Unit: 1 Mendelian Genetics and its Extension.

Principle of inheritance:

Mendel's expiments traced the results of breeding expiments (genetic cross). He used Strains of peas differing in well difine charact-eristics, like shape (round or wrinkled) seed colour (yellow or green), pod shape (inflated or wrinkled) and stem length (long or short). His concentration on well difine differences was of great importance, many breeders had previously tried to follow the inheritance of more gross qualities like body weight and were unable to discover any simple rules about theis trasmission from parent to offspring.

The Mendel's laws of inheritance include law of dominance, law of segregation and law of independent assortment.

① The principle of independent Segregation:

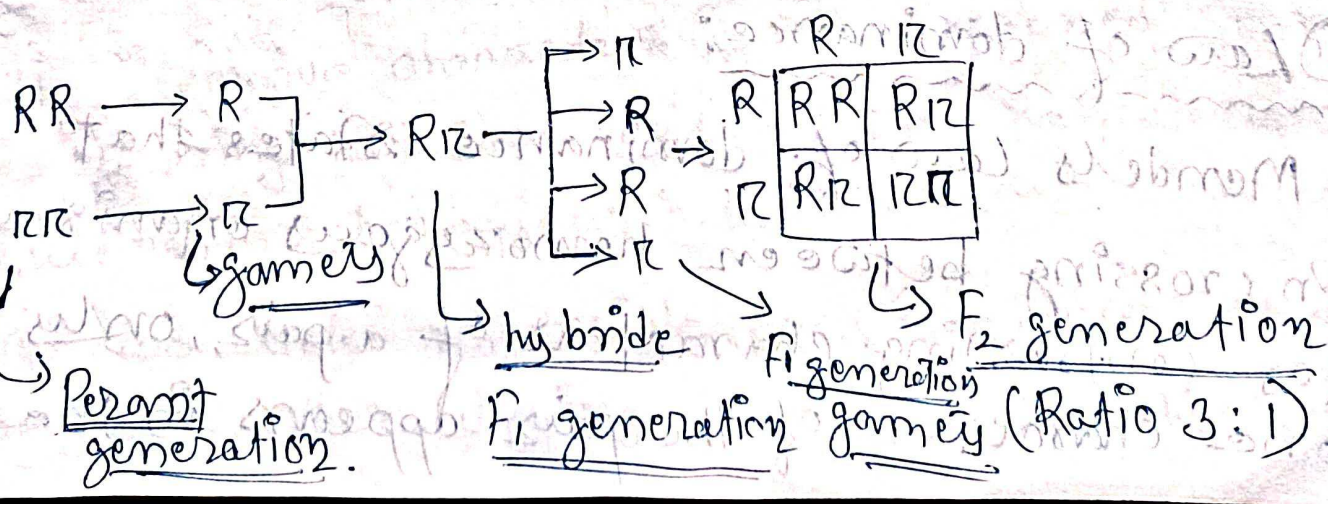
* Mendel performed a numbers of crosses between parents (P) differing in single characteristics (such as seed color or seed shape). All the progeny (F1 = first generation) had a appearance of one parent ~~Character~~ character only.

* The meaning of this results clear when Mendel set up genetic crosses between F1 offspring. These crosses gave the important result that the recessive trait reappeared in approximately 25% of the F_2 progeny, whereas the dominant trait appeared in 75% of these offspring.

* The ratio in F_2 of dominant to recessive traits was always approximately 3:1.

* The reappearance of the recessive characteristics in the F_2 generation indicates the recessive alleles are neither modified nor lost in the F_1 generation. but that the dominant and recessive genes are independently transmitted and so are able to segregate independently during the formation of sex cells. This is called principle of independent segregation and also reffered as a Mendel's first law.

* For example in a cross between peas having yellow seeds and peas having green seeds all the progeny had yellow seeds. The trait that appears in the first progeny is called dominant, whereas the trait that does not appears in F_1 is called recessive.



where R represents the dominant gene and r the recessive genes.

* The appearance or physical structure of an individuals as it's phenotypes and to it's genetic composition as its genotypes. Individuals ~~as~~ ~~its~~ ~~phenotypes~~ with identical phenotypes may possess different genotypes. thus to determine the genotype of an organism it is frequently necessary to perform genetic crosses for several generations. The term homozygous refers to a gene pair in which both the ~~materials~~ maternal and paternal genes are identical. In contrast, those gene pairs in which paternal and maternal genes are different are called heterozygous.

(ii) Law of dominance:

Mendel's law of dominance states that "In crossing between homozygous organisms for contrasting characters of a pair, only one characters of the pair appears in the

first generation.

* The law of dominance is the first law of heredity proposed from the works of Mendel. The law explains that all characters in an individual are controlled by distinct units called factors that occur in pairs.
* The pair can be homozygous or heterozygous and in the case of heterozygous pairs, one of the factors dominates the others.
* The character that dominates is called the dominant character and the one that remains unexpressed is the recessive characters.
* The recessive characters, even though latent is transmitted to the offspring in the same way as the dominant characters.
* The recessive characters is only expressed when the offspring has two copies of the same allele resulting in a homozygous individuals.
* The two alleles responsible for a character are brought together during fertilization, where

one of the alleles comes from the maternal gamete and the other from the paternal gamete.

Mendel's Law of Dominance (characteristics):

- The genes are the unit of inheritance that is transmitted via gametes that controls the expressions of different characters as a result of interaction with other genes.
- The genes occurs within chromosomes which in the case of diploid cells occurs in pairs. Each chromosome in the pairs comes from parent.

Example:

Mendel took a group of contrasting characters like the length of the stem, colour of the seed, shape of the cotyledon etc.

Among these, he took the example of stem length with two contrasting characters,

tallness and dwarfness.

The homozygous allele for tallness is represented by TT and the homozygous allele for dwarfness is represented by tt.

After crossing, the resulting hybrids might have the following possible genotypes, TT, Tt and tt.

The phenotypic ratio of the hybrid is in the ratio of 3:1, whereas the genotypic ratio is 1:2:1.

The tallness is the dominant character as it dominates over dwarfness, whereas dwarfness is only expressed when present in homozygous conditions.

(iii) principle of Independent Assortment:

Mendel start experiments with two strains of peas, each of which bread pure when mated with itself. One of the strains had round yellow seeds; the other, wrinkled green seeds. Since round and yellow dominated over wrinkled green, the entire F_1 generation produced round yellow seeds.

The F_1 generation then crossed within itself to produced a number of F_2 progeny, which were examined for seed appearance.

From above expriment Mendel found that he could interpret the results by the postulate of genes, if he assumed that each gene pair was independently transmitted to the ~~gamete~~ gamet during sex-cell formation. Any one ~~gene~~ contains only one type of allele from each gamete

gene pair. Thus the gametes produced by an F_1 (RrYy) will have the RY, Ry, rY or ry but never

Rr, Yy, YY or RR.

There is no tendancy of genes arising from one parent to stay together. As a result of F_2 progeny phenotypes appears in the ratio 9 round yellow, 3 round green, 3 wrinkle yellow and 1 wrinkle green as depicted in punnett square. This is called principle of independent assorment.

Example:

~~RR × YY~~

↓

RRYY × rryy → RY × ry → RrYy → RY-RY × ry-ry →

↓ Parental generation ↓ Gametes ↓ F_1 generation ↓ f_1 generation gametes.

1, 2

→

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

The ratio of phenotypes in this crossed is

9:3:3:1

The ratio of genotypes is :
RRYY:1, RRYy:2, RrYY:2, RrYy:4, RRyy:1, rrYY:1, Rryy:2, rrYy:2, rryy:1

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Incomplete dominance :

In general most "wild-type" alleles normally found in diploids are dominant because of their advantageous effects on the organism. On the molecular level wild-type gene products are mostly enzymes or other proteins necessary for proper metabolic functioning of the organism, whereas mutant gene products are often non-functional or only partly functional. There is therefore considerable evolutionary advantage for beneficial wild-type alleles to be dominantly expressed in any genotypic combinations in which they find themselves. Nevertheless, as we have seen mutant alleles may also be expressed

Phenotypically as in partial dominance, although the level at which this expression occurs may not always be obvious.

Example of incomplete dominance:

The cross between Red and White dog flower.

Red × White

RR × rr

$F_1 \longrightarrow$ Rr (Pink)

Self pollination done in F_1 (Rr) (Pink)

	R	r
R	RR	Rr
r	Rr	rr

The phenotype ratio 'Red : Pink : White

1 : 2 : 1.

Codominance:

A gene has a both quantitative and qualitative expression in the sense that it usually affects the production of a particular substance having a special structure or function. If we say the two alleles in a gene pair are each associated with different substances, codominace occurs when both substances appear together in the hetero-zygote.

For example:

$L^M L^M = M$ → phenotype, $L^N L^N = N$ → phenotype

$L^M L^N$ — gametes

MN → phenotypes → F_1 (Selfing)

F_2 →

Parents	Offspring		
	M	MN	N
$L^M L^M \times L^M L^M$ or MM × MM	All	—	—
$L^M L^M \times L^M L^N$ or MM × MN	1	1	—
$L^M L^M \times L^N L^N$ or MM × NN	—	all	—
$L^M L^N \times L^M L^N$ or MN × MN	1	2	1
$L^M L^N \times L^N L^N$ or MN × NN	—	1	1
$L^N L^N \times L^N L^N$ or NN × NN	—	—	All

Multiple alleles:

Alleles can be define as a genes that are members of the same gene pair, each kind of allele affecting a pasticulas character somewhat differently than the others. A particular gene pair in a diploid organism contains, by defination, only two alleles at a time, one for each member of the pair.

The grouping of all the different possible alleles that may be present in a gene pair is defined as a System of multiple alleles.

An example of multiple alleles in Drosophilla occurs for one of the gene pairs affecting eye colous for this gene one of the first hereditary changes

Recorded was a recessive mutant allele, white, associated with the complete absence of eye colour in the normal red-eyed fly. Since that time many alleles of white have been found which have a quantitative effect intermediate between white and the wild type red. Among these alleles and their combinations there are two clusters of phenotypic effects. The data are not complete for all possible combinations but show quantitatively that the pigments of heterozygotes (i.e, w^{w}/w^{or} and w^{as}/w^{+a}) fall between the values of their respective homozygotes. Thus dominance in this series appears to be incomplete although it would be difficult to make such a decision without this type of refined analysis.

→

Phenotype	Genotypes
w/w	white
w^t/w^t	tinged
w^a/w^a	apricot
w^{bl}/w^{bl}	blood
w^e/w^e	eosin
w^{ch}/w^{ch}	cherry
w^{a3}/w^{a3}	apricot-3
w^w/w^w	wine
w^{co}/w^{co}	coral
w^{sat}/w^{sat}	satsuma

Lethal alleles:

Detrimental physiological effects are apparently associated with the genes involved (w and vg). Some other genes have no effect on the appearance of the fly, ~~do~~ but do influence viability in some way. Other genes have such serious effects that the organism is unable to live. These are called Lethal genes.

Obviously, if the lethal effect is dominant and immediate in expression, all individuals carrying the gene will die and the gene will be lost. Some dominant lethals, however, have a delayed effect so that the organism lives for time. Recessive lethals carried in the heterozygous condition have no effect but may come to expression when matings between carriers occur.

Example:

Yellow coat colour in Mice:

A cross between pure breeding grey mice and pure breeding white mice produces all F_1 having grey colour. Interbreeding F_1 produced 3:1 ratio of grey and white mice in F_2.

But there was an exception, there were no true breeding yellow mice.

Cuénot also crossed yellow with grey and found that yellow colour was dominant over grey.

Y = Yellow

y = grey

$$Yy \times Yy \longrightarrow \underbrace{YY \times Yy}_{\text{yellow}} \quad \underbrace{yy}_{\text{grey}}$$

The phenotypic ratio is 2:1.

Epistasis:

Gene interaction may include instances where a novel phenotype does not appear but an effect caused by one gene pair interferes with or "hides" an effect caused by another gene pair. This type of interactions called epistasis. It is may be considered the counterpart of dominance relations between alleles. It may be caused by the presence of homozygous recessives at one gene pair or by the presence of a dominant allele at a gene pair. In addition the epistatic effect may be in only one direction, from one particular gene pair to another or in both directions when each gene pair is mutually epistatic to other.

In developmental terms we may envisage the appearance of colour in this example as arising from two sequential ~~of colour in~~ processes, the first of which is controlled by gene A and the other by gene B.

When the first process is inhibited (by aa) the second process cannot occur.

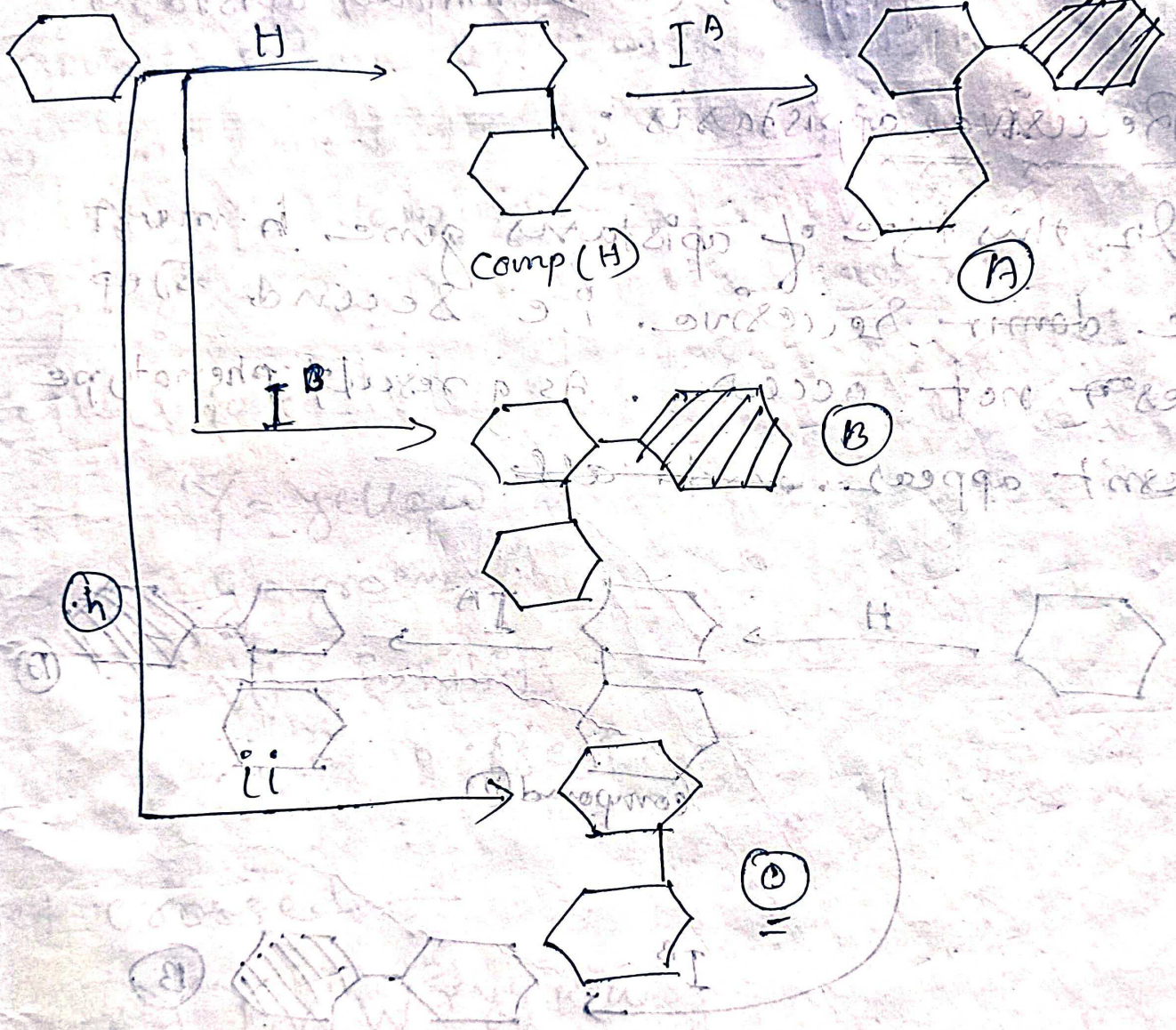
Origin Substances $\xrightarrow[1^{st}\ Step]{B\ gene}$ intermediate Substances $\xrightarrow[2^{nd}\ Step]{B\ gene}$ colour

When aa gene

Original Substances $\xrightarrow[1^{st}\ Step]{aa\ gene}$ No 2nd Step. → colour doesn't appear.

This pattern of ~~epistasis~~ epistatic interaction can also be seen in man, where the appearance of detectable ABO blood type antigen has been shown to depend upon the presence of gene H. Individuals that are homozygous for the very rare recessive allele h show no such antigens and are phenotypically of blood type O

This is called Bombay phenotype.



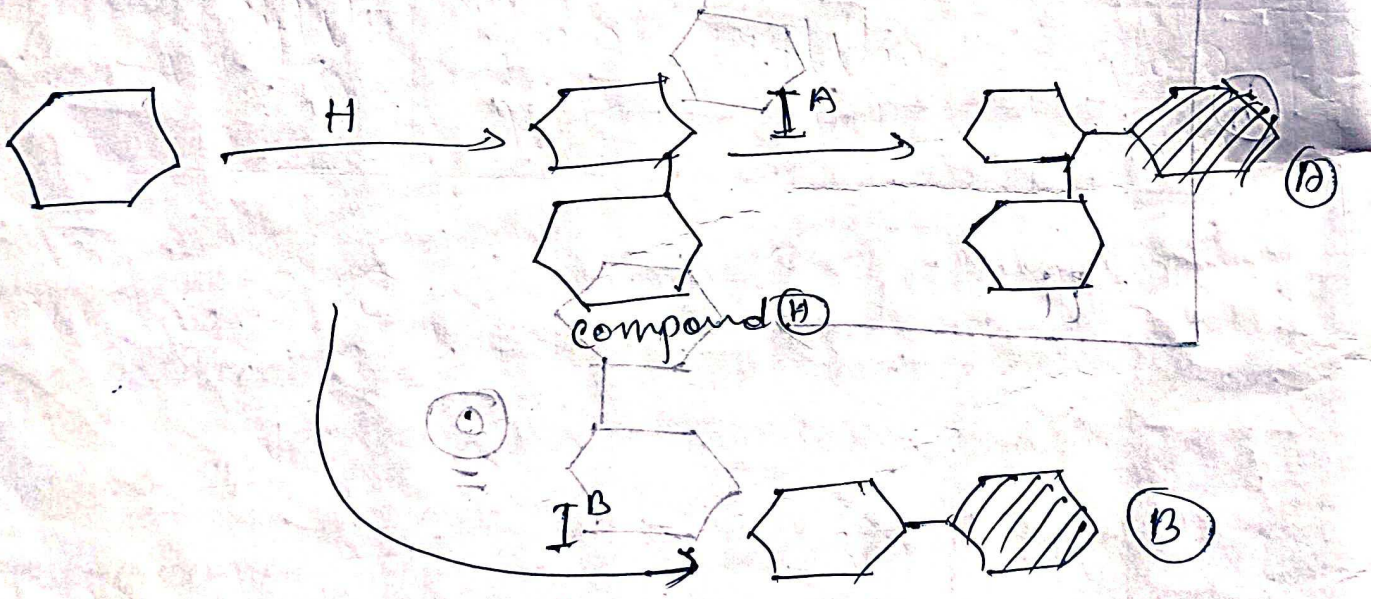
Where (A), (B), (O) = Blood group.

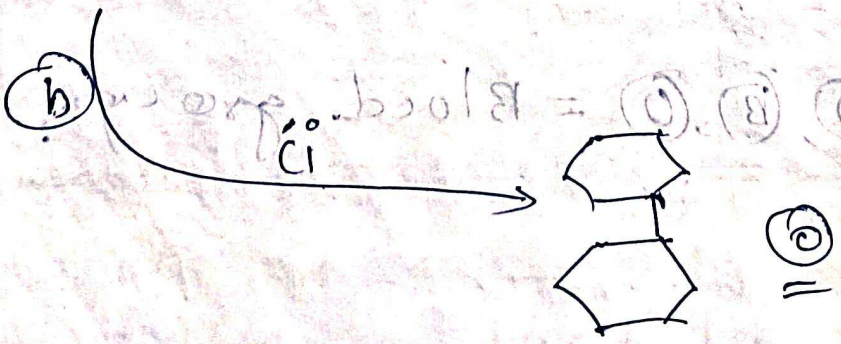
Apistasis are of two types:

(i) Recessive apistasis (ii) Dominant apistasis

(i) Recessive appistasis:

In this type of apistasis gene h must be ~~domin~~ recessive. i.e Second step does not occur. As a result phenotype doesn't appears. ~~and all~~





(11) Dominant epistasis :

In this type of epistasis character gives when at least one gene in intermediate compound must be dominant.

Example :-

Let us take

Y = yellow

y = green

W = inhibits

w = no effect

Get crossed —

→ YYWW × yyww

→ YyWw × yyww

g → YW × yw

F_1 → YyWw

F_2 =

	YW	Yw	yW	yw
YW	YYWW ~~yellow~~ white	YYWw ~~yellow~~ white	YyWw ~~yellow~~ white	YyWw ~~yellow~~ white
Yw	YYWw white	YYww yellow	YyWw white	Yyww yellow
yW	YyWw white	YyWw white	yyWW white	yyWw white
yw	YyWw white	Yyww yellow	yyWw white	yyww green

where, W = dominant epistasis.

This effect is found in Squash.

The phenotypic ratio is = 12 : 3 : 1

12 : 3 : 1 = White : yellow : ~~green~~ green

Sex-linked inheritance :-

Many mutant genes such as the X^g blood alleles in humans have been shown to be located on the X chromosome. Because of their location on the same chromosome as sex determiners, they are said to be Sex-linked. (Traits that are governed by gene located on the sex chromosomes exhibited a very distinctive pattern of transmitio of the one sex chromosomes is abnormal in

(is called Sex-linke inheritance)

female —

$$X^+X \times X^+Y$$

↓

$$\frac{X^+X^+}{F}, \quad \frac{X^+Y}{M} \quad \frac{XX^+}{F} \quad \frac{XY}{M}$$

In this case —

- 50% are males and 50% are affected
- 50% females are carriers and 0% males are carriers.

If x is abnormal in father —

X^+X^+ & xY

↓

F_1 — X^+x, X^+Y

— 100% females are carrier (X^+x)

— 0% males are affected.

If female is carrier and male is affected —

X^+x x xY

↓

X^+x, xx, X^+Y, xY

— 50% males are affected (xY)

— 50% females are carrier (X^+x)

— 50% females are affected (xx)

Sex-influenced inheritance:

The dominance of alleles may be differ in heterozygotes of the two sexes. This phenomenon is called Sex-influenced ~~inheri~~ dominance. Gene products of heterozygotes in the two sexes may be influenced differentially by sex hormones.

Both sexes ~~are~~ are genotypically alike ∈, the gene must behave as dominant in males and as recessive in female, i.e, only one allele is required for its expression ~~is~~ in the male, but the allele must be homozygous for expression in the female.

Dominance of the gene is apparently influenced by sex hormones.

Traits that show the pattern of inheritance characteristic of sex-influenced dominance are controlled by genes located on autosomal chromosomes, not on the sex chromosomes

Example:

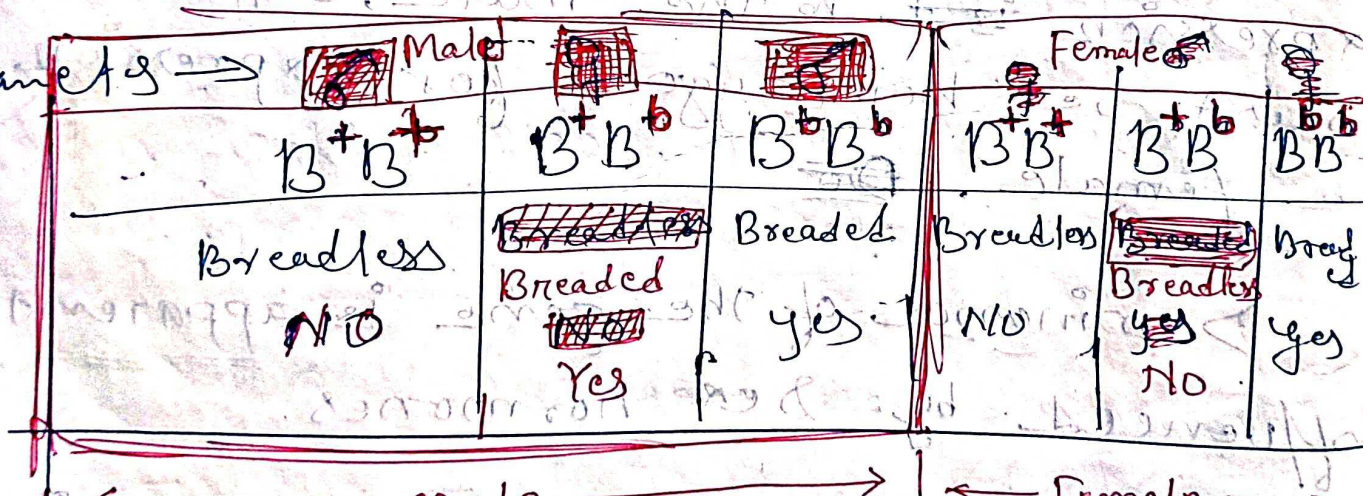
Let us crossed between Breadless (B^+B^+) and ~~(B^bB^b)~~ Breaded (B^bB^b) ~~goats~~ goats

♂ ♀

$B^+B^+ \times B^bB^b$

B^+B^b, B^+B^b

Breaded, Breadless



B^+ is for breadless

B^b → This allele is dominant in male and recessive in female. B^b = produced bread in males

B^bB^b = produced bread in female

Sex-limited inheritance: (It is the extreem form of sex-influenced inheritance)

Sex may be uniform in expression of a particular trait and yet transfer genes that produce a different phenotype in offspring of the other sex. This is called sex-limited gene expression. Sex hormones are apparently limiting factors in the expression of some genes. Other factors may be also involved in controlling the expression of sex-limited characteristics.

Example: plumage pattern in domestic chickens.

Genotype	male phenotype	female phenotype
H H	Hen	Hen
H h	Hen	Hen
h h	cock	hen

Gene maternal effect:

Eggs and embryos are expected to be influence by the maternal environment in which they developed. Even those remove from the body of the mother at an early stage receive cytoplasm and nutrients in the egg from the mother, and special influences on gene action may have already taken effect. Certain potentialities of the egg are known to be determined before fertilization, and in some cases these have been influenced by the surrounding maternal environment. Such predetermination by genes of the mother, & rathas than those of the progeny is called maternal effect. Existance of a maternal effect is commonly substained or disaproved by reciprocal crosses. If a maternal effect is involved, & results from reciprocal crosses will be different from each other, with genes of the mother being expressed.

In some ~~cases~~ cases the maternal effect does not diminish during development but lasts through adults life. Such instances occur when development is started in a particular direction cannot be reversed.

Maternal effects thus arises from egg cytoplasm that has been modified by chromosomally transmitted genes. Its distinguishing characteristic is the difference in the results of reciprocal crosses, so that cytoplasm produced by a particular genotype ~~effect~~ act differently on developing zygot than cytoplasm ~~produced~~ produced by by a different genotype.

In maternal effect, phenotypic changes appears because of differences in egg cytoplasm ~~rather~~ rather than difference in sex chromosomes and often affect both male and female offspring equally.

Among mammals, maternal effects can be more broadly defined, since mammalian mothers may ~~effect the~~ affect the development of their offspring not only through the egg cytoplasm but also through the uterine environment.

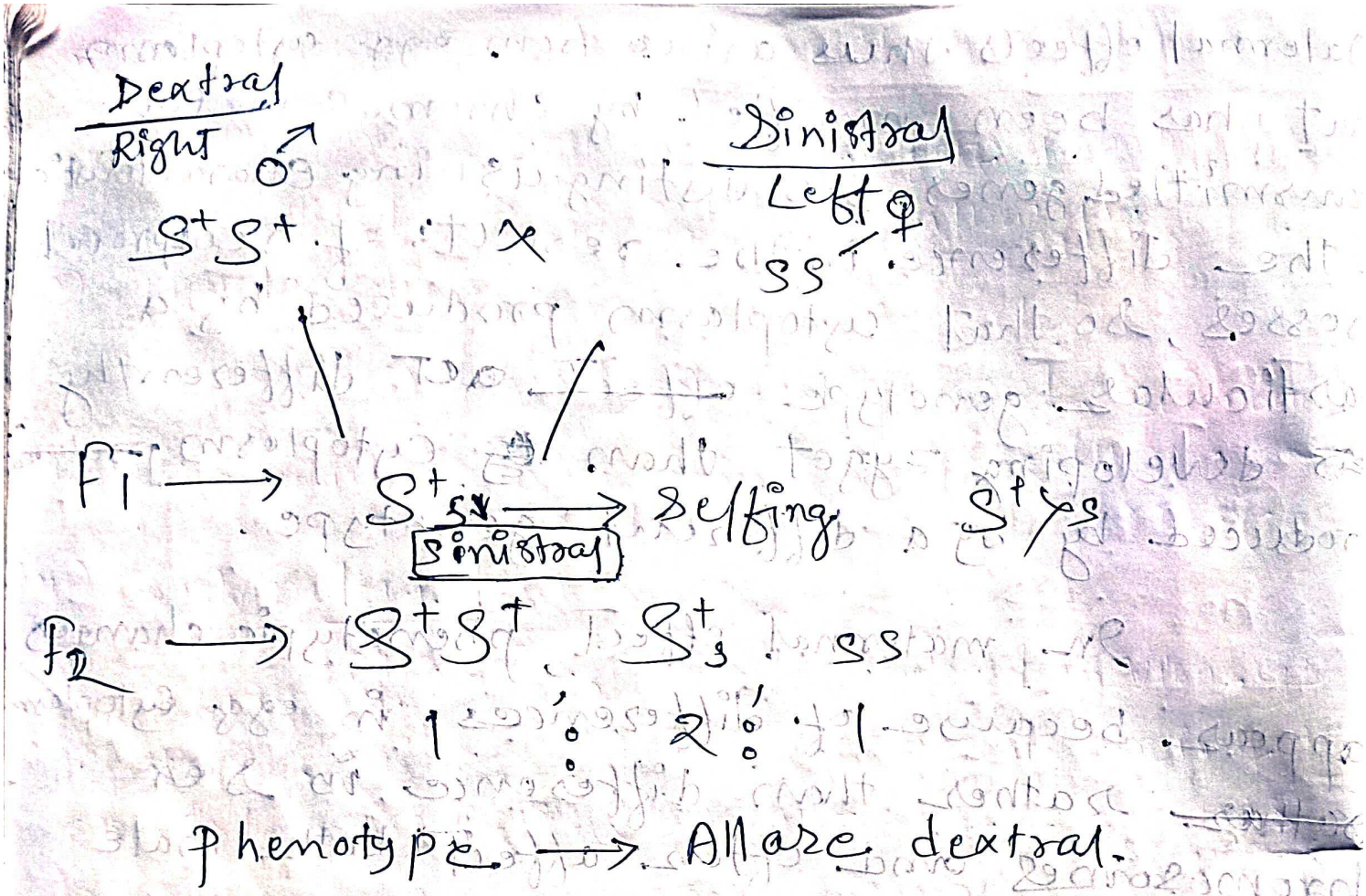


Fig: A cross illustrating a maternal effect in the Snail Limnaea peregra.

Pleiotropy:

- Pleiotropy refers to disorders where multiple seemingly unrelated organ systems are affected.

- Pleiotropy occurs when a single mutation or gene/allele affects more than one phenotypic characteristic.

- pleiotropy means that a single gene affects two or more characters.

pleiotropy broadly divided into 7 types—

(i) Artefactual:

Adjacent but functionally unrelated genes affected by same mutation. Example:- Clarcet

(ii) Secondary:

Simple primary biochemical disorder leading to complex final phenotype.

(iii) Adoptive:

one gene product used for quite different chemical purposes in different tissues.

(iv) Parsimonious:

One gene product used for identical chemical purposes in multiple pathways.

(v) Opportunistic:

One gene product playing a secondary role in addition to its main function.

(vi) Combinatorial:

One gene product employed in various ways and with distinct properties depending on its different protein partners.

(vii) Unifying:

One gene, or cluster of adjacent genes encoding multiple chemical activities that support a common biological function.