ADHIKAANSH ACADEMY

(IITJEE NEET IX X XI XII)

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(CLASS 12TH)

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PRINCIPLES OF INHERITANCE AND VARIATION

Genetics

Genetics is a branch of biology dealing with inheritance and variation of characters from parents of offspring.

Inheritance

Process by which characters are passed on from parent to progeny

Variation

Degree by which the progeny differs from its parents

Mendel's Experiments

- Gregor Johann Mendel known as the father of genetics proposed the laws of inheritance.
- He used garden pea as his sample.
- Large sampling size gave credibility to his collected data.
- Garden pea plant possessed certain completely opposite traits.

Example – tall and dwarf plants

- He worked on the following **seven** traits of garden pea:

S. No.	Character	Dominant	Recessive
1	Stem height	Tall	Dwarf
2	Flower colour	Violet	White
3	Flower position	Axial	Terminal
4	Pod shape	Inflated	Constricted
5	Pod colour	Green	Yellow

6	Seed shape	Round	Wrinkled
7	Seed colour	Yellow	Green

- True breeding pea lines were obtained by continuous self pollination for several generations.
- Fourteen true breeding pea lines were selected as pairs, which were similar except for one character with contrasting traits.
- Artificial cross pollination (hybridisation) was performed on such varieties to obtain first hybrid generation known as the first filial progeny or F_1.

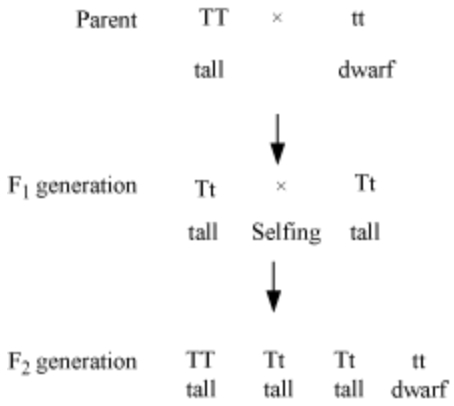
Inheritance of One Gene

- After hybridisation, the F_1 generation so obtained resembled only one of its parents (say, all tall; no dwarf).
- When 2 plants from F_1 generation were self pollinated, the second filial progeny or F_2 generation was obtained.
- Revival of unexpressed trait (dwarf) was observed in some F_2 progeny. Both traits, tall and dwarf, were expressed in F_2 in ratio 3:1.
- Mendel proposed that something is being passed unchanged from generation to generation. He called these things as 'factors' (presently called genes).
- Factors contain and carry hereditary information.
- Alleles – Slightly different form of same factor

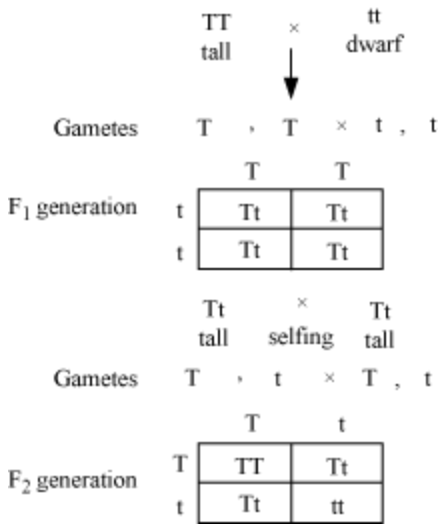
Two alleles code for a pair of two contrasting traits. (e.g., tall and dwarf)

Monohybrid Cross

- Cross that considers only a single character (e.g., height of the part)



- Studying the cross:
    - TT, tt, and Tt are genotypes while the traits, tall and dwarf, are phenotypes.
    - T stands for tall trait while t stands for dwarf trait.
    - Even if a single ‘T’ is present in the genotype, phenotype is ‘tall’. When ‘T’ and ‘t’ are present together, ‘T’ dominates and suppresses the expression of ‘t’. Therefore, T (for tallness) is dominant trait while t (for dwarfness) is recessive trait.
    - TT and tt are homozygous while Tt is heterozygous.
    - From the cross, it can be found that alleles of parental pair separate or segregate from each other and only one allele is transmitted to the gamete.
    - Gametes of TT will have only T alleles; gametes of tt will have only t alleles, but gametes of Tt will have both T and t alleles.
- **Punnett square**
    - Graphical representation to calculate the probability of all possible genotypes of offsprings in a genetic cross
    - Possible gametes are written on two sides, usually at top row and left columns, and combinations are represented in boxes.



- With the help of Punnet square, genotypic ratio in F_2 generation can be found. From the above given Punnet square, it is evident that genotypic ratio TT: Tt: tt is 1:2:1.
- The ratio 1:2:1 or $\frac{1}{4}:\frac{1}{2}:\frac{1}{4}$ of TT: Tt: tt can be derived from binomial expression $(ax + by)^2$.
- Gamete-bearing genes are in equal frequency of $\frac{1}{2}$.
- Hence, the expression can be expanded as

$$\left(\frac{1}{2}T+\frac{1}{2}t\right)^2=\left(\frac{1}{2}T+\frac{1}{2}t\right)\times\left(\frac{1}{2}T+\frac{1}{2}t\right)$$
$$=\frac{1}{4}TT+\frac{1}{2}Tt+\frac{1}{4}tt$$

Law of Dominance, Test Cross, Law of Segregation & Incomplete Dominance

Mendel's Laws of Inheritance

- Based on his experiments, Mendel proposed three laws or principles of inheritance:
  - Law of Dominance
  - Law of Segregation

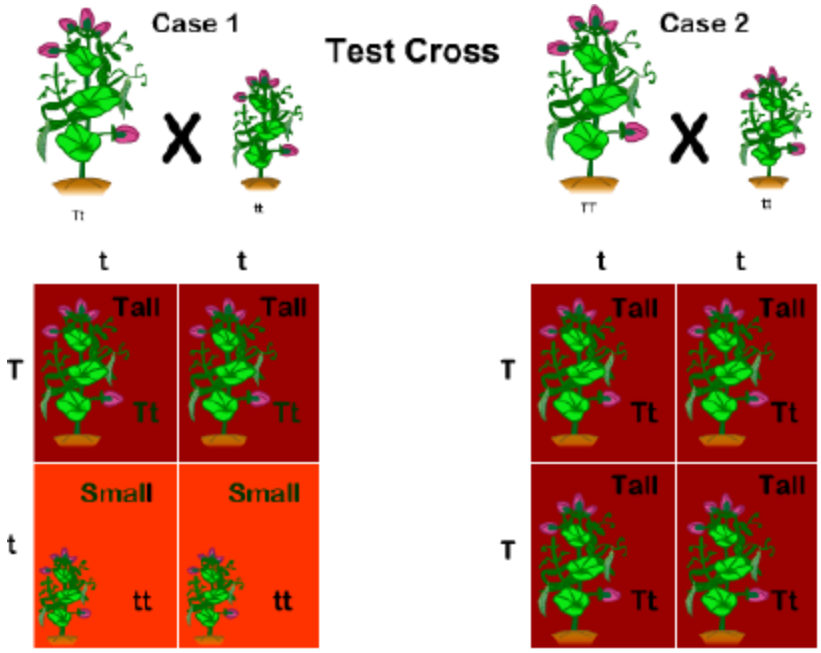
    - Law of Independent Assortment
- Law of dominance and law of segregation are based on monohybrid cross while law of independent assortment is based on dihybrid cross.

Law of Dominance

- According to this law, characters are controlled by discrete units called factors, which occur in pairs with one member of the pair dominating over the other in a dissimilar pair.
- This law explains expression of only one of the parental character in F_1 generation and expression of both in F_2 generation.

Test Cross

- Cross between F_2 progeny and its homozygous recessive parent
- This cross determines whether the dominant character is coming from homozygous dominant genotype or heterozygous genotype. (e.g., tallness coming from TT or Tt)
- When TT is crossed with tt, we obtain all Tt (tall) individuals in the progeny. Whereas when Tt is crossed with tt, we obtain Tt (tall) and tt (dwarf) individuals in the progeny.
- Therefore, if tallness is coming from TT, then we obtain all tall progenies in test cross. We obtain both tall and dwarf varieties in test cross, if tallness is coming from Tt.

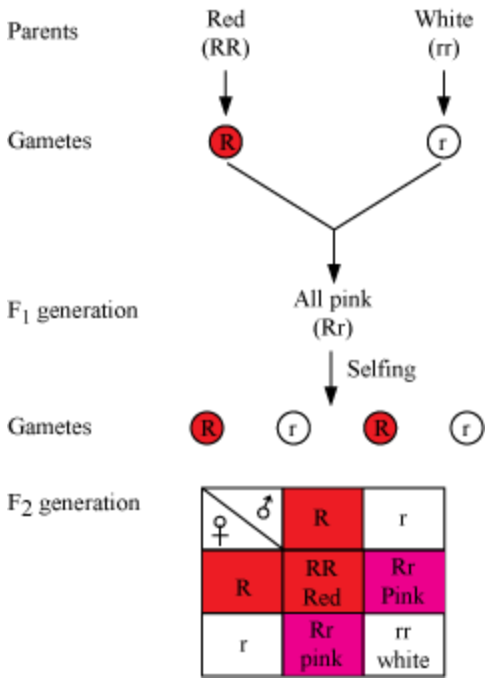


Law of Segregation

- This law states that the two alleles of a pair segregate or separate during gamete formation such that a gamete receives only one of the two factors.
- In homozygous parents, all gametes produced are similar; while in heterozygous parents, two kinds of gametes are produced in equal proportions.

Incomplete Dominance

- In incomplete dominance, F_1 generation has a phenotype that does not resemble either of the two parents, but is a mixture of the two.
- Example – Flower colour in dog flower (snapdragon), where:
    - RR – Red flowers
    - rr – White flowers
    - Rr – Pink flowers
- Here, genotypic ratio remains same as in Mendelian crosses, but phenotypic ratio changes since complete dominance is not shown by R (hence, incomplete dominance).



- Phenotypic Ratio – 1:2:1 that denotes Red: Pink: White
- Genotypic Ratio – 1:2:1 that denotes RR: Rr: rr

What is Dominance?

- A diploid organism produces two copies of a gene, which need not be identical and may have minor alterations.
- Suppose a normal gene produces a product P. Then, the altered version of it must produce a non-functional product P’ or no product at all.
- The altered version of the gene must not perform the functions that a normal gene performs. It must affect the phenotype.
- The original gene is said to be dominant while the modified gene is recessive.

Law of Segregation and Co-dominance

Co-dominance

- In co-dominance, the F_1 progeny resembles both the parents.
- Example: ABO blood groups in human beings
- ABO blood groups are controlled by gene *I*. Gene *I* has three alleles, I^A, I^B and *i*. A person possesses any two of the three alleles.

- I^A and I^B dominate over *i*. But with each other, I^A and I^B are co-dominant.
- I^A and I^B contain A and B types of sugar, while *i* does not contain any sugar.

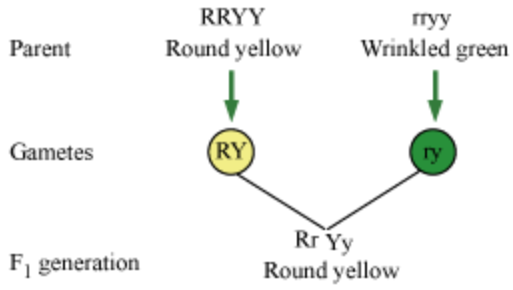
Allele from Parent 1	Allele from Parent 2	Genotype of offspring	Blood type of offspring
I^A	I^A	$I^A I^A$	A
I^A	I^B	$I^A I^B$	AB
I^A	*i*	$I^A i$	A
I^B	I^A	$I^A I^B$	AB
I^B	I^B	$I^B I^B$	B
I^B	*i*	$I^B i$	B
i	*i*	*i i*	O

- Multiple alleles: When more than two alleles control a character, as in human blood groups
    - Multiple alleles are used in population studies.

Inheritance of Two Genes (Dihybrid Cross) & Law of Independent Assortment

Inheritance of Two Genes (Dihybrid Cross)

- In dihybrid cross, we consider two characters. (e.g., seed colour and seed shape)
- Yellow colour and round shape is dominant over green colour and wrinkled shape.



	RY	rY	Ry	ry
RY	RRYY Round yellow	RrYY Round yellow	RRYy Round yellow	RrYy Round yellow
rY	RrYY Round yellow	rrYY Wrinkled yellow	RrYy Round yellow	rrYy Wrinkled yellow
Ry	RRYy Round yellow	RrYy Round yellow	RRyy Round green	Rryy Round green
ry	RrYy Round yellow	rrYy Wrinkled yellow	Rryy Round green	rryy Wrinkled green

- Phenotypic ratio – 9:3:3:1
  Round yellow – 9
  Round green – 3
  Wrinkled yellow – 3
  Wrinkled green –1

Law of independent Assortment

- When two pairs of traits are combined in a hybrid, one pair of character segregates independent of the other pair of character.
- In a dihybrid cross between two plants having round yellow (RRYY) and wrinkled green seeds (rryy), four types of gametes (RY, Ry, rY, ry) are produced. Each of these segregate independent of each other, each having a frequency of 25% of the total gametes produced.

Chromosomal Theory of Inheritance

Rediscovery of Mendel's Work

- Mendel's work remained unrecognised for several years because of the following reasons.
  - Lack of communication and publicity
  - His concept of factors (genes) as discrete units that did not

blend with each other was not accepted in the light of variations occurring continuously in nature.
    - Mendel's approach to explain biological phenomenon with the help of mathematics was also not accepted.
    - In 1990, three scientists Hugo deVries, Correns and Von Tschermak independently rediscovered Mendel's work.

Chromosomal Theory of Inheritance

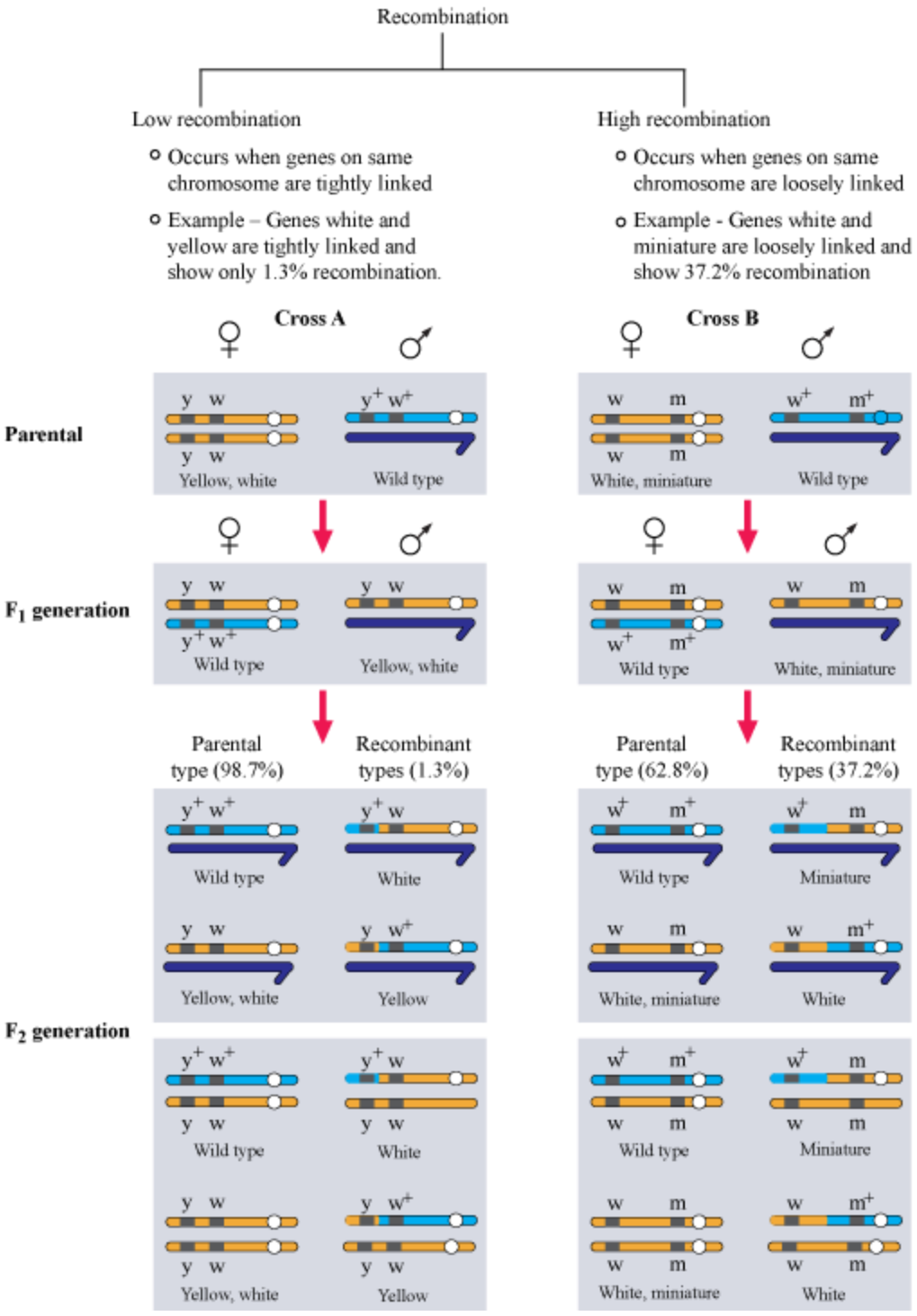
- By 1900, due to the advancement in microscopy, chromosomes were also discovered.
- Sutton and Bovery discovered that the behaviour of chromosomes was parallel to the behaviour of genes.
- Chromosomes and genes both occur in pairs—two alleles of a gene pair are located on **homologous sites of homologous chromosomes**.
- Sutton and Bovery further proposed that it is the pairing and separation of a pair of chromosomes that ultimately leads to segregation of the pair of factors they carry.
- Union of knowledge of chromosomal segregation with Mendelian principles constitutes chromosomal theory of inheritance.

Dihybrid Cross in Drosophila to Study Linkage and Recombination

Linkage and Recombination

- Thomas Hunt Morgan discovered the basis of variations that sexual reproduction produced.
- He worked on fruit flies, *Drosophila melanogaster*. He chose *Drosophila* because of the following reasons:
    - They were suitable to grow on synthetic medium in laboratory.
    - Their life cycle is complete in two weeks.
    - Single mating produces many progeny flies.
    - Clear differentiation of sexes – Easily distinguishable male and female
    - Hereditary variations clearly visible with low power microscopes
- Morgan's experiment

    - Dihybrid cross was carried out on fruit flies. Yellow bodied, white eyed females were crossed with brown bodied, red eyed males.
    - F_1 progeny was obtained, which were inter-crossed.
    - F_2 progeny was obtained and F_2 ratio was observed.
    - F_2 ratio was observed to be significantly different from 9:3:3:1 as observed in Mendelian dihybrid cross.
- Explanation of deviation from Mendelian ratio:
    - Genes involved are located on X chromosome.
    - When two genes are located on the same chromosome, the proportions of parental gene combinations were much higher than those of non-parental.
    - Linkage – Physical association of genes on a chromosome
    - Recombination – Non-parental gene combination



- Alfred Sturtevant utilised the knowledge of frequency of gene recombination as a measure of physical distance between two genes and to map their position on chromosomes.
- In this way, genetic maps were prepared, which are extensively used

today for genome sequencing projects as in human genome project.

Sex Determination in Various Animals Including Humans-Male and Female Heterogamety

Sex Determination

- Henking discovered the genetic/chromosomal basis of sex determination by working on insects. He observed specific nuclear structures during spermatogenesis in insects. He named these structures as X bodies.
- He observed that after spermatogenesis, 50% of the sperm obtained these structures, while 50% did not.
- Later on, it was found that the X body observed by Henking was actually a chromosome and thus, this chromosome was named X chromosome.
- Chromosomes involved in sex determination are called sex chromosomes, while the other chromosomes are called autosomes.
- XO type of sex determination
    - Other than autosomes, at least one X chromosome is present in all insects.
    - Some sperms contain X chromosomes, while some do not.
    - Eggs fertilised by sperms having X chromosomes become females. So, females have two X chromosomes.
    - Eggs fertilised by sperms not having X chromosomes become males. So, males have only one X chromosome.
    - Example of organisms with XO type of sex determination – Insects
- XY type of sex determination
    - Males have X chromosome and its counterpart Y chromosome, which is distinctly smaller. Hence, males are XY.
    - Females have a pair of X chromosomes. Hence, females are XX.
    - Example of organisms with XY type of sex determination – Humans and *Drosophila*
- Male heterogamety – XO and XY types of sex determination are

examples of male heterogamety.
    - In XO type, some gametes have X chromosomes, while some gametes are without X chromosomes.
    - In XY type, some gametes have X chromosomes, while some gametes have Y chromosomes.
- Female heterogamety – ZW type of sex determination is an example of female heterogamety.
    - In ZW type, the female has one Z and one W chromosome, while the male has a pair of Z chromosomes.

Mutation, Pedigree Analysis, & Genetic Disorders

Mutation

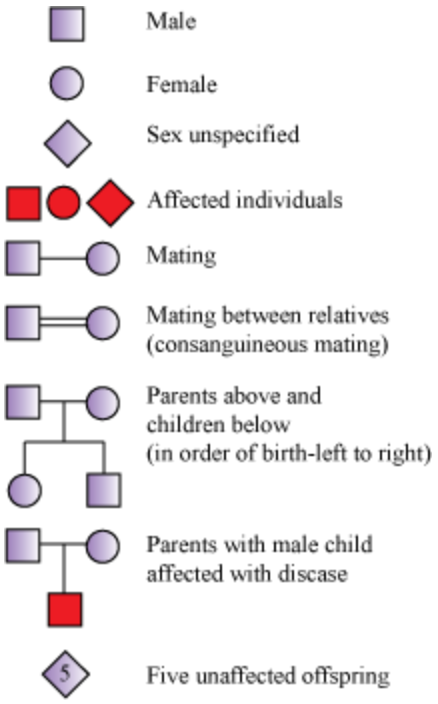
- Alteration of DNA sequence resulting in changes in genotype and phenotype of organisms
- DNA helix runs in a chromatid, hence any change (insertion or deletion) in the DNA sequence affects the chromosome.
- Point Mutation – Mutation arising due to change in single base pair of DNA as in sickle cell anaemia
- Frameshift Mutation – Mutations arising due to deletion or insertion in DNA sequence
- Mutagens – Chemical or physical agents that lead to mutations Example – UV radiations

Pedigree Analysis

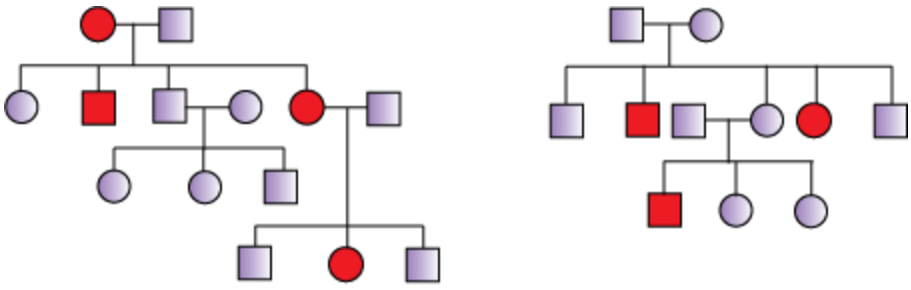
- Pedigree analysis is the analysis of inheritance of traits in several generations of a family.
- A particular trait under study is represented in a family tree.
- By using pedigree analysis, inheritance of a specific trait, abnormality or disease, can be traced.
- DNA is believed to be the carrier of genetic information, which passes unaltered from generation to generation. Mutations occasionally alter the genetic material and genetic diseases are believed to be associated

with these alterations only.

- Standard symbols in pedigree analysis are as follows:



- Pedigree chart is represented as follows:



(a) (b)

Chart (a) represents inheritance of an autosomal dominant trait as in muscular dystrophy.

Chart (b) represents inheritance of an autosomal recessive trait as in sickle cell anaemia.

Genetic Disorders

- Include Mendelian disorders and chromosomal disorders

Mendelian Disorders

- Characterized by mutation in a single gene
- Their mode of inheritance follows the principles of Mendelian genetics.
- Mendelian disorders can be
    - autosomal dominant (muscular dystrophy)
    - autosomal recessive (sickle cell anaemia)
    - sex linked (haemophilia)
- Haemophilia
    - Sex-linked recessive disease
    - Transmission – From unaffected female (carrier) to male progeny
    - Females act as carriers of disease, but rarely suffer from haemophilia since for a female to become haemophilic, the mother should be carrier and father should be haemophilic.
    - In this disease, protein involved in blood clotting is affected. Therefore, even a simple cut results in uncontrolled bleeding.
- Sickle cell anaemia
    - Autosomal recessive disease
    - Transmission – From parent to offspring when both parents are carriers of disease
    - Pair of alleles Hb^A and Hb^S controls the expression of this disease.
      Hb^A and Hb^A – Normal
      Hb^A and Hb^S – Carrier of disease
      Hb^S and Hb^S – Diseased
    - Cause of the disease – Change in gene causes the replacement of GAG by GUG leading to the substitution of Glu by Val at sixth position of beta globin chain of haemoglobin.
    - The mutant haemoglobin so formed polymerises at low oxygen tension, resulting in change in shape of RBC to sickle-like.
- Phenylketonuria
    - Autosomal recessive disease

    - Phenylalanine $\xrightarrow{\text{Enzyme}}$ Tyrosine
      The enzyme responsible for this conversion gets mutated.
    - Phenylalanine accumulates. Then,
      Phenylalanine → Phenylpyruvic acid → Accumulates in brain → Mental retardation
    - Phenylpyruvic acid also gets excreted through urine since kidneys poorly reabsorb it.

Chromosomal Disorders

- Total number of chromosomes in humans = 46 (23 pairs)
- Total 23 pairs = Autosomes (22 pairs) + Sex chromosomes (1 pair)
- Monosomy – Lack of any one pair of chromosomes
- Trisomy – Inclusion of an additional copy of chromosomes
- Aneuploidy – Loss or gain of chromosomes due to failure of segregation of chromatids during cell division

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- Aneuploidy – Loss or gain of chromosomes due to the failure of segregation of chromatids during cell division
- Down's Syndrome
    - Cause: Presence of an additional copy of chromosome 21 (Trisomy of 21)
    - Affected individual has short stature, small, round head, furrowed tongue, partially opened mouth, palm crease, congenital heart disease and mental retardation.
- Klinefelter Syndrome
    - Cause: Additional copy of X chromosome, i.e., 47 chromosomes (XXY)
    - Affected individual has an overall masculine development with gynaecomastia; individual is sterile

- Turner’s Syndrome
    - Cause: Absence of one X chromosome, i.e., 45 chromosomes (XO).
    - Affected females are sterile; have rudimentary ovaries; secondary sexual characters are absent

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