INHERITANCE

Gene

- A unit of inheritance, made up of a segment of DNA containing a sequence of nucleotides that codes for one specific polypeptide / protein that controls a trait in an organism.

Allele

- alternative forms of the same gene.
- alleles occupy the same relative position on a pair of homologous chromosome.

Dominant allele

- traits are expressed in a homozygous or heterozygous condition
- in heterozygous condition, dominant allele will mask / suppress the expression of the recessive allele.

Recessive allele

- Traits are expressed only in the presence of an identical allele in a homozygous condition.

Homologous chromosome

- A pair of chromosome, where one chromosome in the pair comes from the female parent and the other from the male parent.
- They have the same shape, length and position of centromere.
- They have the same sequence of gene loci.

Genotype

- combinations of alleles for a particular gene
    a) Homozygous: having two identical alleles controlling the trait (e.g. DD or dd).
    b) Heterozygous: having two different alleles controlling the trait (e.g. Dd).

Phenotype

- Expressed physical trait of an organism
- <u>Can be seen externally:</u> eye/hair/skin colour, presence of dimples, chin shape, types of earlobes etc
- <u>Cannot be seen externally:</u> blood types, resistance to certain diseases etc

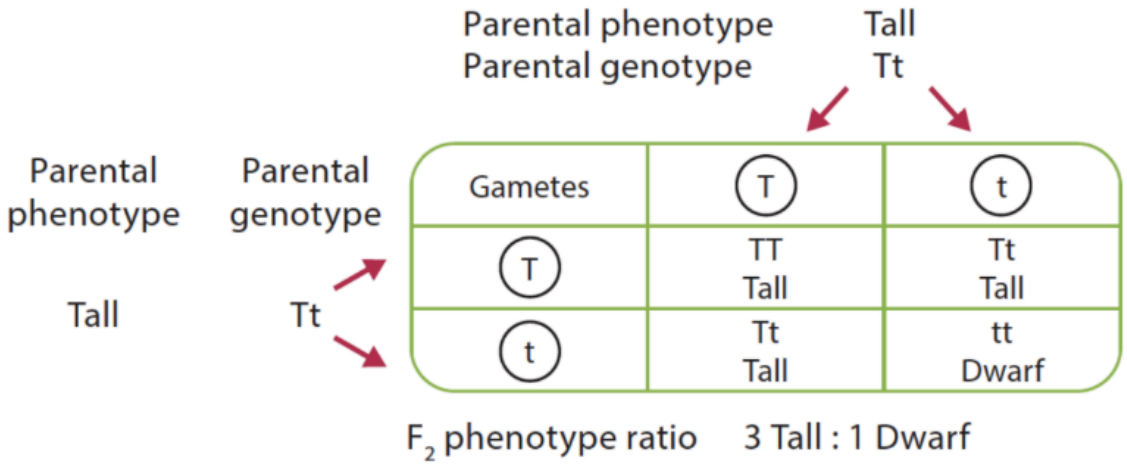
Phenotype is the expressed physical traits of an organism whereas genotype is the specific combination of alleles of a gene.

Pure-bred : Individuals who are homozygous at a particular gene locus.

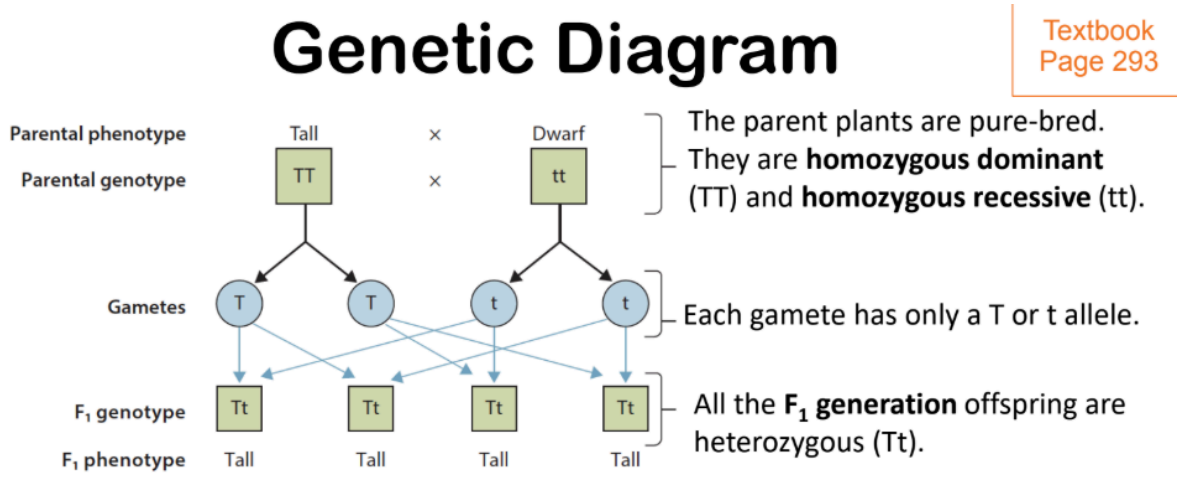
Selfing: fertilisation of gametes from same individual.

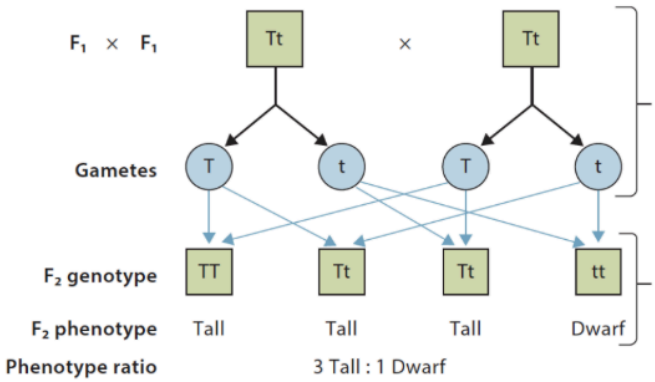
RATIOS

- The observed ratio often differs from the expected ratio of 3:1 because of small sample size and the random nature of fertilisation.



and





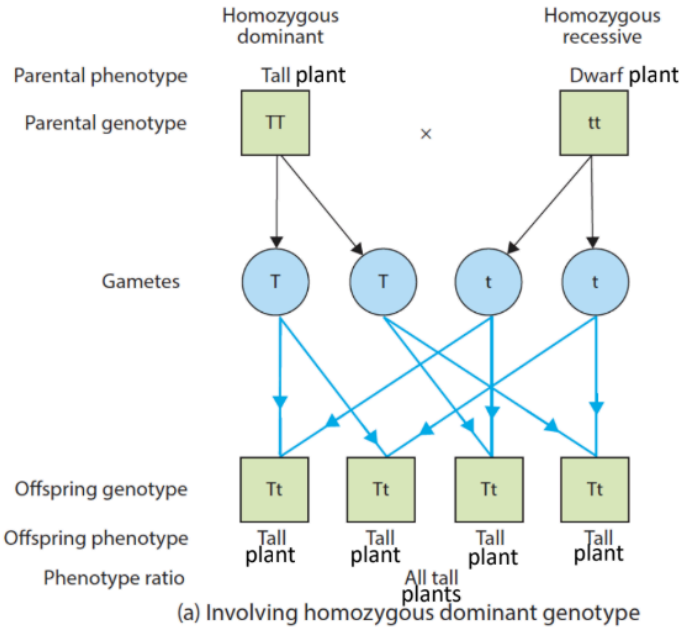
The F_1 generation offspring interbreed with one another.

The phenotype ratio among the **F_2 generation** offspring is **3:1**.

Test Cross

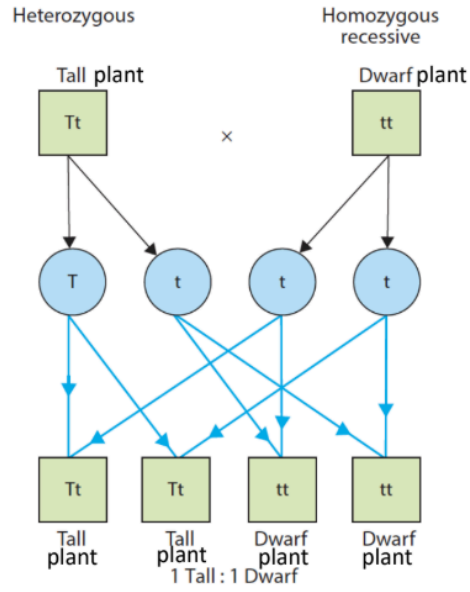
Textbook Page 294

[Possibility 1]: If the organism is **homozygous dominant**, all the offspring should show the dominant trait.



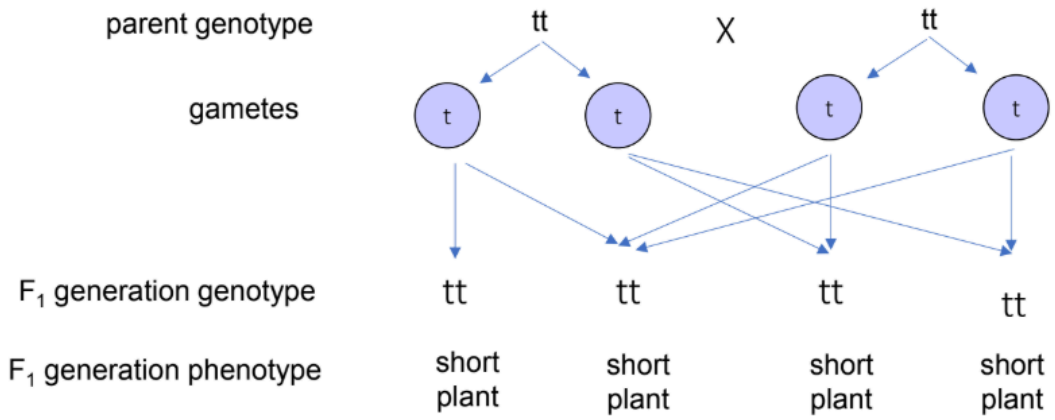
(a) Involving homozygous dominant genotype

[Possibility 2]
If the organism is **heterozygous**, half the number of offspring should show the dominant trait. The remaining half should show the recessive trait.



(b) Involving heterozygous genotype

If a parent is **homozygous recessive** (tt)



Sex determination in man

- Chromosomes that determine gender of an organism is known as sex chromosomes. The rest of the chromosomes are known as autosomes.
- Humans have a pair of sex chromosomes. The Y chromosome determines that the person is a male.

Females have XX chromosome at position 23
Males have XY chromosome at position 23

VARIATIONS

- Variations refers to the differences in traits between individuals of the same species.
- The traits of an individual is dependent on interactions between the genes and the environment.
- Genetic variation is heritable, but variations due to the environment are not.

Discontinuous Variation

- Clear cut phenotypes
- There are few or no intermediate forms of the phenotype.

  Challenge: Give some other examples of discontinuous variations in humans.
  Answer: Fingerprints, gender, ability to roll tongue

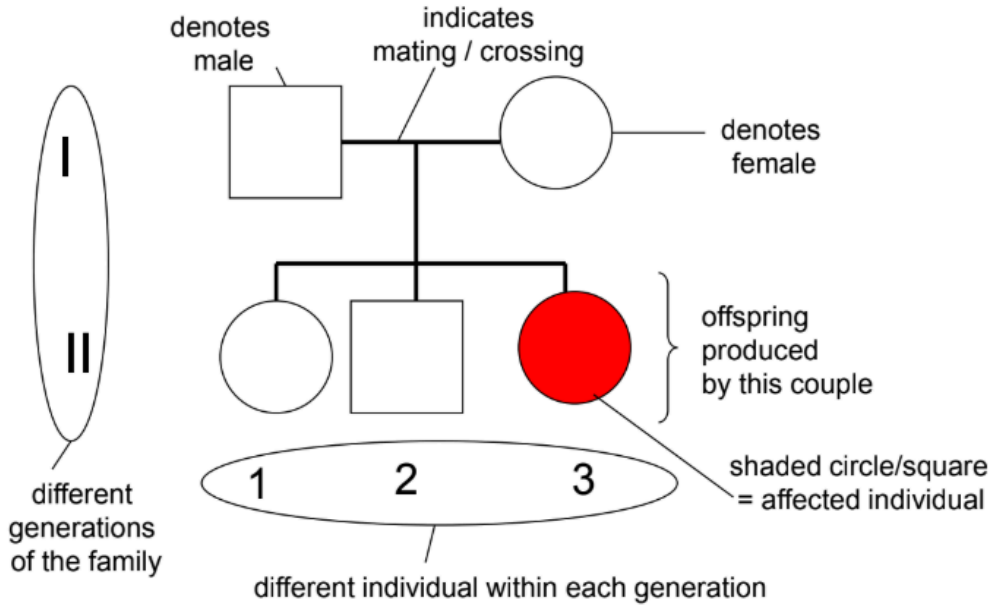
Continuous Variation

- There are many intermediate forms.
- It is controlled by the additive effect of numerous genes.
  (note: discontinuous variation is controlled by one or a few genes)

Differences between Discontinuous Variation and Continuous Variation

Discontinuous Variation	Continuous Variation
Involves a few clear-cut phenotypes (e.g. green or yellow seeds in pea plants)	Involves a range of phenotypes (e.g. human skin colour)
Controlled by one or a few genes	Controlled by many genes
Genes do not show additive effect	Genes show additive effect
Relatively unaffected by environmental conditions	Greatly affected by environmental conditions

Pedigree Chart



Genetic Diagrams vs Pedigree chart

- Genetic diagrams show the probability of genotypes/phenotypes of the offspring due the crossing of two individuals.
- Pedigree chart shows the actual phenotypes of the offspring due to the crossing of two individuals.

In a pedigree chart, the actual genotype of the individual can only be determined when there are enough information of the individual's parents or offspring.

To deduce the genotype of the individual, explanation should include the allele passed on by the individual to his offspring or the allele that he inherited from his parents.

MUTATIONS

Mutation is the sudden, random change in gene structure or chromosome number.
Mutation results in varieties of individuals in a species.

- Mutations may be inherited by the next generation if they occur during gamete production.
- Mutations that occur in somatic cells will not be passed down from parents to offspring.
- Dominant mutations are easily detected unlike recessive mutations, which may not be detectable for generations.
- Mutations may be caused by change in number of chromosome or structure of gene.

TYPES OF MUTATION

Chromosome mutation

- change in number of chromosomes (e.g. Down syndrome)

Gene mutation

- change in sequence of nucleotide bases resulted in a change in gene structure
- may affect amino acids produced → may affect protein formed
- variation introduced because of new alleles of genes
    - e.g. albinism and sickle-cell anaemia

Sickle cell anaemia

- Gene mutation caused by recessive allele.
- Individuals who suffer from sickle cell anaemia are homozygous recessive.

› when oxygen concentration is low, *RBC* of affected individuals are *sickle-shaped*

- reduced surface area to volume ratio results in slower rate of transport of oxygen around the body
- can be fatal

(change in sequence of bases › change one amino acid › haemoglobin S (HbS) protein is produced instead of haemoglobin A (HbA) › changes three dimensional shape of haemoglobin molecule › HbS molecules tend to clump together › red blood cell appear sickle-shaped)

Down Syndrome

- a *chromosome mutation*
- individual has *three copies of chromosome 21*
- linked to reduced life span, varying degrees of mental retardation, heart defects

SYNDROMES

- low muscle tone
- small stature
- upward slant to eyes
- flat facial profile
- single deep crease across centre of palm
- prone to gum disease and dental problems, problem with upper part of spine, poor development of small bowel and blockage of the large intestine and with an inability to pass stool, reduced fertility in males, prone to ADHD, autism, slow learning etc