E Book ZOO - 101 - T: Genetics and Medical Zoology (T)

E Book EDITOR

Dr. Bhausaheb R Ghorpade

Head Department of Zoology

Shri Anand College Pathardi Dist. Ahilyanagar (M.S)

414102 Phone No. (02428) 222736

Mobile No. 8975893234

Fax No. 02428-222730,

E. mail *bhaugorpade2007@gmail.com*

Front page designed by Dr. Dhiraj V. Bhavsar

Learning Outcomes for the e Book – Genetics and Medical Zoology (T)

After the completion of the course, students should be able to:

- ✓ **CO1:** Apply Mendelian genetic principles to predict outcomes of genetic crosses, interpret pedigrees and understand the basics of genetic inheritance.
- ✓ **CO2:** Recognize and explain the inheritance patterns and molecular basis of common genetic disorders, including both Mendelian and complex traits.
- ✓ **CO3:** Understand the concept of non - Mendelian genetics.
- ✓ **CO4 :** Concept and characteristics of multiple alleles, ABO blood group system, Inheritance of Rh antigen, Erythroblastosis foetalis and their medicolegal importance.
- ✓ **CO5:** Understand the structure of chromosomes, chromatin and its types, giant chromosomes and chromosomal aberrations.
- ✓ **CO6:** Successfully solve genetic problems using Punnett squares, probability calculations and pedigree analysis.
- ✓ **CO7:** Understand basic concepts of medical zoology.
- ✓ **CO8:** Understand different epidemic, vector borne and microbial diseases in humans.
- ✓ **CO9:** Understand about investigations and treatments of human physiological disorders.

ZOO - 101 - T : Genetics and Medical Zoology (T)						
Year : I		Semester : I				
Teaching Scheme				Evaluation Scheme		
Course Type	Credits	Number of Teaching hours	Lectures Per week	Internal Assessment	Semester End Exam	Total
Subject - 1	02	30	02	15	35	50

Suggested Readings:

- Genetics: Verma, P. S. and Agrawal, V. K., S. Chand and Co., New Delhi.
- Fundamentals of Genetics: B. D. Singh, Kalyani Publishers, New Delhi.
- Principle of Genetics: Sinnott, Dunn and Dobzhansky, Tata McGraw Hill Edition, New Delhi.
- Genetics: Gupta, P. K., Rastogi Publication, Meerut.
- Genetics: Sarin, C., Tata McGraw Hill, New Delhi.

- Principles of Genetics: Gardner, E. J., Simmons, M. J. and Snustad, D. P., John Wiley and Sons.
- Cytology and Genetics: Dyan Sagar V. R., Tata McGraw Hill Pub. Co. Ltd., New Delhi.
- Baker, F. J. and Silverton, R. E. : Introduction to Medical Laboratory Technology, (6^{th} ed.), Butler Worth and Co. Ltd.
- Chatterjee, K. D. (1995), Parasitology, Protozoology and Helminthology (12th ed).
- Cheesborough, M. (1987), Medical Laboratory Technology for Tropical countries (2^{nd} ed.), Butler Worth and Co. Ltd.
- Garcia, L. S. (2001), Diagnostic Medical Parasitology, (4^{th} ed.), ASM Press, Washington.
- Talib,V. H. (1999), Essential Laboratory Manual, Mehta Publishers, New Delhi.

F Y B Sc Zoology (ZOO - 101 – T)

Genetics and Medical Zoology: Semester I: Syllabus

	4), inhibitory factors (13: 3), duplicate dominant genes (factors) (15: 1). 2.4 Lethal genes in *Mus musculus*.	
3	**Chapter 3. Multiple alleles:** *(02L)*	**30**
	3.1 Concept and characteristics.	
	3.2 ABO blood group system, Inheritance of Rh antigen, Erythroblastosis foetalis and their medico legal importance	
4	**Chapter 4 Chromosomes:** *(05L)*	**37**
	4.1 Introduction: Morphology and types of chromosomes (based on the position of centromere and involvement in sex determination).	
	4.2 Chromatin, its structure and its types (Euchromatin and Heterochromatin).	
	4.3 Giant chromosomes (Polytene chromosome and Lamp brush chromosomes).	
	4.4 Chromosomal Aberrations: Structural (Deletion, duplication, inversion and translocation) and Numerical (Euploidy, monoploidy, polyploidy - auto polyploidy & allopolyploidy and aneuploidy - monosomy, nullisomy, trisomy).	
5	**Chapter 5. Sex Determination:** *(03L)*	**54**

	5.1 genetically controlled sex determination: (Heterogametic males: XX - XY & XX - XO systems, Heterogametic females: ZZ - ZW system), Genetic balance system in Drosophila.	
	5.2 Parthenogenesis and Gynandromorphism.	
6	**Chapter 6 Sex-linked Inheritance:** *(02L)*	**61**
	6.1 Sex-linked inheritance: Characteristics, types (X - linked, Y - linked, and XY - linked).	
	6.2 Examples of Sex-linked inheritance: Hemophilia, Colour blindness and Hypertrichosis	
7	**Chapter 7 Introduction to Medical Zoology:** *(01L)*	**66**
	7.1 Definitions: Parasitology, host, parasite, vector, symbiosis, commensalisms, mutualism, parasitism and zoonosis.	
	7.2. Branches of medical zoology: Medical Protozoology, Medical Helminthology, and Medical Entomology.	
8	**Chapter 8. Epidemic Diseases in Human:** *(03L)* Occurrence, causative organism, symptoms and eradication programs of the following:	**Home Assignment**

	8.1 Typhoid.	
	8.2 Cholera.	
	8.3 Small pox.	
9	**Chapter 9. Vector Borne Diseases in Human:** *(03L)* Occurrence, causative organism, symptoms and eradication programs of the following:	**69**
	9.1 Dengue.	
	9.2 Chicken Guinea	
	. 9.3 Viral Influenza.	
	9.4 Scabies.	
10	**Chapter 10 Microbial Diseases in Human:** *(03L)* Causative organism and clinical features of the following:	**Home Assignment**
	10.1 Tuberculosis.	
	10.2 Hepatitis.	
	10.3 AIDS.	

11	**Chapter 11. Investigations and treatments of human physiological disorders:** *(03L)*	**84**
	11.1 Angiography.	
	11.2 Angioplasty.	
	11.3 Dialysis.	

Question paper pattern for Theory (2 Credit courses):

The students will have to solve the question paper of 35 marks. Including optional questions, the paper setter should set the paper on entire syllabus for total 61 marks.

N. B.: All questions are compulsory.

Max. Time: 2 Hours.

Q. 1) Answer any five of the followings in one sentence - 05 Marks

✓ Attempt any five from six questions.

Q. 2 (a) Attempt any one of the following - 06 Marks

✓ Attempt any one from the two questions.

Q. 2 (b) Attempt any one of the following - 04 Marks

✓ Attempt any one from the two questions.

Q. 3 (a) Solve any one of the following - **06 Marks**

✓ Solve any one from the two questions.

Q. 3 (b) Solve any one of the following - **04 Marks**

✓ Solve any one from the two questions.

Q. 4) Write notes on (Any four) - **10 Marks**

✓ Attempt any four from six questions.

Chapter 1: Recapitulation of Mendelian Genetics:

Mendel's work: Selection of experimental plant.

Mendel was an Augustinian monk and plant breeder who conducted a series of simple, yet elegant, experiments in 1865. He was one of the first to take a quantitative, scientific approach to the study of heredity. He used the garden pea plant, *Pisum sativum* with which to conduct his studies.

This garden pea plant was an excellent choice for Mendel, for the following four reasons:

- Peas had been shown to be true-breeding (all offspring will have the same characteristic generation after generation).
- Peas exhibit a variety of contrasting traits (purple vs. white flowers; round vs. wrinkled seeds).
- The shape of the pea flower protected it from foreign pollen. Peas usually reproduce by self-pollination, in which pollen produced by a flower fertilizes eggs in the same flower.
- Pea plants grow quickly and do not require much space.

The seven (7) traits that Mendel studied are as follows:

- ✓ Form of ripe seed (R) — smooth or wrinkled
- ✓ Colour of seed albumen (Y) — yellow or green
- ✓ Colour of flower (P) — purple or white
- ✓ Form of ripe pods (I) — inflated or constricted
- ✓ Colour of unripe pods (G) — green or yellow
- ✓ Position of flowers (A) — axial or terminal
- ✓ Length of stem (T) — tall or dwarf

Through careful study of patterns of inheritance, Mendel recognized that a single trait could exist in different versions, or **alleles**, even within an individual plant or animal.

Monohybrid Cross

A monohybrid cross is a cross between the single characters or two monohybrid traits (TT and tt). Here plants which have the same characters, but differ in only one character were crossed.

For monohybrid cross, Mendel began with a pair of pea plants with two contrasting traits, i.e., one tall and another dwarf. The cross-pollination of tall and dwarf plants resulted in tall plants and the offspring were called F1 progeny. The trait which is expressed in the phenotype is called the **dominant trait** while the one that is not is called the **recessive trait.**

Mendel's first law: Law of dominance states that when parents with pure, contrasting traits are crossed together, only one form of trait appears in the next generation. The hybrid offspring will exhibit only the dominant trait in the phenotype. "Law of dominance is known as the first law of inheritance. In this law, each character is controlled by distinct units called factors, which occur in pairs. If the pairs are heterozygous, one will always dominate the other. Law of dominance explains that in a monohybrid cross between a pair of contrasting traits, only one parental character will be expressed in the F1 generation and both parental characters

will be expressed in the F2 generation in the ratio 3:1.The one which is expressed in the F1 generation is called the dominant trait and the one which is suppressed is called a recessive trait. In simple words, the law of dominance states that recessive traits are always dominated or masked by the dominant trait. This law can be described by Mendel's experiment. He then continued his experiment with self-pollination of F1 progeny plants. This resulted in both tall and short plants in the ratio of 3:1 which gave rise to the law of segregation.

Mendel's second law: Law of Segregation

Mendel's law of segregation states that ***"During the formation of gamete, each gene separates from each other so that each gamete carries only one allele for each gene."***

Law of segregation is the second law of inheritance. This law explains that the pair of alleles segregate from each other during meiosis cell division (gamete formation) so that only one allele will be present in each gamete.

In a monohybrid cross, both the alleles are expressed in the F2 generation without any blending. Thus, the law of segregation is based on the fact that each gamete contains only one allele.

This law is based on four basic concepts:

- A gene exists in more than one form of an allele.
- When gametes are produced by meiosis, the allelic pairs separate, leaving each gamete with a single allele.
- Every organism inherits two alleles for each trait.
- The two alleles of a pair are different, i.e., one is dominant and one is recessive.

Test cross for monohybrid: (1:1 ratio)

Tall
Tt

Dwarf
tt

	t	t
T	Tt Tall	Tt Tall
t	tt Dwarf	tt Dwarf

Tall (Tt) - 50% dominant Dwarf - (tt) - 50% recessive

Phenotypic ratio - 2 tall : 2 dwarf - 1:1
Genotypic ration - 2Tt :2tt - 1:1

Mendel's third law: law of Independent Assortment.

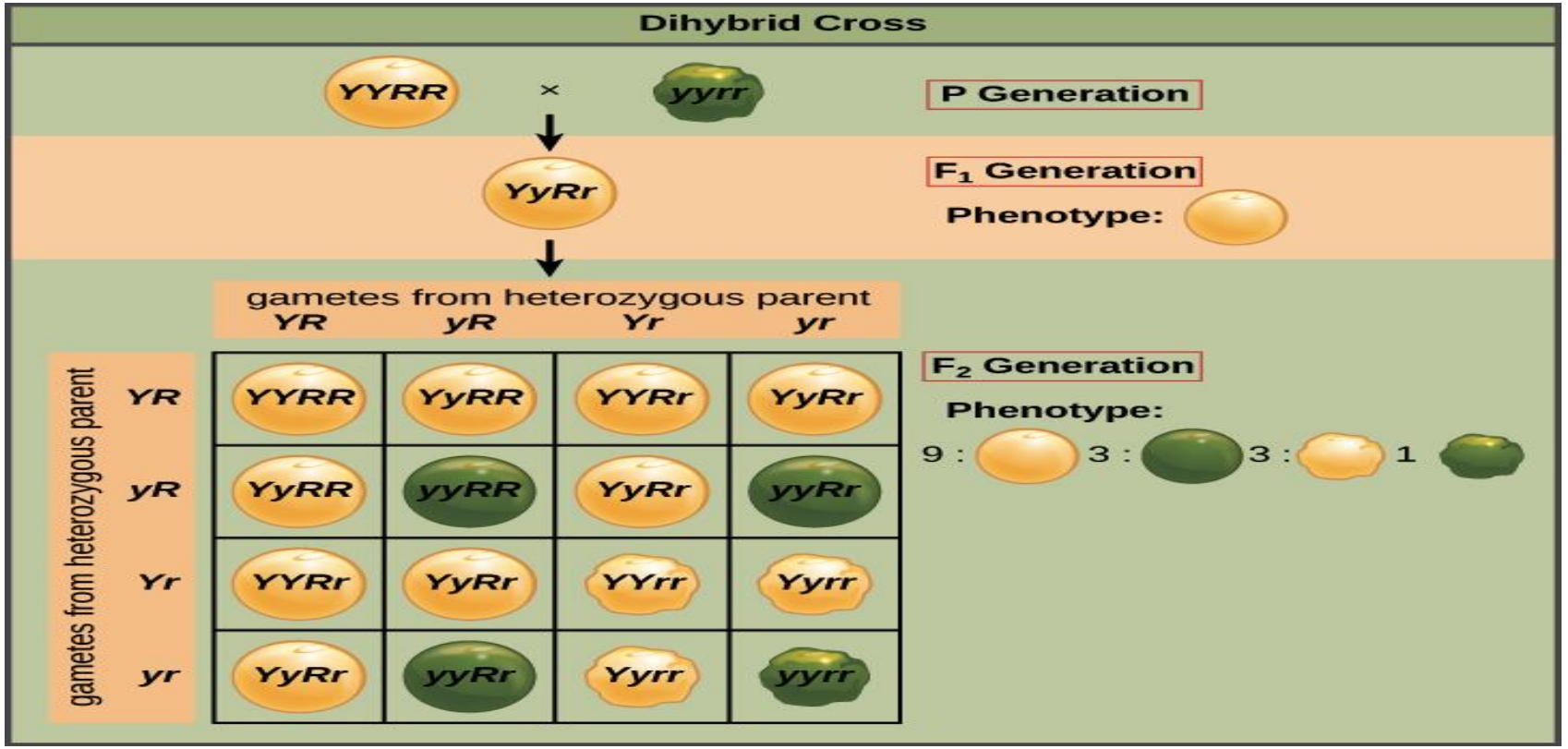
Also known as Mendel's third law of inheritance, the law of independent assortment states that a pair of traits segregates independently of another pair during gamete formation. As the individual **heredity** factors assort independently, different traits get equal opportunity to occur together.

Dihybrid Cross

In a dihybrid cross experiment, Mendel considered two traits, each having two alleles.He crossed two pure-breeding pea plants: one with yellow, round seeds (*YYRR*) and one with green, wrinkled seeds (*yyrr*). He

observed that all the first generation progeny (F1 progeny) were yellow round. This meant that dominant traits were the yellow colour and round shape.

He then self-pollinated the F1 progeny and obtained 4 different traits: yellow round, green round, yellow wrinkled, and green wrinkled seeds in the ratio 9:3:3:1 (F2).

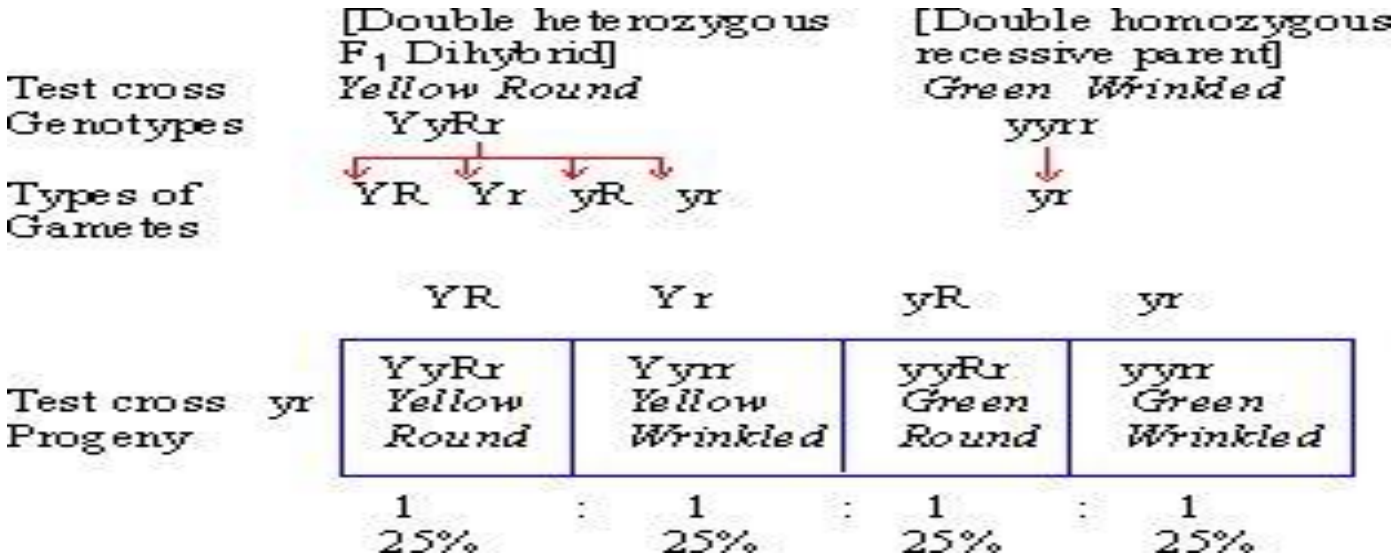


9 yellow round, 3.green round, 3 yellow wrinkled, and 1.green wrinkled seeds in the ratio of 9:3:3:1.

Mendel's fourth law: law of recombination.

Test cross for dihybrid:

In a dihybrid test cross, when the dominant parent of F1 generation is crossed, it gives 1:1:1:1 ratio. Further, on self-pollination of two heterozygous genotype gives 9:3:3:1 ratio in F2 generation.



Chapter 2. Non-Mendelian Genetics:

(Concept of epistasis, complimentary factors (9: 7), supplementary factors (9: 3 : 4), inhibitory factors (13 : 3), duplicate dominant genes (factors) (15 : 1). 2.4 Lethal genes in Mus musculus.

2.1 Concept of Gene Interaction

The expression of a single character by the interaction of more than one pair of genes is called **genic interaction** or **interaction of genes.**

The majority of the traits that comprise living beings are coordinated by various genes. Mendel and other researchers assumed that characters were controlled by a single gene, but it was later found that multiple characters were controlled by two or more genes. Such genes modify the conventional dihybrid (9:3:3:1)

Types of Gene Interaction

Gene interactions are divided into two categories: Intra-allelic interactions and Interallelic interactions.

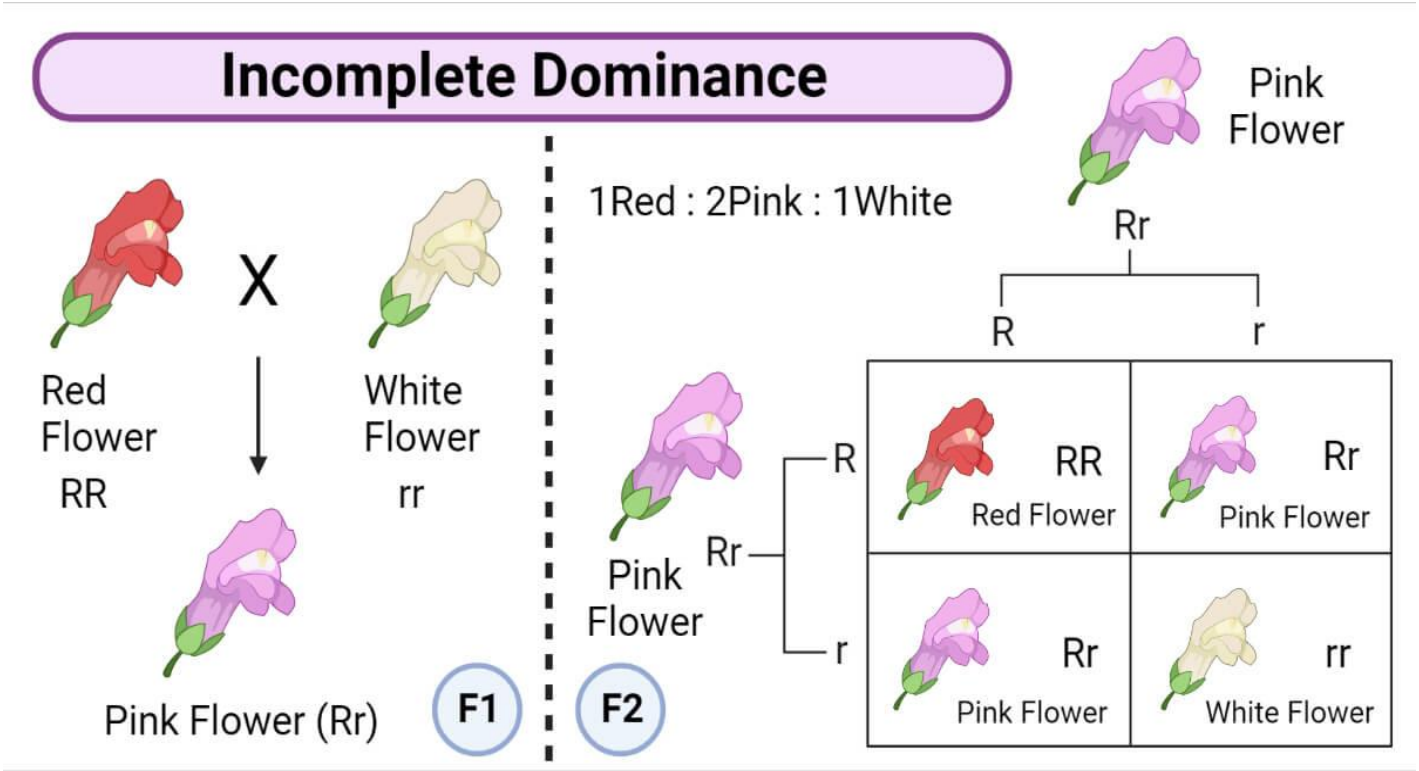
Allelic or Non-epistatic or intra-allelic interactions Gene Interaction

When phenotypic ratios diverge from Mendelian ratios, it is difficult for Mendelian genetics to explain some types of inheritance. This is because specific alleles can often be equally or partially dominant to each other or due to the lethal alleles. Allelic, non-epistatic or intra-allelic interactions are the terms used to describe genetic interactions between alleles of a single gene.

Incomplete Dominance (1:2:1)

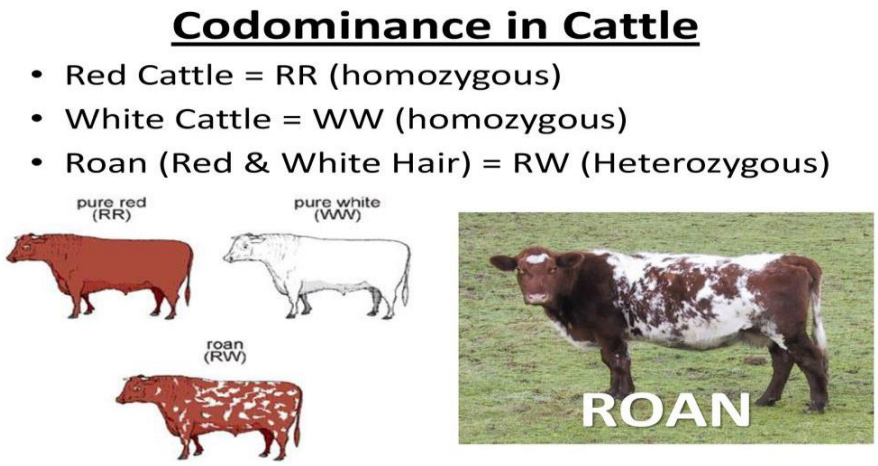
A dominant allele could not entirely suppress the other allele. Thus, a heterozygote is phenotypically differentiated from either homozygote (intermediate phenotype).

In snapdragon and *Mirabilis jalapa*, the hybridisation of pure-bred red-flowered and white-flowered plants results in pink-flowered F_1 seedlings (deviated from parental phenotypes), which are intermediate between the two parents. When the F_1 plant self-fertilises, the F_2 progeny displays three classes of plants in the ratio of 1 red: 2 pink: 1 white rather than 3:1.



Codominance

Coat colour in cattle is a classic example of co-dominance. There are two types, one with a red coat (skin with red colour hair) and the other with a white coat (with white hair). When red cattle (RR) is crossed with white cattle (WW), F1 hybrids (RW) have roan colour. Roans have a mixture of red and white colour hair. Therefore, the alleles that are able to express themselves separately when present together are called codominant alleles, and codominance is called the pattern of inheritance. On inbreeding, three kinds of cattle are provided by the roan hybrids-red, roan, and white in the 1:2:1 ratio.



1:2:1 ratio (1 Red, 2 Roan and 1 White)

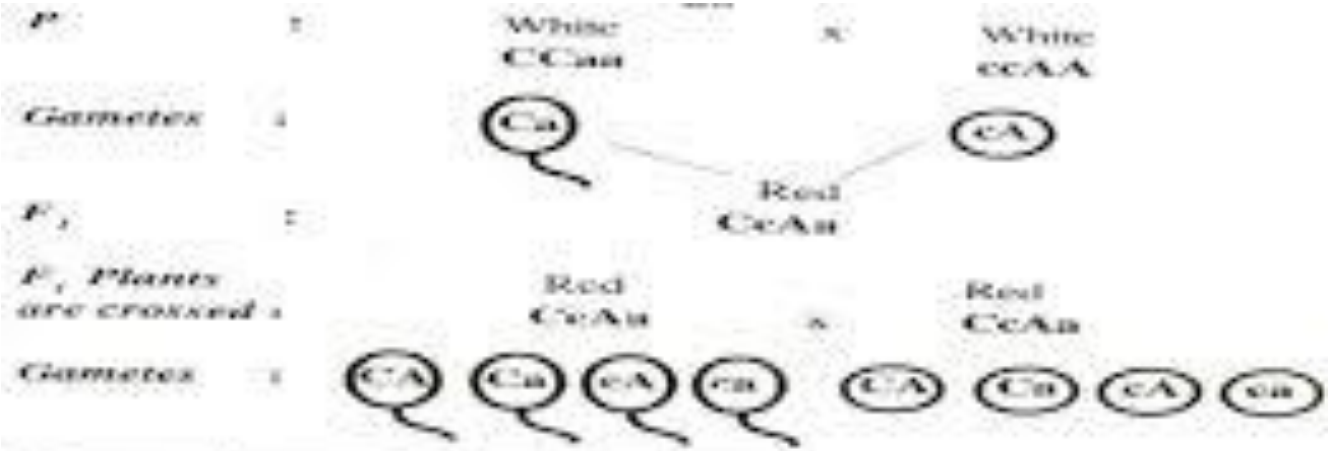
- Complete dominance is an allelic gene interaction that follows Mendel's laws of inheritance. According to this pattern, each gene has two alleles, with one allele completely dominant over the other.
- The presence of the dominant allele masks the effect of the recessive allele, leading to the expression of a single phenotype.

- Archibald Garrod's work in 1903 led to the discovery of complete dominance and its application to human diseases. Garrod studied alkaptonuria, a disease characterized by the excretion of homogentisic acid in urine. By analyzing family histories and urine samples, Garrod observed a 3:1 ratio in affected families, consistent with Mendelian inheritance.

Complementary genes (9:7 ratio)

Complementary genes are a kind of gene interaction in which two genes interact to produce a specific phenotype or observable trait. The term complementary refers to the relationship that exists between the two genes which make up a phenotype. The word complementary comes from the Latin word complement, which means "to complete." Complementary genes are therefore regarded as genes which work together to complete an apparent result.

In **sweet pea** (*Lathyrus odoratus*) two varieties of white flowering plants were seen. Each variety bred true and produced white flowers in successive generations. According to Bateson & Punnett, when two such white varieties of sweet pea were crossed, the offspring were found to have purple coloured flowers in F1 but in F2generation 9 were purple and 7 white. This is again a modification of 9:3:3:1 ratio, where only one character i.e., flower colour is involved. It is clear in the above example that for the production of the purple flower colour both complementary (C and P) genes are necessary to remain present. In the absence of either genes (C or P) the flowers are white. Thus, it is clear that genes C and P interact and presence of both is essential for the purple colour in the flower. These types of genes in which one gene complements the action of the other gene, constitute complementary genes or factors. (Complementation between two non-allelic genes (C and P) are essential for production of a particular or special phenotype i.e., **complementary factor)**



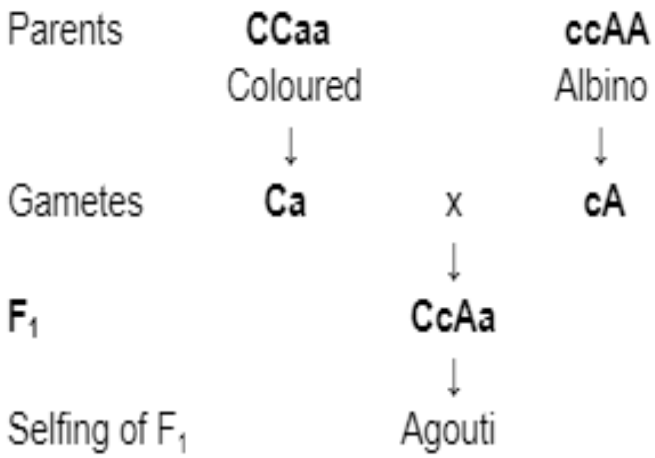
Gametes	CA	Ca	cA	ca
CA	CCAA Red	CCAa Red	CcAA Red	CcAa Red
Ca	CCAa Red	CCaa White	CcAa Red	Ccaa White
cA	CcAA Red	CcAa Red	ccAA White	ccAa White
ca	CcAa Red	Ccaa White	ccAa White	ccaa White

Fig. 5.1 : Inheritance of flower colour in sweet pea (Lathyrus odoratus)

Supplementary Gene Interaction (9:3:4 ratio)

In supplemental gene interaction, the phenotypic effect is produced by the dominant allele of one of the two genes regulating a character. However, the dominant allele of another gene has no independent phenotypic effect.

Therefore, it changes the phenotypic effect brought on by the first gene when it coexists with the dominant allele. For example, Agouti (grey) coat development in mice.



F_2 Generation:

	CA	**Ca**	**cA**	**ca**
CA	CCAA (Agouti)	CCAa (Agouti)	CcAA (Agouti)	CcAa (Agouti)
Ca	CCAa (Agouti)	CCaa (Coloured)	CcAa (Agouti)	Ccaa (Coloured)
cA	CcAA (Agouti)	CcAa (Agouti)	ccAA (Albino)	ccAa (Albino)
ca	CcAa (Agouti)	Ccaa (Coloured)	ccAa (Albino)	ccaa (Albino)

Thus, the phenotypic ratio is 9 (Agouti):3 (coloured):4 (Albino).

Inhibitory factor (13:3 ratio)

: In this type of modification two pairs of genes are involved and one of the non-allelic dominant gene inhibits the expression of the other non-allelic dominant gene. Or A gene which inhibits the expression of an active allele situated at different locus is called as inhibitory gene. Thus, in inhibitory gene action one dominant

inhibitory gene prevents the expression of another dominant gene.

Plumage colour in poultry birds White leghorn and Wyandotte or Plymouth Rock

In poultry white leghorn birds are white but they have a gene for colour plumage ‘C’ which has recessive allele ‘c’. But ‘I’ is inhibiting factor it inhibits the allele for colour ‘C’ is present as epistatic allele, hence they are genetically coloured but phenotypically white. So the gene for colour is hypostatic in white leghorn. Whereas the white plumage gene ‘c’ of white Wyandotte or Plymouth Rock is recessive over the coloured varieties.

When a white leghorn hen IICC is crossed with white Wyandotte rooster iicc, produces only progeny with white plumage ‘IiCc’, since, inhibitor allele not allow the allele C to produce colour. When F1 individuals are interbreeding they produce white and colour birds in the ratio of 13: 3. In F 2 generation genotypes with allele ‘I’ are all white and with recessive allele ‘i’ are coloured. Whereas genotype iicc also white because c is recessive allele for colour allele ‘C. ultimately total number of white birds are 12+1=13 and coloured birds are 3.

Above cross can be illustrated as follows using checker board.

P- Generation Whiteplumage white leghorn X White plumage Plymouth Rock

Genotype IICC iicc

Gametes IC X ic

F_1 Generation IiCc- White plumage

F_1 on selfing IiCc X IiCc

The gametes produced by F_1 Progeny are IC,Ic,iC,ic

The result of **F_2 progeny** are **13-White 3- Coloured**

Gametes	IC	Ic	iC	ic
IC	IICC White	IICc White	IiCC White	IiCcWhite
Ic	IICcWhite	IIccWhite	IiCcWhite	IiccWhite
iC	IiCC White	IiCc White	iiCC Colour	iiCc colour
ic	IiCc White	Iicc White	iiCc Colour	iicc white

Duplicate factors (15:1 ratio)

Sometimes two pairs of genes located on different chromosomes determine the same phenotype. These genes are said to be duplicate of each other. The dominant triangular fruit shape of *Capsella bursa pastoris* (shepherd's purse) is determined by two pairs of genes, say A and B. If any of these genes is present in dominant form, the fruit shape is triangular. In double recessive forms the fruits are top shaped and thus we get a 15 (triangular): 1 (top shaped) ratio in F_2 generation.

P_1 Generation Triangular AABB × Top Shaped aabb

F_1 Generation AaBb × Triangular AaBb

♀ \ ♂	AB	Ab	aB	ab
AB	AABB Triangular	AABb Triangular	AaBB Triangular	AaBb Triangular
Ab	AABb Triangular	AAbb Triangular	AaBb Triangular	Aabb Triangular
aB	AaBB Triangular	AaBb Triangular	aaBB Triangular	aaBb Triangular
ab	AaBb Triangular	Aabb Triangular	aaBb Triangular	aabb Top shaped

– F_2

F_2 Generation = Triangular : 15, Top shaped : 1

Duplicate genes in *Capsella bursa pastoris*

LETHAL GENES

Lethal factors or lethal genes cause survivor defects in the **possessor leading death** when they are homozygous condition. They affect the possessor before attaining the full form. Lethal genes can also alter the basic 9:3:3:1 ratio .The lethal genes may be dominant or recessive. The dominant lethal genes are known to cause their effect either in homo condition or in heterozygous condition. But recessive genes are known to cause defect only homozygous condition, hence their appearance is rare to see.

Dominant Lethal genes: Example: **Yellow Mice.**

The yellow body color in mice is dominant over brown, but the yellow mice are never true breeding. When yellow mice are inbred, (Cross – 1) the progeny consists of yellow and brown mice in the ratio 2:1 which does not fit any of the Mendelian expectations. Moreover, the litter size after inbreeding is smaller by one fourth as compared to litter size resulting from a cross between yellow and brown. When yellow mice were backcrossed to true breeding brown mice (Cross 2), only heterozygous yellow mice were obtained. The reason for not finding homozygous mice was explained by a French geneticist L. Cuenot. He sacrificed Yy pregnant females after inbreeding and examined the embryos to determine if death occurred in embryonic stages or not. Indeed, one-fourth of the embryos were observed to die in late stages of development. Thus only heterozygous yellow and brown mice in the ration 2:1 were being born. The ratio 1:2:1 expected when a cross between two heterozygous is made was never obtained proving the lethal expression of the homozygous yellow gene.

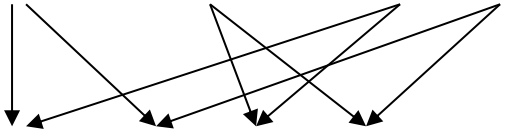
On crossing yellow mice with non-yellow mice production of yellow and non-yellow offspring's in the ratio of 1: 1 resulted. (Cross-2)From this it is clear that yellow colour is dominant over non yellow and that yellow mice are always heterozygous. According to Mendelian ratio 1 pure yellow YY : 2 hybrid yellow Yy : 1 pure non yellow , where as in the above cross we find two yellow heterozygous Yy and one pure non yellow yy . This is because the fact that the homozygous condition produce a lethal effect in the embryo.

Cross 1:

P- generation Yellow heterozygous X Yellow heterozygous

Genotype Yy Yy

Gametes Y y Y y



F1 progeny YY Yy Yy yy

Homozyugous Yellow Heterozygous yellownon yellow

1 2 1

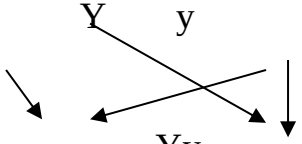
Dies Survives Survives

Cross -2:

P- Generation Yellow heterozygous X NonYellow(Brown)

Genotype Yy yy

Gametes Y y y



F1 progeny Yy yy

Heterozygous non yellow

Yellow

1 1

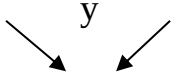
Survives survives

Cross: 3.

P- Generation Non Yellow X NonYellow(Brown)

Genotype yy yy

Gametes y y



F1 progeny yy

Non yellow Survives

Chapter 3 Multiple Alleles

Blood Group System

Karl Landsteiner (1900), discovered the ABO blood group system. In his experiments, he mixed different blood types and noted that the plasma from certain blood type produced agglutinates or formed clusters which were caused by the absence of molecules on red blood cells and resulting in antibodies to defeat that molecule. He then made a note of the agglutination and divided the blood types into 4 different groups. For the discovery of ABO blood group, he was awarded the Nobel Prize.

The blood grouping system is pivotal in blood transfusion. Our immune system recognizes another blood type as foreign and attacks it if introduced in the body causing a ***transfusion reaction***. Any inappropriate match with the Rh and ABO blood types, causes the most serious and life-threatening transfusion reactions. Therefore, before blood transfusion, it is suggested to have a blood group checked.

What are ABO and Rh blood groups?

During the blood transfusion, the two most important group systems examined are the ***ABO-system*** and the ***Rhesus system***.

The ABO blood group system consists of 4 types of blood group – A, B, AB, and O and is mainly based on the antigens and antibodies on red blood cells and in the plasma. Both antigens and antibodies are protein molecules in which antigens are present on the surface of Red Blood Cells and antibodies are present in the plasma which is involved in defending mechanisms.

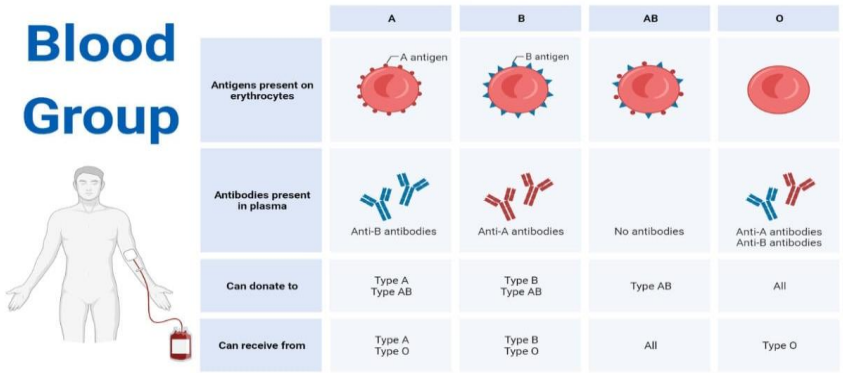
On the other hand, the Rh blood group system consists of 50 defined blood group antigens. In the Rh system, the most important antigens are D, C, c, E, and e. The ABO and Rh blood systems are discussed in detail below.

1. ABO blood Group system

The basis of ABO grouping is of two antigens- Antigen A and Antigen B. The ABO grouping system is classified into four types based on the presence or absence of antigens on the red blood cells surface and plasma antibodies.

- **Group A** – contains A antigen and antibody B.
- **Group B** –contains B antigen and antibody A.
- **Group AB** –contains both A and B antigen and no antibodies (neither A nor B).
- **Group O** – contains no antigen but have both antibodies A and B.

The ABO group system is important during blood donation or blood transfusion as mismatching of blood group can lead to clumping of red blood cells with various disorders. It is important for the blood cells to match while transfusing i.e. donor-recipient compatibility is necessary. For example, a person of blood group A can receive blood either from group A or O as there are no antibodies for A and O in blood group A.



As shown in the above table, individuals of blood group O are called as ***universal donors***, whereas individuals of blood group AB are ***universal recipients***.

- If you have type A blood, you can only receive types A and O blood.
- If you have type B blood, you can only receive types B and O blood.
- If you have type AB blood, you can receive types A, B, AB, and O blood.
- If you have type O blood, you can only receive type O blood.
- If you are Rh+, you can receive Rh+ or Rh- blood.
- If you are Rh-, you can only receive Rh- blood.

Inheritance of blood groups:

Blood groups are inherited from both parents. The ABO blood type is controlled by a single gene (the ABO gene) with three types of alleles inferred from classical genetics: *i*, I^A, and I^B. The *I* designation stands for **isoagglutinogen**, another term for antigen. The I^A allele gives type A, I^B gives type B, and *i* gives type O. As both I^A and I^B are dominant over *i*, only *ii* people have type O blood. Individuals with I^AI^A or I^Ai have type A blood, and individuals with I^BI^B or I^Bi have type B. I^AI^B people have both phenotypes, because A and B express a special dominance relationship: codominance, which means that type A and B parents can have an AB child. A couple with type A and type B can also have a type O child if they are both heterozygous (I^Bi,I^Ai).

Medico legal significance of blood group:

Although blood group studies cannot be used to prove paternity, they can provide unequivocal evidence that a male is not the father of a particular child. Since the red cell antigens are inherited as dominant traits, a child cannot have a blood group antigen that is not present in one or both parents. For example, if the child in question belongs to group A and both the mother and the putative father are group O, the man is excluded from paternity. The table shows the phenotypes (observed characters) of the offspring that can and cannot be produced in the

marriages on the ABO system, considering only the three alleles (alternative genes) *A*, *B*, and *O*. Similar inheritance patterns are seen in all blood group systems. Furthermore, if one parent is genetically homozygous for a particular antigen—that is, has inherited the gene for it from both the grandfather and grandmother of the child—then that antigen must appear in the blood of the child. For example, on the MN system, a father whose phenotype is M and whose genotype is *MM* (in other words, a man who is of blood type M and has inherited the characteristic from both parents) will transmit an *M* allele to all his progeny.

Exclusions of paternity on the ABO system

Marriage between	**Blood gr possible in children**	**Blood gr impossible in children**
O × O	O	A, B, AB
O × A	O, A	B, AB
O × B	O, B	A, AB
O × AB	A, B	O, AB

Exclusions of paternity on the ABO system

Marriage between	Blood gr possible in children	Blood gr impossible in children
A × A	O, A	B, AB
A × B	O, A, B, AB	
A × AB	A, B, AB	O
B × B	O, B	A, AB
B × AB	A, B, AB	O
AB × AB	A, B, AB	O

2. Rh Blood Group System

In addition to the ABO blood grouping system, the other prominent one is the Rh blood group system. About two-thirds of the population contains the third antigen on the surface of their red blood cells known as ***Rh factor*** or ***Rh antigen***; this decides whether the blood group is positive or negative. If the Rh factor is present, an individual is ***rhesus positive*** (Rh+ve); if an Rh factor is absent individual is ***rhesus negative*** (Rh-ve) as they produce Rh antibodies. Therefore, compatibility between donor and individual is crucial in this case as well.

Erythroblastosis foetalis:

Erythroblastosis foetalis is a hemolytic disorder due to Rh (D) incompatibility between the mother and the developing fetus inside her womb. It results in the destruction of the RBCs of the developing fetus.

The foetal RBCs can freely move across the placenta and enter the maternal circulation during foetal development (pregnancy) and child delivery. If the foetus has Rh +ve blood and the mother has Rh –ve blood, then the Rh antigens in the child can trigger an immune reaction in the mother's circulation. The infant's blood may enter the mother's circulation during delivery and also result in a similar immune reaction. Upon the introduction of the foetus's Rh antigens, the mother's body begins to synthesize Rh antibodies. The synthesized Rh antibodies will then attack the foetus's RBCs and cause the destruction of the foetus's RBCs leading to death or haemolytic anaemia in the fetus. The Rh incompatibility between the mother and her first child may be mild and there is rarely serious harm to the foetus and new-borns. However, if there is incompatibility during the second pregnancy also, then the developing infant has a higher chance to suffer from severe erythroblastosis (destruction of RBCs).

Now there are treatments or medication that prevents from the development of Rh antibodies in the mother's body.

Chapter 4 Chromosomes

Chromosomes are filamentous bodies which are typically present in the nucleus and which become visible during cell division. They are the carriers of the genes or units of heredity. Chromosomal mutation represents the structural change in the chromosome which appears phenotypically. However, in this chapter, you will able to read about mutation, chromosomal mutation, Translocation, Inversion, Deletion, Duplication, Euploidy, Aneuploidy, and Polysomy etc.

Introduction:-

During the cell divisions, the chromatin network of the interphase nucleus condenses to form thick rod like structures known as chromosomes. The name chromosome was given by Waldeyer in 1888. The chromosomes play an important role in transmission of heredity characters from one generation to another. Thus chromosomes can be defined as: "The individualized protoplasmic units present in definite number, capable of selfreproduction, maintaining their individuality, morphology and physiological properties throughout, play an important role in heredity". The term mutation was first introduced by a well-known Dutch Botanist Hugo de Vries in1902 while working on a plant Evening primrose (*Oenothera lamarckiana*). He used word mutation for spontaneous inheritable changes which occur suddenly and alter the phenotype of an organism. He performed several experiments on plant evening primrose up to eight years continuously and concluded that suddenly occurred spontaneous inheritable changes in plants and animals are the principle cause of "origin of new species". However, in present condition mutation may be defined as "large spontaneous inheritable sudden changes in the genotype which alter the phenotype of an individual". These mutations may be spontaneous or induced mutation.

Structure of Chromosome

Chromosome number varies from species to species. However, it is constant for a particular number. Size is constant for every species. It ranges from 0.1µ to 30 µ in length. The shape varies at different phases of cell

division. They may be rod-shaped twisted or spiral, curved or filamentous. Each chromosome is comprised of following parts: 1. Pellicle and Matrix 2. Chromonemata 3. Centromere 4. Chromomeres 5. Satellite bodies.

Fig.2.1 morphology of Chromosome

However, chromosomes may be classified in the following category on the basis of centromeres present. 1. Monocentric: The chromosomes which have only one centromere are called monocentric. 2. Dicentric: The chromosomes having two centromeres are termed as dicentric chromosomes. 3. Polycentric: In these, the chromosomes possess many centromeres. 4. Acentric: In these, the chromosomes lack centromeres. Like number, the position of centromeres also varies. Depending upon the position of the centromere the chromosomes are of following types:

1. Acrocentric: When the centromere occupies subterminal end, it is called acrocentric. It is also rod-like in which one arm is very much smaller than the other.

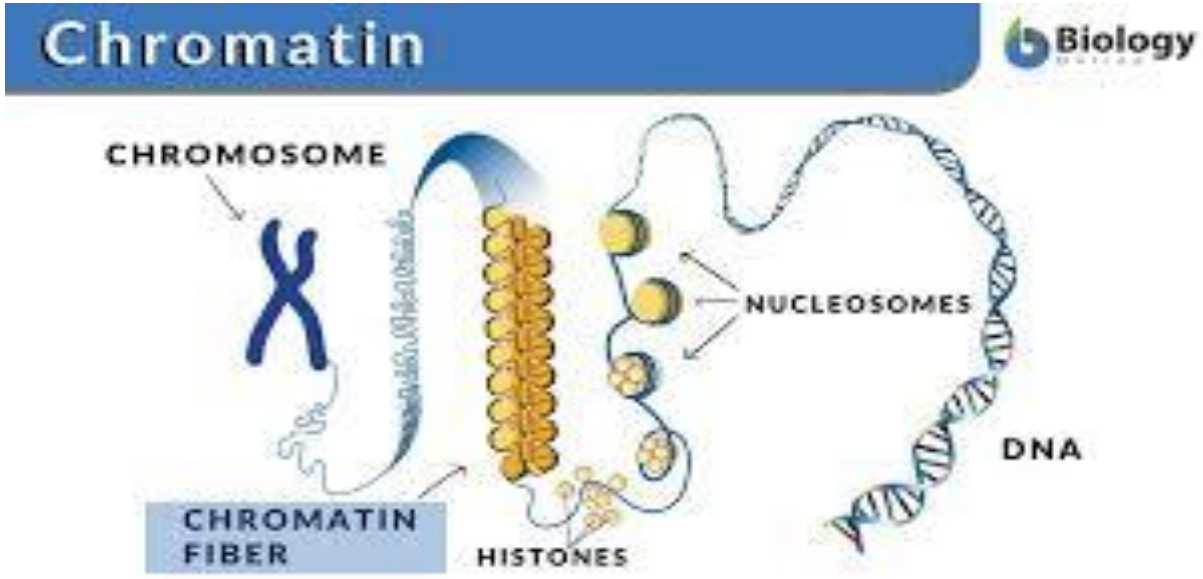
3. Submetacentric: In submetacentric chromosome, the unequal in length giving J or L shaped appearance to the chromosome.

4. Metacentric: In metacentric chromosomes, the centromere lies exactly in the center of the chromosome. These chromosomes are V-shaped having equal arms.

4. Telocentric: When the centromere is situated on the proximal end or terminal end, it is called telocentric. This type of chromosome is rod-like.

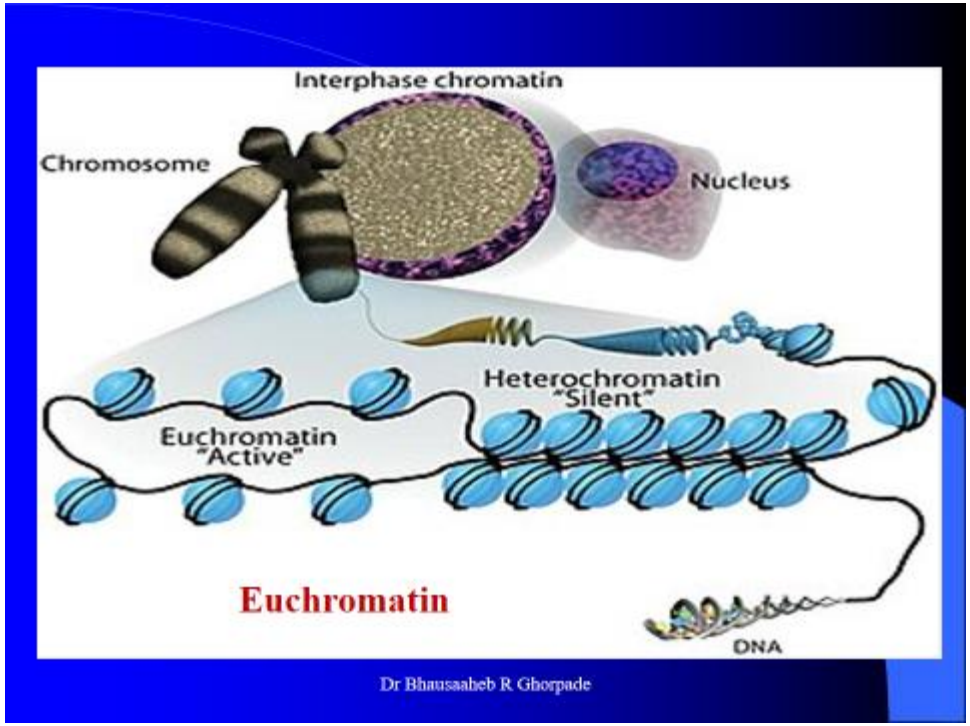
Chromatin (Euchromatin and Heterochromatin)

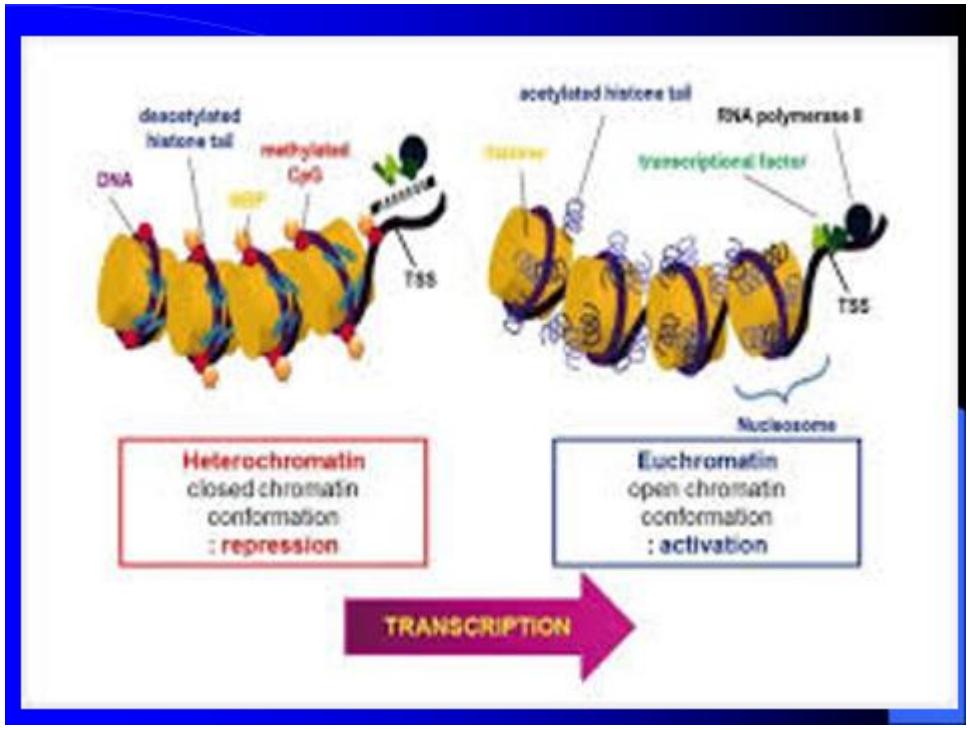
Chromatin is therefore, a complex of macromolecules such as DNA, RNA and proteins, which is assembled together. The primary protein components of chromatin are histones that compact the DNA. Chromatin is only found in eukaryotic cells (more differentiated cells, with well-defined nuclei). Prokaryotic cells have a different organization of their DNA, in which the strand of DNA is localized within the nucleoid region. The prime reason for packing the DNA into a chromatin assembly is to fit the molecule in the much smaller volume of the cell nucleus. DNA, organized as chromatin, is found in two forms along the length of the chromosomes that reflect the level of activity of the cell:



1. Euchromatin:

During interphase, the chromosomes are not distinctly assembled. After mitosis has been completed, most of the chromatin which is in highly compacted mitotic chromosome form, returns to its diffuse interphase condition. Approximately 10 percent of the chromatin, however, generally remains in a condensed, compacted form throughout interphase. This compact, densely stained chromatin is seen at the periphery of the nucleus. Chromatin that remains compacted during interphase is called heterochromatin to distinguish it from euchromatin, which returns to a dispersed state.


Interphase chromatin
Chromosome
Nucleus
Heterochromatin
"Silent"
Euchromatin
"Active"
Euchromatin
DNA
Dr Bhausaaheb R Ghorpade



2. Heterochromatin

In prokaryotes, euchromatin is the only form of chromatin present, there is no heterochromatin. This suggests that the heterochromatin structure evolved later along with the nucleus, possibly as a mechanism to manage the increasing genome size. Heterochromatin thus appears as small, darkly staining, irregular particles scattered throughout the nucleus or accumulated adjacent to the nuclear envelope. On the other hand, heterochromatin is a tightly packed form of DNA.It is commonly found on the peripheral areas of the nucleus. According to some studies, there are probably two or more states of heterochromatin. Inactive satellite sequences are the main constituents of heterochromatin. Heterochromatin is responsible for gene regulation and protection of chromosomal integrity

Giant Chromosomes

Some tissues of certain organisms contain chromosomes which differ significantly from normal ones in terms of either morphology or function; such chromosomes are referred to as specialchromosomes.
1)Giant Chromosomes or Polytene Chromosome:
2)Lampbrush Chromosomes:

Giant Chromosomes or Polytene Chromosome: :

Giant chromosomes are found in salivary glands of larvae, gut epithelium, Malphigian tubules and some Diptera, e.g., Drosophila, Chironomous, Sciara, Rhyncosciara etcThese chromosomes are very long and very thick, hence they are known as giant chromosomes. They were first discovered by Balbiani (1881) in dipteran salivary glands, The giant chromosomes are somatically paired. The giant chromosomes have a distinct pattern of transverse banding which consists of alternate chromatic and achromatic regions. puffs, known aschromosome puffs or Balabiani rings, which are associated with active RNA synthesis. . In Drosophila melanogaster, the giant chromosomes radiate as five long and one short arm from a single more or less amorphous mass known as chromocentre. The chromocentre is formed by fusion of the centromeric regions of all the chromosomes and, in males, of the entire Y chromosomes. The short arm radiating from the chromocentre represents chromosome IV, one of the long arms is due to the X-chromosome, while the remaining four long arms represent the arms of chromosome II and III. The total length of D. melanogaster giant chromosomes is about 2000µ.

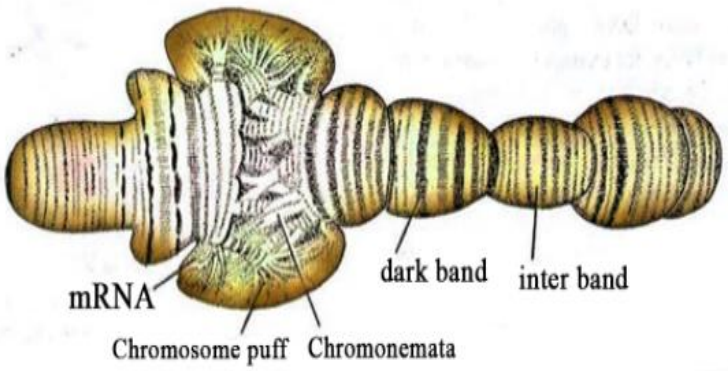
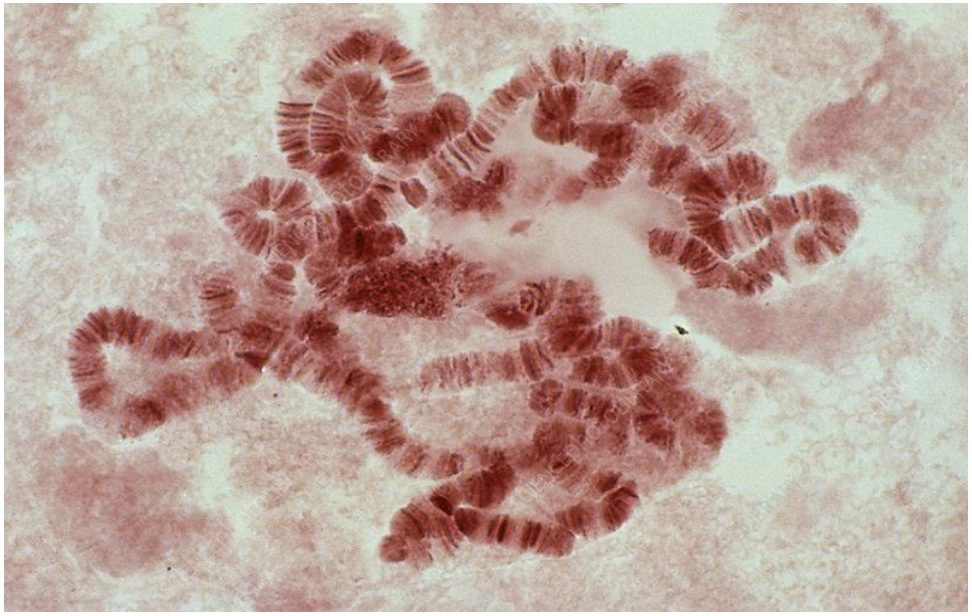


Fig.Polytene chromosome of an insect,showing bands and interbands and a puff or Balbiani ring.



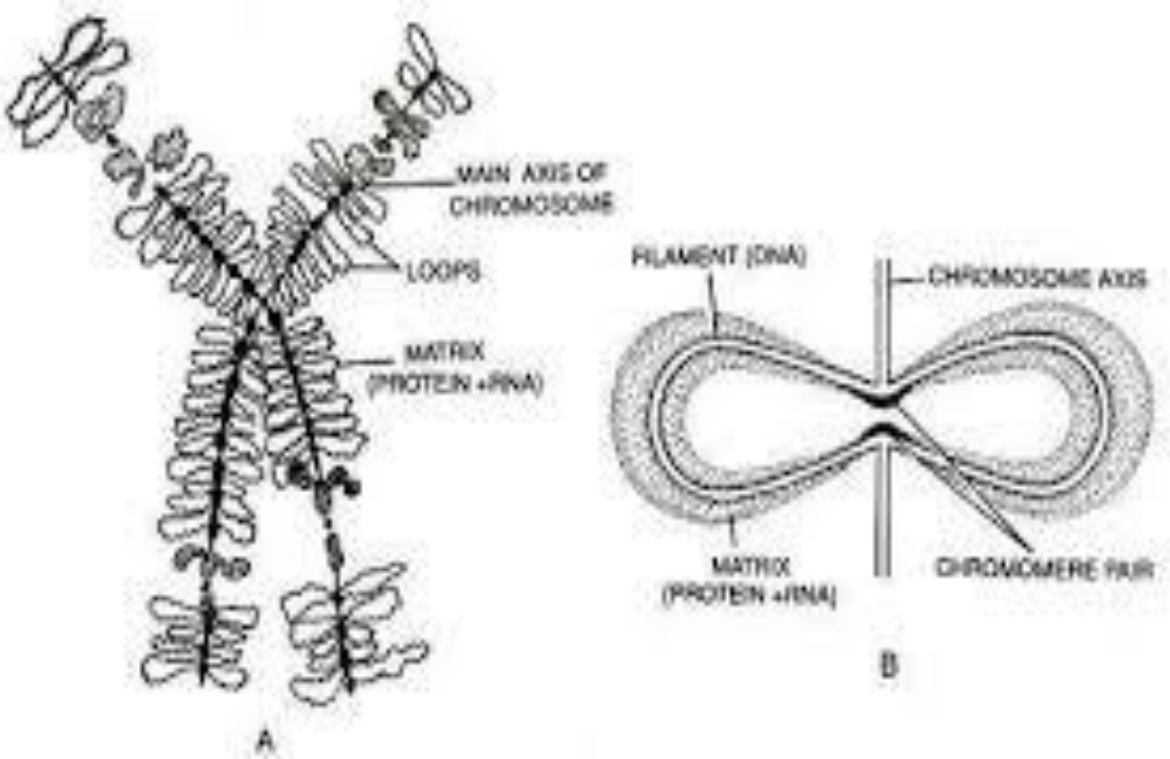
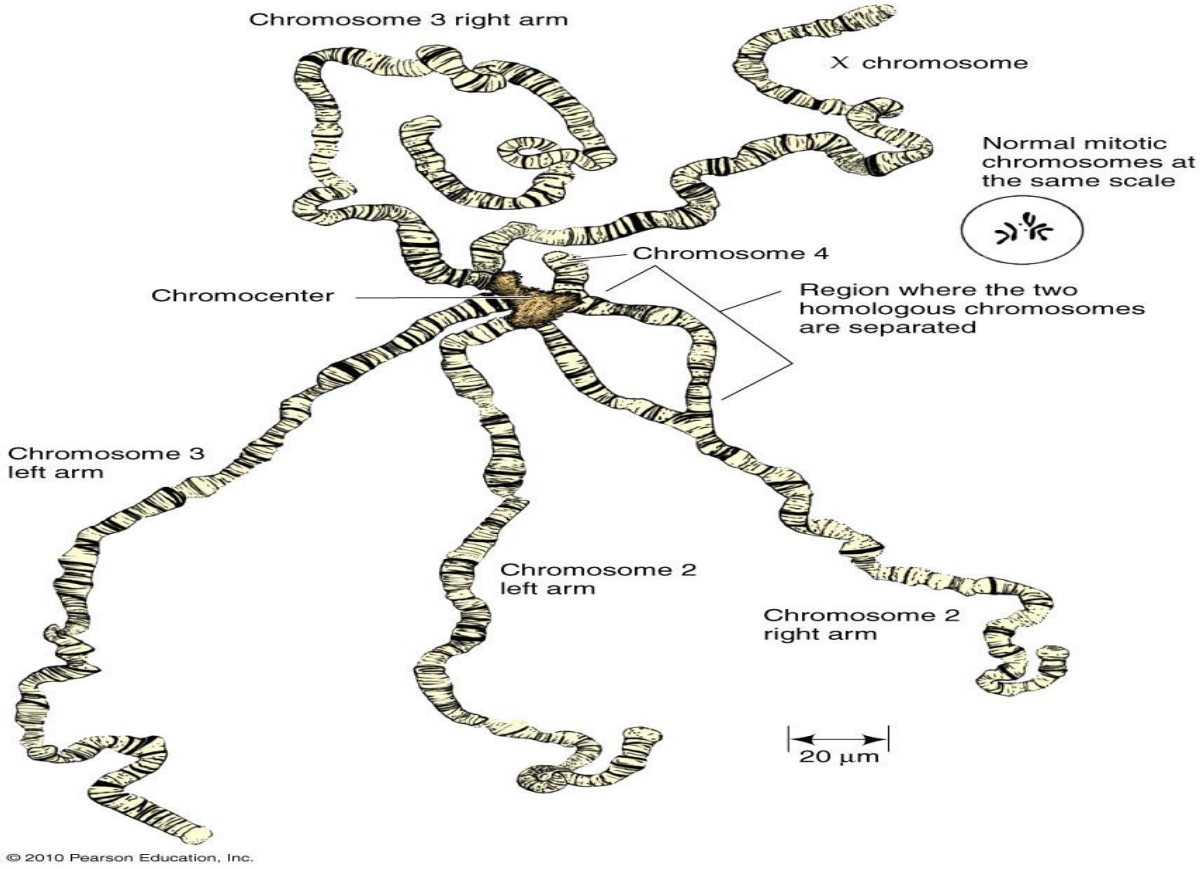


Fig. 8.58. Lampbrush chromosome . A, Enlarged view of a part of lampbrush chromosome. B, One loop of a lampbrush chromosome.

Lampbrush chromosomes are found in oocytes of many invertebrates and all vertebrates, except mammals.

These chromosomes are most distinctly observed during the prolonged diplotene stage of oocytes. During diplotene, the homologous chromosomes begin to separate from each other, remaining in contact only at several points along their length. Each chromosome of a pair has several chromomeres distributed over its length; from each of a majority of the chromomeres generally a pair of lateral loops extends in the opposite directions perpendicular to the main axis of the chromosome. In some cases, up to 9 pairs of loops may emerge from a single chromomere. These lateral loops give the chromosomes the appearance of a lampbrush . These chromosomes are extremely long, the pairs of loops are produced due to uncoiling of the two chromatin fibres which makes their DNA available for transcription (RNA synthesis). Thus each loop represents one chromatid of a chromosome and is composed of one DNA double helix. One end of each loop is thinner (thin end) than the other end (thick end). There is extensive RNA synthesis at the thin ends of loops, while there is little or no RNA synthesis at the thick end.

The chromatin fibre of the chromomere is progressively uncoiled towards the thin end of a loop; the DNA in this region supports active RNA synthesis but later becomes associated with RNA and protein to become markedly thicker. The DNA at the thick end of a loop is progressively withdrawn and reassembled into the chromomere. The number ofpairs of loops gradually increases in meiosis till it reaches maximum in diplotene. As meiosis proceeds further, number of loops gradually decreases and the loops ultimately disappear due to disintegration rather than reabsorption back into the chromomere. Loops represent the sites of gene action (transcription), and the function of lampbrush chromosomes is to produce the large numbers and quantities of proteins and RNA’s stored in eggs.


Chromosome 3 right arm
X chromosome
Normal mitotic chromosomes at the same scale
Chromosome 4
Chromocenter
Region where the two homologous chromosomes are separated
Chromosome 3 left arm
Chromosome 2 left arm
Chromosome 2 right arm
20 µm
© 2010 Pearson Education, Inc.

Structural changes in chromosomal morphology:

1. Deletion:-

When any part or section of a chromosome which containing either one gene or block of genes, is being lost called deletion or deficiency. Various type of structural changes are occurring during meiosis (reduction division) and these changes may be recognized in the pairing of homologous chromosomes (synapsis) in zygotene substage of prophase –I of meiosis. When a chromatid breaks at two places and the end portion fuse leaving out the central point, called intercalary deletion, while terminal, called as deficiency or deletion. When a deletion occurs in one member of a homologous pair, the members will become unequal in length. Genic balance is usually disturbed due to deletion and this affects the phenotype. Deletion can be recognized by distortions of chromosomes during meiotic pairing of homologous chromosomes. Due to a terminal deletion one of the paired chromosomes appears to be much longer than the other, whereas due to intercalary deletion, the normal chromosome forms a loop near the deficient region of its homolog as an only identical regions pair with each other. In present A chromosome part 3, 4 is becoming deleted and during the synapsis, chromosome B becomes curved.

2. Duplication

The presence of a part of a chromosome in excess of the normal complement is known as duplication. A deleted part or segment of broken part or section of a chromosome attaches itself to a normal homologous or non-homologous chromosome in the presence of a centromere it behaves like an independent chromosome and gets included in an otherwise normal nucleus.

Depending on the mode of joining of the duplicated region to a chromosome or its independent existence,

duplication can be of the following types: A. Extra Chromosomal: In the presence of a centromere, the duplicated part of the chromosome may behave as an independent chromosome. B. Tandem: in this case, the duplicated region is situated just by the side of the normal corresponding section of the chromosome and the sequences of genes are the same in the normal and duplicated regions. C. Reverse Tandem: in this case, the sequence of genes in the duplicated section of a chromosome is just the reverse of the normal sequence. D. Displaced: here, the duplicated section is not adjacent to the normal section. E. Transposed: in this case, the duplicated section is attached to a non-homologous chromosome. like deletions, duplications also result into unequal or looped out configuration at the time of pairing of homologous chromosomes.

3. Inversion:-

When a part or segment of the chromosome containing genes rotates on its own axis by 180 degrees called an inversion. Breakage and reunion both are essential for inversion. Sometimes, a chromosome may break at two points and then become reunited at the same point in a reverse order. For example, In chromosome A part 4, and 3 is inverted. During the pairing of chromosomes in zygotene substage of meiosis -I, chromosome a is become inverted at this point at 180 degrees as represented in C chromosome, while other B chromosomes in the form of D chromosome become curved at this portion. During pairing, repulsion occurs at the part where the genes do not match. This is also called as "position effect". Thus, as a result, there is neither a gain nor loss in the genes but a rearrangement of the sequence of the gene take place. Inversion may be of following two types: A. Paracentric inversion B. Pericentric inversion

A. Paracentric inversion: In this type of inversion, the centromere is located outside the inversion loop. When a cross over occurs within the loop, one product contains a centromere and the other does not. At anaphase, this results in an abnormal chromosomal ‘bridge’ and a loss of an entire chromosomal section.

B. Pericentric inversion: Here centromere is located inside the inversion loop. When a cross over between two chromatids occurs within the inversion loop, in the resulting chromatids there are some genes in double number

while others are missing. Due to this imbalance, the cell is not viable. Thus, if the normal sequence of genes in a chromosome is A B C D E F G, The sequence in paracentric and pericentric inversions will be A B C D G F E and A E D C B F G respectively. Inversion has been useful in establishing and maintaining heterozygous condition because in inversion heterozygotes crossing – over is suppressed and only parental progeny is produced. Recessive lethal can be of added advantage because heterozygotes for them are viable but homozygotes non-viable.

4. Translocation

Numerical changes in chromosomes

I)Euploidy:-

Variations that involve an entire set of chromosomes are known as euploidy. Euploids have one or more complete genomes, which may be identical with or distinct from each other. In the diploid state, two copies of the same genome are present in the somatic cells; it is represented as 2x. Euploid variations are designated with reference to the diploid (2x) state.

Some important euploid types are as following:

A. Monoploidy: Monopolids have a single basic set of chromosomes, e.g. 2n = X = 7 in barely and 2n = X = 10 in corn. Haploid, on the other hand, represent individuals with half the somatic chromosome number found in the normal individual. In haploids, each chromosome is represented only once due to which there is no zygotene pairing and all the chromosomes appear as univalent on a metaphase plate at meiosis -I. During anaphase - I, each chromosome moves independently of the other and goes to either of the two poles.

Haploids may originate:

1. Due to the parthenogenetic or androgenic development of gametes.

2. Due to chromosome loss in hybrid embryos and

3. By pollen culture. The most important use of haploids is in the production of homozygous diploids.

B. Polyploidy: In polyploids, each chromosome is represented by more than two homologs. Failure of normal mitotic divisions results into nuclei with increased sets of chromosomes. Depending on whether polyploids are produced by the multiplication of chromosome sets that are initially derived from a single species or from two different species, they are of two types, autopolyploids, and allopolyploids.

1. Autopolyploidy: Autopolyploids are those polyploids which have the same basic set of chromosomes multiplied. For instance, if a diploid species has two similar sets of chromosomes or genomes (AA), an autotriploid will have three similar genomes (AAA) and an autotetraploid four such genomes (AAAA).

2. Allopolyploidy: Polyploidy resulting from the doubling of chromosome number in a F1 hybrid derived from two quite different species is known as allopolyploidy. Allopolyploidy brings two different sets of chromosomes in F1 hybrid. Suppose A represent a set of chromosomes (genome) in species X, and B another genome in species Y. The F1 will then have one A genome and another B genome. The doubling of chromosomes in the F1 hybrid (AB) will give rise to a tetraploid with two A and two B genomes. Such polyploid is called allopolyploid or amphidiploid.

II) Aneuploidy:-

It is the presence of a chromosome number which is different than the multiple of basic chromosome number. This type of variation involves one or a few chromosomes but not the entire set. It is either due to loss of one or more chromosomes or due to the addition of one or more chromosomes to complete chromosome complement.

Aneuploidy is of the following types: 1. Monosomy 2. Nullisomy 3. Trisomy 4. Tetrasomy 1. 1. Monosomy

Monosomics represent the loss of a single chromosome from the diploid set, and they have the chromosome complement 2n -1. Since monosomies lack one complete chromosome, such aberration creates major imbalance and cannot be tolerated in diploids.

2. Nullisomy Nullisomics lack a single pair of the homologous chromosome; have the chromosome complement 2n - 2.

3. Trisomy Trisomies are those organisms that have an extra chromosome (2n + 1) which is homologous to one of the chromosomes of the complement. They are specifically useful in locating genes on a specific chromosome.

4. Tetrasomy Tetrasomy are those organisms which have an extra pair of homologous chromosomes and have the chromosome complement 2n = 2.

Polysomy:-

Polysomy is a condition found in many species, including fungi, plants, insects, and mammals, in which an organism has at least one more chromosome than normal, i.e., there may be three or more copies of the chromosome rather than the expected two copies. Most eukaryotic species are diploid, meaning they have two sets of chromosomes, whereas prokaryotes are haploid, containing a single chromosome in each cell. Aneuploids possess chromosome numbers that are not exact multiples of the haploid number and polysomy is a type of aneuploidy. A karyotype is the set of chromosomes in an organism and the suffix - somy is used to name aneuploid karyotypes. This is not to be confused with the suffix - ploidy, referring to the number of complete sets of chromosomes. Polysomy is usually caused by non - disjunction (the failure of a pair of the homologous chromosome to separate) during meiosis, but may also be due to a translocation mutation (a chromosome abnormality caused by rearrangement of parts between non-homologous chromosomes). Polysomy is found in many diseases, including Down syndrome in humans where affected individuals possess three copies (trisomy) of chromosomes. Polysomic inheritance occurs during meiosis when chiasmata form

between more than two homologous partners, producing multivalent chromosomes. Autopolyploids may show polysomic inheritance of all the linkage groups, and their fertility may be reduced due to unbalanced chromosome numbers in the gametes. In tetrasomic inheritance, four copies of a linkage group rather than two (tetrasomy) assort two-by-two.

TYPES OF POLYSOMY:- Polysomy types are categorized based on the number of extra chromosomes in each set, noted as a diploid (2n) with an extra chromosome of various numbers. For example, a polysomy with three chromosomes is called a trisomy, a polysomy with four chromosomes is called tetrasomy,

Chapter 5 Sex Determination

Introduction:

There are two different types of sexes, which participate in sexual reproduction. It is natural to find it confounding as from which sex the baby inherits, that results in the sex of the child. There are several other procedures followed to determine the sex of a new-born baby. Based on the environmental signals the sex of a baby can be determined. In a few animal species, temperature plays a major role in sex determination. In other animals, like snails, it is possible to change sex as they are not genetically processed. In human beings, the sex of an individual is genetically determined. In other words, the genes which are inherited from their parents decide the sex of the child.

SEX DETERMINATION

Determination of sex at the early stages of life is called as sex determination.

Sex determination on the basis of fertilisation

There are three types of sex determination on the basis of fertilisation

Progamic SyngamicEpigamic

ProgamicWhen sex is determined before fertilisation. Then, it is called Progamic sex determination.

Example - Male honeybee etc.

SyngamicWhen sex is determined during fertilisation . Then, it is called Syngamic sex determination.

Example - Most of the plants and animals.

EpigamicWhen sex is determined after fertilisation . Then, it is called as Epigamic sex determination.

Example - Female honeybees etc.

Discovery of Sex chromosomes

Sex chromosomes (also referred to as allosomes, heterotypical chromosome, onosomes, heterochromosomes, or idiochromosomes) are chromosomes that carry the genes that determine the sex of an individual. The human sex chromosomes are a typical pair of mammal allosomes. They differ from autosomes in form, size, and behavior. Whereas autosomes occur in homologous pairs whose members have the same form in a diploid cell, members of an allosome pair may differ from one another.

Nettie Stevens and Edmund Beecher Wilson both independently discovered sex chromosomes in 1905. However, Stevens is credited for discovering them earlier than Wilson.

Difference between **Autosomes and Sex Chromosomes**

1. Autosomes **are the chromosomes that determine somatic characters such as body weight, length, etc. of an organism.**
2. Sex Chromosomes are the chromosomes that determine sex and sex-related hormonal traits.

Autosomes	Sex chromosomes
1. Determining somatic characters mainly involves the growth of an organism.	1. Determines the gender and sex-related traits.
2. All chromosomes are of the same size, that is, homologous.	2. Chromosomes are partially homologous.
3. Follows Mendelian Inheritance.	3. Shows Non-mendelian inheritance.
4. In humans, 22 pairs of autosomes are present.	4. In humans, only one pair of chromosomes

is present.

Chromosomal theory of the sex-determination

A chromosomal theory of the sex-determination system is a biological system that controls the event of sexual characteristics in an organism. Most organisms that create their offspring using sexual reproduction have two sexes– male and female.

- *A sex-determination system is a biological system that determines the development of sexual characteristics in an organism.*
- *The chromosome theory of sex determination proposed by McClung.*
- *According to this theory, chromosomes play a major role in determination of sex.*
- *In diploid organisms, a pair of chromosomes determines the sex of individual.*
- *They are called the sex chromosomes or allosomes or heterosomes.*

Theory of heterogenesis for sex determination was proposed by Correns.

The **XX/XY sex-determination system**

It is likely the most familiar, as it is found in humans. The XX/XY system is found in most other mammals, as well as some insects. In this system, most females have two of the same kind of sex chromosome (XX), while most males have two distinct sex chromosomes (XY). The X and Y sex chromosomes are different in shape and size from each other, unlike the rest of the chromosomes (autosomes), and are sometimes called allosomes. In some species, such as humans, organisms remain sex indifferent for a period of time after fertilization; in others, however, such as fruit flies, sexual differentiation occurs as soon as the egg is fertilized.

XX/X0 sex chromosome system,

In the variant of the XY system, females have two copies of the sex chromosome (XX) but males have only one (X0). The *0* denotes the absence of a second sex chromosome. Generally in this method, the sex is determined by amount of genes expressed across the two chromosomes. This system is observed in a number of insects, including the grasshoppers and crickets of order Orthoptera and in cockroaches (order Blattodea). A small number of mammals also lack a Y chromosome. These include the Amami spiny rat (*Tokudaia osimensis*) and the Tokunoshima spiny rat (*Tokudaia tokunoshimensis*) and *Sorex araneus*, a shrew species. Transcaucasian mole voles (*Ellobius lutescens*) also have a form of XO determination, in which both sexes lack a second sex chromosome. This mechanism of sex determination is not currently well-understood.

ZW sex-determination system

It is found in birds, some reptiles, and some insects and other organisms. The ZW sex-determination system is reversed compared to the XY system: females have two different kinds of chromosomes (ZW), and males have two of the same kind of chromosomes (ZZ). Like the X/Y system, there are variations on this system. For example, there are moths and butterflies that are ZW, but some have been found female with Z0, as well as female with ZZW. Also, while mammals deactivate one of their extra X chromosomes when female, it appears that in the case of Lepidoptera (butterflies and moths), the males produce double the normal amount of enzymes, due to having two Z chromosomes. Because the use of ZW sex determination is varied, it is still unknown how exactly most species determine their sex.

Despite the similarities between the ZW and XY systems, these sex chromosomes evolved separately. In the case of the chicken, their Z chromosome is more similar to humans' autosome 9. The chicken's Z chromosome also seems to be related to the X chromosome of the platypus. When a ZW species, such as the Komodo dragon, reproduces parthenogenetically (asexually), usually only males are produced. This is due to the fact that the haploid eggs double their chromosomes, resulting in ZZ or WW. The ZZ become males, but the WW are not

viable and are not brought to term.

In both XY and ZW sex determination systems, the sex chromosome carrying the critical factors is often significantly smaller, carrying little more than the genes necessary for triggering the development of a given sex.

Honey bee method (Haplodiploidy)

Haplodiploidy is found in insects belonging to Hymenoptera, such as ants and bees. Sex determination is controlled by the zygosity of a complementary sex determiner (*csd*) locus. Unfertilized eggs develop into haploid individuals which have a single, hemizygous copy of the *csd* locus and are therefore males. Fertilized eggs develop into diploid individuals which, due to high variability in the *csd* locus, are generally heterozygous females. In rare instances diploid individuals may be homozygous, these develop into sterile males. The gene acting as a *csd* locus has been identified in the honeybee and several candidate genes have been proposed as a *csd* locus for other Hymenopterans. Most females in the Hymenoptera order can decide the sex of their offspring by holding received sperm in their spermatheca and either releasing it into their oviduct or not. This allows them to create more workers, depending on the status of the colony.

Gynandromorphs

Gynander or gynandromorphs are the organisms in which the body consists of both male and female parts. Such organisms showing both female and male characteristics are called gynanders or gynandromorphs.

The term is derived from the Greek words (gyne = woman; aner = man and morphe = form). Thus, in these animals one part of the body shows female and the other part male features. These occur in silkworms, bees and fruit flies. Gynandromorphs were first described in Drosophila by Morgan and Bridges.

Types of Gynandromorphs:

Depending upon the position of sex tissue, the gynanders may be of the following types:

1. Bilateral Gynanders:

Some times one half of the body shows female characters while other half shows male characters. Sex intermediates of this type are called bilateral gynanders.

2. Anterior-Posterior Gynanders:

In such gynanders anterior region of the animal body has the characteristics of one sex and posterior half region has the characteristics of the other sex.

3. Sex Piebalds:

In these gynandromorphs the body consists of female tissue having spots of male tissue scattered irregularly. There are certain cases in which a few cells of the body show sex difference.

Origin and Occurrence of Gynandromorphs:

The gynandromorphs are supposed to have produced mainly by two or three methods:

(i) By Elimination of X-chromosome:

Generally, gynander begins its development with two X's. But in later stage of cell division one X gets disappeared or lost in daughter cells. Two X chromosomes in the mitosis become divided in to four X daughters chromosomes.

One daughter cell receives two X's, and the other daughter cell gets only one X while fourth X becomes lost due to abnormal cell division. The daughter cells receiving two X's forms the female tissue while the other daughter cell receiving one X develops in to male tissue. The example is Drosophila.

(ii) By Retention (= holding) of Polar Nucleus in the Egg:

In silk worms female is XY. During Meiosis X and Y get separated, either of one X or Y going to egg and polocyte. But some times polar body nucleus remains in the egg along with egg nucleus.

Thus, these eggs will be bi-nucleate (XY). As a result of fertilization two sperm cells which contain X, fuse with X and Y chromosome separately giving rise to male (XX) and female (XY) tissues respectively.

In bees also bi-nucleate eggs are found either due to fusion of polar body nucleus with egg nucleus or parthenogenetically producing gynanders. If this bi-nucleate egg was fertilized by single X carrying sperm then, only one of the two nuclei will be fertilized and this one will give rise to female tissue. The unfertilized nucleus will give rise to male tissue.

Another possibility is that normal egg is fertilized by two X carrying sperm cells one of which combines with egg nucleus as usual, but the other one does not. The later then might give rise to haploid tissue, this would be male tissue.

The fertilized nucleus would give rise to female tissue. Muller described another possibility of the gynander formation in a parasitic wasp-Habrobracon. In this wasp, female are heterozygous (XY) and males are either homozygous or haploid (XX).

Chapter 6 Sex-linked Inheritance:

Sex-linked inheritance is a biological process which involves the transmission of traits or characters from parents to offspring. X and Y are two sex chromosomes that carry alleles at their gene loci. These traits that are being transferred from one generation to the next generation are present on autosomes or sex chromosomes, i.e., the X chromosome or the Y chromosome.

Sex-linked genes are located on the X chromosome result in X-linkage. Similarly, Y-linkage refers to the gene which is present on the Y chromosome. Since females are homogametic with XX chromosome and males have XY chromosome, the Y-linked traits are transmitted via males only.

There are certain diseases which are linked to the X chromosome but are recessive, where females act as a carrier if they have only one copy of defective genes, such as colour blindness, haemophilia, etc. Males are heterozygous; hence, they are more prone to get sex-linked disorders because only one defective copy of genes is sufficient to cause diseases.

Types of Sex-linked Inheritance

As described earlier, there are two types of sex-linked inheritance: X-linked and Y-linked inheritance.

X-linked Inheritance

The X chromosome is larger than the Y chromosome. Any disorder or trait that is transmitted from the X chromosome is termed an X-linked inheritance. X-linked inheritance can either be recessive or dominant.

X-linked Recessive Inheritance	X-linked Dominant Inheritance
This trait is more common in males as they contain only one X chromosome.	Both males and females are affected by this type of disorder.
Haemophilia A and haemophilia B are examples of X-linked recessive inheritance.	Examples of X-linked dominant inheritance include Incontinentia pigmenti.

Y-linked Inheritance

If the mutated gene is present in the Y chromosome, then this is referred to as Y-linked inheritance. Since the Y chromosome is present in males only, Y-linked disorders are passed from fathers to male offspring. Hypertrichosis (presence of long dark hair on the ears) is an example of Y-linked inheritance.

B. SEX-LINKED DISORDERS Sex-linked recessive inheritance is a mode of inheritance in which a mutation in a gene on the X - chromosome causes the phenotype to be expressed in males (who are necessarily hemizygous for the gene mutation because they have one X and one Y-chromosome and in females who are homozygous for the gene mutation. X-linked inheritance means that the gene causing the trait or the disorder is located on the X chromosome. Females have two X chromosomes, while males have one X and one Y chromosome. Carrier females who have only one copy of the mutation do not usually express the phenotype, although differences in X- chromosome inactivation can lead to varying degrees of clinical expression in carrier females since some cells will express one X- allele and some will express the other. The current estimate of sequenced X-linked genes is 499 and the total including vaguely defined traits is 983. However, some important sex-linked disorders are as following:

1. Hemophilia 2. Color blindness 3. Muscular dystrophy 1. HEMOPHILIA Hemophilia is an uncommon hereditary bleeding disorder which primarily affects male but is transmitted by females.

Haemophilia also spelled hemophilia, is a mostly inherited genetic disorder that impairs the body's ability to make blood clots, a process needed to stop bleeding. This results in people bleeding longer after an injury, easy bruising, and an increased risk of bleeding inside joints or the brain. Those with mild disease may only have symptoms after an accident or during surgery. Bleeding into a joint can result in permanent damage while bleeding in the brain can result in long term headaches, seizures, or a decreased level of consciousness.

There are two main types of hemophilia:-

1. Hemophilia A - occurs due to not enough clotting factor VIII.

2. Hemophilia B - occurs due to not enough clotting factor IX. They are typically inherited from one's parents through an X- chromosome with a nonfunctional gene. Rarely a new mutation may occur during early development or hemophilia may develop later in life due to antibodies forming against a clotting factor. Other types include hemophilia C, which occurs due to not enough factor XI, and parahaemophilia, which occurs due to not enough factor V. Acquired hemophilia is associated with cancers, autoimmune disorders, and pregnancy. Diagnosis is by testing the blood for its ability to clot and its levels of clotting factors. Prevention may occur by removing an egg, fertilizing it and testing the embryo before transferring it to the uterus. Treatment is by replacing the missing blood clotting factors. This may be done on a regular basis or during bleeding episodes. Replacement may take place at home or in the hospital. The clotting factors are made either from human blood or by recombinant methods. Up to 20% of people develop antibodies to the clotting factors which makes treatment more difficult. The medication desmopressin may be used in those with mild hemophilia A. Studies of gene therapy are in early human trials. Hemophilia A affects about 1 in 5,000–10,000, while hemophilia B affects about 1 in 40,000, males at birth. As hemophilia A and B are X- linked recessive disorders, females are very rarely severely affected. Some females with a nonfunctional gene on one of the X - chromosomes may be

mildly symptomatic. Hemophilia C occurs equally in both sexes and is mostly found in Ashkenazi Jews. In the 1800s hemophilia was common within the royal families of Europe.

2. COLOR BLINDNESS

Difficulty in distinguishing between colors, particularly red and green, is an inherited defect. Color blindness, also known as color vision deficiency, is the decreased ability to see color or differences in color. Color blindness can make some educational activities difficult. Buying fruit, picking clothing, and reading traffic lights can be more challenging, for example. Problems, however, are generally minor and most people adapt. People with total color blindness may also have decreased visual acuity and be uncomfortable in bright environments. The most common cause of color blindness is an inherited fault in the development of one or more of the three sets of color-sensing cones in the eye. Males are more likely to be color blind than females as the genes responsible for the most common forms of color blindness are on the X - chromosome. As females have two X chromosomes, a defect in one is typically compensated for by the other, while males only have one X chromosome. Color blindness can also result from physical or chemical damage to the eye, optic nerve or parts of the brain. There is no cure for color blindness. Diagnosis may allow a person's teacher to change their method of teaching to accommodate the decreased ability to recognize color. Special lenses may help people with red–green color blindness when under bright conditions. There are also mobile apps that can help people identify colors. Red–green color blindness is the most common form, followed by blue–yellow color blindness and total color blindness. Red–green color blindness affects up to 8% of males and 0.5% of females of Northern European descent. The ability to see color also decreases in old age. Being color blind may make people ineligible for certain jobs in certain countries. This may include pilot, train driver and armed forces.

Hypertrichosis

Hypertrichosis generally refers to diffuse or localized hair growth in androgen-independent areas of the body. Hypertrichosis develops when vellus hairs convert to terminal hairs. Diffuse hypertrichosis may be caused by medications, anorexia nervosa, or genetic, developmental, and metabolic disorders.

For example, a father may carry the gene for hairy ears, called hypertrichosis pinnae, on his Y chromosome; his daughter will not inherit this gene, but his son would. Some examples of traits caused by holandric genes are: Hypertrichosis pinnae - causes excessive hair in the ear.

Chapter 7 Introduction to Medical Zoology

Introduction to Medical Parasitology

- Definition of Parasitology
- Host
- Parasite
- Vector,
- Symbiosis
- Commensalism,
- Mutualism,
- Parasitism
- Zoonosis

Definition of Medical

Parasitology

Medical parasitology parasites parasitic diseases morphology life cycle pathogenesis diagnosis treatment transmission prevention

➢COMMENSALISM

➢It is a latin term which means eating on the same table..

➢The basis for a commensalism relationship between two organisms may be space, substrate, defense, shelter, transportation or food.

➢Example: An association between sucker fish Remora and large, powerful host Shark.

➢Remora is a carnivorous fish growing to about 50 cm in length. It can swim independently but is more often carried about by the large host Shark.

Example:

➢Another popular example of commensalism is the relationship between cattle Egrets and livestock like Cow, Buffalo, and Horse. ➢The cattle Egret is a common species of Heron that is mostly seen moving along with herds of cattle.

➢The movement of foraging livestock also dislodges various insects like flies, mites and bugs from the field, which cattle Egrets feed on.

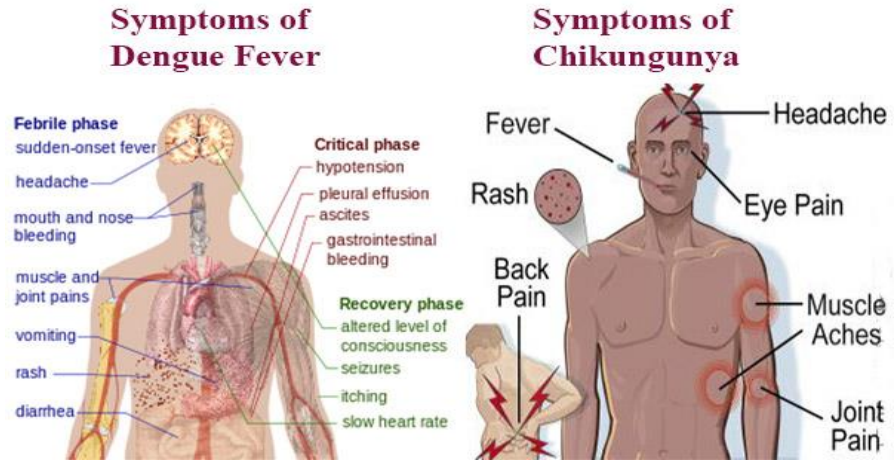
The Remora fish attaches to the shark and gets a free ride.

Birds build nests in trees.

Chapter No.9 Vector Borne Diseases **in Human:**

Occurrence, causative organism, symptoms and eradication programs of the following:

Dengue. Chicken Guinea Viral Influenza Scabies.



Dengue Fever

About Dengue

Dengue is a mosquito-borne viral disease caused by the Dengue virus. In this case, the dengue virus is transmitted by female mosquitoes – *Aedes aegypti*. These dengue mosquitos generally bite during the daytime and are found everywhere (Both inside and outside the house). These mosquitos are found to be at the peak of their activeness at dawn and dusk. The symptoms can develop only after 6 to 10 days after being bitten by an infected mosquito.

Dengue Fever

Dengue fever is transmitted by mosquitos which carry the dengue virus, which has four varied serotypes to infect human beings. The serotypes mentioned above denote a set of microorganisms that are exceptionally closely associated. These microorganisms can only be distinguished due to them having somewhat dissimilar antigens (the alien unit that affects the body and makes us produce antibodies) which prompt the body to create some dissimilar antibodies. Dengue cases are more common in subtropical and tropical regions of our planet, including our country.

Dengue Life Cycle

Following its departure from sylvatic cycles, the dengue virus has spread worldwide, and its primary lifecycle now only involves transmission between people and *Aedes* mosquitoes. The four life stages of the *Aedes aegypti* mosquito are egg, larva, pupa, and adult. About 8 to 10 days are needed to complete the life cycle from egg to adult. Mosquitoes can survive and breed both inside and outside the house.

Eggs

- Above the waterline, adult female mosquitoes lay their eggs on the interior, wet walls of water-filled containers.
- Usually, mosquitoes lay a hundred eggs at a time.
- Eggs are highly resilient; they can withstand drying up to eight months and adhere to container walls like glue.
- A female mosquito can be attracted to very little water. Any object storing water, including cups, bowls, fountains, barrels, tires, vases, and other containers, makes an excellent "nursery."

Larva

- Mosquito eggs hatch into larvae only once the water level rises enough to cover the eggs. Humans or rains will cause the larvae to emerge from containers containing eggs.
- Larvae consume aquatic bacteria as food. The larva develops into a pupa after going through three moults.

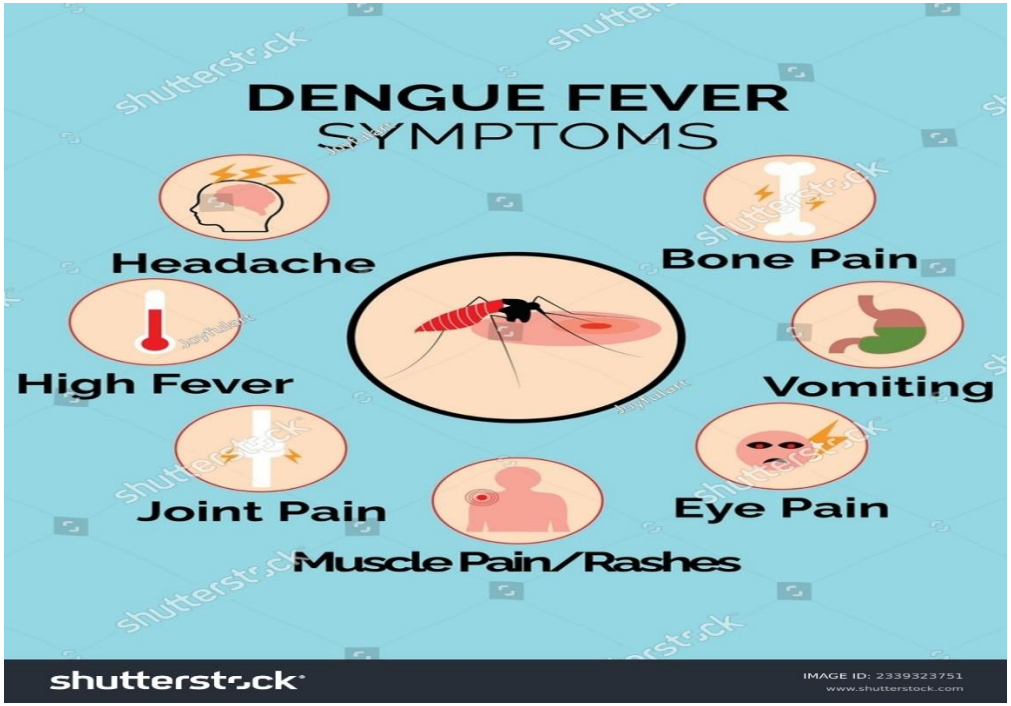
Pupa

Pupae continue to grow until the newly formed adult flying mosquito's body breaks through the pupal skin and exits the water.

Adult

- The adult female mosquito feeds on human and animal blood to generate eggs, while the male mosquito feeds on nectar from flowers.
- Female mosquitoes search for water sources to lay additional eggs after feeding.
- Throughout its lifespan, Aedes aegypti flies only a few blocks.

- Aedes aegypti mosquitoes prefer to attack humans above other mosquito species.
- Mosquitoes of the Aedes aegypti species favour areas with humans. They can be found inside residences, buildings, and workplaces when windows and doors lack screens or are left open.



Sign and Symptoms of Dengue Fever

Dengue has an unexpected attack, viz. a sudden start and these symptoms could be an indicator of its onset.

- Loss of appetite
- Diarrhoea and vomiting
- Gum and nose bleedings
- Severe joint and muscle pain
- Fatigue, nausea, and vomiting
- A sudden drop in blood pressure
- Multiple rashes and wounds on the skin
- Pain behind the eyes coupled with extreme headaches
- The patient might feel weak with a high fever for 3-7 days

Diagnosis of Dengue Fever

The presence of the Dengue virus in the blood cells can be diagnosed by isolation of the virus, testing serum samples, and other molecular methods. A patient with this syndrome is allowed to have a few blood tests to check the total count of red blood cells, and blood platelets, and other physical examinations conducted by the physician to evaluate whether the symptoms are caused by a dengue infection.

Treatment of Dengue Fever

Till today, there is no definite treatments or specific medicine to treat dengue infection. In general, the doctor may generally recommend regulating the pain and fever by using paracetamol instead of aspirin (as it might stimulate bleeding) and increasing fluid ingestion. Children below the age of 12 should not be given aspirin until and unless specially prescribed by the doctor.

In severe cases, blood transfusions, intravenous (IV) fluid supplementation and 24 hours hospitalization are required.

Prevention of Dengue Fever

The patient should take proper bed rest, especially during the days when the fever is at its peak and take leave from work, school preschool or childcare.

People suffering from dengue must stay away from places where they could get bitten by mosquitoes and should stay at home until they are no longer infectious (around 3-5 days).

For avoiding this illness, make sure your surroundings are free of any water logging issues, as the *Aedes* mosquito prefers to breed in stagnant clean water that could be found easily nearby our habitats.

Until now, no vaccine has been developed to prevent the Dengue virus. The only prevention is to avoid mosquito bites.

1. Cover your skin by wearing long pants, and long-sleeved shirts.
2. Use of mosquito repellents, traps, and nets.
3. Keep all the doors and windows closed especially at dawn, dusk, and early evening to avoid the entry of Dengue mosquitoes.
4. Keep your surroundings clean by removing all the waste and cleaning the standing water.

Chikungunya is an infection caused by the Chikungunya virus (CHIKV). It is mainly spread by the *Aedes albopictus and Aedes aegypti* mosquitoes

Cases of chikungunya are usually reported in Africa and Asia but there have been few instances of it happening in North America and Europe as well.

This article will give details about Chikungunya within the context of the IAS Exam.

Recorded outbreaks of the Chikungunya Disease

Each year, outbreaks of Chikungunya cause about 3 million infections every year. It's the developing nations in Africa and Asia that report the most cases. The transmission of the pathogen is high in urban environments where contact between humans and mosquitoes is at its peak.

There is no data available to ascertain when did Chikungunya find its way into Asia from Africa or how it did find its way, but the Asian strain outbreaks primarily happen in India and Southeast Asia.

To know more in detail about Viruses, candidates can checkout the linked article

Transmission and Diagnosis of Chikungunya

As stated before, Chikungunya is mainly spread by the *Aedes albopictus* and *Aedes aegypti* mosquitoes which bite during the day. Another form of transmission is vertical transmission as in, mother to child during birth or pregnancy. Although in theory it is possible for an infection to happen through tainted blood samples and organ donation, no cases have been reported as of late.

Chickungunya:-

A chikungunya diagnosis is done through blood testing for the virus's RNA to find antibodies of the virus. At times, symptoms can be mistaken for dengue and Zika virus fever as well. It has been reported that those previously infected with Chikungunya become immune to the virus

At present there is no proven method to test for chronic symptoms associated with Chikungunya fever although nonspecific laboratory findings such as C reactive protein and elevated cytokines can correlate with disease activity.

Symptoms of Chikungunya

The following are the common symptoms of Chikungunya are as follows:

- High fever
- Joint pain
- Rash
- Headache

- Fatigue
- Digestive problems,
- Conjunctivitis

In about 40-50% cases, rashes occur after two to five day after onset of symptoms. Abdominal pain, nausea vomiting or diarrhea may also occur. Normal activity is limited due to fatigue.

Sometimes, inflammation of the eyes may occur in the form of iridocyclitis, or uveitis, and retinal lesions may occur.

Treatment and Prevention of Chikungunya

There is no specific method of treatment for chikungunya at present. Supportive care and treatment of its symptoms are the best known methods to fight against chikungunya.

No vaccine against the disease has been made and the most effective methods of prevention is protection against contact with disease-carrying mosquitoes and controlling mosquito populations by destroying or limiting their habitats. This involves eliminating standing water bodies, preventing mosquitoes from laying eggs. If this isn't possible then insecticides and repellents need to be employed.

Other preventive measures include:

- Wearing bite-proof clothes such as long sleeves and trousers
- Treating garments with pyrethroids (a type of insect repellent)
- Securing windows or any other points of entry with meshes or anti mosquito nets.

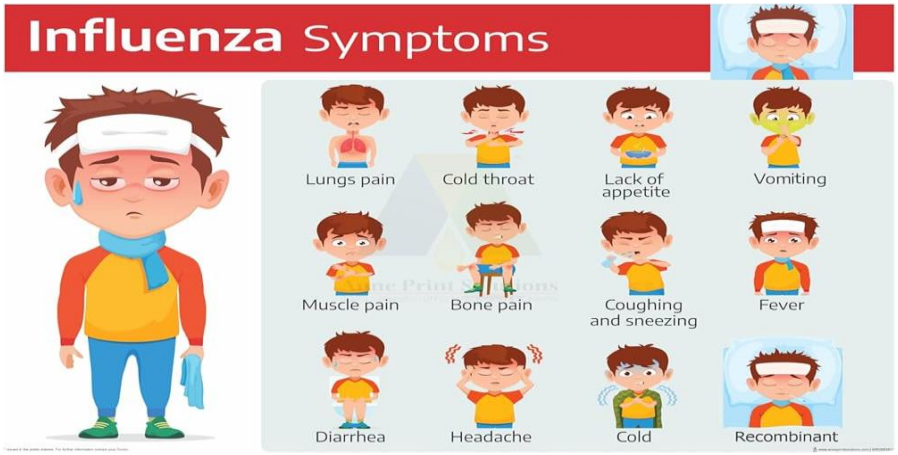
Overview of the Flu

The flu, also called Influenza, is a viral disease that affects the nose, throat and the lungs. It is quite contagious

and normally, this disease is not life-threatening. But different strains of the virus might be more potent and cause complications that can potentially lead to death. Of these strains, types A and B are the ones usually responsible for causing an outbreak. Furthermore, flu symptoms are harder to distinguish from other diseases as their symptoms are almost identical.

Causes of Flu

Flu is caused by viruses that spread from one person to the other. It is spread through the cough or sneeze droplets from the infected person. It can also spread if a healthy person touches the objects containing the virus.



Flu Symptoms

The flu can be contracted by everyone, but people over the age of 65 are more vulnerable. Even infants and children under 5 are at risk of contracting this disease. These are some of the most common symptoms of the

flu:

1. Cough
2. Fever accompanied by chills
3. Blocked or running nose
4. Sore throat
5. Body pain
6. Headaches
7. Fatigue and tiredness
8. Loss of appetite
9. Secondary bacterial infections
10. Nausea and Vomiting

Other problems associated with Flu

In some cases, flu develops other complications as well. These include:

- Ear infection
- Bronchitis
- Sinus
- Pneumonia
- Encephalitis

In conclusion, these are some of the characteristic influenza symptoms to watch out for. The best treatment for

flu is to take the flu shot or vaccine every year as a precautionary measure. But once it infects, the best remedy is to consume enough fluids and get adequate rest.

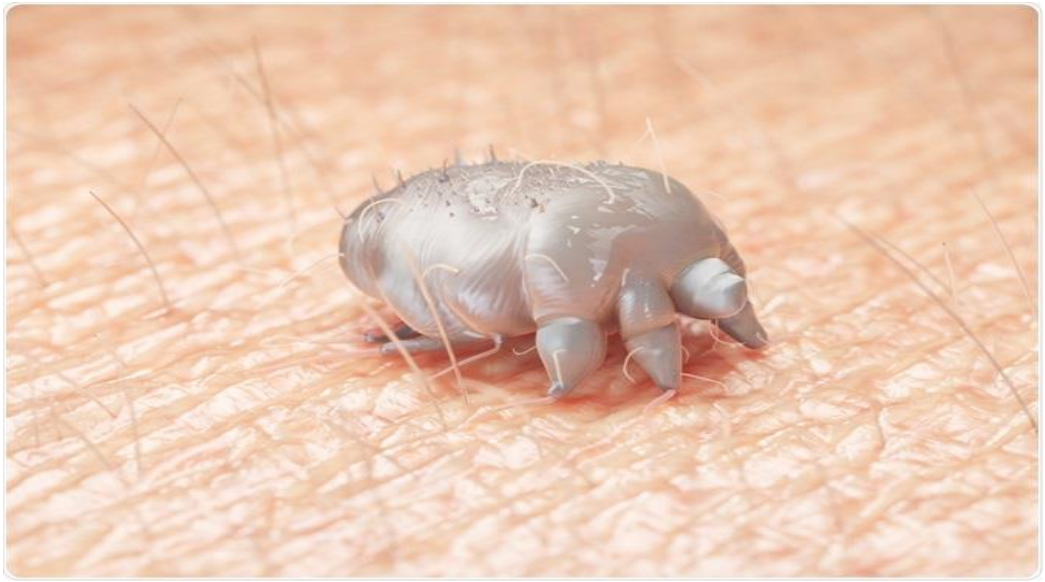
Scabies

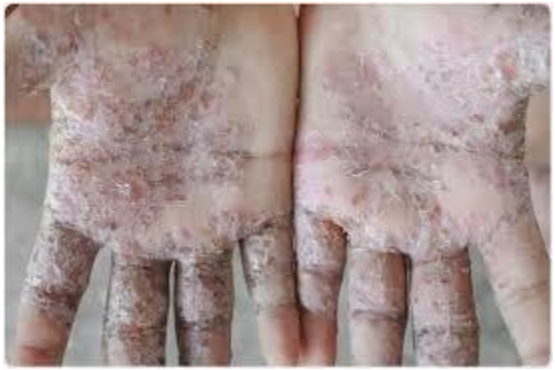
- Itching, mainly at night: Itching is the most common symptom. ...
- Rash: Many people get the scabies rash. ...
- Sores: Scratching the itchy rash can cause sores. ...
- Thick crusts on the skin: Crusts form when a person develops a severe type of scabies called crusted scabies.

Introduction

Scabies is a kind of skin irritation or inflammation caused by the parasite *Sarcoptes scabiei*. The *Sarcoptes scabiei* falls under the order Sarcoptiformes, the class Arachnida and the subclass Acari. This parasite enters the epithelial tissue during the night hours and causes severe skin irritation that makes the host rigorously scratch the infected area. Scabies is transmitted through physical contact, as the parasite passes from one individual to the other. This parasite is also known as the 'itch mite', that passes through close contact with an infected person. The parasite *Sarcoptes scabiei* measures up to 0.35 mm in length, which is not visible from the naked eye.

Let's look at the life of scabies to get a better understanding.





Life Cycle of Scabies

1) The Egg – Stage 1 –

After fertilization, the female parasite deposits 2 to 3 eggs daily under the skin. The size of the eggs are oval in shape and measure up to 0.15 mm in length. Under favourable conditions, the eggs hatch in three to four days and about 10% of the eggs transform into adult itch mites.

2) The Larvae – Stage 2 –

After the eggs hatch, the larvae mite migrates to the skin's surface and burrows itself into the stratum corneum; the outermost layer of the skin. This creates moulting pouches, which are invisible burrows. The larvae after hatching have only three pairs of legs and last for about three to four days. Later, the larva moults and transforms into a nymph.

3) The Nymph – Stage 3 –

In this stage, the nymph has four pairs of legs. The nymphs moult into much larger nymphs before it transforms into adult mites. The nymphs and larvae of itch mites are mostly found in the moulting pouches. The nymphs look similar to adults and are also found in hair follicles as well.

4) The Adult – Stage 4 –

When the nymph enters the adult stage, they appear to be round, sac-like eyeless itch mites. The female adult mites measure up to 0.45 mm in length. The male adult mites measure twice the size of the females. Mating process in mites occurs only once in their lifetime. The process of mating in mites occurs after the male mite has penetrated into the moulting pouch of the female mite. After mating, the female mite is fertilized for the rest of her remaining life.

These fertilized female mites detach their moulting pouches and roam on the surface of the skin until they find a suitable spot for burrowing permanently. The adult mites grab on to the skin through sucker-like pulvilli, which is attached to the two pairs of anterior legs.

After the female mite finds the desired spot, she burrows into the skin, laying the eggs in the process. After the fertilized female has burrowed into the skin, she can survive up to 1 to 2 months in that host. The male mites are hardly seen during their adult stage. They make shallow pits to feed, till the time they find a female's burrow and start mating.

Conclusion

Apart from the parasite *Sarcoptes scabiei*, there are other species that affect other mammals like cats, dogs, pigs, etc. As the scabies cases have been reported for the past 2,500 years, it is one of the earliest human diseases known to mankind. Since the transmission of this disease is close, physical contact, approximately 300 million cases are reported every year.

Chapter 11

Investigation and Treatments of Human Physiological Disorders

Angiography, Angioplasty and Dialysis

Angiography

- Angiography is used to check the health of your blood vessels and how blood flows through them. It can help to diagnose or investigate several problems affecting blood vessels, including: atherosclerosis – narrowing of the arteries, which could mean you're at risk of having a stroke or heart attack.

- **Angiography involves the use of x-ray imaging to examine blood vessels**. The images generated during an angiography procedure are known as angiograms.

Your doctor may recommend angiography if you experience symptoms of a blocked, damaged or malformed artery. An angiography test assists your doctor in determining the source of the problem and the amount of damage to your blood vessels. An angiography test can help your doctor diagnose and/or plan treatment for diseases like:

- Coronary artery disease (CAD) is a widespread cardiovascular condition characterized by the narrowing of coronary arteries due to the buildup of fatty deposits.

- Proper management of CAD is essential to prevent complications such as heart attack and improve overall heart health. In this comprehensive guide, we will delve into various CAD treatment options, including medications, lifestyle changes, and surgical interventions, providing valuable insights into how individuals with

CAD can effectively manage their condition.

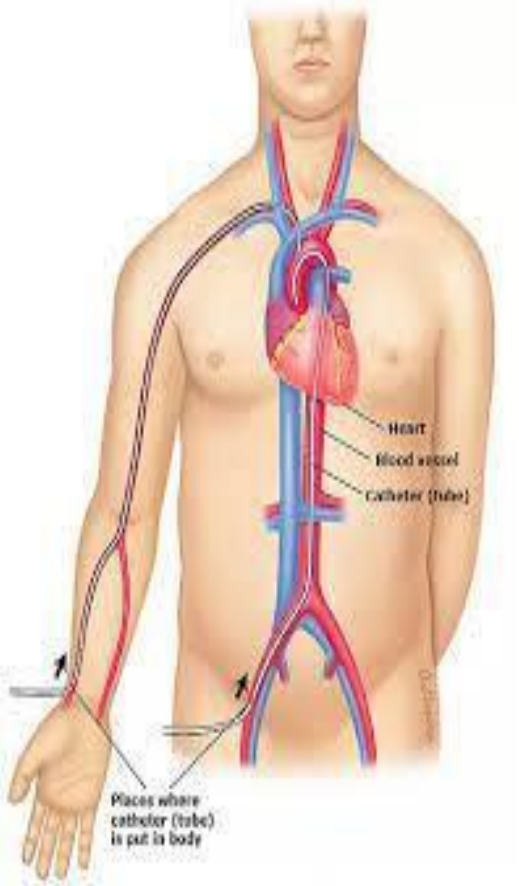
- An Aortic Aneurysm is defined as a tiny bulge, similar to a balloon, that developes on any part of the aorta when its walls become weak. If it is not treated in time, the damage can worsen leading to rupture and internal bleeding, which can pose a serious threat to the patients life.
- Aortic aneurysms can be broadly classified into two different types

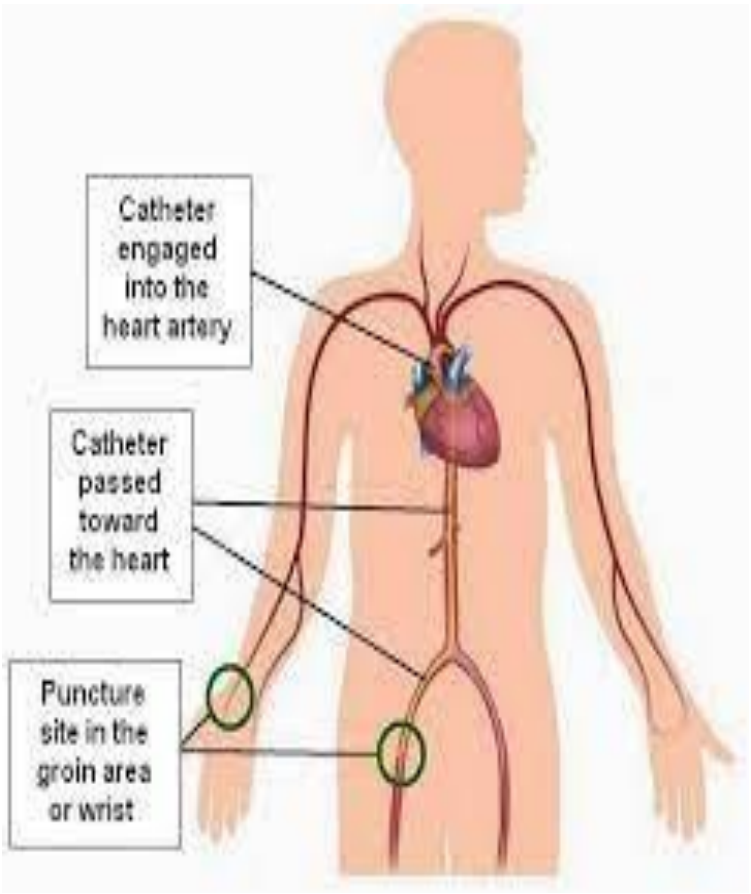
- abdominal aortic aneurysms and thoracic aortic aneurysms.

- Atherosclerosis –
- This occurs when the arteries become narrower and less flexible due to fatty plaque formation. Plaque is formed by the substances like fat, cholesterol, and scar tissue in the blood.
- This plaque builds up in the walls of arteries which makes them

sticky. This hinders the proper blood flow to other body parts.

- The chemical present in the smoke thickens the blood arteries making the movement of blood cells through the artery more difficult. It also causes inflammation.
- Procedure of angiogram : During the procedure a catheter (thin flexible tube) is inserted into an artery in your arm or groin and then threaded carefully into the heart.
- The blood vessels of the heart are then studied by injection of contrast media through the catheter.
- A rapid succession of X-rays (fluoroscopy) is taken to view blood flow.


Heart
Blood vessel
Catheter (tube)
Places where catheter (tube) is put in body


Catheter engaged into the heart artery
Catheter passed toward the heart
Puncture site in the groin area or wrist

- Safety : Angiography is generally a safe procedure, but minor side effects are common and there's a small risk of serious complications.
- You'll only have the procedure if the benefits outweigh any potential risk. Speak to your doctor about the risks with having angiography.
- Angiography uses: Angiography is used to check the health of your blood vessels and how blood flows through them.
- It can help to diagnose or investigate several problems affecting blood vessels, including: atherosclerosis – narrowing of the arteries, which could mean you're at risk of having a stroke or heart attack.

A coronary angiogram is a special procedure that takes dynamic x- ray pictures of your heart. The purpose of this procedure is to see if the coronary arteries are narrowed or blocked and to look for abnormalities of heart muscle or heart valves.

A coronary angiogram can help your cardiologist look for blockages in your coronary arteries. X-ray images from your procedure will help your provider make a diagnosis and decide if you need medicine, a stent or surgery. Plan on spending a few hours at the hospital for the procedure and recovery.

our provider may do coronary angiography when deciding if you need:

Medicine and a healthier lifestyle. Angioplasty or stenting.

Coronary artery bypass surgery (CABG).

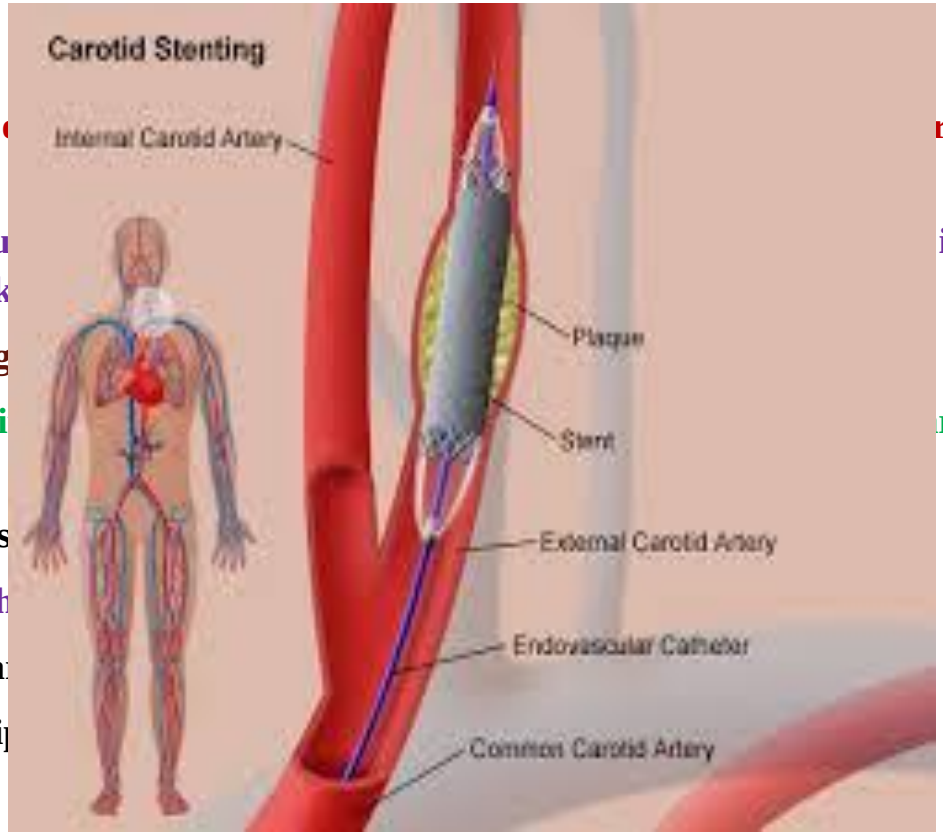
- **When would a coronary angiogram be needed?**
- **You may need a coronary angiogram when:**
- **Your stress test or electrocardiogram (EKG) wasn't normal.**
- **Your provider diagnoses you with a heart attack, a problem with a heart valve, or heart failure.**
- **You have heart surgery coming up and your provider thinks you may have coronary artery disease.**
- **You have chest pain (angina) that recently started or has changed in some way.**
- **You're having unusual chest discomfort or shortness of breath, but other tests don't show anything wrong.**
- **Who performs a coronary angiogram?**
- **A healthcare provider who's a heart expert — a cardiologist — will perform your coronary angiogram.**
- **How does a coronary angiogram work?**
- **Contrast dye that's injected into your coronary arteries through a small catheter allows your provider to see (through X-ray images) if there is blockage of your coronary arteries.**
- **The most common cause of narrowing of the coronary** arteries is cholesterol plaque (atherosclerosis).

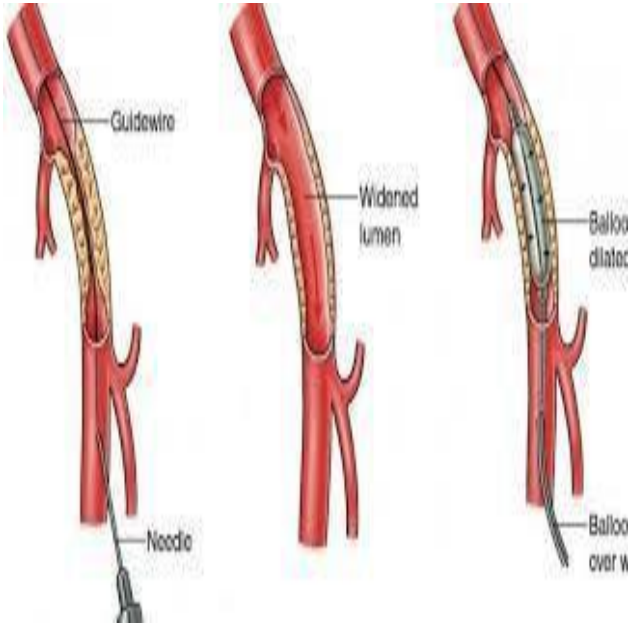
- **How do I prepare for a coronary angiogram?**
- **Your provider will most likely tell you not to eat or drink anything for eight hours before your coronary angiogram procedure.**
- **If your provider tells you to do so, you may need to avoid these medicines for at least one day before your procedure:**
- **Anticoagulants.**
- **Diabetes medications.**
- **Diuretics**

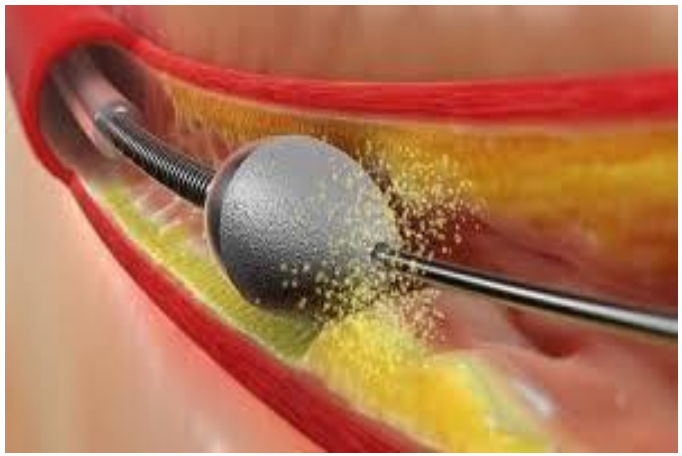
- **What are the risks of a coronary angiogram?**
- **An experienced healthcare provider can do coronary angiography safely.**
- **Serious complications are rare.**
- **People who are older or who have diabetes or kidney disease** are more likely to have complications.
- **The risks of a coronary angiogram include:**
- **Low blood pressure.**
- **Heart attack.**
- **An injured blood vessel.**

- **Stroke.**
- **Blood clots.**
- **Abnormal heartbeats.**
- **Kidney damage, including the need for dialysis.**
- **Pain, bleeding or infection where a needle or catheter broke your skin.**

- **Angioplasty**
- **Angioplasty is a procedure used to** **ry disease.**
- **It restores blood flow to the heart mu** **in an emergency setting, such as a heart attack**
- **Angioplasty can be done in an emerg**
- **Or it can be done as elective surgery i** **rt disease.**
- **Angioplasty is also called percutaneous**
- **For angioplasty, a long, thin tube (cath**
- **It is then guided to the blocked corona**
- **The catheter has a tiny balloon at its ti**




Guidewire
Widened
lumen
Needle



- **Once the catheter is in place, the balloon is inflated at the** narrowed area of the heart artery.
- **This presses the plaque or blood clot against the sides of the artery.**
- **The result is more room for blood flow.**
- **The healthcare provider uses fluoroscopy during the surgery.**
- **It helps the healthcare provider find the blockages in the heart arteries as a contrast dye moves through the arteries.**
- **This is called coronary angiography.**
- **In atherectomy, the healthcare provider may use a catheter with a rotating tip.**
- **The plaque is broken up or cut away to open the artery once the catheter reaches the narrowed spot in the artery.**

- **Coronary stents**
- **A stent is a tiny, expandable metal mesh coil.**
- **It's put into the newly opened area of the artery to help keep the artery from narrowing or closing again.**
- **Tissue will start to coat the stent like a layer of skin once the** stent has been placed.
- **The stent will be fully lined with tissue within 3 to 12 months.**
- **length of time depends on if the stent has a medicine coating or not.**
- **You may be prescribed medicines called antiplatelets to decrease the "stickiness" of platelets.**
- **Platelets are special blood cells that clump together to stop bleeding.**
- **The medicine can also prevent blood clots from forming inside the stent.**
- **Your healthcare team will give specific instructions on which medicines need to be taken and for how long.**

- **Some stents don't have medicine coating and are called bare** metal stents.
- **They may have higher rates of stenosis.**
- **But they don't need long-term use of antiplatelet medicines.**

- **This may be the preferred stent in people who are at high risk**

of bleeding.

- **Stents can become blocked.**
- **It's important to talk with your healthcare team about what you need to do if you have chest pain after a stent placement.**
- **You may need a repeat procedure if scar tissue does form inside the stent.**
- **This may be using either balloon angioplasty or with a second stent. In rare cases, radiation therapy may be given through a catheter placed near the scar tissue to stop the growth of scar tissue.**
- **It also opens up the vessel.**

- **Why might patient need angioplasty?**
- **Angioplasty is done to restore coronary artery blood flow.**
- **This is done when the narrowed artery is in a place that can be reached in this manner.**
- **Not all coronary artery disease (CAD) can be treated with** angioplasty.
- **Your healthcare provider will decide the best way to treat your CAD based on your circumstances.**
- **Risks of angioplasty: Possible risks linked to angioplasty, stenting, atherectomy, and related procedures include:**
- **Bleeding at the site where the catheter is put into the body (usually the groin, wrist, or arm)**
- **Blood clot or damage to the blood vessel from the catheter**
- **Blood clot within the treated blood vessel**
- **Infection at the catheter insertion site**
- **Abnormal heart rhythms**
- **Heart attack**
- **Stroke**
- **Chest pain or discomfort**
- **Rupture of the coronary artery or complete closing of the coronary artery, needing open-heart surgery**

- **Allergic reaction to the contrast dye used**
- **Kidney damage from the contrast dye**
- **Preparation for Angioplasty Your healthcare team will explain the procedure to you and you can ask questions.**
- **You will be asked to sign a consent form that gives your permission to do the procedure.**
- **Tell your healthcare team if you have ever had a reaction to any contrast dye, or if you are allergic to iodine.**
- **Tell your healthcare team if you are sensitive to or are allergic to any medicines, latex, tape, and local or general anesthesia.**

- **Follow any directions you are given for not eating or drinking**

before surgery.

- **Tell your healthcare team if you are pregnant or think you could be.**
- **Radiation exposure during pregnancy may lead to birth**

defects.

- **Tell your healthcare team if you have any body piercings on your chest or belly (abdomen).**
- **Tell your healthcare team about all prescription and over-the- counter medicines, vitamins, herbs, and supplements that you are taking.**
- **Tell your healthcare team if you have a history of bleeding disorders or if you are taking any blood-thinning medicines (anticoagulant or antiplatelet),**

- **Your provider may request a blood test before the procedure**

to find out how long it takes your blood to clot.

- **Other blood tests may be done as well.**
- **Tell your healthcare team if you have a pacemaker or other implanted device.**
- **You may get a sedative before the procedure to help you relax.**
- **Based on your health condition, your healthcare provider may give you other instructions on how to get ready.**

- **Procedure of angioplasty :**
- **You will be asked to remove any jewelry or other objects that may interfere with the procedure.**
- **You may wear your dentures or hearing aid if you use either of these.**
- **You will be asked to remove your clothing and will be given a gown to wear.**

- **You will be asked to empty your bladder before the procedure.**
- **If there is a lot of hair at the area of the catheter insertion (often the groin area), the hair may be shaved off.**
- **An IV (intravenous) line will be started in your hand or arm before the procedure.**
- **It will be used for injection of medicine and to give IV fluids, if needed.**
- **You will be placed on your back on the procedure table.**

- **You will be connected to an electrocardiogram (ECG) monitor that records the electrical activity of your heart and keeps track of your heart rate using electrodes that stick to your skin.**
- **Your vital signs (heart rate, blood pressure, breathing rate, and oxygen level) will be tracked during the procedure.**
- **There will be several monitor screens in the room, showing your vital signs, the images of the catheter being moved through your body into your heart, and the structures of your heart as the dye is injected.**
- **You will get a sedative in your IV line to help you relax. But you will likely stay awake during the procedure.**
- **Your pulses below the catheter insertion site will be checked and marked so that the circulation to the limb below the site can easily be checked during and after the procedure.**

- **Local anesthesia will be injected into the skin at the insertion**

site.

- **This may be in your leg, arm, or neck.**
- **You may feel some stinging at the site for a few seconds after the local anesthetic is injected.**

- **Once the local anesthesia has taken effect, a sheath, or introducer, will be put into the blood vessel (often at the groin). This is a plastic tube through which the catheter will be threaded into the blood vessel and advanced into the heart.**
- **The catheter will be threaded through the sheath into the blood**

vessel.

- **The healthcare provider will advance the catheter through the aorta into the heart.**
- **Fluoroscopy ("live" X-ray) will be used to help see the catheter**

advance into the heart.

The catheter will be threaded into the coronary arteries.

Once the catheter is in place, contrast dye will be injected through the catheter into your coronary arteries to see the narrowed area.

These effects include a flushing sensation, a salty or metallic taste in the mouth, or a brief headache.

These effects usually last only a few moments.

Tell your healthcare provider if you feel any breathing trouble, sweating, numbness, itching, nausea or vomiting, chills, or heart palpitations.

After the contrast dye is injected, a series of rapid X-ray images of the heart and coronary arteries will be taken.

- **Any chest discomfort or pain should go away when the balloon**

is deflated.

- **But tell your healthcare provider right away if you notice any continued discomfort or pain**
- **The healthcare provider may inflate and deflate the balloon**

several times.

- **In some cases, the stent may be put into the artery before the balloon is inflated.**
- **Then the inflation of the balloon will open the artery and fully**

expand the stent.

- **When the healthcare provider locates the narrowed artery, the catheter will be advanced to that location and the balloon will be inflated to open the artery.**
- **You may have some chest pain or discomfort at this point because the blood flow is temporarily blocked by the inflated balloon.**

- **The healthcare provider will take measurements, pictures, or**

angiograms after the artery has been opened.

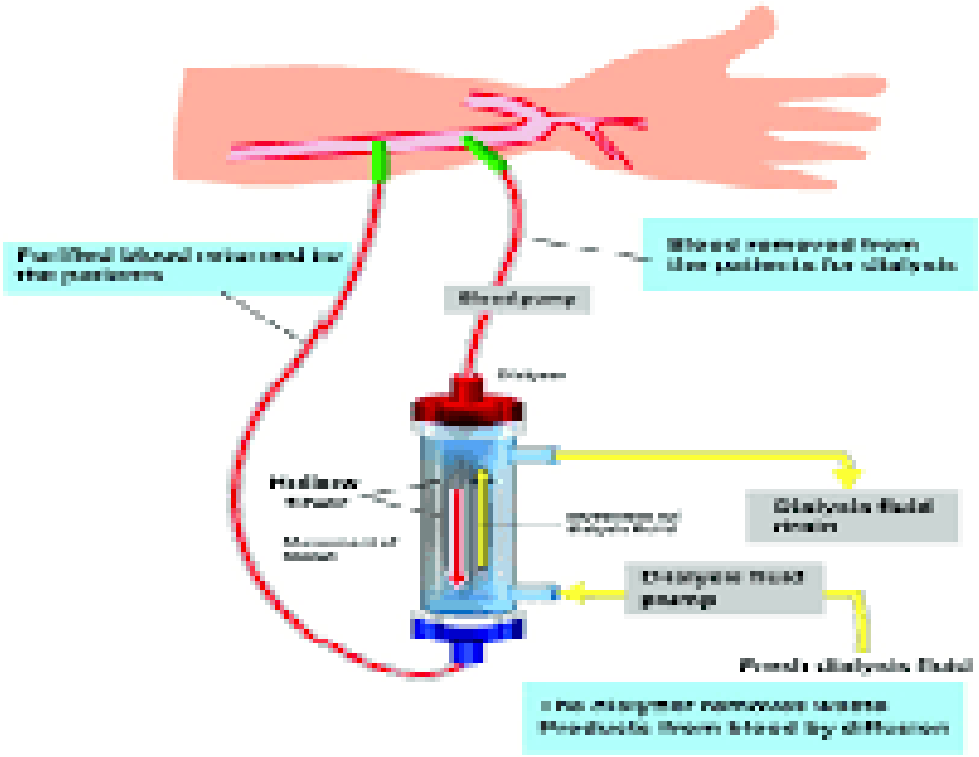
- **Once it has been determined that the artery is opened sufficiently, the catheter will be removed.**

➢ may be closed with a
cl by the use of stitches
(s the blood vessel from
bl

➢ for
y

➢ pass in a week's time.
O taking the prescribed
m

➢ ɔut this can c
n



➢ **Dialysis**

➢ **Dialysis is a treatment for people whose kidneys are failing. When you have kidney failure, your kidneys don't filter blood the way they should.**

➢ **Dialysis does the work of your kidneys, removing waste products and excess fluid from the blood.**

➢ **Acute kidney injury (AKI): a sudden episode of kidney failure or kidney damage that happens within a few hours or days. AKI is usually treated in a hospital setting with intravenous fluids (given through the vein).**

➢ **Kidney failure: when 10-15% of your kidney function remains, measured by an estimated glomerular filtration rate (eGFR) of less than 15 mL/min.**

➢ **At this stage, your kidneys are no longer able to keep you alive**

without some extra help.

With kidney failure, dialysis is only able to do some of the work of healthy kidneys, but it is not a cure for kidney disease.

- **How it works**
- **Dialysis performs some of the duties that your kidney usually does to keep your body in balance, such as:**
- **removing waste and extra fluids in your body to prevent them from building up in the body keeping safe levels of minerals in your blood, such as potassium, sodium, calcium, and bicarbonate**
- **helping to regulate your blood pressure. Types**
- **1 Hemodialysis (HD) In hemodialysis, a dialyzer (filtering machine) is used to remove waste and extra fluid from your blood, and then return the filtered blood into your body.**

- **Before starting hemodialysis, a minor surgery is needed to create a vascular access site (opening into one of your blood vessels), usually in your arm.**
- **This access site is important to have an easy way to get blood**

from your body, through the dialyzer, and back into your body.

- **Hemodialysis can be done at a dialysis center or at home. Treatments usually last about four hours and are done three times per week.**
- **2 Peritoneal Dialysis (PD)**
- **In peritoneal dialysis, your blood is filtered inside your own body instead of using a**

dialyzer machine.

- **For this type of dialysis, the lining of your abdomen or belly area (also called the peritoneum) is used as a filter.**
- **Before starting peritoneal dialysis, a minor surgery is needed to place a catheter (soft tube) in your belly.**
- **During each treatment, your belly area is slowly filled with dialysate (a cleansing fluid made from a mixture of water, salt, and other additives) through the catheter.**
- **As your blood flows naturally through the area, extra fluid and waste products are pulled out of the blood vessels and into the belly area by the dialysate (almost like a magnet).**
- **After a few hours, the fluid mixture is drained from your belly using the same catheter and bag that was used at the beginning of the treatment.**

Peritoneal dialysis can be done almost anywhere if you have the supplies required to perform the treatment.

- **Effectiveness**
- **Dialysis is a very effective treatment option for clearing waste products and extra fluid from your blood.**
- **All types of dialysis are equally effective, but your medical condition and personal preferences may match one treatment approach better than others.**
- **You and your doctor will discuss this and decide which type of dialysis and which place is best.**

- **Side Effects**
- **Every person responds differently to dialysis, and your level of risk for each side effect will differ from others.**
- **If you have concerns about any of these risks, talk to your doctor and dialysis team about ways you can lower your risk.**
- **Although these side effects may sound scary, they should be compared to the risks that come from continuing to live with untreated kidney failure.**
- **Most people on dialysis are able to keep a regular routine**

except for the time needed for treatments.

- **Dialysis often makes people feel better because it helps clear the waste products t have built up in the blood between treatments.**