Lecture 11. Lethal genes; Pleiotropy; Phenocopy; Multiple alleles; Self incompatibility in plants; Pseudo alleles; iso-alleles; Penetrance and Expressivity.

Lethal Genes

NOT 3 : 1 it is 2 : 1

A lethal gene causes the death of all the individuals carrying this gene in the appropriate genotype before these individuals reach adulthood

Some genes are lethal when both alleles are present.

Lethality can occur before or after birth

An example is the "creeper" allele in chickens, which causes the legs to be short and stunted.

Creeper is a dominant gene, heteroyzous chickens display the creeper phenotype

If two creeper chickens are crossed, one would expect to have (from Mendelian genetics) 3/4 of the offspring to be creeper and 1/4 to be normal

Instead the ratio obtained is 2/3 creeper and 1/3 normal.

This occurs because homozygous creeper chickens die.

Lethal Genes

NOT 3 : 1 it is 2 : 1

Creeper x **Creeper**

Pp **Pp**

We Expect

3 : 1 of Creeper : Normal

	P	p
P	PP lethal	Pp creeper
p	Pp creeper	pp normal

BUT actually

2: 1

Pleiotropism

Single gene controlling more than one character is called as **Pleiotropy**

Manifold effect of a gene

A gene having more than one phenotypic effect

Gene for awn in variety also enhances yield and seed weight

Pleiotropy- A gene can sometimes affect another characteristic. This ability of having multiple effects of gene is called pleiotropy.

Eg. i. Sickle-cell anemia
ii. Genes that control fur pigmentation in cats may have an influence on the cats eyes and brain.

Sickle-cell anemia - Is most frequent in African Americans. The location of gene is known. It causes an abnormal hemoglobin formation from single incorrect amino acid. The abnormal hemoglobin leads to poor circulation and can lead to strokes and heart attacks certainly pain. Pre-natal testing is available, but there is no cure

Phenocopy

- The phenotype becomes altered by the **environment** in such a way that the new phenotype resembles another phenotype produced by known genes. The induced phenotype is not inherited, this is called as phenocopy.

  Eg:-Generally the body colour of the fruit fly Drosophila melanogaster is light brown.

- When the larvae of the normal brown bodied fruit flies were reared on food with silver salts, the emerging adults had yellow body

Pseudoalleles

- Pseudoalleles are non alleles so closely linked as often inherited as one gene, but shown to be separate by cross over studies.
- One of the first demonstration of pseudo-allelic condition was that of star-asteroid analysis by Lewis in 1951.
- He found a recessive mutation in Drosophila producing a small rough eye when homozygous. It was at locus 1.3 in the second chromosomes.
- This was also the identical location of gene star, a dominant mutation also affecting the morphology of the eye.

- The eye was rough and had a slight gleam, hence the name star, with gene symbol S.
- In crosses among these flies, recombination between star and asteriod occurred at a low frequency of one in five thousand.
- Pseudoallelic effects are found in Drosophila, corn, cotton, Aspergillus, Neurospora, bacteria and in viruses.

Isoalleles

- Usually wild type allele (represented as +) is dominant over its recessive allele.
- In **some natural populations, different wild type alleles affecting the same character were found and these wild type alleles had similar allelic dominance or they may differ in their degree of expression that could be detected in special combinations. Such alleles are called isoalleles.**

Examples for Isoalleles

- Timofeev - Ressovsky and Muller found that the wild type Drosophila from different natural populations had different dominant (red eye) alleles as judged by their stability or by their different effects in combinations.

- Stern found three different wild type alleles of another Drosophila mutant, cubitus interruptus, which showed different degrees of dominance over the same mutant allele.

- He called such alleles as isoalleles because they were alike in their homozygous effect and their differences appeared only in special combinations.

Penetrance

The ability of a gene to express uniformly in all individuals

The ability of a gene or gene combination to be expressed phenotypically to any degree is called penetrance.

The frequency with which a gene produces a phenotypic or visible effects in the individual that carry it. OR

The proportion of individuals which exhibit phenotypic effects of a specific gene carried by them.

Penetrance is calculated in percentage

Complete Penetrance:

When all the individuals which carry a particular gene exhibit its phenotypic effect.

Homozygous dom. (carriers) – one phenotypic effect; hom. recessive show another phenotypic effect; recessive genes have no penetrance in heterozygous condition

Most dominant and recessive genes in homozygous condition and many completely dominant gene even in heterozygous condition give their phenotypic expression.

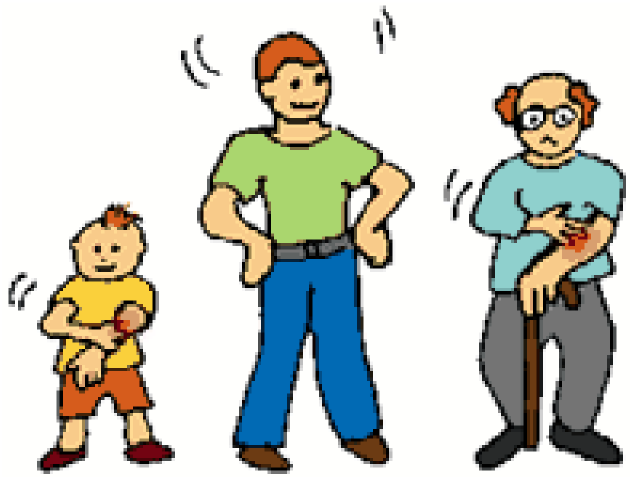
Heterozygous condition also produce the normal phenotype.

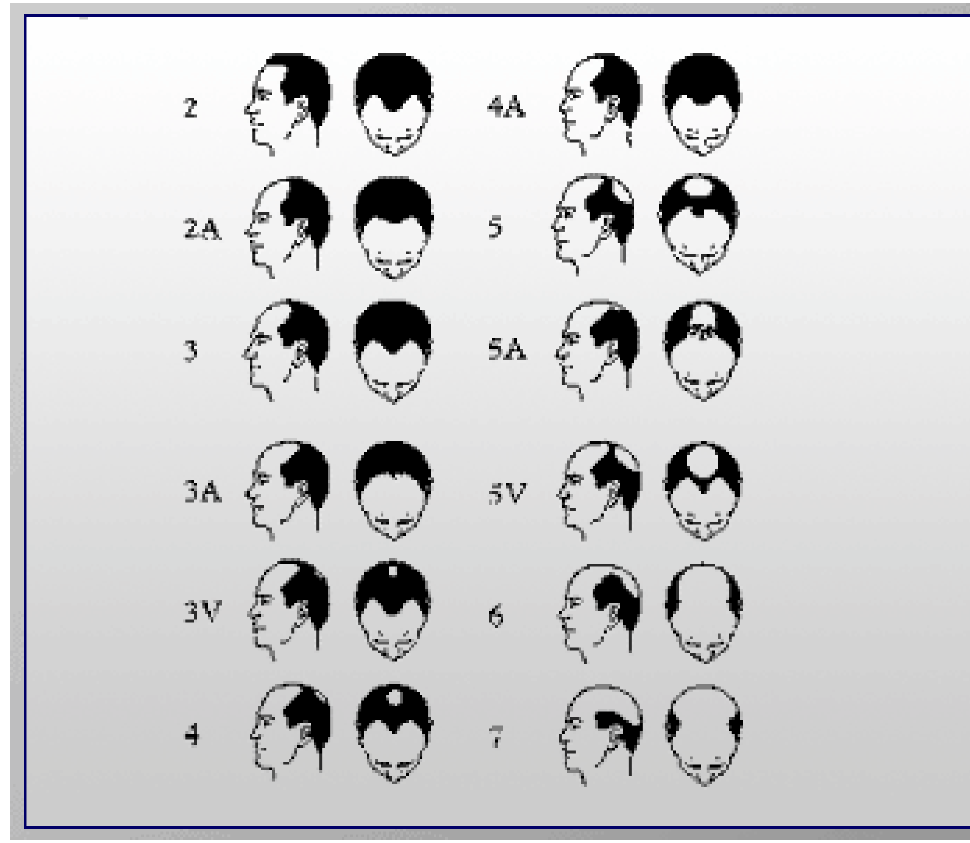
Incomplete penetrance

Heterozygous condition unable to express fully the normal phenotype.

Eg: In the case of polydactyly in man where one extra finger is present in the palm or foot, the heterozygous condition (**Pp**) brings-forth polydactyly in some and normal condition in others.

Some heterozygotes individuals were not polydactyles some had an incomplete penetrance





Penetrance can be age-dependent. The older a person is, the more likely he or she is to develop the condition if carrying a susceptible genotype. Male pattern baldness is an example of age-dependent penetrance. Men generally don't show signs of this X-linked recessive condition until later in life.

Expressivity

The degree of phenotypic expression of a gene in the different individuals is known as expressivity

The degree of effect produced by the Penetrant genotype

Degree of expression of a penetrant gene in its carriers

Change of temperature, nutrition, hormone deficiency etc., influence the expressivity of the curly wing in *Drosophila*.

Eg:- In man polydactyl condition may be penetrance in left hand (6 fingers) not in right hand (5 fingers) or may be penetrance with the feet but not in the hand.

Types of expressivity

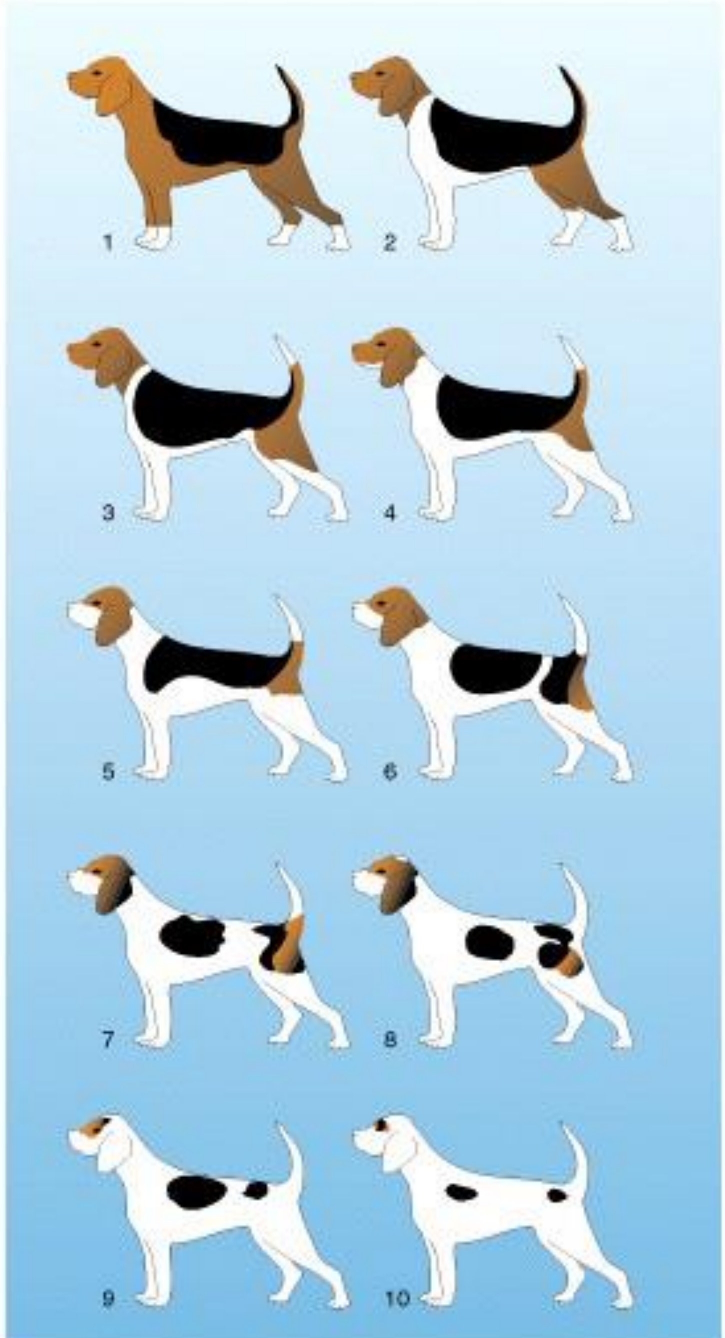
- **Uniform :** when the phenotypic expression of a gene is identical or similar in all the individuals which carry such genes
  Eg. Qualitative characters exhibit uniform expressivity
- **Variable :** when the phenotypic expression differs in different carriers of a gene
  Eg. Lima bean –dom. Gene for chlorophyll deficiency on leaf tips, leaf margins, or entire cotyledonary leaf

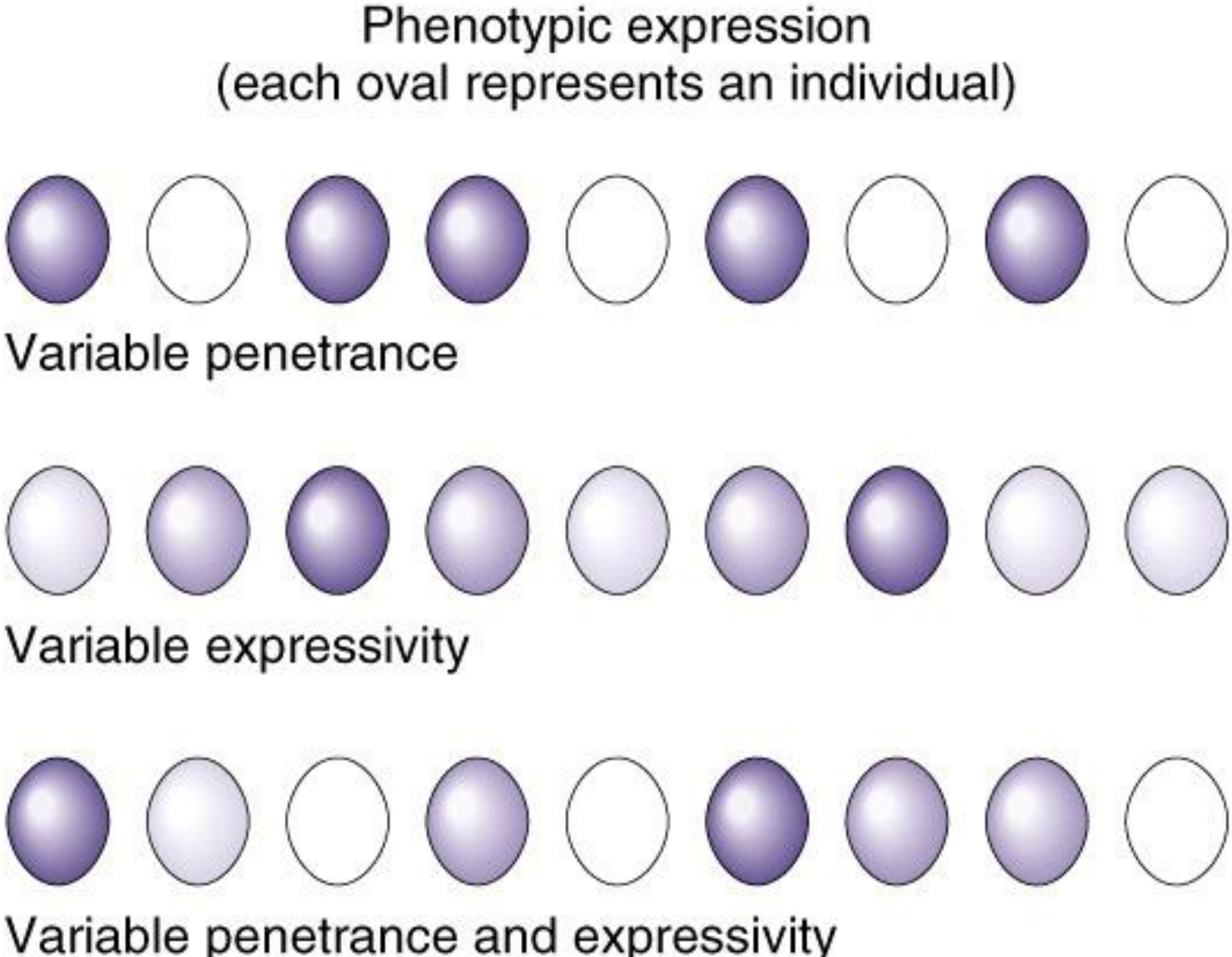
Variable Expressivity

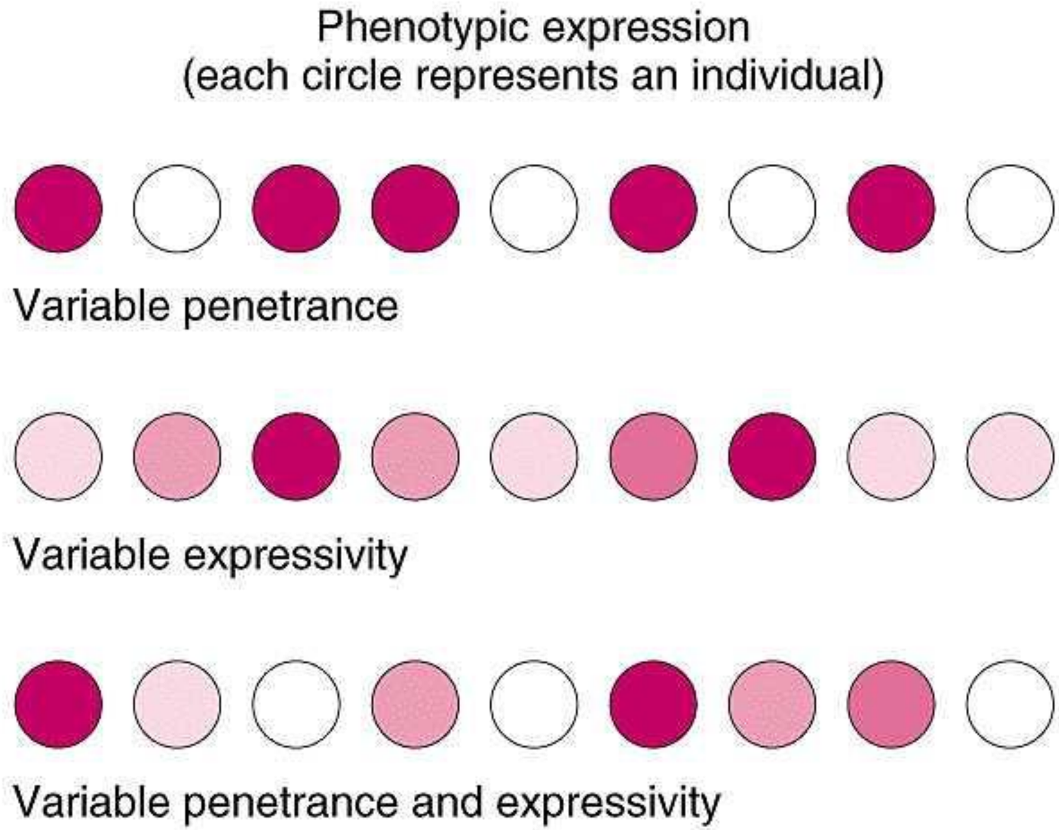
Variable expressivity refers to the degree to which a condition or disorder is expressed in an individual. Expressivity differs from penetrance.

Think of penetrance as a light switch that can only be on or off, and expressivity as a dimmer on that light switch.

Though some disorders are expressed with little variation, it is worthwhile to note that the majority of autosomal dominant conditions do appear to exhibit some variation in expressivity.


1
2
3
4
5
6
7
8
9
10


Phenotypic expression
(each oval represents an individual)
Variable penetrance
Variable expressivity
Variable penetrance and expressivity



Penetrance vs expressivity
Penetrance: What fraction of individuals with a genotype show the expected phenotype
Expressivity: What is the extent of expression
Example: a human disease

If only 75% of AA individuals have disease, penetrance = 75%
Variation in severity of disease among AA individuals showing disease = expressivity

Multiple alleles – characteristic features, study of blood groups, coat colour in rabbits and self incompatibility in plants

Allele is the short term for allelomorph

allelon **- of one another**

morphus **- form**

Allelomorph - Bateson

- **Allele is the alternative form of a gene**
- **Alleles are of two types**
  1. **dominant or recessive**
  2. **wild or mutant type**

Multiple alleles

- **More than two alleles at the same locus gives rise to a multiple allelic series.**
- **The existence of more than two alleles at the same locus of a homologous chromosome is referred to as multiple alleles**
- **Presence of multiple alleles adds variability for a character**

The number of possible genotypes in a series of multiple alleles can be calculated by using the formula

$$\tfrac{1}{2}\{n \times 1\ (n + 1)\}$$

where, n is the number of identified alleles in that group.

Main features of multiple alleles

- **Belong to the same locus**
- **Always control the same character of an individual. However, the expression of the character will differ depending on the allele present.**
- **There is <u>no crossing over in a Multiple allelic series</u>. When two alleles are involved in a cross, the same two alleles are recovered in the F2 or test cross progeny. This is based on the classical concept of the gene, according to which crossing over takes place between gene and not within a gene.**

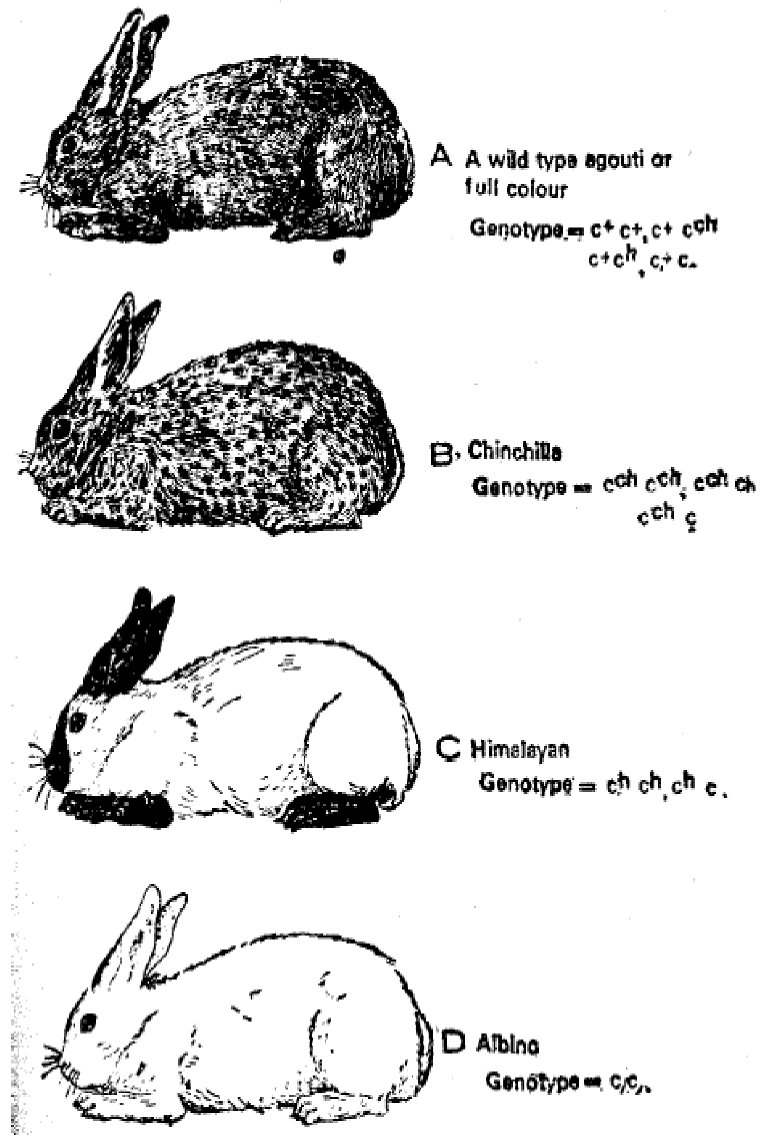
- In a series of Multiple alleles, wild type is always dominant. Rest of the alleles in the series may exhibit dominance or intermediate phenotypic expression when two alleles are involved in a cross.
- The cross between two mutant alleles will always produce mutant phenotype (intermediate). Such cross will never produce wild phenotype.
- In other words, Multiple alleles do not show complementation (Complementation refers to appearance of wild phenotype when two mutants are crossed)

Examples of Multiple Alleles

- **Fur color in Rabbits**
- **Wing type in Drosophila**
- **Eye colour in Drosophila**
- **Self incompatability alleles in Plants**
- **ABO Blood group in man**

Fur colour in Rabbits

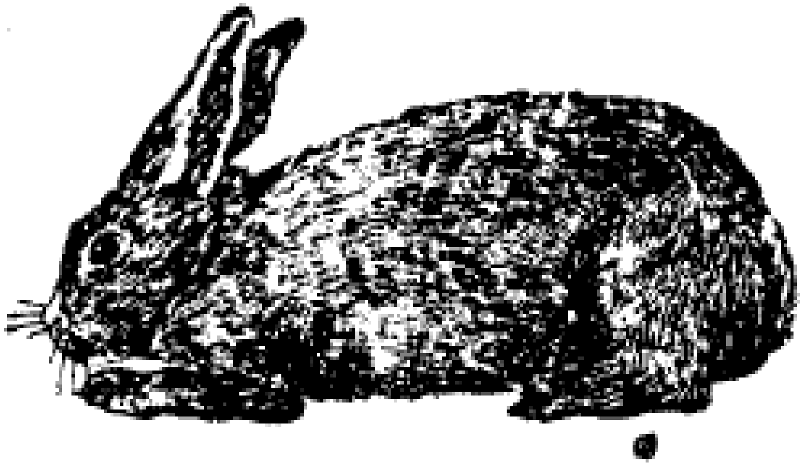
- **Classical example for multiple alleles**
- The fur colour is of four types
  - **Agouti**
  - **Chinchilla**
  - **Himalayan**
  - **Albino**



Different coat colours in rabbits (after Burns, 1969).

Agouti

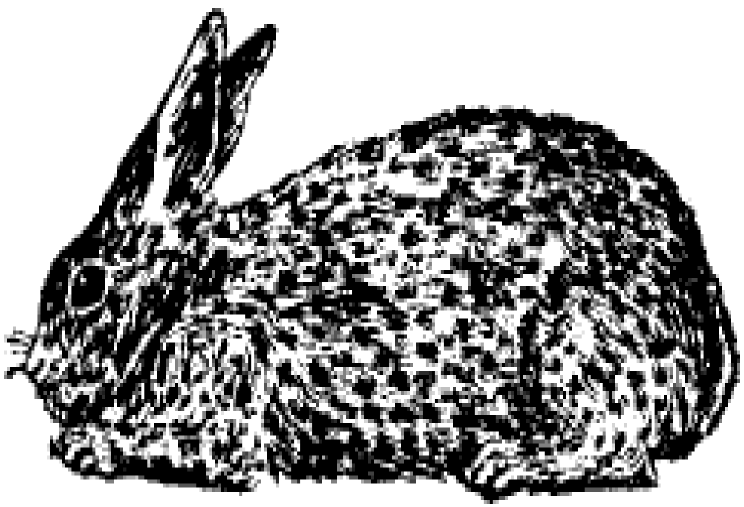
- This has full colour, known as wild type
- This colour is dominant over all the remaining colours
- Produces agouti colour in F_1 and 3:1 in F_2 when crossed with any of the other three colours in rabbits.
- This colour is represented by C



Agouti

Chinchilla

- This is lighter than Agouti.
- Silver grey due to mixture of black and white hairs
- This colour is dominant over Himalayan and albino
- Produces chinchilla in F_1 and 3:1 ratio in F_2 when crossed with either Himalayan or albino.
- This colour is represented by c^{ch}



Chinchilla c^{ch}

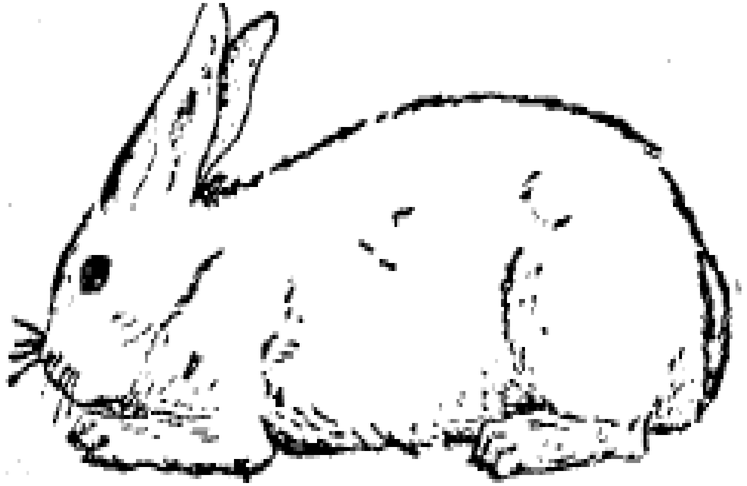
Himalayan

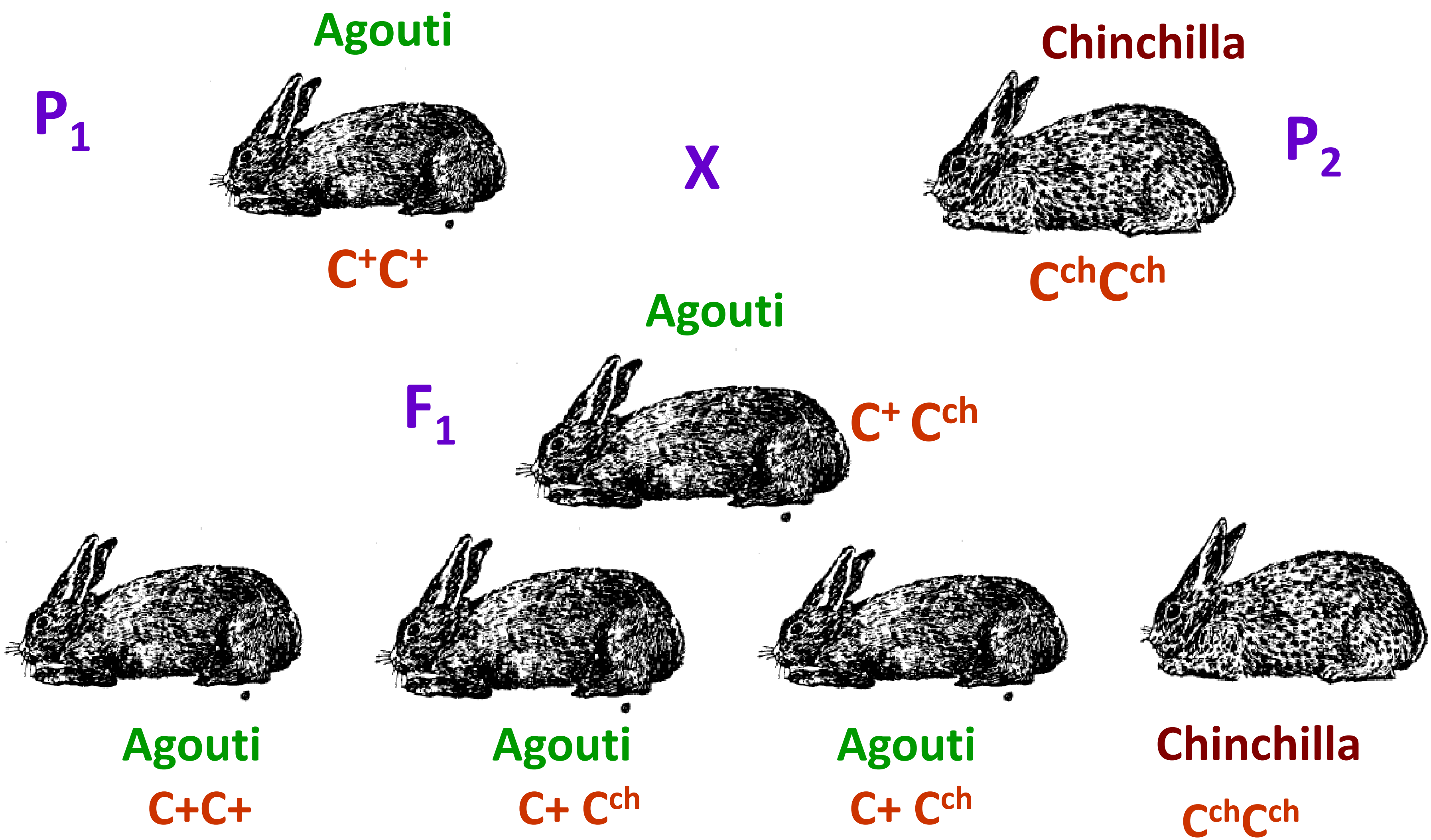
- The main body is white while the tips of ear, feet and tail and snout are coloured
- This colour is dominant over albino
- Produces 3:1 ratio in F_2 when crossed with albino.
- This is represented by ch

Himalayan

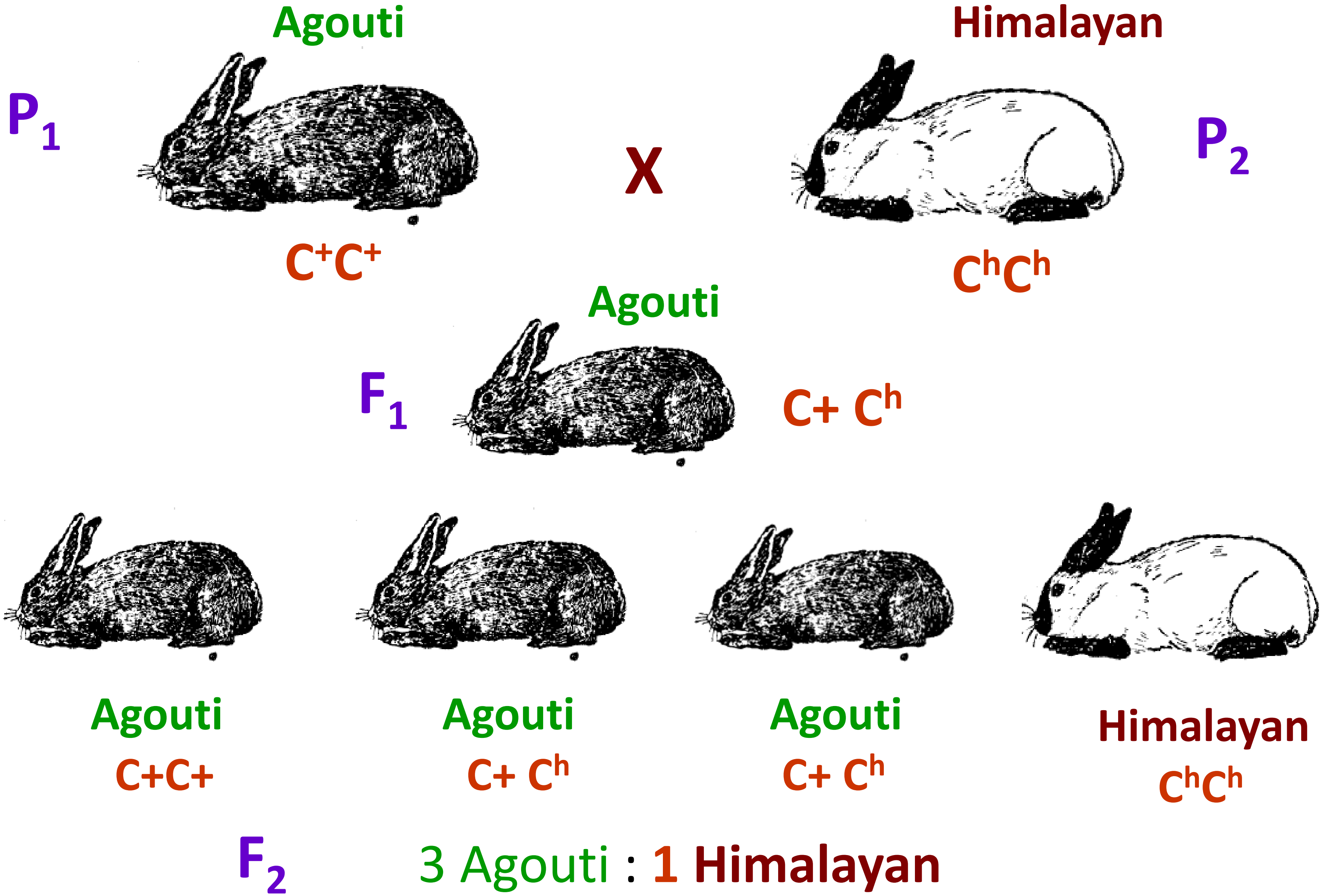
Albino

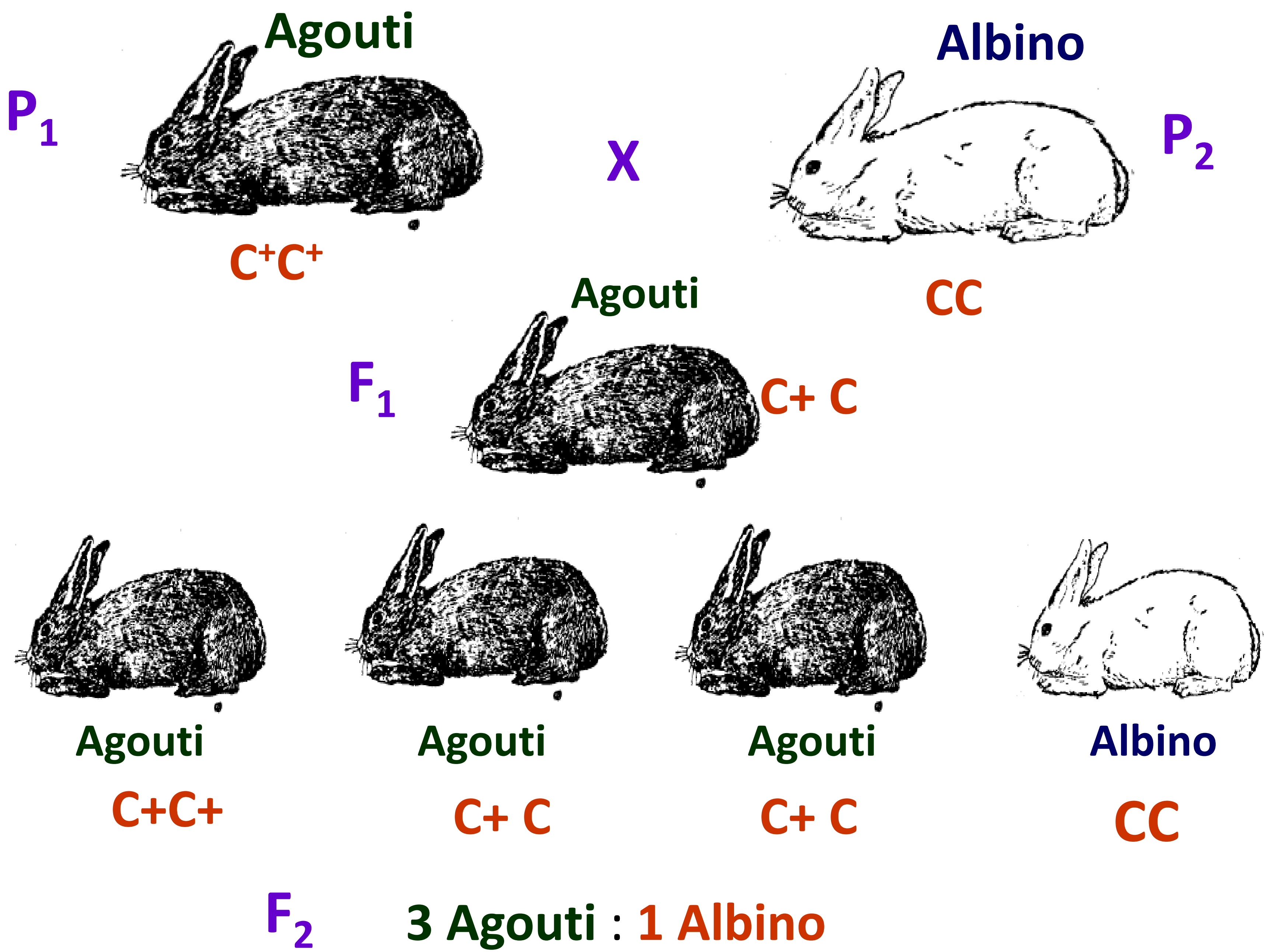
- This has pure white fur colour and is **recessive** to all other types.
- This is represented by **c**





F_2 3 Agouti : 1 Chinchilla


Agouti
Himalayan
P_1
X
P_2
C^+C^+
C^hC^h
Agouti
F_1
C+ C^h
Agouti
C+C+
Agouti
C+ C^h
Agouti
C+ C^h
Himalayan
C^hC^h
F_2
3 Agouti : 1 Himalayan


P_1
Agouti
C^+C^+
X
Albino
P_2
CC
F_1
Agouti
C+ C
Agouti
C+C+
Agouti
C+ C
Agouti
C+ C
Albino
CC
F_2
3 Agouti : 1 Albino

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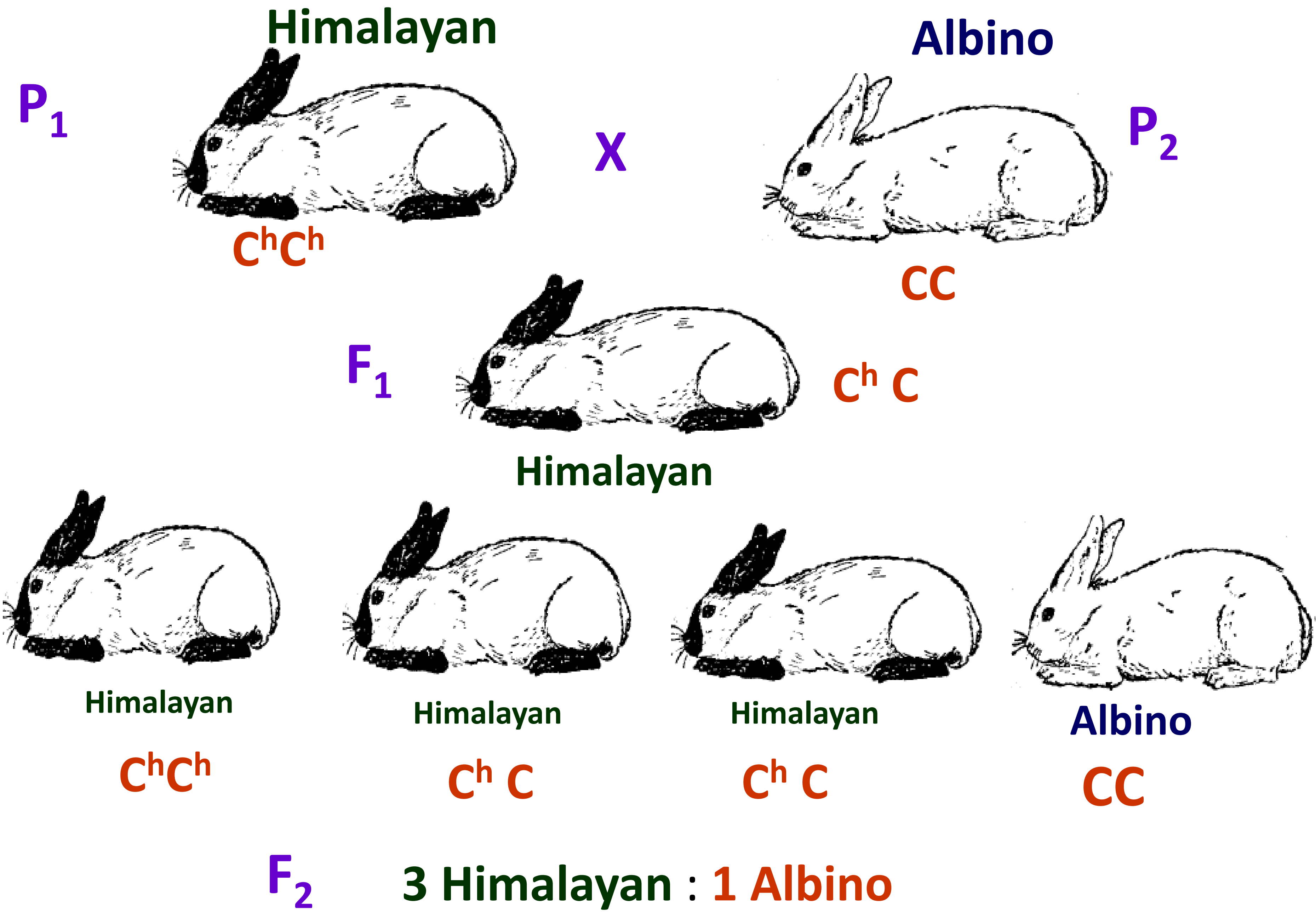
Himalayan
Albino
P_1
X
P_2
C^hC^h
CC
F_1
$C^h\,C$
Himalayan
Himalayan
Himalayan
Himalayan
Albino
C^hC^h
$C^h\,C$
$C^h\,C$
CC
F_2
3 Himalayan : 1 Albino

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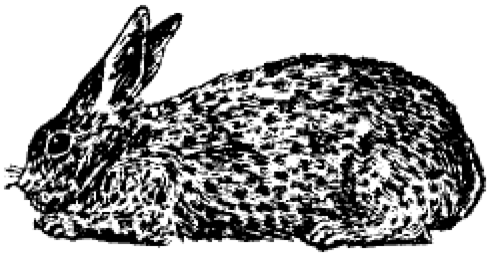
Chinchilla

Himalayan

P_1



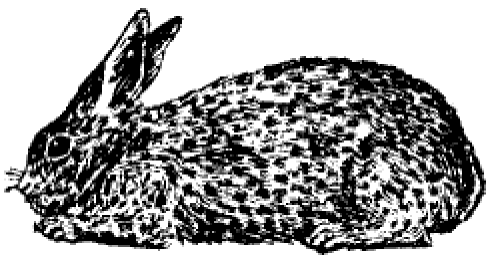
X

P_2

F_1

Light grey

Chinchilla | Light grey | Light grey | Himalayan



F_2

1 Chinchilla : 2 Light Grey : 1 Himalayan

Incomplete dominance of Chinchilla on Himalayan

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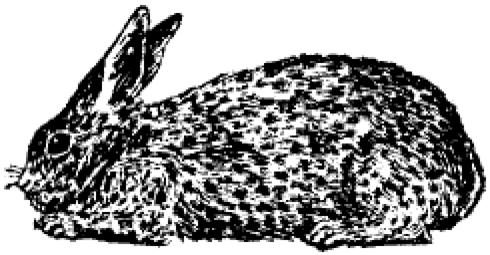
Chinchilla

Albino

P_1



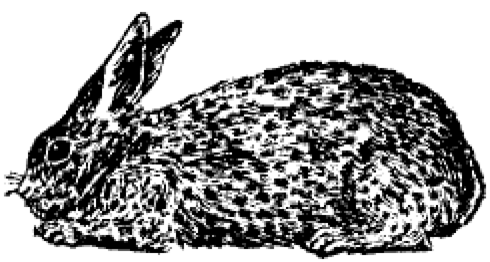
X

P_2

F_1

Light grey

Chinchilla	Light grey	Light grey	Albino



F_2

1 Chinchilla : 2 Light Grey : 1 Albino

Incomplete dominance of Chinchilla on Albino

Phenotypes and genotypes of multiple allelic series

PHENOTYPES	GENOTYPES
Agouti	C^+C^+, C^+C^{ch}, C^+C^h, C^+C
Chinchilla	$C^{ch}C^{ch}$,
Light grey	$C^{ch}C^{h}$, $C^{ch}C$
Himalayan	C^hC^h, C^hC
Albino	CC

The alleles of this multiple allelic series have following dominance hierarchy

$$c^{+} > c^{ch} > c^{h} > c$$

Blood groups in human beings

- One of the most firmly established series of multiple alleles in human beings involving the genetic locus controlling blood types A, B, AB and O
- Antigen is a specific protein found on the surface of the RBCs
- Antibody is another kind of specific protein found in the plasma
- There are two kinds of antigen A&B
- Two kinds of antibodies A&B
- Agglutination which is antigen- antibody reaction is highly specific one.
- **‘A** antigen’ can react with ‘**A** antibody’ alone. Likewise ‘B antigen’ react with ‘B antibody’ only

Blood groups in human beings

Antigen	Antibody	Blood group
A antigen	B antibody	A group
B antigen	A antibody	B group
A and B antigen	No antibody	AB group
No antigen	A and B antibodies	O group

Inheritance of Blood group in human beings

- The blood group is determined by a series of three alleles I^A I^A, I^B and i

Blood group	Possible genotypes
A	I^AI^A, $I^A i$
B	$I^B I^B$, $I^B i$
AB	$I^A I^B$
O	i i

The I^A controls production of 'A antigen'

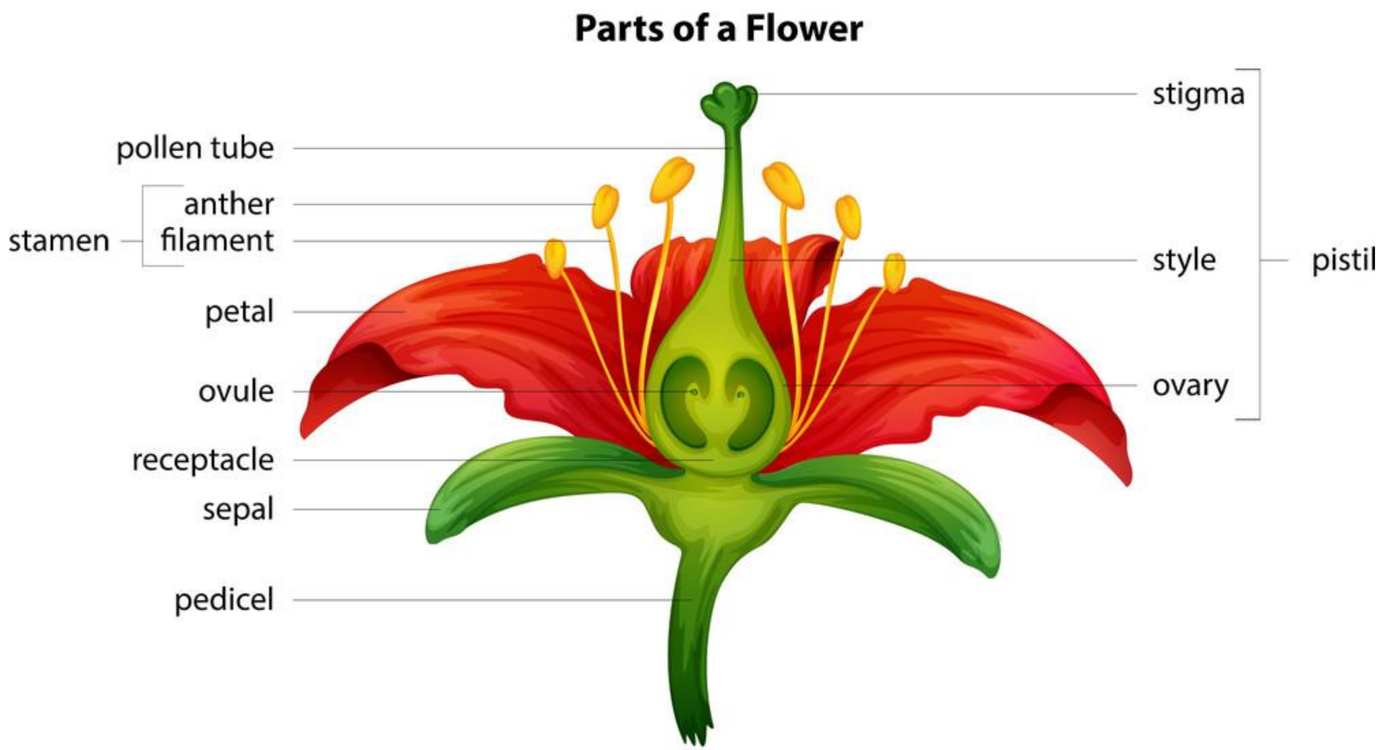
Allele I^B controls the production of 'B antigen'

Both I^A and I^B dominant over recessive allele i.

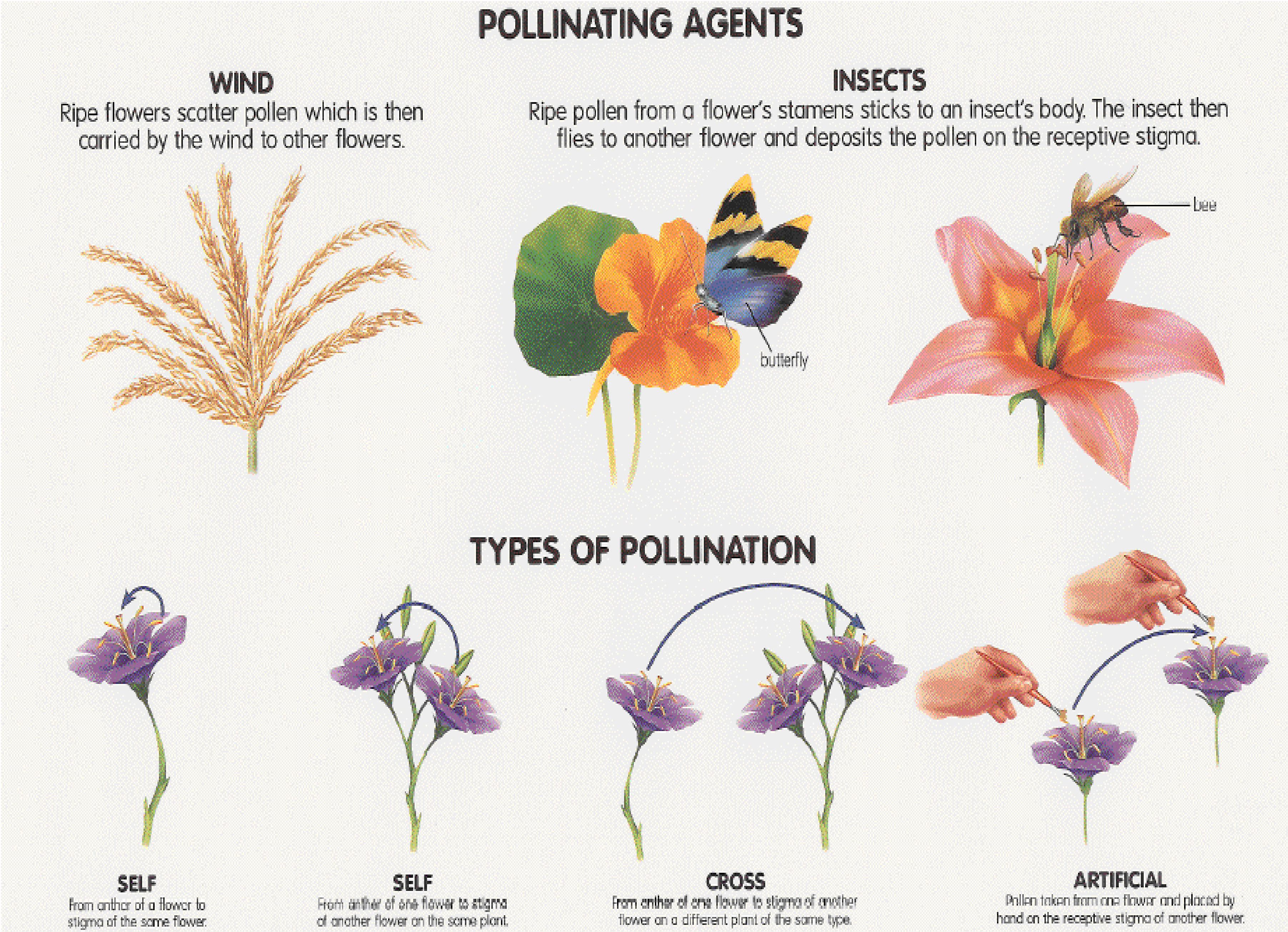
The heterozygote I^A and I^B is not intermediate, but both alleles expressed
equally (Co dominance)

Other Examples for multiple alleles

- Self incompatibly alleles in plants are controlled by multiple allelic series eg. Nicotiana, Brassica
- Eye color in Drosophila ranging from red (w+ or W), Coral (W^{co}), blood (W^{bl}), Eiosin (w^{e}), cherry (w^{c}), apricot(w^{a}), honey(w^{h}), tinged (w^{t}), pearl(w^{p}), ivory (w^{i}) and white(w)
- Red eye color being completely dominant
- ABO Blood group in human beings controlled by three alleles I^{A}, I^{B} and i form of multiple allelic series.
- Feather color in fowls is governed by a sex linked multiple allelic series B^{a} (ash red),(Blue- wild type) and b (chocolate).



PARTS OF A FLOWER


POLLINATING AGENTS
WIND
Ripe flowers scatter pollen which is then carried by the wind to other flowers.
INSECTS
Ripe pollen from a flower's stamens sticks to an insect's body. The insect then flies to another flower and deposits the pollen on the receptive stigma.
bee
butterfly
TYPES OF POLLINATION
SELF
From anther of a flower to stigma of the same flower.
SELF
From anther of one flower to stigma of another flower on the same plant.
CROSS
From anther of one flower to stigma of another flower on a different plant of the same type.
ARTIFICIAL
Pollen taken from one flower and placed by hand on the receptive stigma of another flower.

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Self-incompatibility (SI)

It refers to the inability of a plant with functional pollen to set seeds when self pollinated. It is the failure of pollen from a flower to fertilize the same flower or other flowers of the same plant.

SELF INCOMPATABILITY in plants

- The term Self Incompatibility was coined by Stout in 1917
- Koelreuter reported in *Verbascum phoeniceum* plants
- Self incompatibility enhance cross pollination.
- Stages **of SI**
- a) failure to germinate at stigmatic surface.
- b) failure to penetrate stigma
- C) slow growth of Pollen tube
- D) embryo degenerates.

Pollination

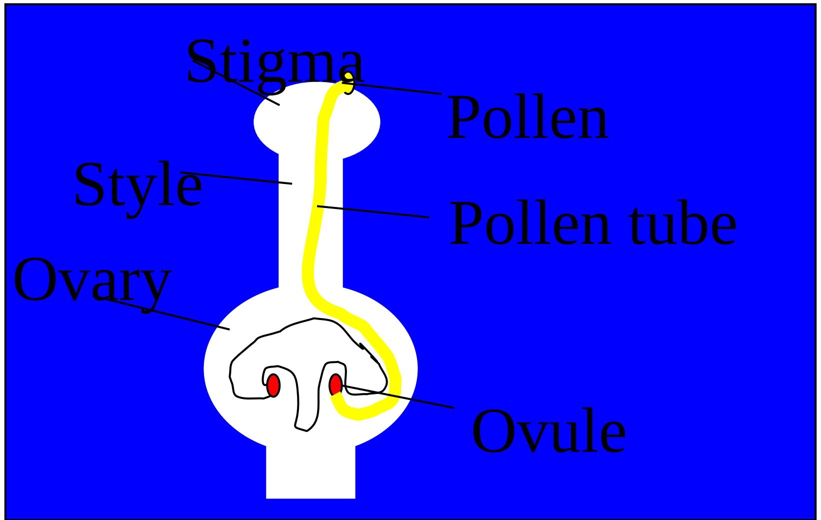


TABLE 9.1

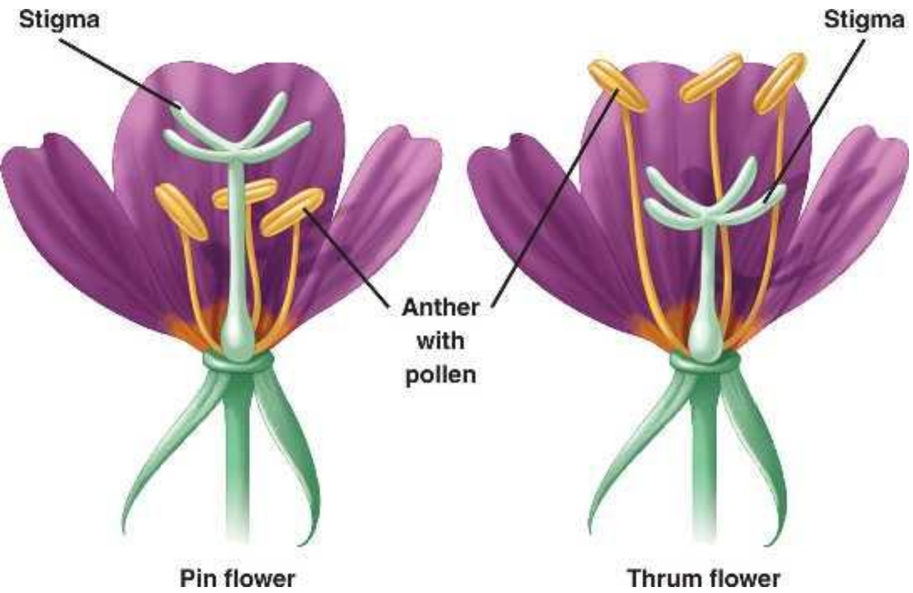
Classification of self incompatibility

Basis of Classification	*Types of self incompatibility*	*Brief Description*
1. Flower Morphology	(*a*) Heteromorphic	SI is associated with differences in flower morphology.
	(*i*) Distyly	Styles and stamens are of two types, *i.e.* short and long.
	(*ii*) Tristyly	Styles and stamens have three positions, *i.e.* short, medium and long.
	(*b*) Homomorphic	The flowers do not differ in Morphology.
	(*i*) Spororphytic	SI is governed by genotype of pollen producing plant.
	(*ii*) Gametophytic	SI is governed by the genetic constitution of gametes.
2. Genes Involved	(*a*) Monoallelic	SI is controlled by single gene.
	(*b*) Diallelic	SI is governed by two genes.
	(*c*) Polyallelic	SI is governed by several genes.
3. Site of Expression	(*a*) Stigmatic	SI genes express on the stigma
	(*b*) Stylar	SI genes express in the style
	(*c*) Ovarian	SI genes express in the ovary.
4. Pollen Cytology	(*a*) Binucleate	The pollen grains have two nuclei.
	(*b*) Trinucleate	The pollen grains have three nuclei.

SI = Self incompatibility.

Thrum: GSL_1L_2PA/gsl_1l_2pa

Pin: gsl_1l_2pa / gsl_1l_2pa



Incompatibility reaction of style	L_1/l_1
Incompatibility reaction of pollen	L_2/l_2
Length of style	G(Short)/g(long)
Size of stigmatic cells	S(Small)/s(Large)
Height of anthers	A(High)/a (Low)
Size of pollens	P(Large)/p(small)

TABLE 9.2

Results of four types of crosses in Primula

Crosses between	*Genotypes*	*Results*
Pin × pin	ss × ss	Incompatible
Thrum × Thrum	Ss × Ss	Incompatible
Thrum × pin	Ss × ss	1 Thrum : 1 Pin
Pin × Thrum	ss × Ss	1 Thrum : 1 Pin

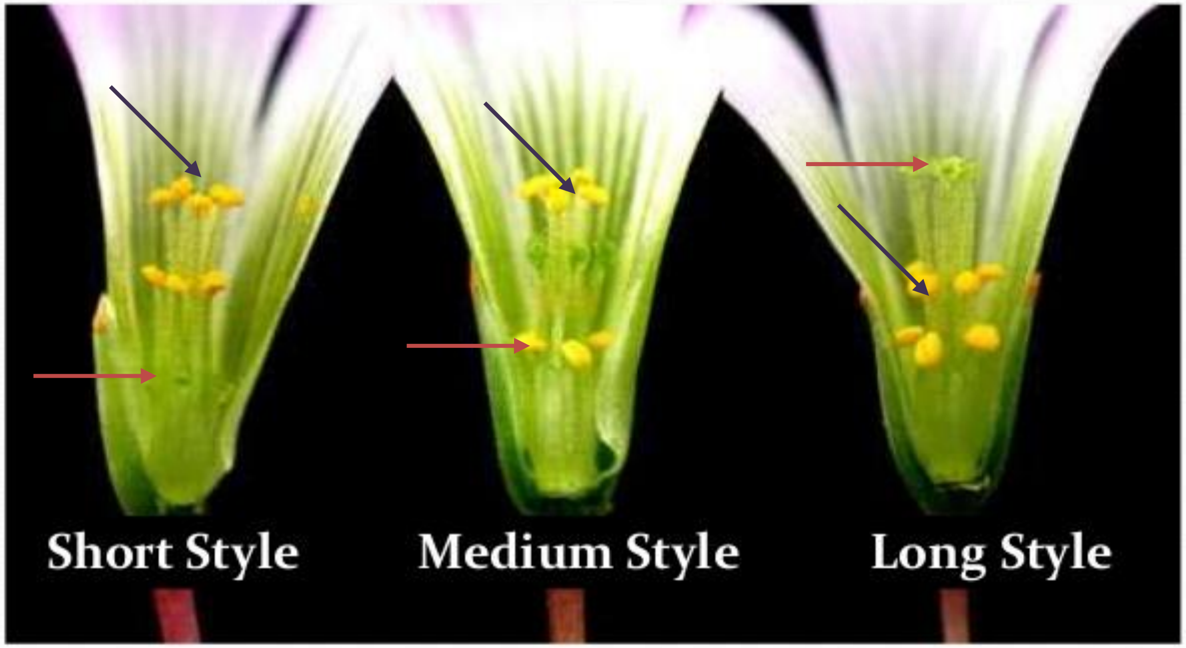
What is Tristyly?

In tristyly, styles and stamens have three different positions.

It is determined by **two genes S and M**, each with two alleles.

- S gives rise to short style,
- S and M to medium style and
- s and m to long style.
- The number of possible genotypes is greater, but a 1:1 ratio exists between individuals of each SI type.

Tristyly	*Lythrum salicaria*



Genetically controlled by two genes (Ss and Mm)

Position of style	No.of genes	Genotypes	
Short	S	Ssmm,SsMm or SsMM	
Medium	s and M	ssMm or ssMM	
Long	s and m	ssmm	

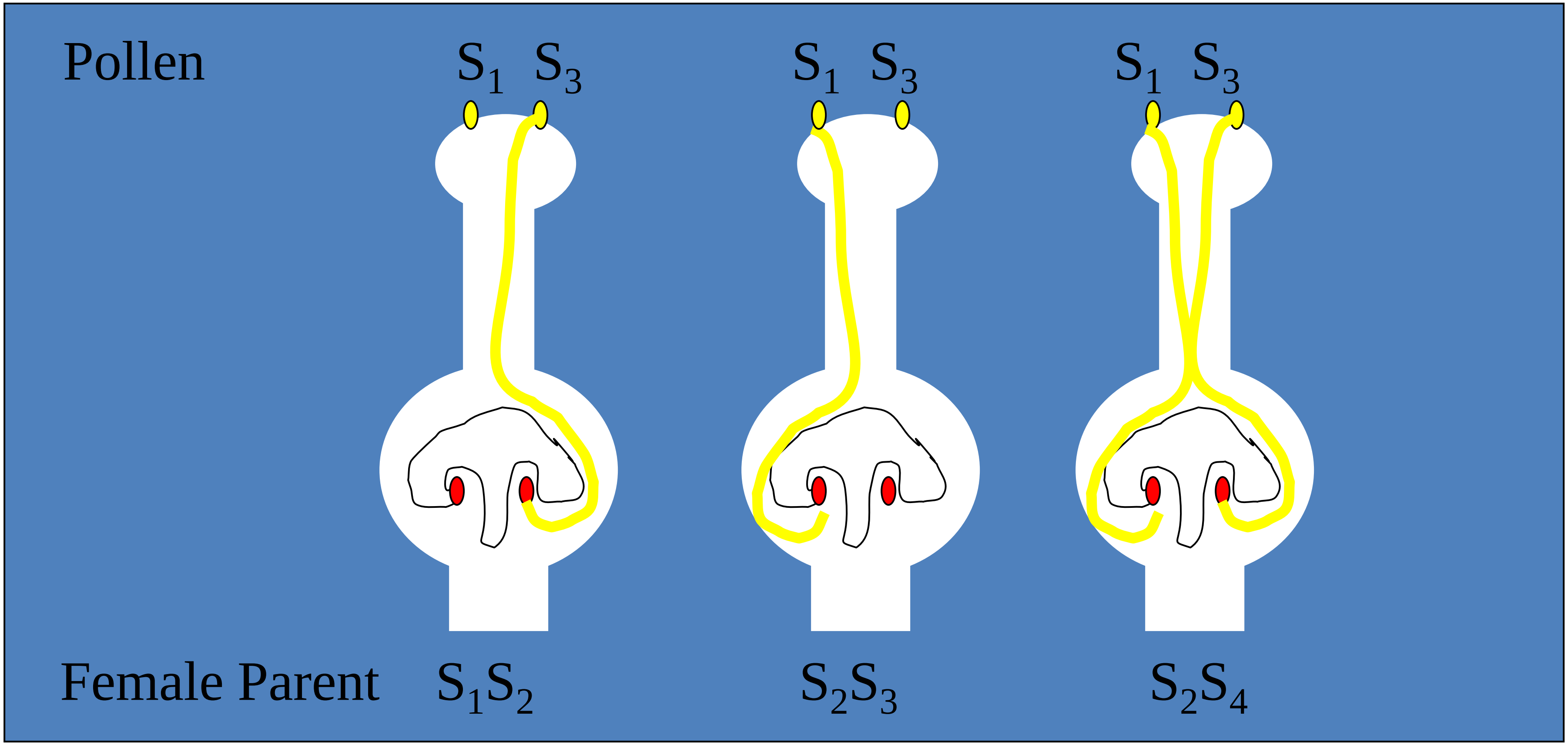
Site of gene expression

- The genes which govern SI may express only at different locations in the flower, *viz.,*
- 1) on the stigma (Stigmatic inhibition) – Radish, Cabbage, Sunflower, Cauliflower, *Linum grandiflorum*.
- 2) in the style (Stylar inhibition) – Bougainvillea, Helianthus
- 3) in the ovary (Ovarian inhibition) – Lillium, *Theobroma cacao*, Gasteria.

- **Homomorphic system**
- No morphological difference among flowers.
  1. **Gametophytic control**
- East and Mangelsdorf (1925) in *Nicotiana sandreae*
- Incompatibility reaction is determined by the pollen genotype.
- Single gene with multiple alleles with co-dominance effects.

GAMETOPHYTIC Self-incompatibility

In gametophytic, it is the single S allele of the pollen that determines pollination. If the S allele of the pollen grain matches either of the female alleles, there is no germination



- **Sporophytic system**
- Hughes and Babcock (1950) in *Crepis foetida and* Gerstel (1950) in Parthenium
- Controlled by S gene with multiple alleles.
- 30 alleles reported in *B. oleracea*
- Dominance is assumed.
- Incompatibility is determined by the genotype of the plant and not by genotype of pollen.
- **Crops:** Radish, Brassica and Spinach, sunflower.

SPOROPHYTIC Self-incompatibility

In sporophytic, it is the combined S alleles of the all pollen that determines pollination (i.e. it is the alleles of the male plant). If the S allele of any pollen grains matches the female, no pollination

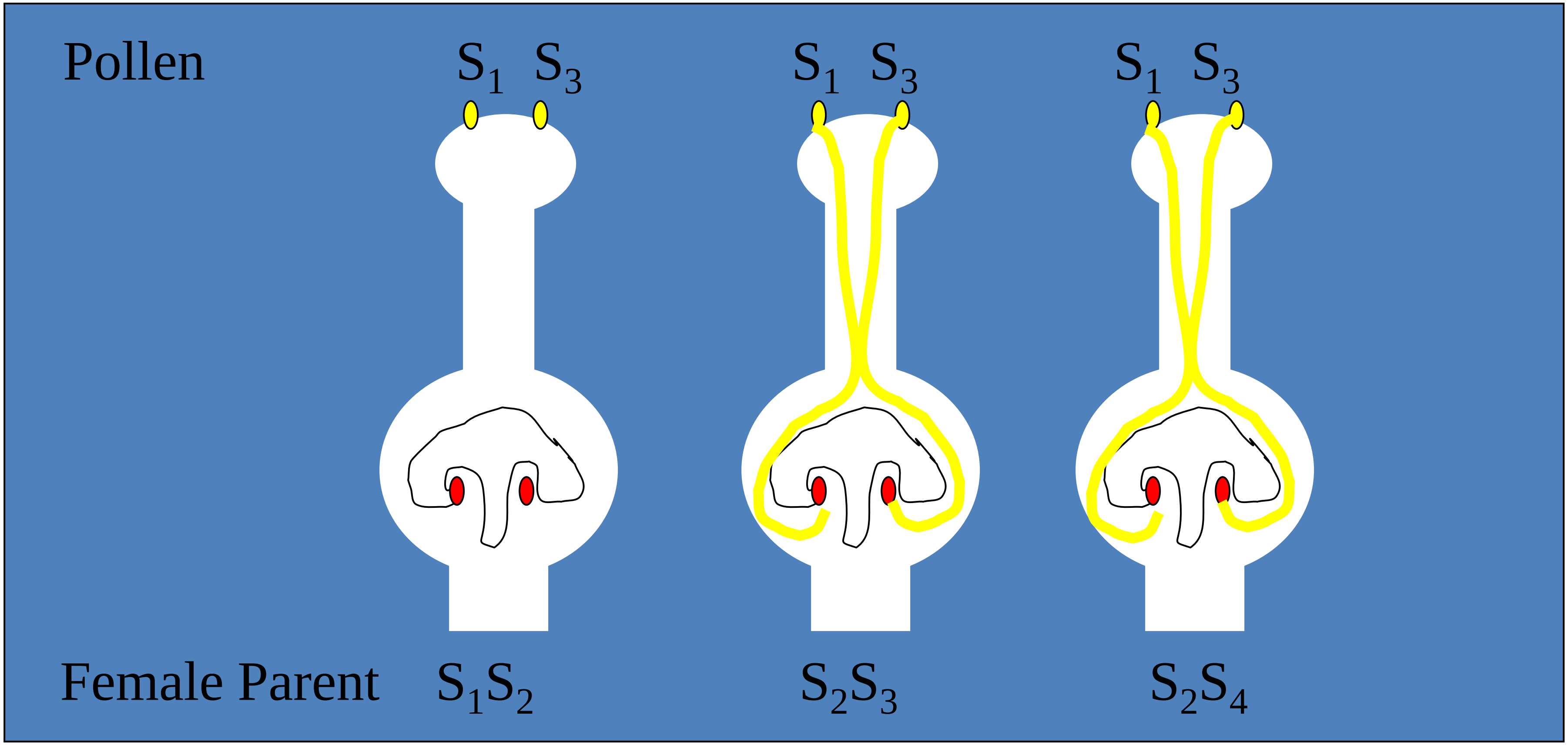


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TABLE 9.6

Comparison of gametophytic and sporophytic systems of self-incomatibility

Sl. No.	Particulars	Gametophytic System	Sporophytic System
1.	SI is controlled by	Genetic constitution of pollen	Genotype of sporophyte
2.	Genes involved	S gene with multiple alleles	S gene with multiple alleles
3.	Site of gene action	Style	Pollen or style
4.	Type of gene action	Individual	Dominance, individual or interaction
5.	Inhibition of pollen tube growth	In the style or ovary	On the stigma
6.	Pollen structure	Binucleate	Trinucleate
7.	Reciprocal differences	Not observed	Observed
8.	Recovery of parents from crosses	Male parent only	Both male and female
9.	Production of homozygotes	Not Possible	Possible
10.	Types of crosses	Sterile, partially fertile and fully fertile	Either fertile or sterile
11.	Effect of polyploidy	Fertility is restored	Fertility is not restored
12.	Production of chemical associated wit SI	During pollen formation	Before pollen formation
13.	Examples	Rye, Potato, Tomato, Red clover, White clover, etc.	Radish, Cabbage, Cauliflower, etc.

Application of Self Incompatibility in Plant breeding

- For hybrid seed production - *Brassica* (Cabbage and Brussels sprouts) and Cruciferous crops
- Combining desirable genes