SUBJECT CODE: 18BBO54C-UNIT-III

Extrachromosomal Inheritance

Definition:

- *"The extrachromosomal DNA present in the cytoplasm and not on chromosomes which follows the non-Mendelian pattern of inheritance is known as extrachromosomal inheritance."*
- Unlike nuclear DNA, the organelle DNA or the extrachromosomal DNA has its own replication and transcription machinery. It synthesised their won DNA.

- **What is Sterility?**
  - ✓ An inability of a living organism to effect sexual reproduction.
- **What is Male Sterility?**
  - ✓ It is the failure of plants to produce functional anthers, pollen, or male gametes.
  - ✓ Occurs mainly in bisexual plants.
  - ✓ **J.K. Koelreuter (1763)** observed anther abortion within species & species hybrids.
  - ✓ More prevalent than female sterility.

Phenotypic Expression Of Male Sterility

- Absence of male sex organs.
- Lack of normal anther sac.
- Inability of the pollen to mature.
- Inability to develop normal pollen.

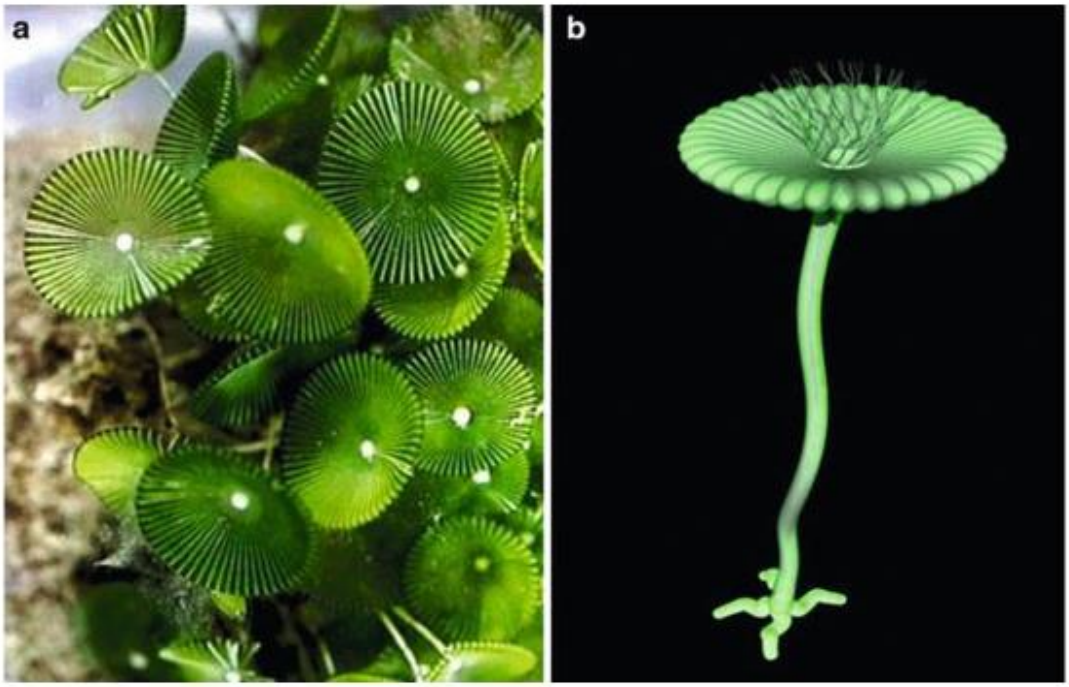
INTRODUCTION

Male Sterility in Plants

Male sterility is characterised by nonfunctional pollen grains, if and where produced, while female gametes function normally.

Features of Male Sterility

- Prevents self pollination, permits cross pollination.
- Leads to heterozygosity
- Female gametes function normally
- Assayed through staining techniques
- In nature, occur due to spontaneous mutations
- Can be induced artificially



- Hammerling's experiment with the single celled green algae, *Acetabularia*, showed that **the nucleus of a cell contains the genetic information that directs cellular development**.
- *A. mediterranea* has a smooth, disc shaped cap, while *A. crenulata* has a branched, flower-like cap. Each *Acetabularia* cell is composed of three segments: the "foot" or base which contains the nucleus, the "stalk," and the "cap."

  In his experiments, Hammerling grafted the stalk of one species of *Acetabularia* onto the foot of another species. In all cases, the cap that eventually developed on the grafted cell matched the species of the foot rather than that of the stalk. In this example, the cap that is allowed to grow on the grafted stalk looks like the base species one... *A. mediterranea.*
- This experiment shows that the base is responsible for the type of cap that grows. The nucleus that contains genetic information is in the base, so the nucleus directs cellular development.

Cytoplasmic Male Sterility

Cytoplasmic male sterility is governed by cytoplasmic factors.In fertilization process, zygote is formed by the fusion of egg cell and one male gamete. The zygote carries equal number of chromosomes from both (male & female) gametes, but cytoplasm of egg cell (female). So, the cytoplasmic male sterility shows maternal inheritance.

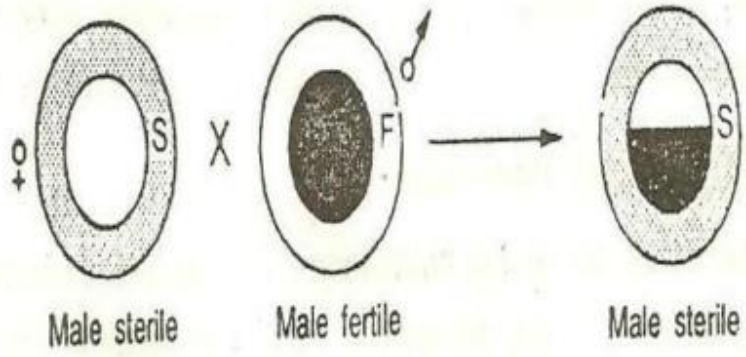
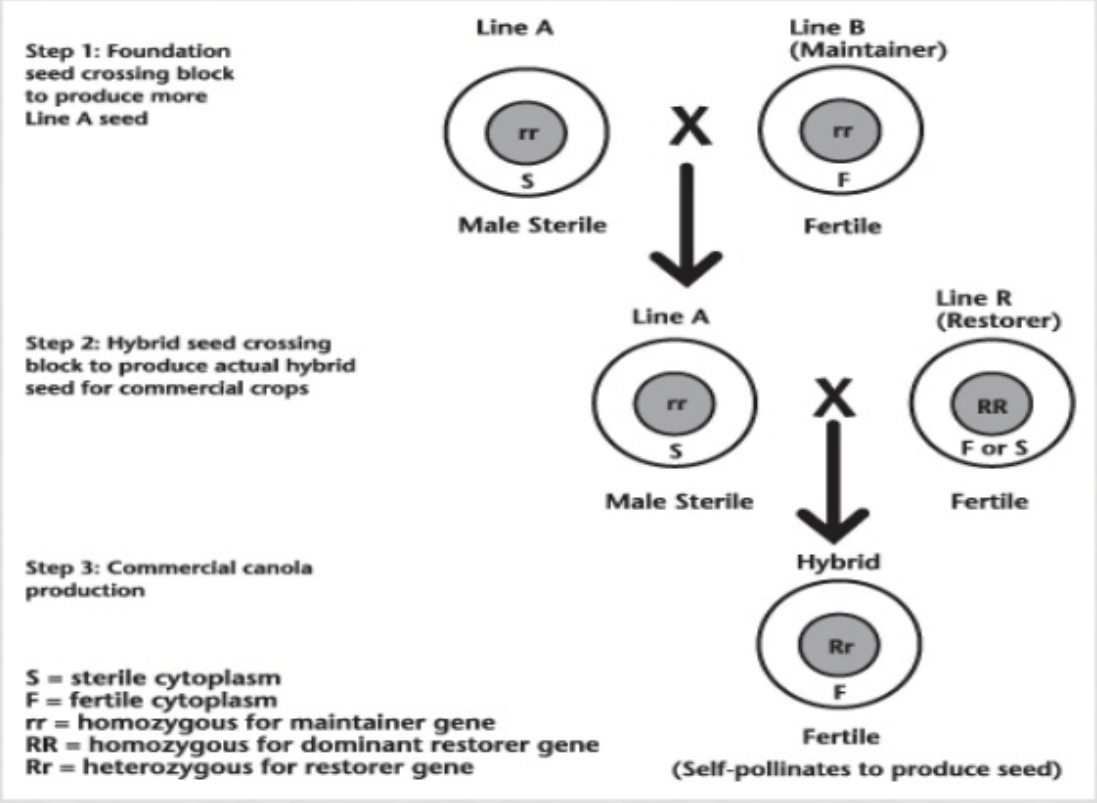
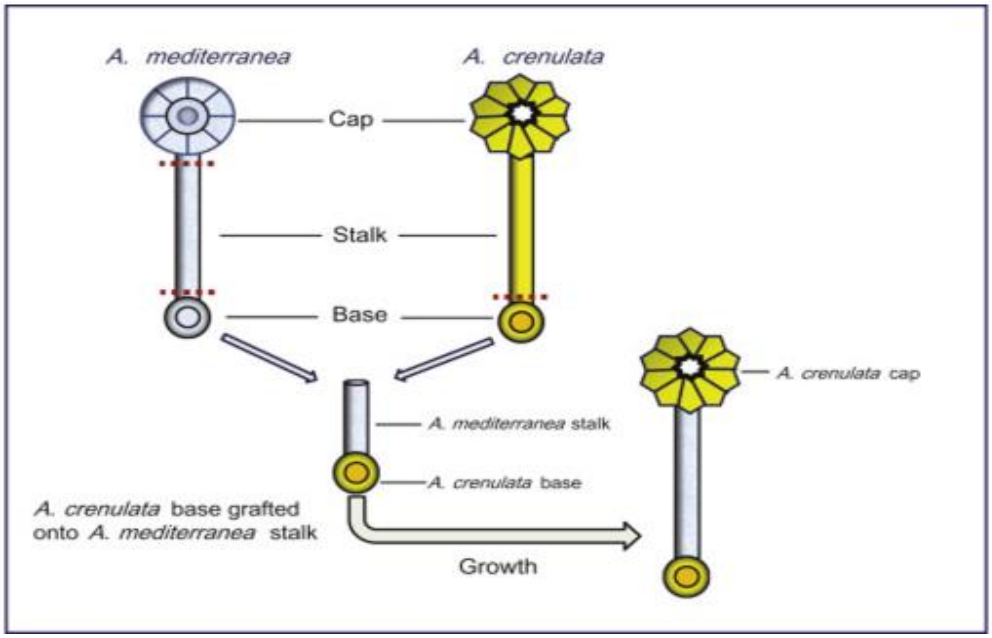
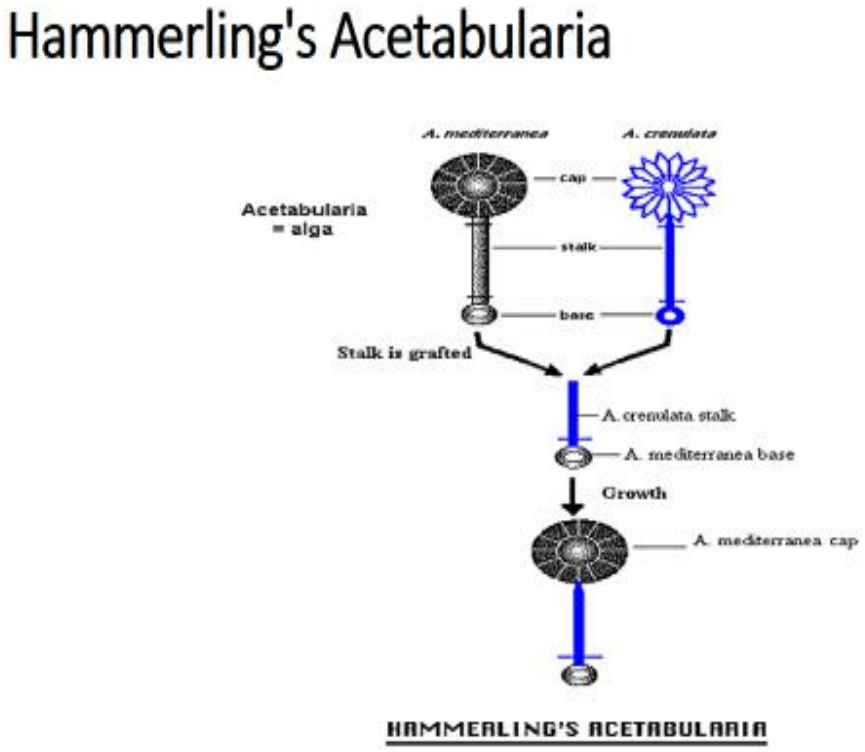


Fig. 4.1. Inheritance of cytoplasmic male-sterility.

Use of CMS lines







Maternal Effects:

In case of cytoplasmic inheritance, distinct maternal effects are observed. This is mainly due to more contribution of cytoplasm to the zygote by female parent than male parent. Generally ovum contributes more cytoplasm to the zygote than sperm.

Plastid Inheritance:

Chloroplasts are the important plastids. Plastids have green pigments called chloroplasts. Plastids self-duplicate, have some amount of DNA and play an important role in cytoplasmic inheritance.

(i) Mirabilis jalapa:

The first conclusive evidence of cytoplasmic inheritance was reported by Correns in 1909 for leaf colour in four 'o' clock plant (Mirabilis jalapa). This plant has three types of leaves, viz., and green, white and variegated. Three types of results were obtained from crosses between these genotypes as given below.

1. When green was used as female and either green, white or variegated as male, all individuals in F_1 were green.

2. When white was used as female and either green, white or variegated as male, all individuals in F_1 were white.

3. When variegated was used as female and either green, white or variegated as male, various proportions of green, white and variegated individuals were obtained in F_1 (Table 11.3).

TABLE 11.3. Inheritance of leaf colour in Mirabilis jalapa

Crosses between three leaf colours			*Expression of leaf colour in F_1*
Female		*Male*	
Green	×	Green	Green
	×	White	Green
	×	Variegated	Green
White	×	Green	White
	×	White	White
	×	Variegated	White
Variegated	×	Green	Green, white and variegated in various ratios in each cross
	×	White	
	×	Variegated	

The inheritance is governed by chloroplasts which are originated from proplastids. If the proplastids are normal, they will develop into normal chloroplasts and when proplastids are mutants, they will produce white chloroplasts. This suggests that green leaf branches have normal chloroplasts; white branches have mutant chloroplasts and variegated have a mixture of both normal and mutant chloroplasts.

Since cytoplasm is contributed to the zygote mainly by female parent, the plastids are transmitted to the zygote from the female parent. These plastids are responsible for variation in the crosses of green, white and variegated leaves.

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	×	Variegated	

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Introduction

- ✓ Sex-determination system is a biological system that determines the development of sexual characteristics in an organism.
- ✓ In many cases, sex determination is genetic: males and females have different alleles or even different genes that specify their sexual morphology
- ✓ Camerarius (1694) gave first description of reproductive organs in plants (maize)

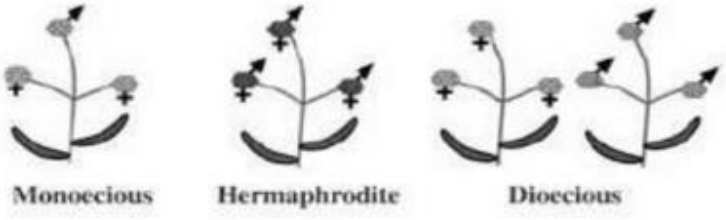
What is sex determination?

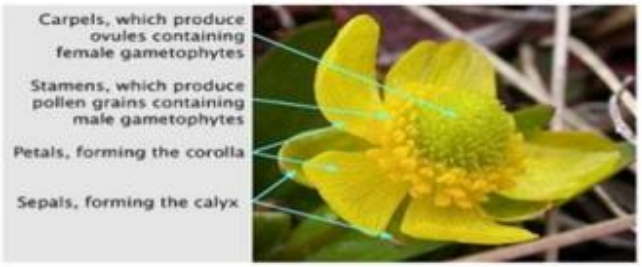
- The method by which distinction between male and female is established in an organism under genetic control.
- The sex chromosomes are responsible for determination of separate sexes.
- It is a biological system that determines the development of sexual characteristics in an organisms.

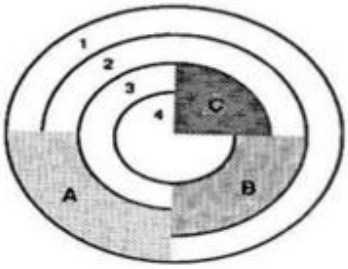
Traditionally, the symbol ♂ designates male

and the symbol ♀ designates female.

Sex determination in Angiosperms







SEX DETERMINATION

SEX DETERMINATION → The natural event by which an individual of a dioecious species become male or female

Two main mechanisms

- Enviromental Sex: In some species sex is determined after fertilization by environment factors
- Genetic sex determination: Sex is determined at fertilization by the combination of genes that the zygote recieves

Mechanisms

- ✓ Environmental
- ✓ Chromosomal
  1. homomorphic chromosomes
     - Heterogametic male (XX, XY)
     - Heterogametic female(XY,XX)
  2. heteromorphic chromosomes
     - XX, XY (active Y)
     - XX, XY_1Y_2 (X/autosome balance)
     - $X_1X_1X_2X_2$, $X_1X_2Y_1Y_2$
- ✓ Genic
  - Single locus
  - Multiple loci

6

Homomorphic chromosomes :Morphologically identical members of a homologous pair of chromosomes.
The homomorphic chromosome is the **identical chromosome** that is paired during the process of mitosis and meiosis. These chromosomes are of the same length and have the **same genetic composition, the genes are identical to each other**, but they **may have little variation** in them.
For Example: One gene may have the mutation.

Heteromorphic chromosomes :A chromosome pair with some homology but differing in size, shape, or staining properties. Homologous chromosome pair which are not morphologically identical (eg the sex chromosomes).
The heteromorphic chromosome is **the non-identical chromosome** that is paired during the process of meiosis or mitosis. These chromosomes have **different size, shape, and genetic composition**. These chromosomes **determine the sexual nature of the organism**.

HOMOMORPHIC CHROMOSOMES

Meaning :-morphologically identical members of a homologous pair of chromosomes

Homogametic (XX) Female , Heterogametic (XY)Male

Species	Features of Y chromosomes	Mechanism of sex Determination
Asparagus officinalis	Similar to X	Y active
spinach	Similar to X	Y active

Heterogametic (XY) Female, Homogametic (XX) Male

Fragaria elateria	------	----

Sex Determination in *Sphaerocarpos*

- Sex determination was first described in the bryophyte ***Sphaerocarpos sps*** which has heteromorphic chromosomes. The gametophyte is haploid and heteromorphic. The male gametophyte as well as the female gametophyte is an haploid organism.

with 8 chromosome (n=8). The diploid sporophyte is always heterogametic. Seven autosomes are similar in both male and female gametophyte. But the eighth chromosome of female is X which is larger than the seven autosomes. The eighth chromosome of male is Y which is comparatively smaller than autosomes. The sporophyte containing XY combination produces two types of meiospores, that is some with X and others with Y chromosomes. The meiospores with X chromosomes produce female gametophyte and those with Y chromosome produces male gametophyte.

Examples

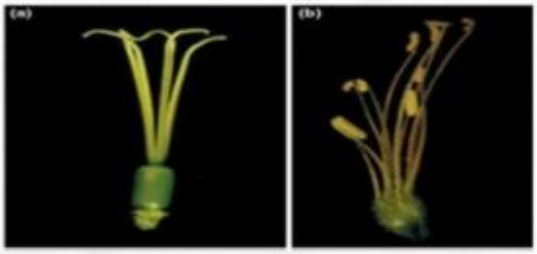
(ii) XX-XY types:

Silene latifolia **(Melandrium album)**

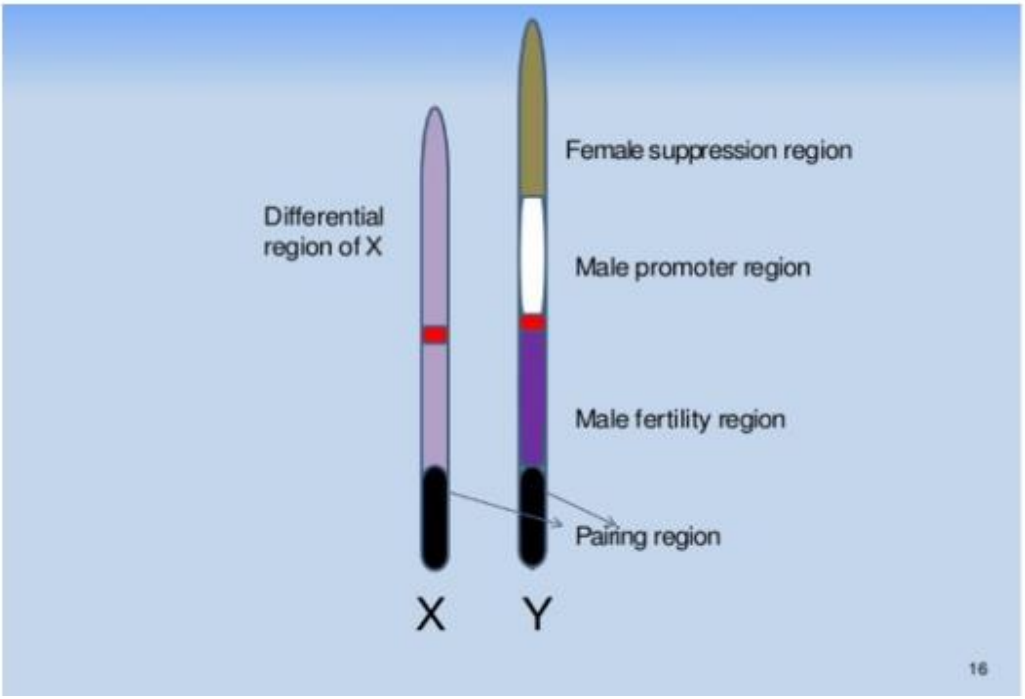
- **Most well studies dioeceous plant for sex determination** this species.
- sex determination is by sex chromosomes similar to that in many animals, males are XY and felames are XX. on mechanisms.

♀ = XX

♂ = XY



The dioecious model plant Silene latifolia. (a) Female and (b) male flowers; the calyx and corolla were removed to display pistils and stamens, respectively.



Sex determination in papaya

- Recently researchers in Hawaii discovered sex chromosomes in Papaya. Papaya (Carica papaya, 2n=36). Papaya has 17 pairs of autosomes and one pair of sex chromosomes. Male papaya plants have XY and female plants have XX chromosome. Unlike human sex, papaya sex chromosomes look like autosomes and it is evolved from autosome. The sex chromosomes are functionally distinct because the Y chromosome carries the genes for male organ development and X bears the female organ developmental genes

In papaya sex determination is controlled by three alleles. They are m, M_1 and M_2 of a single gene.

Genotype	Dominant/ recessive	Modification	Sex
mm	Homozygous recessive	Restrict maleness	Female
M_1m	Heterozygous	Induces maleness	Male
M_2m	Heterozygous	Induces both the sex	Bisexual (rare)
M_1M_1 or M_2M_2 or M_1M_2	Homozygous/ Heterozygous dominant	Inviable plants	Sterile

Table 3.6 : Sex determination in Papaya

Papaya

In papaya, a single gene with three alleles (m, M_1 and M_2) is suggested to control the sex differentiation.

♀ = mm produces female plants

♂ = M_1m produces male plants

⚥ = M_2m produces hermaphrodite plants

Genotypes M_1M_1, M_1M_2 and M_1M_2 are inviable, i.e. M_1 and M_2 alleles are recessive lethals.

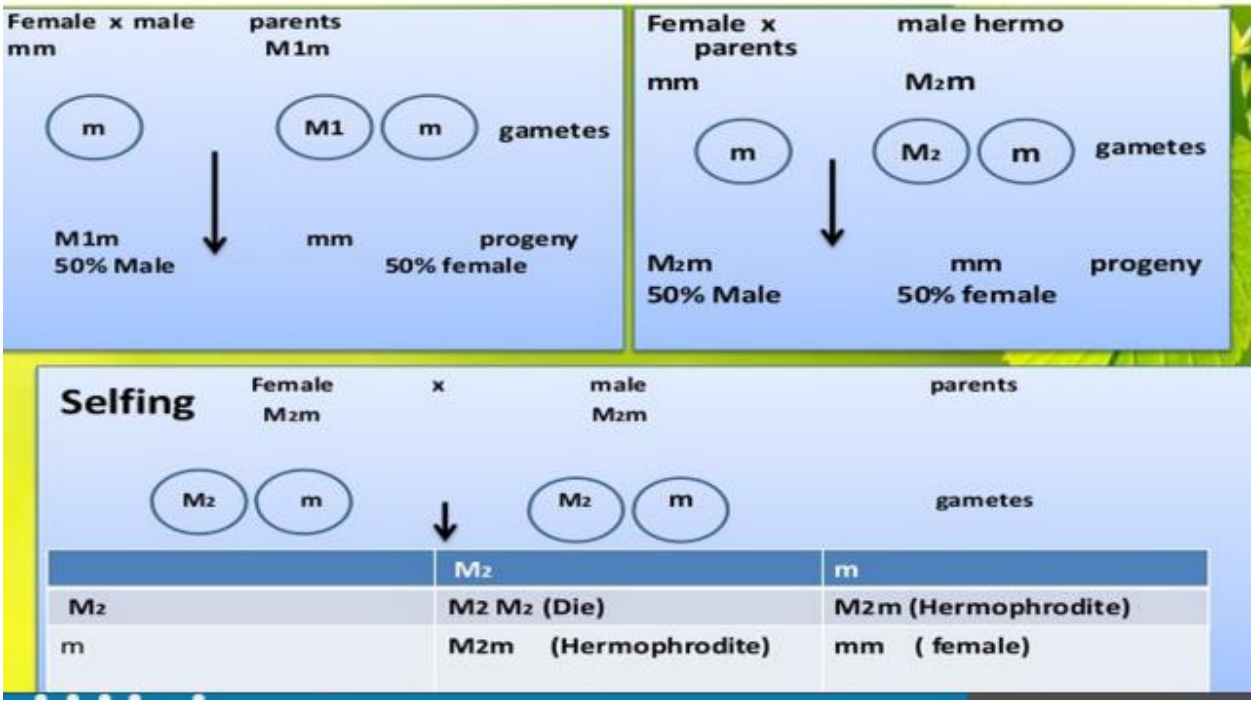
Papaya

- can produce male, female and hermaphrodites depending on the genotype of a single sex determining focus. This focus is thought to be clustered with several genes that play a role in sex determination.

mm

M_2m

M_1m



Sex determination in maize

Zea mays (maize) is an example for monoecious, which means male and female flowers are present on the same plant. There Tassel are two types of inflorescence.

The terminal inflorescence which bears staminate florets develops from Ear shoot apical meristem called tassel.

The lateral inflorencence which develop pistillate florets from axillary bud is called ear or bud.

Inflorescence of *Zea mays*

2 Sex determination in maize

- Maize being a monoecious plant bears both female (silk) and male (tassel) inflorescences on the same plant.
- A recessive gene **ba** (barren cob) in homozygous condition **(baba)** makes the **cobs barren** or non-functional.

  Similarly,
- a recessive gene **ts** in homozygous condition **(tsts)** converts the **male flowers** of tassel into **female flowers**.

Genotype	Dominant/ recessive	Modification	Sex
ba/ba ts/ts	Double recessive	Lacks silk on the stalk, but transformed tassel to pistil	Rudimentary female
ba/ba ts^+/ts^+	Recessive and dominant	Lacks silk and have tassel	Male
ba^+/ba^+ ts^+/ts^+	Double dominant	Have both tassel and cob	Monoecious
ba^+/ba^+ ts/ts	Dominant and recessive	Bears cob and lacks tassel	Normal female

Table 3.7: Sex determination in Maize
(Superscript (+) denotes dominant character)

Unisexuality in maize occurs through the selective abortion of stamens in ear florets and pistils in tassel florets. A substitution of two single gene pairs '**ba**' for barren plant and '**ts**' for tassel seed makes the difference between monoecious and dioecious (rare) maize plants. The allele for barren plant (ba) when homozygous makes the stalk staminate by eliminating silk and ears. The allele for tassel seed (ts) transforms tassel into a pistillate structure that produce no pollen. The table-3.7 is the resultant sex expression based on the combination of these alleles. Most of these mutations are shown to be defects in gibberellin biosynthesis. Gibberellins play an important role in the suppression of stamens in florets on the ears.

1.ENVIRONMENTAL

- Sex determination induced by the environment is one of the most direct ways in which progeny sex ratios can vary.
- Some plants Sex Determination is greately affected by the Environment. Eg:-*Equisetum*

♂ Plants If grown under "adverse" condition.

♀ Plants If grown under "optimum" condition.

Clip slide

- Others like **Cucumber, Melons ,Cannabis etc.** the sex of the flowers is affected by many environmental factors like **Temperature, Day-Length, Ethylene,GA_3, some ions(Ca^{+2} Mg^{+2}) etc.**
- In general treatment with **Ethylene or GA_3** promotes the production of **Female flowers.**
- In ***cannabis*** GA_3 treatment induces devlopment of **only female flowers** in otherwise male flower.

I. Population genetics: definitions

Population **–** group of interbreeding individuals of the same species that are occupying a given area at a given time.

Population genetics is the study of the allele frequency distribution and change under the influence of the 4 evolutionary forces: ***natural selection, mutation, migration (gene flow), and genetic drift***. Population genetics is concerned with gene and genotype frequencies, the factors that tend to keep them constant, and the factors that tend to change them in populations.

All the genes at all loci in every member of an interbreeding population form ***gene pool***. Each gene in the genetic pool is present in two (or more) forms **– *alleles.***

Individuals of a population have same number and kinds of genes (except sex genes) and they have different combinations of alleles *(phenotypic variation).*

The applications of Mendelian genetics, chromosomal abnormalities, and multifactorial inheritance to medical practice are quite evident. Physicians work mostly with patients and families. However, as important as they may be, genes affect populations, and in the long run their effects in populations have a far more important impact on medicine than the relatively few families each physician may serve.

Why is population genetics important for human health and diseases?

(a) Human evolution has been underlain by adaptive and non-adaptive changes in gene frequencies

(b) Diseases are due to effects of alleles interacting with environments - is a spectrum from single-locus disorders to polygenic disorders

Gene frequencies tend to remain constant from generation to generation when disturbing factors are not present. Factors that disturb the natural equilibrium of gene frequencies include mutation, migration (or gene flow), random genetic drift, and natural selection. A mutation is a spontaneous change in the gene frequency that takes place in a population and

occurs at a low rate. Migration is a local change in gene frequency when an individual moves from one population to another and then interbreeds. Random genetic drift is a change that takes place from one generation to another by a process of pure chance. Mutation, migration, and genetic drift alter gene frequencies without regard to whether such changes increase or decrease the likelihood of an organism surviving and reproducing in its environment. They are all random processes.

II. Hardy-Weinberg Law

In 1908 German physician *Wilhelm Weinberg* (1862-1937) and the British mathematician *Godfrey.H. Hardy* independently formulated the *Hardy-Weinberg principle*. They pointed out that under ideal conditions it could easily predict genotype frequencies from allele frequencies, at least for a diploid sexually reproducing species such as humans.

For many human autosomal recessive traits the heterozygote cannot be distinguished from the normal dominant homozygote. When this occurs the Hardy-Weinberg equilibrium is assumed to apply. These authors used different approaches but came to the same conclusions. They made *several assumptions of the population*:

1. The population follows the Laws of Mendelian genetics
2. Size of the population approaches infinity
3. There is no effect of recurrent mutation
4. There is no migration in or out of the population
5. There is no selection against any phenotype
6. Mating is totally random

Under these assumptions, Hardy and Weinberg found that the gene frequency and the genotype frequency in the population do not change from generation to generation.

Furthermore, if the frequency of the dominant allele **A** in the founding population was **p** , and the frequency of the recessive allele **a** in the founding population was **q**, then after one generation of random mating the genotype frequencies would remain fixed and would be in the ratio:

p^2 (AA) 2pq (Aa) q^2 (aa)

Since there are only two alleles in the population,

p(A)+q(a) = 1 *and* p^2(AA) + 2pq(Aa) + q^2(aa) = 1

Thereby, Hardy-Weinberg law is fundamental principle in population genetics stating that ***the genotype frequencies and gene frequencies of a large, randomly mating population remain constant provided migration, mutation, natural selection and genetic drift do not take place.***

Importance of "H-W" Formula

Example: If the incidence of PKU is 1 in 10,000 live births, and it is a recessive condition then:

q^2 = 1/10,000 = 0.0001

q = $\sqrt{}$ 0.0001 = 0.01

Where q is the gene frequency for PKU.

The frequency of heterozygote carriers is $2pq$ as:

$p + q = 1$

$p = 1 - 0.01 = 0.99$

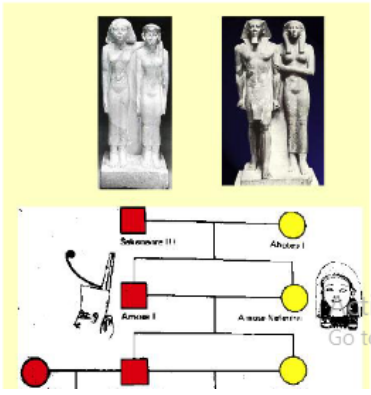
$2pq = 2\ (0.99 \times 0.01) = 0.0198$

III. Factors Affecting Gene Frequency in a Population:

1. Non-random mating.
2. Altered mutation rate.
3. Natural selection.
4. Small populations.
5. Migration.

1. *Non-random mating* – decreases the proportion of heterozygotes, violates assumption of Hardy-Weinberg equilibrium.

1. ***Assortative mating*** – non-random mating based on phenotypic similarities (in humans pair bonds established by religion, ethnic background, professional interest, etc.) **Assortative mating** (also called **assortative pairing**) takes place when sexually reproducing organisms tend to mate with individuals that are like themselves in some respect (*positive assortative mating*) or dissimilar (*negative assortative mating*). In evolution, these two types of assortative mating have the effect, respectively, of reducing and increasing the range of variation, or trait variance, when the assorting is cued on heritable traits. Positive assortative mating, therefore, results in disruptive natural selection, and negative assortative mating results in stabilizing natural selection.
2. ***Inbreeding*** – breeding between close relatives. If practiced repeatedly, it leads to an increase in homozygosity of a population. A higher frequency of recessive, deleterious traits in homozygous form in a population can, over time, result in inbreeding depression. This may occur when inbred individuals exhibit reduced health and fitness and lower levels of fertility.



Coefficient of Inbreeding is the probability that an individual being homozygous for a certain locus by receiving both alleles of this locus from ancestral source. Offspring of first-cousin marriage is homozygous at 1/16 of his loci.

Genetic consequences of inbreeding

1. Reduction in genetic variability
   a. Increase in frequency of homozygous recessives
   b. Recessive alleles exposed to natural selection
2. Social consequences
   a. Concentration of wealth and power
   b. Reduction of social ties with other groups

2. *Altered mutation rate*

Mutation (heritable changes in DNA) give rise to new alleles. Mutation rate is low: for a single locus the average frequency of mutation is about 0.0001. They may be lethal, neutral, or advantageous. Mutations are ultimate source of genetic variation. If mutation is advantageous the natural selection favours it and allele frequency changes.

Eradication of dominant disorders.

Huntington's disease (and any other dominant disorder) could in principle be eliminated in one generation by aborting every foetus carrying the gene. However, this would not prevent spontaneous mutations occurring (in Huntington's, ±1 in 100,000) unless the entire population was screened for them.

3. *Natural Selection*

Natural selection is process by which a particular genotype/phenotype combination confers an advantage to individuals in a population, thereby determining its survival and reproduction. Natural selection is often geographically-restricted.

Genetic selection acts on the individual **phenotypes** and either favours or hinders reproduction and thus the propagation of that individual's **genotype**. Natural selection acts by modifying an individual's *biological fitness*.

For an *autosomal dominant trait*, any increase in fitness, will rapidly alter the gene frequency over the next few generations to a new equilibrium.

Selection against a *recessive* genotype is less effective and results in a slow change in gene frequencies.

For *X-Linked recessive trait*, the situation is intermediate between autosomal dominant and recessive.

4. *Small Populations*

For religious, geographical (spatial, e.g., due to islands, mountains, and glaciers that isolate local populations), tribal or other reasons a small group of individuals may become genetically isolated from the rest of the population **(genetic isolates).**

By chance one allele may fail to be passed on to the next generation and so disappear **(extinction)** leaving only the alternative allele at that locus **(fixation).**

Gene frequencies in small isolate populations do not reflect those of the larger founding population from which they were derived because of two factors, ***founder effect*** and ***random genetic drift***.

Founder effect refers to the loss of genetic variation when a new colony is established by a very small number of individuals from a larger population, i.e. the population grew from a few founding individuals.

5. Migration

Until recently most people stayed in the place they were born and married someone nearby. The last half of 20th century was characterizes by breakups of local population isolates. This major change in breeding patterns has resulted in increased in genetic heterogeneity.

***Migration** is the movement of individuals **in** or **out** of a population.*

Migration is necessary to keep a species from fragmenting into several different species. Even as low a level as one individual per generation moving between populations is enough to keep a species unified. Migration can be thought of as combining two populations with different allele frequencies and different numbers together into a single population. After one generation of random mating, the combined population will once again be in H-W equilibrium.

Gene flow – movement of alleles into and out of populations by migration of individuals. It balances genetic differences that arise through mutation. Genetic flow also helps keep separated populations genetically similar (ie. the same species.

Genetic drift is random (by chance) change of allele frequencies from generation to generation. It leads to homozygous condition – loss of genetic diversity. The effect of genetic drift is greater in small than large populations. Migrant individuals will modify the gene pool of their descendents.

As population size increases the effects of genetic drift decrease. For example, the chance of completely loosing an allele (fixation) by chance is great in a population of 25 individuals. In a population of 500 individuals, the probability that one of the alleles will be lost by chance is low and, in the absence of other influences (natural selection etc.) they tend to be maintained in the population at constant relative frequency.