5) Principles of Inheritance & Variation

Genetics deals with inheritance & variation of characters from parents to offspring.

Inheritance is process by which characters are passed on from parent to progen; it is basis of heredity.

Variation is the degree by which progeny differ from their parents.

★ Mendel's Laws of Inheritance

Reasons for Mendel's Success

→ First time, statistical analysis & mathematical logic were applied in biology.
→ Large sampling size gave credibility to his data.
→ Also, his confirmation of inferences from experiments on successive generations of his test plant proved that his results showed general rules of inheritance rather than unsubstantiated ideas.

★ True breeding line means one which having undergone succesive self-pollination, shows stable trait inheritance and expression for several generations.

DOMS | Page No.
Date / /

★ Reason's for choosing garden pea :-
1) Pea has many clear contrasting traits.
2) Produces large number of seeds and ~~produces~~ completes life cycle in one season.
3) Flowers show self pollination.
4) Easy to cross-pollinate them. So hybrid produced is also fertile.

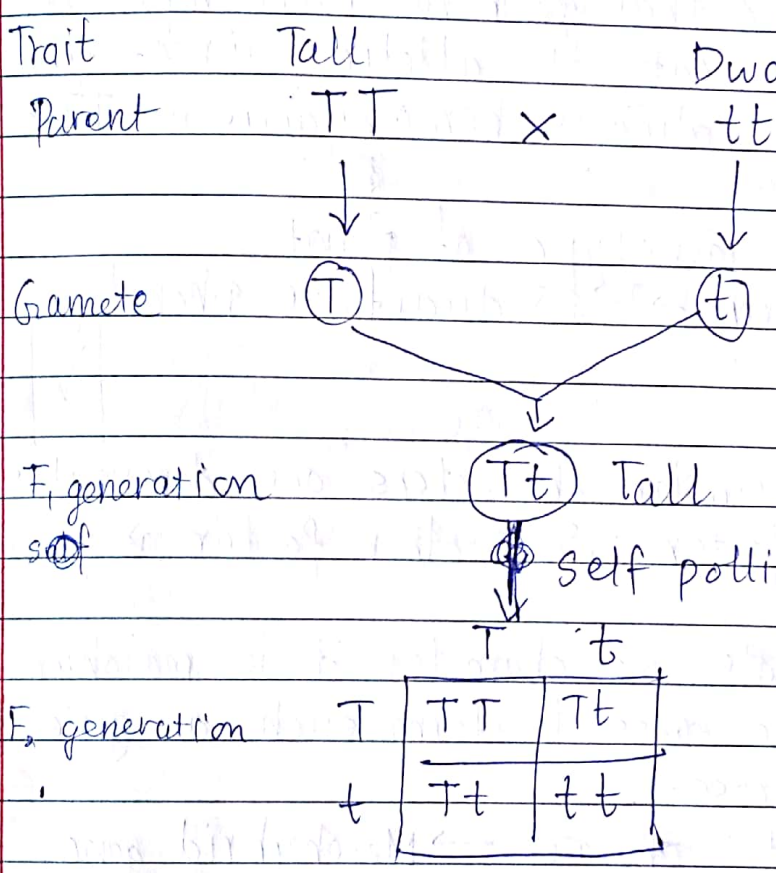
	Charater	Dominant Trait	Recessive Trait
1)	Seed Shape	Round	Wrinkled
2)	Seed colour	Yellow	Green
3)	Flower colour	Violet	White
4)	Pod Shape	Full (inflated)	Constricted
5)	Pod Colour	Green	Yellow
6)	Flower Positin	Axial	Terminal
7)	Step Height	Tall	Dwarf

Inheritance of One Gene :-

Tall & dwarf plants are crossed.
Seeds produced due to this cross are collected and grown to get plants of first hybrid generation. This generation is called Fillial$_1$ of F_1 generation.
All F_1 progeny are tall.
Same observation of F_1 being similar to only one of the parent and not other with different contrasting traits.

Then, Self pollination of F_1 tall plants.
F_2 generation, dwarf character seen inspite of its absence in F_1 generation.
1/4th of F_2 plants → Dwarf } → Both identical to
3/4th of F_2 plants → Tall } original parents & no blending i.e medium sized seen.

Similar results of F_1 similar to one parent & F_2 in ration 3:1 seen with diff. traits.
No blending at F_1 or F_2



Phenotypic ratio :- Tall : Dwarf
3 : 1
Genotypic ratio → TT : Tt : tt
1 : 2 : 1

DOMS Page No.
Date / /

Gene → called as factors by Mendel
- → Stably passed down from parents to offsprings, unchanged, for generations
- → Units of inheritance.
- → Contain information required for expressing a trait.

Allele → Genes which code for pair of contrasting traits
- → slightly diff. forms of same gene.

→ Mendel also said that in true breeding, tall or dwarf pea variety the allelic pair of genes for height is always homozygous i.e. TT or tt.

→ TT & tt are genotype of plant

→ Descriptive terms → Tall & dwarf are phenotype of plant.

→ In pair of dissimilar characters one dominates and is dominant factor while other factor is recessive.

→ If gene controls one character it is monohybrid gene and the cross to form such a gene is monohybrid cross.
Tt → here only tall charater → Monohybrid gene
TT x tt → Monohybrid cross

→ When plants produce gamete by meiosis alleles of parental pair separate or segregate and only one allele is transmitted. Segregation of allele is random process.

→ Plants having alleles having contrasting traits are heterozygous. Eg:- Tt → Tall plant

→ Punnet square is a graphical representation to calculate probability of all possible genotypes of offsprings in a genetic cross.

From monohybrid cross, for character → height of pea plant we have,

TT : Tt : tt

1/4 : 1/2 : 1/4

This ratio can be mathematically condensed to the form of binomial expression $(ax+by)^2$, that has gametes having T & t in equal frequency of (1/2)

$$\left[(1/2 \times T) + (1/2 \times t)\right]^2 = (1/2\,T + 1/2\,t) \times (1/2\,T + 1/2\,t)$$

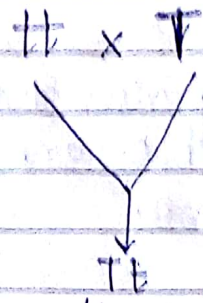
$$= 1/4\,TT + 1/2\,Tt + 1/4\,tt$$

Mendel self pollinated F_2 plants and found that dwarf F_2 plants continued to generate dwarf plants in F_3 & F_4 generations.

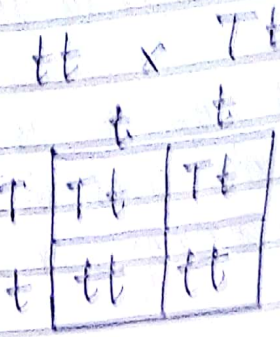
∴ dwarf plant genotype → tt

For tall plants, to find genotype, Mendel crossed tall plants of F_2 generation with a dwarf plant. This he called test cross.

Eg,



All plants are tall



50% are tall and 50% are dwarf

Hence, using test cross we predict genotype of Dominant phenotype.

★ Principles of Inheritance

1) Law of Dominance

→ Characters are controlled by discrete units called factors.
→ Factors occur in pairs
→ In dissimilar pair of factors one member dominates over other.

2) Law of Segregation.

→ Law is based on the fact that alleles do not show blending and both characters are recovered in F_2 generation & one of them is not seen in F_1 generation.
→ Parents have 2 alleles during gametogenesis and the allels segregate in such a way that gamete gets only one of the two genes or factors.

☆ Incomplete Dominance

→ Some cases are observed where F_1 generation's phenotype is intermediate between dominant and recessive phenotype & it does not resemble either of the two parents. This is incomplete dominance

Egs:- Antirrhinium or dog flower ⟶ flower colour

Parents (True breeding line): Red RR × White rr

Gametes: R, r

F_1 generation: Rr All pink

↓ selfing

F_2 generation

	R	r
R	RR	Rr
r	Rr	rr

Phenotypic ratio :- Red : Pink : White
1 : 2 : 1

Genotypic ratio → RR : Rr : rr
1 : 2 : 1

Here genotypic ratio are ideal as per Mendel's law but phenotypic are not, as 'R' is not dominant over 'r' completely

☆ Concept of Dominance :-

Every gene contains information to express a trait. In a allele there are 2 genes if they are different they modify particular information.

Eg:- Gene having info. about producing an enzyme. Now normal allele will produce normal enzyme to transform substrate 'S'. Now if allele is modified :-

① If modified allele = Unmodified allele, then it will produce same or normal enzyme to transform 'S'

② If modified allele produces non-functional or no enzyme then, phenotype will be only dependent on original unmodified allele which becomes dominant & modified allele becomes recessive.

☆ Co-dominance :-

F1 generation resembles both parents.

Eg:- ABO blood group system

→ They are controlled by gene I having 3 alleles, I^A, I^B & i

I^A & I^B produce slightly diff. sugar & i doesn't produce

→ If I^A & I_B express with 'i' only I^A or I^B expresses as they are dominant over 'i'

→ If I^A & I^B are present together both express because of co-dominance. So, in humans there are 6 genotype of ABO system & 4 phenotypes

Genotypes - I^Ai, I^Bi, I^AI^A, I^BI^B, I^AI^B, ii

Phenotypes - Blood grp :- A, B, AB, O

☆ Multiple Allelism :-

Prescence of more than 2 alleles or Multiple alleles governing same character is Multiple Allelism.

Eg:- ABO blood group system.
3 alleles I^A, I^B & i govern same character of producing sugars in RBC.

⇒ Dihybrid Cross :-

Parents: Round Yellow RRYY × Wrinkled green rryy

Gametes: RY, ry

F1 generation: RrYy → Round Yellow

↓ selfing

F2 generation:

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

Phenotypic Ratio :- Round Yellow : Round Green : Wrinkled Yellow : Wrinkled G
9 : 3 : 3 : 1

★ Law of Independent Assortment :-

→ When 2 pairs of traits are combined in a hybrid, segregation of one pair of characters is independent of the othe pair of characters.

→ Explaination :-
Consider segregation of one pair of Genes R & r in a F_1 RrYy plant. 50% gametes have R & other 50% gametes have r and also besides this, they have allele Y or y. Here the segregation of 50% R & 50% r is independent of segregation of 50% Y & 50% y.

50% 'r' gametes must have 50% Y & 50% y.
Also, 50% 'R' gametes have 50% Y & 50% y.

Thus; there are four genotypes of gametes each with frequency 25% or 1/4th of total gametes produced.

★ Chromosomal Theory of Inheritance

→ Reason's for Mendel's work being unacceptable then :-
1) Communication was a problem & his work could not be widely published
2) Concept of genes (factors) as stable & discrete units that controlled expression of traits & of pair of alleles which do not blend was not accepted by contemporaries as an explanation for continuous variation.

→ 1900 → de Vries, Correns & von Tschermak independently rediscovered Mendel's result on Inheritance

→ Walter Sulton and Theodore Boveri
→ They stated that behaviour of chromosomes was similar or parallel to behaviour of genes and used chromosome movement to tell about Mendelian law's.
→ Also, chromosomes as well as genes occur in pairs. Two alleles of gene pair are located on homologous sites on homologous chromosomes
→ During Anaphase of meiosis-I, the two chromosomes pairs align at metaphase plate independently of each other (Refer pg 82, NCERT)
→ They argued that pairing and segregation of a pair of chromosomes would lead to segregation of a pair of factors they carried.

→ Sutton united knowledge of chromosomal segregation with mendelian principles and called it Choromosomal Theory of Inheritance

★ Linkage and Recombination

T.H. Morgan and his colleques did experimental verification of chromosomal theory by carrying out several dihybrid crosses in Drosophila melanogaster.

DOMS | Page No.
Date / /

☆ → Reasons for choosing Drosophila Melanogaster:
1) They could be grown on simply synthetic medium in Laboratory.
2) Complete life cycle in 2 weeks and single mating produces large number of progenies
3) Clear differentiation of male & female sexes are easily distinguishable
4) Has many hereditary variations
5) Can be observed using low power Microscope.

→ Morgan did dihybrid cross of yellow bodied, white eyed females to brown bodied, red eyed males and intercrossed their F_1 progeny.
→ He observed that genes did not segregate independently F_2 ratio deviated significantly from 9:3:3:1
→ He observed that in two genes grouped on same chromosome, propotion of parental gene combination was higher that non-parental.
Here he coined two terms:-
☆ Linkage → physical association of genes on a chromosome
☆ Recombination → Generation of non-parental gene combinations.
→ In genes grouped on same chromosome;
some where tightly linked → Very low Recombination
Loosely linked → Higher recombination.

→ Alfred measured distance b/w genes and prepared maps with position of genes on chromosomes based on percentage of recombinants. These are genetic Maps

Sex Dermination Mechanisms

Male Heterogamety

→ Male produces 2 different types of gametes

- XX-XY type
- XX-XO type

Female Heterogamety

→ Female produces 2 different types of gametes.

- ZZ-ZW type
- ZZ-ZO type

1) XX - XY Type

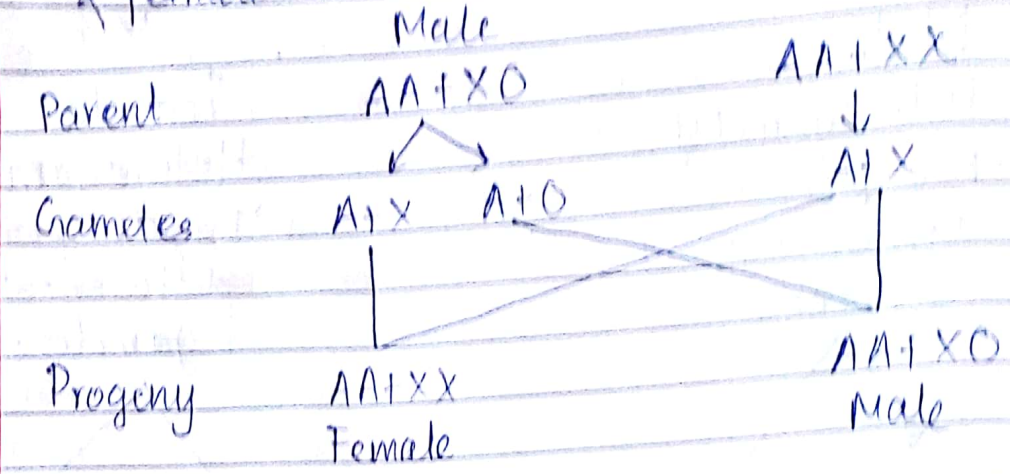
→ Males have X & Y chromosomes along with autosoms & females have pair of X chromosomes.

	Male	Female
Parents	AA+XY	AA+XX
Gametes	A+X, A+Y	A+X
Progeny	AA+XX Female	AA+XY Male

Egs:- Humans, Drosophila, insects etc.

2) XX - XO type

→ Male only one X chromosome besides autosomes & female 2 X chromosomes



Eg:- Grasshopper.

3) ZZ-ZW type

→ Females have one Z & one W & males have 2 Z chromosomes

→ Eg:- Birds, Fowls & fishes

* Diagram similar to XX - XY

4) ZO - ZZ

→ Females have one chromosome Z & autosomes & males have 2 Z chromosomes

→ Eg:- Cockroaches

* Diagram similar to XO - XX type.

* Mutation is a phenomenon which results in alteration of DNA sequences and consequently results in changes of genotype and phenotype of organism. It also leads to variation in DNA.

DOMS | Page No.
Date / /

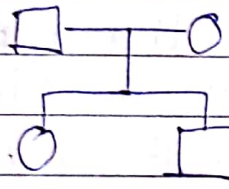
✱ Pedigree Analysis

Analysis of traits in several generations of a family is Pedigree analysis. Here, the inheritance of a particular trait is represented in a family tree over generations.

→ Help to trace the inheritance of a specific trait, abnormality or disease.
→ Helps in genetic counselling to avoid disorder.

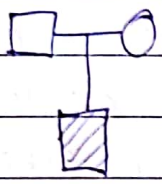
Symbols :-

□ → Male ○ → female
◇ → Sex-unspecified
■ ● ◆ → Affected individuals
□—○ → Mating □=○ → Mating b/w relatives



Parents

children (in order of birth left-right)



Male child affected

◇5 Five unaffected offsprings.

Mendelian Disorders

1) Haemophilia :-

→ Sex linked recessive disease
→ Shows transmission from unaffected carrier female to male progeny.
→ Single protein which is part of a cascade of proteins involved in clotting of blood is affected.
→ Single cut also causes non-stop bleeding in affected individuals.
→ Heterozygous female (carrier - X^cX) may transmit disease to sons (X^cY)
→ Chances of female being haemophilic (X^cX^c) is very less as for this mother has to be atleast carrier (X^cX) & father haemophilic (X^cY)
→ Queen Victoria → carrier of disease.

2) Sickle-cell Anaemia :-

→ Autosome linked recessive trait.
→ Parents must be heterozygous or carrier of gene (having pair Hb^A & Hb^S) i.e parent should be atleast (Hb^AHb^S) carrier to transmit disease to child and make him diseased (Hb^SHb^S)
Heterozygous are unaffected but are carriers
→ Caused by substitution of Glutamic acid by Valine at sixth position of β-globin chain of haemoglobin.
Substitution results due to single base substitution at sixth codon of beta globin gene from GAG to GUG.
The mutant haemoglobin molecule undergoes polymerisation under low oxygen tension causing change in shape of RBC.

5) Phenylketonuria :- Inborn error of metabolism also inherited as autosomal recessive trait.
→ Affected individual lacks enzyme that converts amino acid phenylamine into tyrosine.
So, [phenylamine is accumulated] → converted → [phenylpyruvic acid]
Accumulation of this acid in brain causes mental retardation.

Chromosomal Disorders :-

→ Failure of segregation of chromatids during cell division cycle results in gain or loss of chromosomes called aneuploidy.
→ Failure of cytokinesis after telophase, results in increase in whole set of chromosomes in organism, this is polyploidy.
→ Additional copy of a chromosome → Trisomy
Lack of any chromosomes → Monosomy.

1) Down's Syndrome :- by Langdon Down (1866)
Cause :- Prescence of additional copy of chromosome no. 21 (trisomy of 21)
Symptoms :- Short height, small round head, ~~for~~ furrowed head, partially open mouth, broad palm, physical & mental growth retarded
2) Klinefelter's syndromy :- Additional X chromosome resulting in karyotype of 47, XXY
Symptoms :- Overall masculine development but feminine development also seen (like gynaecomastia), sterile.
3) Turner's Syndrome :- Caused due to absence of one of X chromosomes i.e. 45 with XO.
Symptoms :- Females are sterile with rudimentary ovaries. Lack other 2° sexual characters.