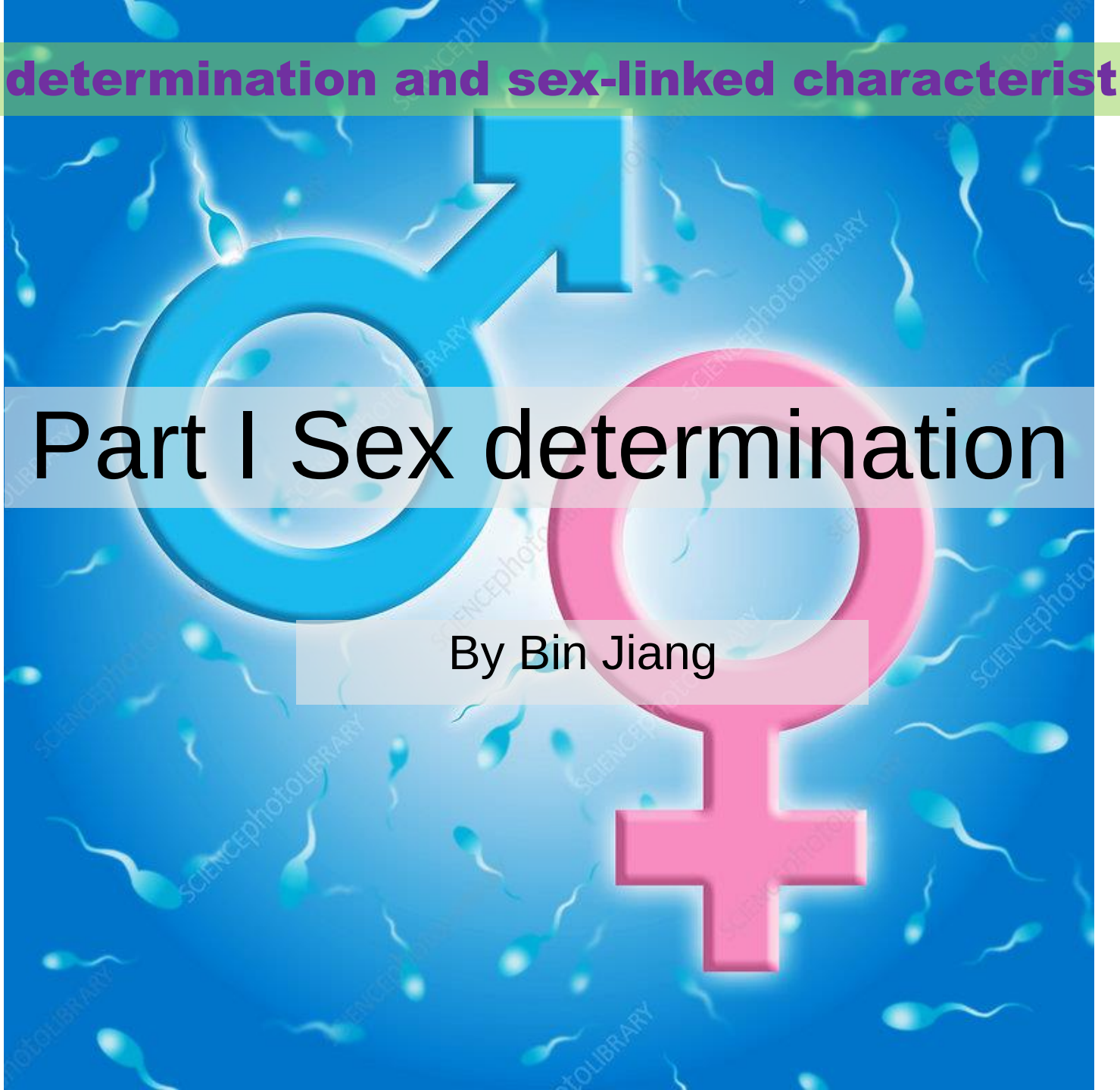
Chapter 5 Sex determination and sex-linked characteristics



Part I Sex determination

By Bin Jiang

Outline

- Different sex determining system on chromosome
- Sex determination in human
- Sex determination in *drosophila*
- Sex determination by environment
- Sex-linked traits

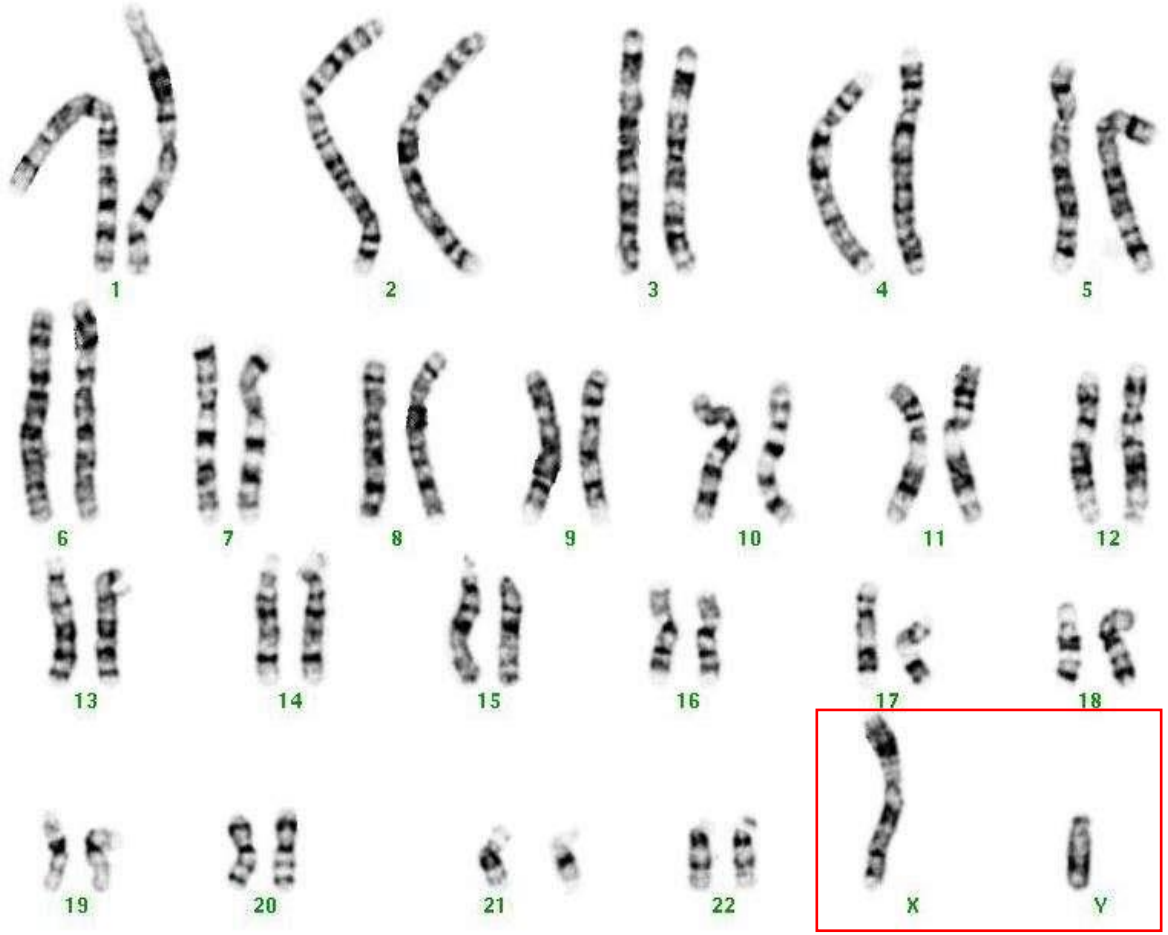
Men are from Mars, Women are from Venus

Bezeichnung
der Himmelskörper.

☉ Sonne.	♃ Jupiter.
☾ Mond.	♄ Saturn.
☿ Merkur.	♅ Uranus.
♀ Venus.	♆ Neptun.
♁ Erde.	⚵ Asträa.
♂ Mars.	♆ Hebe.
⚶ Vesta.	⚘ Iris.
⚵ Juno.	⚷ Flora.
⚴ Pallas.	⚙ Metis.
⚳ Ceres.	

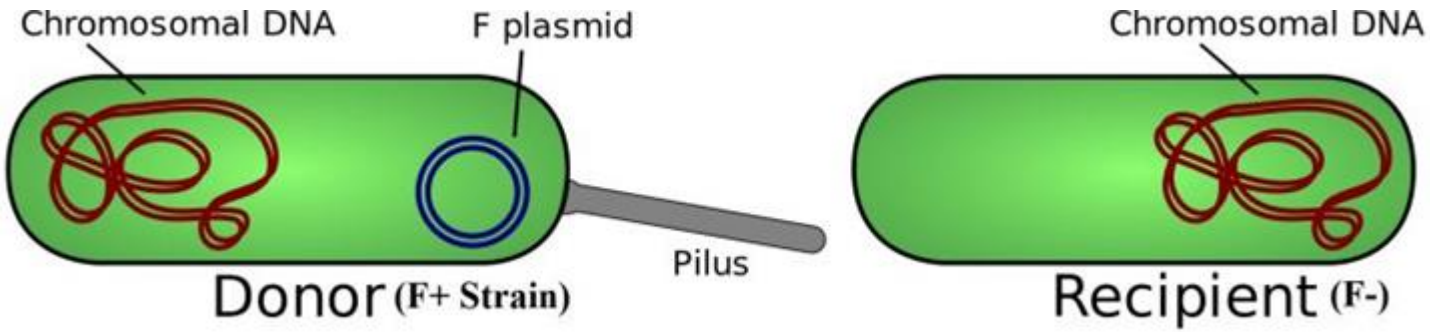
Chromosomal sex determining system (XY)

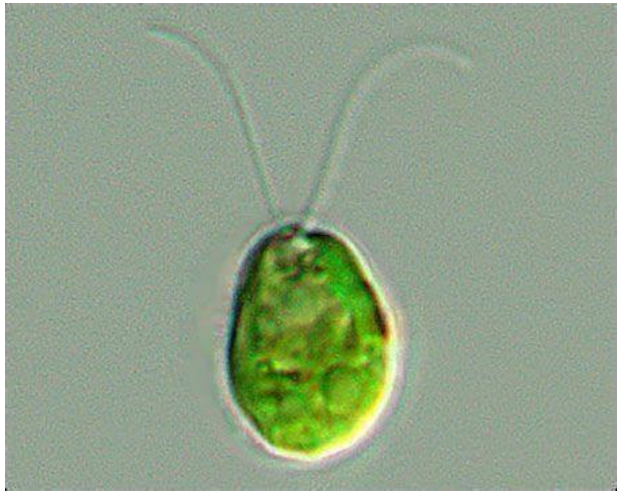
Sex chromosome vs. Autosome



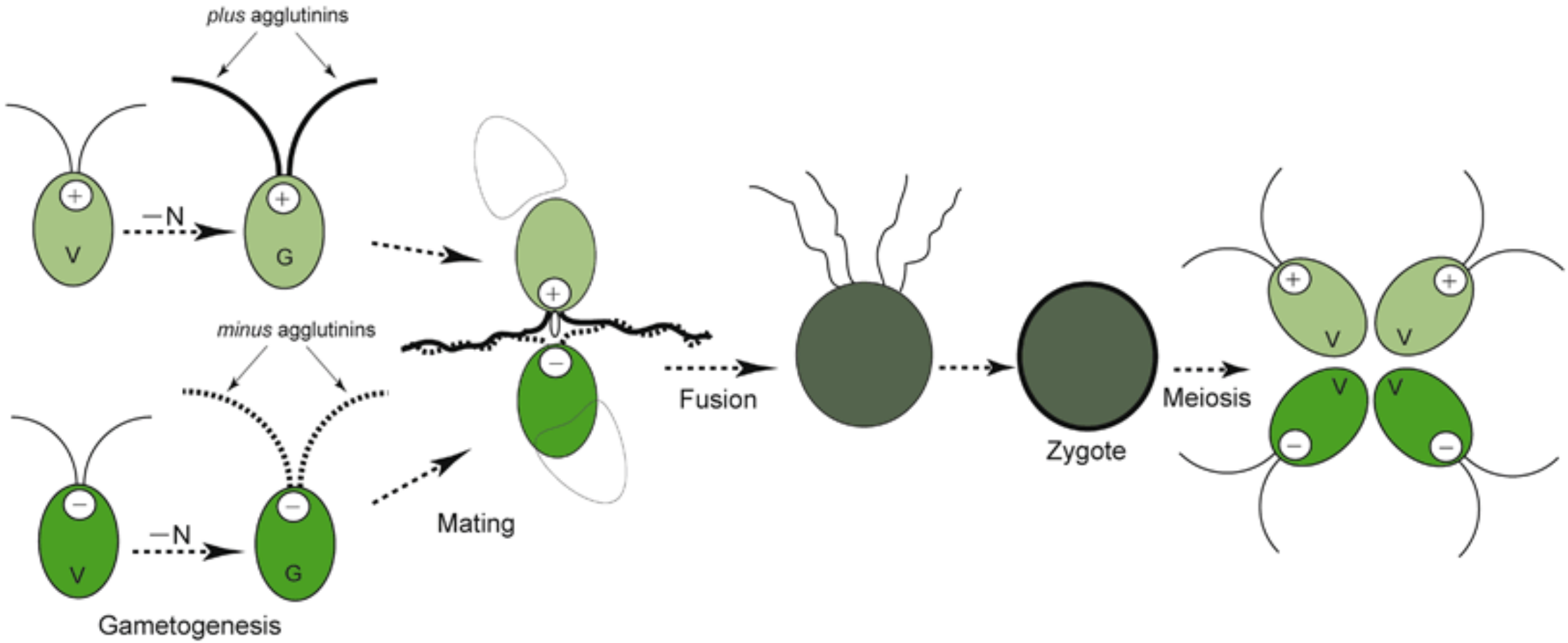
Sexual differentiation is very common

Sexual differentiation occurs in organisms as low as on the evolutionary scale as bacteria and single-celled eukaryotic algae.





Sexual reproduction in *Chlamydomonas reinhardtii*

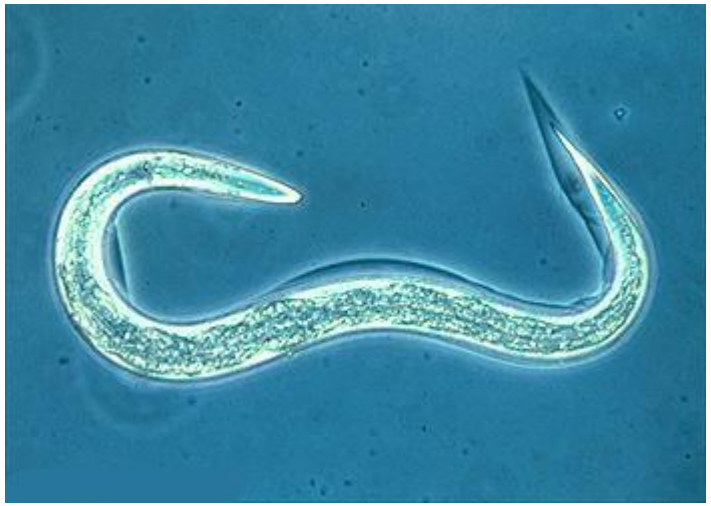


Sexual dimorphism

Secondary sexual differentiation

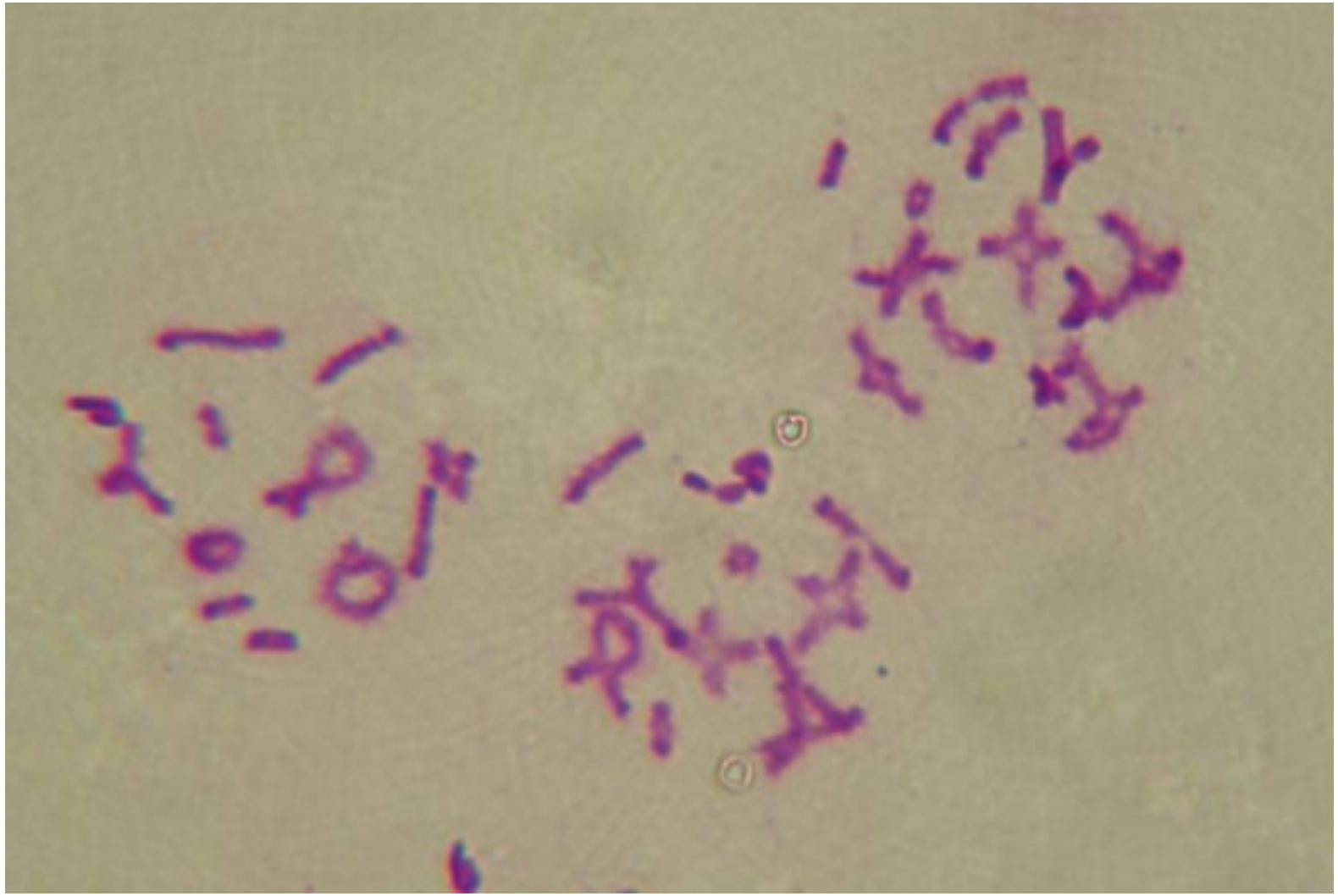
Primary sexual differentiation (gonads)

Unisexual vs. Bisexual



Hermaphrodite (雌雄同体)

1. Different sex determining system



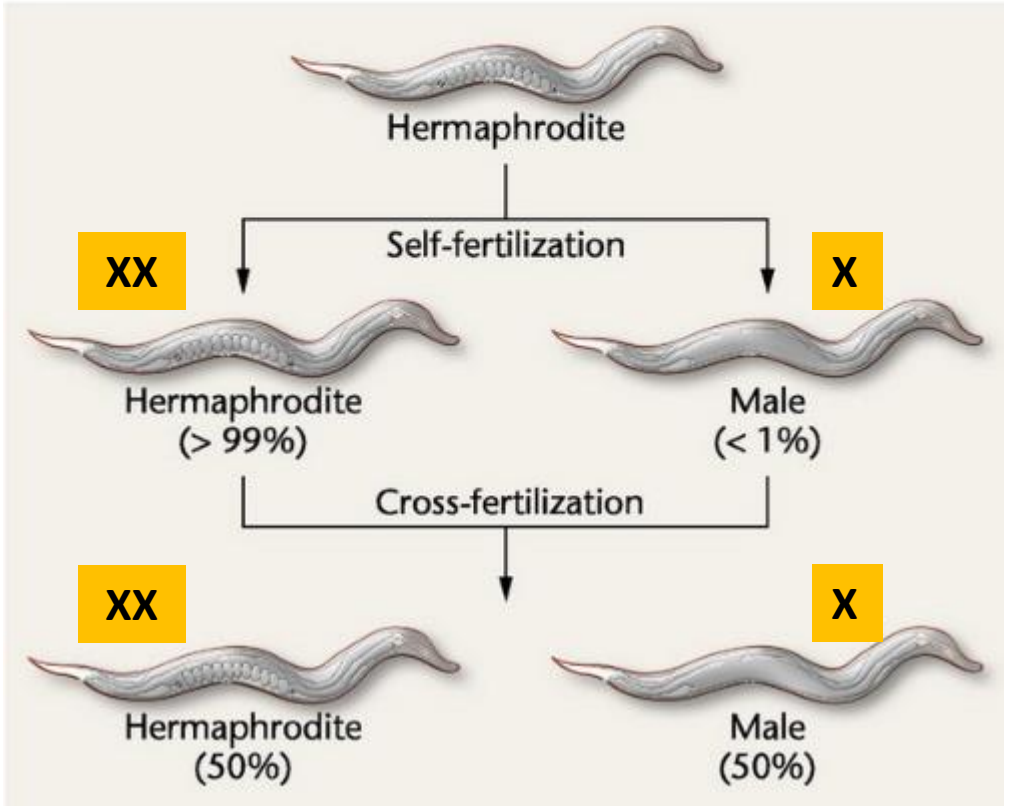
Nuclear structure of the Grasshopper sperm

Eg. 1 Nematodes

- *Caenorhabditis elegans*
- XX/XO system
- Ratio of sex chromosomes to autosomes
  - ♀ 2X/2A=1
  - ♂ 1X/2A=0.5

(a)

(b)



Protenor butterfly (蓝凤蝶)

♀: XX+12A

x

♂: X +12A

Gametes

X +6A

6A

X+6A

♀ XX+12A

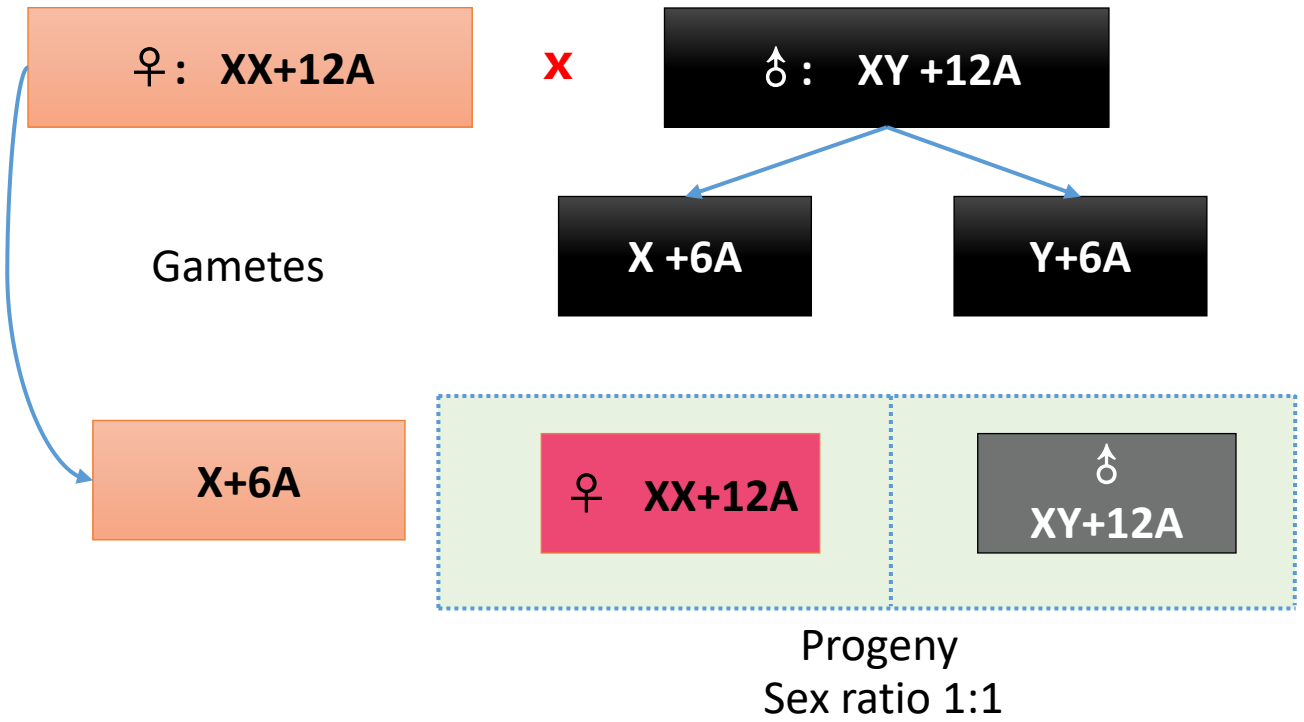
♂ X+12A

Progeny

Sex ratio 1:1

Eg. 2 Milkweed bug (大乳草长蝽)

- *Lygaeus turcicus*
- XX/XY system
- Other example

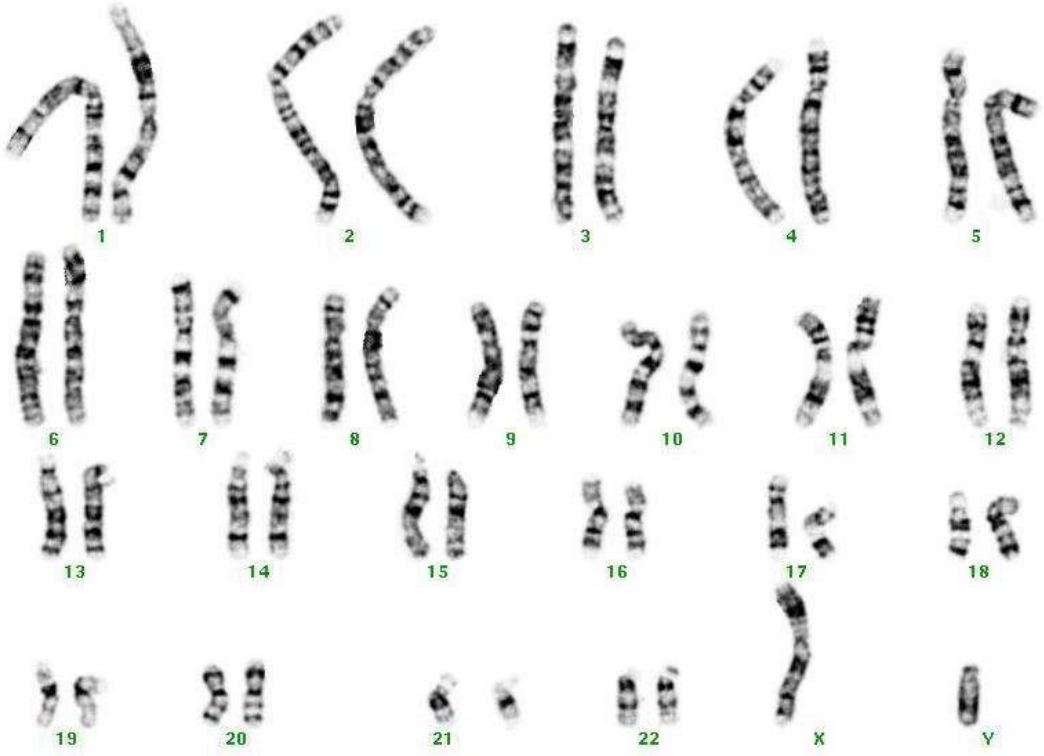


Heterogametic sex
Homogametic sex

Eg. 3 Chickens

- ~78 chromosomes
- Female is the heterogametic sex.
- ZZ/ZW system
- Other organisms: silkworm (2n=28), certain moth and butterflies, most birds, some fish, reptiles, amphibians, one species of plant (东方草莓).

2. Sex determination in human



- XX/XY
- What determines maleness?

Lack of a second X chromosome

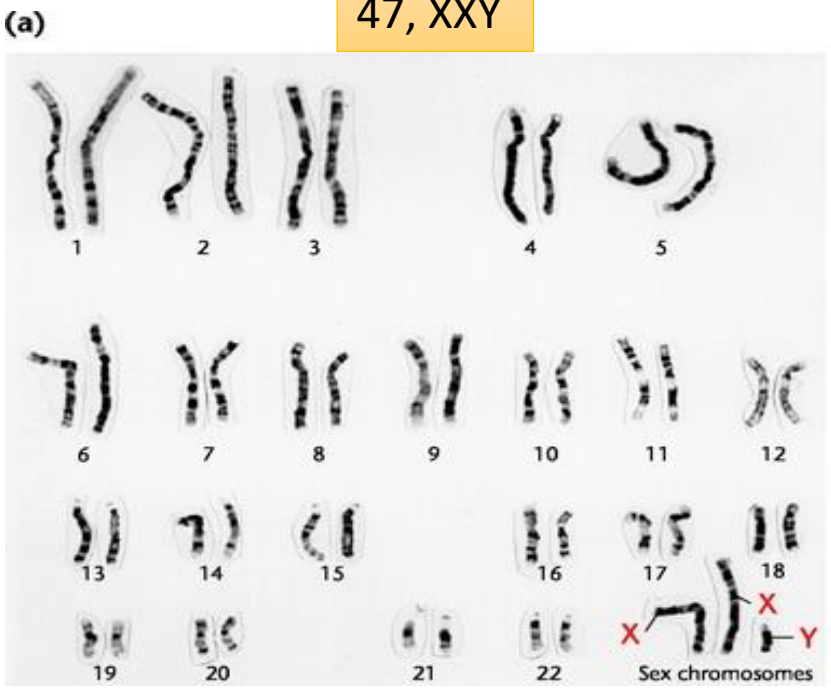
Presence of Y chromosome

How to prove this?

Klinefelter and Turner Syndromes

Presence of Y chromosome

47, XXY



- Male genitalia, rudimentary testes
- Tall and long arms
- Feminine sexual development：slight enlargement of breasts, rounded hips

Lack of a second X chromosome

45, X

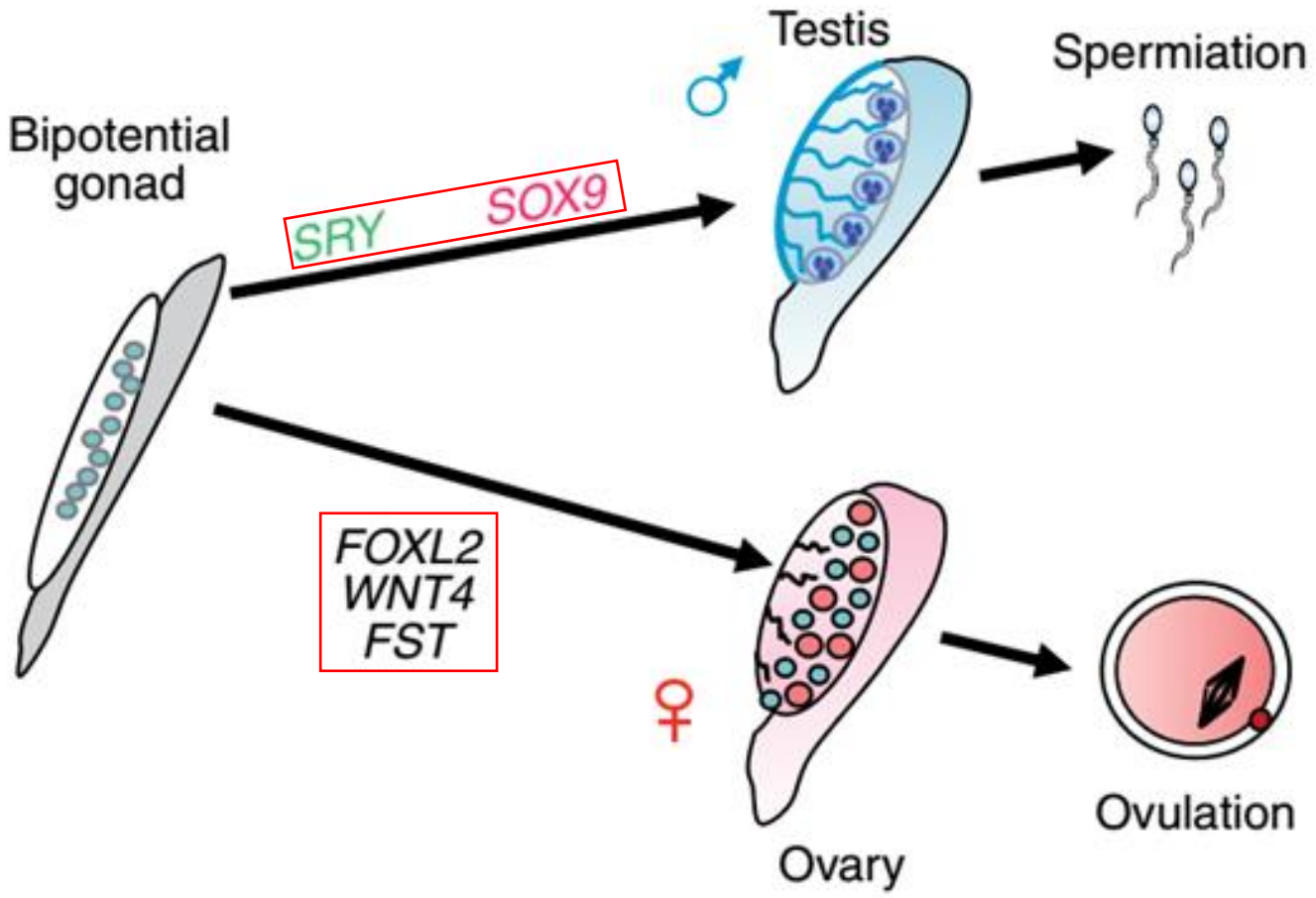
- Female appearance
- Female genitalia, rudimentary ovaries
- Short
- Skin flaps on the back of the neck

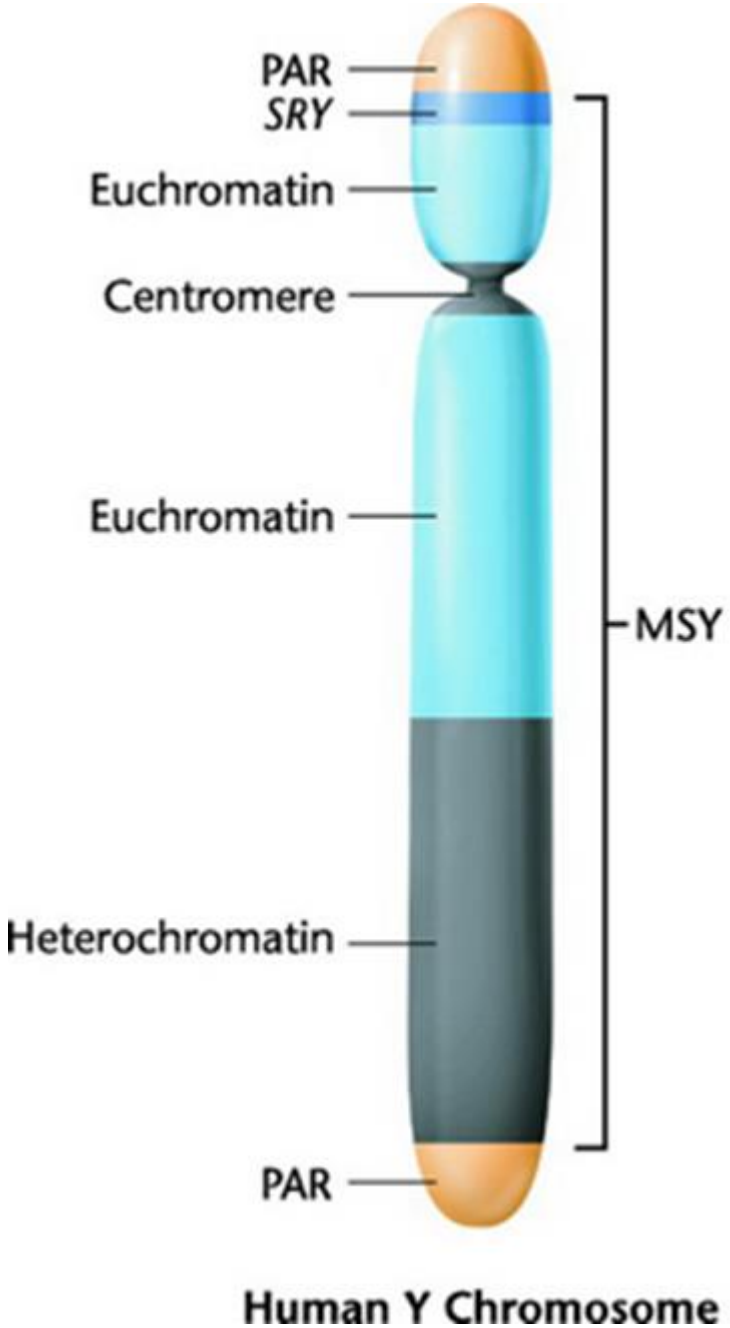
XYY syndrome （47,XYY）

- Patricia Jacobs (Scottish geneticist)
- In a Scottish maximum security prison: 9/315 males with 47,XYY karyotype.
- Characteristic:
  - Usually be dangerous, violent individual with criminal propensities.
  - **7/9**: subnormal intelligence; **all** suffered personality disorders.

Sexual differentiation in humans

5th week of embryo

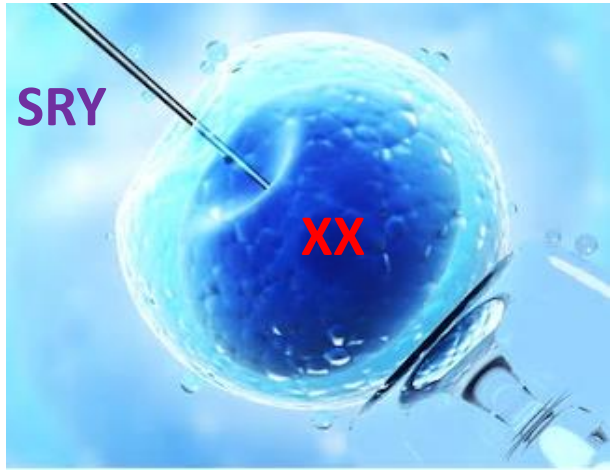




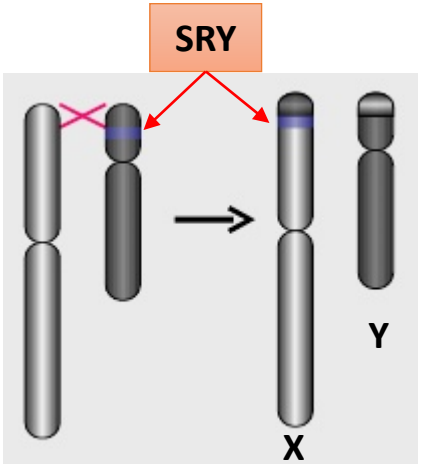
Y chromosome (p132)

- **PAR (pseudo-autosomal region)**: share homology with regions on the X (recombine with X during meiosis)
- ★**SRY (sex-determining region Y)**: coding testis-determining factor (TDF)
- MSY (male-specific region of the Y)

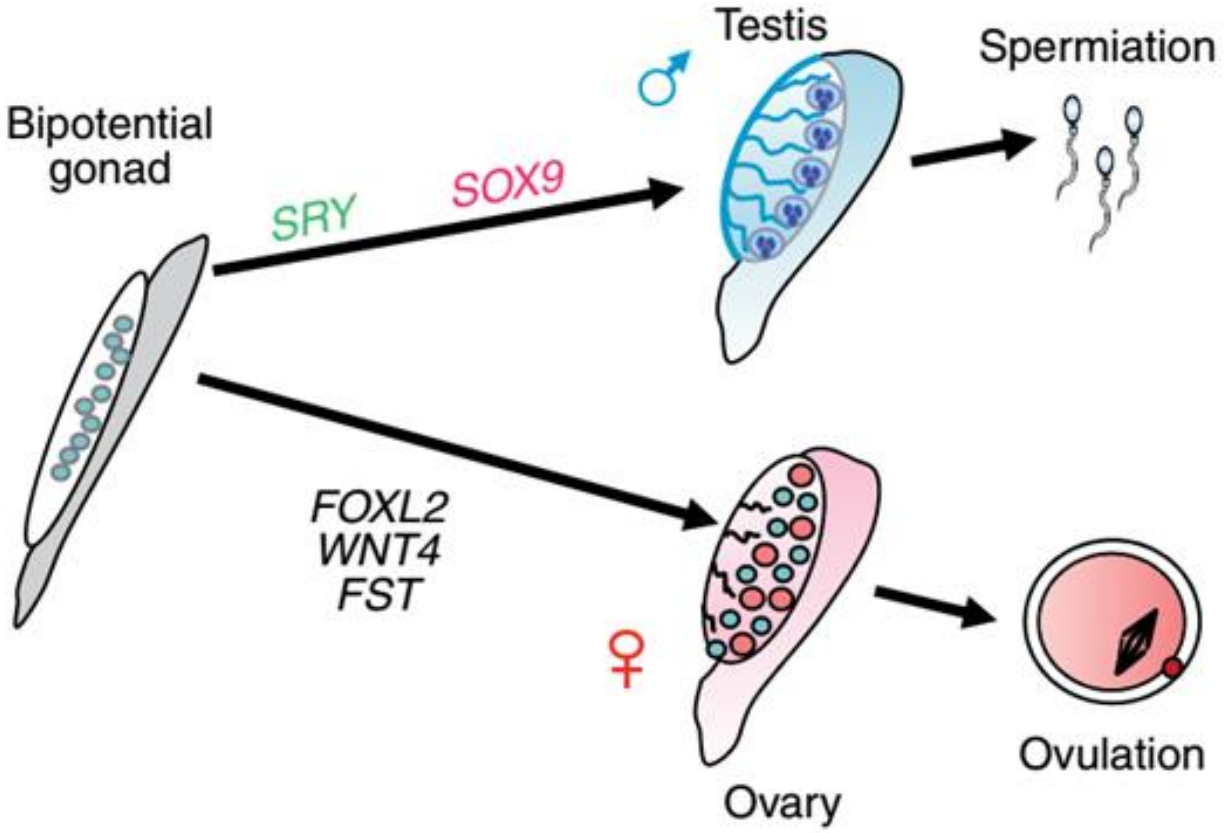
- Q1: What sexuality will the embryo develop into?



- Q2: What will happen if SRY were translocated to X during crossing-over in meiosis? （同源染色体的交叉互换）



The presence or absence of a Y chromosome that contains an intact *SRY* gene is responsible for causing maleness in humans.



SRY Gene

encode

TDF protein

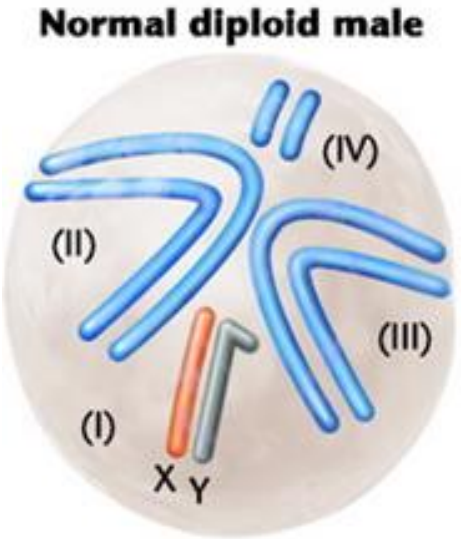
regulate

Genes downstream:
e.g. *SOX9*

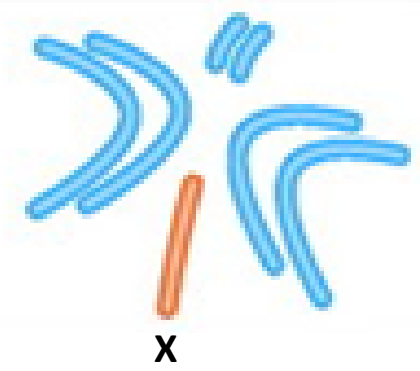
SOX9 on chromosome 17

3. Sex determination in *Drosophila* p103

- Males are XY; female are XX.
- Question: Does Y chromosome cause maleness in fruit flies?



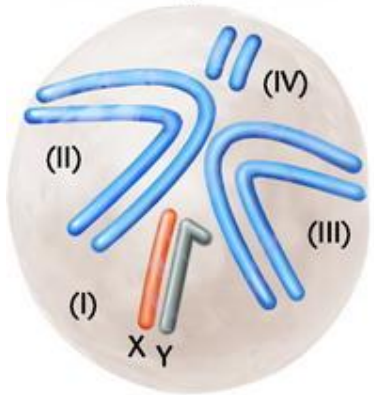
2 sets of autosomes
+
X Y



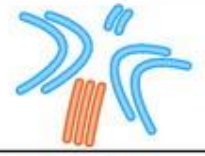
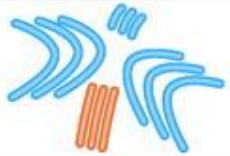
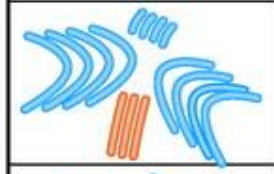
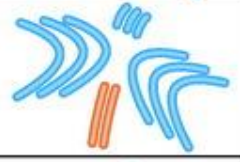
maleness

Calvin Blackman Bridges
Morgen's lab

Normal diploid male

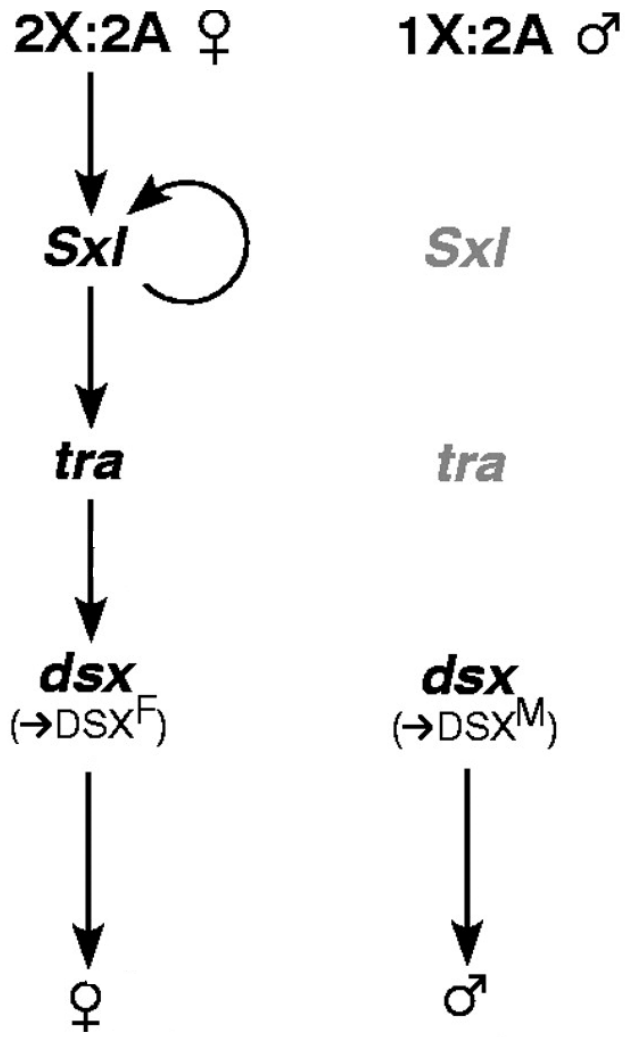


2 sets of autosomes
+
X Y

Chromosome composition	Chromosome formulation	Ratio of X chromosomes to autosome sets	Sexual morphology
	$3X/2A$	1.5	Metafemale
	$3X/3A$	1.0	Female
	$2X/2A$	1.0	Female
	$3X/4A$	0.75	Intersex
	$2X/3A$	0.67	Intersex
	$X/2A$	0.50	Male
	$XY/2A$	0.50	Male
	$XY/3A$	0.33	Metamale

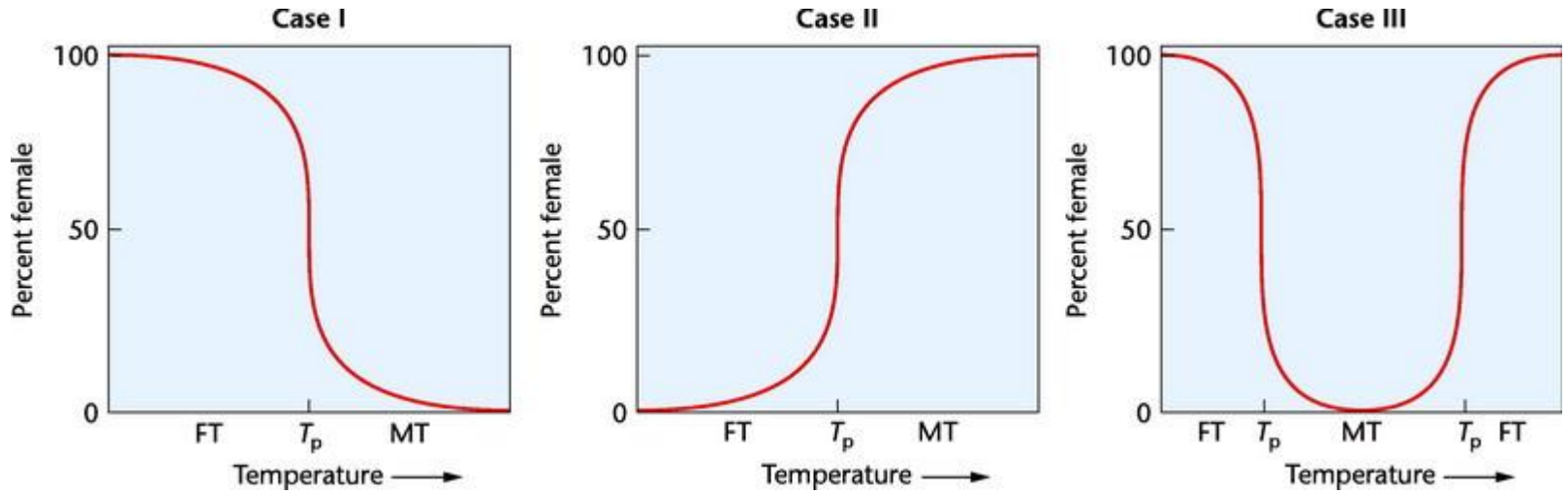
Gene regulation in sex determination

- ***Sex-lethal* gene (*Sxl*):** “master switch” in sex determination. X-linked.
- Gene on autosome :
  - ***Transformer (tra)***
  - ***Doublesex* (*dsx*)**
- RNA splicing/ alternative splicing



4. Sex determination by environment

- Temperature-dependent sex determination (TSD)
  - All crocodiles, most turtles, some lizards
  - **Aromatase** (芳香化酶): androgen (雄性激素)→estrogen (雌性激素)
  - **Thermosensitive factor** mediates the transcription of aromatase.



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Current Biology

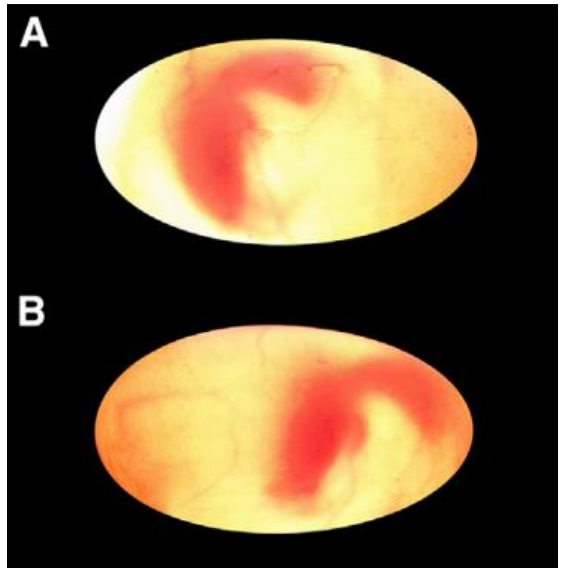
The Embryos of Turtles Can Influence Their Own Sexual Destinies

Highlights

- By moving within the egg, a turtle embryo can influence its own sex
- That ability expands the range of nest conditions producing an equal sex ratio
- Thermoregulation by embryos should buffer populations against climate change

Authors

Yin-Zi Ye, Liang Ma, Bao-Jun Sun, Teng Li, Yang Wang, Richard Shine, Wei-Guo Du

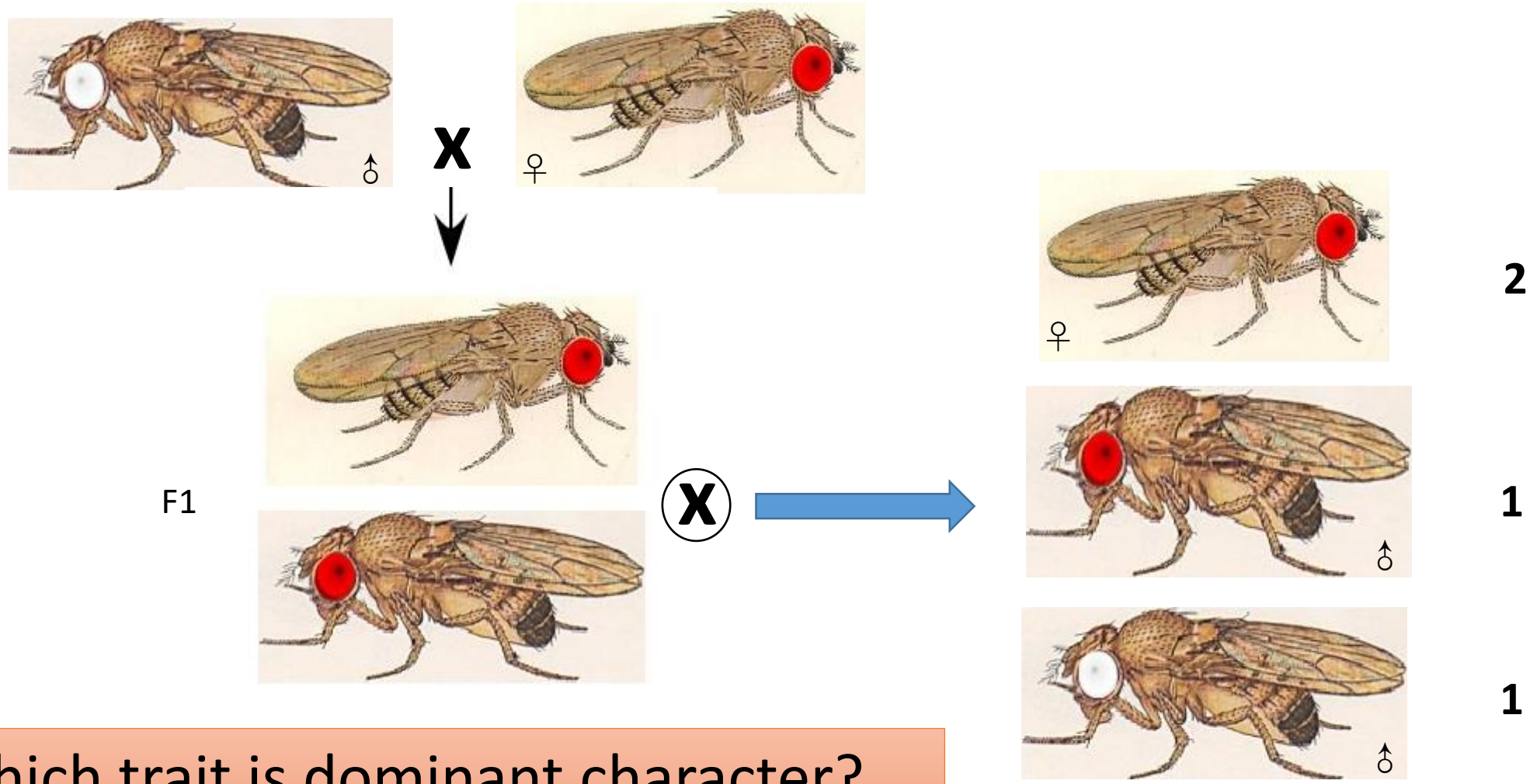


Conclusion in this part

1. Sex determining system
    1. By sex chromosomes：XX/XO，XX/XY，ZZ/ZW
    2. By environment：temperature and day length (p127)
    3. Gene regulation in sex determination

II. Sex-linked inheritance (伴性遗传)

1. Sex-linked inheritance in *drosophila*



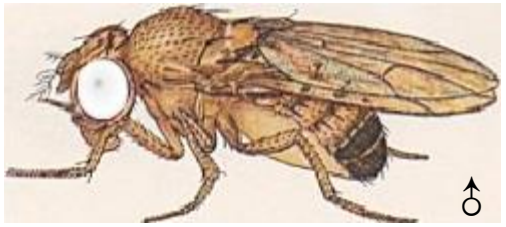
Which trait is dominant character?

Morgan's hypotheses

- White-eye gene is recessive gene (w).
- This gene is on X chromosome (X^w).
- It's allele (X^+) which controls red eye trait.

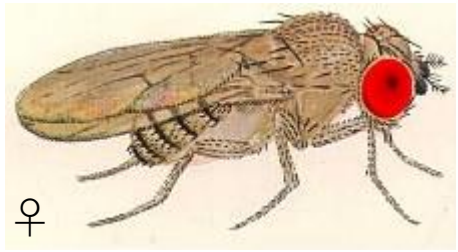
How to prove?

Reciprocal cross

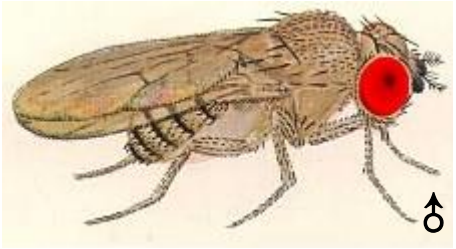


White male

X

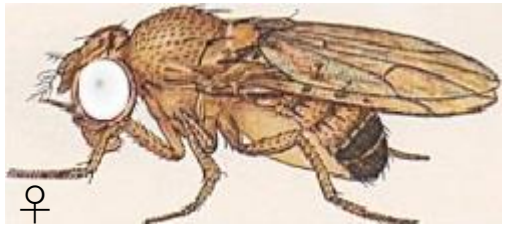


Red female



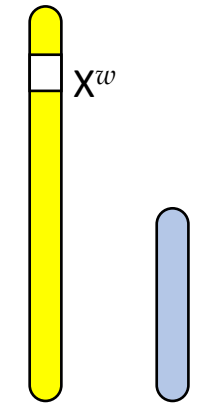
Red male

X

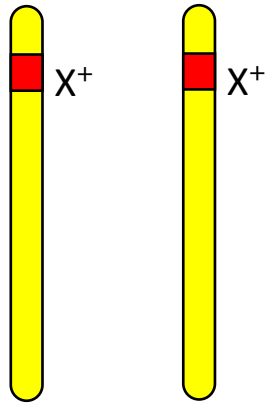


White female

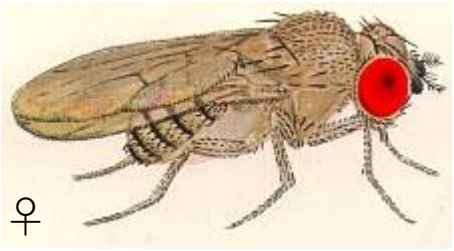
Genotype of different traits in *drosophila*

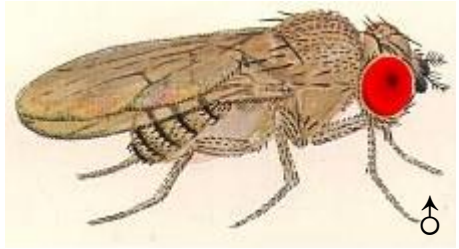


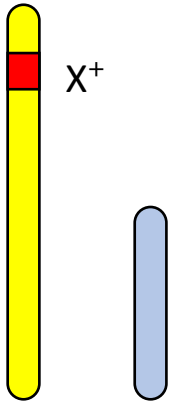
White male



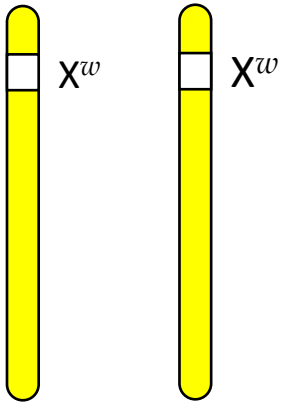
Red female



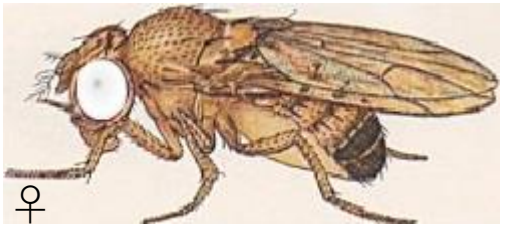




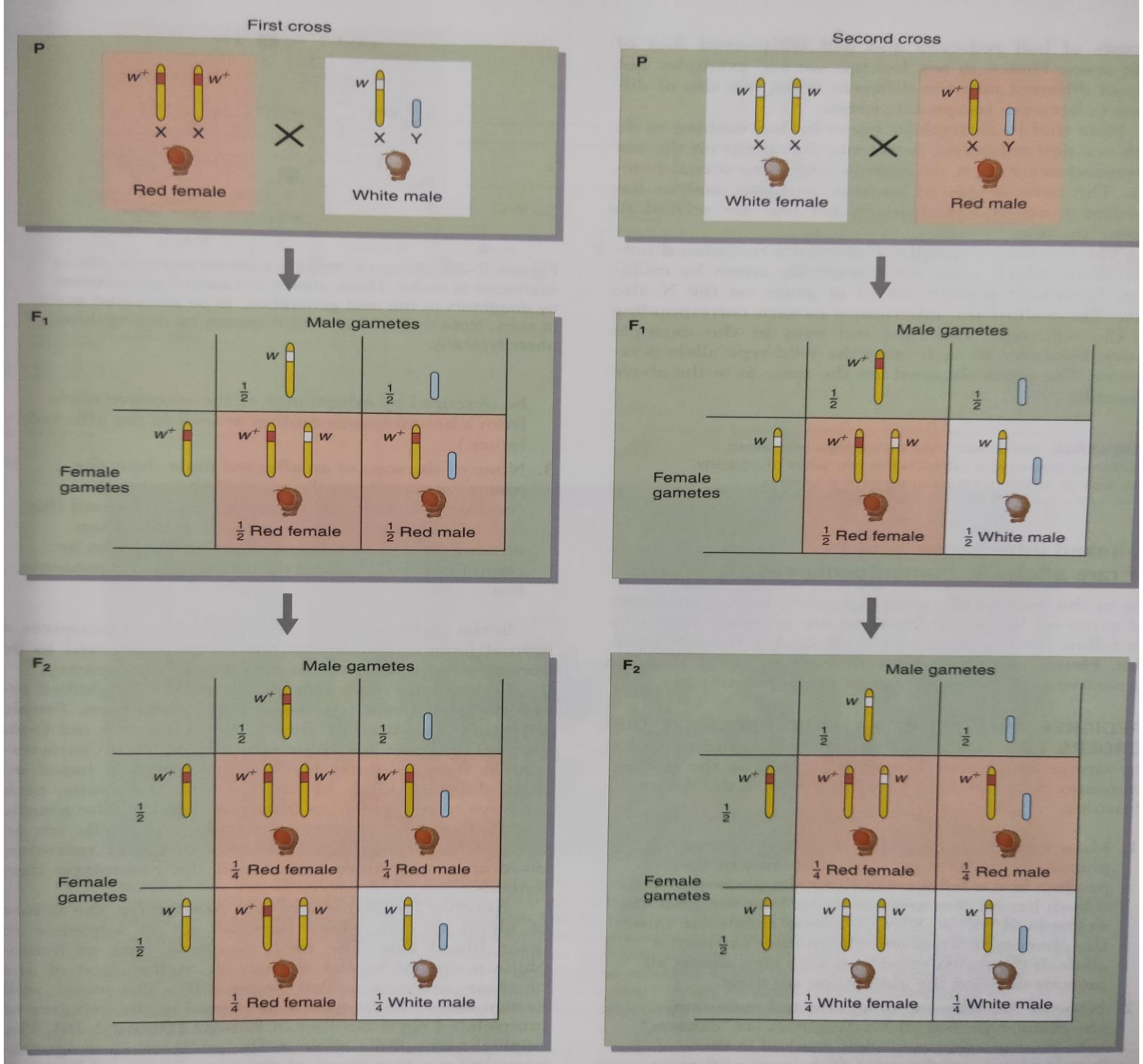
Red male



White female



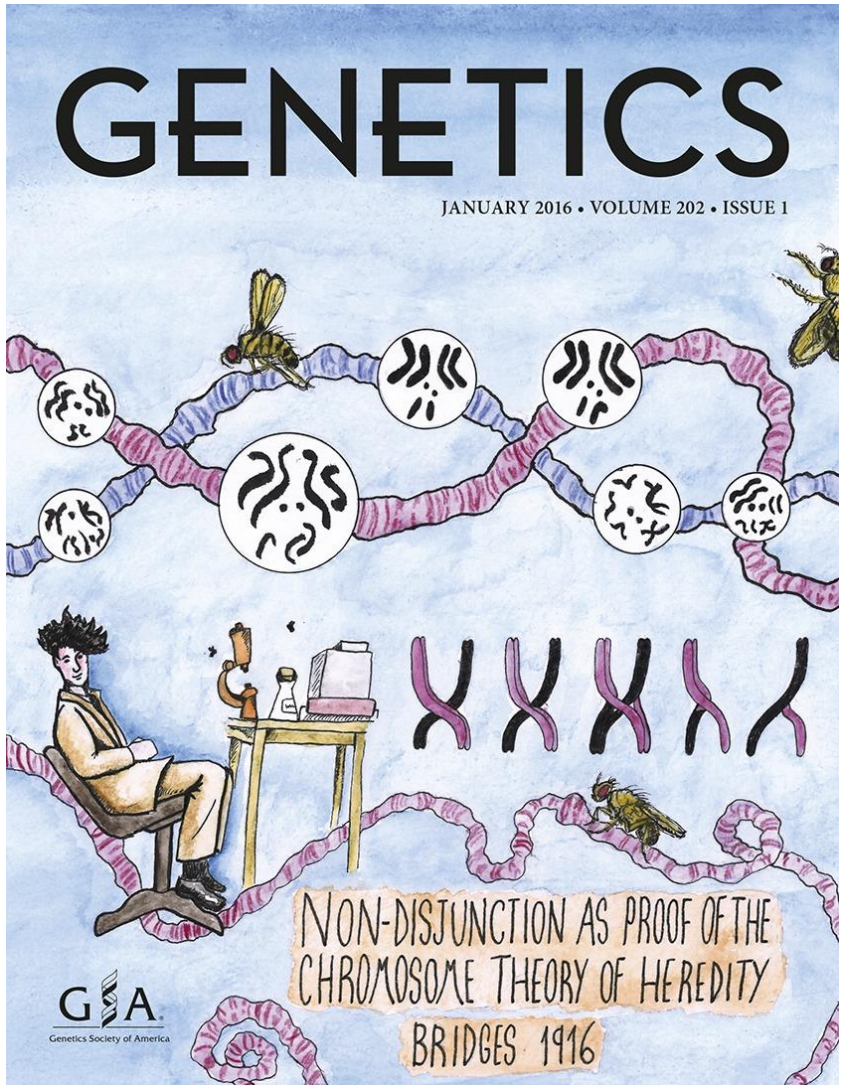
Prove the hypotheses



Practice

14. In *Drosophila*, an X-linked recessive mutation, *scalloped* (*sd*), causes irregular wing margins. Diagram the F_1 and F_2 results if (a) a scalloped female is crossed with a normal male; (b) a scalloped male is crossed with a normal female. Compare these results to those that would be obtained if the *scalloped* gene were autosomal.
15. Another recessive mutation in *Drosophila*, *ebony* (*e*), is on an autosome (chromosome 3) and causes darkening of the body compared with wild-type flies. What phenotypic F_1 and F_2 male and female ratios will result if a scalloped-winged female with normal body color is crossed with a normal-winged ebony male? Work this problem by both the Punnett square method and the forked-line method.

阅读Bridges的贡献



The Centenary of *GENETICS*: Bridges to the Future

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*Department of Genetics, University of Wisconsin, Madison, Wisconsin 53706, †Stowers Institute for Medical Research, Kansas City, Missouri 64110, and ‡Department of Molecular and Integrative Physiology, University of Kansas Medical Center, Kansas City, Kansas 66160

One hundred years ago, in the first paper in the first edition of *GENETICS*, Calvin Blackman Bridges provided evidence for the chromosome theory of inheritance, laying the groundwork for much of the genetics research that has followed (Bridges 1916). As we discuss a paper that is arguably a cornerstone of modern genetic analysis, it is well worth remembering that this two-part paper was the report of Bridges's Ph.D. thesis work (indeed, we find it sobering to compare the impact of our own theses to that of Bridges's). Bridges's 1916 paper described nondisjunction (improper chromosome segregation), explained how evidence of nondisjunction during meiosis provided proof that chromosomes contained the genetic material and illustrated how sex determination works in *Drosophila melanogaster*. The scientific insights Bridges made in this seminal paper were instrumental to subsequent experimental studies of meiosis, and his influence is still felt in genetics labs today.