Sex Determination in Drosophila & Human

Dr. Kaustav Chakraborty

Assistant Professor (W.B.E.S.)
D/o Zoology
S.B.S. Govt. College, Hili

Chromosomal Sex-Determination Systems:

- **XX-XO system**:
  - XX – female
  - XO – male

  Example: Grasshoppers

- **XX-XY system**:
  - XX – female
  - XY – male

  Example: Mammals

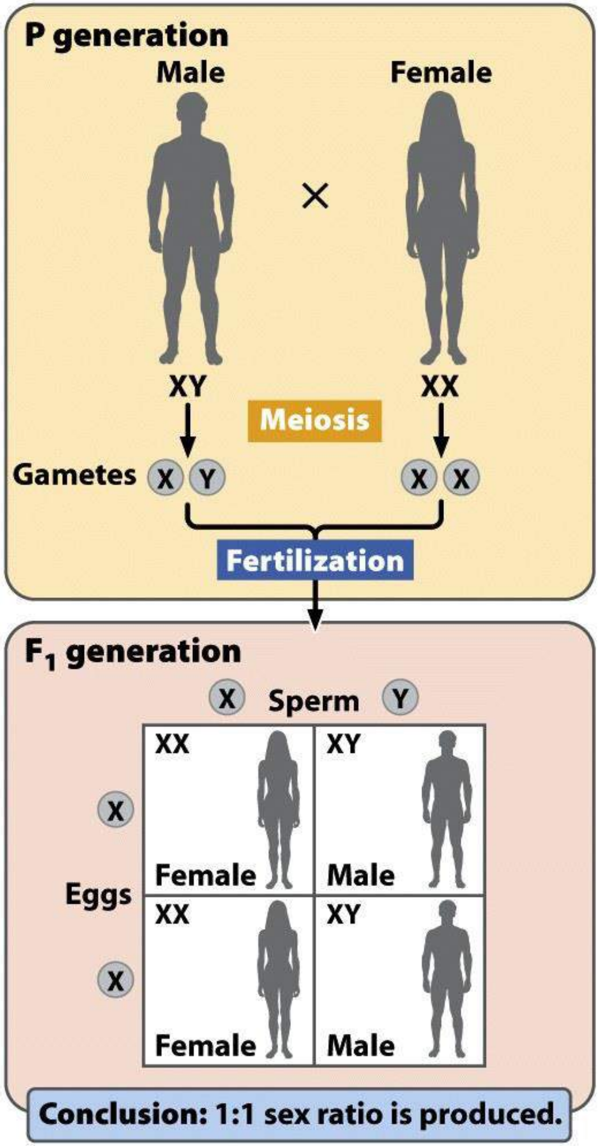

Figure 4-4
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Chromosomal Sex-Determination Systems

- **ZZ-ZW system**:
  - ZZ – male
  - ZW – female

  Example: Birds, snakes, butterflies, some amphibians, and fishes.
- **Haplodiploidy system**:
  - Haploid set – male
  - Diploid set – female

  Example: Bees, wasps, and ants

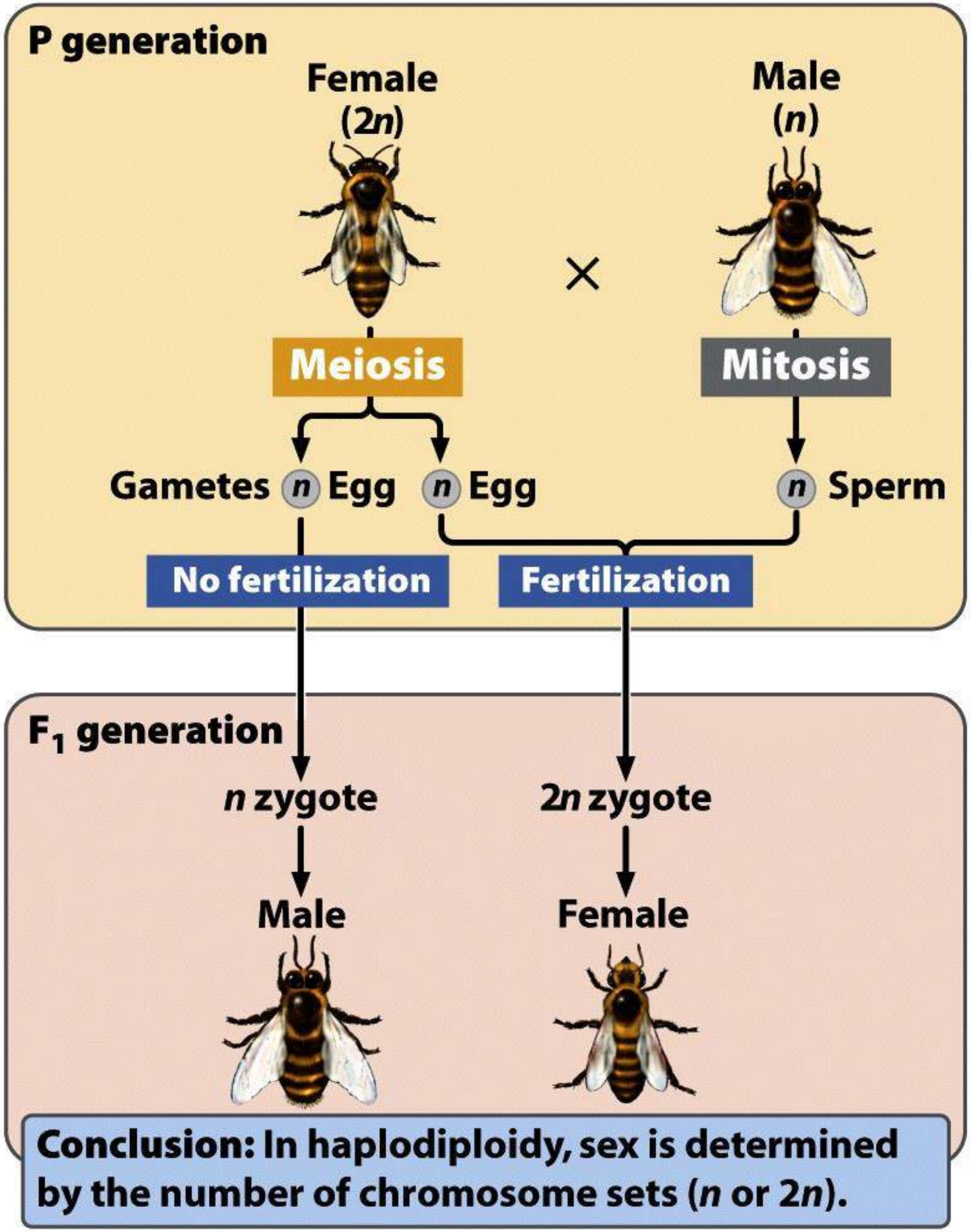

Figure 4-6

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Sex Determination in *Drosophila melanogaster*

- Sex in Drosophila is determined by the ratio of number of X chromosomes (X) to that of the number of sets of autosomes (A) - **Genic Balance System**, proposed by **Calvin Bridges**, 1926.

- 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

Table 4-1
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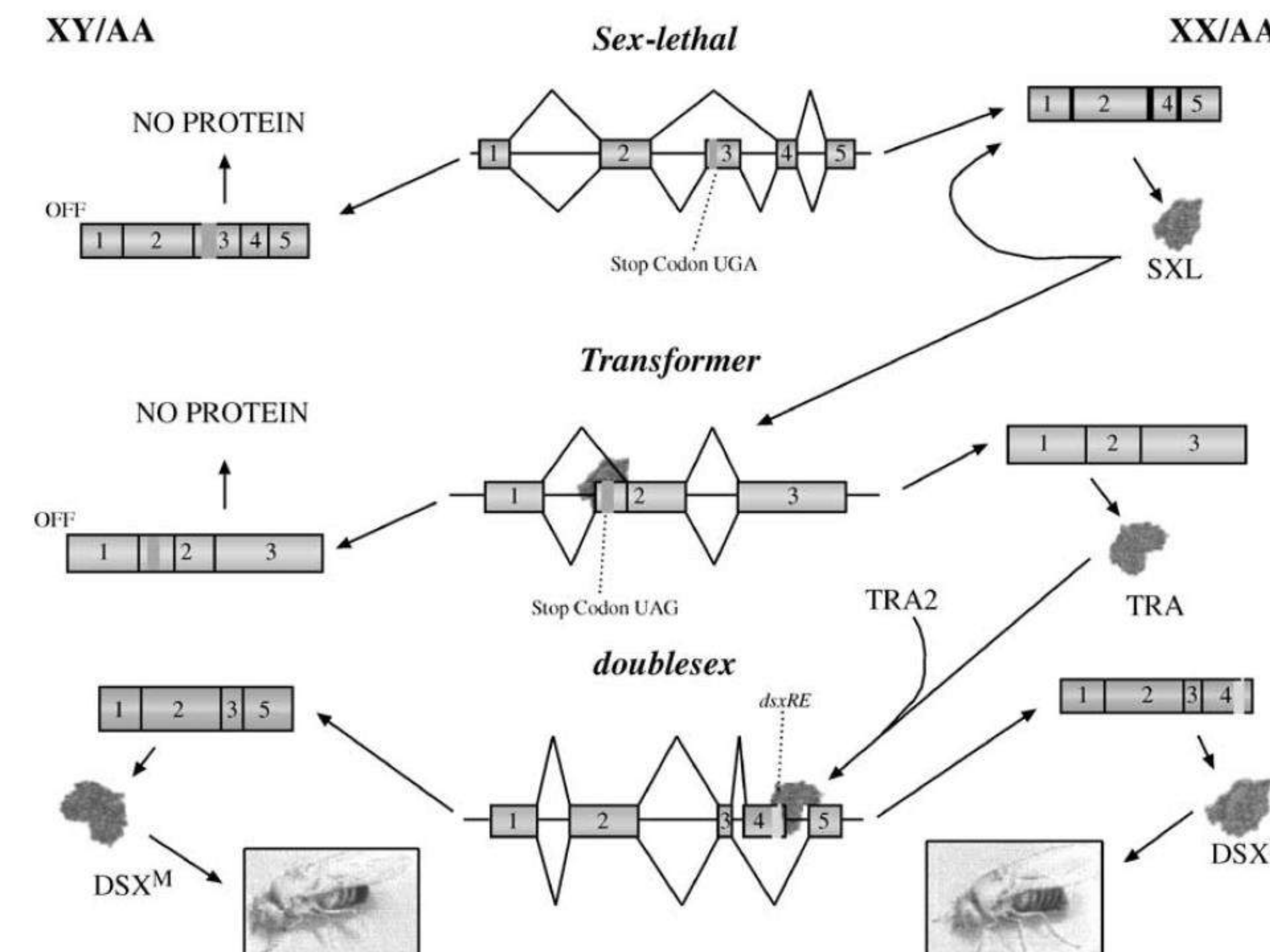
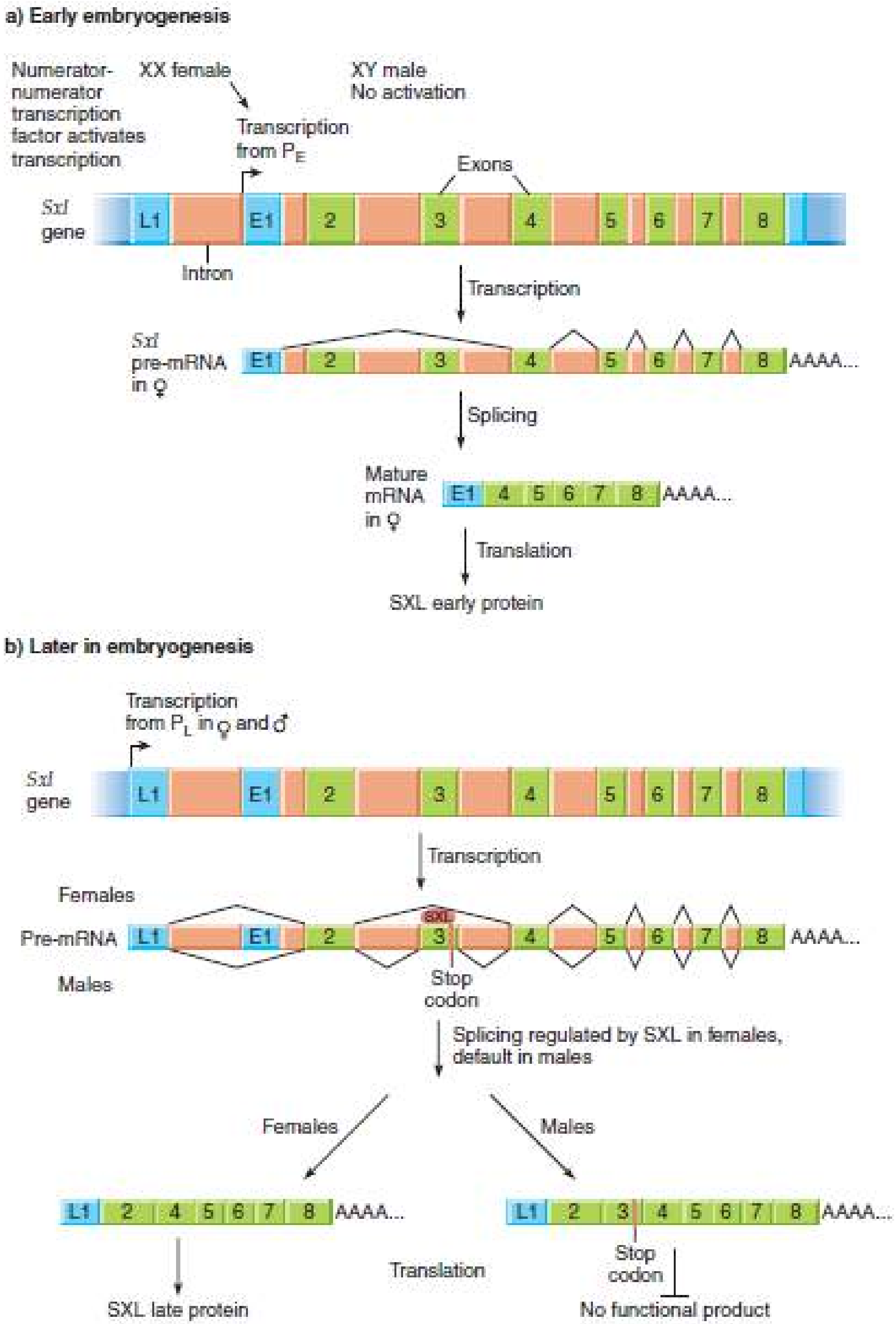
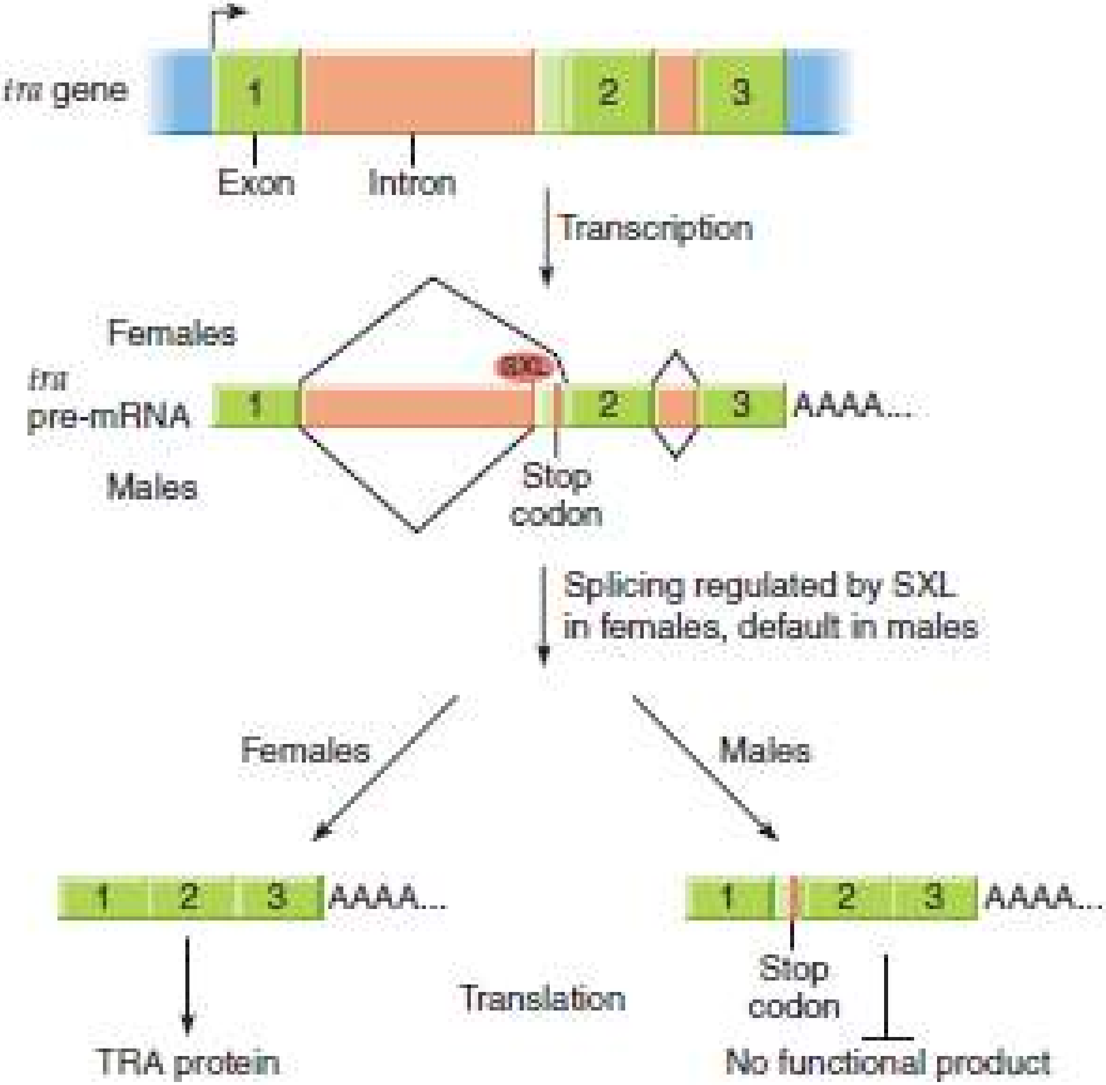
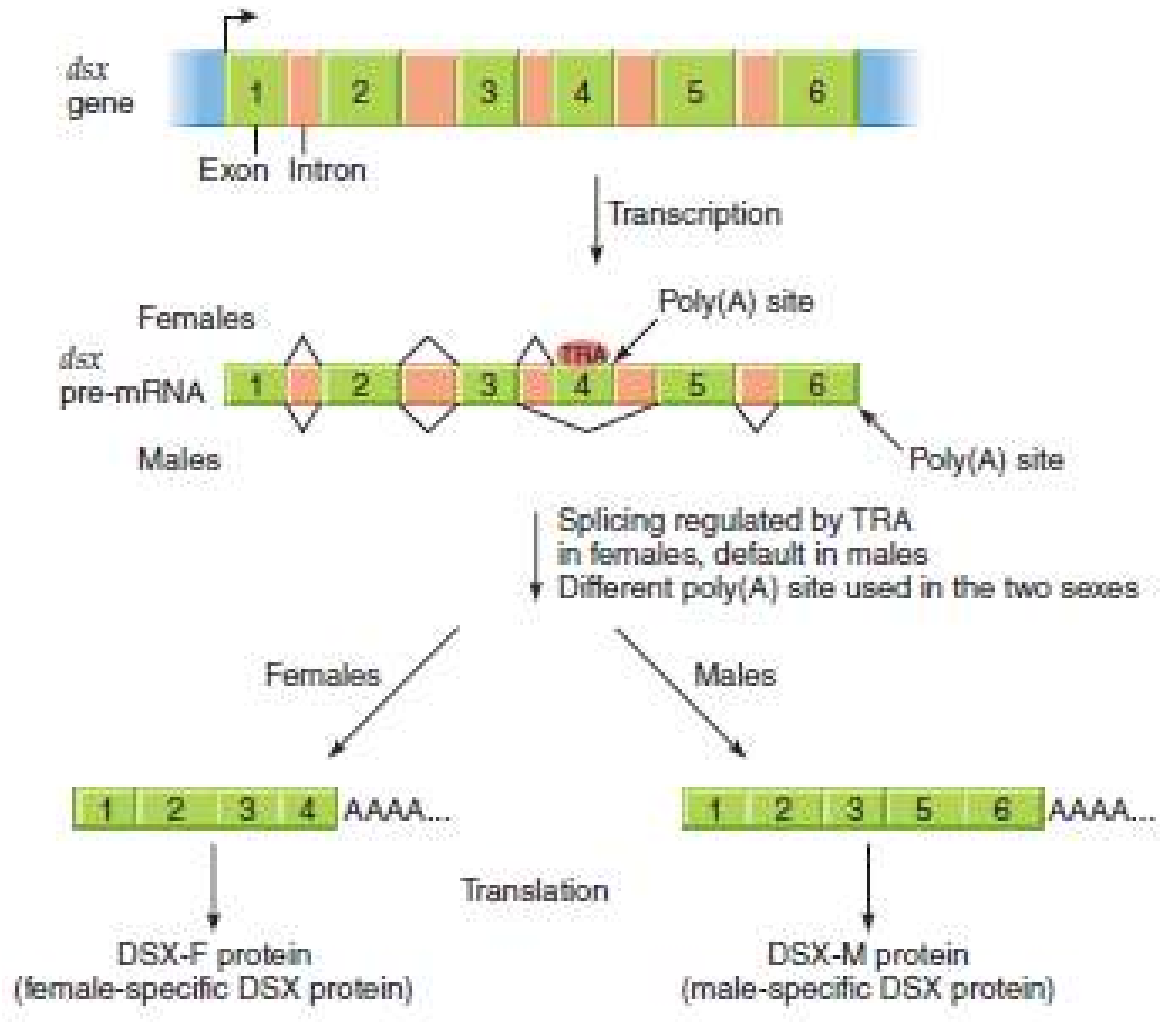

XY/AA
Sex-lethal
XX/AA
NO PROTEIN
OFF
1 2 3 4 5
Stop Codon UGA
SXL
Transformer
NO PROTEIN
OFF
1 2 3
Stop Codon UAG
TRA2
TRA
doublesex
dsxRE
1 2 3 5
1 2 3 4
1 2 3 4 5
DSX^M
DSX



Role of Sex-lethal (sxl) during embryogenesis.



Expression of transformer (tra) during embryogenesis.



Expression of doublesex (dsx) during embryogenesis.

Sex Determination in Human (XX-XY)

- **SRY (sex-determining region Y)** gene on the short arm of the Y chromosome determines maleness.

- SRY encodes a gene product that somehow triggers the undifferentiated gonadal tissue of the embryo to form testes – **testis-determining factor (TDF)**.

- Autosomal genes such as **SOX9, SF-1, WT-1** are believed to be part of a cascade of genetic expression initiated by SRY.

The male-determining gene in humans

- Sex-determining region Y (SRY) gene

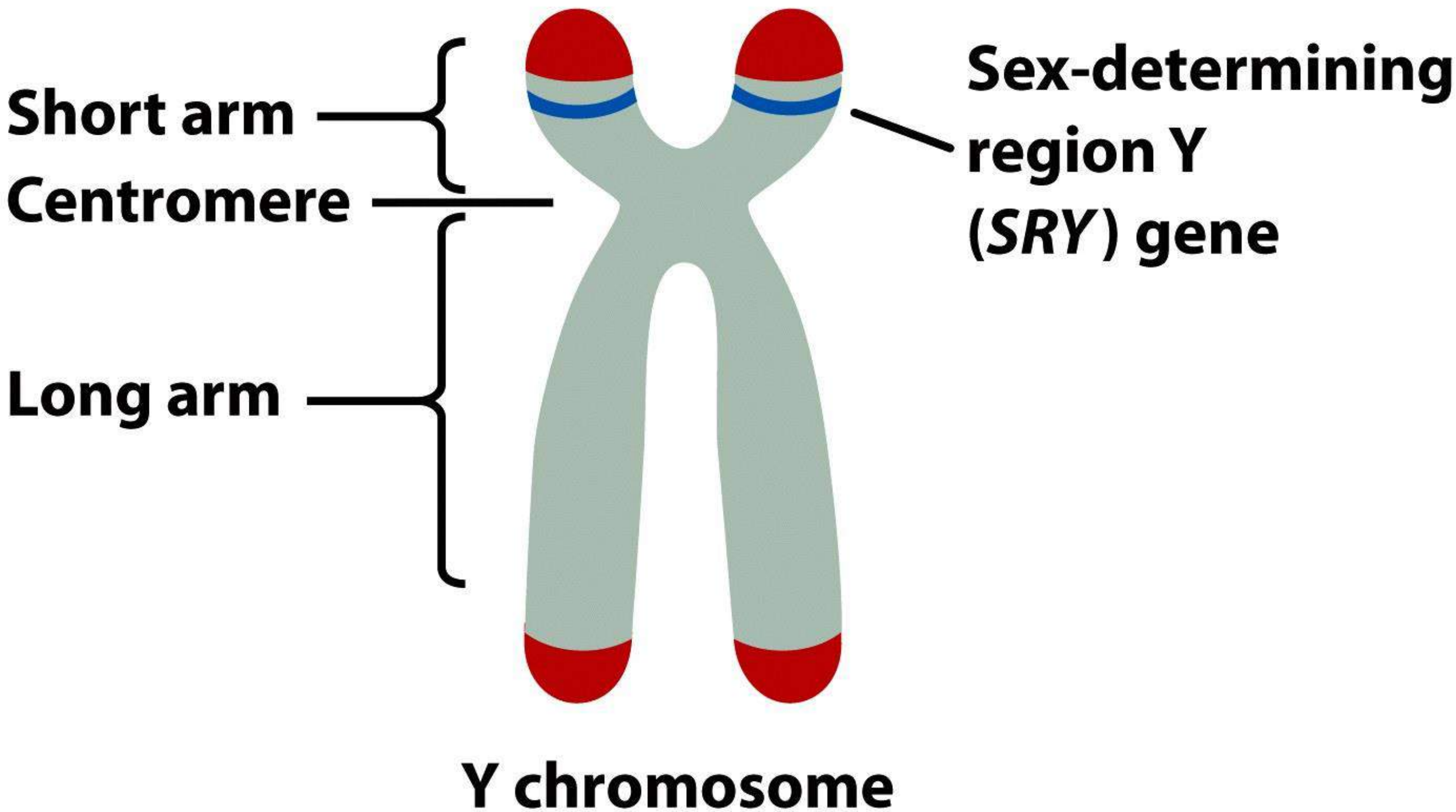


Figure 4-10
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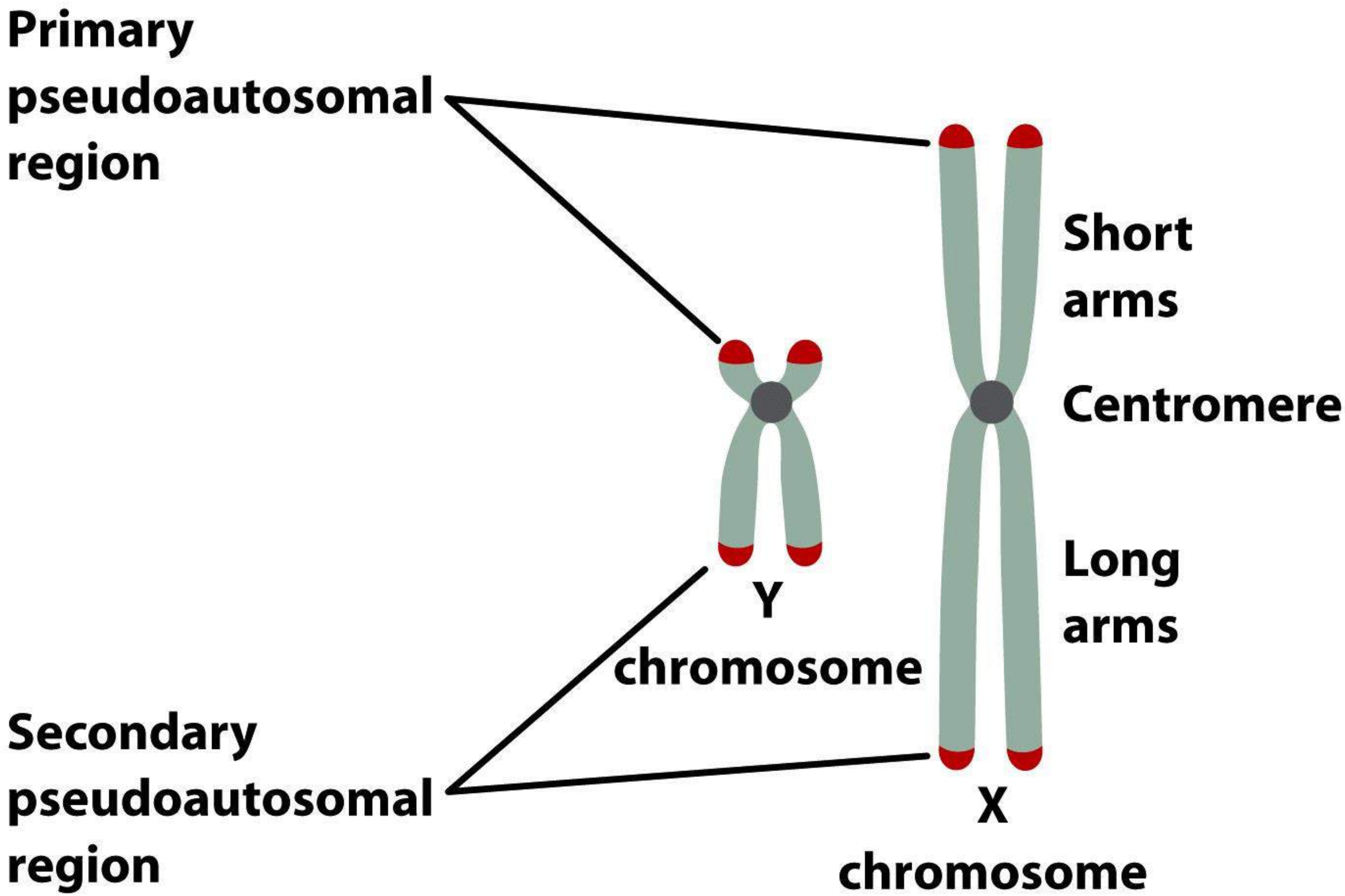

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The Role of Sex Chromosomes

- The X chromosome contains genetic information essential for both sexes; at least one copy of an X is required.

- The male-determining gene is located on the Y chromosome. A single Y, even in the presence of several X, still produces a male phenotype.

- The absence of Y results in a female phenotype.

XIC (X-Inactivation Centre)

- Cytogenic location: Xq13.2

- Genetic expression of XIC supports inactivation of X-chromosome.

- 1 Mb in length, contains several putative regulatory units.

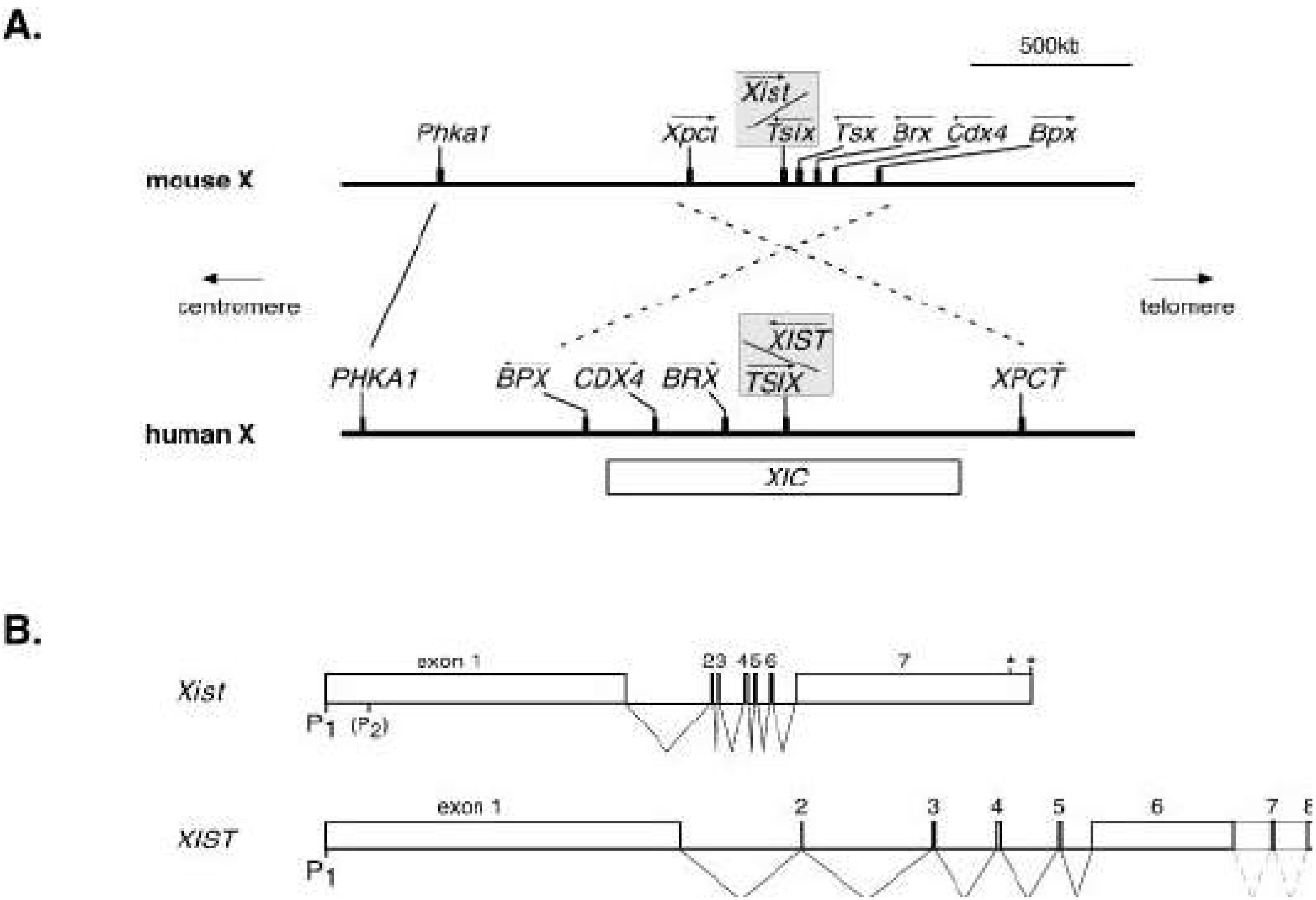
- https://ghr.nlm.nih.gov/gene/XIST#location

XIST (X-Inactive Specific Transcript)

- RNA product of this gene is quite large & lacks ORF (open reading frame – includes the info necessary for translation of RNA product into protein). So, RNA is not translated, leads to chr. Inactivation.

- RNA products of XIST spread over & coat the X-chromosome bearing the gene that produced it, leading to inactivation (cis acting).

- Transcription of XIST occurs initially at low levels on all X-chr. As the inactivation process begins; enhanced only in the X-chromosome that becomes inactivated.


A.
500kb
Xist
Tsix
Phka1
Xpct
Tsx
Brx
Cdx4
Bpx
mouse X
centromere
telomere
XIST
TSIX
PHKA1
BPX
CDX4
BRX
XPCT
human X
XIC
B.
Xist
exon 1
2 3
4 5 6
7
P1
(P2)
XIST
exon 1
2
3
4
5
6
7
P1

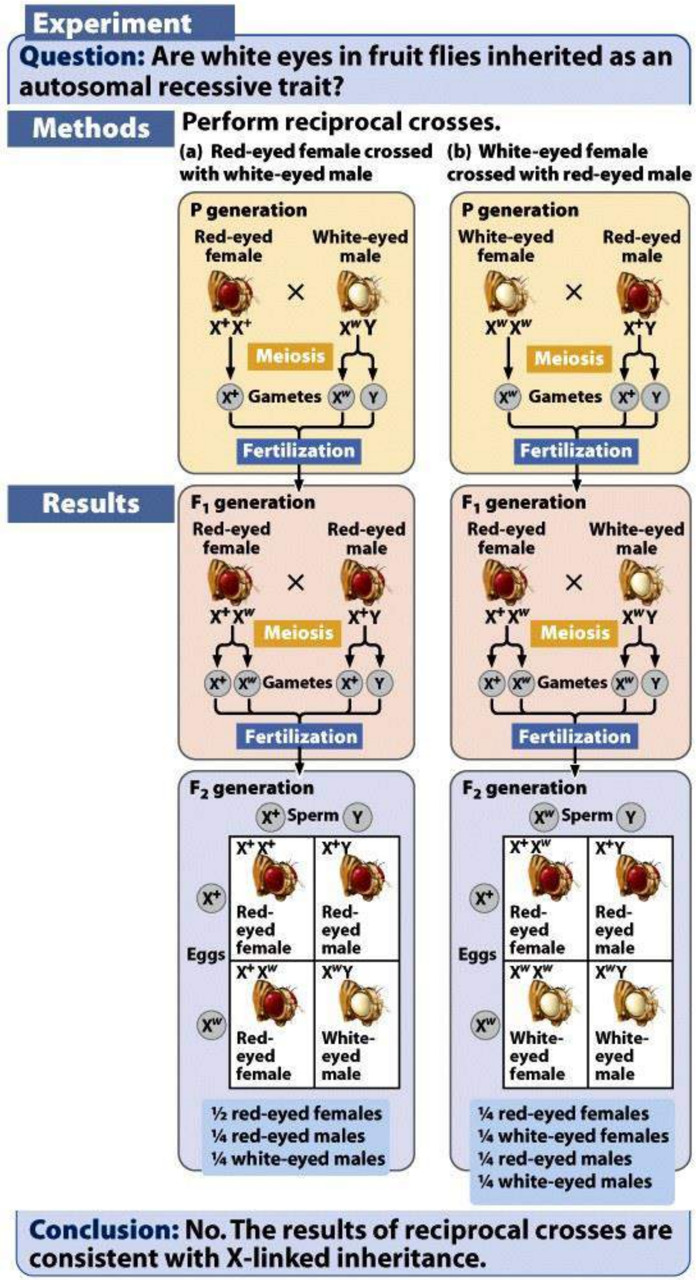


Figure 4-12
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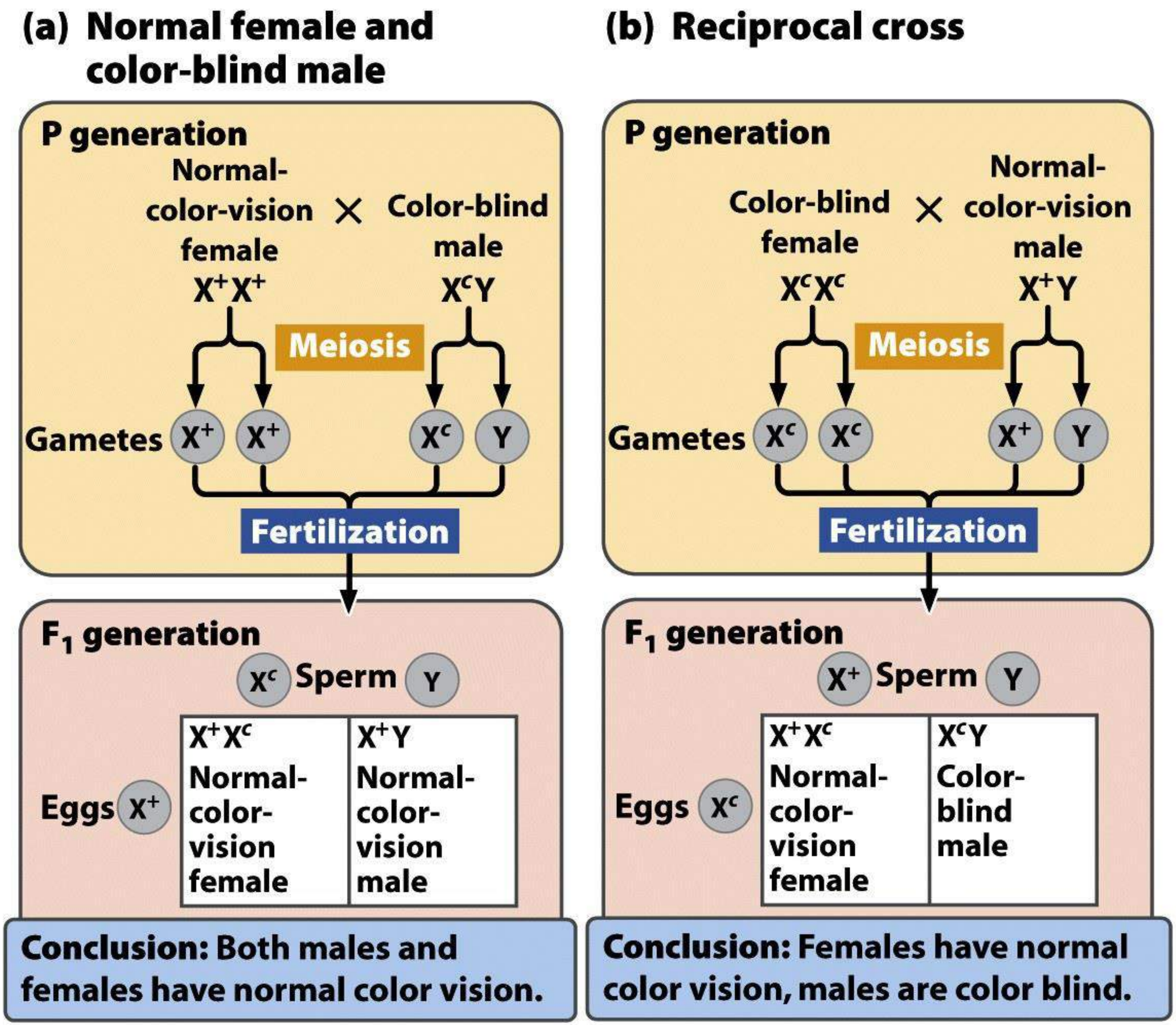

Figure 4-15

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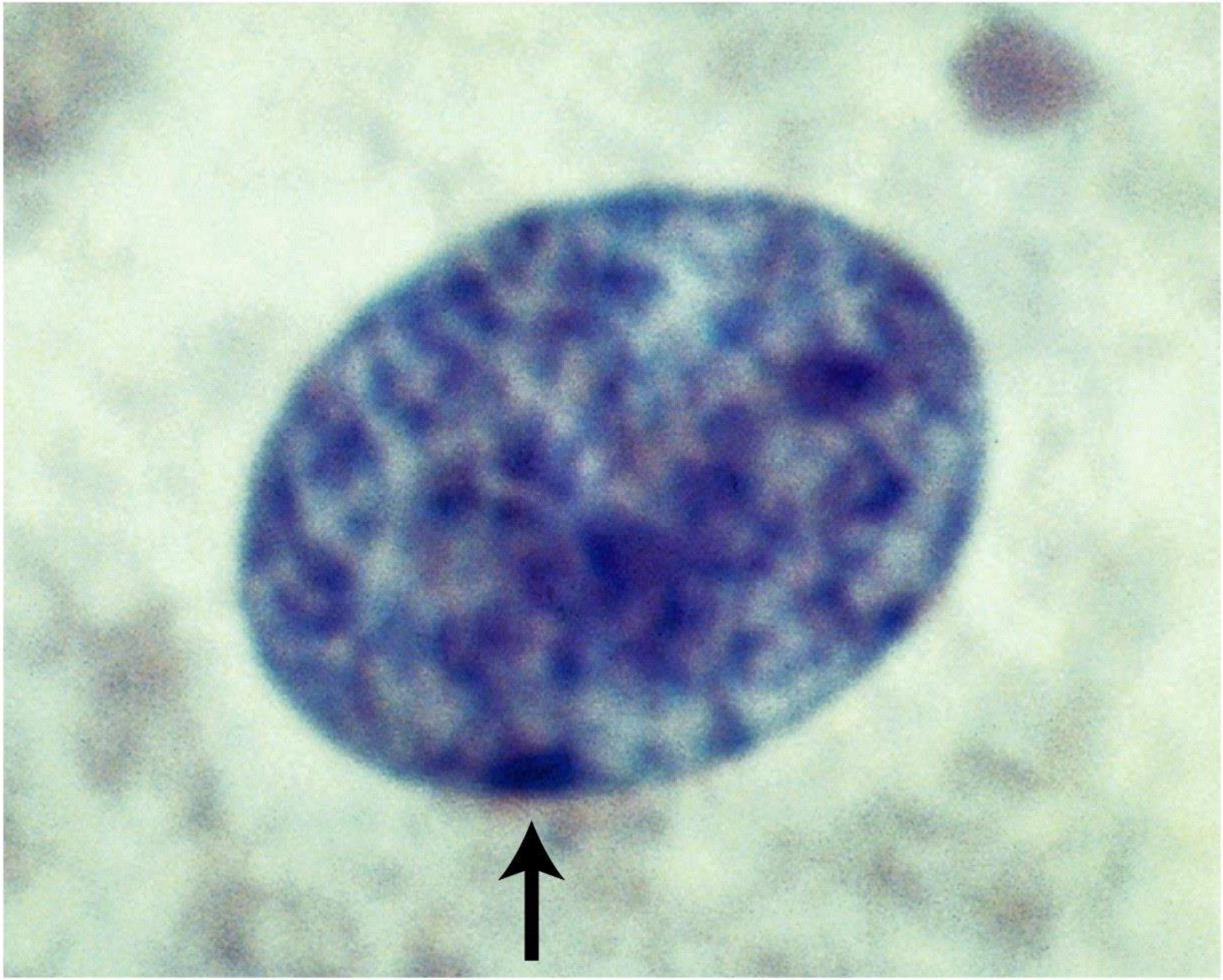

Figure 4-16a
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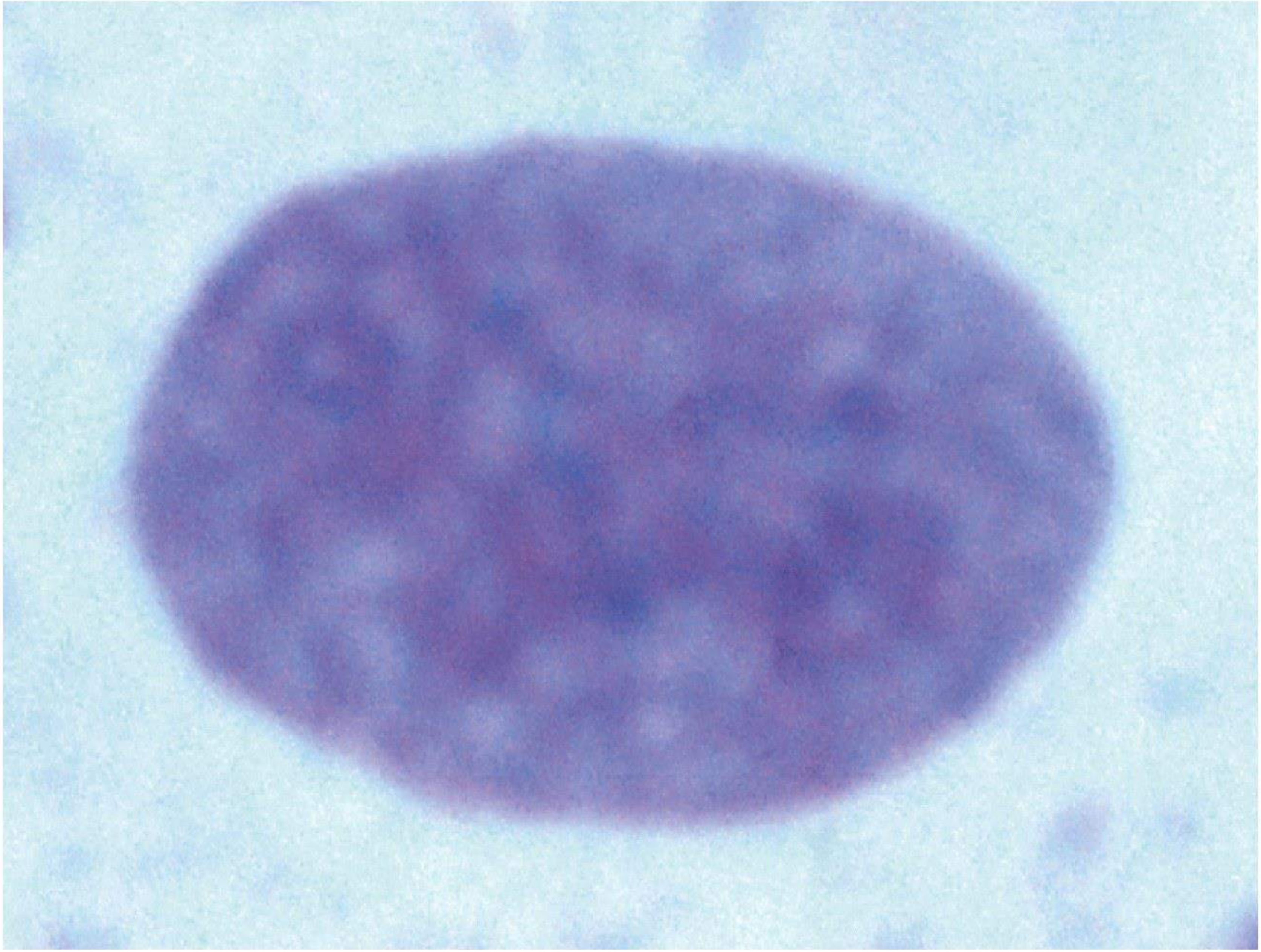

Figure 4-16b
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