

Sex Determination and Sex- Linked Characteristics



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Introduction

- *Mechanisms of Sex Determination*
 - Chromosomal
 - Environmental
 - Genetic

- *Sex-linked Characteristics*

Sex Determination

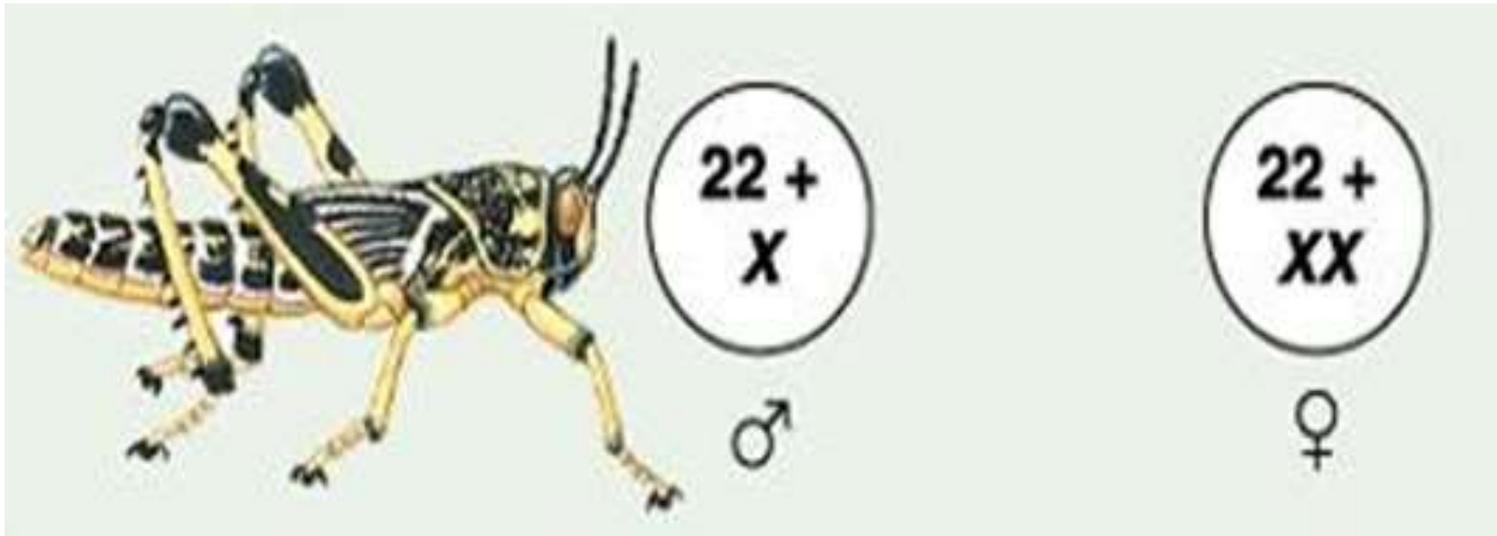
- Sexual reproduction is the results of meiosis and fertilization
- Sex determination – mechanism by which sex is established
- **Monoecious** – organisms with both male and female reproductive structures (**hermaphroditism**)
- **Dioecious** – organism has male or female reproductive structures
- In many organisms, sex is determined by a pair of chromosomes – sex chromosomes (**Allosomes**) and Non-sex chromosomes (**Autosomes**)
- **Heterogametic sex** – gametes differ with respect to sex chromosomes
- **Homogametic sex** – gametes are the same with respect to sex chromosomes

How does the heterogametic sex differ from the homogametic sex?

- The heterogametic sex is male; the homogametic sex is female.
- Gametes of the heterogametic sex have different sex chromosomes; gametes of homogametic sex have the same sex chromosome.
- Gametes of the heterogametic sex all contain a Y chromosome.
- Gametes of the homogametic sex all contain an X chromosome.

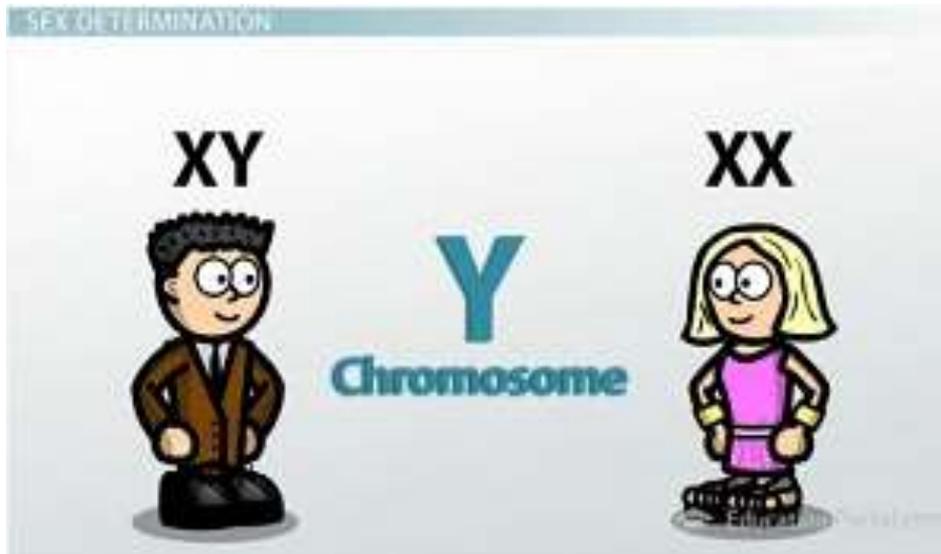
Chromosomal Sex-Determination

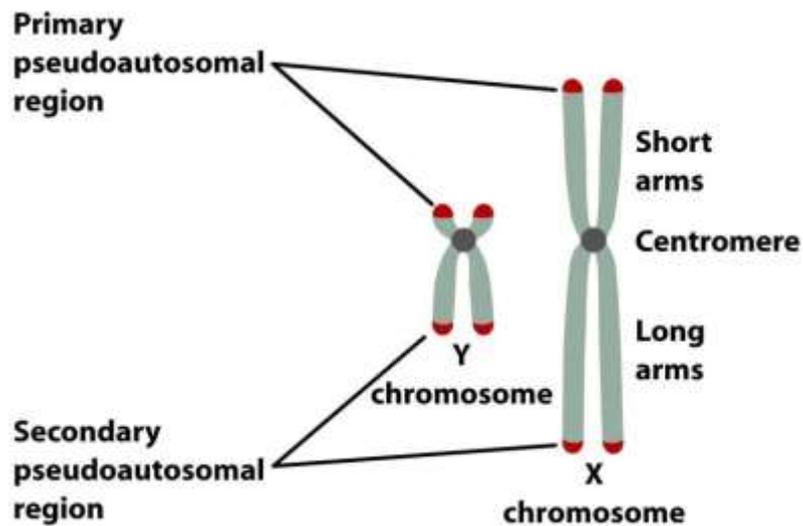
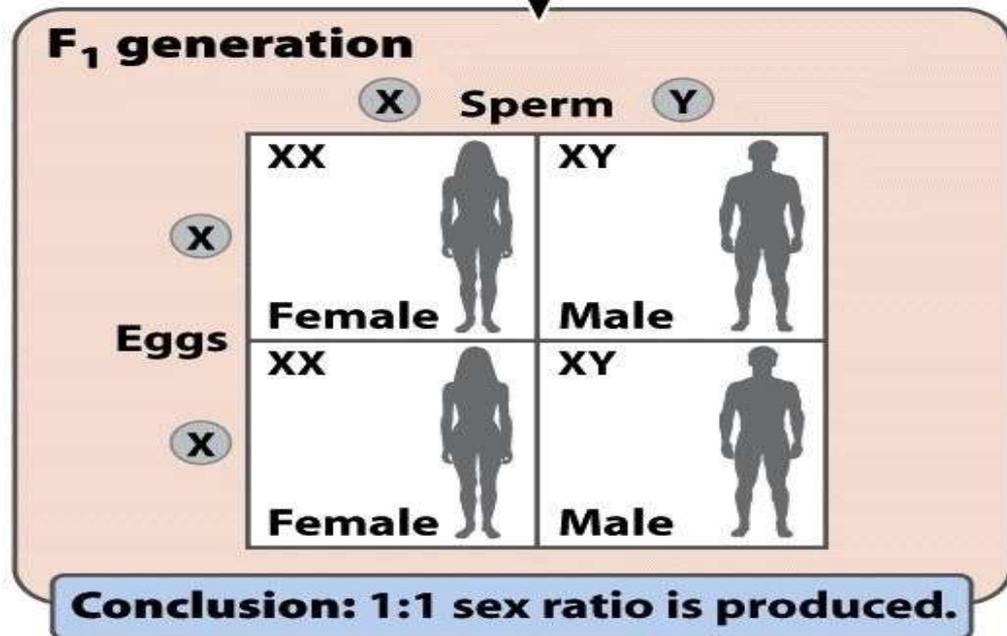
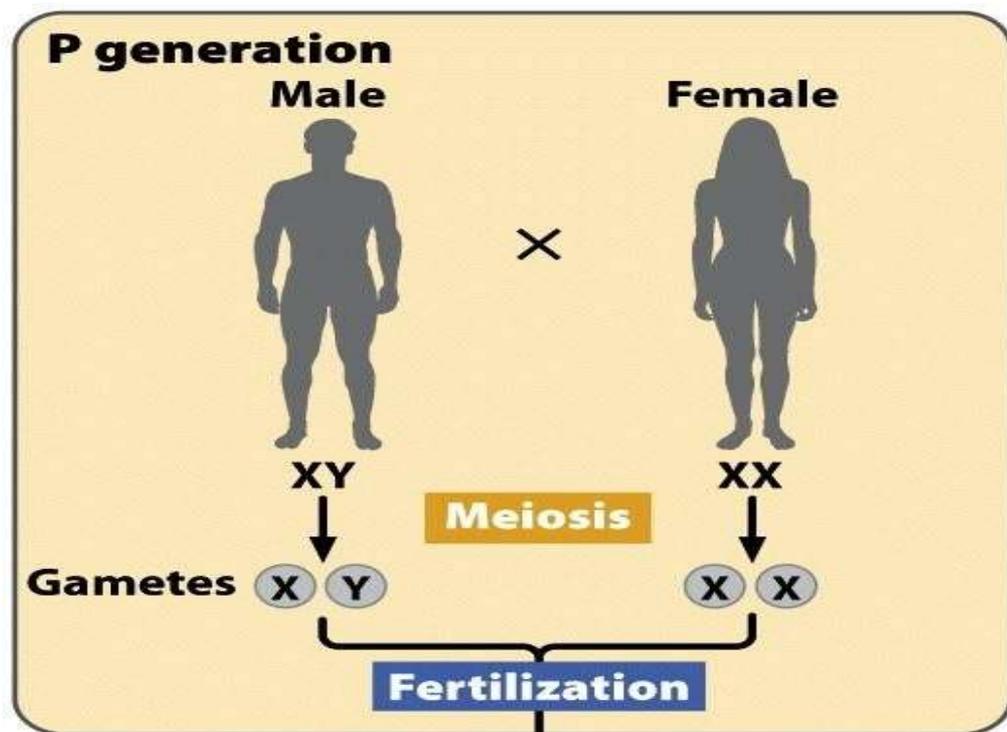
- **XX-XO sex determination** – Ex. grasshoppers
 - XX - **female**; homogametic
 - XO - **male**; heterogametic
 - (O = absence of chromosome)



Chromosomal Sex-Determination

- **XX-XY sex determination** – many species incl. mammals
 - XX – female; homogametic
 - XY – male; heterogametic





Chromosomal Sex-Determination

- **ZZ-ZW sex determination** – birds, snakes, amphibians, butterflies, isopods, some fish
 - ZZ – male; homogametic
 - ZW – female; heterogameic



76
+
ZW



76
+
ZZ

Chromosomal Sex-Determination

➤ **Haplodiploidy** – bees, wasps, ants

➤ Unfertilized eggs,

haploid set – male

➤ Fertilized eggs,

diploid set – female

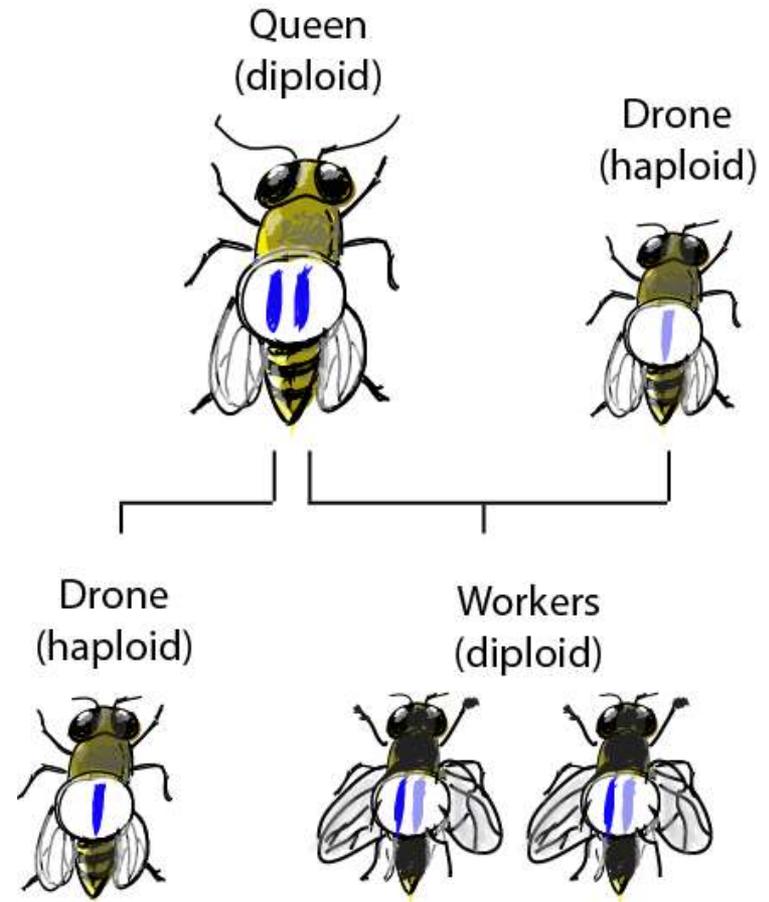
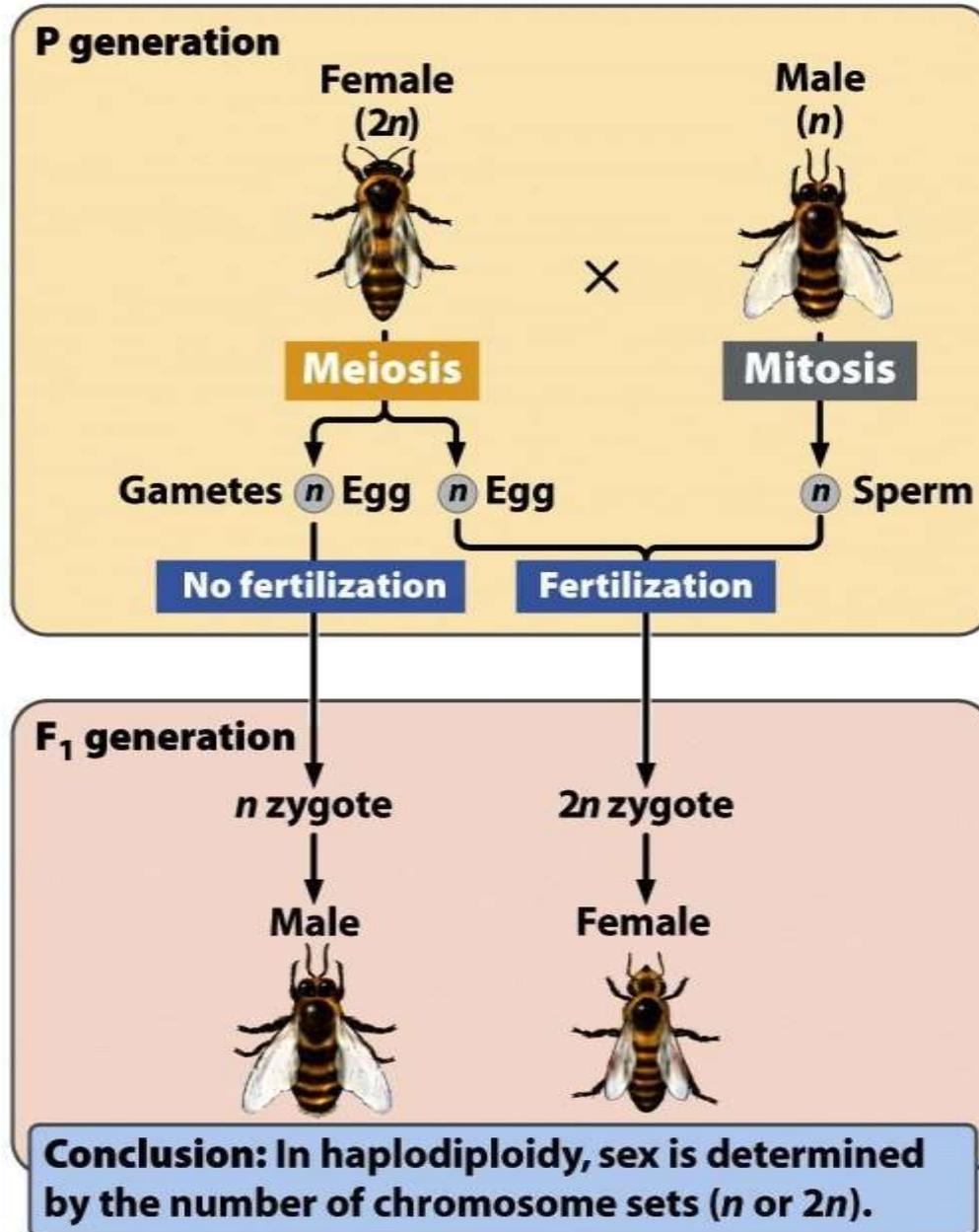
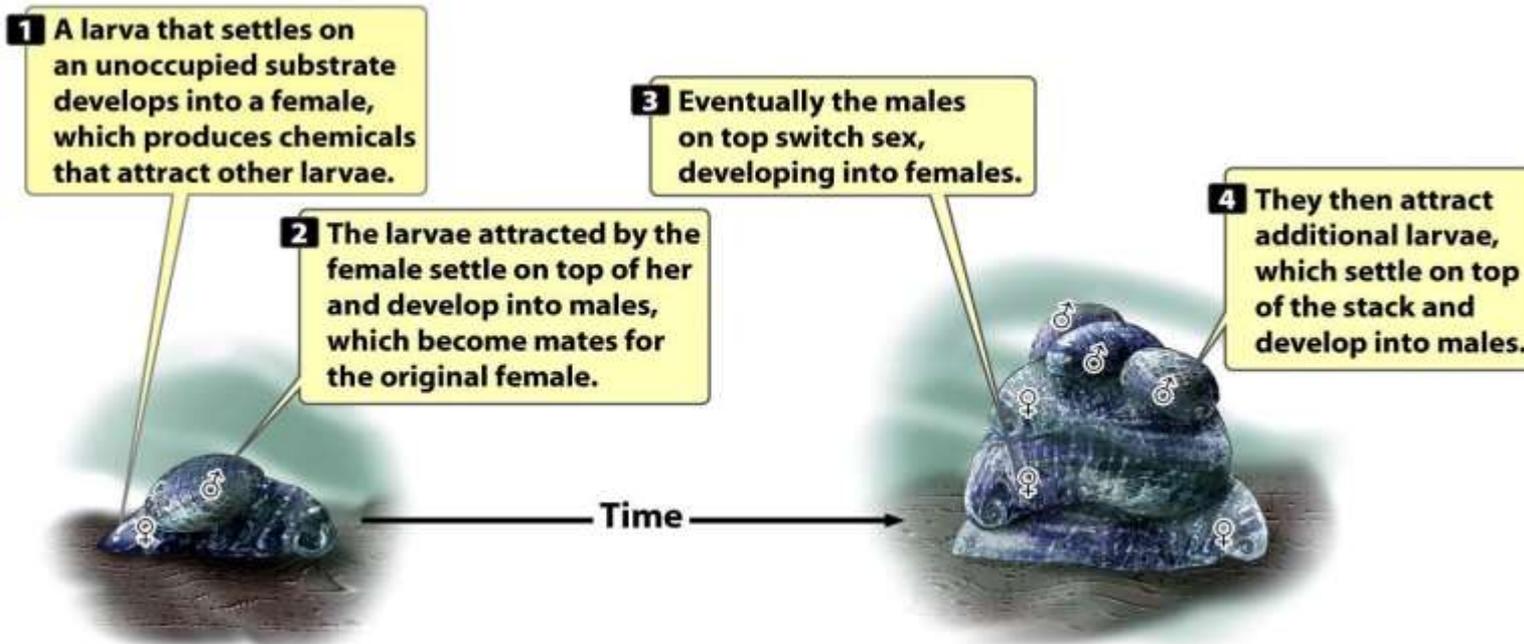
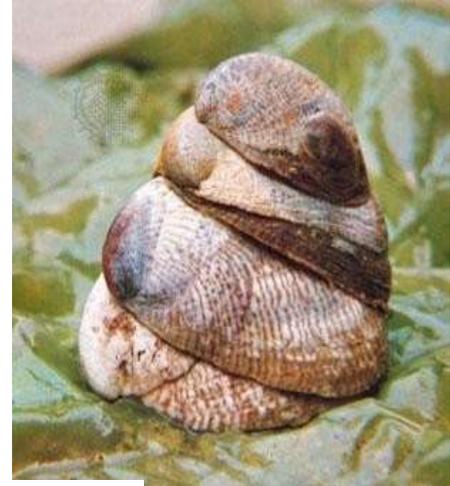


Fig 4.6



Environmental Sex Determination

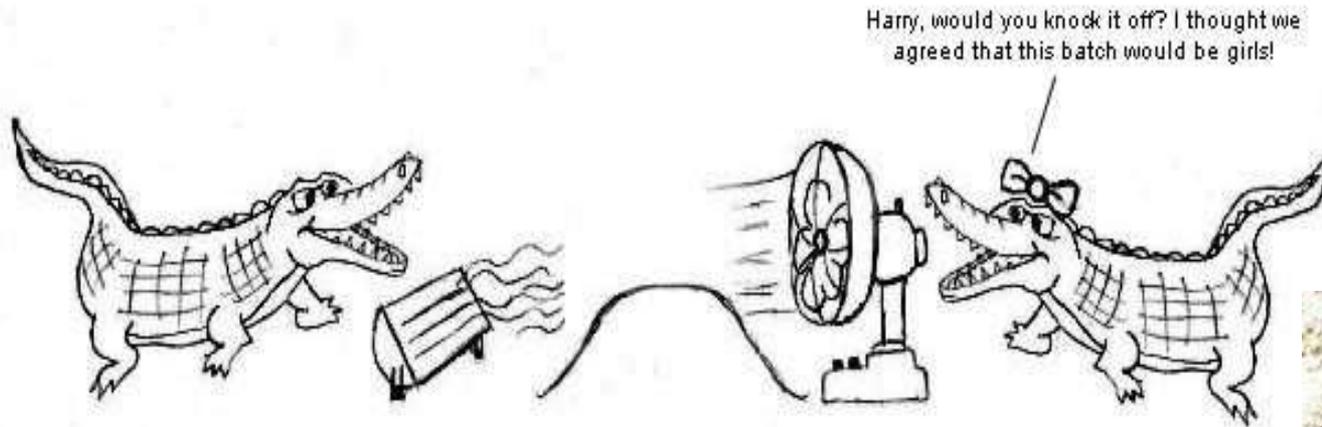
- Limpets – stack position determines sex
 - Bottom of stack female, top male
 - But males then become females and new individuals on top settle as males
 - **Sequential hermaphroditism**



Environmental Sex Determination

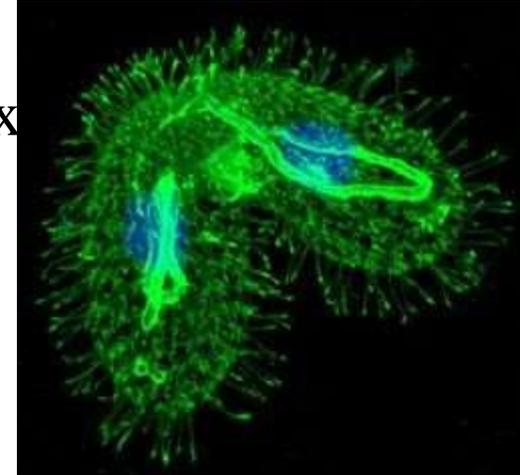
- **Reptiles** – temperature during development
 - Turtles: warm temps produce females
 - Alligators: warm produce males

The Finest Technology in Gender Determination in Reptiles



Genetic Sex Determination

- Sex is genetically determined, But no sex chromosomes
- Sex is still determined by genes
 - As in chromosomal sex determination
 - Genes at one or more loci determine sex of individual
- Found in some plants and protozoans



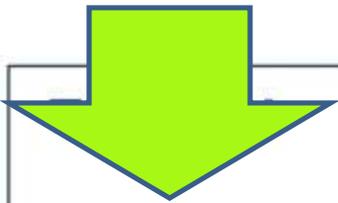
Drosophila melanogaster – fruit fly

➤ XX-XY system

- But presence of Y does not determine maleness
- **Genetic balance system** – sex determined by a balance between genes on autosomes and genes on X

➤ X : A ratio

- X = number of X chromosomes
- A = number of haploid sets of autosomes



Chromosome complements and sexual phenotypes in *Drosophila*

Sex-Chromosome Complement	Haploid Sets of Autosomes	X : A Ratio	Sexual Phenotype
XX	AA	1.0	Female
XY	AA	0.5	Male
XO	AA	0.5	Male
XXY	AA	1.0	Female
XXX	AA	1.5	Metafemale
XXXY	AA	1.5	Metafemale
XX	AAA	0.67	Intersex
XO	AAA	0.33	Metamale
XXXX	AAA	1.3	Metafemale

NORMAL

Appear normal;
male
sterile

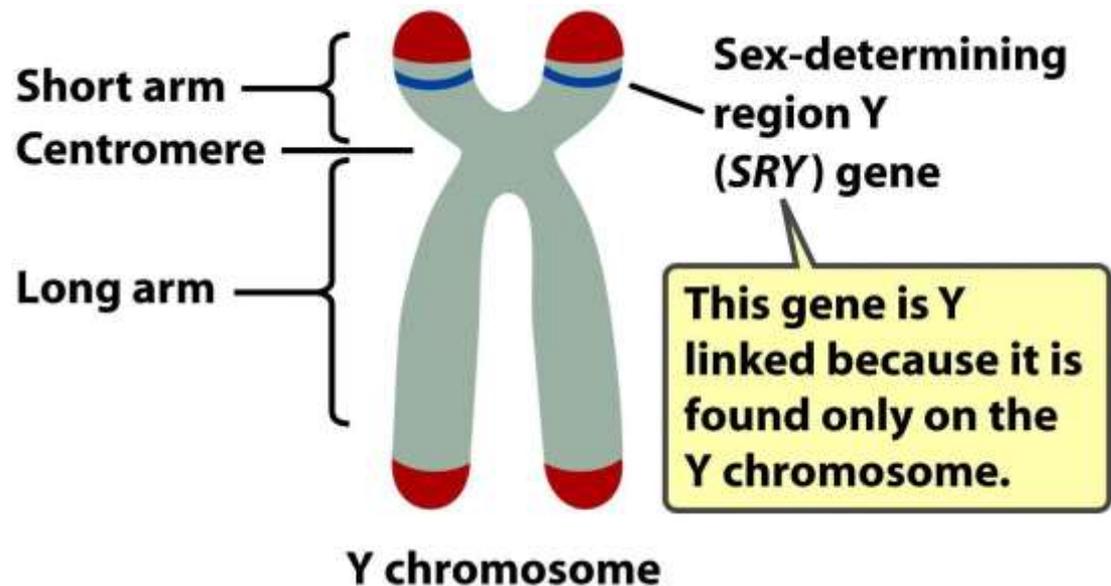
>1, female but
devel. problems

btw 0.5-1, intersex
<0.5 weak, sterile

Results of the sex-determination in abnormal flies confirms that the Y chromosome does not determine sex in *Drosophila*

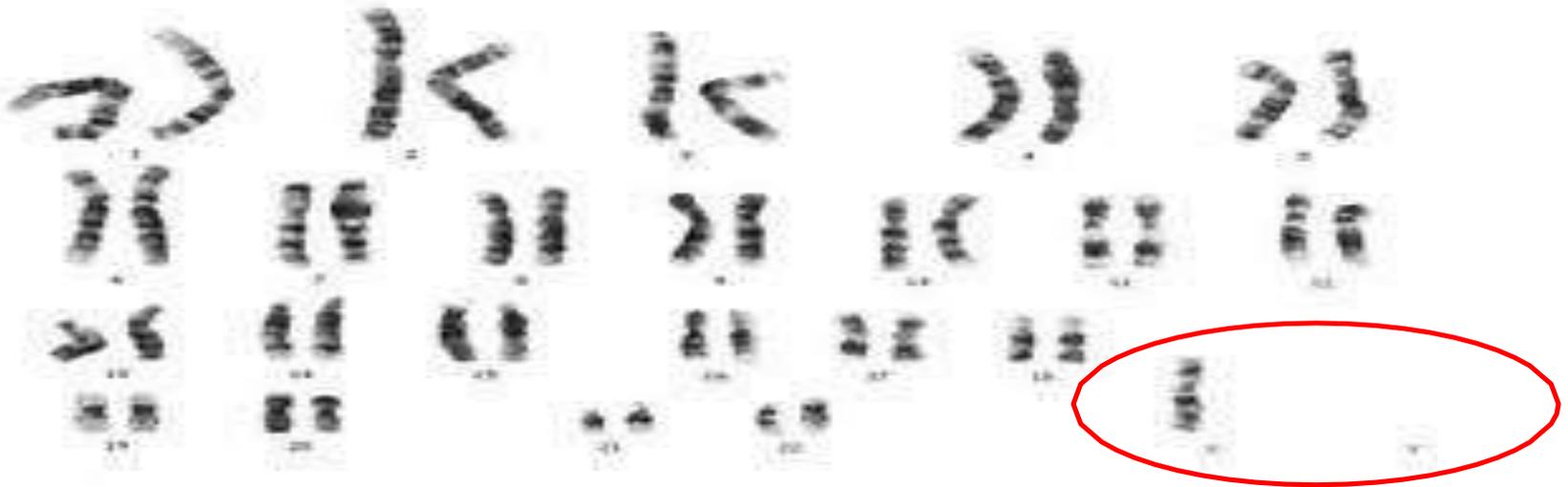
Sex Determination - Humans

- **XX-XY** sex determination
- **SRY** gene on Y chromosome determines maleness



- Phenotypes from abnormal sex chromosome numbers illustrate the importance of *SRY*
 - **Turner syndrome**: XO; female
 - **Klinefelter syndrome**: XXY, or XXXY, or XXXXY, or XXYY; male

Turner syndrome



Role of Sex Chromosomes - Humans

- X contains genetic info essential for both sexes
- Male determining gene on Y chromosome, Even if multiple Xs, still male
- Absence of Y results in female
- Genes affecting fertility – on both X and Y, 2 copies of X required for female fertility
- Additional X may upset normal development in both male and female

Inheritance of Sex-linked Characteristics

- In humans and other mammals, biological sex is determined by a pair of **sex chromosomes**: XY in males and XX in females.
- Genes on the X chromosome are said to be **X-linked**. X-linked genes have distinctive inheritance patterns because they are present in different numbers in females (XX) and males (XY).
- X-linked human genetic disorders are much more common in males than in females due to the X-linked inheritance pattern.
- When a gene is present on the X chromosome, but not on the Y chromosome, it is said to be X-linked. X-linked genes have different inheritance patterns than genes on non-sex chromosomes (autosomes). That's because these genes are present in different copy numbers in males and females.

Inheritance of Sex-linked Characteristics

➤ **Thomas Hunt Morgan** – first to explain White eyes in *Drosophila*

Experiment

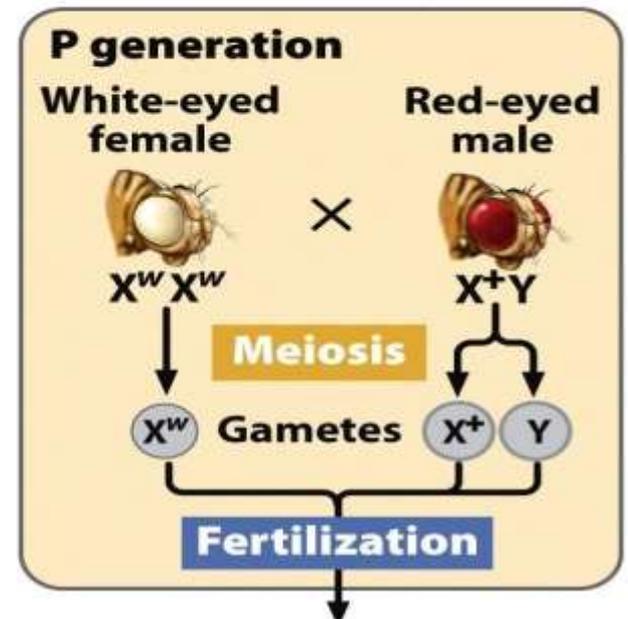
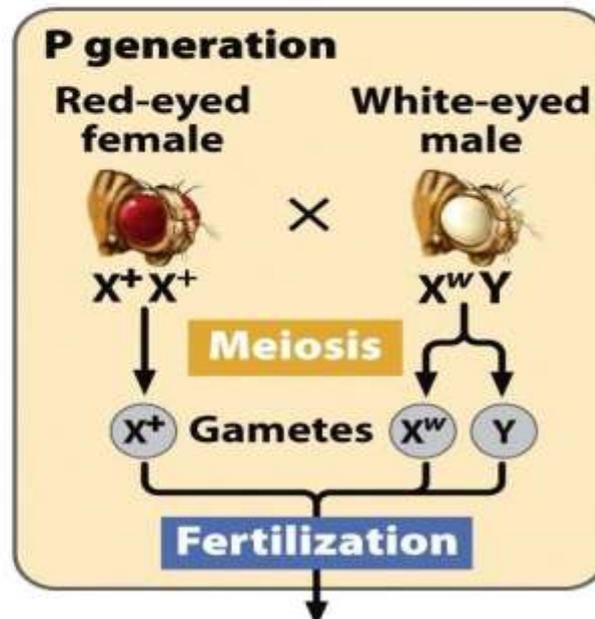
Question: Are white eyes in fruit flies inherited as an autosomal recessive trait?

Methods

Perform reciprocal crosses.

(a) Red-eyed female crossed with white-eyed male

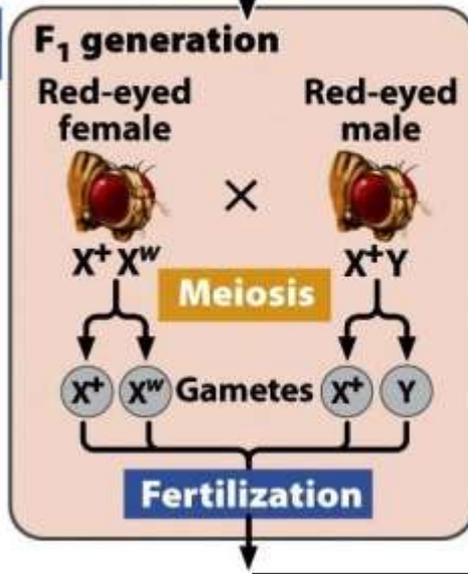
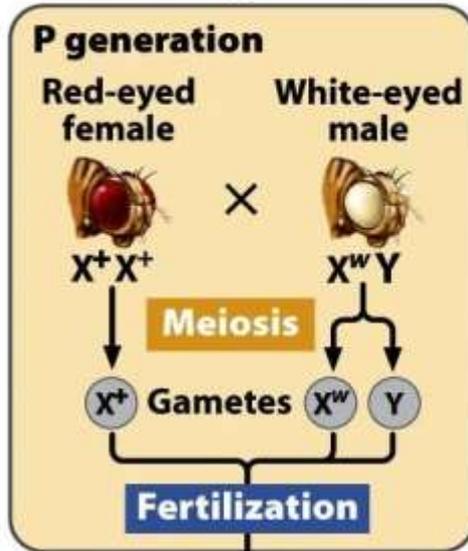
(b) White-eyed female crossed with red-eyed male



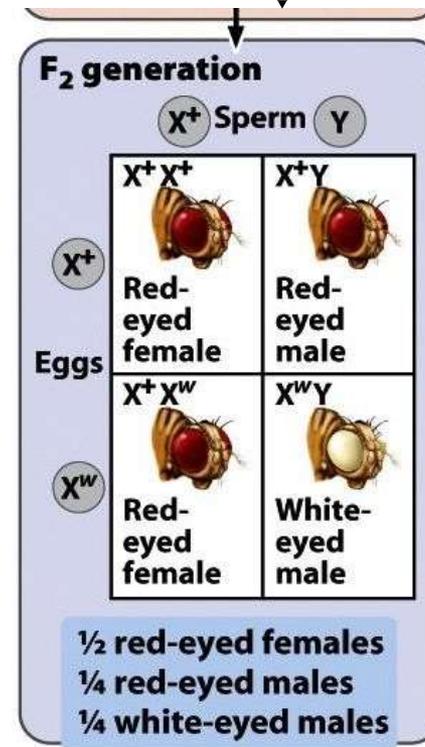
Locus for eye colour is X-linked

Males are **hemizygous**

(a) Red-eyed female crossed with white-eyed male

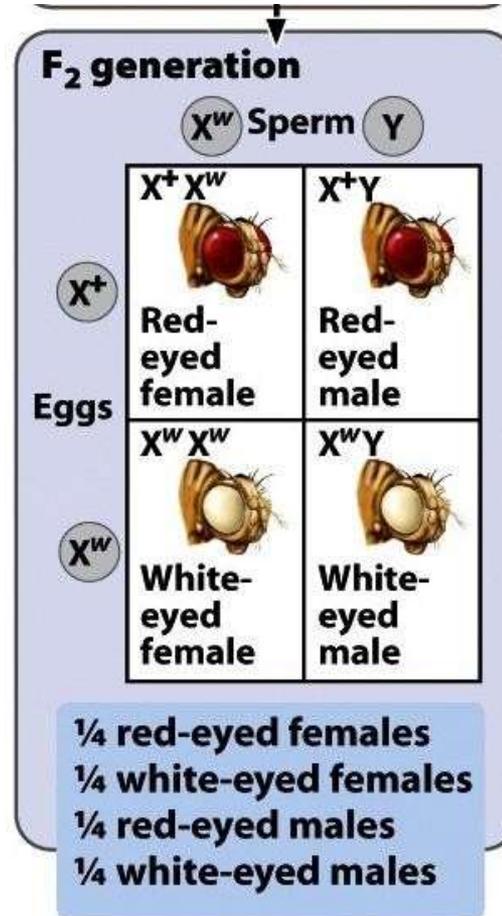
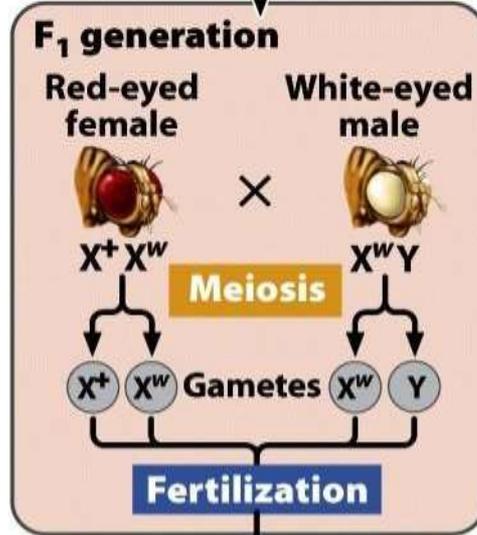
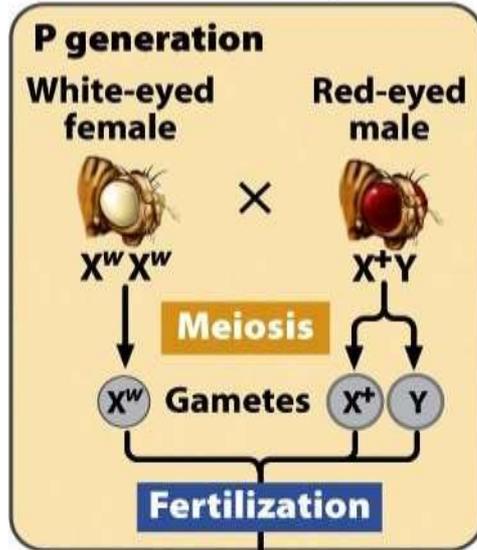


Results



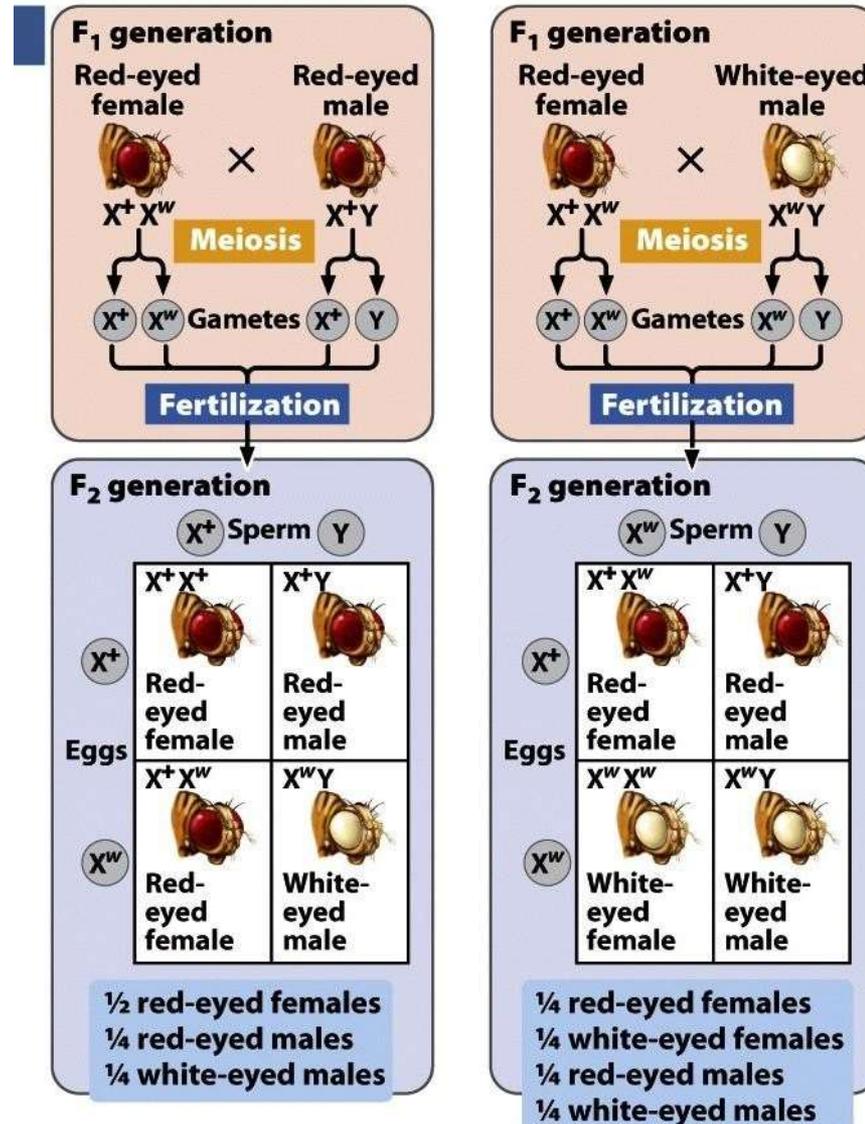
I (b) White-eyed female crossed with red-eyed male

Reciprocal cross



Reciprocal crosses – produced different results in F1 and F2

-consistent with X-linked inheritance



Non-disjunction

Chromosomes fail to separate in anaphase 1 – non-disjunction

Non-disjunction of 2Xs in XXY female flies

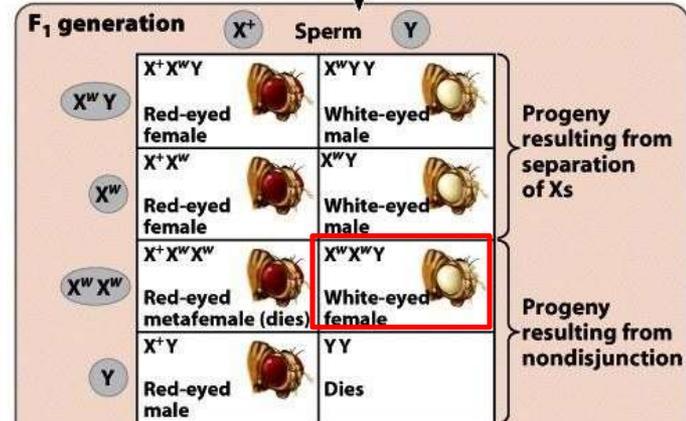
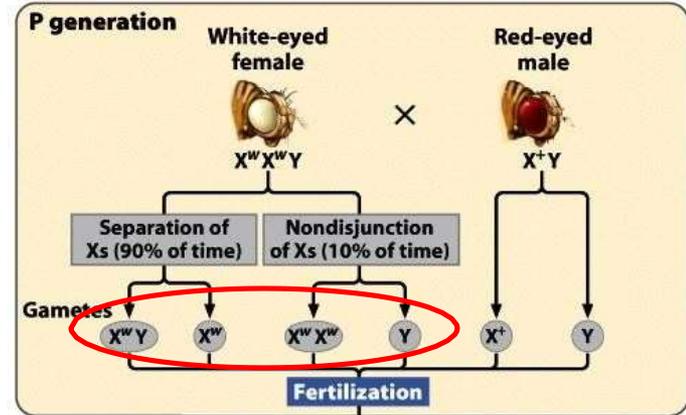
Confirms evidence of sex-linked gene for eye-color on the X chromosome

Experiment

Question: In a cross between a white-eyed female and a red-eyed male, why are a few white-eyed females and red-eyed males produced?

Methods

Hypothesis: White-eyed females and red-eyed males in F_1 result from nondisjunction in an XXY female.



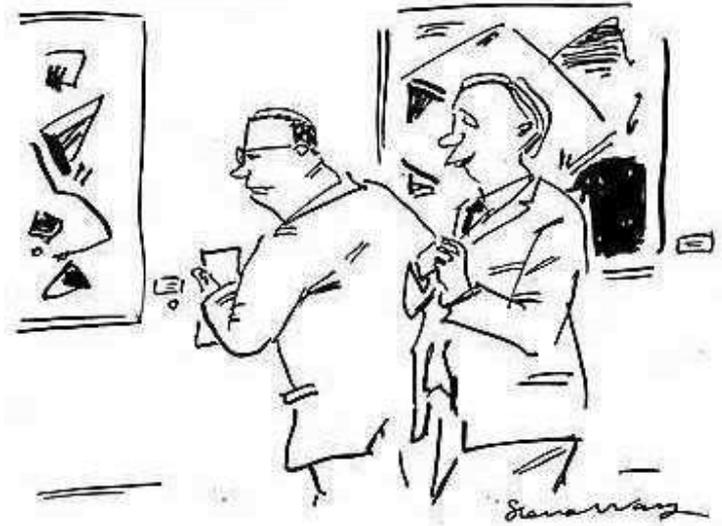
Results

F ₁ flies	Chromosomes observed
Red-eyed females	$\frac{1}{2} X X, \frac{1}{2} X X Y$
White-eyed males	$\frac{1}{2} X Y, \frac{1}{2} X Y Y$
White-eyed females	$X X Y$
Red-eyed males	$X Y$

Conclusion: The white-eyed females and red-eyed males in the F_1 result from nondisjunction of the X chromosomes in an XXY female.

Red-green Colour Blindness

- Cone cell pigments (3)
 - Blue – chromosome 7
 - Green and Red – close together on X
- Sex-linked, recessive trait



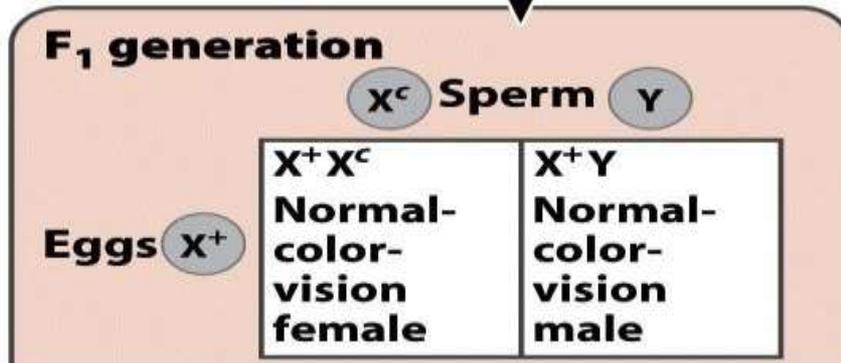
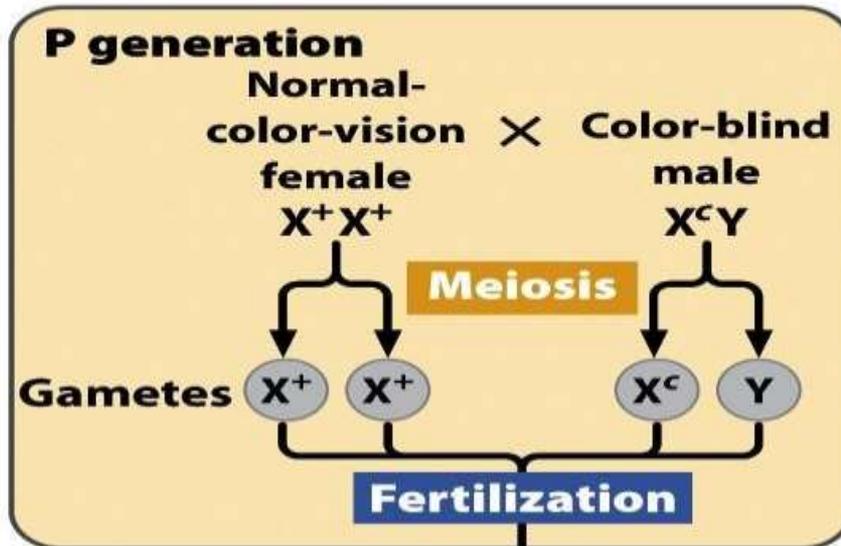
"The green dot indicates it's just been sold to someone colour-blind..."



X^+ - normal (wild-type)

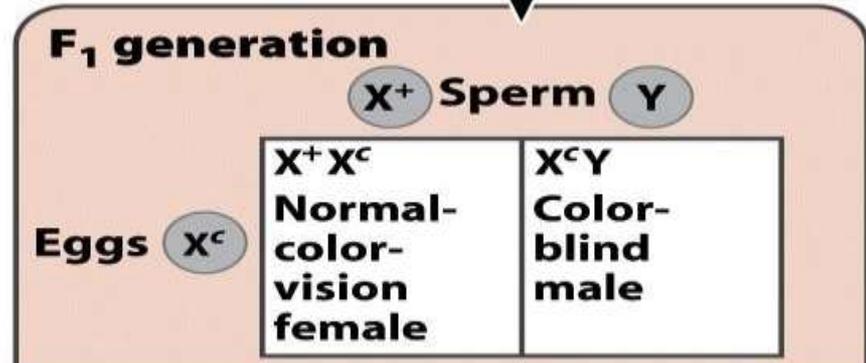
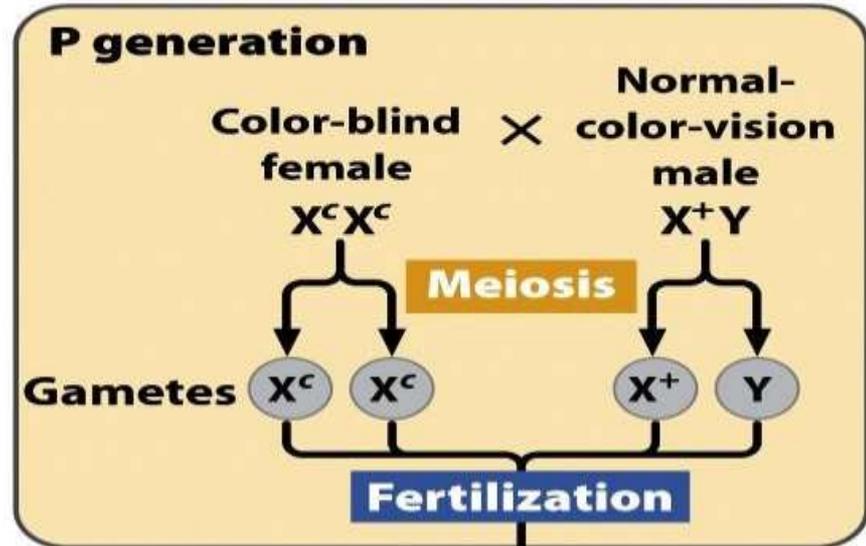
X^c - color-blindness

(a) Normal female and color-blind male



Conclusion: Both males and females have normal color vision.

(b) Reciprocal cross



Conclusion: Females have normal color vision, males are color blind.

Colour-blindness

- Affected woman passes the trait to sons
- Affected men pass trait to grandsons through daughters but never to sons
- X-linked recessives may appear to alternate between sexes
 - Females in one generation
 - Males in next generation

Dosage Compensation

- Different numbers of X chromosomes in males and females – potential problem.
 - the amount of protein produced by X-linked genes would differ in the two sexes.
 - Females would produce twice as much
 - This difference could be highly detrimental
- Dosage compensation:
 - e.g. in fruit flies - double the activity of the X linked genes in males
 - *Caenorhabditis elegans* – halve the activity of X linked genes in females
 - Placental mammals – inactivate genes on one X

Barr body – condensed, dark staining body in the nuclei of female cats – inactive X chromosome

- Females functionally hemizygous at cellular level – proteins from both Xs produced, but not in the same cell
- Random X-inactivation occurs early in development – mitotic divisions (remains inactive) – can get spotty distributions

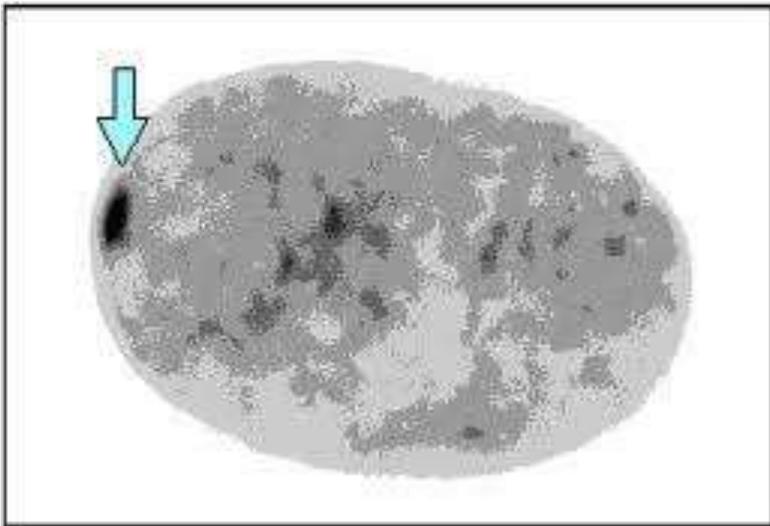


Fig 4.17

THANK

YOU

