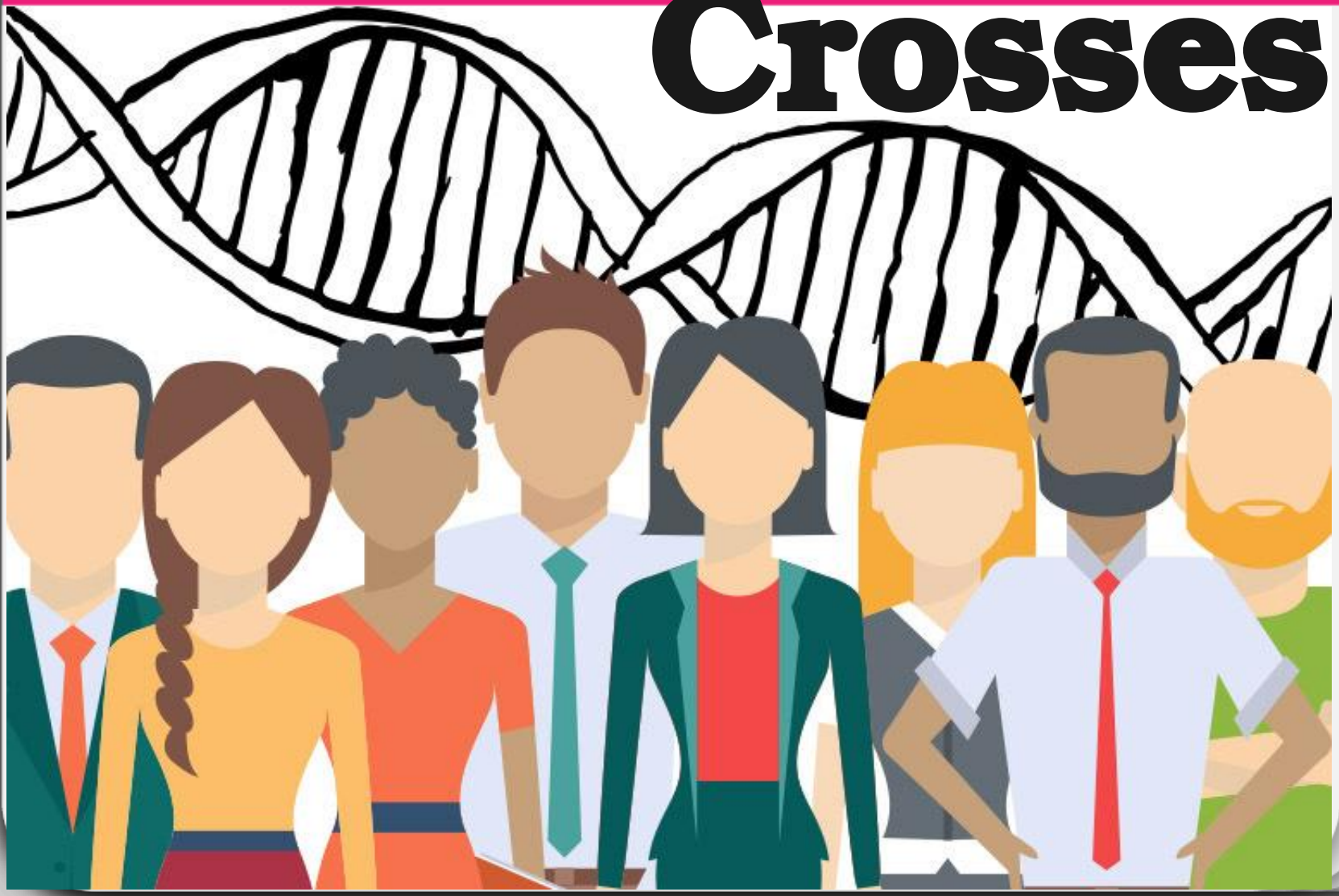
Genetic Crosses

23



Mendelian Ratios

- Simple Mendelian genetics depends on character traits which are determined by one gene (located on an autosome), for which there are two alleles, one completely dominant to the other.

	Flower color	Flower position	Seed color	Seed shape	Pod shape	Pod color	Stem length
P	Purple × White	Axial × Terminal	Yellow × Green	Round × Wrinkled	Inflated × Constricted	Green × Yellow	Tall × Dwarf
F_1	Purple	Axial	Yellow	Round	Inflated	Green	Tall

Exceptions to simple Mendelian Genetics

- Deviations from standard Mendelian ratios are observed when
  (1) Alleles are not completely dominant or recessive ; incomplete dominance or codominance
  (2) A particular gene has more than 2 alleles ; multiple allelic condition
  (3) A gene is carried on the sex chromosomes Sex-linked genes

GENETIC CROSS

Monohybrid

1 gene (on autosome)→ 1 trait
1 gene → 2 alleles
(completely dominant or recessive)
Total = 2 phenotypes

Expected

Aa x Aa → 3 : 1
&
Aa x aa → 1 : 1

Deviations

1. Incomplete dominance } Alleles not completely dominant or recessive
2. Co-dominance
3. Multiple alleles → Gene has more than 2 alleles
4. Sex-linkage → Gene on sex chromosome

Dihybrid

2 genes (on different autosome)→ 2 traits
Each gene → 2 alleles
(completely dominant or recessive)
2 genes, 4 alleles hence…
Total = 4 phenotypes

Expected

AaBb x AaBb → 9 : 3 : 3 : 1
&
AaBb x aabb → 1 : 1 : 1 : 1

Deviations

Not in H1 syllabus

Copy onto blank space on Pg 23

Exceptions to simple Mendelian genetics

- In the previous lectures, you learnt about the standard Mendelian ratios such as :

  3 : 1 (monohybrid)

  9 : 3 : 3 : 1 (dihybrid) when 2 heterozygotes cross ;

  1 : 1 (monohybrid)

  1 : 1 : 1 : 1 (dihybrid) when heterozygote cross with homozygous recessive individual in test cross)

Standard Mendelian ratios will not be observed.

What I need to know …01

- 2(n)
  Use genetic diagrams to solve problems in dihybrid crosses, including those involving **incomplete dominance, codominance, multiple alleles** and **sex linkage**

Exceptions

- Incomplete dominance
- Codominance
- Multiple alleles
- Sex-linked genes

(1) Incomplete dominance

- The situation where the **heterozygote (Aa)** exhibits a **phenotype** which is **intermediate between homozygous dominant (AA) and homozygous recessive (aa)** forms.

- For incomplete dominance, one allele of a gene may code for **functional gene product** while the other allele may code for **non-functional or mutated gene product**. **Neither allele of an allelic pair is dominant or recessive with respect to each other**, both alleles contribute equally to the phenotype of the heterozygote.

(1) Incomplete dominance

- Thus when a cross between parents with two different phenotypes, some offspring with a "third" phenotype that is an **intermediate** of the parental traits would result.

Snapdragon flower

Incomplete dominance in snapdragon

- Flower colour in snapdragon is controlled by **two alleles** of a **flower-colour gene**
- The two alleles are C^R and C^r.

Addition to notes; way of allele representation: allele symbol (R or r) SUPERSCRIPTED on a common alphabet for the gene (e.g. C)

Gene locus encoding for flower colour → C^R or C^r allele

Incomplete dominance in snapdragon

- Individuals that are homozygous for the red-flower allele (C^RC^R) have red flowers

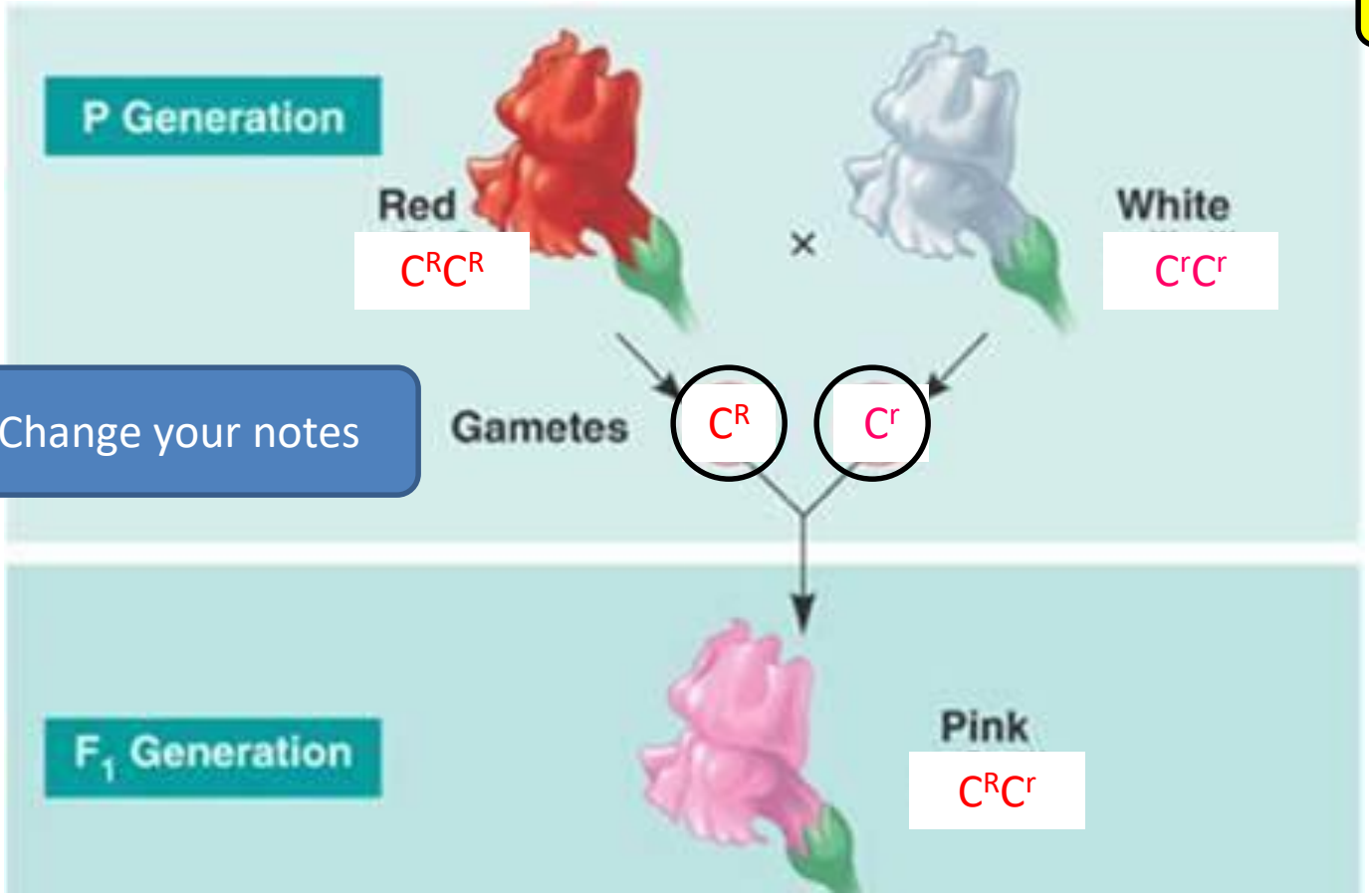
C^R — — C^R

Incomplete dominance in snapdragon

- Those that are homozygous for the white-flower alelle (C^rC^r) have white flowers.

C^r ▬ ▬ C^r

Pg 24



Change your notes

Incomplete dominance in snapdragon

C^R C^r

Incomplete dominance in snapdragon

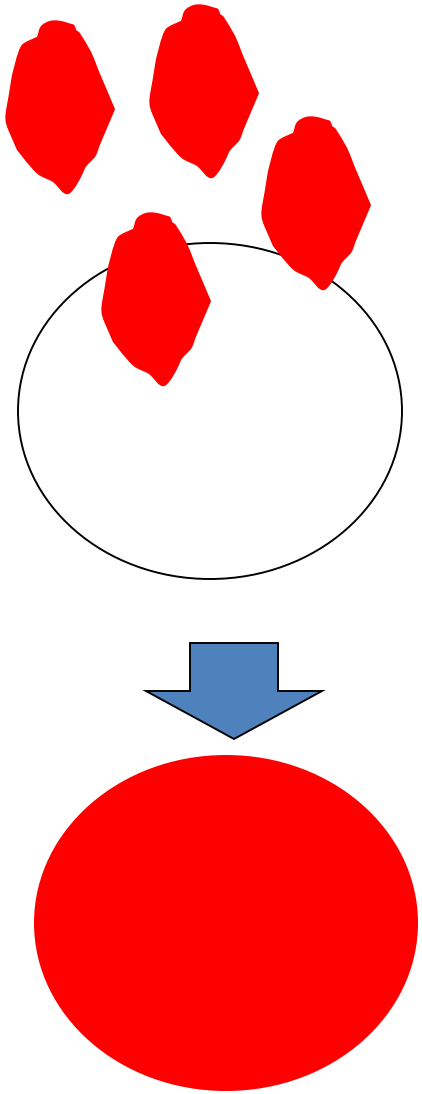
Biochemical explanation for this

- Allele C^R encodes for a functional enzyme that converts white pigment to red
- Allele C^r encodes for a non-functional / mutated enzyme that cannot catalyse the reaction leading to red pigment synthesis

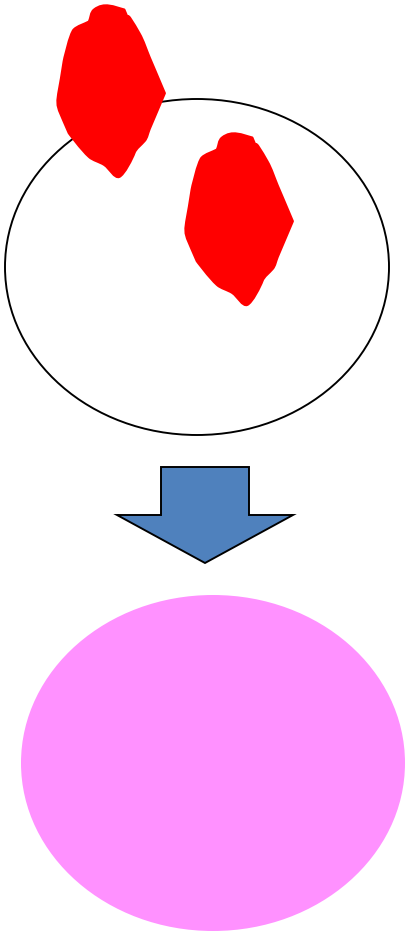
Incomplete dominance in snapdragon

Biochemical nature for this :

- Heterozygotes (C^RC^r) produce inadequate quantity of enzyme to synthesise enough red pigment so its flowers are pink ;
- Pink is a flower colour intermediate between red and white.



2 copies of allele C^R



1 copy of allele C^R

(in the heterozygote)

30 s

Exercise [Pg.24]

Punnett Square to show the fusion of gametes produced by the F_1 generation

30

Gametes		

Exercise [Pg.24]

Punnett Square to show the fusion of gametes produced by the F_1 generation

Gametes	C^R	C^r
C^R	C^RC^R (Red)	C^RC^r (Pink)
C^r	C^RC^r (Pink)	C^rC^r (White)

Phenotypes of F_2 generation:

	red flowers	:	pink flowers	:	white flowers
Phenotypic ratio	1	:	2	:	1

Other example – Wavy hair in humans

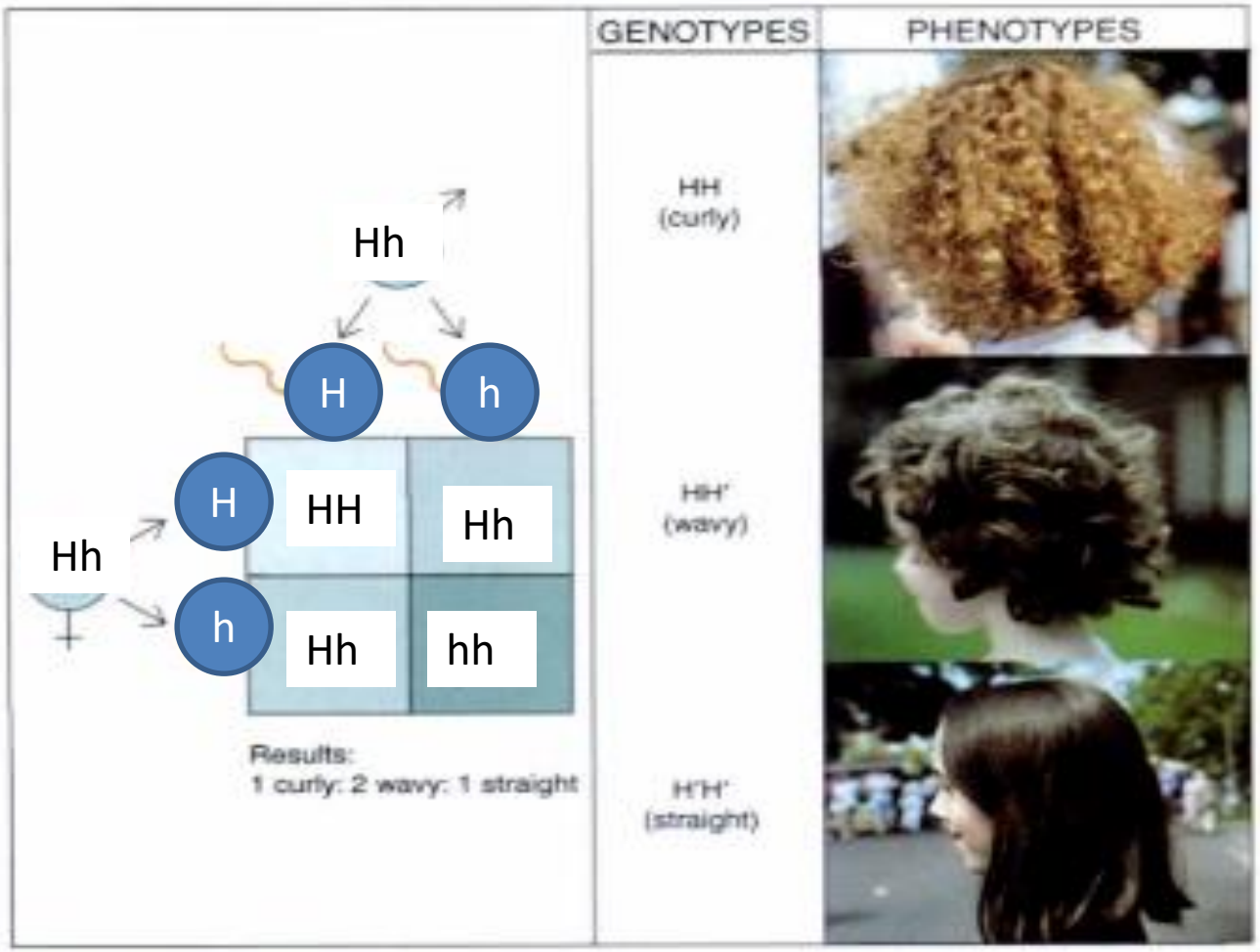


Figure 21.16 **Incomplete dominance.** Sometimes, alternative alleles are not dominant or recessive. For example, neither straight nor curly hair is dominant in Caucasians. The predicted ratio of the offspring of two wavy-haired people will be one curly-haired individual, one straight-haired individual, and two wavy-haired individuals. Wavy hair is the phenotypic intermediate.

Source: http://tinyurl.com/je3qnl9

Exceptions

- Incomplete dominance
- Codominance
- Multiple alleles
- Sex-linked genes

???

(2) Codominance

- Codominance (without a hyphen in the spelling) are displayed only in the phenotype of the heterozygote.
- Codominance occurs when the 2 alleles of a gene are **equally dominant**, and hence both alleles are equally expressed in the phenotype of the heterozygote, e.g. the human ABO and MN blood group systems and coat colour in cattle.
- The two alleles both affect the phenotype in separate, distinguishable ways.
- This occurs when a cross between organisms with two different phenotypes produces offspring with a **phenotype in which both of the parental traits appeared together** in the offspring.
- For codominance, both alleles of a gene code for functional products, therefore both contribute equally to the phenotype of the heterozygotes.

Roan colour in Shorthorn cattle

Codominance in Shorthorn cattle

- Cattle hair colour is controlled by **a gene** with **two alleles**
- The two alleles are C^W and C^R.

Add in to notes; way of allele representation: 2 different allelic symbols (W or R) SUPERSCRIPTED on a common alphabet for the gene (e.g. C)

Why not just R vs r???
Both alleles are equally dominant wrt to each other; both allele codes for diff functional products

Gene locus for coat colour → C^R or C^W

Codominance in Shorthorn cattle

- An animal which is homozygous for the C^R allele has red hair

C^R C^R

Codominance in Shorthorn cattle

- An animal which is homozygous for the C^W allele has white hair

C^W C^W

Codominance in Shorthorn cattle

- An animal with a heterozygous genotype C^RC^W is roan in colour, meaning its coat contains both red and white hairs

C^R C^W

Predict what happens for incomplete dominance?

?????

C^R C^W

Codominance in Shorthorn cattle

- The roan coat colour is not intermediate between the white and red hair phenotypes.
- The roan coat colour is the result of the equal phenotypic expression of both types of alleles in the heterozygote.

Pg 26 Blank space

1 min

Exercise [Additional]

Complete the Punnett Square to illustrate a cross between 2 ROAN animals; show phenotypic ratio

Punnett Square to show the fusion of gametes produced by the F_1 generation


X


Gametes		

1:00

Pg 25 Blank space

Exercise [Additional]

Punnett Square to show the fusion of gametes produced by the F_1 generation


X


Gametes	C^R	C^W
C^R	C^RC^R Red	C^RC^W Roan
C^W	C^RC^W Roan	C^WC^W White




Phenotypes of F_2 generation:

	red coat :	roan coat :	white coat
F_2 Phenotypic ratio	: 1	: 2	: 1

MN blood group system in humans

MN blood group system in humans

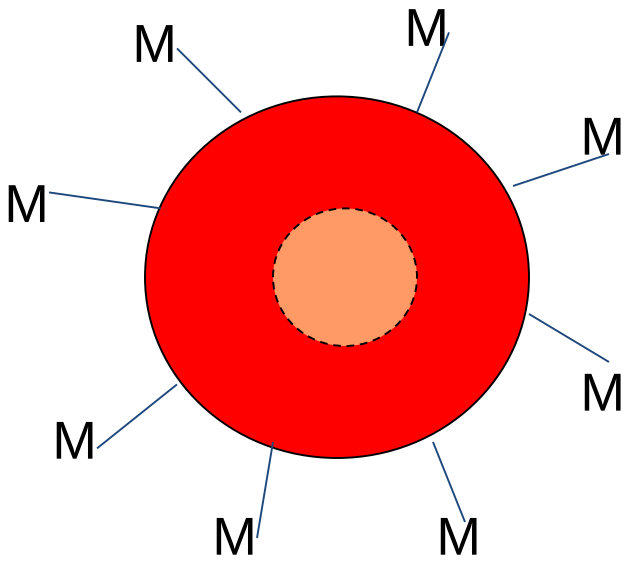
- Ability to produce the M and N antigens is determined by a gene with two alleles
- The two alleles are L^M and L^N

Antigens = molecules recognized by antibodies of the immune system

Gene locus encoding for M and N antigens → L^M or L^N

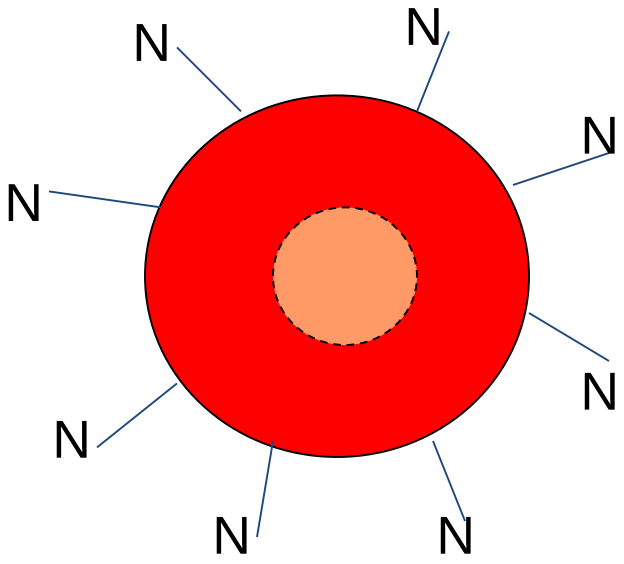
MN blood group system in humans

- L^M allele results in the production of glycoprotein M on the red blood cell membrane



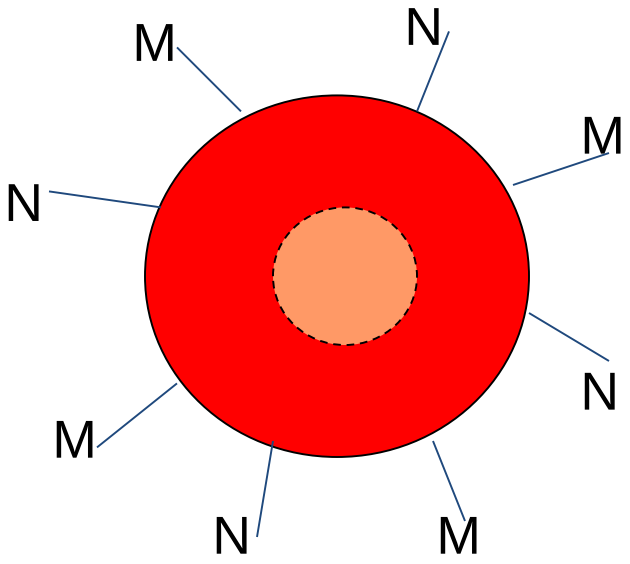
MN blood group system in humans

- L^N allele results in the production of glycoprotein N on the red blood cell membrane



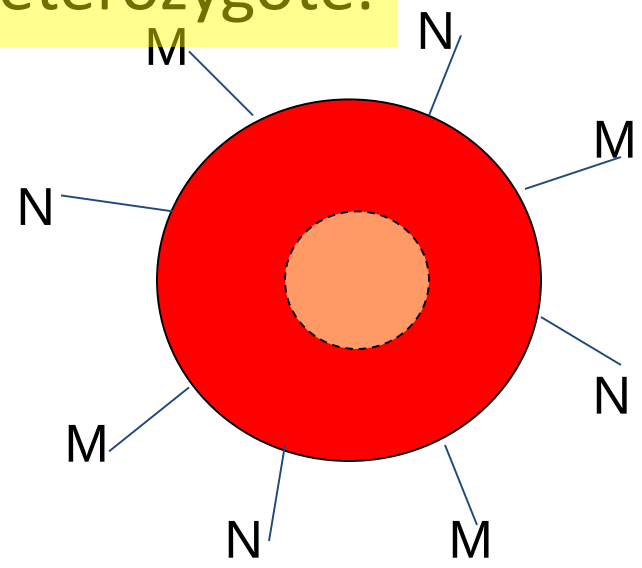
MN blood group system in humans

- For the genotype $L^M L^N$, both types of glycoproteins exists on the red blood cell membrane.



MN blood group system in humans

- The MN phenotype is not intermediate between the M and N phenotypes. The MN blood type is the result of the equal phenotypic expression of both alleles in the heterozygote.



STANDARD CONVENTION FOR WRITING GENE/ALLELES IN INCOMPLETE DOMINANCE OR CODOMINANCE:

Using one alphabet is allowed e.g. C^R and C^r if allele C^r codes for non-functional product

- Where codominance or incomplete dominance exist, the **gene should be designated by an uppercase letter** and the **alleles** indicated by appropriate **superscript** letters, e.g C^R and C^W for the two alleles involved in roan coat colour in cattle.

1 min

TASK:

Without looking at P26,

Scan through P24 and P25,

Compare between codominance and incomplete dominance

1:00

Comparison of codominance and incomplete dominance

SIMILARITY

- Both are displayed only in the phenotype of the heterozygote

Comparison of codominance and incomplete dominance

DIFFERENCES

- For **codominance**, both alleles of a gene code for functional products, therefore both contribute equally to the phenotype of the heterozygotes; the effects of both alleles can be seen in codominance.
- In contrast**, incomplete** dominance has one of the alleles coding for a non-functional protein, thus the heterozygote displays an intermediate phenotype.

Test yourself

Incomplete Dominance or Codominance in Andalusian fowls?

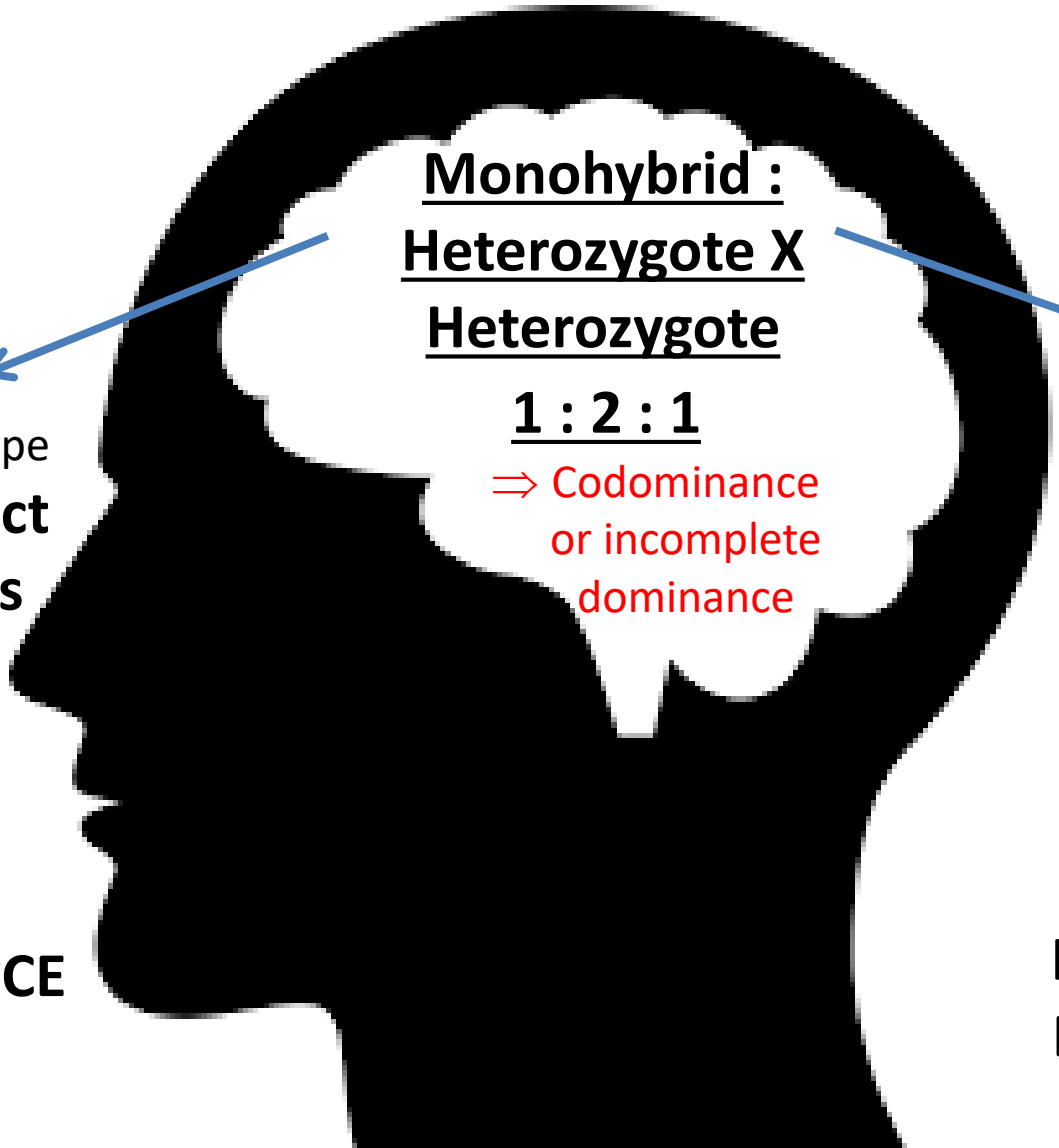
Parental Generation

X

F_1 Generation

Codominance!

Add-on to important ratios [Pg.22]



Monohybrid :
Heterozygote X
Heterozygote

1 : 2 : 1

⇒ Codominance or incomplete dominance

Heterozygote express a phenotype in which the **effect of both alleles** are seen

CODOMINANCE

Heterozygote express an **intermediate** phenotype between the two homozygous forms

INCOMPLETE DOMINANCE

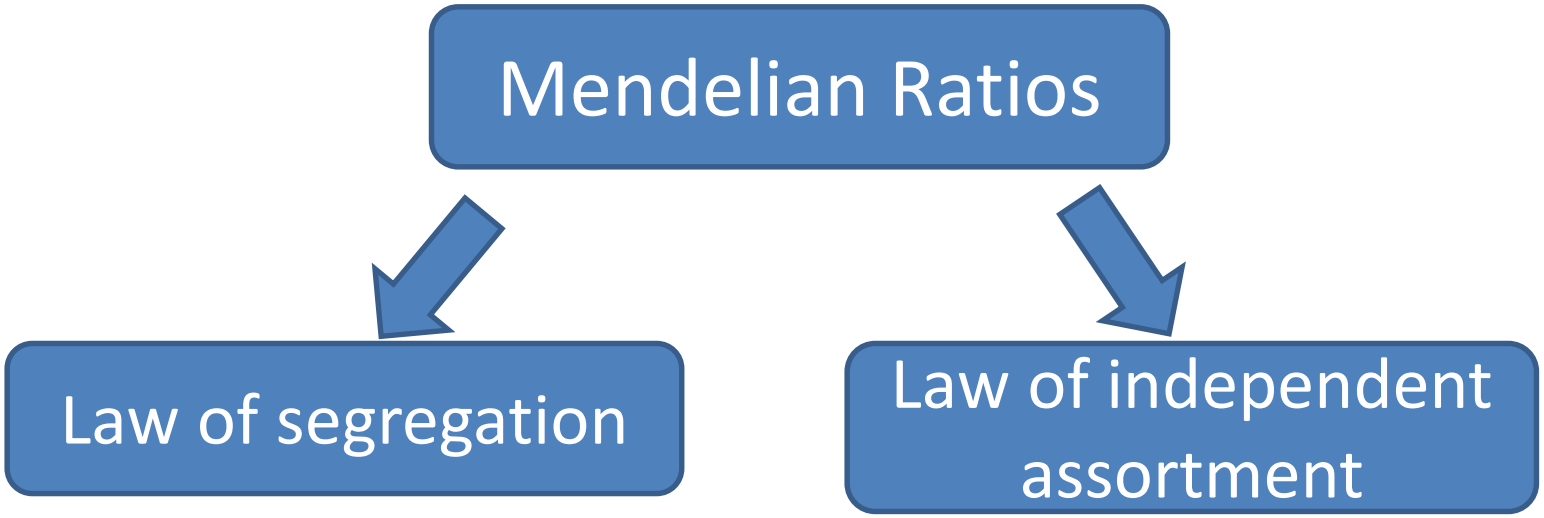
Recall: List down the expected Mendelian phenotypic ratios

- Monohybrid hetero x hetero cross (Aa x Aa):
- Dihybrid hetero x hetero cross (AaBb x AaBb):
- Monohybrid Test cross involving heterozygote (Aa x aa):
- Dihybrid Test cross involving heterozygote (AaBb x aabb):

30 s

Recall: List down the expected Mendelian phenotypic ratios

- Monohybrid hetero x hetero cross (Aa x Aa): 3:1
- Dihybrid hetero x hetero cross (AaBb x AaBb): 9:3:3:1
- Monohybrid Test cross involving heterozygote (Aa x aa):1:1
- Dihybrid Test cross involving heterozygote (AaBb x aabb): 1:1:1:1



Exceptions

- Incomplete dominance and codominance
- Multiple alleles
- Sex-linked genes

2 alleles: norm

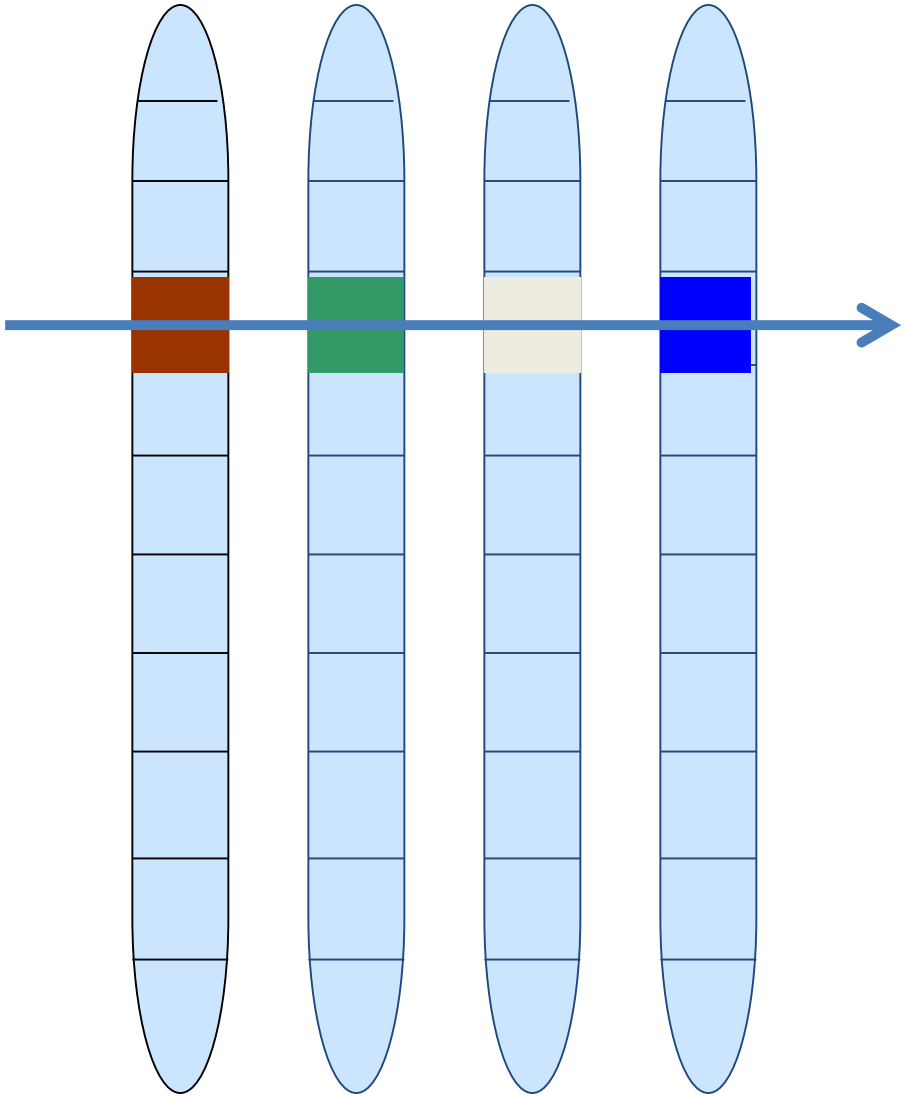
(3) Multiple alleles

- Most genes exist in more than two allelic forms

e.g. gene for eye color

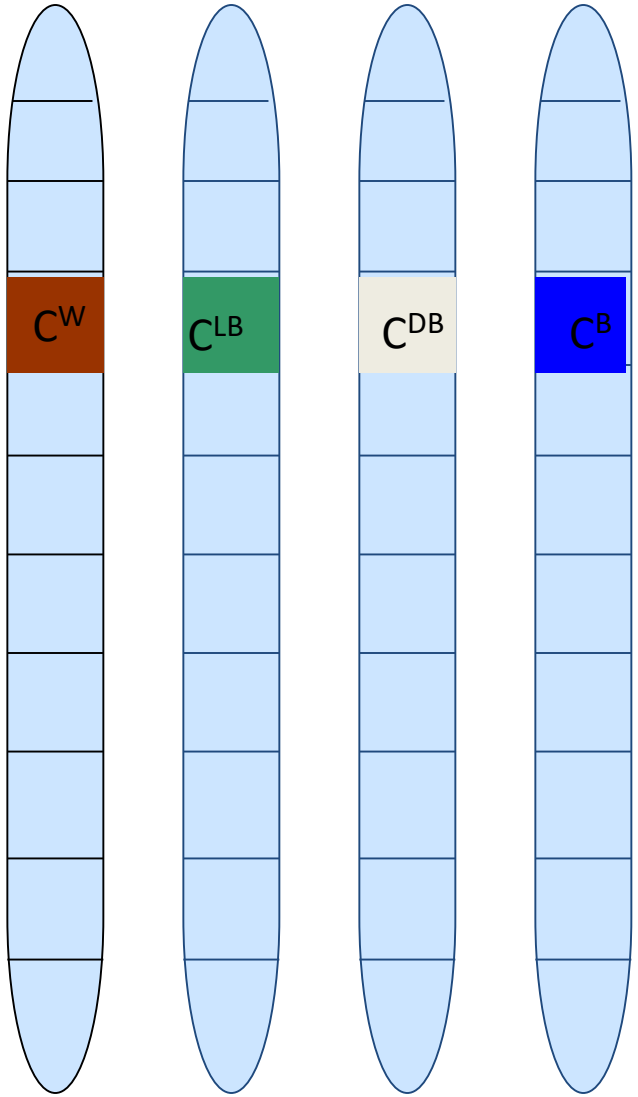
- If a gene controlling a particular characteristic has three or more alleles, these alleles are called multiple alleles

(3) Multiple alleles



Gene locus for particular Characteristic
E.g. fur coat colour

(3) Multiple alleles



Key:

C^w: White fur
CL^B: light brown fur
C^{DB}: Dark brown fur
C^B: Black fur

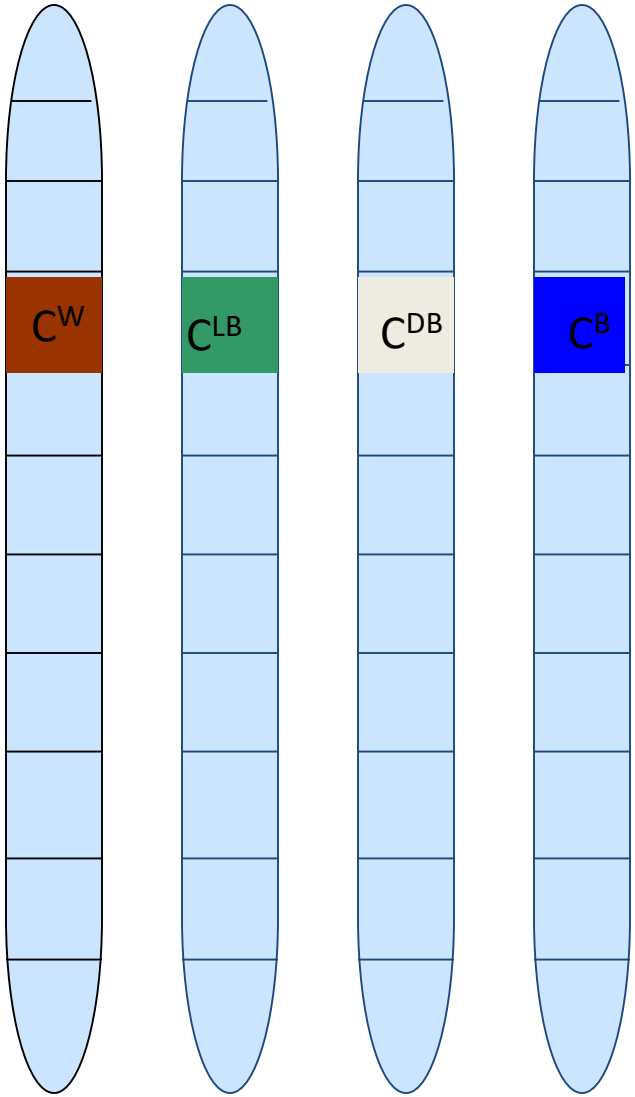
4 possible alleles for fur color in this gene locus

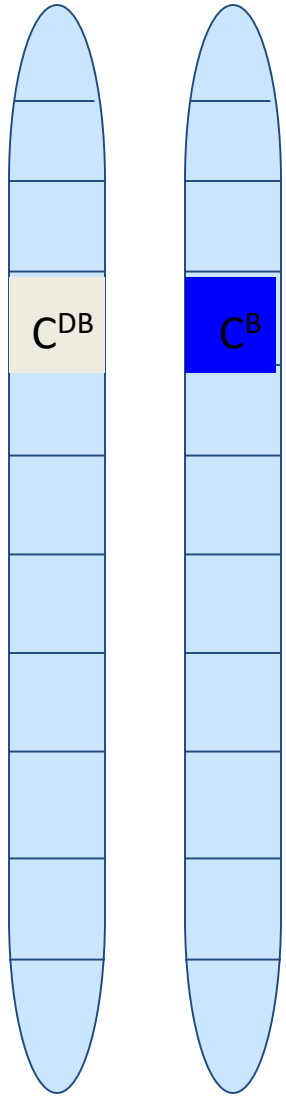
(3) Multiple alleles

1 paternal chromosome set and 1 maternal chromosome set

- However, in any one diploid individual, **at most two (alleles)** can be present, where they occupy the same gene loci on homologous chromosomes.

(3) Multiple alleles





IMPORTANT

Only 2 alleles in a diploid organism

Paternal chromosome

Maternal chromosome

(3) Multiple alleles

- Standard convention for writing multiple alleles.

If several alleles are being considered e.g. human blood groups, the gene should be allocated an uppercase letter and each allele an appropriate superscript, e.g. I represents the gene; A, B and o represent alleles; thus the alleles are indicated I^A, I^B and I^o.

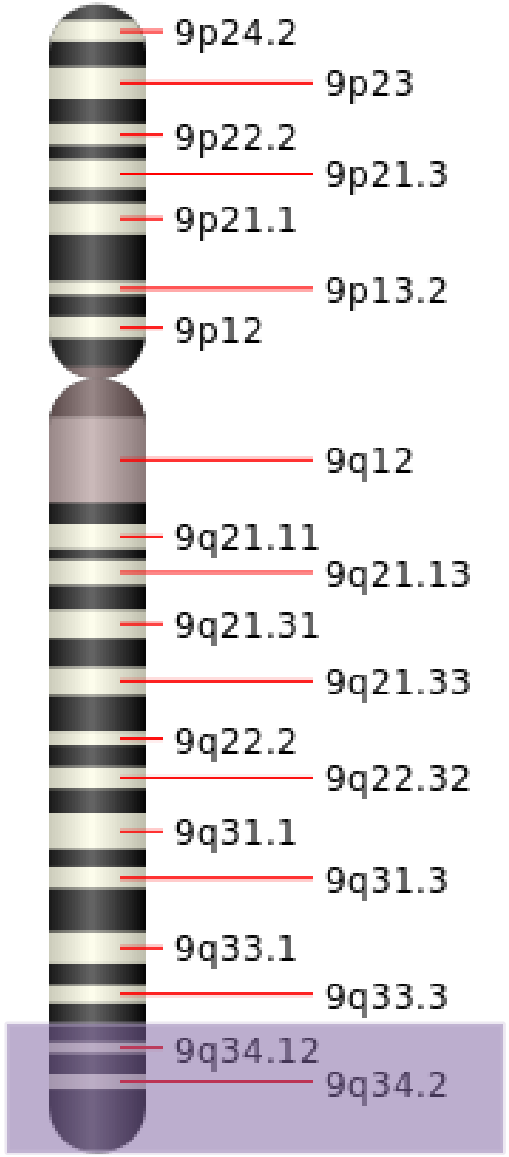
Note, however, that in the ABO blood group system I^A and I^B are codominant, but both are dominant over I^o. (Note that the superscript 'o' is lowercase to denote its recessive character).

Human ABO Blood Group

- All humans belong to one of four inherited blood groups : A, B, AB and O.

Human ABO Blood Group

- The ABO blood group in humans is determined by multiple alleles of a single gene
- This gene is found on chromosome 9
- This gene encodes an **enzyme** that **adds sugar molecules to lipids** on the surface of red blood cells

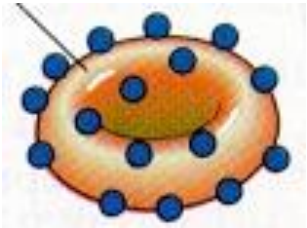


9q34.1-q34.2

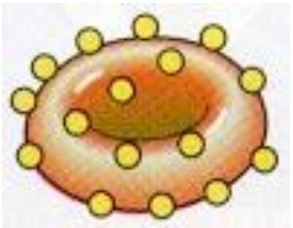
Human ABO Blood Group

- The letters A and B refer to the kind of antigen found on an individual's red blood cells.
  E.g. blood group type A is due to the presence of only type A antigen on red blood cells

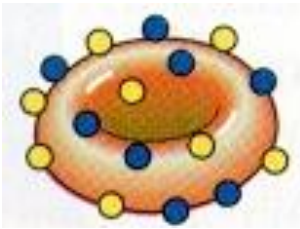
Type A



Type B



Type AB



Type O

Human ABO Blood Group

- The A and B antigens are actually carbohydrate groups (sugars) that are bound to lipid molecules (fatty acids) protruding from the membrane of the red blood cell

Human ABO Blood Group

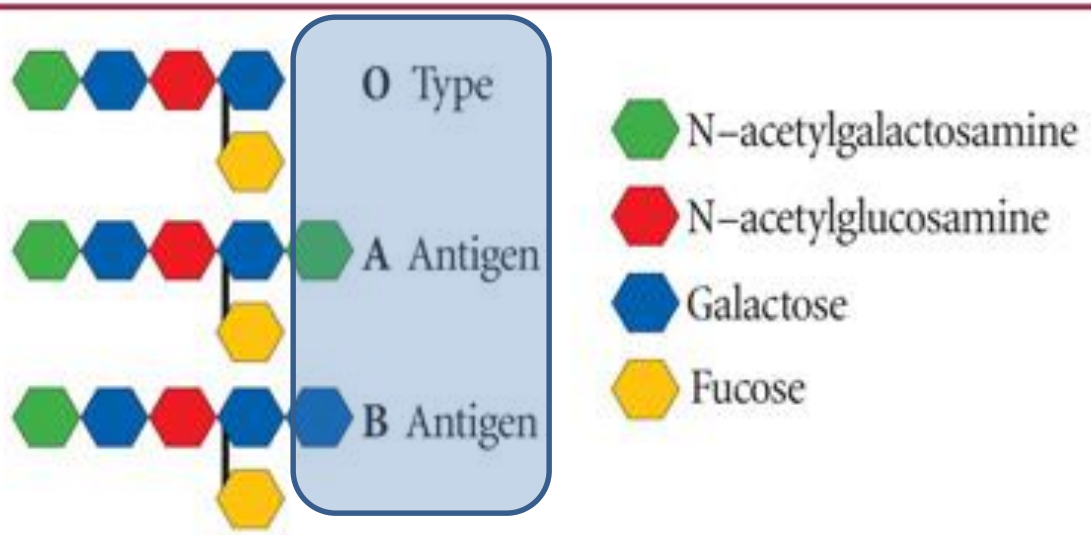


Figure 1. ABO antigen specificity. The ABO antigens differ by just one sugar at the antigen terminus. Only the carbohydrate portion of the antigen is illustrated.

Human ABO Blood Group

- The specificity of the A and B antigens is based on the terminal sugar of the carbohydrate group.
- These sugars act as recognition markers for the immune system.

Antigens = molecules recognized by antibodies of the immune system

Human ABO Blood Group

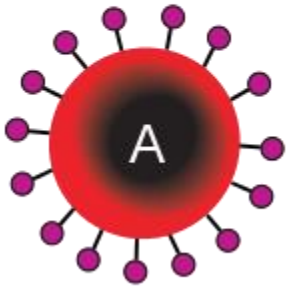
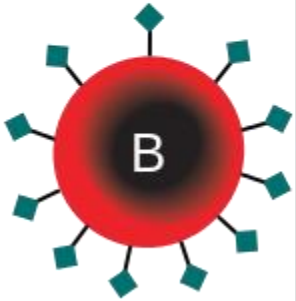
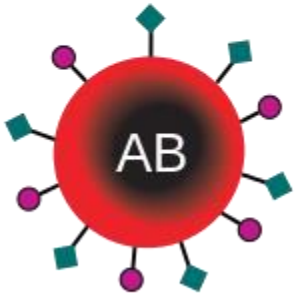
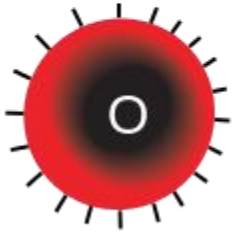
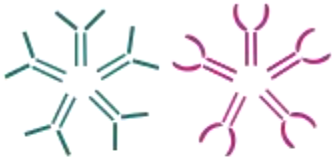
- The gene coding for these surface antigens has three alleles :
  - I^A : allele for the production of type A antigen
  - I^B : allele for the production of type B antigen
  - I^O : allele that produces neither antigen

Human ABO Blood Group

- Alelles I^A and I^B are codominant, but each is dominant to I^O.
- **Only two** of the three alleles can be present in a diploid individual.
- For blood group AB, both type A and B antigens are present on the red blood cell membrane.

Human ABO Blood Group

Pg 27

	I^AI^A OR I^Ai^o	I^BI^B OR I^Bi^o	I^AI^B	i^oi^o
	Group A	Group B	Group AB	Group O
Red blood cell type				
Antibodies in Plasma	 Anti-B	 Anti-A	None	 Anti-A and Anti-B
Antigens in Red Blood Cell	 A antigen	 B antigen	 A and B antigens	None

Example [Pg.28]

0:30

- Consider the phenotypes that can result when a couple with the genotypes I^AI^O and I^BI^O have children.

	Female	Male
Parental Phenotypes:	Blood group A	Blood group B
Parental Genotypes:		

Gametes:

Offspring Genotypes:

Offspring Phenotypes: Group AB Group A Group B Group O

Example [Pg.28]

- Consider the phenotypes that can result when a couple with the genotypes I^AI^O and I^BI^O have children.

	Female		Male
Parental Phenotypes:	Blood group A		Blood group B
Parental Genotypes:	I^AI^O	X	I^BI^O

Gametes: I^A I^O X I^B I^O

Offspring Genotypes:	I^AI^B	I^AI^O	I^BI^O	I^OI^O
Offspring Phenotypes:	Group AB	Group A	Group B	Group O

- Probability of a child with blood group A

EXTENSION

30s

Example [Pg.28]

- Consider the phenotypes that can result when a couple with the genotypes I^AI^O and I^BI^O have children.

	Female		Male
Parental Phenotypes:	Blood group A		Blood group B
Parental Genotypes:	I^AI^O	X	I^BI^O

Gametes: I^A I^O X I^B I^O

Offspring Genotypes:	I^AI^B	I^AI^O	I^BI^O	I^OI^O
Offspring Phenotypes:	Group AB	Group A	Group B	Group O

Blank space on Pg 28

- Probability of a child with blood group A

  = ¼

EXTENSION

30s

Example [Pg.28]

- Consider the phenotypes that can result when a couple with the genotypes I^AI^O and I^BI^O have children.

	Female		Male
Parental Phenotypes:	Blood group A		Blood group B
Parental Genotypes:	I^AI^O	X	I^BI^O

Gametes: I^A I^O X I^B I^O

Offspring Genotypes:	I^AI^B	I^AI^O	I^BI^O	I^OI^O
Offspring Phenotypes:	Group AB	Group A	Group B	Group O

Blank space on Pg 28

EXTENSION

- Probability of having a son with blood group A

Cannot deduce gender from the cross

30s

Example [Pg.28]

- Consider the phenotypes that can result when a couple with the genotypes I^AI^O and I^BI^O have children.

	Female		Male
Parental Phenotypes:	Blood group A		Blood group B
Parental Genotypes:	I^AI^O	X	I^BI^O
Gametes	I^A I^O	X	I^B I^O

Offspring Genotypes:	I^AI^B	I^AI^O	I^BI^O	I^OI^O
Offspring Phenotypes:	Group AB	Group A	Group B	Group O

Blank space on Pg 28

EXTENSION

- Probability of having a son with blood group A

  = ¼ X ½ = 1/8

1 out of 2 possibilities of getting a boy

30s

Example [Pg.28]

- Consider the phenotypes that can result when a couple with the genotypes I^AI^O and I^BI^O have children.

	Female		Male
Parental Phenotypes:	Blood group A		Blood group B
Parental Genotypes:	I^AI^O	X	I^BI^O

Gametes: (I^A) (I^O) X (I^B) (I^O)

Offspring Genotypes:	I^AI^B	I^AI^O	I^BI^O	I^OI^O
Offspring Phenotypes:	Group AB	Group A	Group B	Group O

Blank space on Pg 28

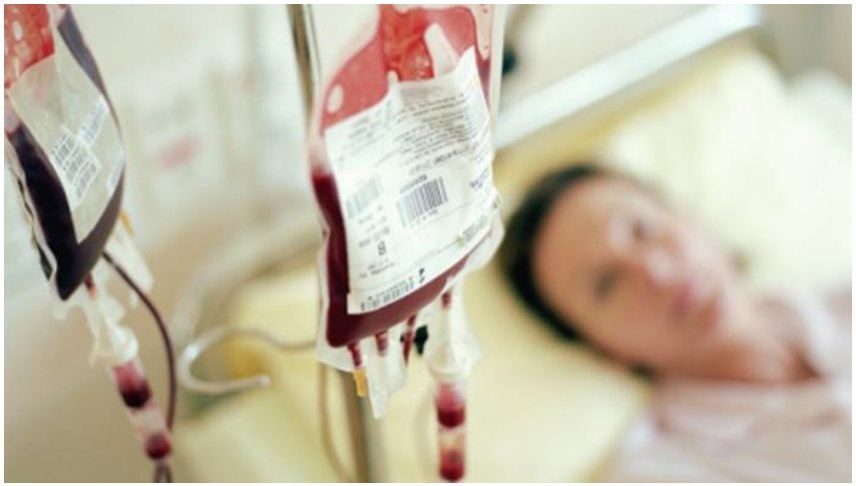
Genotypes and phenotypes

Genotype	Phenotypes
I^AI^A , I^AI^O	Blood group A (Antigen A, anti-B serum antibodies)
I^BI^B , I^BI^O	Blood group B (Antigen B, anti-A serum antibodies)
I^AI^B	Blood group AB (Antigen A and antigen B, no anti-A or anti-B serum antibodies)
I^OI^O	Blood group O (No antigens, anti-A and anti-B serum antibodies)

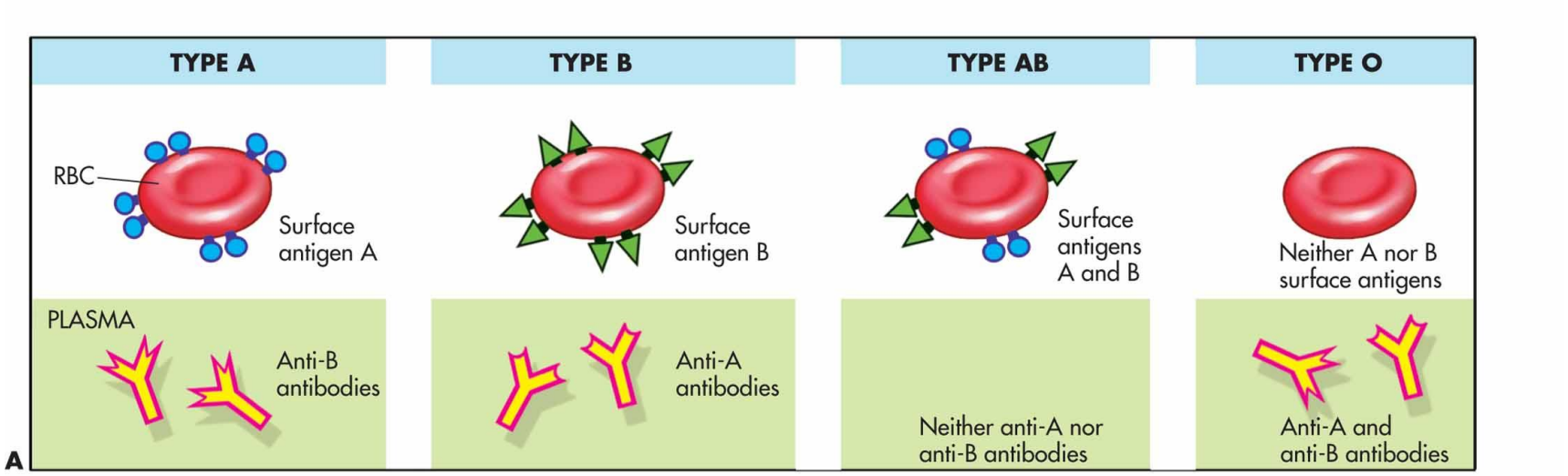
A body should not make antibodies which recognise the antigen types present

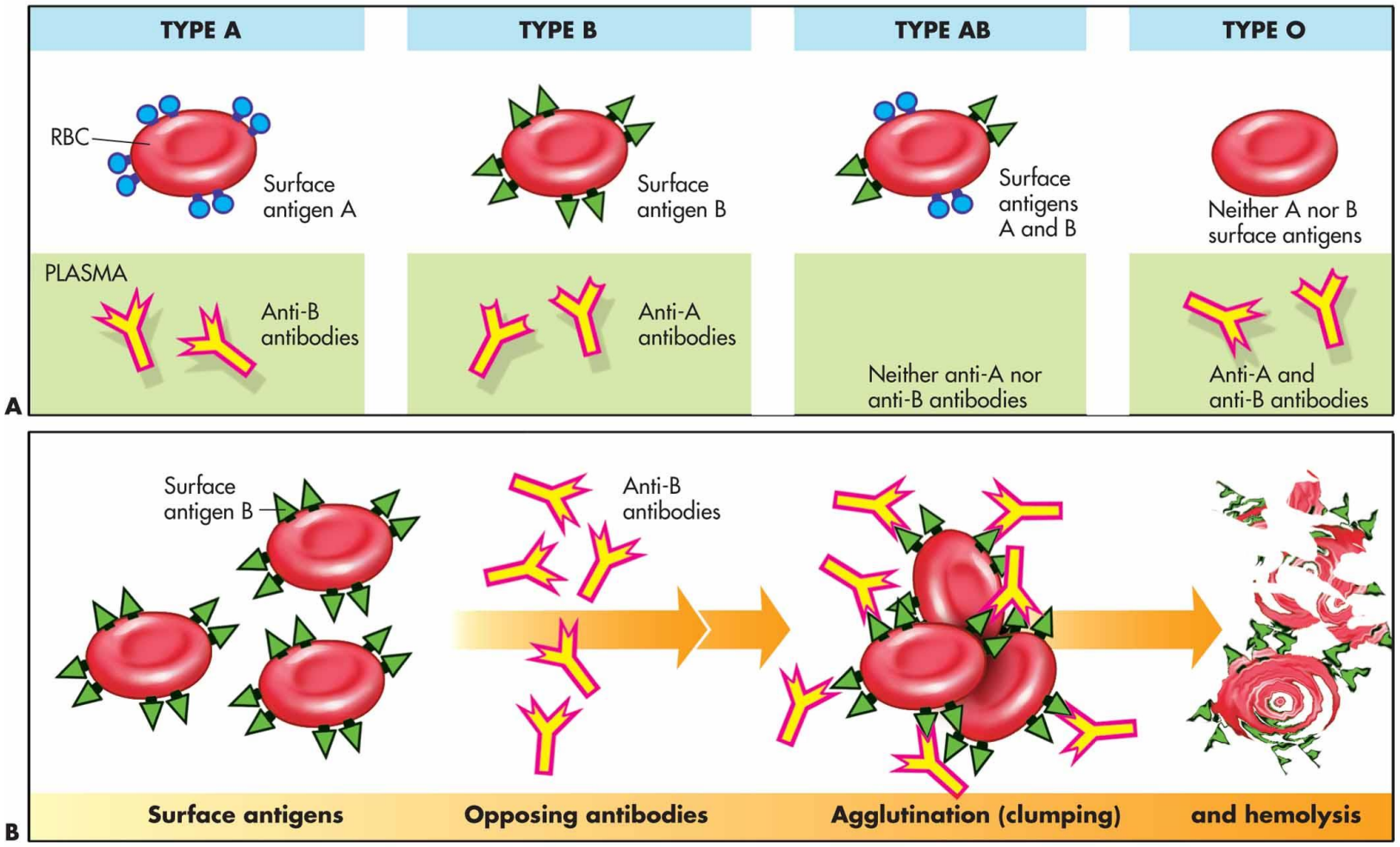
What happens if antibody A meets antigen A during mistakes in blood transfusion??

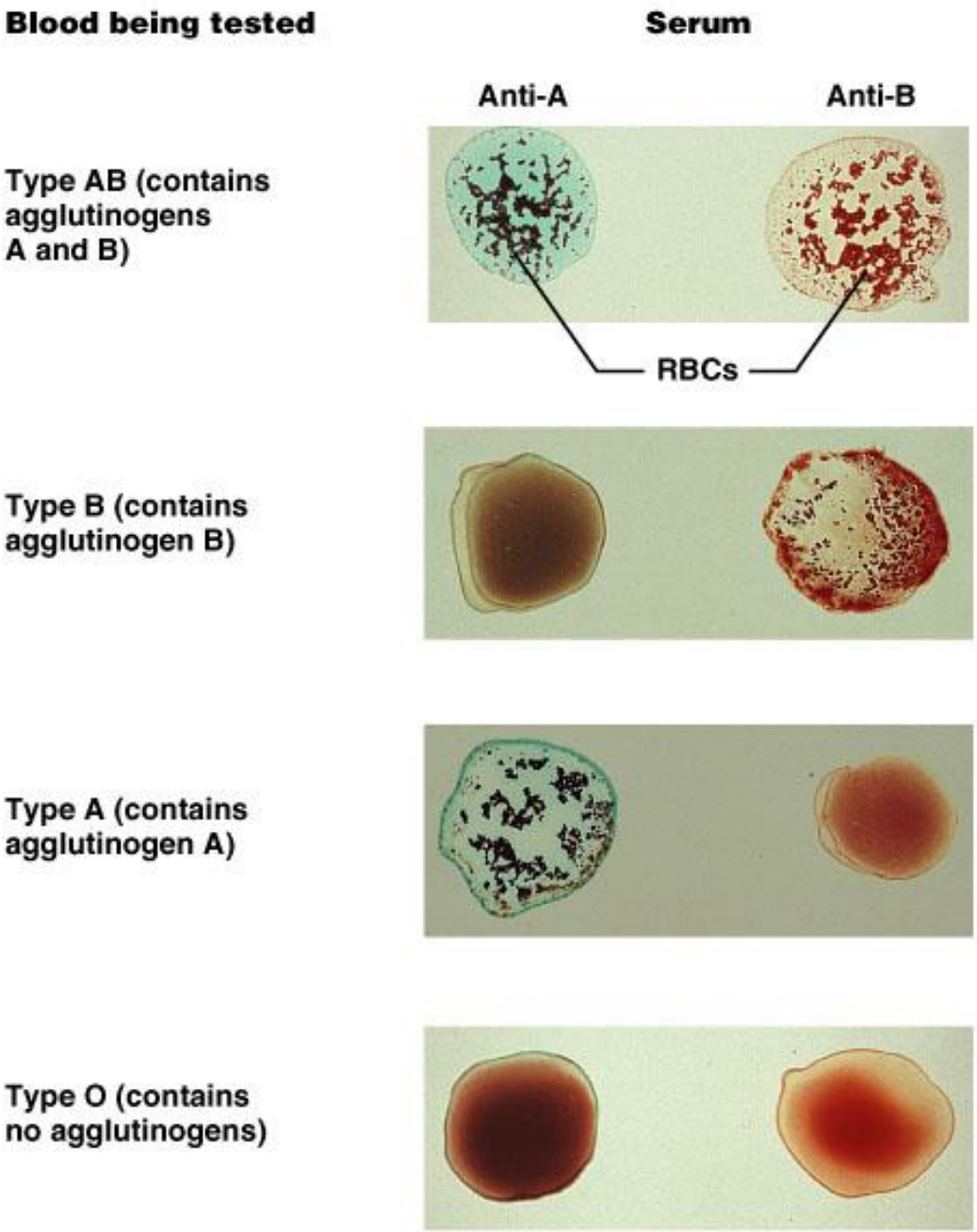
Agglutination occurs
➔ fatal



Not in notes


TYPE A
TYPE B
TYPE AB
TYPE O
RBC
Surface antigen A
Surface antigen B
Surface antigens A and B
Neither A nor B surface antigens
PLASMA
Anti-B antibodies
Anti-A antibodies
Neither anti-A nor anti-B antibodies
Anti-A and anti-B antibodies
A


TYPE A
TYPE B
TYPE AB
TYPE O
RBC
Surface antigen A
Surface antigen B
Surface antigens A and B
Neither A nor B surface antigens
PLASMA
Anti-B antibodies
Anti-A antibodies
Neither anti-A nor anti-B antibodies
Anti-A and anti-B antibodies
A
Surface antigen B
Anti-B antibodies
Surface antigens
Opposing antibodies
Agglutination (clumping)
and hemolysis
B

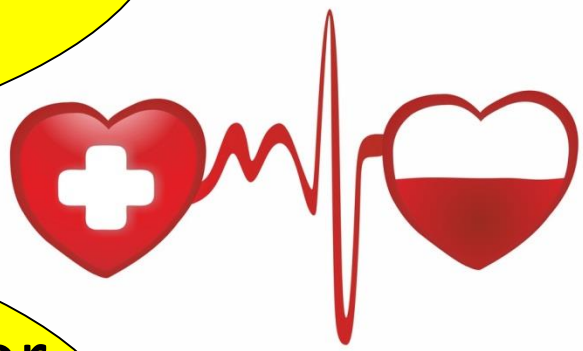


Importance of Human Blood Groups

Not in notes

BLOOD AB is the universal Acceptor (both antigens A and B; no antibodies)

Can accept all blood groups. Can only donate to ppl with AB blood group



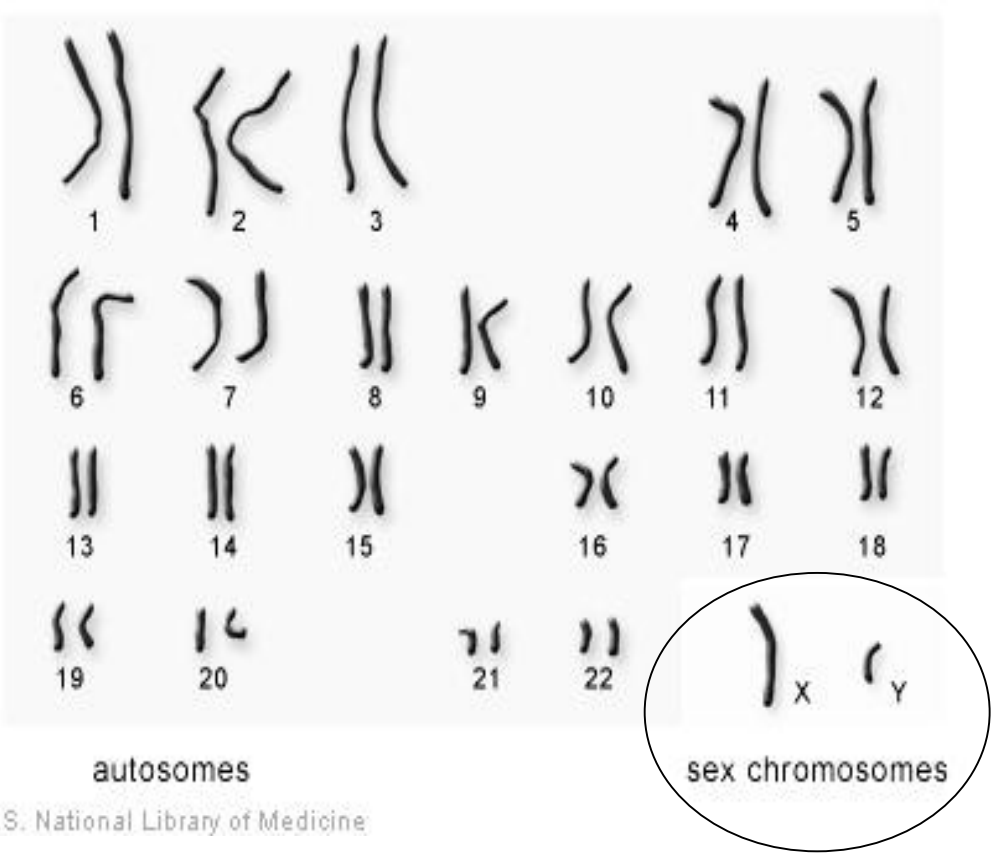
BLOOD O is the universal donor (no antigens; both antibodies Against antigens A and B)

Can donate to ppl with all blood groups! Can accept only blood group O.

Exceptions

- Incomplete dominance and codominance
- Multiple alleles
- Sex-linked genes

Sex determination in Humans

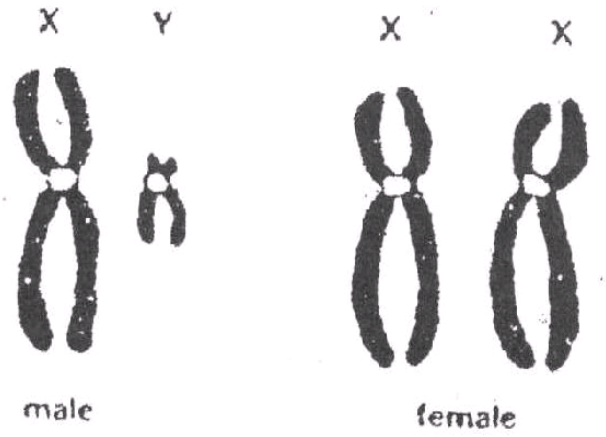


Sex determination in Humans

- There are 23 pairs of chromosomes in humans.
- Of these, the first 22 pairs appear identical in both the male and the female. These chromosomes are known as autosomes or autosomal chromosomes, and are not involved in sex determination.
- The 23rd pair is the sex chromosome and carries genes that control sexual development.

Sex determination in Humans

- In a female, the two sex chromosomes appear identical and are called X chromosomes
- In the male, there is one X chromosome and one Y chromosome. The Y chromosome is shorter than the X chromosome and carries very few genes as compared to the X chromosomes.



Sex determination in Humans

- In humans, females produce only one type of gamete.
- All the egg cells carry an X chromosome.
- Hence the female is described as the homogametic sex.

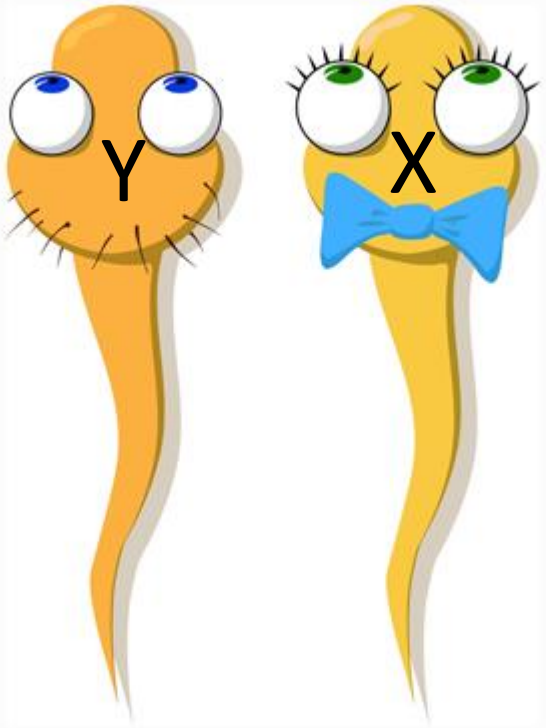
Sex determination in Humans

- In humans, males produce only two types of gamete.
- Half of the sperm cells carry an X chromosome while the other half carries a Y chromosome
- Hence the male is described as the heterogametic sex.

So, is it the father or mother who determines our gender???

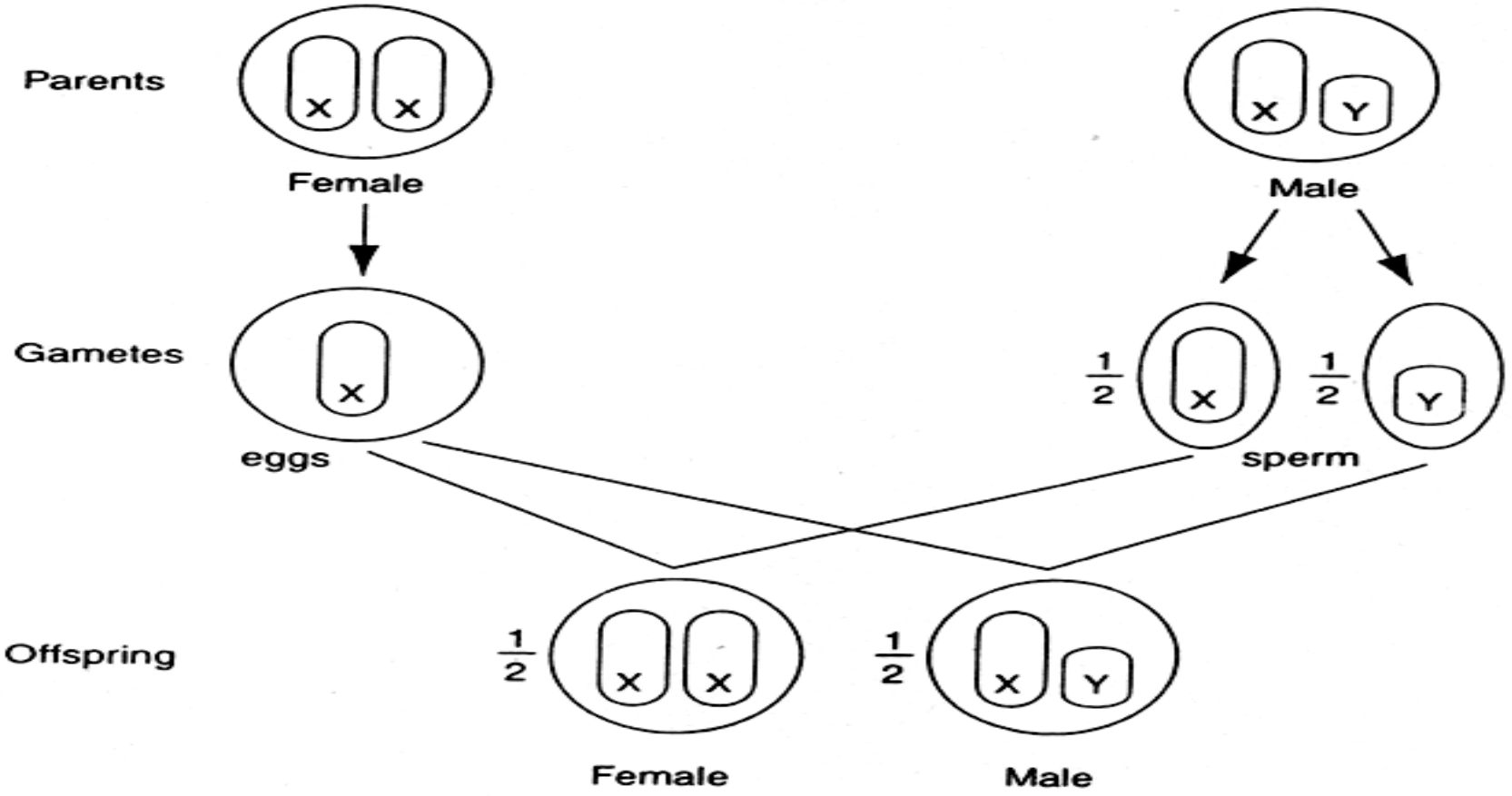
Sex determination in Humans

- The sex of an offspring is determined by the



SPERM that fertilises the egg cell.

Sex determination in Humans



Probability of a girl or boy = ½

Sex determination in Humans

X or Y

- Sex-linked genes : **genes located on the sex chromosomes** that result in a characteristic being expressed mainly in one sex. A trait determined by a gene on the sex chromosome is said to be said to be sex-linked.
- A trait determined by a gene on the X-chromosome is said to be X-linked.

X only

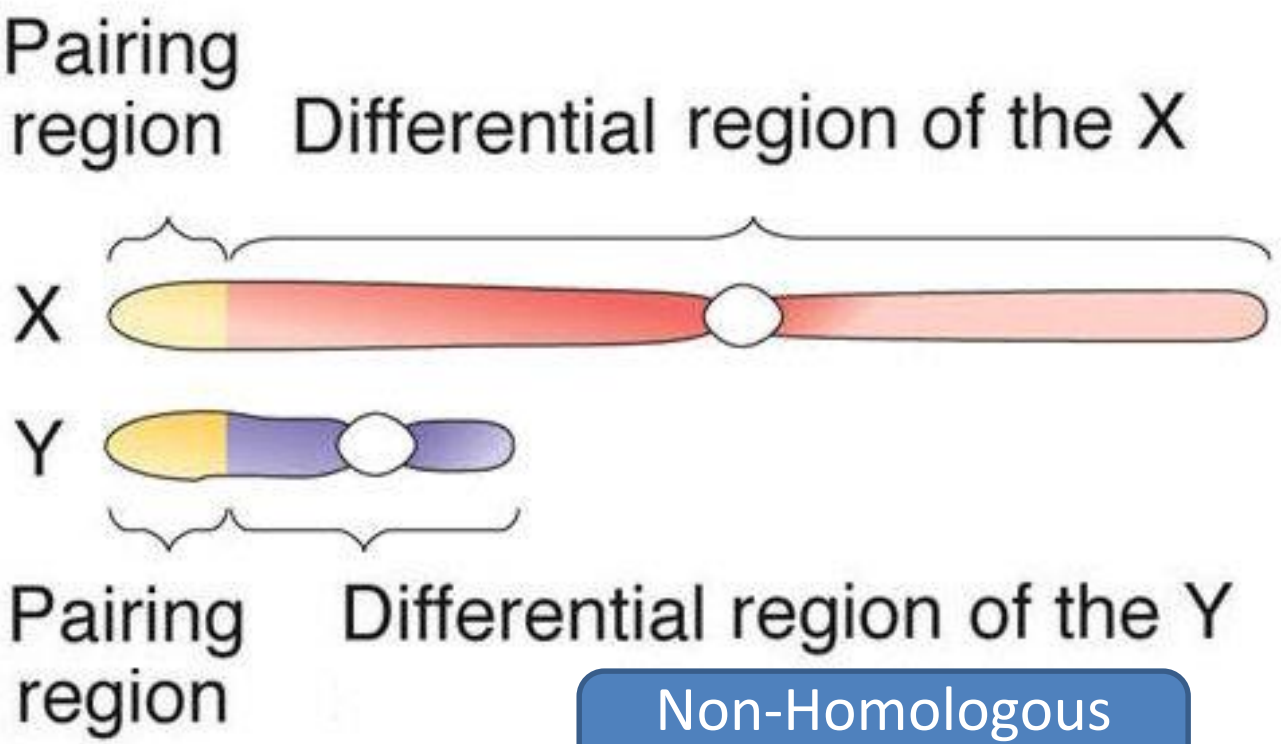
[Do not confuse sex-linked genes with (autosomal) linked genes. Linked genes: Genes coding for different characteristics but are situated on the same chromosome; they tend to be inherited together in genetic crosses].

Sex determination in Humans

- X chromosomes carry genes controlling sexual development as well as other characteristics such as colour vision.
- Y chromosomes carry genes controlling the development of male characteristics but few other genes.

Long ear hair

Sex determination in Humans

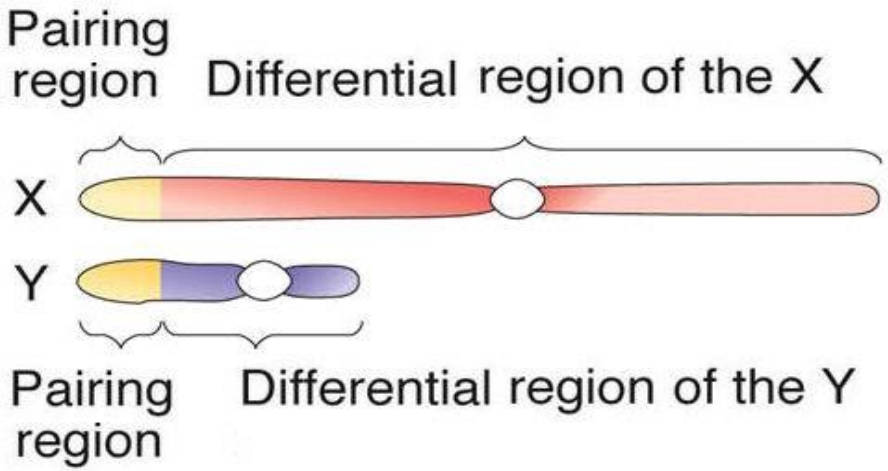


Homologous regions

Non-Homologous regions

Sex determination in Humans

- Short arm of Y chromosome carries a male determining gene, the SRY (sex determining region of the Y)
- The Y-linked SRY gene encodes for a protein that switches on other genes that direct the development of male structures in the embryo

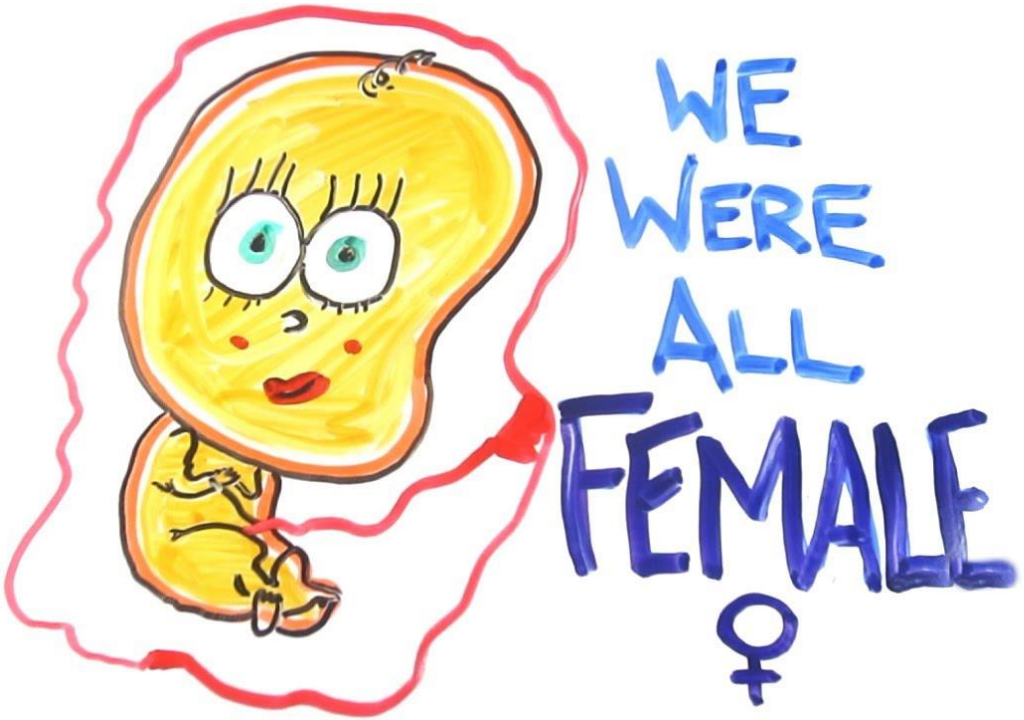


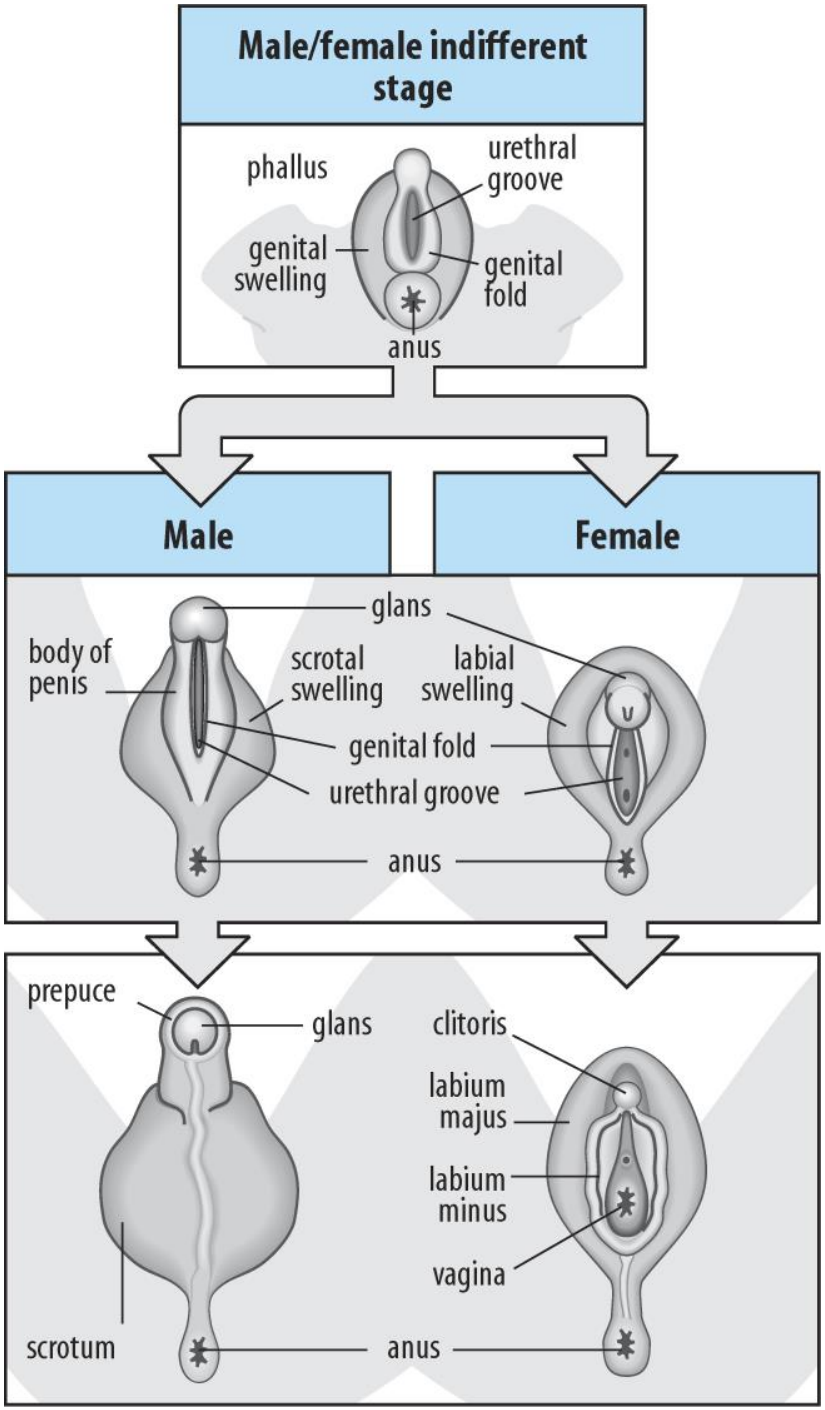
Sex determination in Humans

- In the absence of the product encoded by SRY, as in females, undifferentiated gonads develop into ovaries

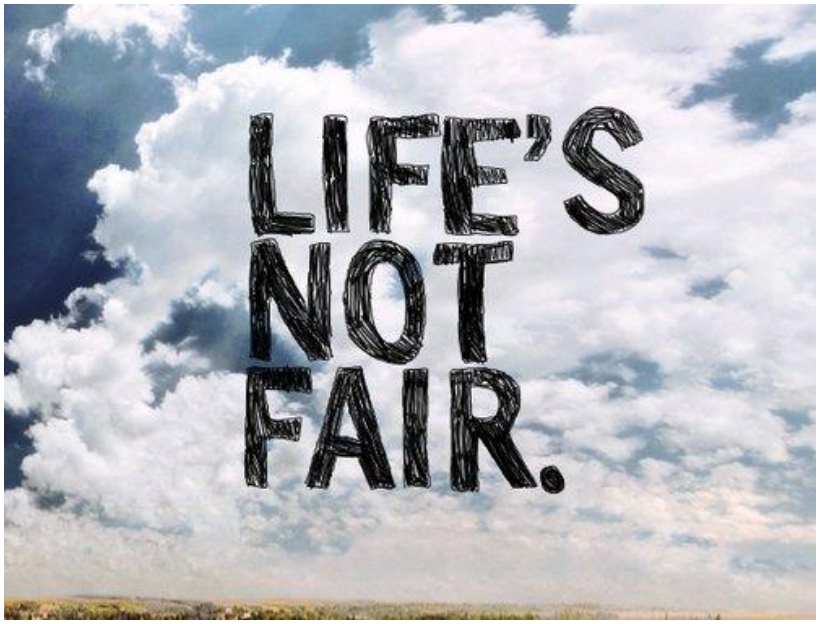
All embryos start off as FEMALE in the first few months of development

SRY gene is only expressed later in development

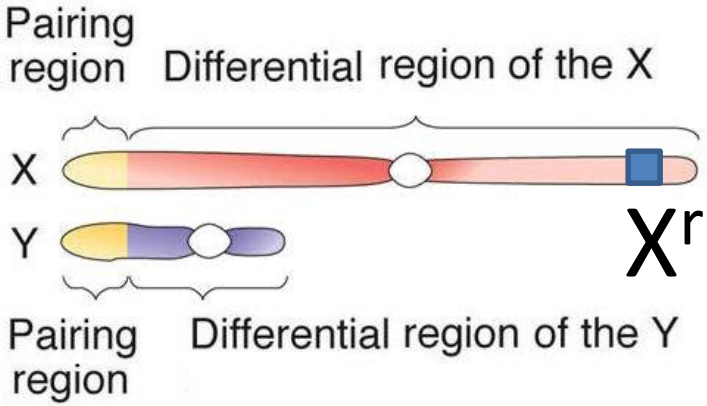



Male/female indifferent stage
phallus
urethral groove
genital swelling
genital fold
anus
Male
Female
glans
body of penis
scrotal swelling
labial swelling
genital fold
urethral groove
anus
prepuce
glans
clitoris
labium majus
labium minus
vagina
scrotum
anus

Explain why sex-linked recessive diseases are more common in human males



Sex determination in Humans

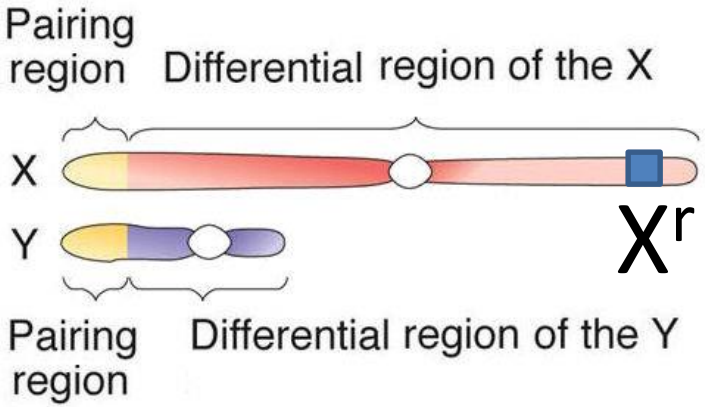


X^r → recessive allele that code for disease

- Recessive alleles occurring on the longer arm of the X chromosome are expressed phenotypically in males since there are no corresponding dominant alleles in the Y chromosome to mask the recessive alleles.

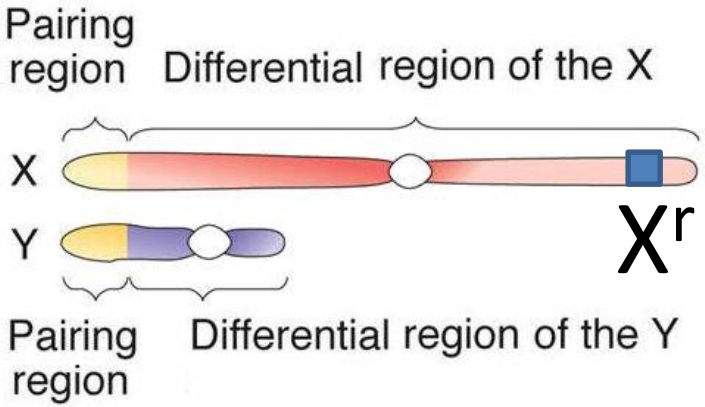
By coding for functional protein

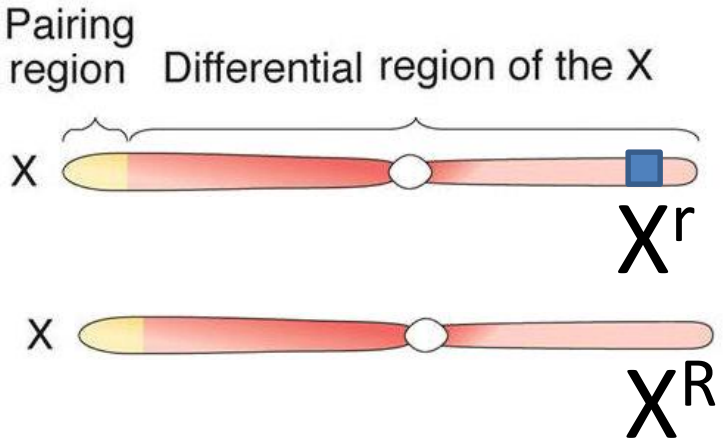
Sex determination in Humans



- Males have a higher tendency to inherit ~~sex~~X-linked diseases as they only have one X chromosome, whereas females have two X chromosomes.

Sex determination in Humans





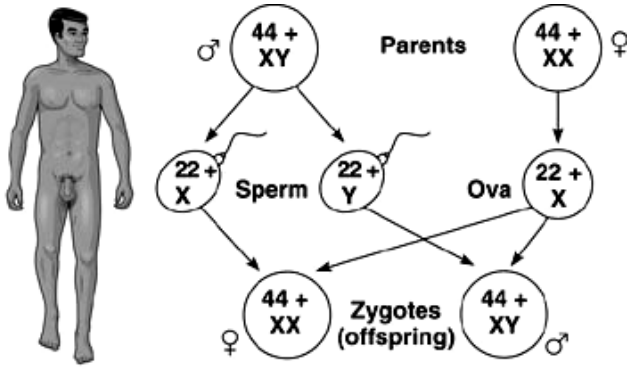
- In females, the other X chromosome may carry dominant alleles which then mask the expression of these recessive alleles.

Sex determination in other animals

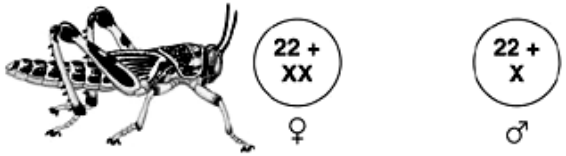
- In Drosophila, the female is XX, the male is XY.
- The male is not always the heterogametic sex. Example in birds (e.g. chickens), the male is the homogametic sex (ZZ) and the female is the heterogametic sex (ZW).

REMEMBER

Sex determination in other animals



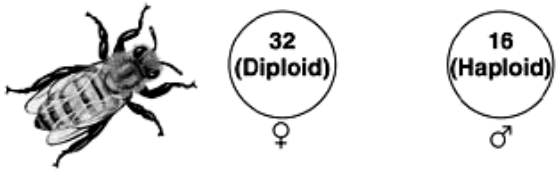
(a) The X-Y system



(b) The X-0 system

Popular trap in MCQs

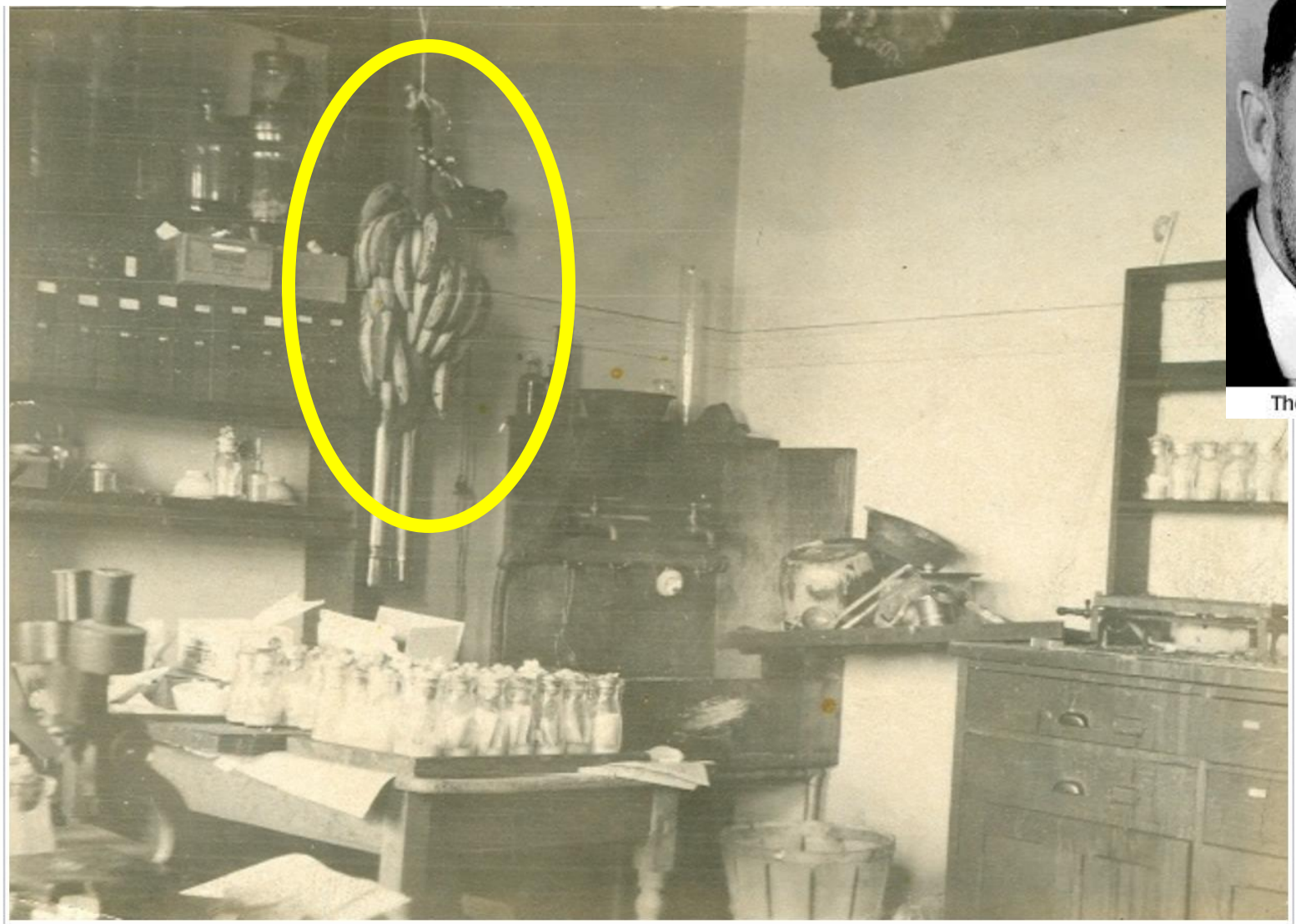
(c) The Z-W system

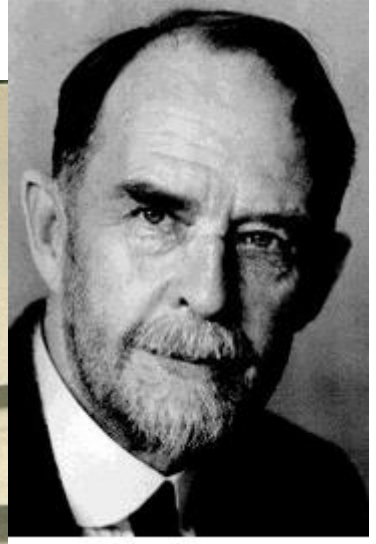


(d) The haplo-diploid system

Example of X-linked genes

(1) Eye colour in Drosophila



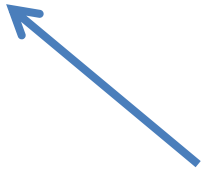


Thomas Hunt Morgan

Figure 1: Columbia University Fly Room.
This faded black and white photograph shows the Fly Room at Columbia University with bunches of bananas featured prominently.

(1) Eye colour in Drosophila

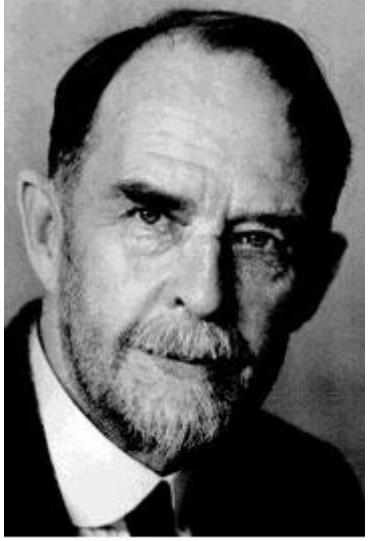
- The allele for red eye colour is dominant to that for white eye colour.
- Red eye colour is considered to be the normal eye colour (i.e. the "wild type").



Phenotype which is prevalent among individuals under natural conditions

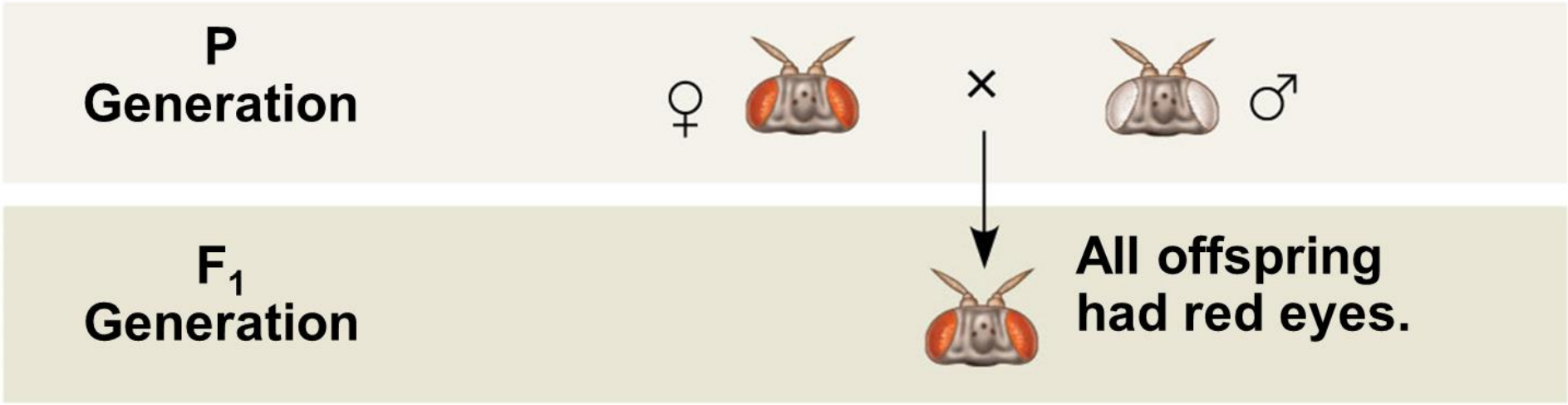
Pg 32

Morgan's Experiment


Thomas Hunt Morgan

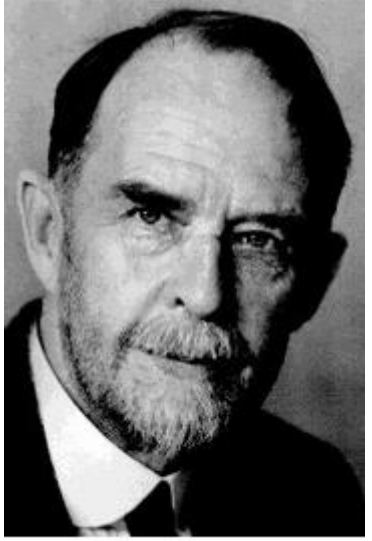
- When a white-eyed male fly is mated with a red-eye female fly, all the F1 offspring have red eyes, suggesting that the wild-type (red eye) allele is dominant.

EXPERIMENT



Pg 32

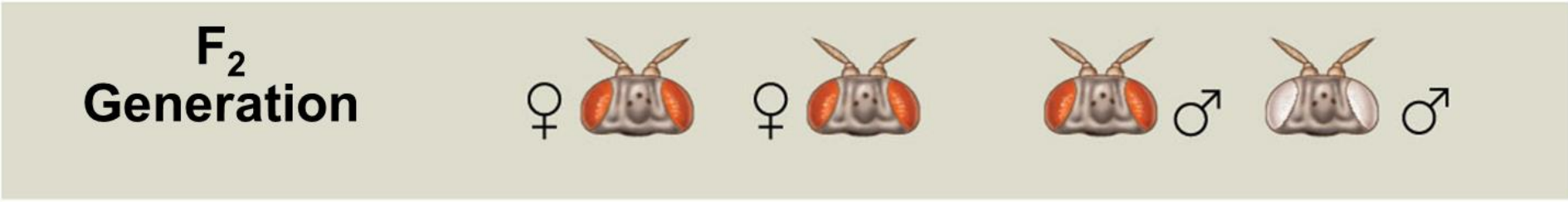
Morgan's Experiment



Thomas Hunt Morgan

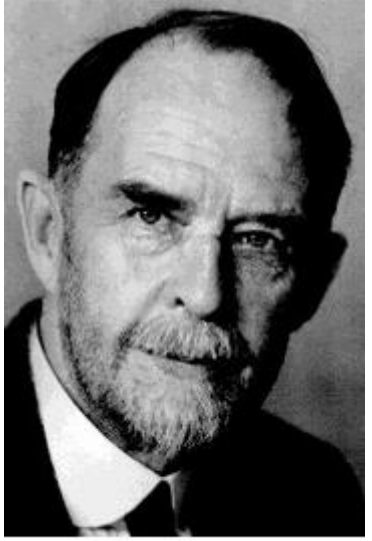
- When the F1 generation are bred to each other, the classic 3:1 phenotypic ratio among the F2 offspring is observed

RESULTS



Pg 32

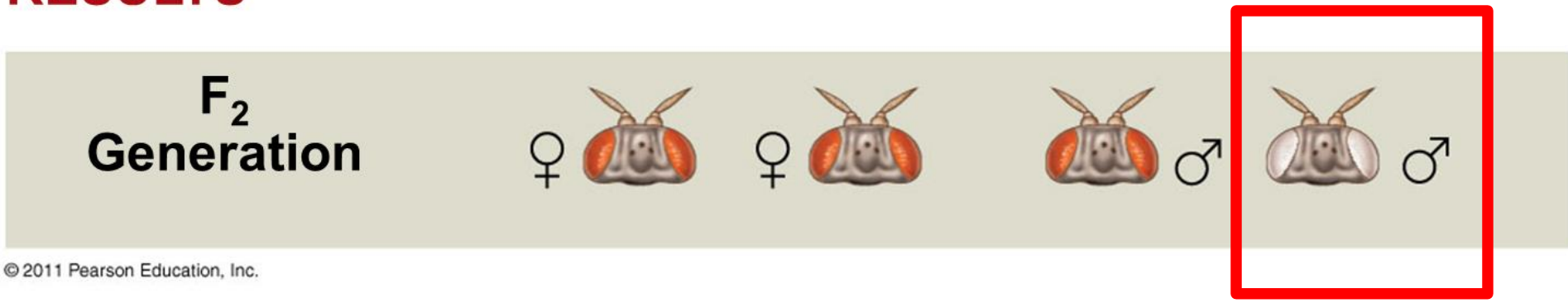
Morgan's Experiment



Thomas Hunt Morgan

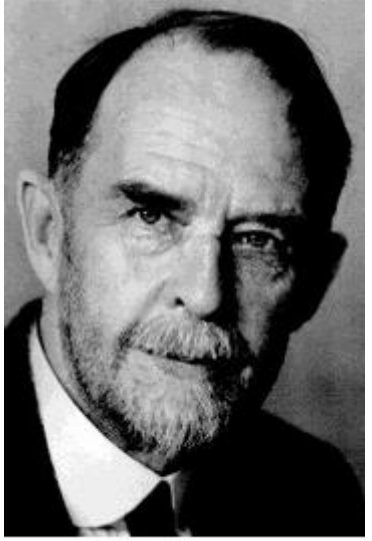
- However, the white-eyed trait shows up only in males.
- All the F2 females have red eyes while half the males have red eyes and the other half have white eyes.

RESULTS



Pg 32

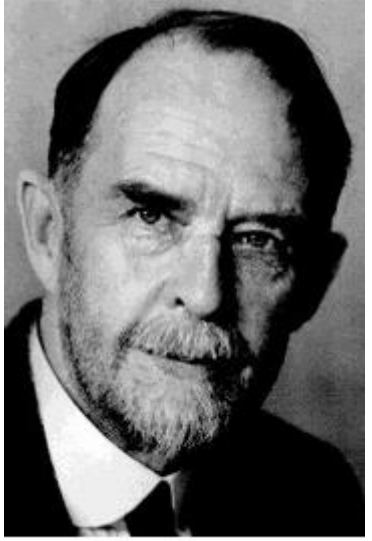
Morgan's Experiment


Thomas Hunt Morgan

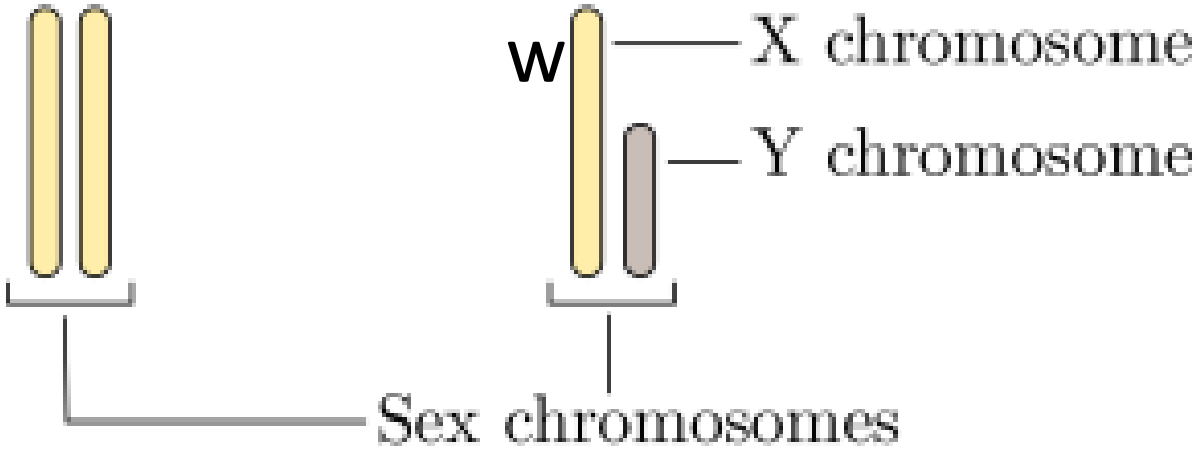
- Conclusion :
  A fly's eye colour is linked to its sex ; the allele for eye colour is located on the X chromosome, with no corresponding allele present on the Y chromosome

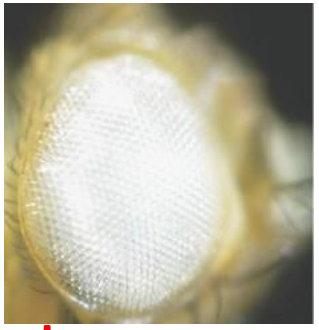
Pg 32

Morgan's Experiment


Thomas Hunt Morgan

- Conclusion :

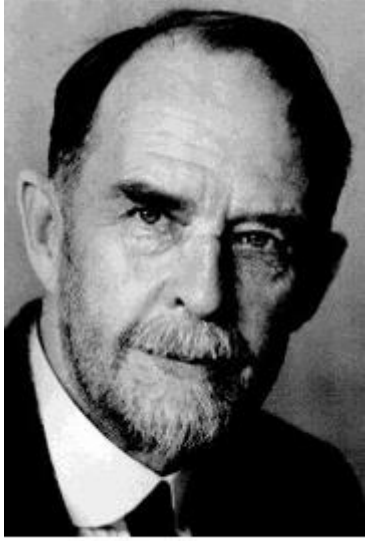




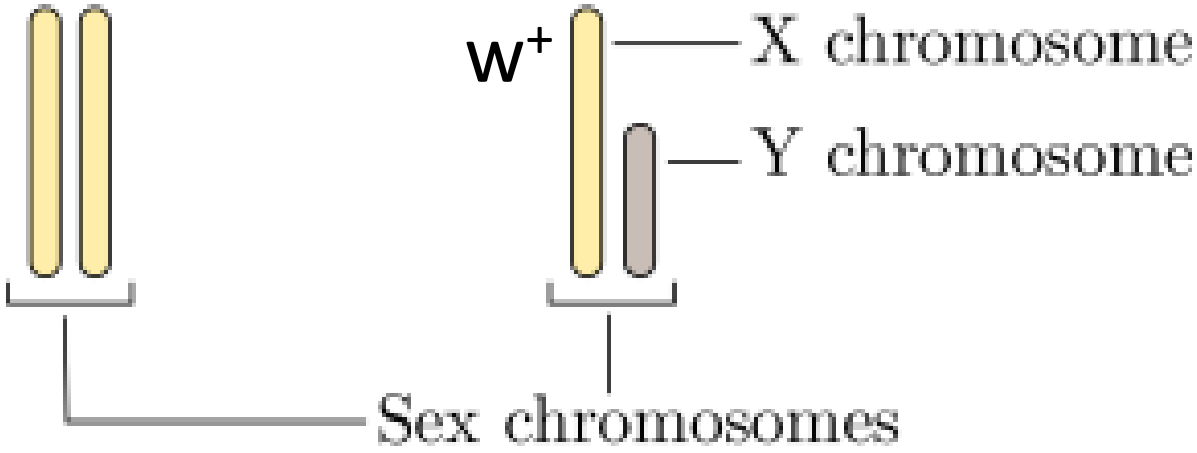
For a male, a single copy of the mutant allele would confer white eyes since a male has only one X chromosome, there can be no wild-type allele present (on the Y chromosome) to mask the effect of the recessive allele.

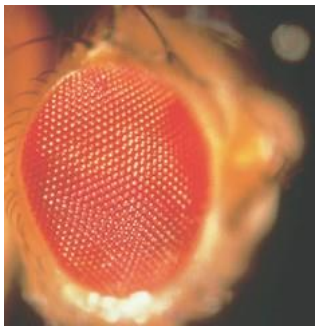
Pg 32

Morgan's Experiment


Thomas Hunt Morgan

- Conclusion :



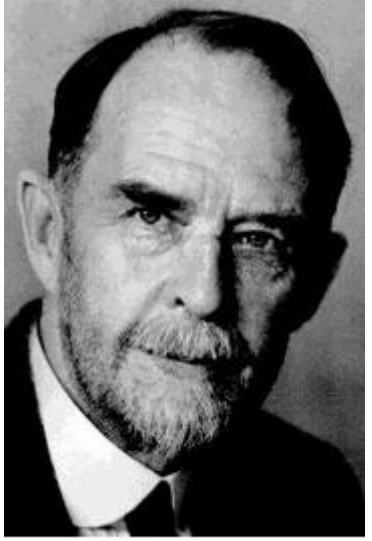


For a male, a single copy of the wild-type allele would confer red eyes since a male has only one X chromosome.

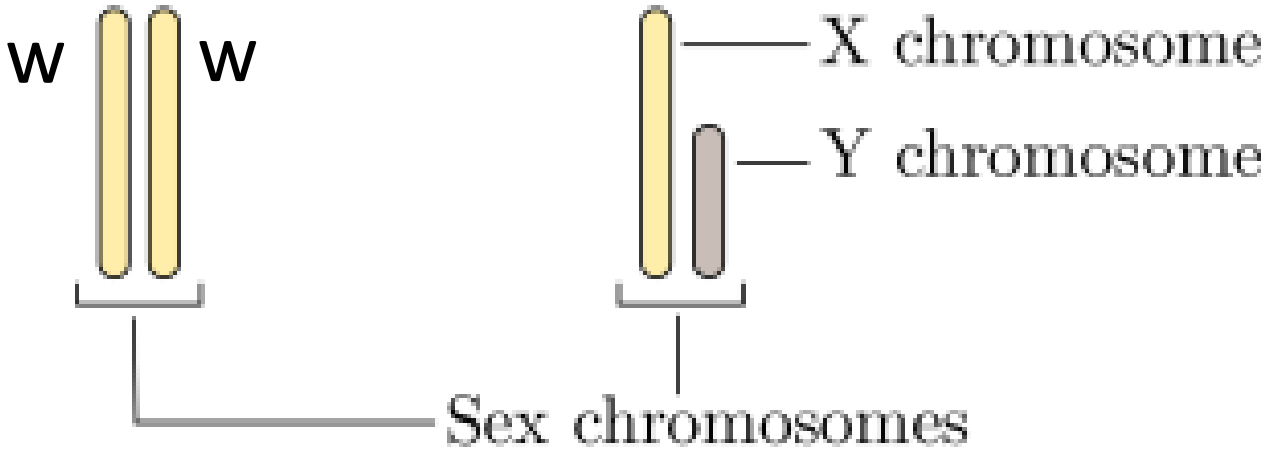
Can females have white eyes too???

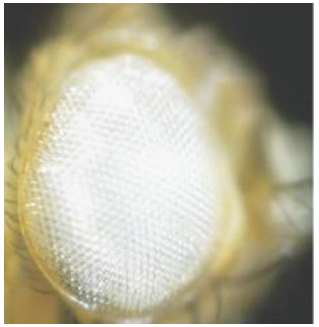
Pg 33

Morgan's Experiment


Thomas Hunt Morgan

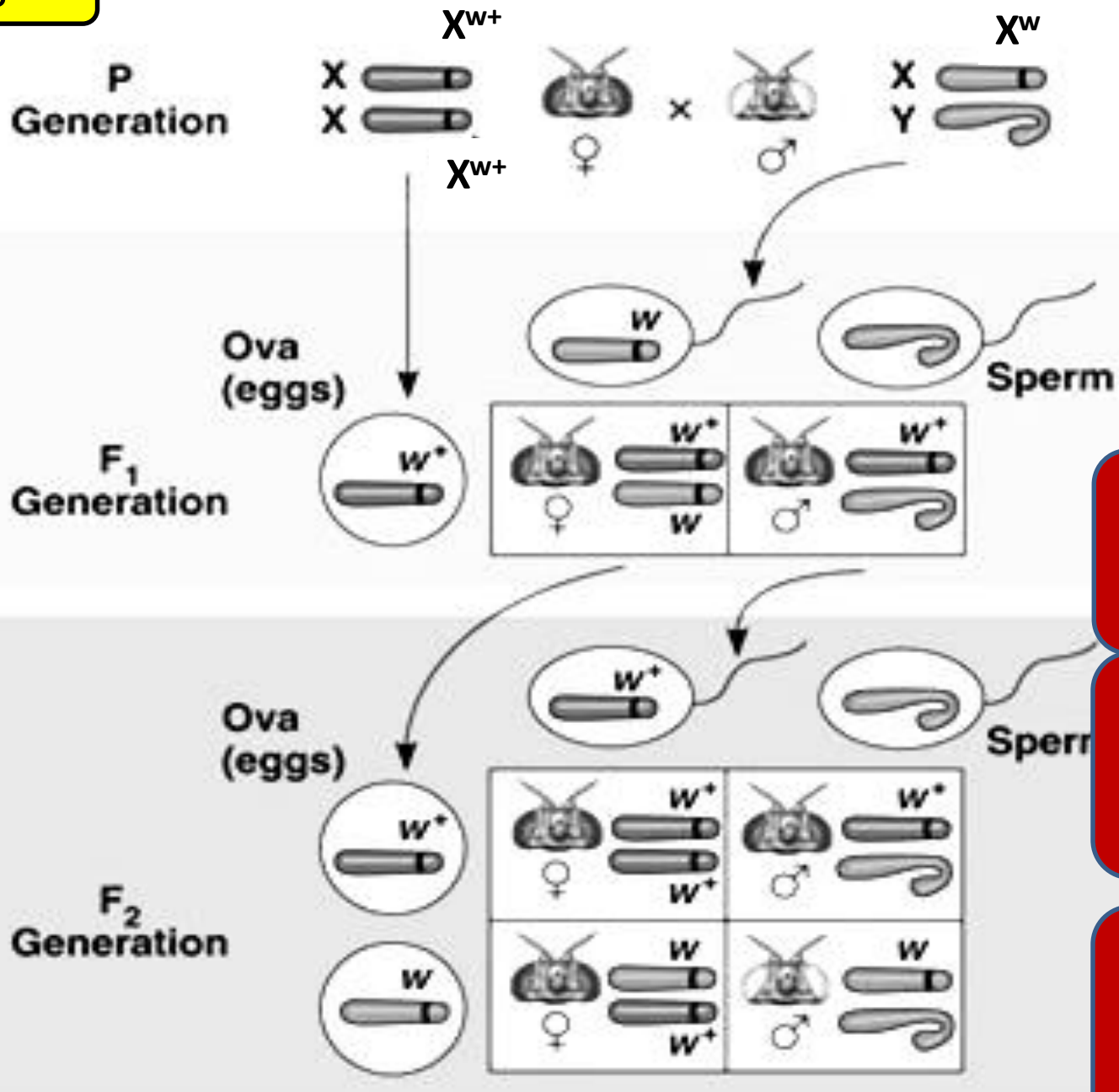
- Conclusion :





A female could have white eyes only if both her X chromosomes carried the recessive mutant allele (i.e. the white eye allele).

Pg 33



Key :

X^{w+}

X^{w}

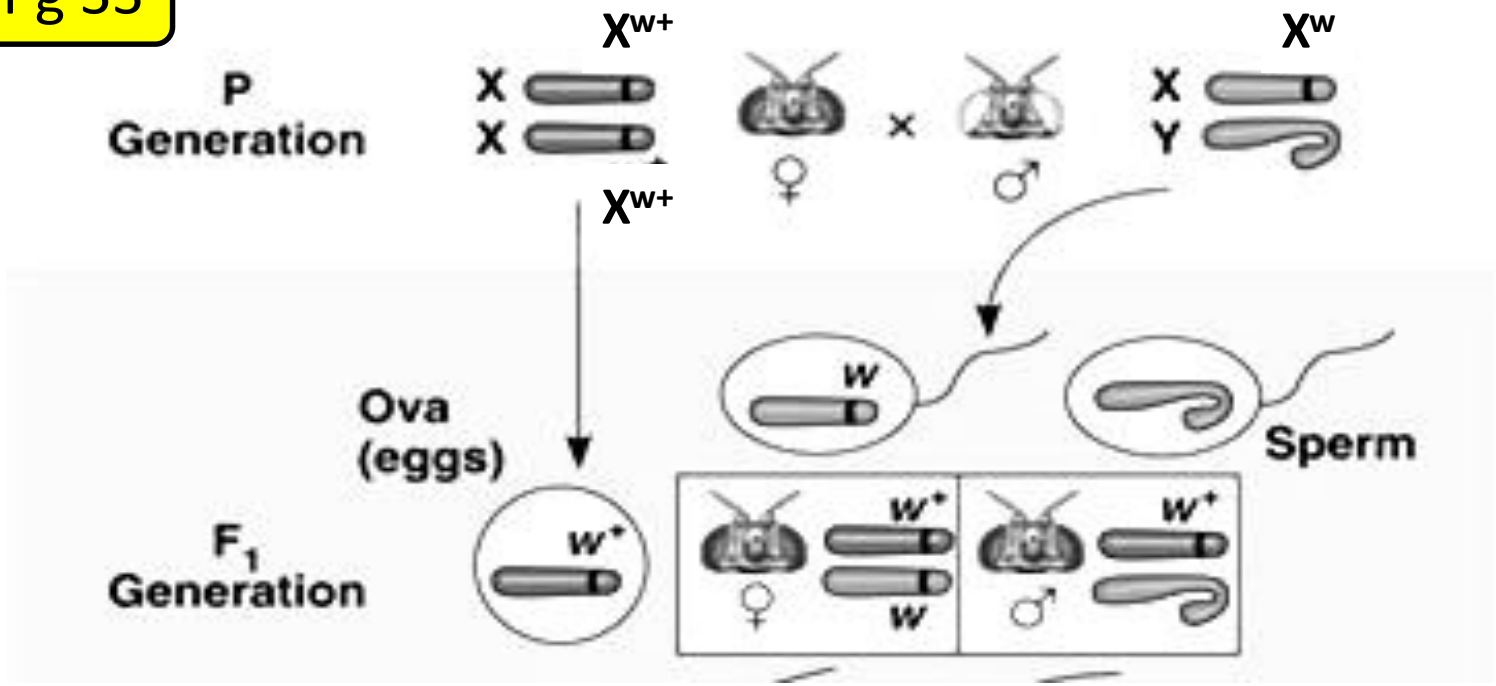
Y

Super script letter on the X chromosome

Unless instructed, avoid using W+ vs W

Use Capital letter vs small letter eg A vs a

Pg 33



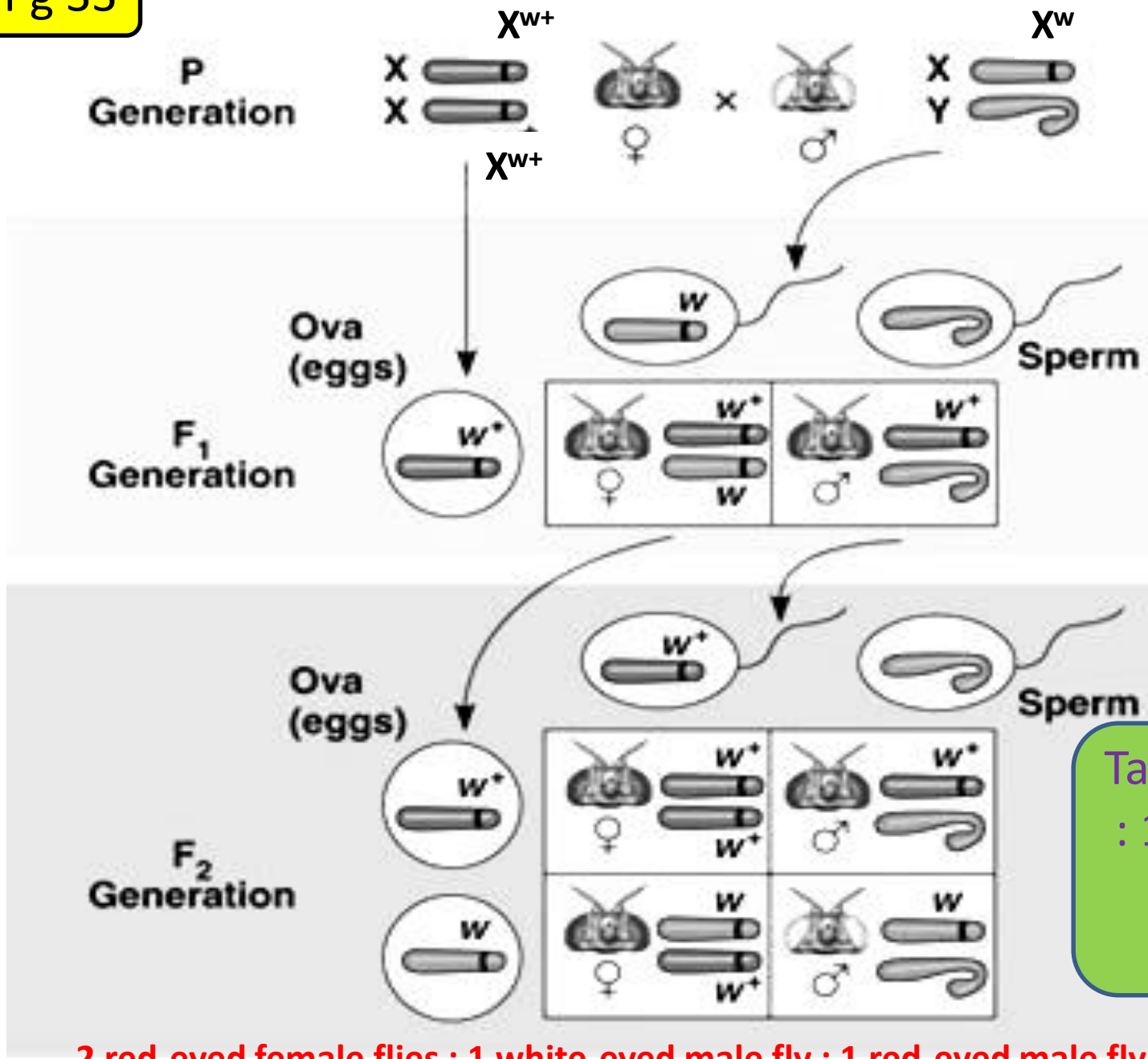
Key :

X^{w+}

X^{w}

Y

Pg 33



Key :

X^{w+}

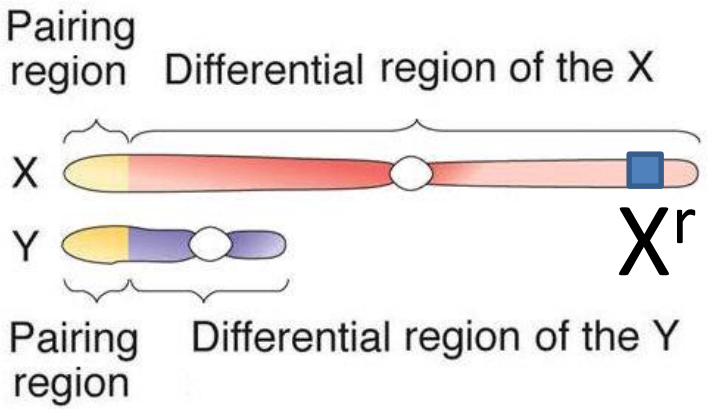
X^{w}

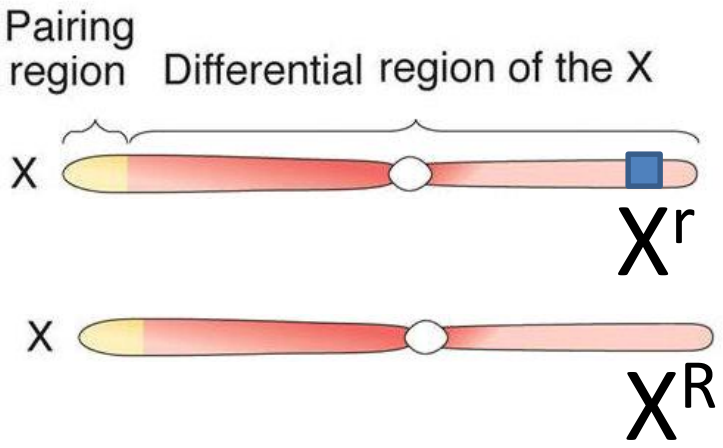
Y

Tallies with 3 red : 1 white eye fly that Morgan observed

2 red-eyed female flies : 1 white-eyed male fly : 1 red-eyed male fly

Males express X-linked recessive traits more easily than females





- In females, the other X chromosome may carry dominant alleles which then mask the expression of these recessive alleles.

Recap

Exercise [Pg.33]

- What is the phenotypic ratio of the F2 generation when a red-eyed male Drosophila is crossed with a white-eyed female , followed by sibling mating of the F1 generation?

Super script letter on the X chromosome

Unless instructed, avoid using W+ vs W

Pg 33

Key:

$X^{W+}X^{W+}$ = female with the allele for red eye (w^+)

$X^{W}Y$ = male with the allele for white eye (w)

$X^{W}X^{W}$ = female with neither allele

1:00

Use Capital letter vs small letter eg A vs a

Parental Phenotypes:	red-eyed male	X	white-eyed female
Parental Genotypes:	$X^{W+}Y$	X	$X^{W}X^{W}$

Gametes: X^{W+} , Y X X^{W}

Circled gametes

Genotypes (F1): $X^{W+}X^{W}$ $X^{W}Y$ $X^{W+}X^{W}$ $X^{W}Y$

Genotypic ratio of F_1 generation => 2 $X^{W+}X^{W}$: 2 $X^{W}Y$

Phenotypic ratio of F_1 generation => 1 red-eyed female : 1 white-eyed male

1 min

Complete p32

Sibling mating

Pg 34

Sibling mating of F1:

F1 Genotypes: $X^{W+}X^{W}$ X $X^{W}Y$

F1 gametes

Punnett square to show fusion of gametes produced by sibling-mating of F_1 generation

Female Gametes / Male Gametes	X^{W+}	X^{W}
X^{W}	$X^{W+}X^{W}$	$X^{W}X^{W}$
Y	$X^{W+}Y$	$X^{W}Y$

WRONG: 2 red eyed flies ; 2 white eyed flies

F2 genotypes	$X^{W+}X^{W}$:	$X^{W}X^{W}$:	$X^{W+}Y$:	$X^{W}Y$
F2 Genotypic ratio	1 :	1 :	1 :	1
F2 Phenotype	red-eyed female:	white-eyed female:	white-eyed male:	red-eyed male
F2 Phenotypic ratio	1 :	1 :	1 :	1

Remember to separate phenotypes by gender!

Pg 34

Sibling mating of F1:

F1 Genotypes: $X^{W+}X^{W}$ X $X^{W}Y$

F1 gametes

Punnett square to show fusion of gametes produced by sibling-mating of F_1 generation

F / Male Gamet	X^{W+}	X^{W}
	$X^{W+}X^{W}$	$X^{W}X^{W}$
	$X^{W+}Y$	$X^{W}Y$

No need IF genotypes are represented in punett sq

F2 genotypes: $X^{W+}X^{W}$: $X^{W}X^{W}$: $X^{W+}Y$: $X^{W}Y$

F2 Genotypic ratio: 1 : 1 : 1 : 1

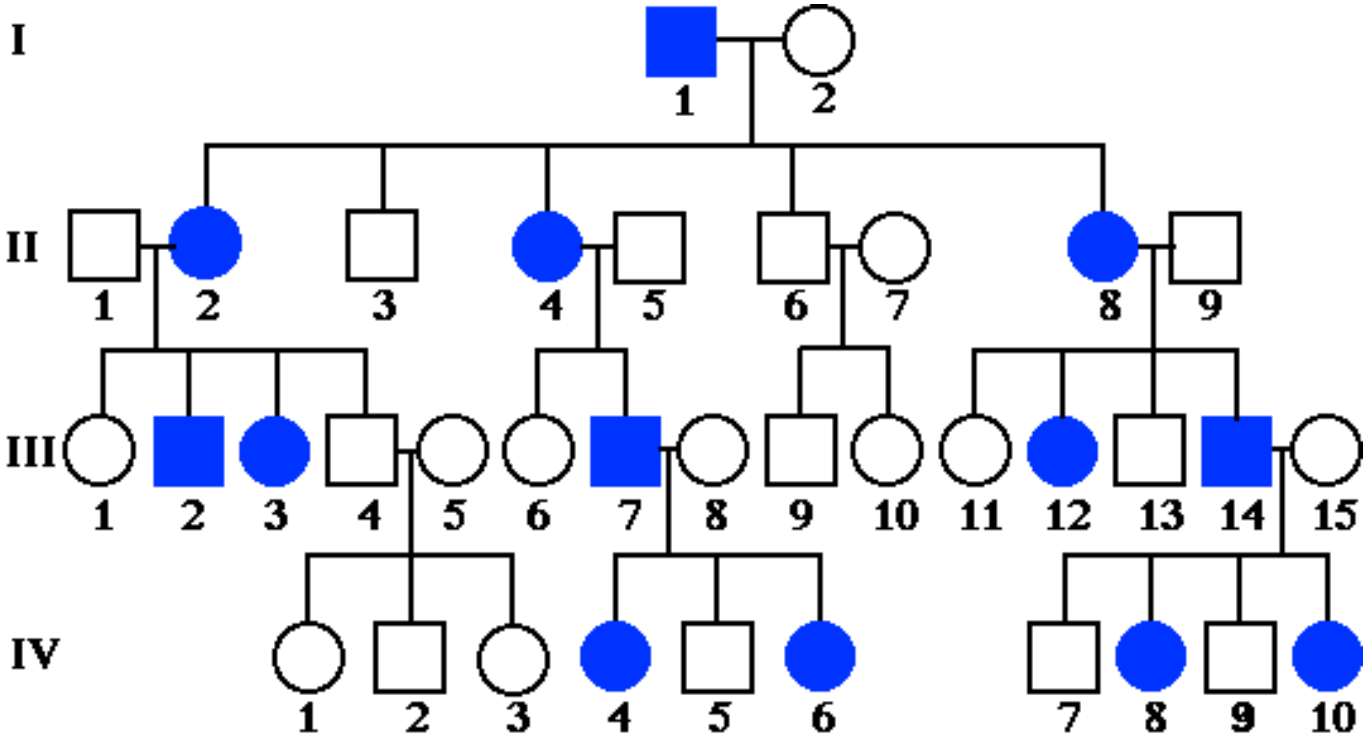
F2 Phenotype: red-eyed female: white-eyed female: white-eyed male: red-eyed male

F2 Phenotypic ratio: 1 : 1 : 1 : 1

Can combine into 1 line: phenotypic ratio

Pedigree

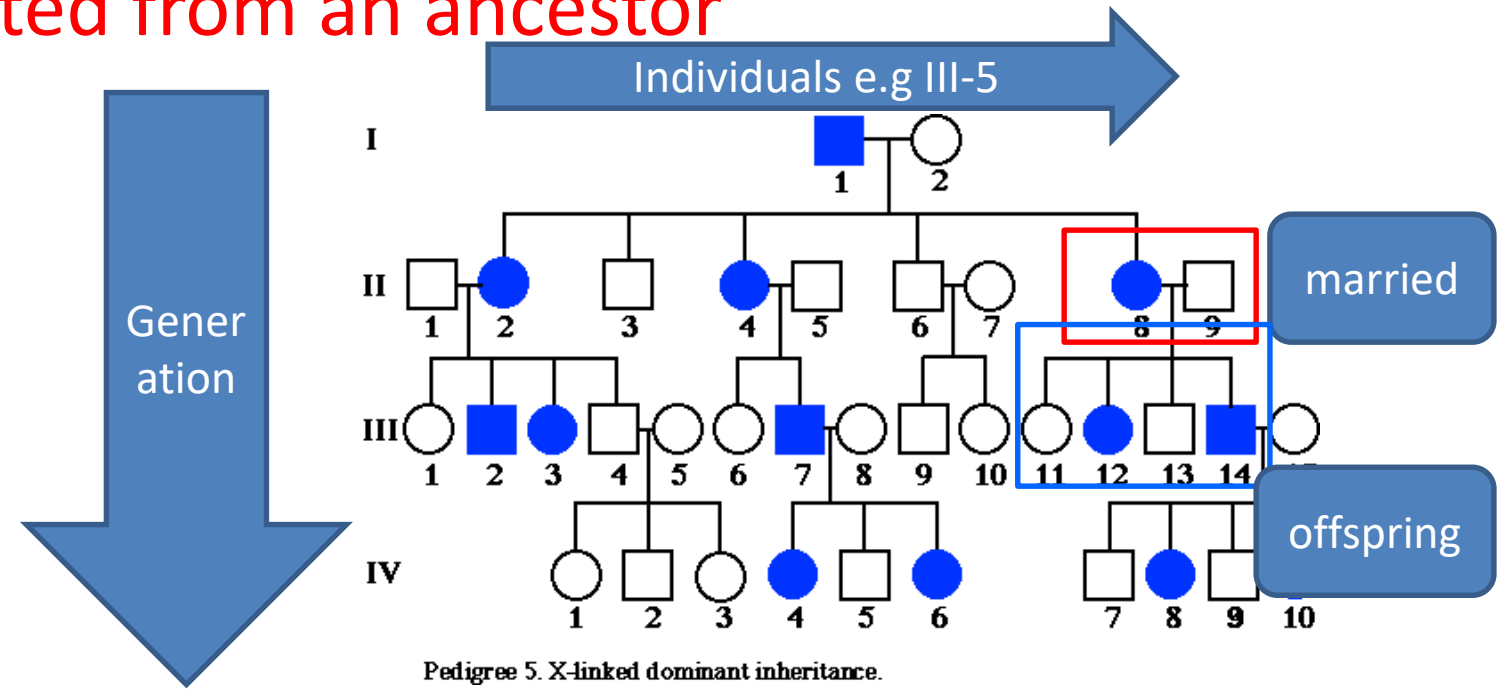
- A pedigree is a chart that shows a record of the family of an individual.



Pedigree 5. X-linked dominant inheritance.

Pedigree

- It can be used to show how family members are related to each other and how a particular characteristic (trait) or disease has been inherited from an ancestor



Pedigree 5. X-linked dominant inheritance.

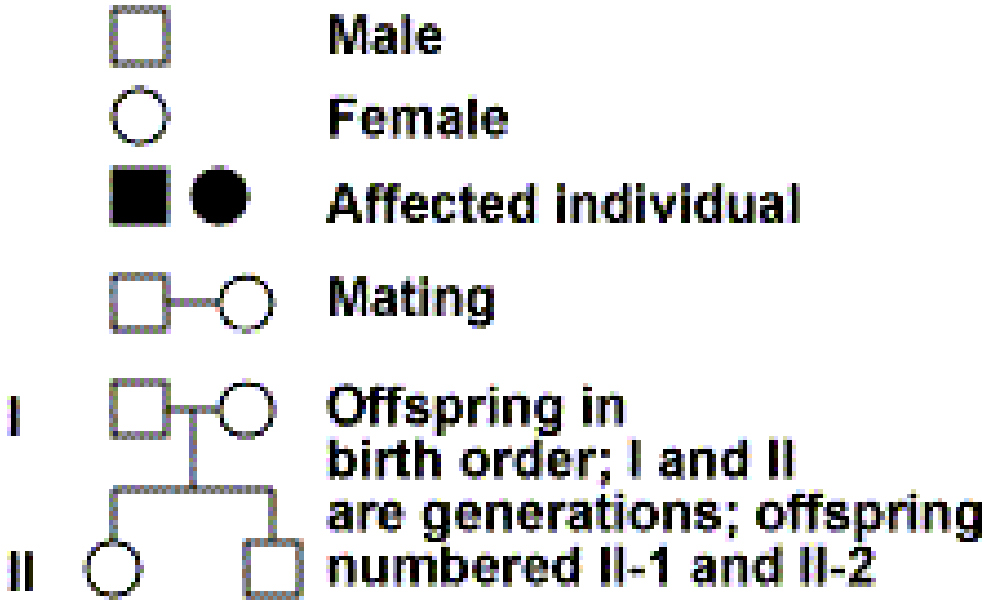
Pedigree

For analysis purposes

- It is particularly useful when
  (1) there are large families and
  (2) a good family record over several generations.

Pedigree

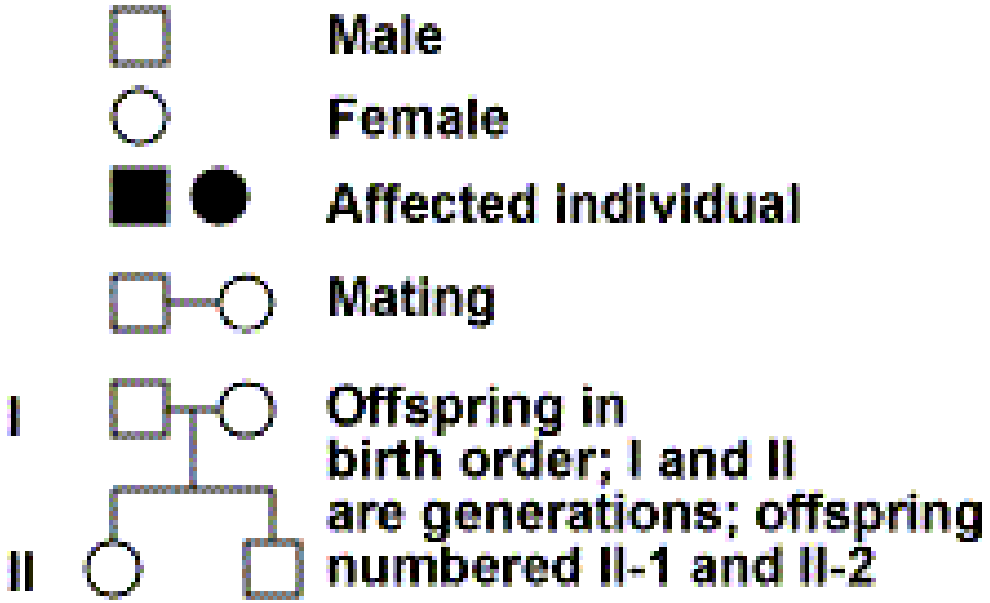
- In pedigree diagrams, males are represented by squares and females by circles. Trait under investigation is shaded if expressed

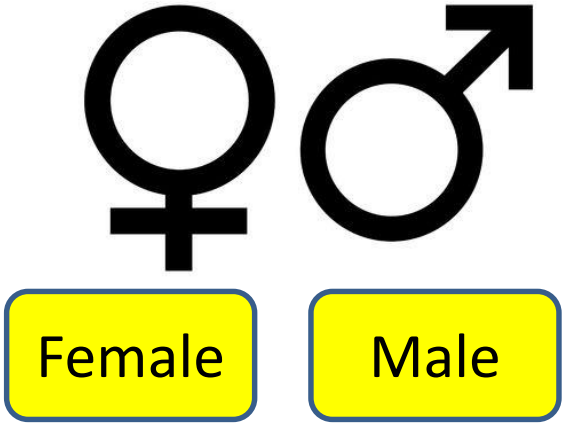


Male or female?

Pedigree

- In pedigree diagrams, males are represented by squares and females by circles. Trait under investigation is shaded if expressed

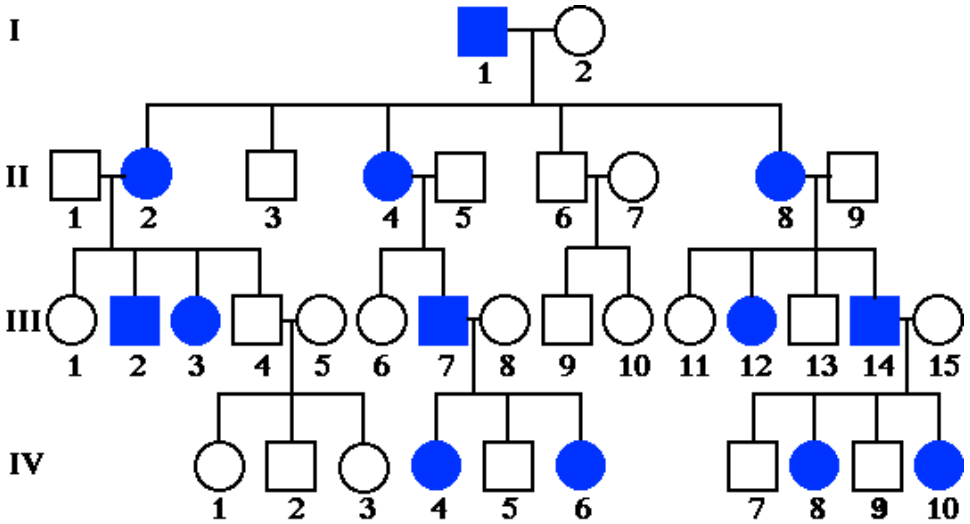




Pedigree

- In pedigree diagrams, males are represented by squares and females by circles. Trait under investigation is shaded if expressed

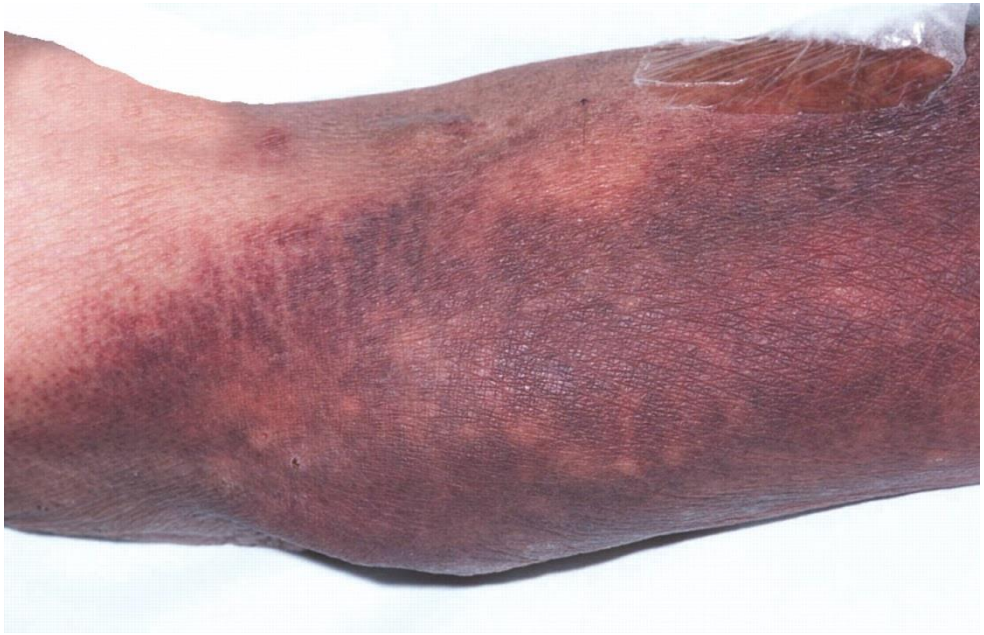
Shaded = diseased



Pedigree 5. X-linked dominant inheritance.

Individual organisms should be numbered from left to right for each generation e.g II-1, II-2, and each generation should be shown by Roman numerals e.g. I, II

Example of X-linked genes



(2) Haemophilia in Humans

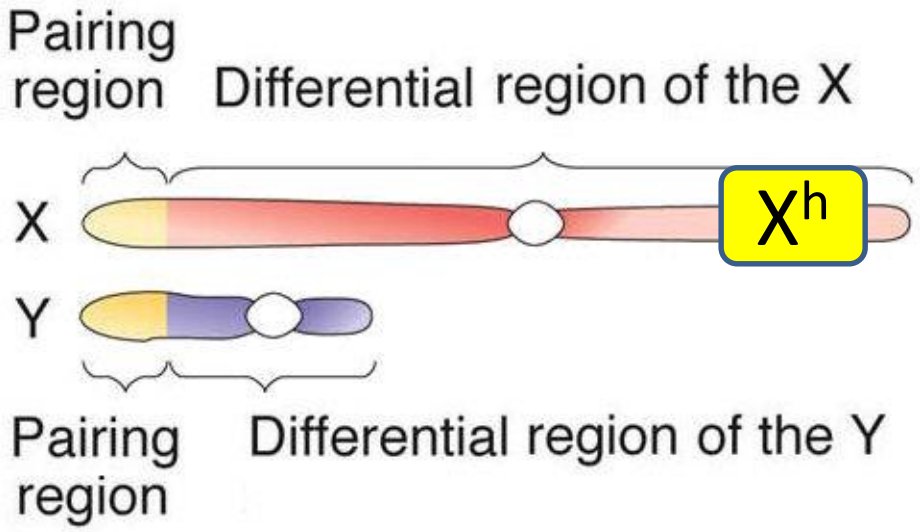
(2) Haemophilia in Humans

- A blood clotting disorder due to deficiency of one of the blood clotting factors, **VIII**
- The gene encoding for **VIII** is located on the longer arms of the X chromosome
- The allele for normal blood clotting (X^H) is dominant to that for haemophilia (X^h)

Memorise this key

(2) Haemophilia in Humans

- With only a single X chromosome, males who inherit the recessive allele (always from their mother) will be unable to produce factor VIII and suffer from extensive bleeding

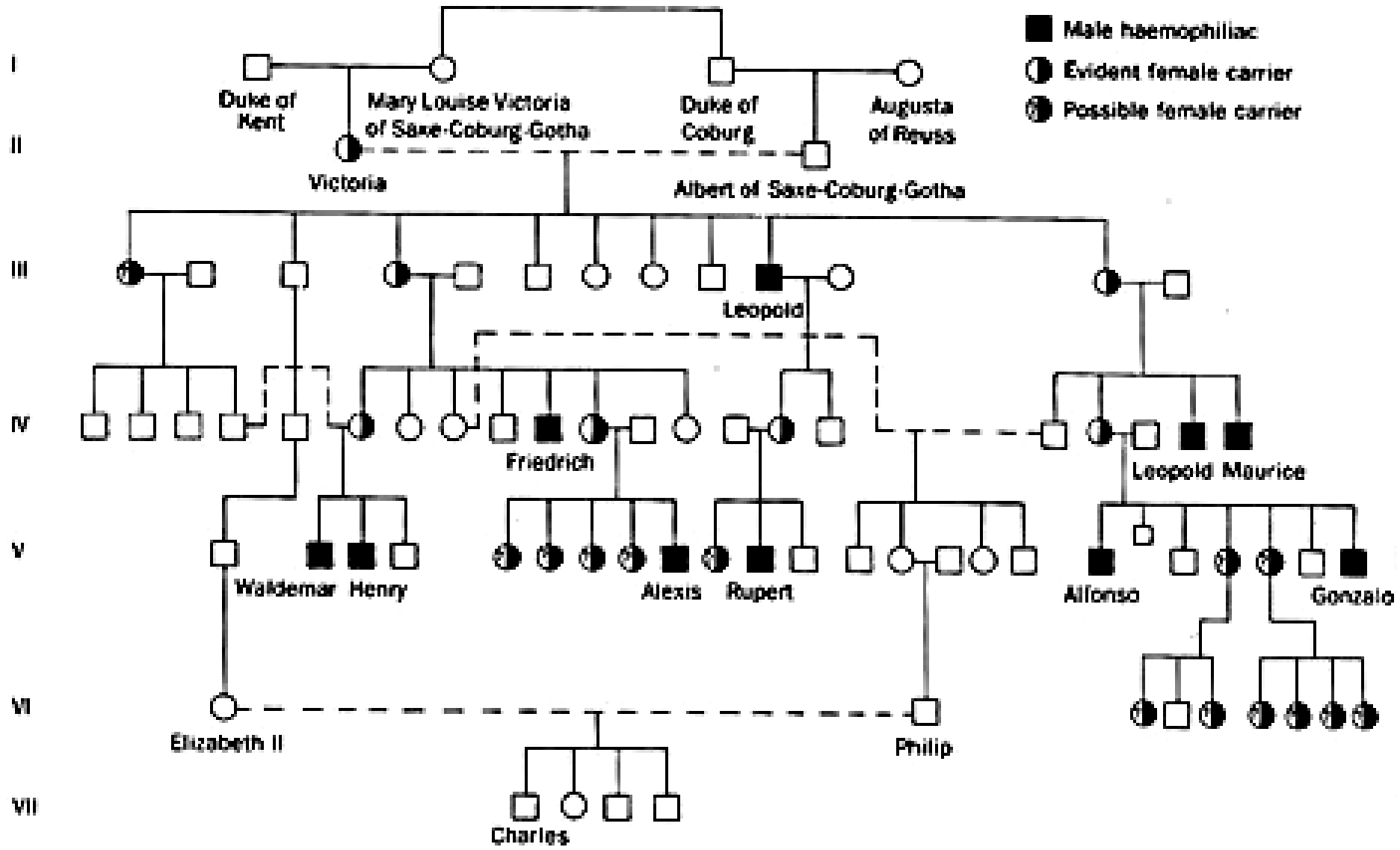


(2) Haemophilia in Humans

- For female to have haemophilia, the recessive allele must be present on both X chromosomes.
- Heterozygous females are called carriers because although they do not suffer from the disease, half their gametes carry the recessive allele.

(2) Haemophilia in Humans

- Famous case of X-linked gene type of disease e.g. haemophilia
- Queen Victoria of Great Britain was a carrier and transmitted the mutant allele to 3 out of 9 children
- Through inter-marriage, the mutated allele was transmitted from the British royal family to the German, Russian and Spanish royal families.


Male haemophiliac
Evident female carrier
Possible female carrier
I
II
III
IV
V
VI
VII
Duke of Kent
Mary Louise Victoria of Saxe-Coburg-Gotha
Duke of Coburg
Augusta of Reuss
Victoria
Albert of Saxe-Coburg-Gotha
Leopold
Friedrich
Leopold Maurice
Waldemar
Henry
Alexis
Rupert
Alfonso
Gonzalo
Elizabeth II
Philip
Charles

(2) Haemophilia in Humans

- X^H : X chromosome carrying allele for normal blood clotting
- X^h : X chromosome carrying allele for haemophilia

Memorise this key: universal

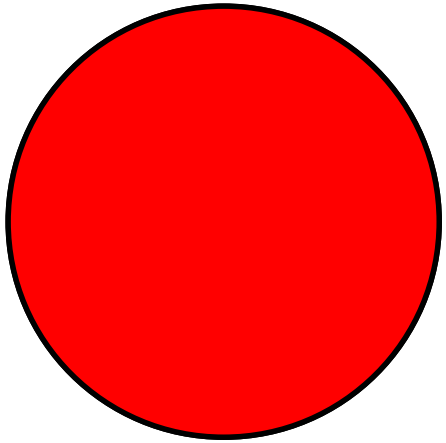
All X linked genes are represented by superscript alleles on the X

2 min

Exercise [Pg.37]

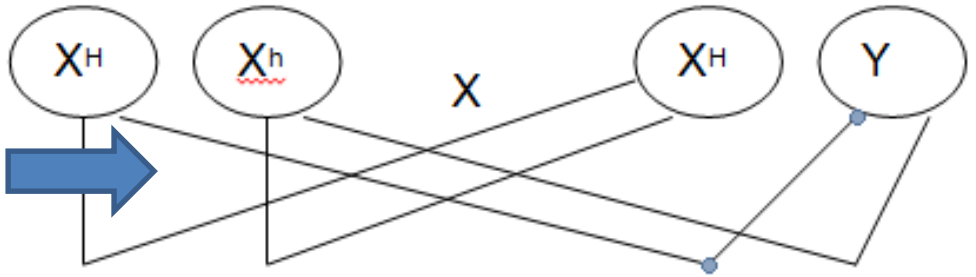
- Predict the probability of having haemophiliac offspring if a carrier female married a normal male

2 minutes



Key: X^H represents X chromosome with normal allele
X^h represents X chromosome with allele for haemophilia
Y represents Y chromosome

	Female	X	Male
Parental Phenotypes	Normal	X	Normal
Parental Genotypes	$X^H X^h$	X	$X^H Y$
Gametes			



Offspring genotypes	X^HX^H	X^HX^h	X^HY	X^hY
Offspring phenotypes	Normal female	Normal female (carrier)	Normal male	Haemophilic male

NO NEED for such lines

Probability of haemophiliac offspring = ¼

Separate phenotype by gender

Probability (male with haemophilia)?

Key: X^H represents X chromosome with normal allele
X^h represents X chromosome with allele for haemophilia
Y represents Y chromosome

A "condition" is listed in front: males

Parental Phenotypes	Normal	X	Normal
Parental Genotypes	$X^H X^h$	X	$X^H Y$
Gametes	X^H X^h	X	X^H Y
Offspring genotypes	X^HX^H	X^HX^h	X^HY X^hY
Offspring phenotypes	Normal female	Normal female (carrier)	Normal male, Haemophilic male

NO NEED for such lines

Probability of haemophiliac offspring = ¼

Separate phenotype by gender

Probability (male with haemophilia)?

Key: X^H represents X chromosome with normal allele
X^h represents X chromosome with allele for haemophilia
Y represents Y chromosome

A "condition" is listed in front: males

Parental Phenotypes	Normal	X	Normal
Parental Genotypes	$X^H X^h$	X	$X^H Y$
Gametes	X^H X^h	X	X^H Y
Offspring genotypes	$X^H X^H$ $X^H X^h$		$X^H Y$ $X^h Y$
Offspring phenotypes	Normal female; Normal female (carrier)		Normal male; Haemophilic male

NO NEED for such lines

Probability of haemophiliac offspring = ¼

Probability (male with haemophilia) = ½

Identify CONDITIONS in Question stems

P (Haemophilia offspring)

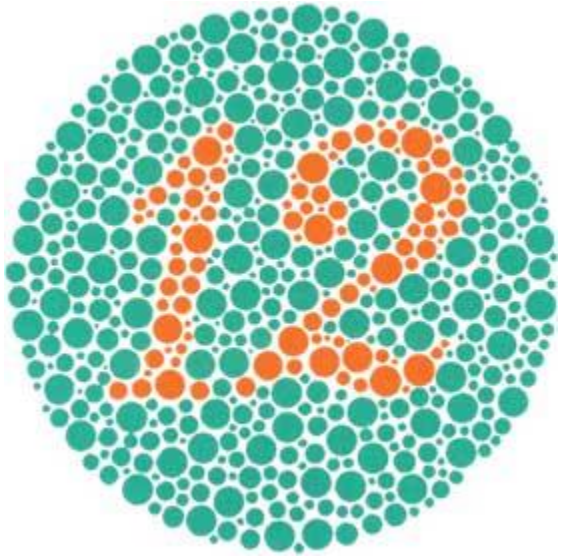
No condition

vs

P (boy with haemophilia)

Condition listed in front

Example of X-linked genes

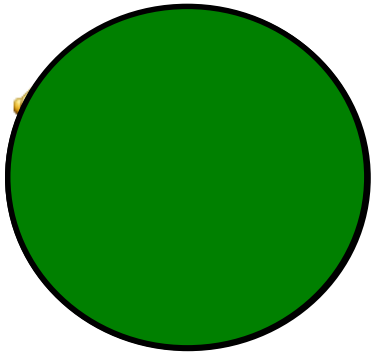


Red green colour blindness in Humans

Red-green colour blindness in Humans

- The inability to distinguish between red and green
- Allele for normal vision is dominant to that for red-green colour blindness

2 minutes



Exercise [Pg.38]

X^cX^c or X^CX^c ?

- A colour-blind man marries a woman with normal vision whose father is colour blind. If they have a daughter, what is the probability that she will be colour-blind?

X^cY

X^c passed to daughter

Condition is always listed in front

Pg 38

Key:

X^C represents X chromosome with the allele for normal vision (C).

X^c represents X chromosome with the allele for colour blindness (c).

Y represents Y chromosome

Rem the key: universal

	Female		Male
Parental Phenotypes	Normal vision (Carrier)		Colour-blind
Parental Genotypes	X^CX^c	X	X^cY
Gametes	X^C X^c	X	X^c Y
Offspring Genotypes	X^CX^c X^cX^c		X^CY X^cY
Offspring Phenotypes	Female Normal (carrier) / Female Colour-blind		Male Normal / Male Colour-blind

Ans 0.5

List phenotype by gender, identify carriers

Red-green colour blindness in Humans

Pg 38

- The allele for colour blindness is passed from one sex to the other at each generation
- The colour-blind father passes the recessive allele to all his daughters who thus become carriers X^cY
- Heterozygous females pass the X-linked recessive allele to half their sons and half their daughters X^CX^c
- Their sons are thus colour-blind (if they inherit recessive allele)

Diseased mothers X^cX^c always have diseased sons

Recap!

- Incomplete dominance
- Codominance
- Multiple alleles
- Sex-linked genes

How to test if a trait is X linked?

Reciprocal Cross

- A reciprocal cross is one where the same genetic features are used, but the sexes are reversed.
- For e.g. the reciprocal cross for crossing a red-eyed female with a white-eyed male is … Crossing a red-eye male with a white-eyed female

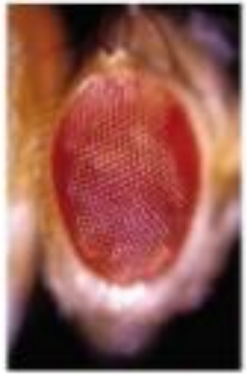
Perform 2 "halve" crosses → Compare results of each cross

QUESTION: How to test if a trait is X-linked???

White-eyed x Red-eyed

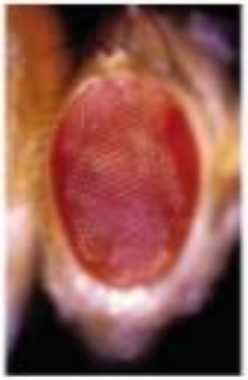
X^rY

X^RX^R



Red-eyed x White-eyed

X^RY



X^rX^r

Reciprocal Cross

- Purpose of a reciprocal cross Observation of the offspring from the two halves of the reciprocal cross will allow determination of whether a characteristic is X-linked or not.

NOT Sex-linked
(too vague)

Reciprocal Cross

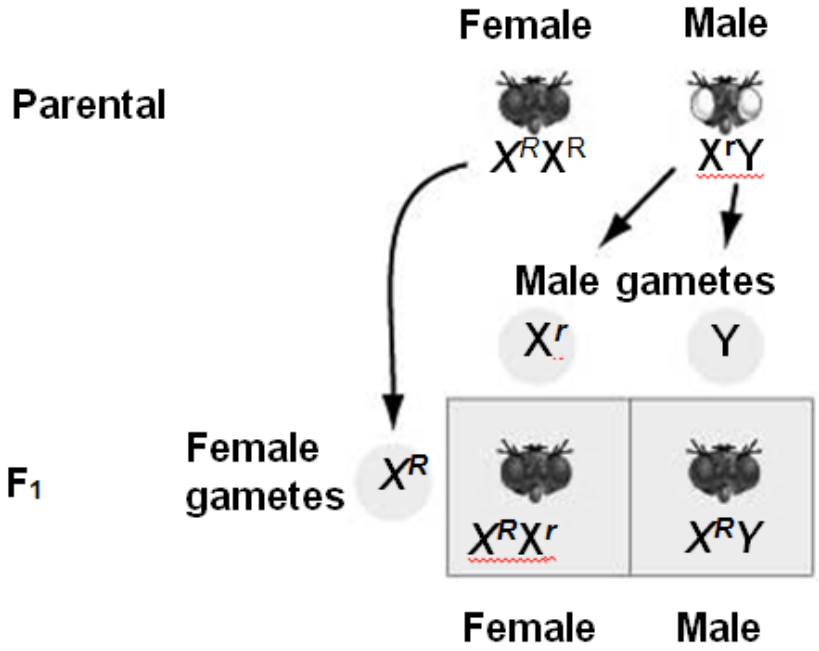
Autosomal

- If the trait for eye colour is **NOT X-linked**, results for both halves of the reciprocal cross will be the **SAME**. (RR x rr for both halves; all offspring Rr ➔ same phenotype eg all red eyed flies)
- If the trait for eye colour **IS X-linked**, the second half of the reciprocal cross will give a **DIFFERENT** result from the first half.

Reciprocal Cross

Observations if trait is X-linked

First half of reciprocal cross

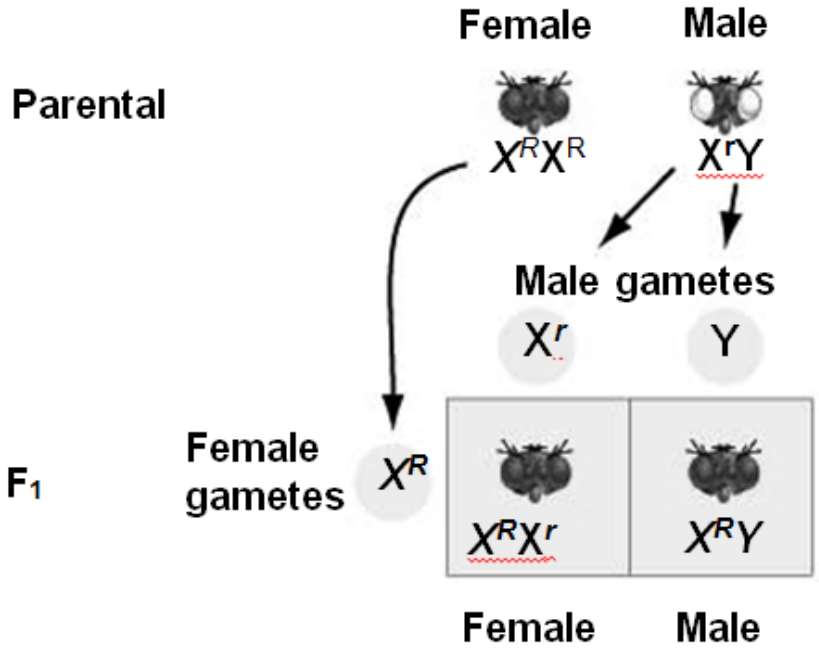


Resulting phenotypes: All offspring are red-eyed

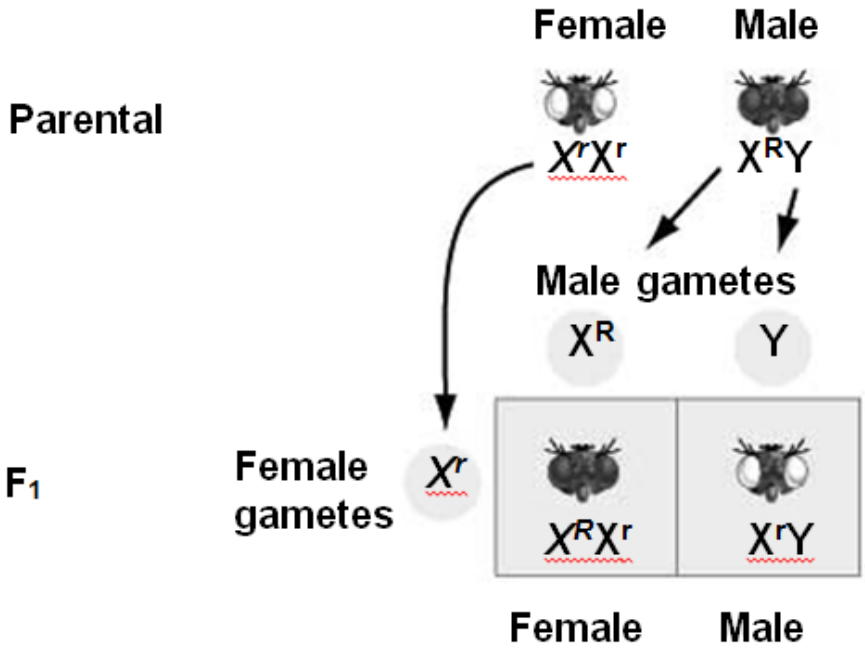
Reciprocal Cross

Observations if trait is X-linked

First half of reciprocal cross



Second half of reciprocal cross



Resulting phenotypes: All offspring are red-eyed

Resulting phenotypes: F_1 females are red-eyed, F_1 males are white-eyed

Reciprocal Cross

Observations if trait is X-linked

First half of reciprocal cross

Parental: Female X^RX^R × Male

F_1: Female gametes X^R × Male gametes

	X^R	
X^R	X^RX^r	X^RY

F_1: Female X^RX^r, Male X^RY

Second half of reciprocal cross

Parental: Female X^rX^r × Male X^RY

F_1: Female gametes X^r × Male gametes X^R, Y

	X^R	Y
X^r	X^RX^r	X^rY

F_1: Female X^RX^r, Male X^rY

DIFFERENT OUTCOMES FOR EACH HALVE CROSS! = X linked trait

Resulting phenotypes: All offspring are red-eyed

Resulting phenotypes: F_1 females are red-eyed, F_1 males are white-eyed

Exceptions to Mendelian genetics

- Incomplete dominance
- Codominance
- Multiple alleles
- Sex-linked genes

Mendelian Ratios NOT observed

What I need to know … 01

- 2(n)
  Use genetic diagrams to solve problems in dihybrid crosses, including those involving **incomplete dominance, codominance, multiple alleles** and **sex linkage**

GENETIC CROSS

Monohybrid

Expected

1 gene (on autosome)→ 1 trait
1 gene → 2 alleles
(completely dominant or recessive)
Total = 2 phenotypes

Aa x Aa → 3 : 1
&
Aa x aa → 1 : 1

Deviations

1. Incomplete dominance } Alleles not completely dominant or recessive
2. Codominance
3. Multiple alleles → Gene has more than 2 alleles
4. Sex-linkage → Gene on sex chromosome

Dihybrid

2 genes (on different autosome)→ 2 traits
Each gene → 2 alleles
(completely dominant or recessive)
2 genes, 4 alleles hence…
Total = 4 phenotypes

Expected

AaBb x AaBb →
9 : 3 : 3 : 1
&
AaBb x aabb →
1 : 1 : 1 : 1

Deviations

Not in H1 syllabus