Sex Determination and Sex-Linked Characteristics

Chapter 4

Lecture Outline

- Mechanisms of Sex Determination
  - Chromosomal
  - Genetic
  - Environmental
- Sex-linked Characteristics

Sex Determination

- Sexual reproduction is the results of meiosis and fertilization
- Sexual phenotypes (the sexes) – male and female
  - Differ in gamete size
- Sex determination – mechanism by which sex is established
- Monoecious – organisms with both male and female reproductive structures (hermphroditism)
- Dioecious – organism has male or female reproductive structures
  - Chromosomal, genetic or environmental sex determination

- In many organisms, sex is determined by a pair of chromosomes – sex chromosomes
  - Non-sex chromosomes = Autosomes

- Heterogametic sex – gametes differ with respect to sex chromosomes

- Homogametic sex – gametes are the same with respect to sex chromosomes

Concept Check 1

How does the heterogametic sex differ from the homogametic sex?

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

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Chromosomal Sex-Determination

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

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

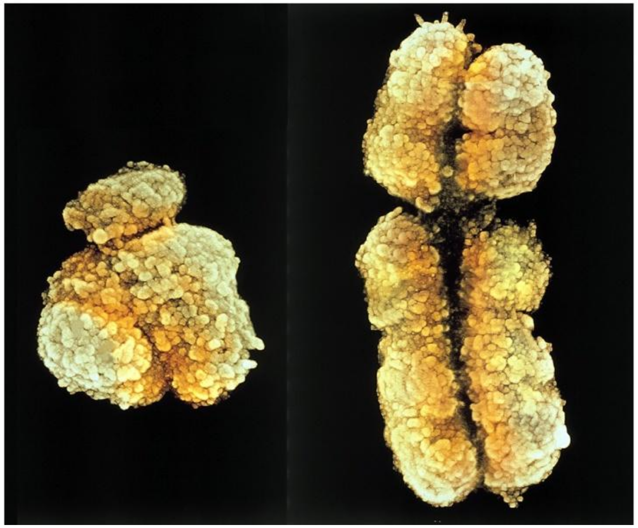


Fig 4.1

Fig 4.4

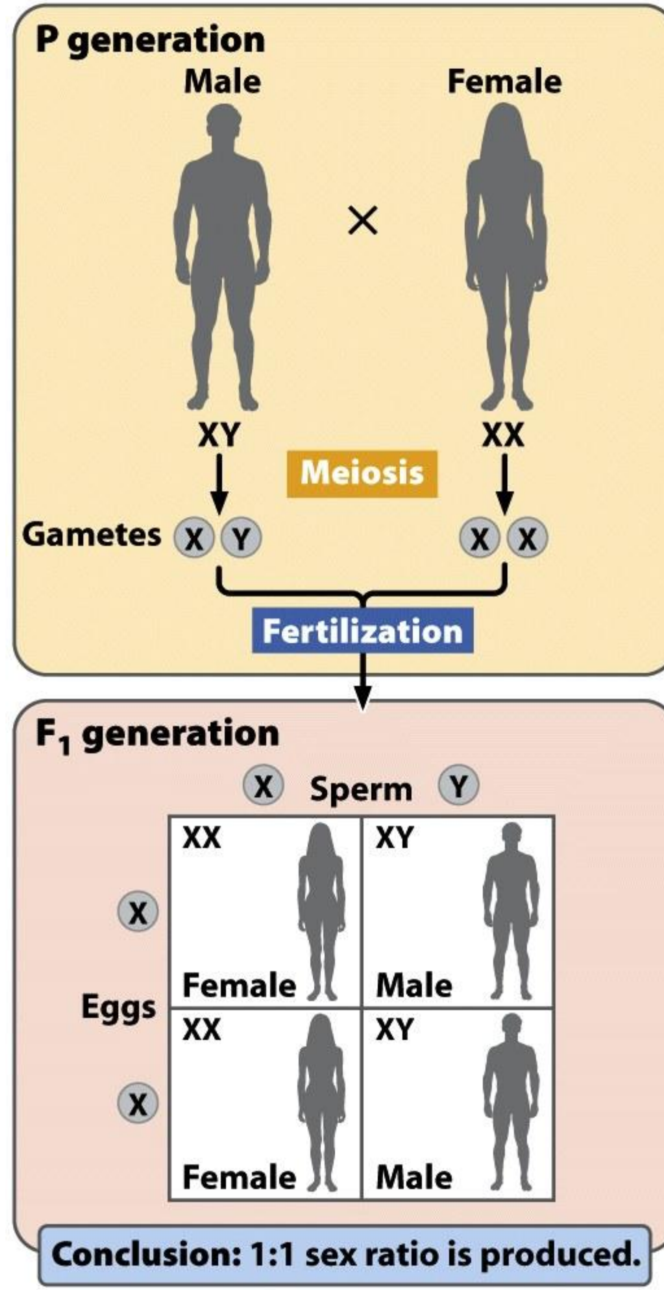
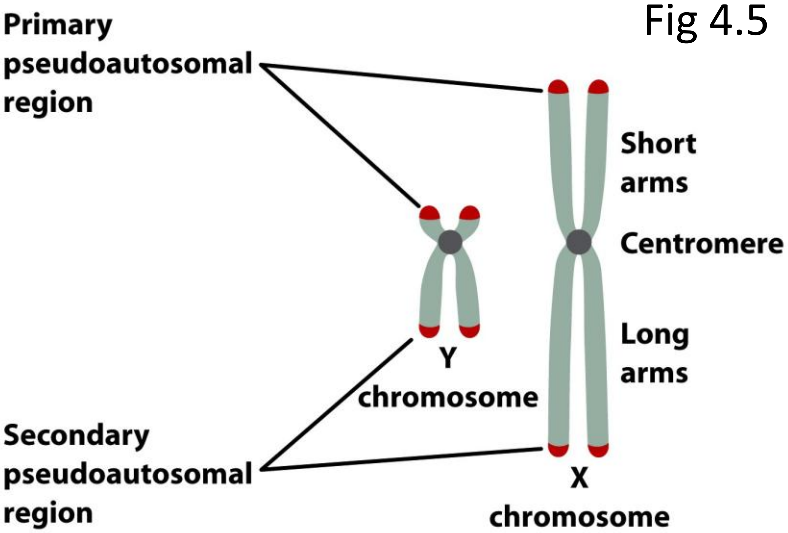


Fig 4.5

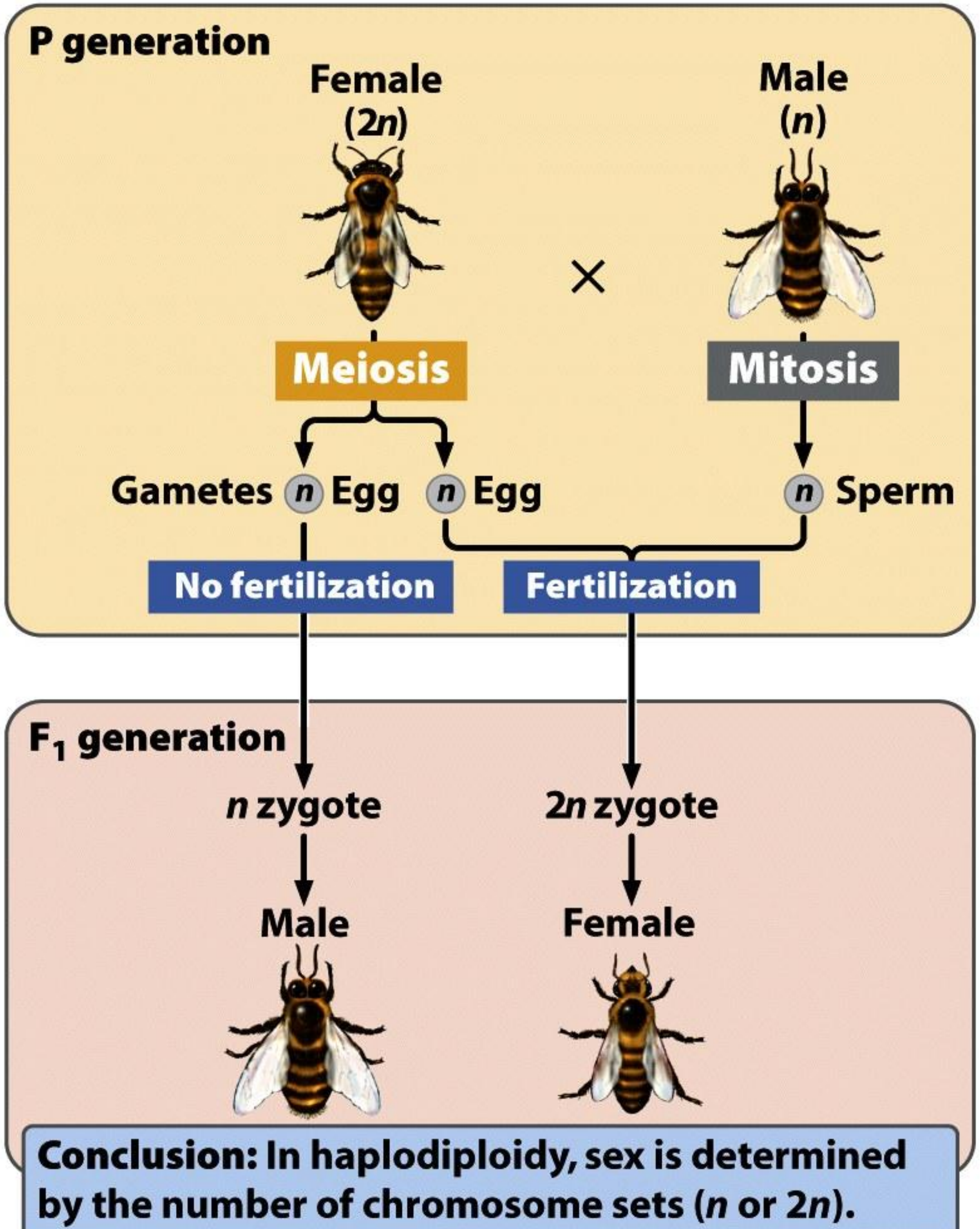


Chromosomal Sex-Determination

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

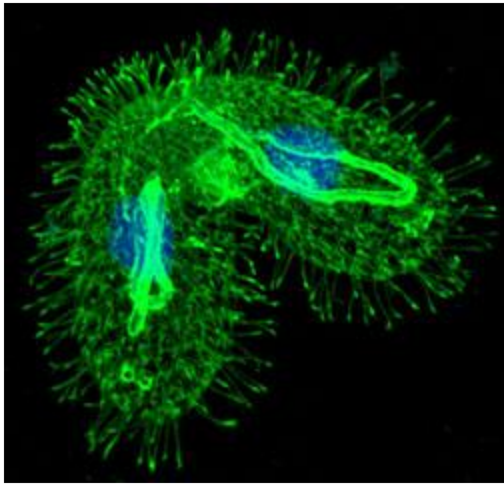
- Haplodiploidy – bees, wasps, ants
  - Unfertilized eggs, haploid set – male
  - Fertilized eggs, diploid set – female

Fig 4.6



P generation
Female
(2*n*)
Male
(*n*)
×
Meiosis
Mitosis
Gametes
n Egg
n Egg
n Sperm
No fertilization
Fertilization
F1 generation
n zygote
2*n* zygote
Male
Female
Conclusion: In haplodiploidy, sex is determined by the number of chromosome sets (*n* or 2*n*).

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

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

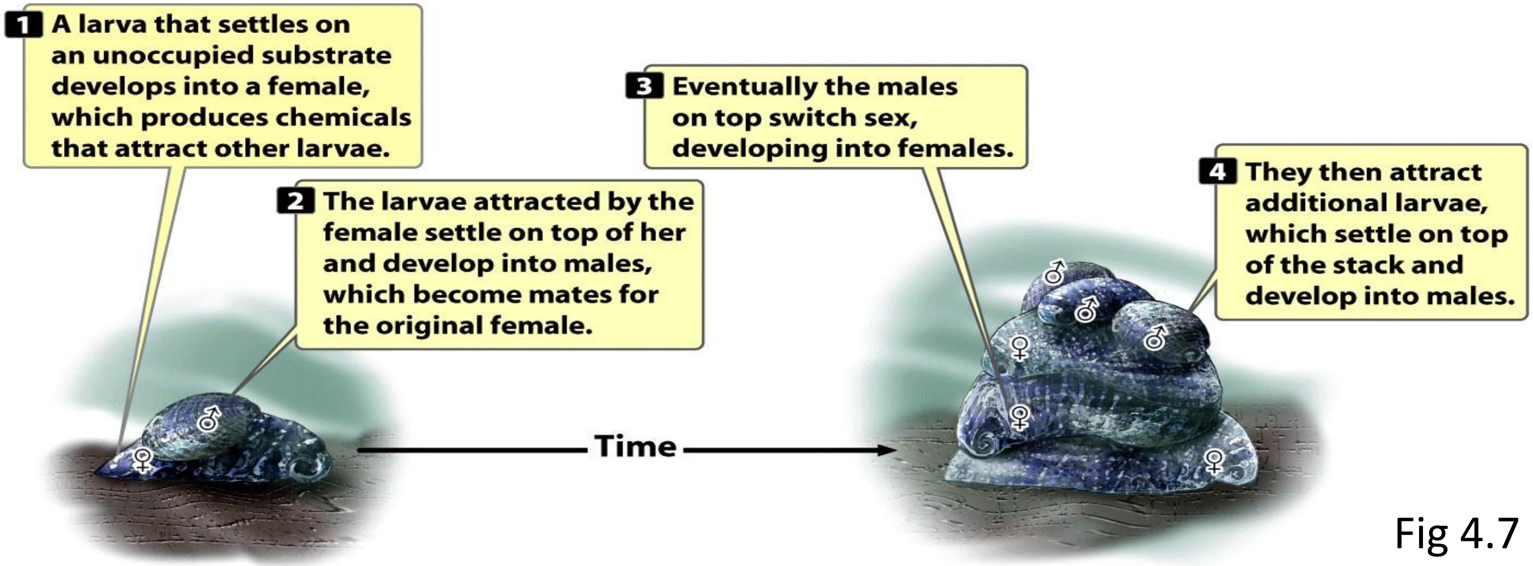
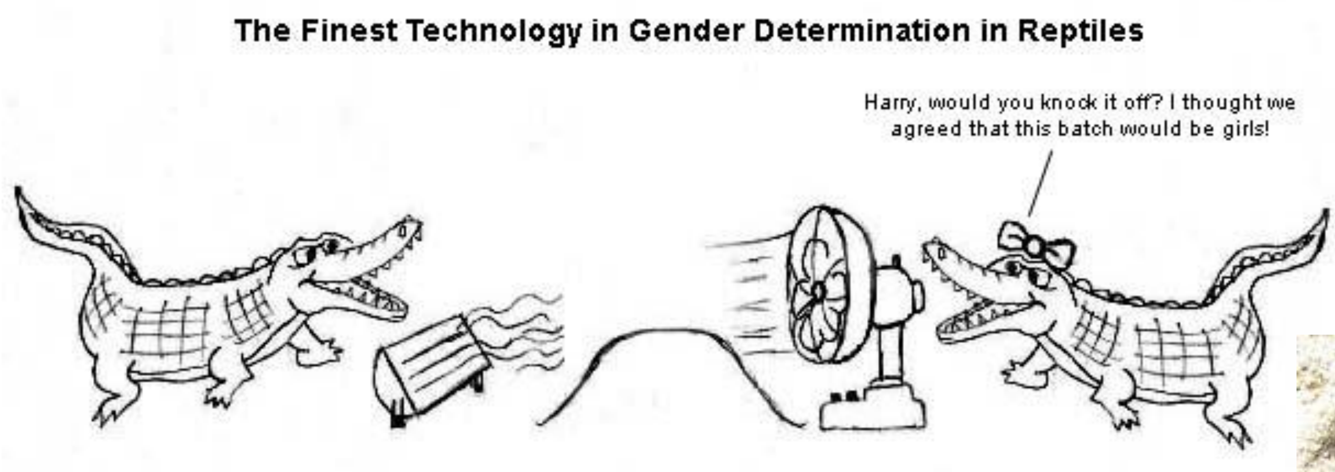


Fig 4.7

Environmental Sex Determination

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



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

Table 4.1 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*

Concept Check 2

What will be the sexual phenotype of a fruit fly with XXYYY sex chromosomes and two sets of autosomes?

a. male
b. female
c. Intersex
d. metamale

Concept Check 2

What will be the sexual phenotype of a fruit fly with XXYYY sex chromosomes and two sets of autosomes?

a. male
b. female
c. Intersex
d. metamale

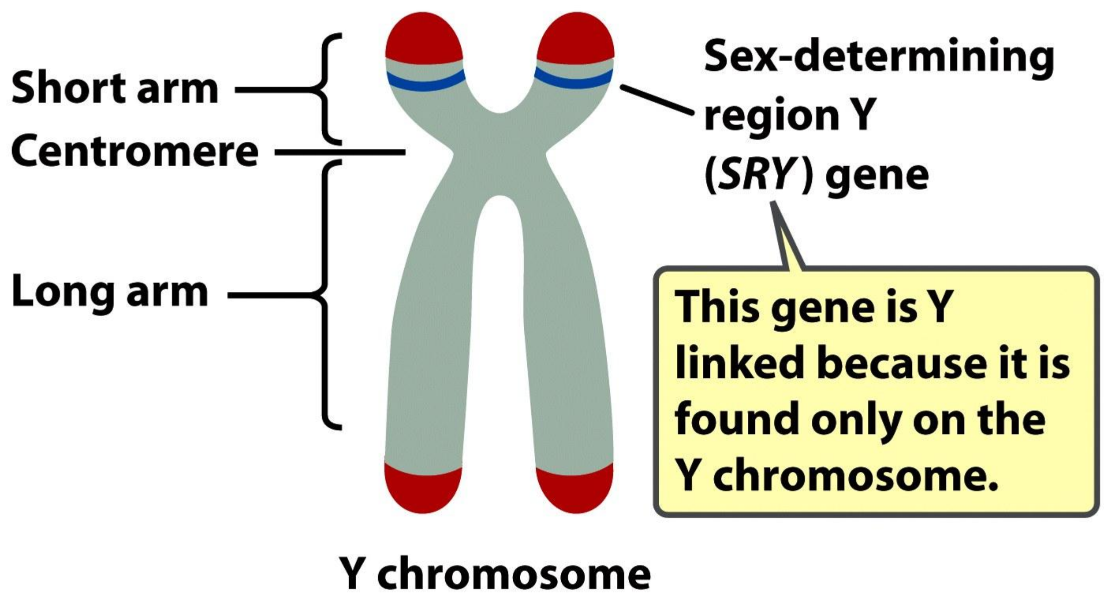
2X and 2 sets autosomes

X:A = 2:2 = 1.0

Sex Determination - Humans

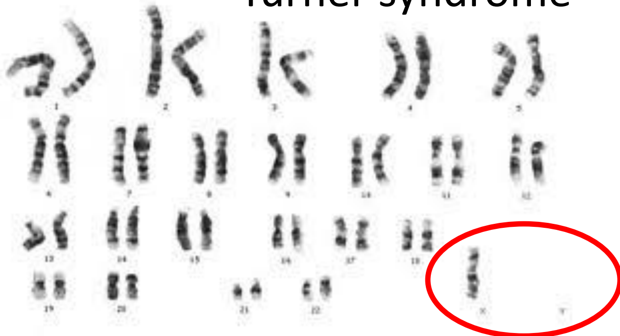
- XX-XY sex determination
- *SRY* gene on Y chromosome determines maleness

Fig 4-10



- Phenotypes from abnormal sex chromosome numbers illustrate the importance of *SRY*
  - Turner syndrome: XO; 1/3000 female births
  - Klinefelter syndrome: XXY, or XXXY, or XXXXY, or XXYY; 1/1000 male births
  - Poly-X females: 1/1000 female births

Turner syndrome



Role of Sex Chromosomes - Humans

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

SRY – sex determining region Y

- Rare males with XX?
  - Small piece of Y attached to another chromosome
- Y chromosome becomes active at week 6
  - Gonad tissue develops into testes
    - Produce testosterone and Mullerian-inhibiting substance
- *SRY* codes for protein that binds and bends DNA- affects expression of genes encoding testes formation
- Other genes also important for fertility and sexual characteristics

Androgen-insensitivity Syndrome

- Appear as normal females – but XY
  - Lack uterus, oviducts and ovaries
  - Testis in abdominal cavity, produce male testosterone levels
  - Androgen receptor is defective so cells insensitive to testosterone – develop female characteristics
  - Gene for receptor- on X so maternally inherited
- Genes for most secondary sex characteristics on autosomal chromosomes – key in control of expression

Inheritance of Sex-linked Characteristics

- Thomas Hunt Morgan – first to explain
  - White eyes in Drosophila

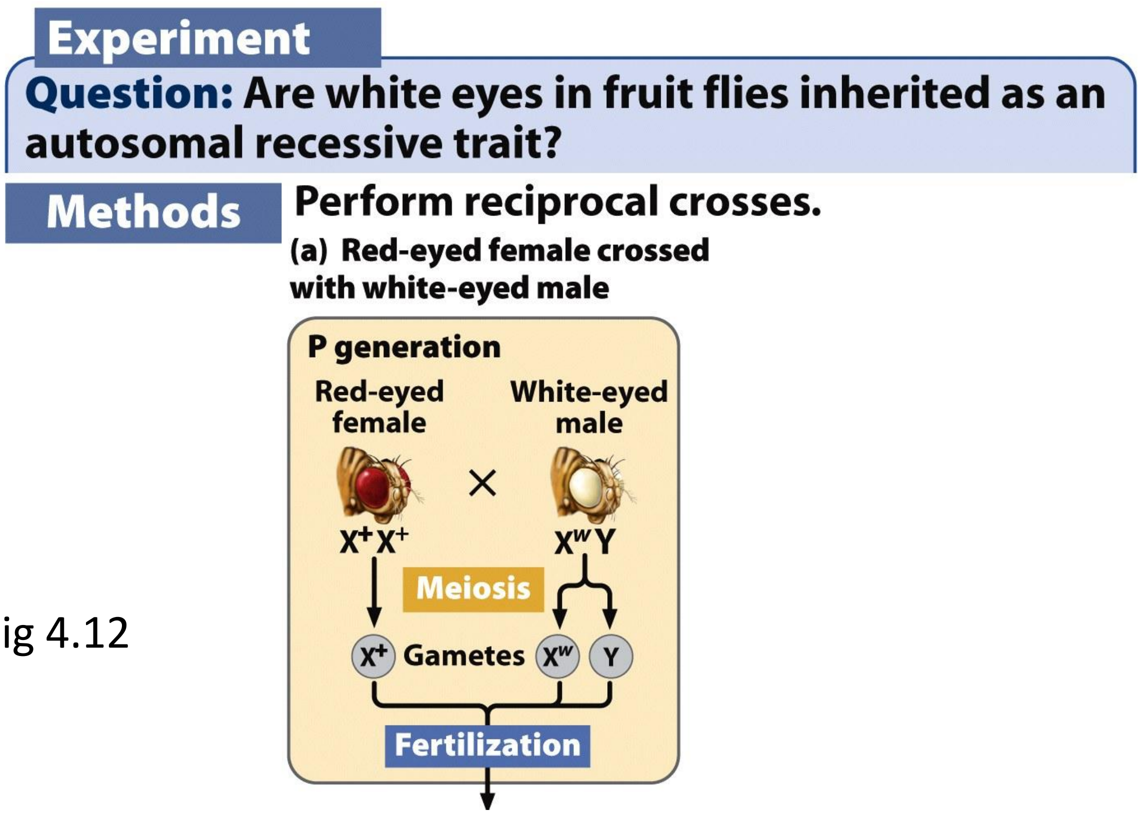
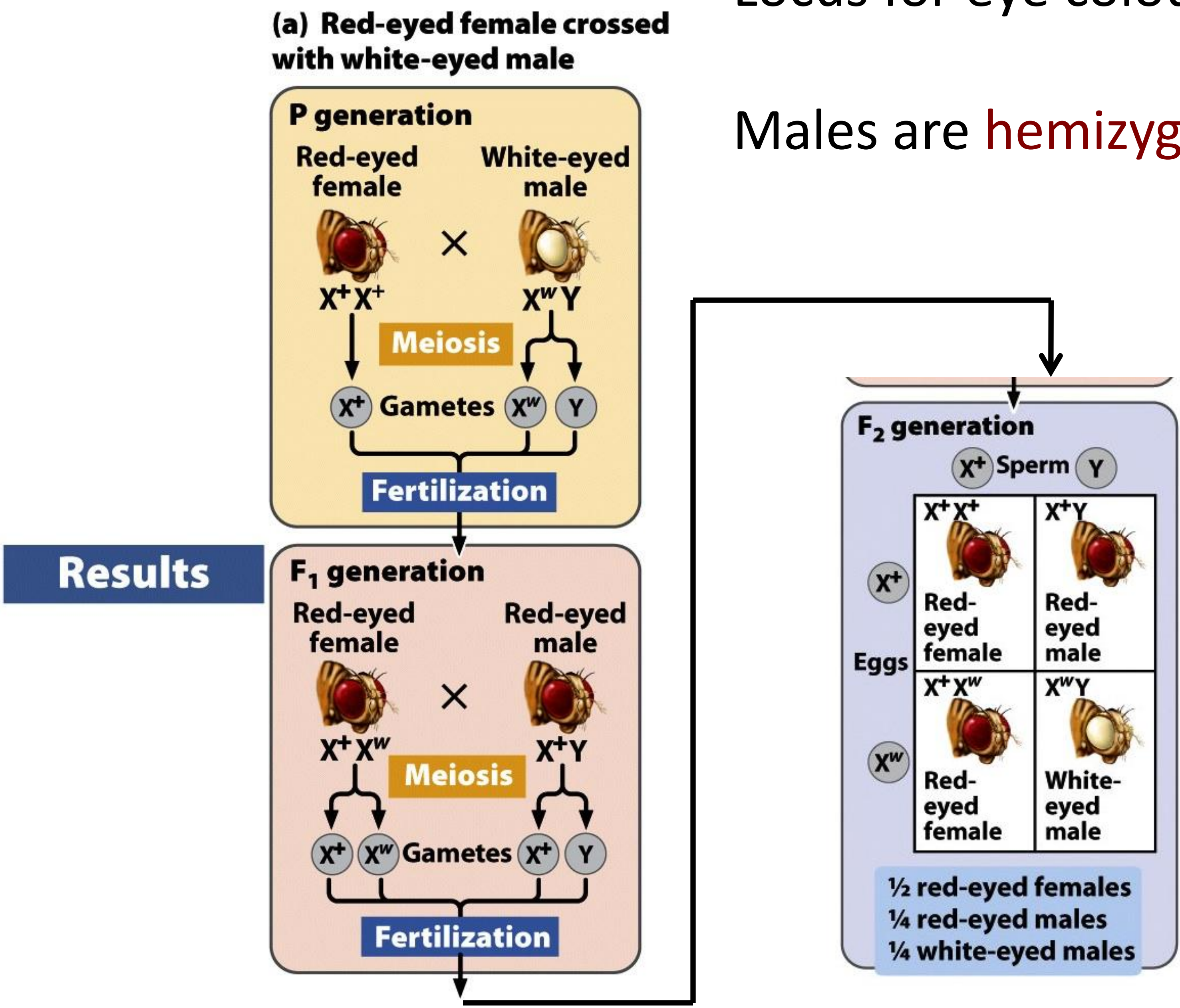


Fig 4.12

Fig 4.12

Locus for eye colour is X-linked

Males are hemizygous



Reciprocal cross

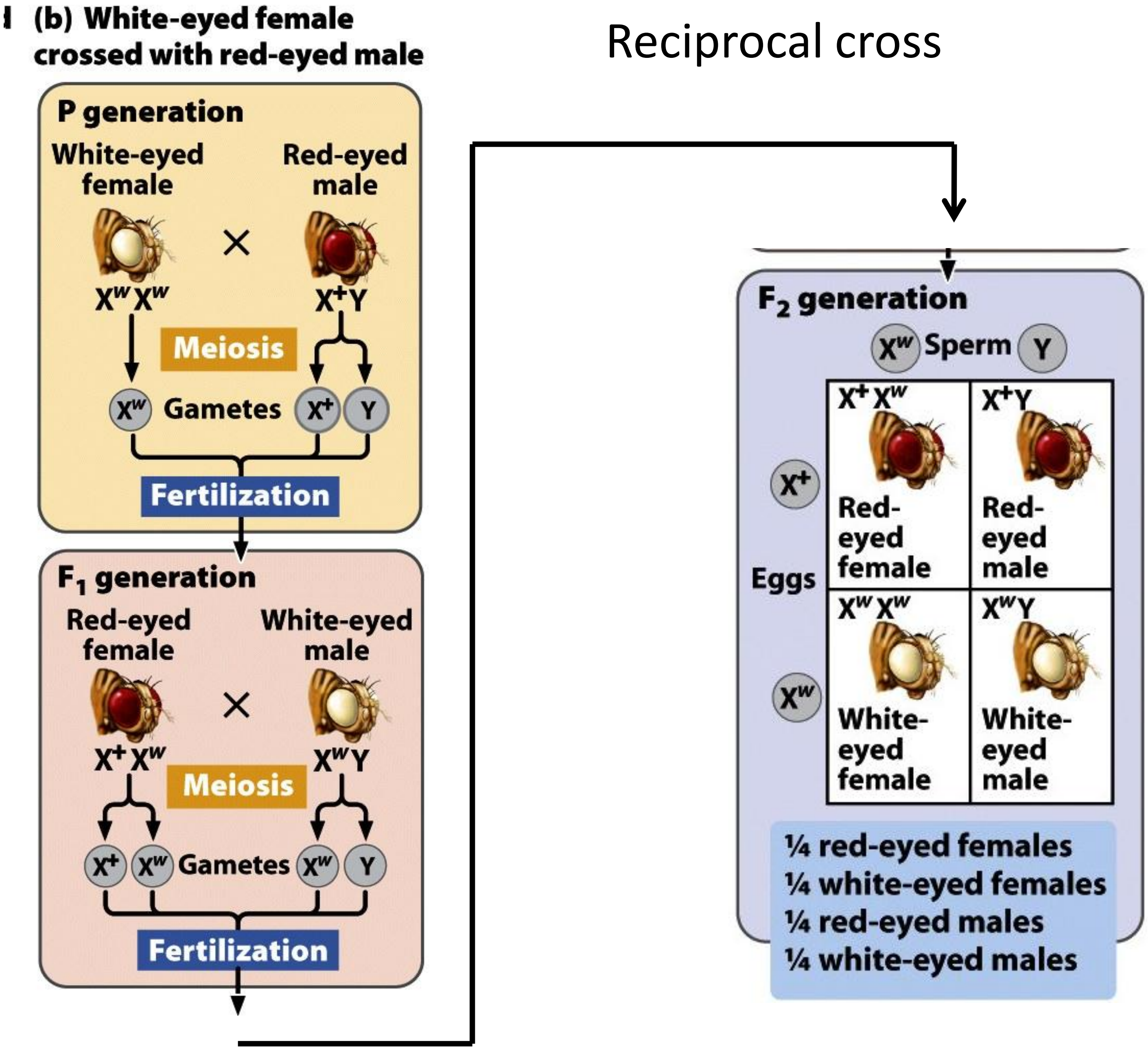


Fig 4.12

Reciprocal crosses – produced different results in F1 and F2

-consistent with X-linked inheritance

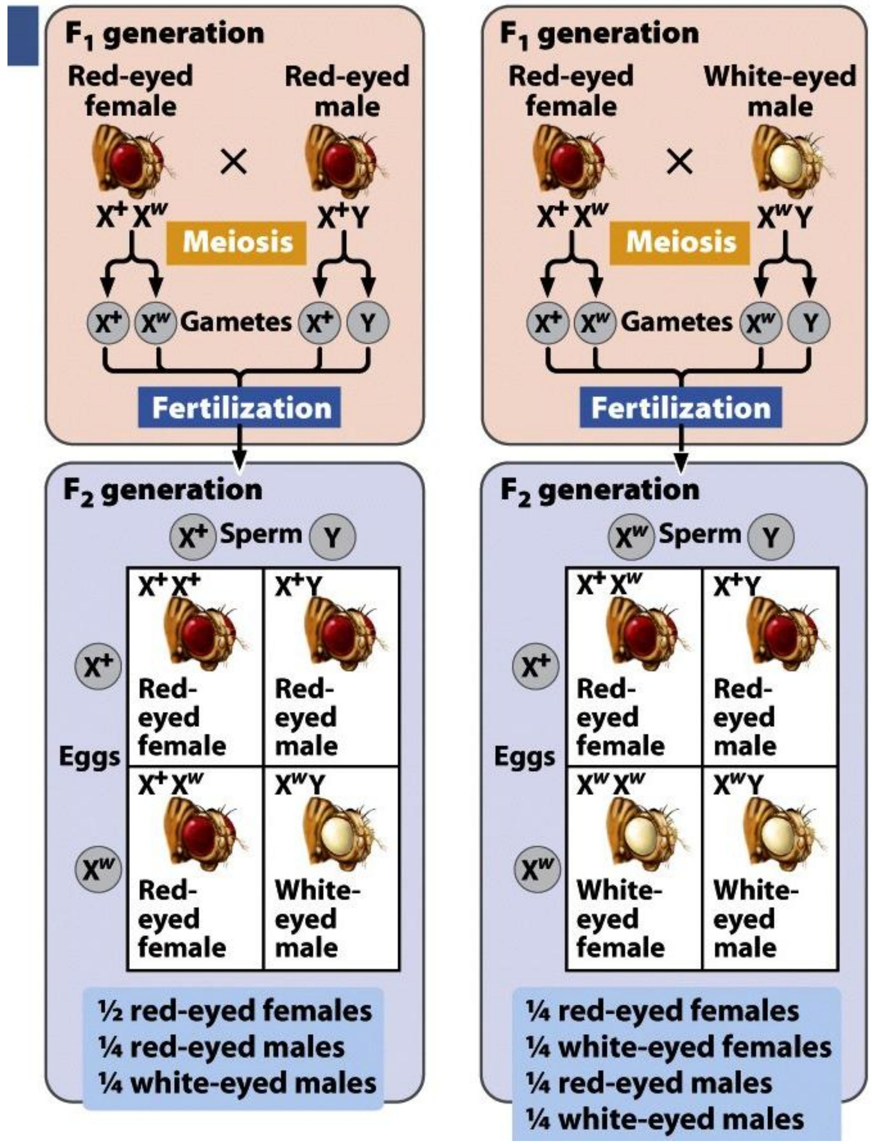


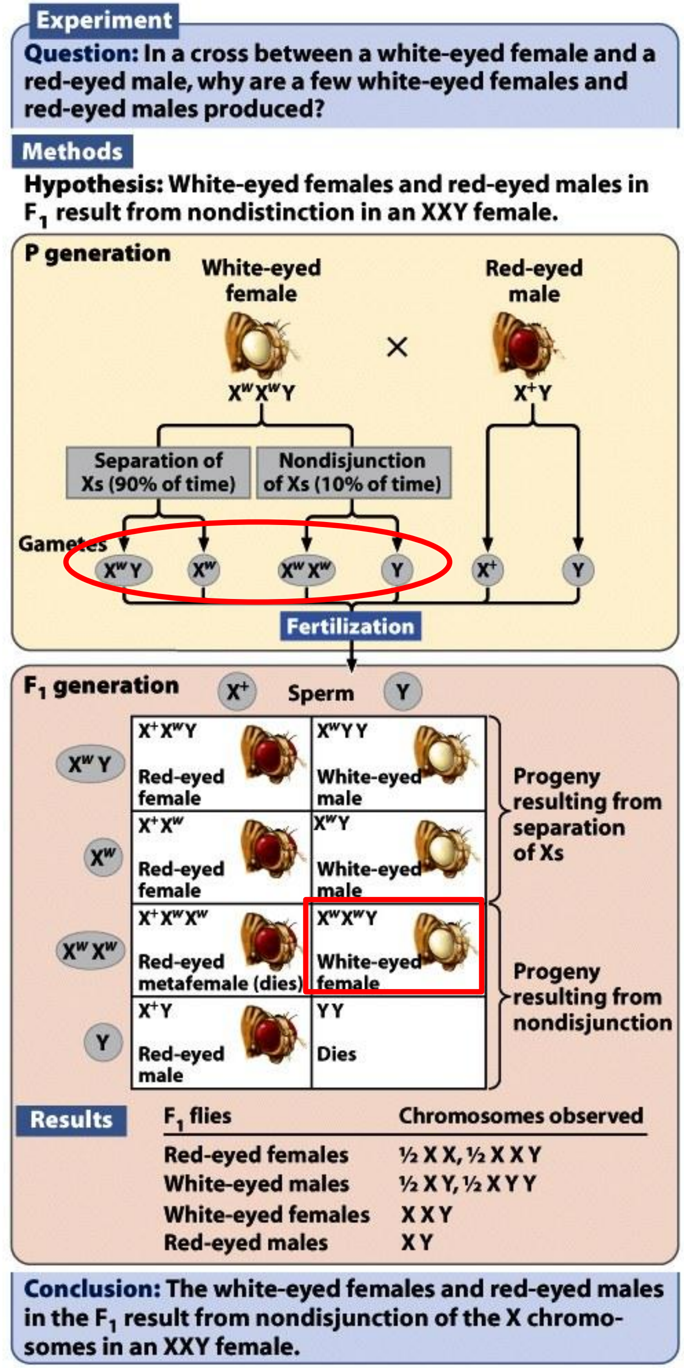
Fig 4.12

Non-disjunction

Chromosomes fail to separate in anaphase 1 – non-disjunction

Non-disjunction of 2Xs in XXY female flies

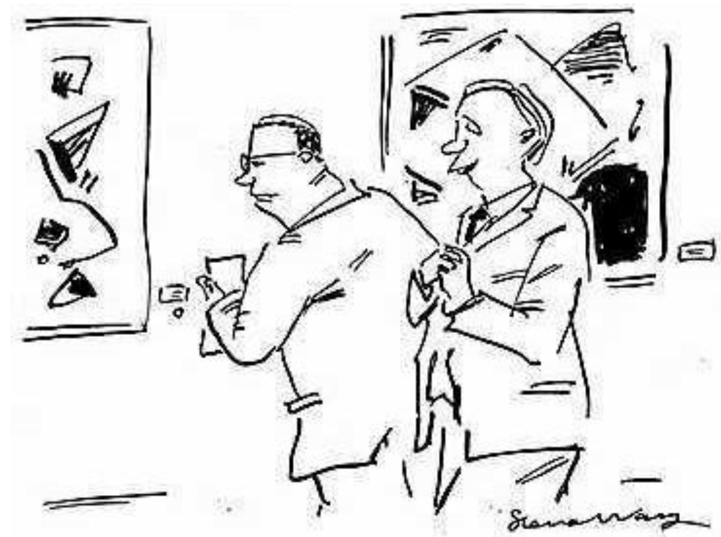
Confirms evidence of sex-linked gene for eye-color on the X chromosome



Red-green Colour Blindness

- Cone cell pigments (3)
  - Blue – chromosome 7
  - Green and Red – close together on X

- Sex-linked, recessive trait

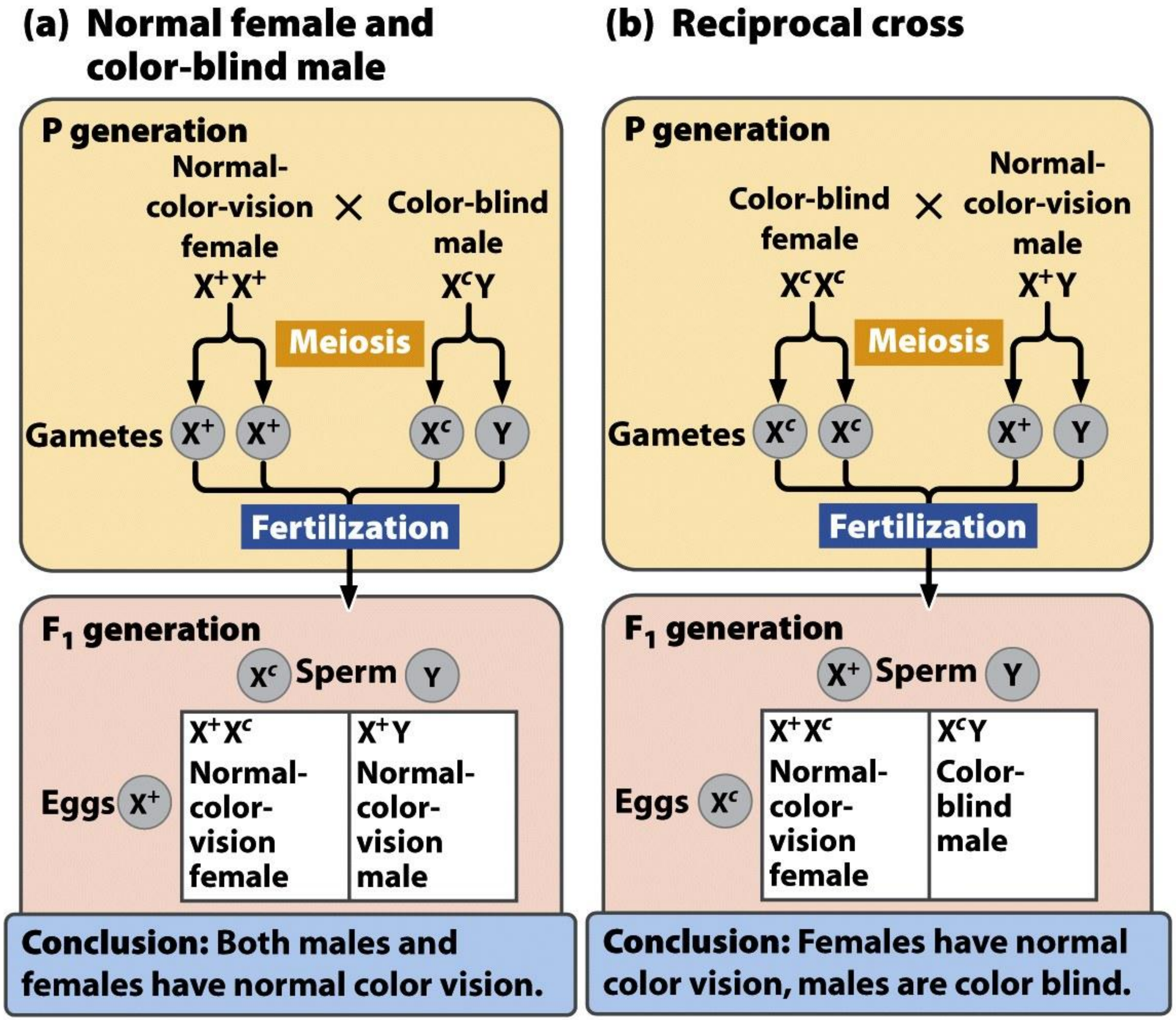


"The green dot indicates it's just been sold to someone colour-blind..."

X^+ - normal (wild-type)

X^c – color-blindness

Fig 4.15



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

Concept Check 4

Hemophilia (reduced blood clotting) is an X-linked recessive disease in humans. A woman with hemophilia mates with a man who exhibits normal blood clotting.

1) What is the probability that their child will have hemophilia?

2) What is the probability that their male child will have hemophilia?

Concept Check 4

Hemophilia (reduced blood clotting) is an X-linked recessive disease in humans. A woman with hemophilia mates with a man who exhibits normal blood clotting.

1) What is the probability that their child will have hemophilia? 1/2

2) What is the probability that their male child will have hemophilia? 100%

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

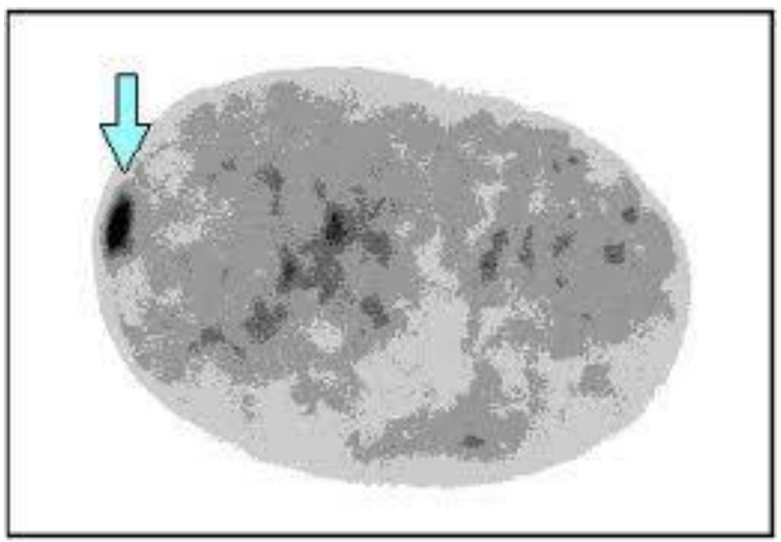


Fig 4.17

Z-linked Characteristics

- The same as in X-Y
  - Patter of inheritance in males and females is reversed
  - Females are heterogametic sex

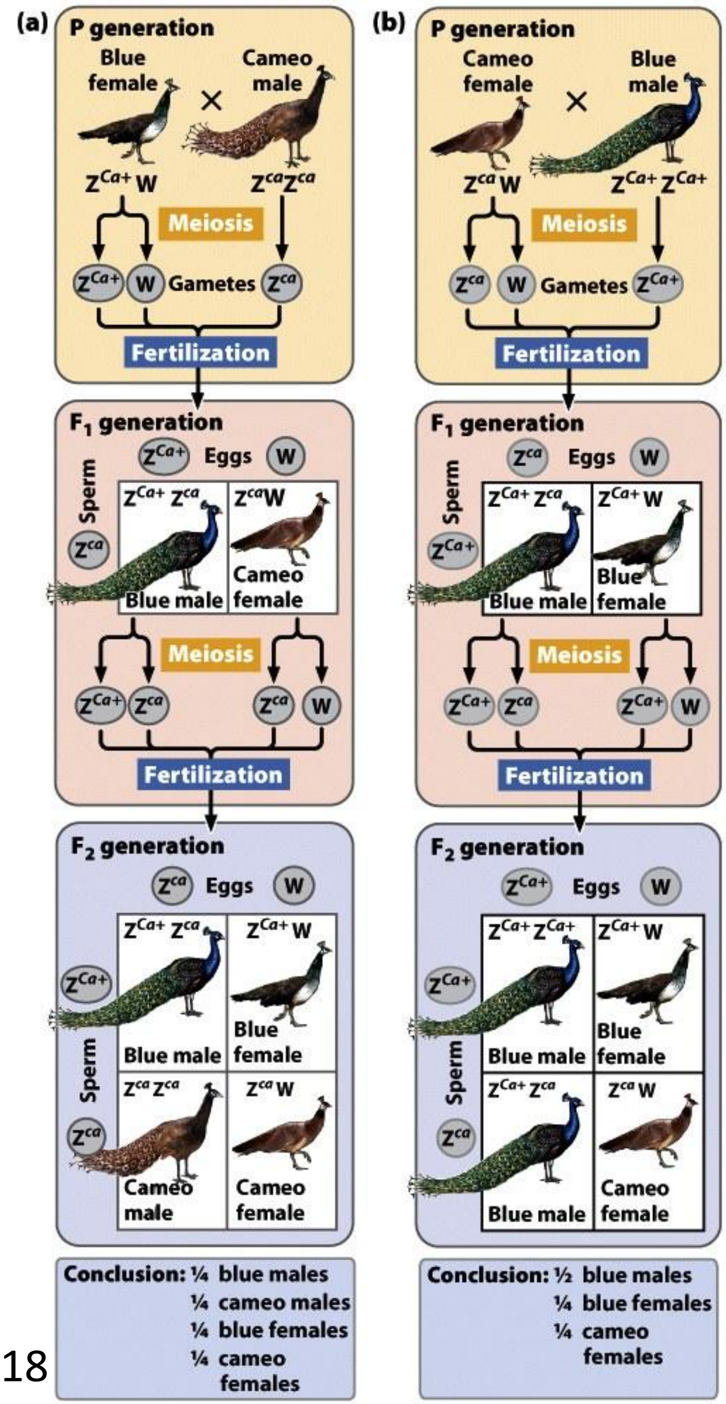


Fig 4.18

Y-linked Characteristics

- Only about 150 genes on Y
  - Many influence male sexual development and fertility
  - Function of most poorly understood
  - Useful for determining genetic relationships
    - e.g. origins of Lemba – African tribe in South Africa and Zimbabwe – Judaic origin

Chapter 4 Questions

- Comprehension Questions: 1-15
- Application Questions: 16-18, 20-27, 29-34
- Challenge Questions: 42, 45