 9: 3: 3: 1 (d) 7:9:9:9:3:3:3:1

79. In his experiment, Mendel obtained wrinkled pea. The wrinkling was due to deposition of sugar instead of starch. This happened due to the enzyme [CBSE AIPMT 2001]
(a) amylase (b) invertase
(c) diastase (d) absence of starch-branching enzyme

80. Which of these do not follow independent assortment? [CBSE AIPMT 2001]
(a) Genes on non-homologous chromosomes and absence of linkage

(b) Genes on homologous chromosomes
(c) Linked genes on same chromosome
(d) Unlinked genes on same chromosome

81. Extranuclear inheritance occurs in [CBSE AIPMT 2001]
(a) Killer Paramecium (b) Killer Amoeba
(c) Eug/ena (d) Hydra

82. Number of Barr bodies in XXXX female [CBSE AIPMT 2001]
(a) 1 (b) 2 (c) 3 (d) 4

83. Male XX and female XY sometime occur due to [CBSE AIPMT 2001]
(a) deletion (b) transfer of segments in X and Y chromosomes
(c) aneuploidy (d) hormonal imbalance

84. Two non -allelic genes produce the new phenotype when present together but fail to do so independently, it is called [CBSE AIPMT 2001]
(a) epistasis (b) polygene
(c) non-complementary gene (d) complementary gene

85. A and B genes are linked. What shall be the genotype of progeny in a cross between AB/ab and ab/ab? [CBSE AIPMT 2001]
(a) AAbb and aabb (b) AaBb and aabb
(c) AABB and aabb (d) None of these

86. Ratio of complementary genes is [CBSE AIPMT 2001]
(a) 9 : 3 : 4 (b)12 : 3 : 1 (c) 9 : 3 : 3 : 4 (d) 9 : 7

87. During organ differentiation in Drosophila, an organ is modified to another organ (such as wings may be replaced by legs). Genes responsible for such metamorphosis are called [CBSE AIPMT 2000]
(a) double dominant genes (b) plastid genes
(c) complementary genes (d) homeotic genes

88. Drosophila flies with XXY genotype are females, but human beings with such genotype are abnormal males. It shows that [CBSE AIPMT 2000]
(a) V-chromosome is essential for sex determination in Drosophila
(b) V-chromosome is female determinating in Drosophila
(c) V-chromosome is male determining in human beings
(d)V-chromosome has no role in sex determination either in Drosophila or in human beings

89. Mutation generally produces [CBSE AIPMT 2000]
(a). recessive genes (b) lethal genes
(c) polygenes (d) dominant genes

90. Which one of the following characters studied by Mendel in garden pea was found to be dominant? [CBSE AIPMT 2000]
(a) Green seed colour (b) Terminal flower position
(c) Green pod colour (d) Wrinkled seed

91. Hybridisation between Ttx tt gives rise to the progeny of ratio [CBSE AIPMT 1999]
(a) 1 : 1 (b) 1 : 2 : 1 (c) 1 : 2 (d) 4 : 1

92. Haemophilic man marries a normal woman. Their offspring will be [CBSE AIPMT 1999]
(a) all boys haemophilic (b) all normal
(c) all girls haemophilic (d) all haemophilic

93. Which of the following is the main category of mutation? [CBSE AIPMT 1999]
(a) Somatic mutation (b) Genetic mutation
(c) Zygotic mutation (d) All of these

94. A woman with two genes (one on each X -chromosome) for haemophilia and one gene for colour blindness on the X -chromosomes marries a normal man. How will the progeny be?[CBSE AIPMT 1998]
(a) All sons and daughters haemophilic and colour blind
(b) Haemophilic and colour blind daughters
(c) 50% haemophilic colour blind sons and 50% haemophilic sons
(d) 50% haemophilic daughters and 50% colour blind daughters

95. Albinism is known to be due to an autosomal recessive mutation. The first child o f a couple with normal skin pigmentation was an albino. What is the probability that their second child will also be an albino? [CBSE AIPMT 1998]
(a) 100% (b) 25% (c) 50% (d) 75%

96. Crossing over in diploid organism is responsible for [CBSE AIPMT 1998]
(a) dominance of genes (b) linkage between genes
(c) segregation of alleles (d) recombination of linked alleles

97. How many types of genetically different gametes will be produced by a heterozygous plant having genotype AABbCc? [CBSE AIPMT 1998]
(a) Two (b) Four (c) Six (d) Nine

98. Mental retardation in man, associated with sex chromosomal abnormality is usually due to [CBSE AIPMT 1998]
(a) reduction in X-complement
(b) increase in X-complement

(c) moderate increase in Y-complement
(d) large increase in Y-complement

99. Which base is responsible for hotspots for spontaneous point mutations? [CBSE AIPMT 1998]
(a) Guaine (b) Adenine (c) 5-bromouracil (d) 5-methylcytosine

100. If Mendel had studied the seven trai ts using a plant with 12 chromosomes instead of 14, in what way would his interpretation have been different? [CBSE AIPMT 1998]
(a) He would have mapped the chromosome
(b) He would have discovered blending or incomplete dominance
(c) He would not have discovered the law of independent assortment
(d) He would have discovered sex-linkage

101. When a single gene influences more than one traits it is called [CBSE AIPMT 1998]
(a) pleiotropy (b) epistasis (c) pseudodominance (d) None of these

102. Loss of an X-chromosome in a particular cell, during its development, results into [CBSE AIPMT 1998]
(a) diploid individual (b) triploid individual
(c) gynandromorphs (d) Both (a) and (b)

103. The formation of multivalents at meiosis in diploid organism is due to [CBSE AIPMT 1998]
(a) monosomy (b) inversion (c) deletion (d) reciprocal translocation

104. A fruit fly heterozygous for sex -linked genes, is mated with normal female fruit fly. Male specific chromosome will enter egg cell in the proportion [CBSE AIPMT 1997]
(a) 1 : 1 (b) 2 : 1 (c) 3 : 1 (d) 7 : 1

105. Foetal sex can be determined by examining cells from the amniotic fluid by looking for [CBSE AIPMT 1997]
(a) Barr bodies (b) autosomes (c) Chiasmata (d) kinetochore

106. A mutation at one base of the first codon of a gene produces a non-functional protein. Such a mutation is referred as [CBSE AIPMT 1997]
(a) frameshift mutation (b) mis-sense mutation
(c) non-sense mutation (d) reverse mutation

107. The hereditary material present in the bacterium E. coli is [CBSE AIPMT 1997]
(a) single stranded RNA (b) double stranded RNA
(c) single stranded DNA (d) double stranded DNA

108. Genetic identity of a human male is determined by [CBSE AIPMT 1997]
(a) autosome (b) nucleolus (c) sex chromosome (d) cell organelles
109. Different mutations referrable to the same locus of chromosome give rise to [CBSE AIPMT 1997]
(a) pseudoalleles (b) polygenes (c) oncogenes (d) multiple alleles
110. After crossing two plants, the progenies are found to be male sterile. This phenomenon is found to be maternally inherited and is due to some genes which are present in [CBSE AIPMT 1997]
(a) nucleus (b) chloroplast (c) mitochondria (d) cytoplasm
111. HJ Muller was awarded Nobel Prize for his [CBSE AIPMT 1996]
(a) discovery that chemicals can induce gene mutations
(b) discovery that ionizing radiations can induce gene mutations
(c) work on gene mapping in Orosophila
(d) efforts to prevent the use of nuclear weapons
112. A man with a certain disease marries a normal woman. They have eight children (3 daughters and 5 sons). All the daughters suffer from their father's disease but none of the sons are affected. Which of the following mode of inheritance do you suggest for this disease? [CBSE AIPMT 1996, 2002]
(a) Sex-linked recessive (b) Sex-linked dominant
(c) Autosome dominant (d) Sex-limited recessive
113. A person with 47 chromosomes due to an additional Y -chromosome suffers from a condition called [CBSE AIPMT 1996, 97]
(a) Down's syndrome (b) Super female
(c) Turner's syndrome (d) Klinefelter's syndrome
114. Alleles that produce independent effects in their heterozygous condition are called [CBSE AIPMT 1996]
(a) codominant alleles (b) epistatic alleles
(c) complementary alleles (d) supplementary alleles
115. An individual exhibiting both male and female sexual characteristics in the body is known as [CBSE AIPMT 1996]
(a) hermaphrodite (b) intersex (c) gynandromorph (d) bisexual
116. T he most striking example of point mutation is found in a disease called [CBSE AIPMT 1995]
(a) thalassemia (b) night blindness
(c) Down's syndrome (d) sickle-cell anaemia
117. The polytene chromosomes were discovered for the first time in [CBSE AIPMT 1995]

(a) Drosophila (b) Chironomus
(c) Musca nebulo (d) Musca domestica

118. When two genetic loci produce identical phenotypes in cis and trans position, they are considered to be [CBSE AIPMT 1995]
(a) pseudoalleles (b) different genes
(c) multiple alleles (d) parts of same gene

119. In a dihybrid cross AABB x aabb, F_2 progeny of AABB, AABb, AaBB and AaBb occurs in the ratio of [CBSE AIPMT 1994]
(a) 1 : 1 : 1 : 1 (b) 9 : 3 : 3 : 1 (c) 1 : 2 : 2 : 1 (d) 1 : 2 : 2 : 4

120. A cross between pure tall pea plant with green pods and dwarf pea plant with yellow pods will produce dwarf F_2 plants out of 16 [CBSE AIPMT 1994]
(a) 9 (b) 3 (c) 4 (d) 1

121. Out of A=T, G==C pairing, bases of DNA may exist in alternate valency state owing to arrangement called [CBSE AIPMT 1994]
(a) analogue substitution (b) tautomerisational mutation
(c) frameshift mutation (d) point mutation

122. A colourblind woman marries a normal visioned male. In the offspring [CBSE AIPMT 1994]
(a) both son and daughter are colour blind
(b) all daughters are colour blind
(c) all sons are normal
(d) all sons are colour blind

123. Genes located on Y-chromosome are [CBSE AIPMT 1994]
(a) mutant genes (b) sex-linked genes
(c) autosomal genes (d) holandric genes

124. A woman with albinic father marries an albinic man. The proportion of her progeny is [CBSE AIPMT 1994]
(a) 2 normal: 1 albinic (b) all normal
(c) all albinic (d) 1 normal: 1 albinic

125. A child of blood group 0 cannot have parents of blood groups [CBSE AIPMT 1994]
(a) AB and AB/a (b) A and B
(c) Band B (d) a and a

126. Which of the following is suitable for experiment on linkage? [CBSE AIPMT 1993]
(a) aaBB × aaBB (b) AABB × aabb
(c) AaBb × AaBb (d) AAbb × AaBB

127. Two dominant non -allelic genes are 50 map units apart. The linkage is [CBSE AIPMT 1993]
(a) cis type (b) trans type (c) complete (d) absent/incomplete

128. Mendel studied inheritance of seven pairs of trai ts in pea which can have 21 possible combinations. If you are told that in one of these combinations, independent assortment is not observed in later studies, your reaction will be [CBSE AIPMT 1993]
(a) independent assortment principle may be wrong
(b) Mendel might not have studied all the combinations
(c) it is impossible
(d) later studies may be wrong

129. Of both normal parents, the chance of a male child becoming colour blind are [CBSE AIPMT 1993]
(a) no
(b) possible only when all the four grand parents had normal vision
(c) possible only when father's mother was colour blind
(d) possible only when mother's father was colour blind .

130. Mr. Kapoor has Bb autosomal gene pair and d allele sex -linked. What shall be proportion of Bd in sperms? [CBSE AIPMT 1993]
(a) 0 (b) 1/2 (c) 1/4 (d) 1/8

131. Sex is determined in human beings [CBSE AIPMT 1993]
(a) by ovum
(b) at the time of fertilisation
(c) 40 days after fertilisation
(d) seventh to eight week when genitals differentiate in foetus

132. Of a normal couple, half the sons are haemophilic while half the daughters are carriers. The gene is located on [CBSE AIPMT 1993]
(a) X-chromosome of father (b) V-chromosome of father
(c) one X-chromosome of mother (d) both the X-chromosomes of mother

133. A polygenic inheritance in human beings is
[CBSE AIPMT 1993, 99, 2006, 07]
(a) skin colour (b) phenylketonuria
(c) colour blindness (d) sickle-cell anaemia

134. In human beings 45 chromosomes/single X/XO abnormality causes
[CBSE AIPMT 1992]
(a) Down's syndrome (b) Klinefelter's syndrome
(c) Turner's syndrome (d) Edward's syndrome

135. A colourblind mother and normal father would have [CBSE AIPMT 1992, 99, 2006]
(a) colour blind sons and normal/carrier daughters
(b) colour blind sons and daughters
(c) all colour blind
(d) all normal

136. Down's syndrome is due to [CBSE AIPMT 1992, 2000, 02, 03]
(a) crossing over (b) linkage
(c) sex-linked inheritance (d) non-disjunction of chromosomes

137. When a certain character is inherited only through female parent, it probably represents [CBSE AIPMT 1992]
(a) multiple plastid inheritance (b) cytoplasmic inheritance
(c) incomplete dominance (d) Mendelian nuclear inheritance

138. Out-of 8 ascospores formed in Neurospora the arrangement is Za : 4a : Za showing [CBSE AIPMT 1992]
(a) no crossing over (b) some meiosis
(c) second generation division (d) first generation division

139. An organism with two identical alleles is [CBSE AIPMT 1992]
(a) dominant (b) hybrid (c) heterozygous (d) homozygous

140. Segregation of Mendelian factors (no linkage, no crossing over) occurs during [CBSE AIPMT 1992]
(a) anaphase-I (b) anaphase-Ii (c) diplotene (d) metaphase-I

141. An allele is dominant if it is expressed in [CBSE AIPMT 1992, 2002]
(a) both homozygous and heterozygous states
(b) second generation
(c) heterozygous combination
(d) homozygous combination

142. A gene pair hid es the effect of another. The phenomenon is [CBSE AIPMT 1992,95,99]
(a) epistasis (b) dominance (c) mutation (d) None of these

143. RR (red) Antirrhinum is crossed with WW (white) one. Offspring RW are pink. This is an example of [CBSE AIPMT 1991]
(a) dominant-recessive (b) incomplete dominance
(c) hybrid (d) supplementary genes

144. The allele which is unable to express its effect in the presence of another is called [CBSE AIPMT 1991]
(a) codominant (b) supplementary
(c) complementary (d) recessive

145. The contrasting pairs of factors in Mendelian crosses are called [CBSE AIPMT 1991]
(a) multiple alleles (b) allelomorphs (c) alloloci (d) paramorphs
146. First geneticist/father of genetics was [CBSE AIPMT 1991]
(a) De Vries (b) Mendel (c) Darwin (d) Morgan
147. Mendel's last law is
(a) segregation (b) dominance
(c) independent assortment (d) polygenic inheritance
148. A dihybrid condition is [CBSE AIPMT 1991]
(a) It Rr (b) Tt rr (c) It rr (d) Tt Rr
149. A colourblind girl is rare because she will be born only when [CBSE AIPMT 1991]
(a) her mother and maternal grandfather were colourblind
(b) her father and maternal grandfather were colourblind
(c) her mother is colour blind and father has normal vision
(d) parents have normal vision but grand parents were colourblind and Variation
150. Blue eye colour is recessive to brown eye colour. A brown eyed man whose mother was blue eyed marries a blue eyed woman . The children shall be
(a) both blue eyed and brown eyed 1 : 1 (b) all brown eyed
(c) all blued eyed (d) blue eyed and brown eyed 3 : 1
151. Multiple alleles control inheritance of [CBSE AIPMT 1991]
(a) phenylketonuria (b) colour blindness
(c) sickle-cell anaemia (d) blood groups
152. Which one is a hereditary disease? [CBSE AIPMT 1990]
(a) Cataract (b) Leprosy (c) Blindness (d) Phenylketonuria
153. Haemophilia is more common in males because it is a [CBSE AIPMT 1990]
(a) recessive character carried by V-chromosome
(b) dominant character carried by V-chromosome
(c) dominant trait carried by X-chromosome
(d) recessive trait carried by X-chromosome
154. Both husband and wife have normal vision though their fathers were colour blind. The probability of their d aughter becoming colour- blind is [CBSE AIPMT 1990]
(a) 0% (b) 25% (c) 50% (d) 75%
155. In Down's syndrome of a male child, the sex complement is [CBSE AIPMT 1990]

(a) XO (b) XV (c) XX (d) XXV

156. A normal green male maize is crossed with albino female. The progeny is albino because [CBSE AIPMT 1989]
(a) trait for albinism is dominant
(b) the albinos have biochemical to destroy plastids derived from green male
(c) plastids are inherited from female parent
(d) green plastids of male must have mutated

157. Two linked genes a and b show 20% recombination. The individuals of a dihybrid cross between ++/++ × ab/ab shall show gametes [CBSE AIPMT 1989]
(a) ++80 : ab :20 (b) ++50 : ab: 50
(c) ++ 40 : ab 40: + a 10 : + b : 10 (d) + + 30 : ab 30: + a 20: + b : 20

158. Diploid chromosome number in humans is [CBSE AIPMT 1989]
(a) 46 (b) 44 (c) 48 (d) 42

159. A family of five daughters only is expecting sixth issue. The chance of its beings a son is [CBSE AIPMT 1988]
(a) Zero (b) 25% (c) 50% (d) 100%

160. Which contribute to the success of Men del? [CBSE AIPMT 1988]
(a) Qualitative analysis of data
(b) Observation of distinct inherited traits
(c) His knowledge of Biology
(d) Consideration of one character at one time

161. Haploids are able to express both recessive and dominant alleles/mutations because there are [CBSE AIPMT 1988]
(a) many alleles for each gene
(b) two alleles for each gene
(c) only one allele for each gene in the individual
(d) only one allele in a gene

162. If there are 999 bases in an RNA that codes for a protein with 333 amino acids and the base at position 901 is deleted such that the length of the RNA becomes 998 bases, how many eo dons will be altered? [NEET 2017]
(a) 1 (b) 11 (c) 33 (d) 333

163. The final proof fo r DNA as the genetic material came from the experiments of [NEET 2017]
(a) Griffith (b) Hershey and Chase
(c) Avery, MacLeod and McCarty (d) Hargobind Khorana

164. The association of histone H1 with a nucleosome indicates [NEET 2017]
(a) transcription is occurring
(b) DNA replication is occurring
(c) the DNA is condensed into chromatin fibre
(d) the DNA double helix is exposed

165. Spliceosomes are not found in cells of [NEET 2017]
(a) plants (b) fungi (c) animals (d) bacteria

166. DNA replication in bacteria occurs [NEET 2017]
(a) during S-phase (b) within nucleolus
(c) prior to fission (d) just before transcription

167. Which of the following RNAs should be most abundant in animals cell? [NEET 2017]
(a) rRNA (b) tRNA (c) mRNA (d) miRNA

168. Which one of the following is the starter codon? [NEET2016,Phase I]
(a) UGA (b) UAA (c) UAG (d) AUG

169. A complex of ribosomes attached to a single strand of RNA is known as [NEET 2016, Phase I]
(a) polymer (b) polypeptide
(c) okazaki fragment (d) polysome

170. Which of the following is not required for any of the techniques of DNA fingerprinting available at present? [NEET 2016, Phase I]
(a) Zinc finger analysis (b) Restriction enzymes
(c) DNA-DNA hybridisation (d) Polymerase chain reaction

171. Which of the following is required as inducer(s) for the expression of lac operon? [NEET 2016, Phase I]
(a) galactose (b) lactose (c) lactose and galactose (d) glucose

172. DNA-dependent RNA polymer ase catalyses transcription on one strand of the DNA which is called the [NEET 2016, Phase II]
(a) template strand (b) coding strand (c) alpha strand (d) anti strand

173. A molecule that can act as a genetic material must fulfill the traits given below, except [NEET 2016, Phase II]
(a) it should be able to express itself in the form of 'Mendelian characters'
(b) it should be able to generate its replica
(c) it should be unstable structurally and chemically
(d) it should provide the scope for slow changes that are required for evolution

174. Which of the following r RNAs act as structural RNA as well as ribozyme in bacteria? [NEET 2016, Phase II]
(a) 5 srANA (b) 18 srANA (c) 23 srANA (d) 58 srANA

175. The equivalent of a structural gene is [NEET 2016, Phase II]
(a) muton (b) cistron (c) operon (d) recon

176. Taylor conducted the experiments to prove semiconservative mode of chromosome replication on [NEET 2016, Phase II]
(a) Vinca rosea (b) Vicia faba
(c) Drosophila melanogaster (d) E.coli

177. Which one of the following is not applicable to RNA? [CBSE AIPMT 2015]
(a) Complementary base pairing (b) 5' phosphoryl and 3' hydroxyl ends
(c) Heterocyclic nitrogenous bases (d) Chargaff's rule

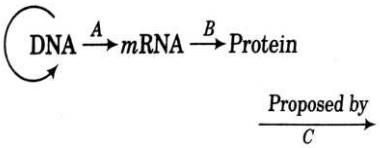
178. Identify the cor rect order of organisation of genetic material from largest to smallest. [CBSE AIPMT 2015]
(a) Chromosome, gene, genome, nucleotide
(b) Genome, chromosome, nucleotide, gene
(c) Genome, chromosome, gene, nucleotide
(d) Chromosome, genome, nucleotide, gene

179. Satellite DNA is important because it [CBSE AIPMT 2015]
(a) codes for proteins needed in cell cycle
(b) shows high degree of polymorphism in population and also the same degree of polymorphism in an individual, which is heritable from parents to children
(c) does not code for proteins and is same in all members of the population
(d) codes for enzymes needed for DNA replication

180. Which one of the following is wrongly matched?[CBSE AIPMT 2014]
(a) Transcription-Writing information from DNA to tRNA
(b) Transiation-Usinq information in mRNA to make protein
(c) Repressor protein-Binds to operator to stop enzyme synthesis
(d) Operon-Structural genes, operator and promoter

181.Transformation was discovered by [CBSE AIPMT 2014]
(a) Meselson and Stahl (b) Hershey and Chase
(c) Griffith (d) Watson and Crick

182. The diagram shows an important concept in the genetic implication of DNA Fill in the blanks A to C. [NEET 2013]



(a) A -transcription, B -replication, C - James Watson
(b) A -translation, B -transcription, G - Erwin Chargaff
(c) A-transcription, B-translation, C-Francis Crick
(d) A-translation, 8-extension, C-Rosalind Franklin

183. Which enzyme/s will be produced in a cell in which there is a non - sense mutation in the lac Y-gene? [NEET 2013]
(a) p-galactosidase (b) Lactose permease
(c) Transacetylase (d) Lactose permease and transacetylase

184. Removal of RNA polymerase -III from nucleoplasm will affect the synthesis of [CBSE AIPMT 2012]
(a) tRNA (b) hnRNA (c) mRNA (d) rANA

185. Which one of the following is not a part of a transcription unit in DNA? [CBSE AIPMT 2012]
(a) The inducer (b) A terminator (c) A promoter (d) The structural gen

186. Ribosomal RNA is actively synthesised in [CBSE AIPMT 2012]
(a) lysosomes (b) nucleolus
(c) nucleoplasm (d) ribosomes

187. If one strand of DNA has the nitrogenous base sequence as ATCTG, what would be the complementary RNA strand sequence? [CBSE AIPMT 2012]
(a) TIAGU (b) UAGAC (c) AACTG (d) ATCGU

188. Removal o f introns and joining of exons in a defined order during transcription is called [CBSE AIPMT 2012]
(a) looping (b) inducing (c) slicing (d) splicing

189. What are the structures called that give an appearance as 'beads on string' in the chromosom es when viewed under electron microscope? [CBSE AIPMT 2011]
(a) Genes (b) Nucleotides (c) Nucleosomes (d) Base pairs

190. Select the two statements out of the four (I -IV) given below about lac operon.
I. Glucose or galactose may bind with the repressor and inactivate it.
II. In the absence of lactose, the repressor binds with the operator region.
III. The z-gene codes for permease.
IV. This was elucidated by Francois Jacob and Jacques Monad.
The correct statements are [CBSE AIPMT 2010]

(a) I and III (b) I and III (c) II and IV (d) I and II

191. What is not true for genetic code? [CBSE AIPMT 2009]
(a) A codon in mRNA is read in a non-contiguous fashion
(b) It is nearly universal
(c) It is degenerate
(d) It is unambiguous

192. Whose experiments cracked the DNA and discovered unequivocally that a genetic code is a triplet? [CBSE AIPMT 2009]
(a) Nirenberg and Matthaei (b) Hershey and Chase
(c) Morgan and Sturtevant (d) Beadle and Tatum

193. Polysome is formed by [CBSE AIPMT 2008]
(a) several ribosomes attached to a single mRNA
(b) many ribosomes attached to a strand of endoplasmic reticulum
(c) a ribosome with several subunits
(d) ribosomes attached to each other in a linear arrangement

194. Which one of the following pairs of codons is correctly matched with their function or the signal for the particular amino acid? [CBSE AIPMT 2008]
(a) GUU, GCU Alanine (b) UAG, UGA Stop
(c) AUG, ACG Start/methionine (d) UUA, UCA Leucine

195. A sequential expression of a set of human genes occurs when a steroid molecule binds to the [CBSE AIPMT 2007]
(a) transfer RNA (b) messenger RNA
(c) DNA sequence (d) ribosome

196. One gene-one enzyme relationship was established for the first time in [CBSE AIPMT 2007]
(a) Neurospora crassa (b) Salmonella typhimurium
(c) Escherichia coli (d) Diplococcus pneumoniae

197. The Okazaki fragments in DNA chain growth [CBSE AIPMT 2007]
(a) result in transcription
(b) polymerise in the 3' to 5' direction and forms replication fork
(c) prove semi-conservative nature of DNA replication
(d) polymerise in the 5' to 3' direction and explain 3' to 5' DNA replication

198. Differentiation of organs and tissues in a developing organism is associated with [CBSE AIPMT 2007]
(a) developmental mutations (b) differential expression of genes
(c) lethal mutations (d) deletion of genes

199. The length of DNA molecule greatly exceeds the dimensions of the nucleus in eukaryotic cells. How is this DNA accommodated? [CBSE AIPMT 2007]
(a) Deletion of non-essential genes (b) Super-coiling in nucleosomes
(c) DNAse digestion (d) Through elimination of repetitive DNA

200. Molecular basis of organ differentiation depends on the modulation in transcription by [CBSE AIPMT 2007]
(a)RNA polymerase(b) ribosome (c) transcription factor (d) anticodon

201. Telomere repetitive DNA sequences control the function of eukaryotic chromosomes because they. [CBSE 2007]
(a) act as replicons (b) are RNA transcription i nitiator
(c) help chromosome pairing (d) prevent chromosome loss

202. One gene-one postulated by
(a) R Franklin (b) Hershey and Chase
(c) A Garrod (d) Beadle and Tatum

203.Amino acid sequence, in protein synthesis is decided by the sequence of [CBSE AIPMT 2006]
(a) tRNA (b) mRNA (c) cDNA (d) (RNA

204. Which one of the following makes use of RNA as a template to synthesise DNA? [CBSE AIPMT 2005]
(a) Reverse transcriptase (b) DNA dependant RNA polymerase
(c) DNA polymerase (d) RNA polymerase

205. Which one of the following hydrolyses internal phosphodiester bonds in a polynucleotide chain? [CBSE AIPMT 2005]
(a) lipase (b) Exonuclease (c) Endonuclease (d) Protease

206. During transcription holoenzyme RNA polymerase binds to a DNA sequence and the DNA assumes a saddle like structure at that point. What is that sequence called? [CBSE AIPMT 2005]
(a)CAAT box (b)GGTI box (c) AAAT box (d) TATA box

207. Telomerase is an enzyme which is a [CBSE AIPMT 2005]
(a) repetitive DNA (b) RNA (c) simple protein (d) ribonucleoprotein

208. During replication of a bacterial chromosome DNA synthesis starts from a replication origin site and [CBSE AIPMT 2004]
(a) RNA primers are involved (b) is facilitated by telomerase
(c) moves in one direction of the site (d) moves in bi-directional way

209. After a mutation at genetic locus the character of an organism changes due to the change in [CBSE AIPMT 2004]

(a) protein structure (b) DNA replication
(c) protein synthesis pattern (d) RNA transcription pattern

210. The telomeres of eukaryotic chromosomes consist of short sequences of [CBSE AIPMT 2004]
(a) thymine rich repeats (b) cytosine rich repeats
(c) adenine rich repeats (d) guanine rich repeats

211. The following ratio is generally constant for a given species [CBSE AIPMT 2004]
(a) A + G/C + T (b) T + C/G + A (c) G + CIA + T (d) A + C/T + G

212. During transcription, the nucleotide sequence of the DNA strand that is being coded is AT ACG, then the nucleotide sequence in the mRNA would be [CBSE AIPMT 2004]
(a) TATGC (b) TCTGG (c) UAUGC (d) UATGG

213. DNA fingerprinting refers to [CBSE AIPMT 2004]
(a) molecular analysis or profiles of DNA samples
(b) analysis of DNA samples using imprinting device
(c) techniques used for molecular analysis of different specimens of DNA
(d) techniques used for identification of finger-prints of individuals

214. What does 'lac' refer to in what we call the lac operon? [CBSE AIPMT 2003]
(a) Lac insect (b) The number, 1,00,000
(c) Lactose (d) Lactase

215. During transcription, the DNA site at which RNA polymerase binds is called [CBSE AIPMT 2003]
(a) receptor (b) enhancer (c) promoter (d) regulator

216. Chromosomes in a bacterial cell can be 1-3 in number and [CBSE AIPMT 2003]
(a) can be either circular or linear, but never both within the same cell
(b) can be circular as well as linear within the same cell
(c) are always circular
(d) are always linear

217. In the genetic code dictionary, how many codons are used to code for all the 20 essential amino acids? [CBSE AIPMT 2003]
(a) 61 (b) 60 (c) 20 (d) 64

218. What would happen if in a gene encoding a polypeptide of 50 amino acids, 25^{th} codon (UAU) is mutated to UAA? [CBSE AIPMT 2003]
(a) A polypeptide of 49 amino acids will be formed

(b) A polypeptide of 25 amino acids will be formed
(c) A polypeptide of 24 amino acids will be formed
(d) Two polypeptides of 24 and 25 amino acids will be formed

219. Degeneration of a genetic code is attributed to the [CBSE AIPMT 2003]
(a) entire cod on (b) third member of a cod on
(c) first member of a codon (d) second member of a codon

220. During translation initiation in prokaryotes, a GTP molecule is needed in [CBSE AIPMT 2003]
(a) association of 30S, rrflNA with formyl met tRNA
(b) association of 50S subunit of ribosome with initiation complex
(c) formation of formyl met *t*RNA
(d) binding of 30S subunit of ribosome with *m*RNA

221. Which one of the following triplet c odes, is correctly matched with its specificity for an amino acid in protein synthesis or as 'start' or 'stop' codon? [CBSE AIPMT 2003]
(a) UGU-Leucine (b) UAC-Tyrosine (c) UCG-Start (d) UUU-Stop

222. Exon part of mRNAs have code for [CBSE AIPMT 2002]
(a) protein (b) lipid (c) carbohydrate (d) phospholipid

223.Which of the following reunites the exon segments after RNA splicing? [CBSE AIPMT 2002]
(a) RNA polymerase(b)RNA primase (c) RNA ligase (d) RNA protease

224. Nucleus of a donor em bryonal cell/somatic cell is transferred to an enucleated egg cell. Then after the formation of organism, what shall be true? [CBSE AIPMT 2002]
(a) Organism will have extra-nuclear genes of the donor cell
(b) Organism will have extra-nuclear genes of recipient cell
(c) Organism will have extra -nuclear genes of both donor and recipient cell
(d) Organism will have nuclear genes of recipient cell

225. Which statements is correct for bacterial transduction? [CBSE AIPMT 2002]
(a) Transfer of some genes f rom one bacteria to another bacteria through virus
(b) Transfer of genes from one bacteria to another bacteria by conjugation
(c) Bacteria obtained its DNA directly
(d) Bacteria obtained DNA from other external source

226. In a DNA percentage of th ymine is 20. What is the percentage of guanine? [CBSE AIPMT 2002]
(a) 20% (b) 40% (c) 30% (d) 60%
227. Jacob and Monod studied lactose metabolism in E. coli and proposed Operon concept. Operon concept applicable for [CBSE AIPMT 2002]
(a) all prokaryotes
(b) all prokaryotes and some eukaryotes
(c) all prokaryotes and all eukaryotes
(d) all prokaryotes and some protozoans
228. In E. coli, during lactose metabolism repressor binds to [CBSE AIPMT 2002]
(a) regulator gene(b)operator gene(c)structural gene(d) promoter gene
229. Sequence of which of the following is used to know the phylogeny? [CBSE AIPMT 2002]
(a) mRNA (b) rRNA (c) tRNA (d) DNA
230. In negative operon [CBSE AIPMT 2001]
(a) co-repressor binds with repressor
(b) co-repressor does not bind with repressor
(c) co-repressor binds with inducer
(d) cAMP has negative effect on lac operon
231. In which direction mRNA is synthesised on DNA template? [CBSE AIPMT 2001]
(a) 5' -7 3' (b) 3' -7 5' (c) Both (a) and (b) (d) Any of above
232. Gene and cistron words are sometimes used synonymously because [CBSE AIPMT 2001]
(a)one cistron contains many genes(b) one gene contains many cistrons
(c) one gene contains one cistron (d) one gene contains no cistron
233.E.coli about to replicate was placed in a medium containing radioactive thymidine for five minutes. Then it was made to replicate in a normal medium. Which of the following observation shall be correct? [CBSE AIPMT 2001]
(a) Both the strands of DNA will be radioactive
(b) One strand radioactive
(c) Each strand half radioactive
(d) None is radioactive
234. Due to discovery of which of the following in 1980's the evolution was termed as RNA world?[CBSE AIPMT 2001]
(a) nBNA, tRNA, rRNA synthesise proteins

(b) In some viruses, RNA is genetic material
(c) Some RNAs have enzymatic property
(d) RNA is not found in all cells

235. 'Signal hypothesis' for the biosynthesis of secretory type of proteins was proposed by [CBSE AIPMT 2000]
(a) Camillo Golgi (b) Blobel and Sabatini
(c) Baltimore (d) Sheeler and Bianchi

236. During replication of DNA, its two strands separate. Each of these serves as a template for the formation of new strands. Such type of replication is called [CBSE AIPMT 2000]
(a) non-conservative (b) semi-conservative
(c) flexible (d) conservative

237. The Pneumococcus experiment proves that [CBSE AIPMT 1999]
(a) DNA is the genetic material
(b) RNA sometime controls the production of DNA and proteins
(c) bacteria undergo binary fission
(d) bacteria do not reproduce sexually

238. In DNA when AGCT occurs, their association is as per which of the following pair? [CBSE AIPMT 1999]
(a) ACGT (b) AGCT (c) ATGC (d) All of these

239. DNA elements, which can switch their position, are called [CBSE AIPMT 1998]
(a) exons (b) introns (c) cistrons (d) transposons

240. Genes that are involved in turning on or off the transcription of a set of structural genes are called [CBSE AIPMT 1998]
(a) polymorphic genes (b) operator genes
(c) reductant genes (d) regulatory genes

241. Protein synthesis in an animal cell takes place [CBSE AIPMT 1997]
(a) only in the cytoplasm
(b) in the nucleolus as well as in the cytoplasm
(c) in the cytoplasm as well as in mitochondria
(d) only on ribosomes attached to a nucleus

242.The RNA that picks up specific amino acid from amino acid pool in the cytoplasm to ribosome during protein synthesis is called [CBSE AIPMT 1997]
(a) nBNA (b) tRNA (c) rRNA (d) RNA

243.The condons causing chain termination are [CBSE AIPMT 1997]
(a) TAG, TAA, TGA (b) GAT, AAT, AGT

(c) AGT, TAG, UGA (d) UAA, UAG, UGA

244.Genes are packaged into a bacterial chromo- some by [CBSE AIPMT 1997]

(a) histones (b) basic protein (c) acidic protein (d) actin

245.An enzyme that joins the ends of two strands of nucleic acid is a [CBSE AIPMT 1996, 2002]

(a) polymerase (b) synthetase (c) helicase (d) ligase

246. Okazaki fragments are seen during [CBSE AIPMT 1996]

(a) transcription (b) translation (c) replication (d) transduction

247. The translation termination triplet is [CBSE AIPMT 1996]

(a) UAU (b) UAA (c) UAC (d) UGC

248. The basis for DNA fingerprinting is [CBSE AIPMT 1996]

(a) occurrence of Restriction Fragment Length Polymorphism (RFLP)

(b) phenotypic differences between individuals

(c) availability of cloned DNA

(d) knowledge of human karyotype

249.In split genes, the coding sequence are called [CBSE AIPMT 1995]

(a) introns (b) operons (c) exons (d) cistrons

250. If the sequence of bases in DNA is ATTCGATG, then the sequence of bases in its transcript will be [CBSE AIPMT 1995]

(a) CAUCGAAU (b) UAAGCUAC

(c) GUAGCUUA (d) AUUCGAUG

251.Anticodon is an unpaired triplet of bases in an expo sed position of [CBSE AIPMT 1995]

(a) mRNA (b) rRNA (c) tRNA (d) sRNA

252.The wild type E. coli cells are growing in normal medium with glucose. They are transferred to a medium containing only lactose as sugar. Which of the following changes takes place? [CBSE AIPMT 1995]

(a) The lac operon is repressed (b) All operons are induced

(c) The lac operon is induced (d) E. coli cells stop dividing

253.Which is not involved in protein synthesis? [CBSE AIPMT 1994]

(a) Transcription (b) Initiation

(c) Elongation (d) Termination

254.Protein helping in opening of DNA double helix in front of replications fork is

(a) DNA gyrase (b) DNA polymerase-I

(c) DNA ligase (d) topoisomeras

255.DNA template sequence of CTGATAGC is transcribed over mRNA as [CBSE AIPMT 1994]
(a) GUCTUTCG (b) GACUAUCG (c) GAUTATUG (d) UACTATCU
256.In Escherichia coli lac Operon is induced by [CBSE AIPMT 1994]
(a) lactose (b) promoter gene
(c) βgalactosidase (d) I-gene
257. Reverse transcriptase is [CBSE AIPMT 1994]
(a) RNA dependent RNA polymerase
(b) DNA dependent RNA polymerase
(c) DNA dependent DNA polymerase
(d) RNA dependent DNA polymerase
258.The number of base substitution possible in amino acid eo dons is [CBSE AIPMT 1994]
(a) 261 (b) 264 (c) 535 (d) 549
259.Initiation codon of protein synthesis (in eukaryotes) is [CBSE AIPMT 1993,94,99, 2000]
(a) GUA (b) GCA (c) CCA (d) AUG
260.Nucleosome core is made of [CBSE AIPMT 1993]
(a) H1, H2A, H2B and H3 (b) H1, H2A, H2B and H4
(c) H1, H2A, H2B, H3 and H4 (d) H2A, H2B, H3 and H4
261. A DNA with unequal nitrogen bases would most probably be [CBSE AIPMT 1993]
(a)singlestranded(b)double stranded(c)triple stranded (d) four stranded
262.The process of translation is [CBSE AIPMT 1993]
(a) ribosome synthesis (b) protein synthesis
(c) DNA synthesis (d) RNA synthesis
263. During DNA replication, the strands separate by [CBSE AIPMT 1993]
(a) DNA polymerase (b) topoisomerase
(c) unwindase/helicase (d) gyrase
264. Who proved that DNA is basic genetic material? [CBSE AIPMT 1993]
(a) Griffith (b) Watson (c) Boveri and Sultan (d) Hershey and Chase
265. Because most of the amino acids are represented by more than one codon, the genetic code is [CBSE AIPMT 1993, 2002]
(a) overlapping (b) wobbling (c) degenerate (d) generate
266.Nucleotide arrangement in DNA can be seen by [CBSE AIPMT 1993]
(a) X-ray crystallography (b) electron microscope
(c) ultracentrifuge (d) light microscope

267.The transforming principle of Pne umococcus as found out by A very, Mac Lead and McCarty was [CBSE AIPMT 1993]
(a) mRNA (b) DNA (c) protein (d) polysaccharide

268.Experimental material in the study of DNA replication has been [CBSE AIPMT 1992]
(a) Escherichia coli (b) Neurospora crassa
(c) Pneumococcus (d) Drosophila melanogaster

269. Khorana first deciphered the triplet codons of [CBSE AIPMT 1992]
(a) serine and isoleucine (b) threonine and histidine
(c) tyrosine and tryptophan (d) phenylalanine and methionine

270. Escherichia coli fully labelled with N^{15} is allowed to grow in N^{14} medium.The two strands of DNA molecule of the first generation bacteria have [CBSE AIPMT 1992]
(a) different density and do not resemble parent DNA
(b) different density but resemble parent DNA
(c) same density and resemble parent DNA
(d) same density but do not resemble parent DNA

271. In RNA, thymine is replaced by [CBSE AIPMT 1992]
(a) adenine (b) guanine (c) cytosine (d) Uracil

272. A nucleotide is formed of [CBSE AIPMT 1991]
(a) purine. pyrimidine and phosphate (b) purine, sugar and phosphate
(c)nitrogen base, sugar and phosphate (d) pyrimidine, sugar and phosphate

273. The process of transfer of genetic information from DNA to RNN formation of RNA from DNA is [CBSE AIPMT 1991]
(a) transversion (b) transcription (c) translation (d) translocation

274.In the genetic dictionary, there are 64 codons as [CBSE AIPMT 1990]
(a) 64 amino acids are to be coded
(b) 64 types of tRNAs are present
(c) there are 44 non-sense codons and 20 sense codons
(d) genetic code is triplet

275. DNA replication is [CBSE AIPMT 1989]
(a) conservative and discontinuous (b) semi-conservative and semidiscontinuous
(c) semi-conservative and discontinuous (d) conservative

276. Genetic code consist of [CBSE AIPMT 1988]
(a) adenine and guanine (b) cytosine and uracil
(c) cytosine and guanine (d) All of the above

Answers

1. (a)	2. (b)	3. (c)	4. (c)	5. (a)	6. (a)	7. (a)
8. (a)	9. (b)	10. (c)	11.(d)	12.(a)	13. (c)	14. (c)
15. (d)	16. (a)	17. (c)	18. (d)	19. (b)	20.(d)	21. (a)
22.(a)	23. (b)	24. (b)	25. (a)	26. (c)	27. (d)	28. (c)
29. (b)	30. (d)	31. (a)	32.(d)	33. (c)	34. (a)	35. (c)
36. (a)	37. (d)	38. (d)	39. (b)	40. (a)	41. (a)	42.(d)
43. (d)	44. (a)	45. (c)	46. (b)	47. (d)	48. (d)	49. (d)
50. (d)	51. (c)	52.(d)	53. (d)	54.(b)	55. (a)	56. (a)
57. (c)	58. (a)	59. (c)	60. (c)	61. (b)	62. (d)	63. (d)
64. (b)	65. (a)	66. (b)	67. (d)	68. (c)	69. (a)	70. (a)
71. (d)	72.(d)	73. (c)	74. (a)	75 (b)	76. (c)	77. (b)
78. (a)	79. (d)	80. (c)	81. (a)	82.(c)	83. (d)	84. (d)
85. (b)	86. (d)	87. (d)	88. (c)	89. (a)	90. (c)	91. (a)
92. (b)	93. (b)	94. (c)	95. (b)	96. (d)	97. (b)	98. (b)
99. (d)	100. (c)	101. (a)	102. (c)	103. (d)	104.(a)	105. (a)
106. (b)	107.(d)	108.(c)	109. (d)	110.(c)	111. (a)	112. (b)
113.(d)	114. (a)	115. (c)	116.(d)	117. (b)	118. (a)	119.(d)
120. (c)	121. (b)	122.(d)	123. (d)	124. (d)	125.(a)	126. (b)
127. (d)	128.(b)	129. (d)	130. (c)	131. (b)	132. (c)	133. (a)
134.(c)	135. (a)	136. (d)	137.(b)	138. (c)	139. (d)	140.(a)
141. (a)	142. (a)	143.(b)	144. (d)	145. (b)	146.(b)	147.(c)
148. (d)	149.(b)	150. (a)	151. (d)	152.(d)	153.(d)	154. (a)
155.(b)	156. (c)	157. (c)	158.(a)	159.(c)	160. (d)	161. (c)
162. (c)	163. (b)	164. (c)	165. (d)	166. (c)	167. (a)	168. (d)
169. (d)	170. (a)	171. (b)	172. (a)	173. (c)	174. (c)	175. (b)
176. (b)	177. (d)	178. (c)	179. (b)	180. (a)	181. (c)	182. (c)
183. (a)	184. (a)	185. (a)	186. (b)	187. (b)	188. (d)	189. (c)
190. (c)	191. (a)	192. (a)	193. (a)	194. (b)	195. (c)	196. (a)
197. (a)	198. (b)	199. (b)	200. (c)	201. (d)	202. (d)	203. (b)
204. (a)	205. (c)	206. (d)	207. (d)	208. (a)	209. (a)	210. (d)
211. (c)	212. (c)	213. (a)	214. (c)	215. (c)	216. (c)	211. (c)
217. (a)	218. (c)	219. (b)	220. (a)	221. (b)	222. (a)	223. (c)
224. (b)	225. (a)	226. (c)	227. (b)	228. (b)	229. (b)	230. (a)
231. (a)	232. (c)	233. (b)	234. (c)	235. (b)	236. (b)	237. (a)
238. (c)	239. (d)	240. (d)	241. (c)	242. (b)	243. (d)	244. (b)
245. (b)	246. (c)	247. (b)	248. (a)	249. (c)	250. (b)	251. (c)
252. (c)	253. (a)	254. (a)	255. (b)	256. (a)	257. (d)	258. (d)
259. (d)	260. (d)	261. (a)	262. (b)	263. (c)	264. (d)	265. (c)
266. (a)	267. (b)	268. (a)	269. (b)	270. (b)	271. (d)	272. (c)
273. (b)	274. (d)	275. (b)	276. (d)			