

BIOLOGY

I Human Genetic Diseases

Inheritance of Sex

- Males and females do not have identical chromosome
- Males contain XY chromosomes
- Females contain XX chromosomes
- Y chromosome has testis determining factor (Tdf)

Sex-Linked Genes

- Y chromosome is smaller than X
- In fruit flies, X chromosome contains from 100 to 200 genes
- Y chromosome contains fewer genes
- Some non-sexual traits are carried on the X chromosome

Linkage

- Two or more genes located on nonhomologous chromosomes tend to assort independently of each other and end up in different gametes
- Two or more genes on the same chromosome tend to be transmitted together into a gamete

Crossing Over

- Crossing over is the breakage and exchange of corresponding segments between homologous chromosomes
- The farther apart two genes are on a chromosome, the greater will be the frequency of crossing over and recombination between them.

Euploidy : Variations from the normal diploid number ($2n$) may either be haploids or polyploids ($3n$, $4n$, $5n$, $6n$ etc.)

Type	Homologues present
Monoploid (haploid)	one (n)
Diploid	two ($2n$)
Polyploid	
(a) triploid	three ($3n$)
(b) tetraploid	four ($4n$)
(c) pentaploid	five
(hexaploid, septaploid, octoploid etc.)	

Polyploidy is classified as

- **Autoploids** : In autoploids all the chromosome sets are homologous (ABC, ABC, ABC, ABC).
- **Allopolyploids** : Allopolyploids arise from a stock that is a heterozygote (ABC, ABC, A ϕ B ϕ C ϕ , A ϕ B ϕ C ϕ).

Aneuploidy : Variations in chromosome number involving individual chromosomes are included in aneuploidy.

Type	Chromosome Number
Disomic (normal)	$2n$
Monosomic	$2n - 1$
Double monosomic	$2n - 1 - 1$
Nullisomic	$2n - 2$
Polysomic	more than $2n$
(a) Trisomic	$2n + 1$
(b) Double trisomic	$2n + 1 + 1$
(c) Tetrasomic	$2n + 2$
(d) Pentasomic	$2n + 3$
(Hexasomic, Septasomic etc.)	

PKU (Phenylketonuria)

- Recessive autosomal
- Phenylketonuria - Increased phenylalanine in blood and urine.
- Inability to properly metabolize phenylalanine
- Affects chromosome 12
- Damage to nervous system and mental retardation
- Can be overcome by diet

Alkaptonuria

- Black urine disease
- Caused due to deficiency of alkapton oxidase
- Inability in metabolism of homogentisic acid
- Cause arthritis and other damages

Albinism

- Autosomal recessive
- Example of epistasis

- Missing enzyme results in unpigmented skin, hair and eyes
- Affects 1 in 20,000
- Those affected must alter life-style

Tay Sach's disease

- Autosomal recessive
- Caused due to deficiency of b-D-N acetyl hexosaminidase
- Accumulation of ceramide in brain cells
- Damage to nervous system - brain and spinal cord.

Sickle-Cell Disease

- Abnormal haemoglobin
- Anaemia and blocked circulation
- One amino acid change in b chain of haemoglobin
- Resistance to malaria
- Chromosome 11 affected

Thalassemia

- Autosomal recessive
- Chromosome 16 in a thalassamia and 11 in b thalassamia affected
- Abnormal haemoglobin and anaemia
- Bone and spleen enlargement
- Some protection from malaria
- Lack glucose-6-phosphate dehydrogenase

Galactosemia

- Autosomal recessive
- Cannot breakdown galactose
- Occurs in about 1 in every 100,000 newborns
- Cannot convert galactose-1-phosphate to glucose-1-phosphate
- Damages brain, liver and eyes
- Both males and females can carry the recessive allele on an autosome - not sex linked
- Heterozygotes of either sex are symptom free
- Homozygous of both sexes are affected
- When both parents are heterozygous, there is a fifty-percent chance that each child will be heterozygous also.
- There is twenty five percent chance that each child will be a homozygous recessive. When both parents are homozygous for the recessive allele, all of their children will be affected.

Huntington's Disorder

- Autosomal dominant

- Causes progressive degeneration of the nervous system - abnormal speech and respiration
- Generally onset is observed after 40 years of age
- Children of affected parent have a 50% chance of developing the disease
- Caused by dominant gene mutation on short arm of 4th chromosome.

Achondroplasia

- Autosomal dominant gene
- Affects 1 in 10,000
- Type of dwarfism due to cartilage formation.

Polydactyly and Brachydactyly

- More than five fingers and toes
- Small sized fingers.

Marfan syndrome

- Dominant mutation of structural gene (fibrillin) on chromosome 15
- Very long extremities
- Spider like finger
- Dislocation of eye and cardiovascular disease.

Haemophilia A

- In females, two defective genes are necessary for the disease to develop. i.e. that condition is lethal.
- In males, there is only one copy of the X-chromosome, so if they inherit a defective gene from their mother, they will develop the disease.
- Heterozygous females are phenotypically normal. Males are affected
- A normal male and heterozygous female have a 50% chance that females will be carriers and 50% chance that males will be affected
- A homozygous female and normal male will have all daughters carriers and all males affected
- Only 1 in 7,000 males is affected
- Also called bleeder disease.

Colour blindness

- Recessive sex linked abnormality
- Caused by recessive X-linked gene
- Mainly of three types
Protan, deutan and tritan (opia) *i.e.*, defective perception of red, green and blue respectively.
- Occur in females only in homozygous condition

- Inheritance pattern is same as in haemophilia

Duchenne Muscular Dystrophy

- Recessive sex linked
- Failure to produce the protein, dystrophin
- Progressive muscle degeneration
- Attempting to treat with myoblasts

Cystic Fibrosis

- Defective autosomal
- Defect on chromosome 7 which code for chloride transport factor (CFTR)
- Defective membrane protein
- Excessive mucous production
- Respiratory and digestive failure

Down's Syndrome

- Results when a gamete receives two copies of chromosome 21 due to nondisjunction
- Individual possesses 3 copies of chromosome 21. This is called trisomy 21. Condition is $(2X + 1)$
- Most affected individuals show moderate to severe mental retardation
- Not an inherited condition
- Increased risk in mothers past 35 and fathers past 55

Turner's Syndrome

- Monosomic
- Only affects females
- Affected individuals have only a single X chromosome ($44 + XO$)
- Affects about 1 in 5,000
- Affected individuals are sterile and have shortened life span

Klinefelter's Syndrome

- Results from nondisjunction
- Males who are XXY
- Condition is $(44 + XXY)$
- Affected individuals are sterile and suffer slight to moderate mental impairment
- One barr body present

Edward Syndrome

- Trisomy of 18th chromosome
- Mental retardation, micrognathia, short sternum, malformed ears etc.

Patau's Syndrome

- Trisomy of 13th chromosome

- Hare lip, cleft palate, polydactyly etc. are the symptoms.

- Death usually occurs soon after birth or may survive upto 3 months.

Super females

- Due to presence of extranumerary chromosomes
- Mentally retarded and congenital abnormalities like underdeveloped external genitalia, uterus and vagina
- Condition is $45 + XXX$
- Presence of two barr bodies

XYY Condition or super males

- Affects 1 in 1,000 males
- Results from nondisjunction
- Affected individuals tend to be taller than normal and may show mild retardation
- At one time, affected individuals were thought to be predisposed to crime

Cri-du Chat Syndrome

- Rare deletion on chromosome 5
- Malformed larynx
- Affected children's crying sounds like meowing cat
- Causes mental retardation.

Fragile X Chromosome

- Found in 15% of males with pure mental retardation
- Portion near the end of the X chromosome breaks off
- Second only to Down's syndrome as a cause of mental retardation

Hypertrichosis

- Excessive hairs on ear pinna
- Y-linked disease. Genes called holandric gene
- Affects only males

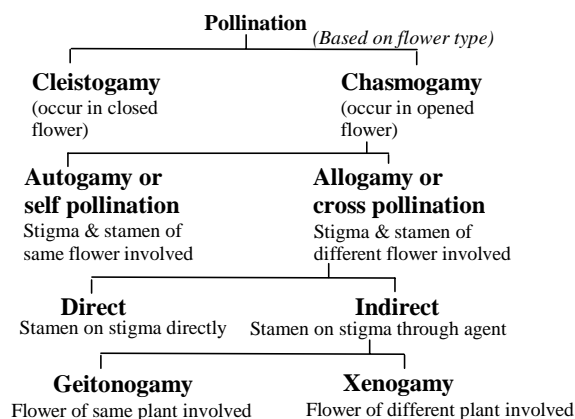
Erythroblastosis foetalis

- An Rh factor incompatibility occur when father is Rh^+ and mother is Rh^-
- Caused due to disadjustment of Rh factor
- Occur in human embryo
- In developing foetus, erythroblastosis foetalis is caused by haemolysis
- Human do not produce Rh antibody naturally.

II Pollination

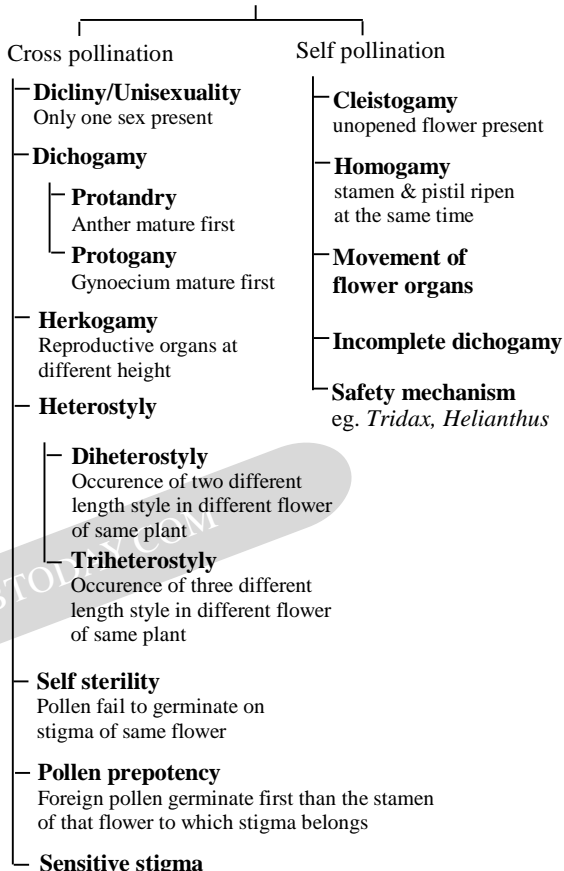
POLLINATION

- The process of transfer of pollen grains from the anther to the stigma is called pollination.
- Types of pollination**



- Genetically, Geitonogamy is considered as self pollination. But sometime considered as cross pollination as an agency is involved in pollination.
- Mechanism of insect pollination**
 - (i) Piston mechanism – *Papilionaceae*
 - (ii) Liver – *Salvia*
 - (iii) Trap – *Aristolochia*
 - (iv) Pit fall – *Arum*
 - (v) Translator – *Calotropis*

Contrivances (artificial or natural mechanism which help) for



Types of pollination	Agency involved	Characteristic of flower	Example
1. Anemophily	Air	Spike, catkin or spadix inflorescence flower small & unattractive, flower produces neither nectar nor scent, large quantity of pollen. Stigma branched & feathery & sticky.	eg. <i>Pilea</i> <i>Urtica</i>
2. Hydrophily	Water	Pollen grains needle like.	<i>Zostera</i> , <i>Vallisneria</i> <i>Ceratophyllum</i>
3. Zoophily (i) Entomophily	Animals Insect	Big flower, bright coloured, strong smelling, possess nectary gland and produce lots of nectar.	<i>Calotropis</i> <i>Jasmine</i> <i>Ocimum</i> <i>Ixora</i>

(ii) Ornithophily	Birds	Big flower producing plenty of nectar.	<i>Erythrina</i> <i>Delonix</i>
(iii) Chiropteriphily	Bats	Tropical flower with juicy stamen.	<i>Musa</i> <i>Anthocephalus</i> <i>Kagelia</i>
(iv) Melacophily	Slugs & snails	Spadix inflorescence.	<i>Lemna</i>
(v) Ophiophily	Snake	–	<i>Arisaemia</i>

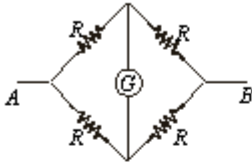
DIFFERENCE BETWEEN SELF POLLINATION AND CROSS POLLINATION

S.N.	Self Pollination	Cross Pollination
(i)	It is the transfer of pollen grains from anthers to the stigma of either the same or genetically similar flower.	Cross pollination involves the transfer of pollen from anther of one flower to the stigma of a genetically different flower.
(ii)	Both the anthers and stigmas mature simultaneously.	The anthers and stigmas mature at different times.
(iii)	Self pollination can occur even in closed flower.	It occurs only when the flowers are open.
(iv)	External agency is not required for self pollination.	An external agent is essential for carrying the pollen grains from anthers to the stigma.
(v)	Self pollination is economical for the plant.	Cross pollination is not economical as the plant has to produce a lots of pollen grains, nectar, scent, bright colour, etc.
(vi)	The plants ultimately become homozygous.	The plants remain heterozygous.
(vii)	Self pollination produces pure lines.	It gives rise to offspring having variations among themselves.
(viii)	Self pollination cannot eliminate useless or harmful characters.	Cross pollination dilutes or eliminates the useless and harmful characters.
(ix)	Highly useful characters are preserved by self pollination.	Cross pollination is unable to preserve all the highly useful characters since they tend to get diluted.
(x)	Adaptability to changed environment is absent as self pollination does not produce variability.	Plants are better adapted to changed environment and struggle for existence due to introduction of variations.
(xi)	Immunity of the race towards disease and yield of plant falls with time.	Immunity of the race towards diseases and yield of plant is usually maintained.
(xii)	It does not help in producing new races, varieties and species.	Cross pollination is a mechanism of producing new races, varieties and even species.

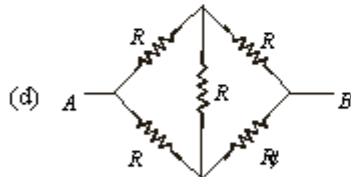
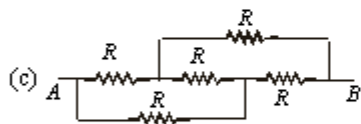
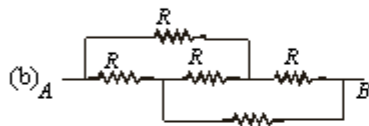
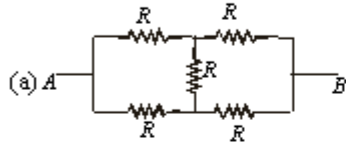
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Some standard forms of grouping of resistances which are generally asked

1.



It is a case of balanced Wheatstone bridge. It can be asked in different forms as given below:



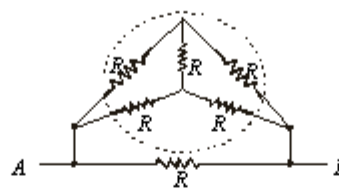
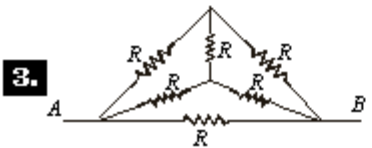
In above all cases, equivalent resistance across A and $B = R$.

2.



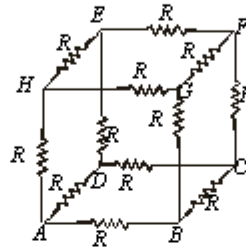
Equivalent resistance across A and $B = R/3$.

3.



The dotted part of circle is a balanced Wheatstone bridge. Hence equivalent resistance across

4.

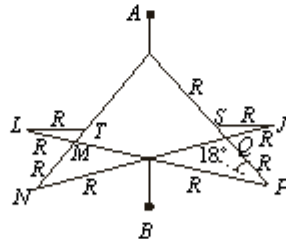


Case 1: Equivalent resistance across A and $F = 5R/6$.

Case 2: Equivalent resistance across A and $C = 3R/4$.

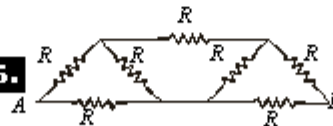
Case 3: Equivalent resistance across A and $B = 7R/12$.

5.



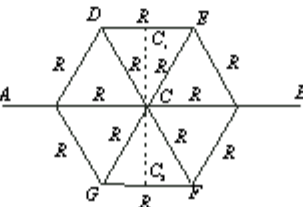
Equivalent resistance across A and $B = 0.97 R$.

6.



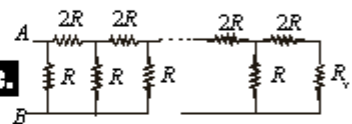
Equivalent resistance across A and $B = 8R/7$.

7.



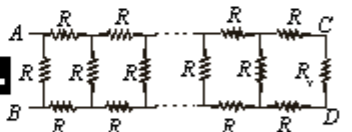
Resistance between A & $C =$
Resistance between C & $B = 0.4 R$
∴ resistance between A and $B = 0.8 R$

8.



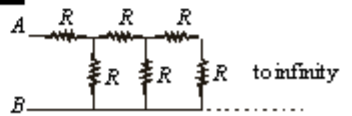
The value of R_x so that the total resistance between points A and B be independent of the number of sets is equal to $R(\sqrt{3} - 1)$.

9.



The value of R_x so that the resistance of the entire circuit between A and B does not change with the number of elementary sets used is equal to $(\sqrt{3} - 1) R$.

10.



Equivalent resistance across A and $B = \frac{(\sqrt{3} + 1) R}{2}$.

Different types of parallel plate capacitors

SNo.	Combination	Capacitance
1.		$C = \frac{\epsilon_0 A}{d}$
2.		$C = \frac{\epsilon_0 K A}{d}$
3.		$C = \frac{\epsilon_0 A}{\left(d - t + \frac{t}{K}\right)}$
4.		$C = \frac{\epsilon_0 A}{\left(\frac{t_1}{K_1} + \frac{t_2}{K_2} + \frac{t_3}{K_3}\right)}$

Multiple capacitors

	Eq. Capacitance	Combination
1.	$C_0 = \frac{2\epsilon_0 A}{d}$	
2.	$C_0 = \frac{3\epsilon_0 A}{d}$	
3.	$C_0 = \frac{3\epsilon_0 A}{2d}$	
4.	$C_0 = \frac{2\epsilon_0 A}{d}$	

Different combinations of capacitors

	Combination	Eq. Capacitance
1.		$K = \frac{K_1 + K_2}{2}$ $C_0 = \frac{\epsilon_0 A (K_1 + K_2)}{2t}$
2.		$K = \frac{2K_1 K_2}{K_1 + K_2}$ $C_0 = \frac{2\epsilon_0 A}{t} \left(\frac{K_1 K_2}{K_1 + K_2} \right)$
3.		It is a case of balanced Wheatstone's bridge. So Eq. capacitance = C.
4.		$C_0 = C.$
5.		$C_0 = \frac{2}{3} \frac{\epsilon_0 A}{d}$
6.		$C_0 = \left(\frac{1 + \sqrt{5}}{2} \right) C$
7.		$C_0 = \left(\frac{\sqrt{5} - 1}{2} \right) C$
8.		$C_0 = 2C.$