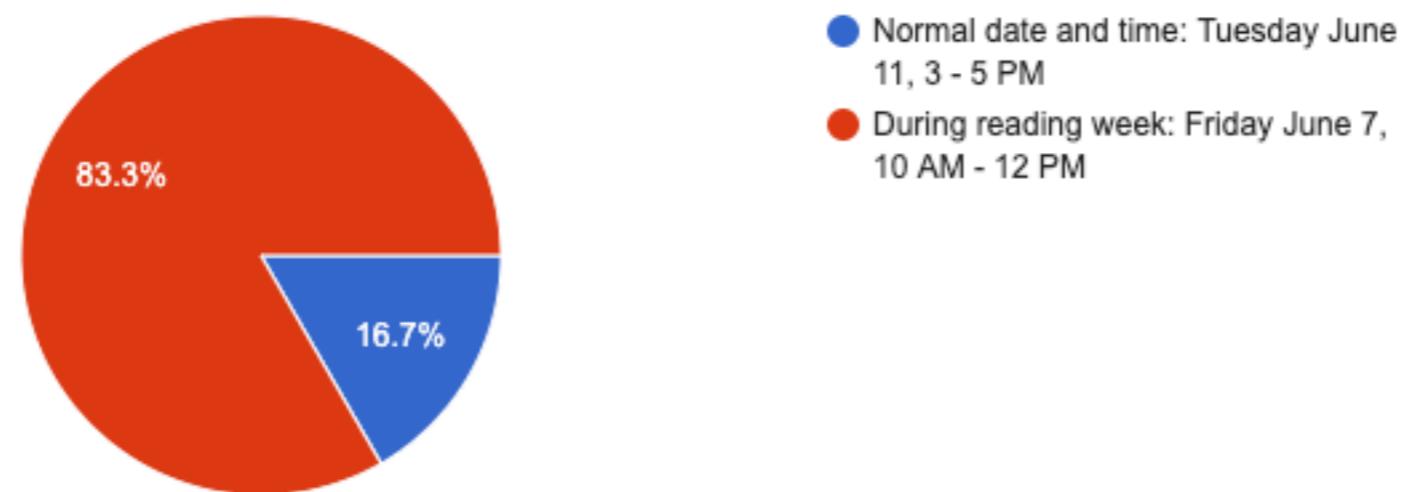


Course website:
bio393.andersenlab.org

Problem set #1 is out.

Genetics terms are online.

Final will be on June 7th

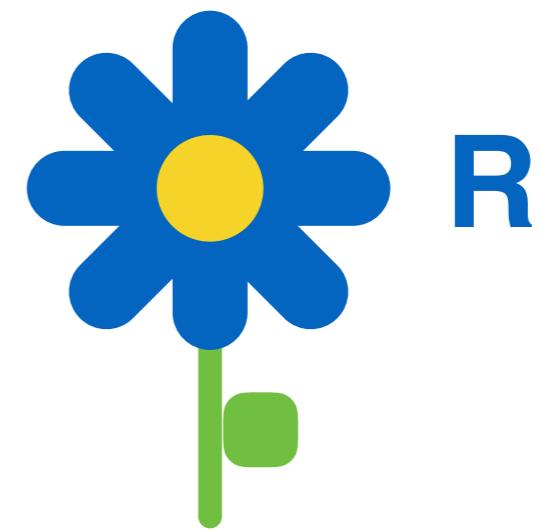


Gregor Mendel was lucky!

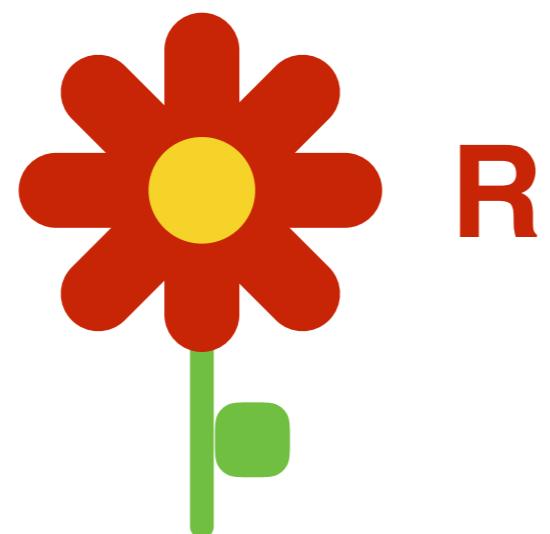


1. Peas are diploid (two copies of every chromosome).
2. Traits could have been multigenic (controlled by many genes).
3. Genes could have been linked (violate Law of Ind. Assortment).
4. Traits could have been co-dominant or incomplete dominant.

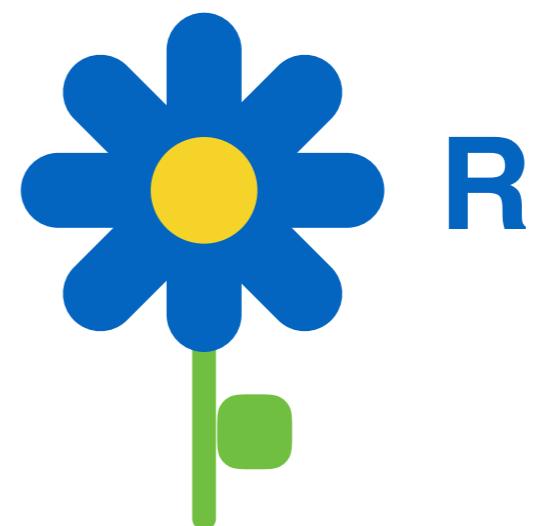
Three different types of dominance



Three different types of dominance



Complete



Incomplete

Co-dominant

Three different types of dominance



Complete

RR

Red



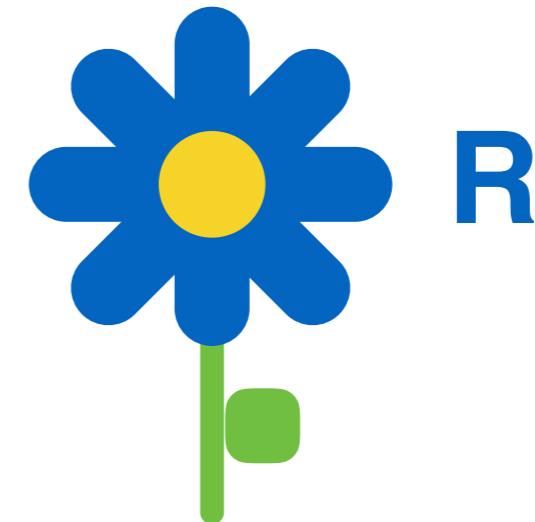
Incomplete

Red

Co-dominant

Red

Three different types of dominance



Complete

RR

Red

Incomplete

RR

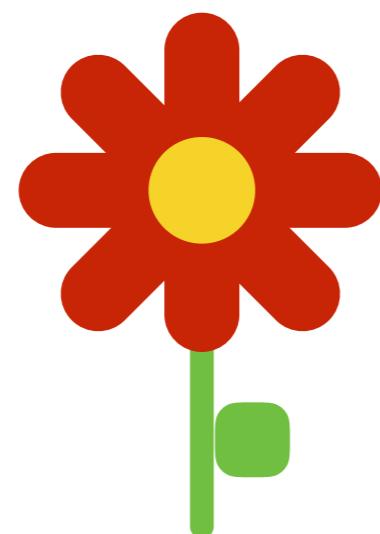
Blue

Co-dominant

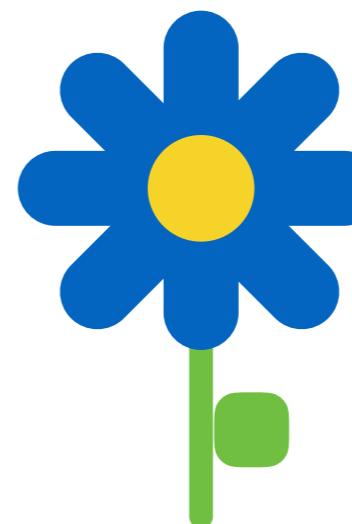
Red

Blue

Three different types of dominance



R



R

Complete

Incomplete

Co-dominant

RR

Red

Red

Red

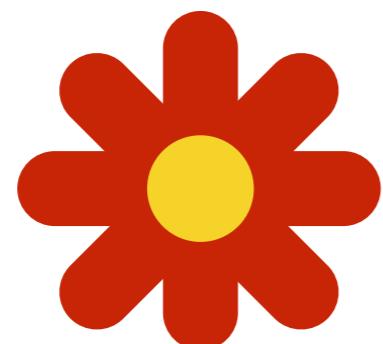
RR

Blue

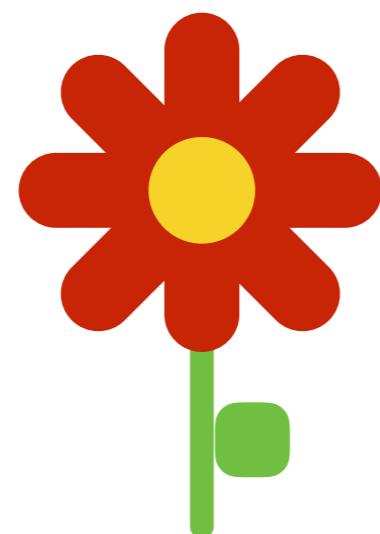
Blue

Blue

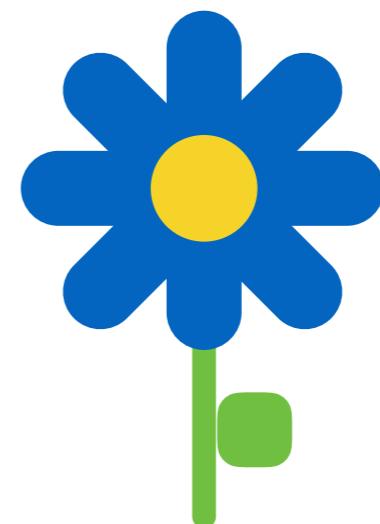
RR



Three different types of dominance



R



R

Complete

Incomplete

Co-dominant

RR

Red

Red

Red

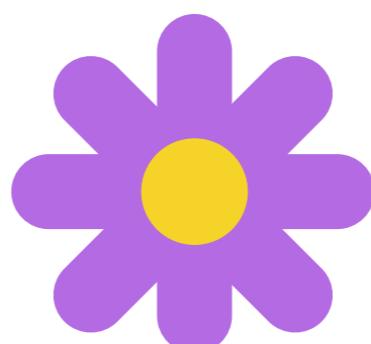
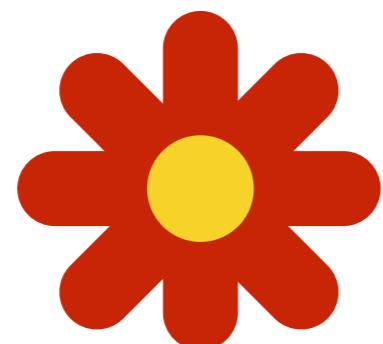
RR

Blue

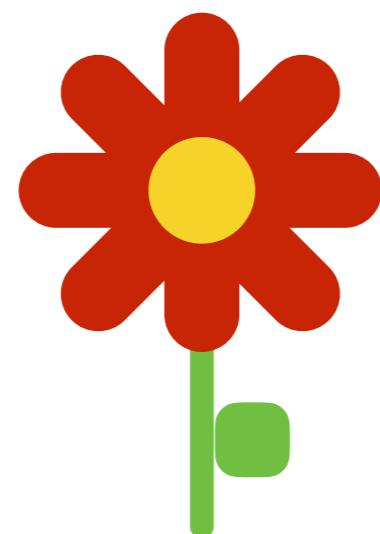
Blue

Blue

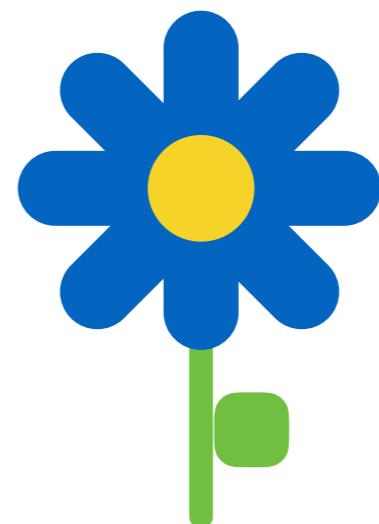
RR



Three different types of dominance



R



R

Complete

Incomplete

Co-dominant

RR

Red

Red

Red

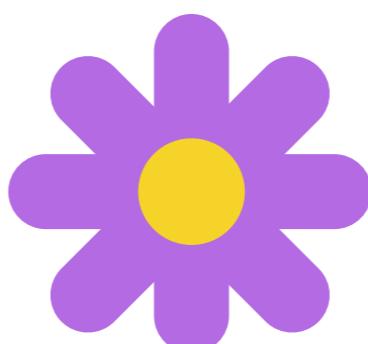
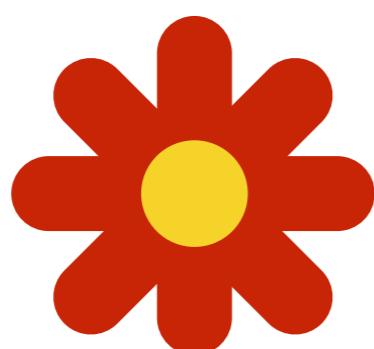
RR

Blue

Blue

Blue

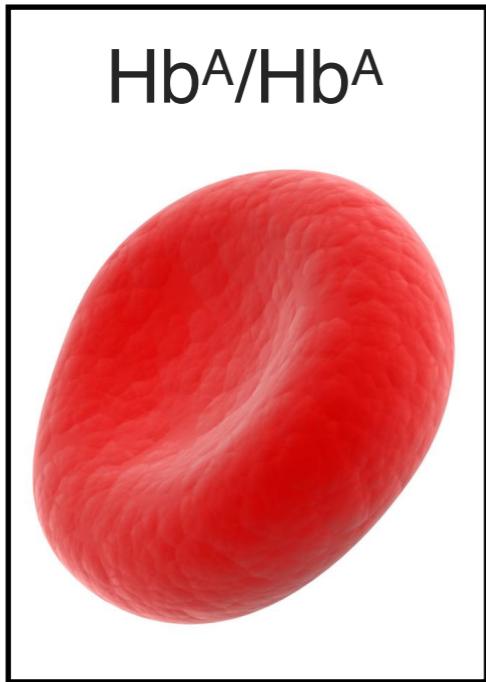
RR



Incomplete dominance: Different alleles confer a mixed phenotype



Hemoglobin sickle-cell allele

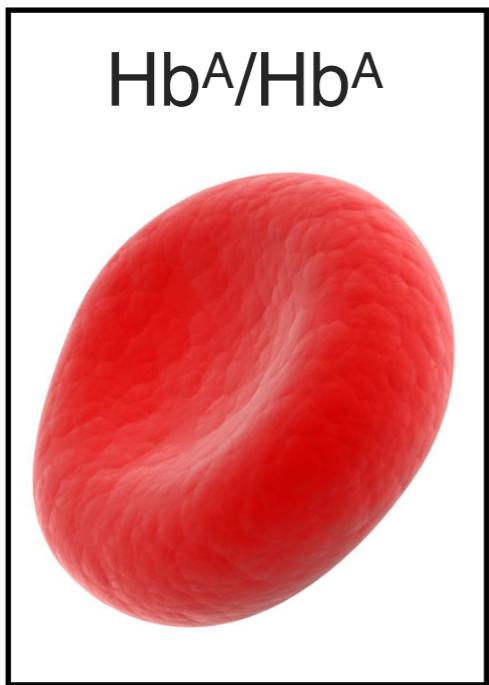


Normal
RBCs

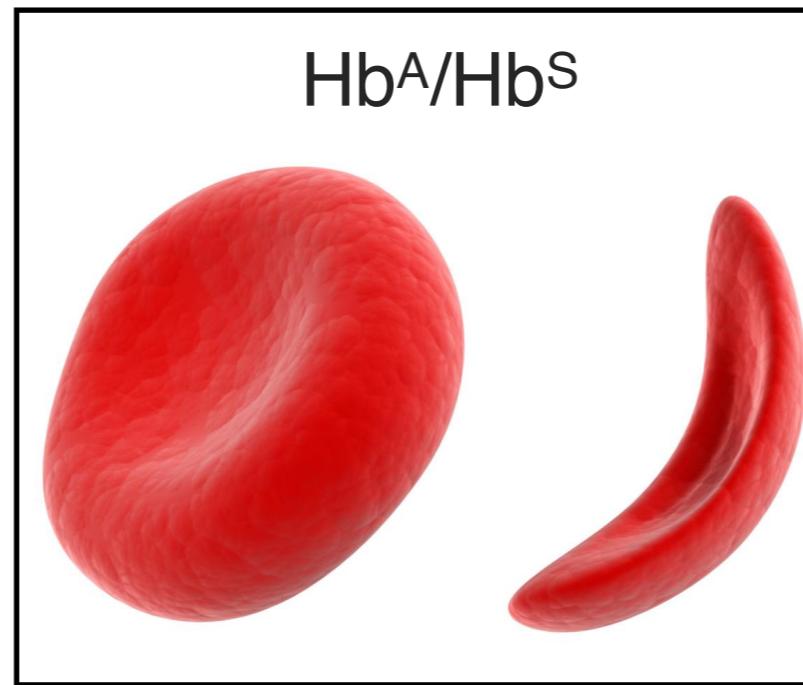


Mostly sickle
RBCs

Hemoglobin sickle-cell allele



$\text{Hb}^{\text{A}}/\text{Hb}^{\text{A}}$



$\text{Hb}^{\text{A}}/\text{Hb}^{\text{S}}$



$\text{Hb}^{\text{S}}/\text{Hb}^{\text{S}}$

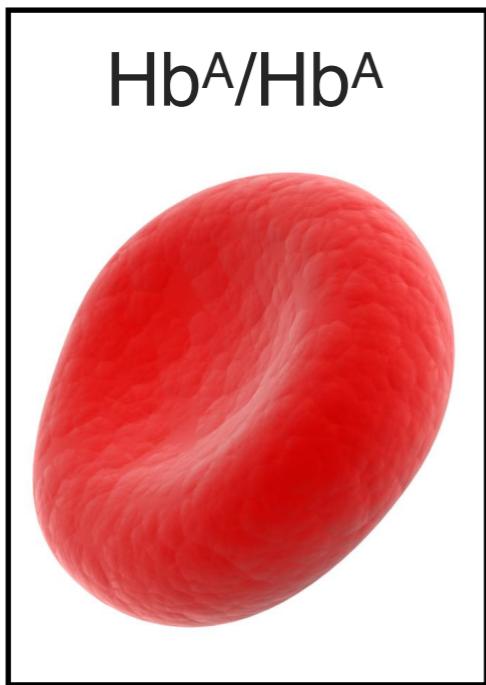
Normal
RBCs

Both normal and sickle
RBCs

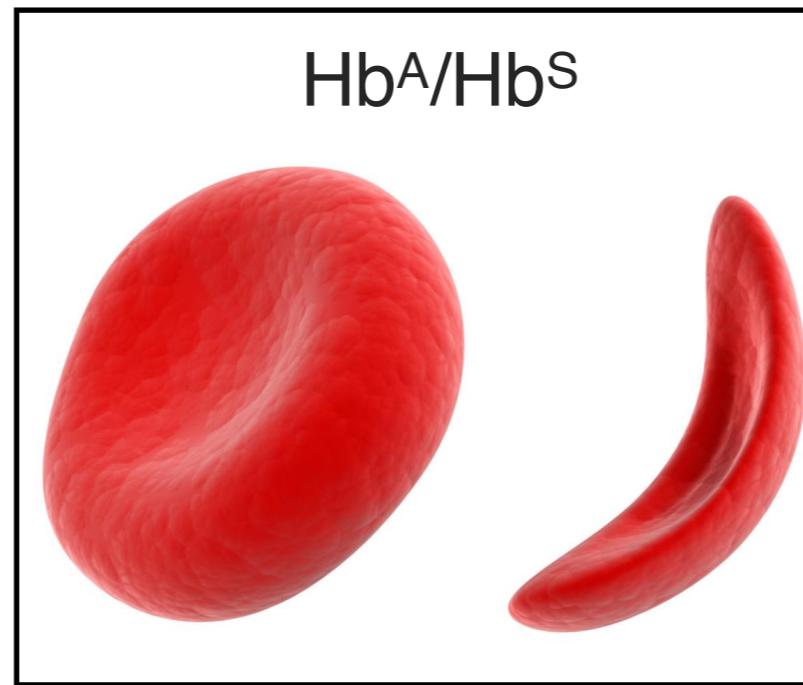
Mostly sickle
RBCs

Co-dominant

Hemoglobin sickle-cell allele



$\text{Hb}^{\text{A}}/\text{Hb}^{\text{A}}$



$\text{Hb}^{\text{A}}/\text{Hb}^{\text{S}}$



$\text{Hb}^{\text{S}}/\text{Hb}^{\text{S}}$

Normal
RBCs

Both normal and sickle
RBCs

Mostly sickle
RBCs

Malaria-sensitive

Malaria-resistant

Malaria-resistant

Dominant
Co-dominant

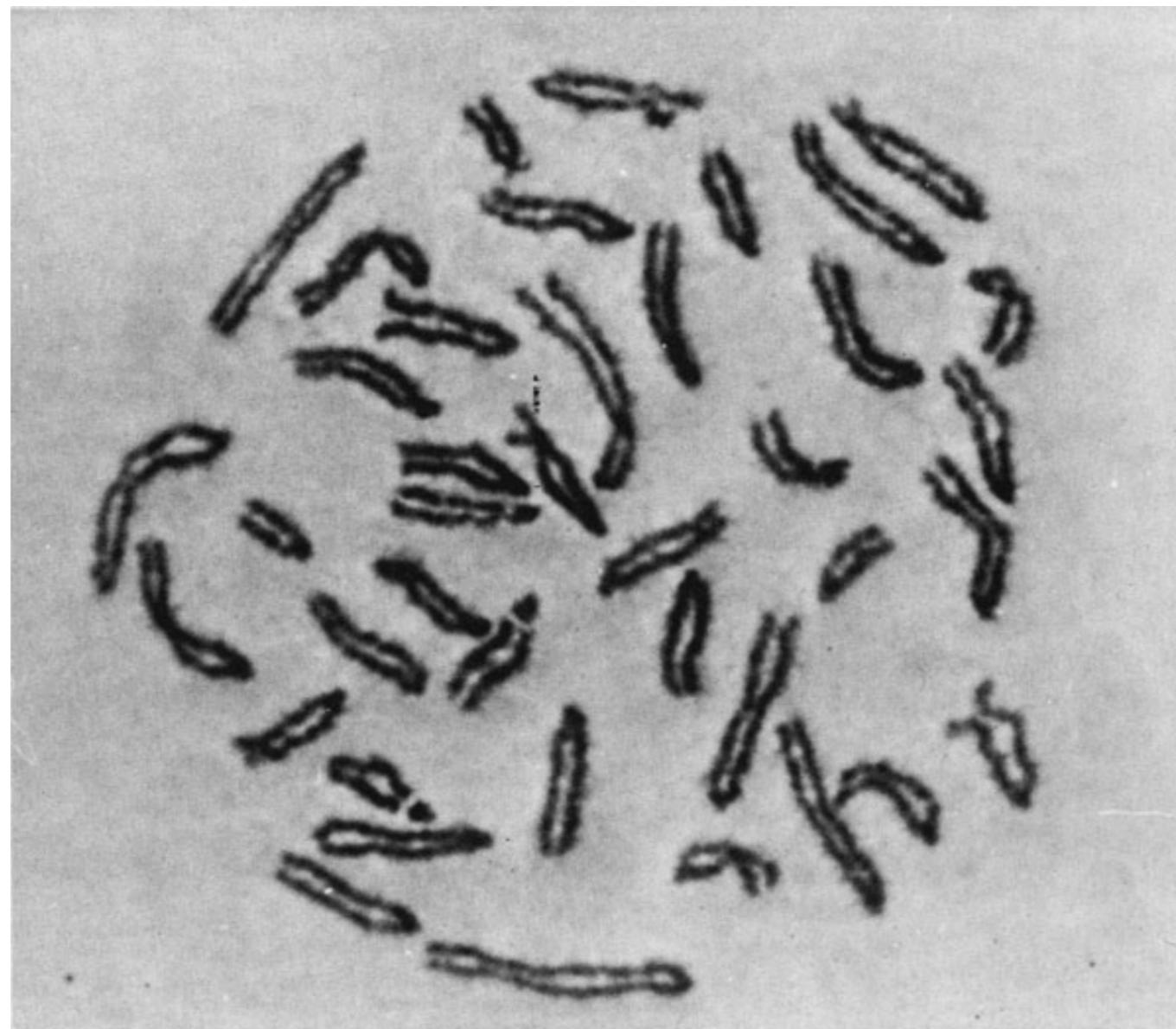
Hemoglobin sickle-cell allele

Hb^A/Hb^A	Hb^A/Hb^S	Hb^S/Hb^S
		
Normal RBCs	Both normal and sickle RBCs	Mostly sickle RBCs
Malaria-sensitive	Malaria-resistant	Malaria-resistant
No sickling disease	No sickling disease	Severe sickling disease

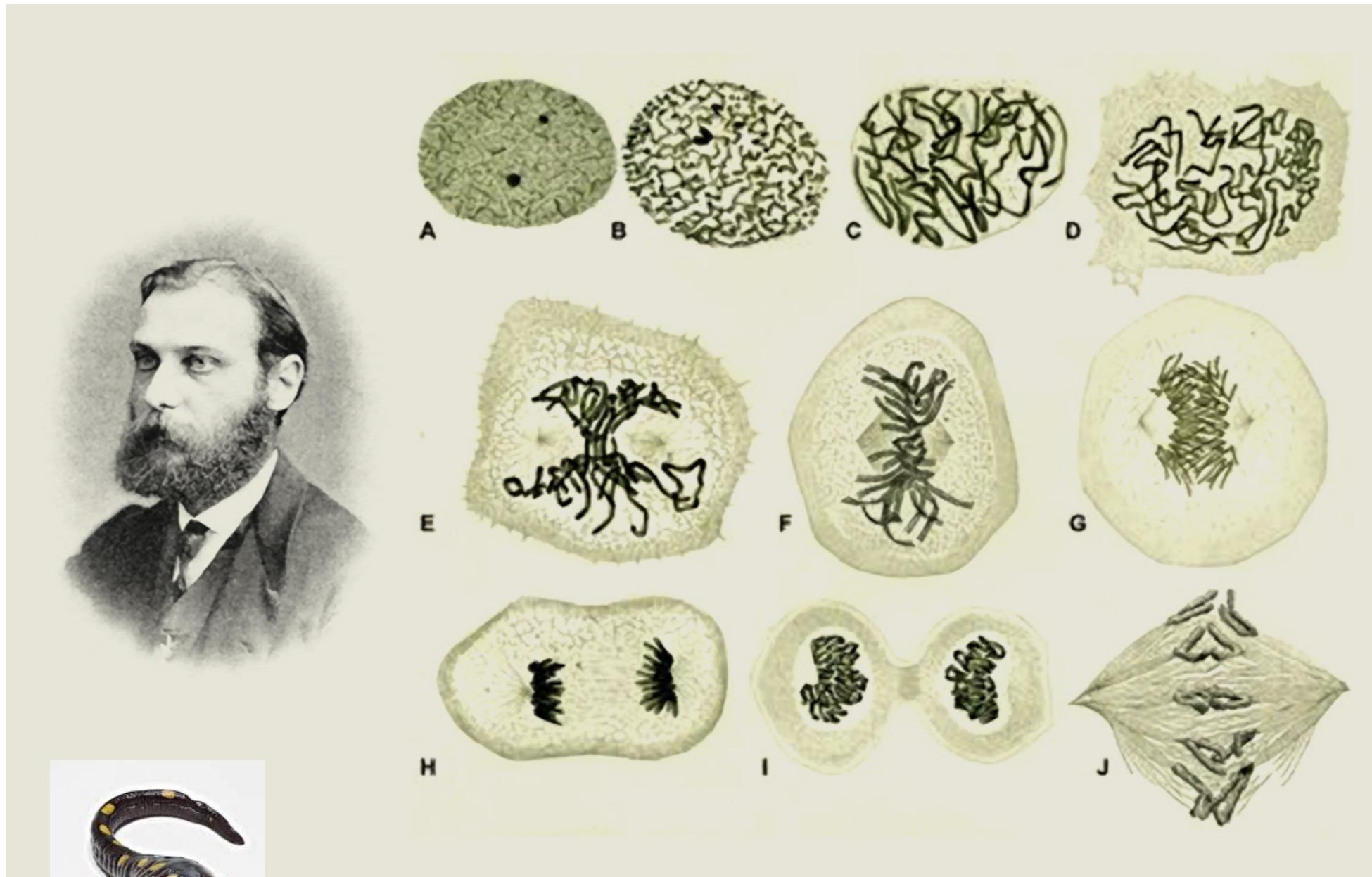
Is Hb^S a dominant or recessive allele?

Bio393: Biomedical Genetics

Chromosome theory, recombination, and mapping



Walther Flemming stained cells



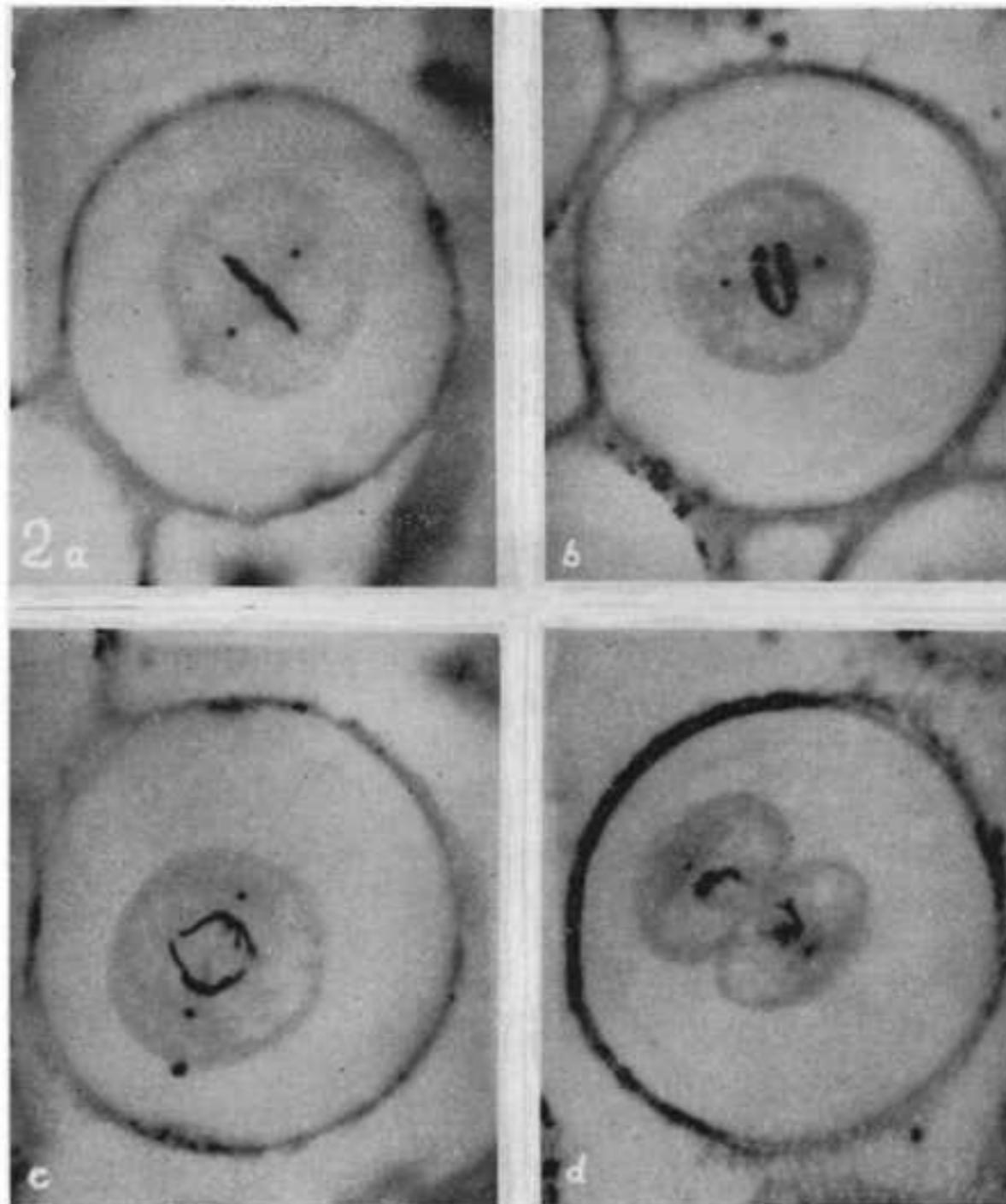
Walther Flemming, 1882



Cells divide their chromosomes with high fidelity



Theodor Boveri

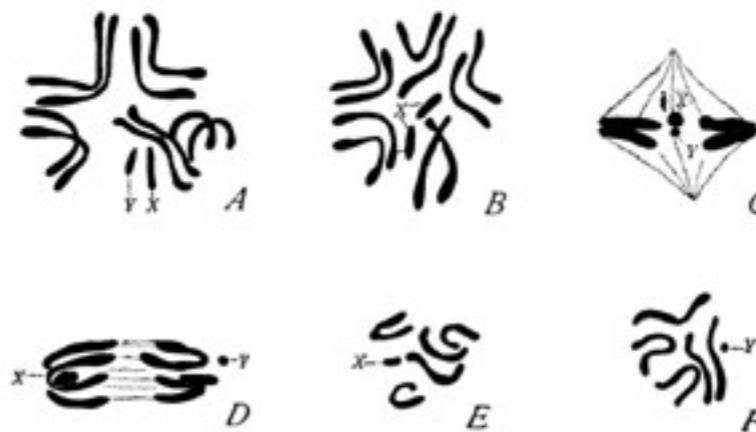


Discovery of sex chromosomes



Courtesy of the Marine Biological Laboratory.
Noncommercial, educational use only.

Nettie Stevens



Tenebrio melitor

then 50 beetle species and nine species of fly!

Lecture 2

Gametes have half the chromosomes of the soma



Theodor Boveri



Parascaris equorum



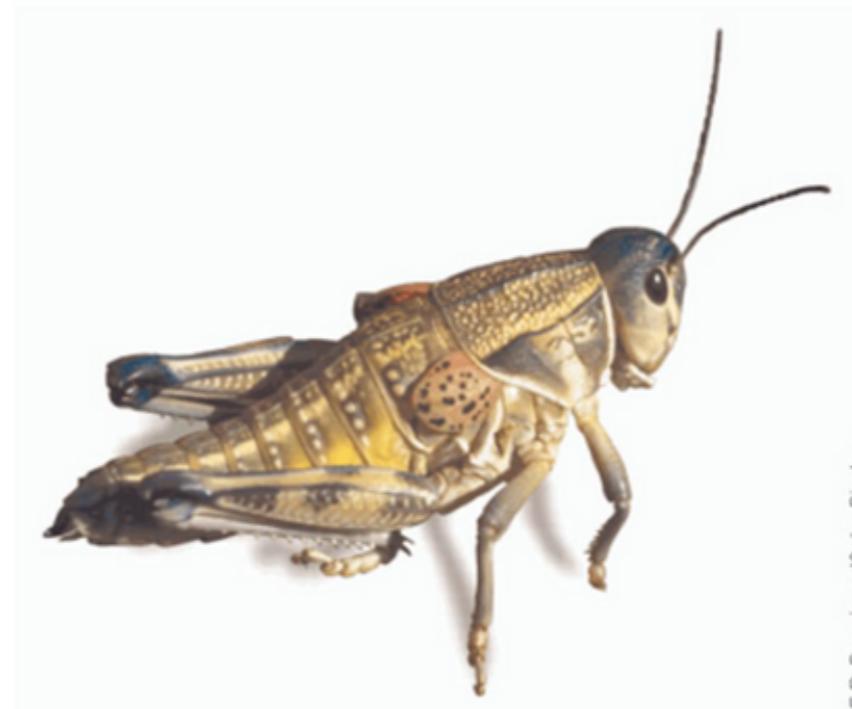
Discovery of a connection to Mendel's principles



Walter Sutton



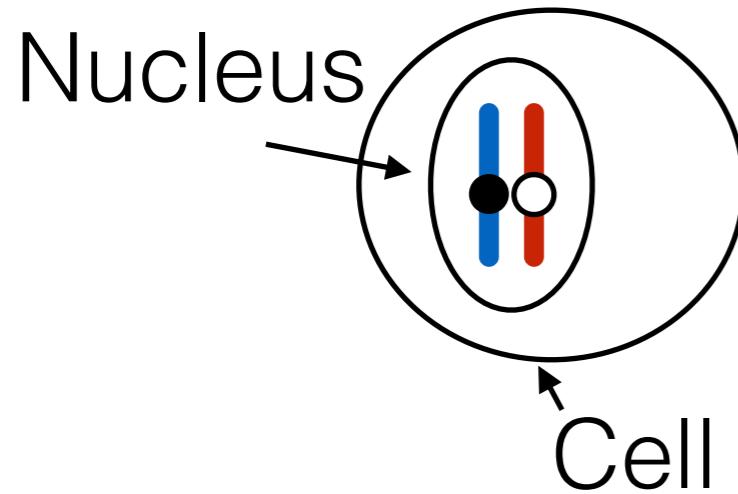
Michael Abbey/Photo Researchers, Inc.



E.R. Deggingen/Color-Pic, Inc.

- Gametes have half chromosome complement of somatic cells
- Homolog separation to gamete was random

Terms for mitosis and meiosis



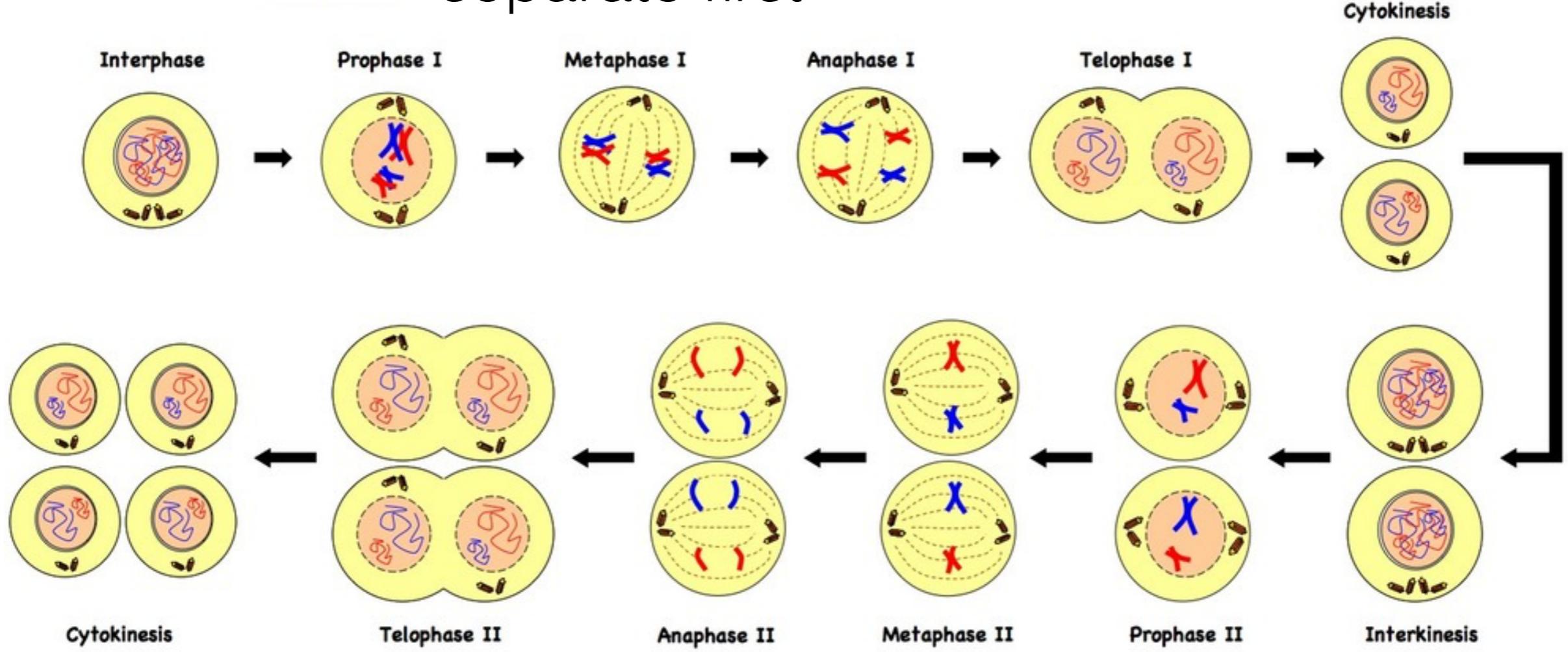
- Chromosome
- Pair of homologs (2N)
- Sister chromatids

- Ploidy (N)
- Diploid (2N)
- Haploid (1N)
- Polyplloid (>2N)
- Gamete

Meiosis: A reductional division in two acts

Homologs

MEIOSIS I separate first



Sisters
separate last

Keep track of
centromere

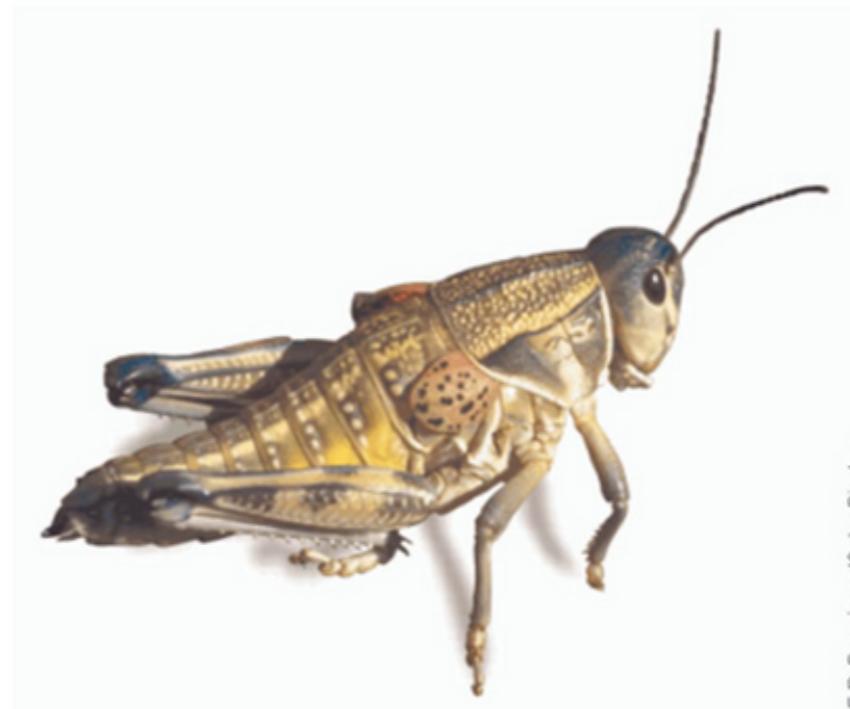
Discovery of a connection to Mendel's principles



Walter Sutton



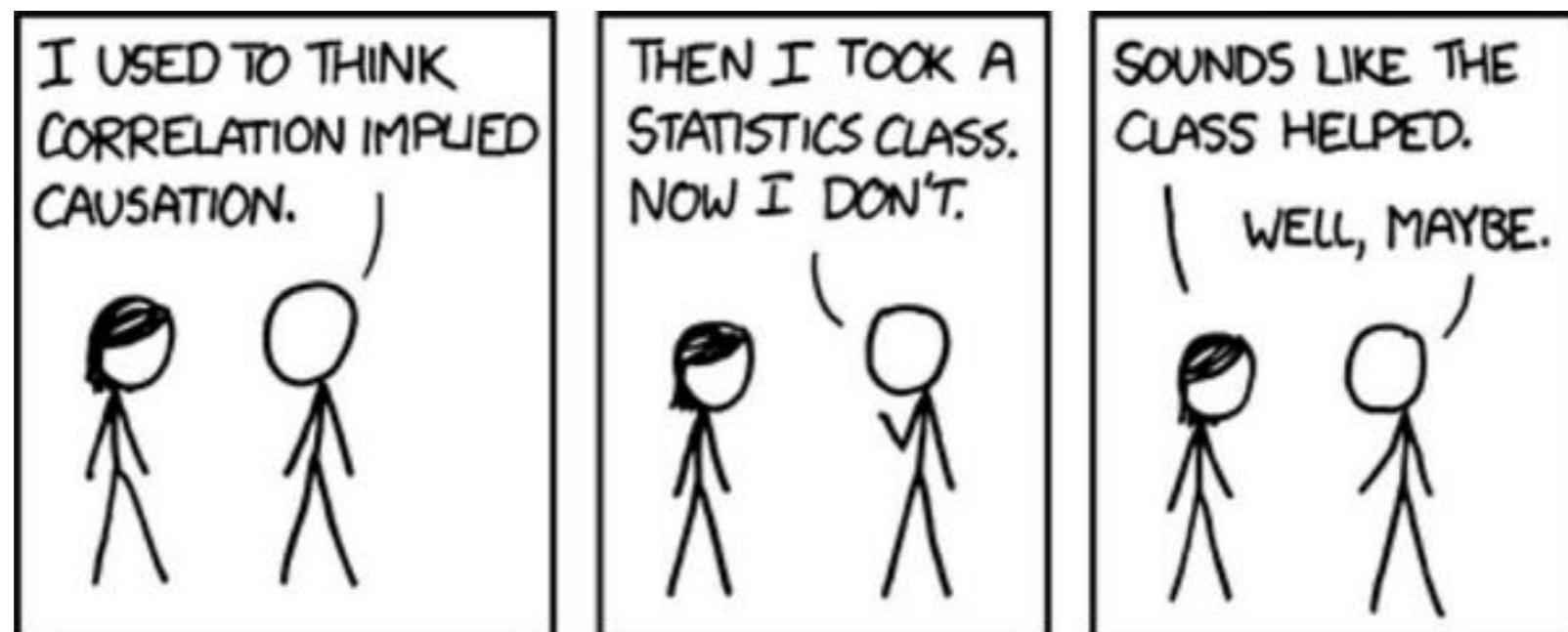
Michael Abbey/Photo Researchers, Inc.



E.R. Deggingen/Color-Pic, Inc.

- Gametes have half chromosome complement of somatic cells
- Homolog separation to gamete was random

Correlation does not mean causation



xkcd.com



Thomas Hunt Morgan

Drosophila melanogaster: genetics superstar



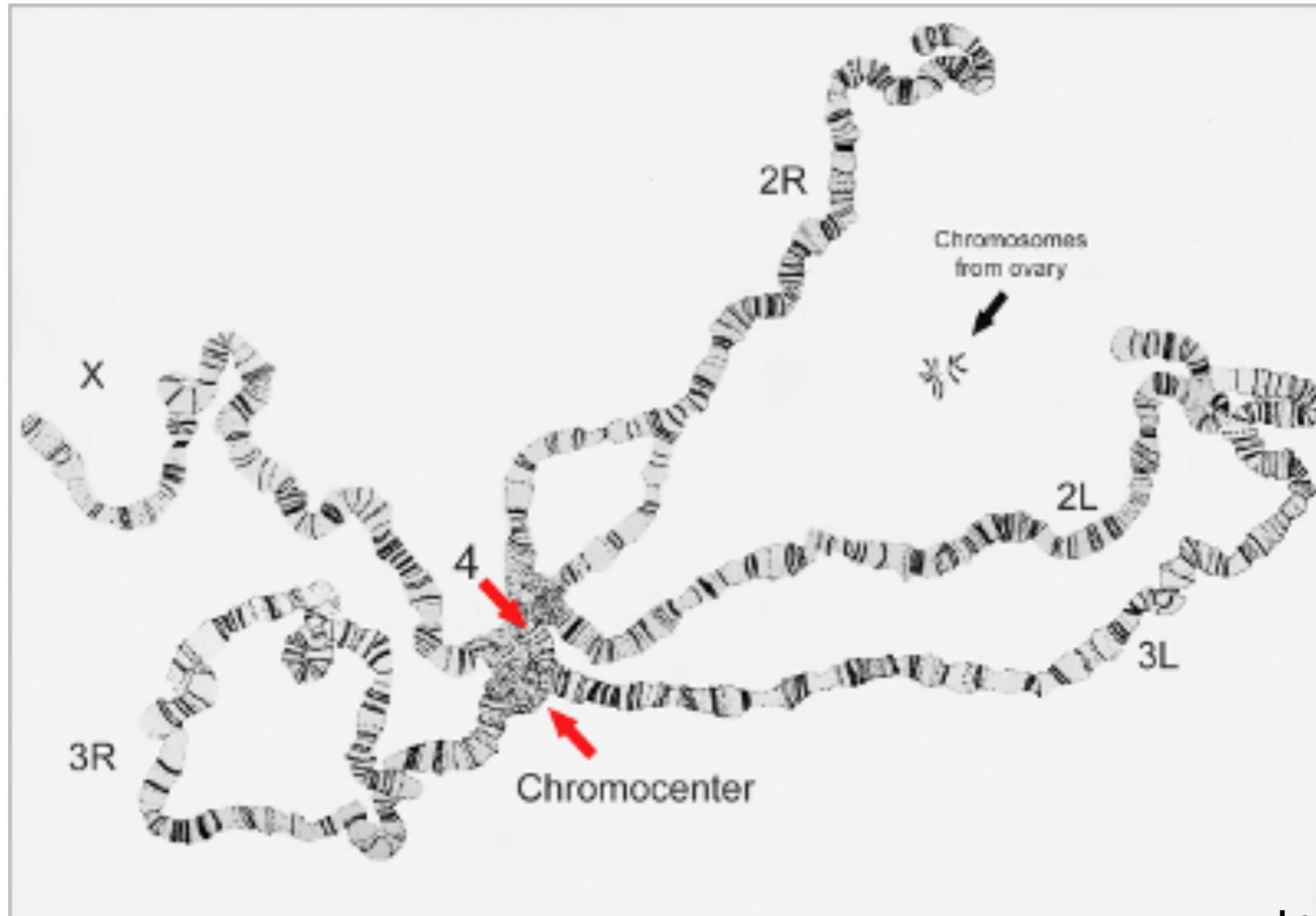
Courtesy of the Marine Biological Laboratory.
Noncommercial, educational use only.

Nettie Stevens

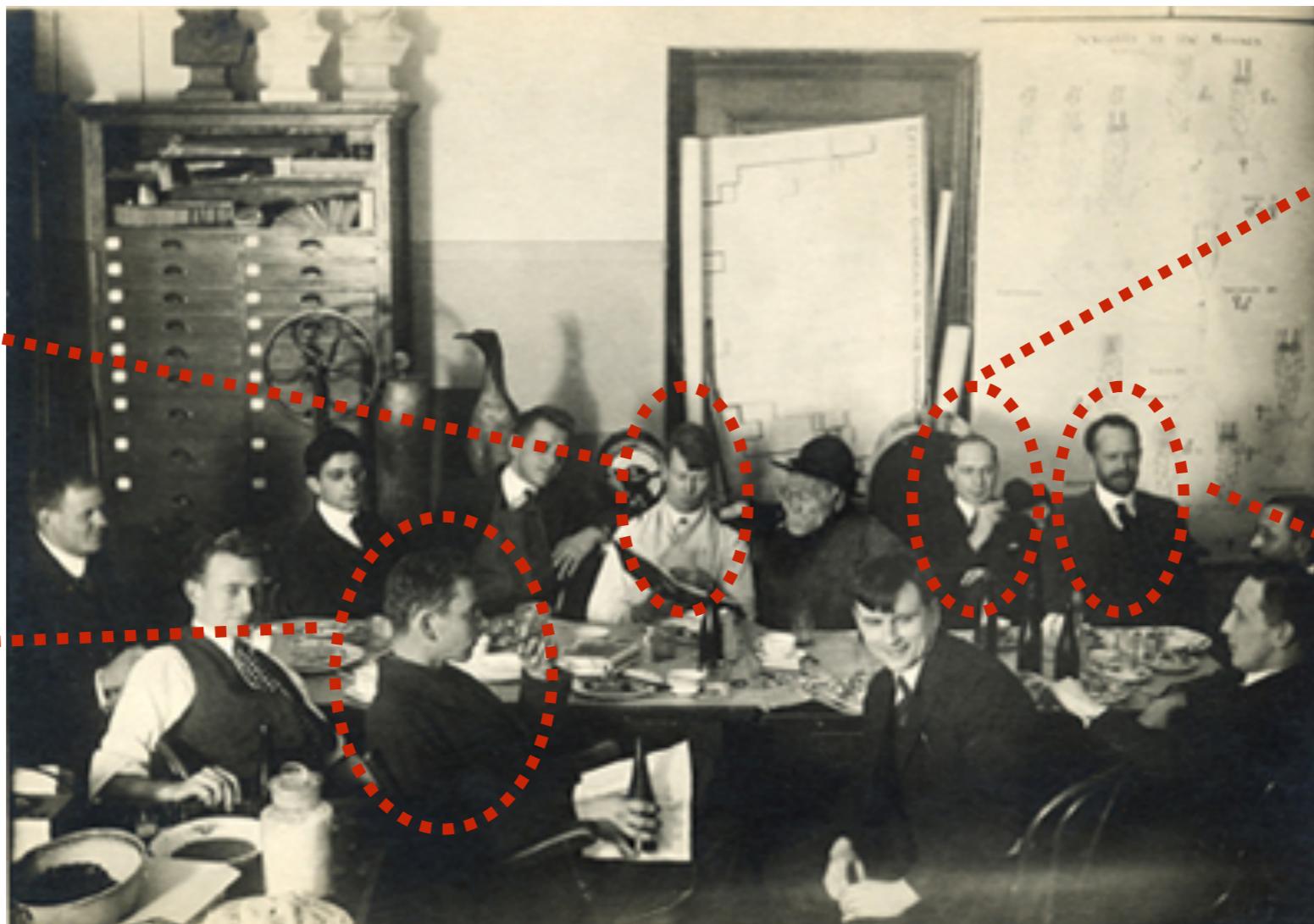


Thomas Hunt Morgan

Drosophila polytene chromosomes allow us to directly visualize genetic principles



The fly room at Columbia



Calvin Bridges



Alfred Sturtevant

Hermann Muller



Thomas H. Morgan



***W*⁺**

W



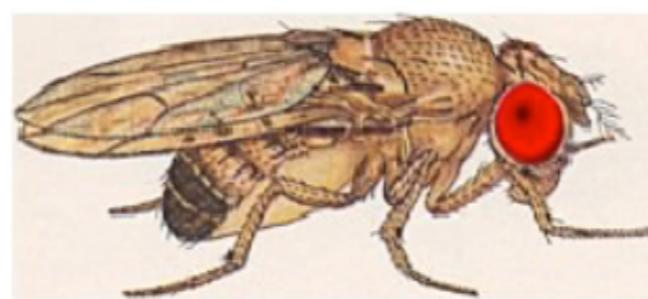


♂

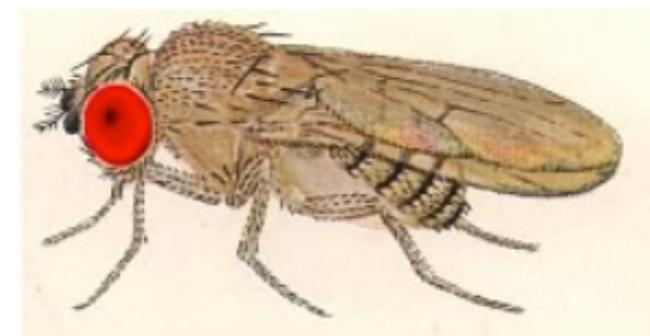
X



♀



♂



♀

What is dominance relationship of *white* mutant allele?



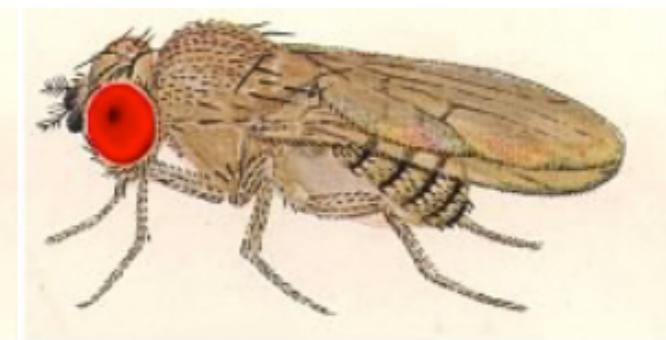
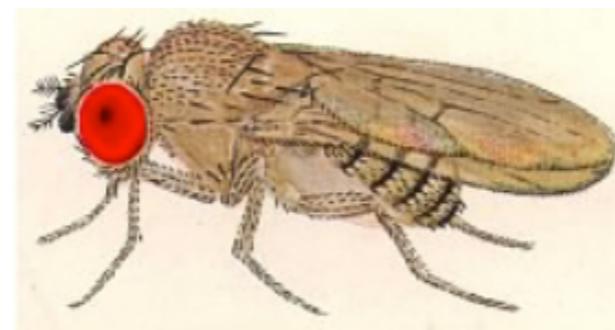
X



♂



♀



♂

♂

♀

♀

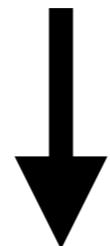
The reciprocal cross



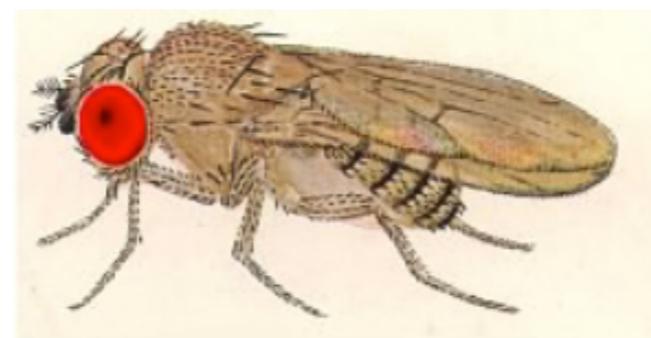
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♂

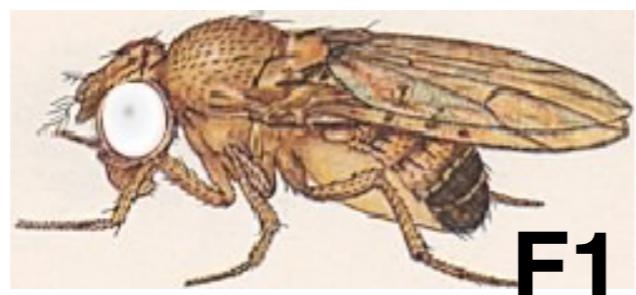


♀



♂

♀



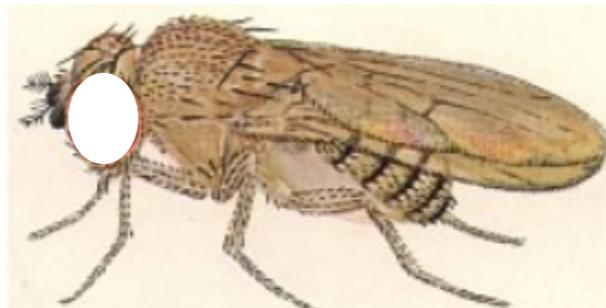
X



♂



♀



♂

♂

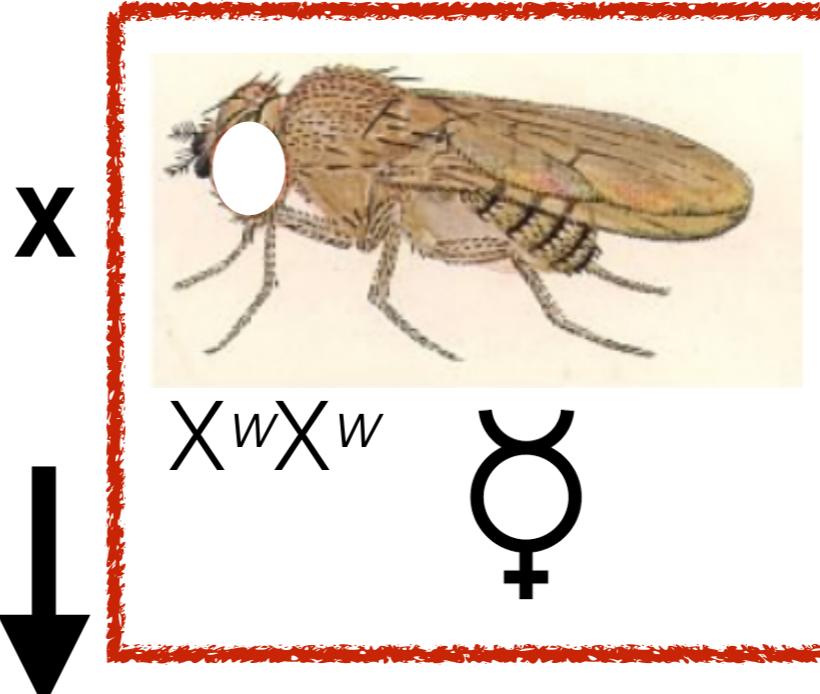
♀

♀

Equal ratios of each sex and eye color



$X^{w+}Y$ ♂



1999/2000
offspring



X^wY ♂

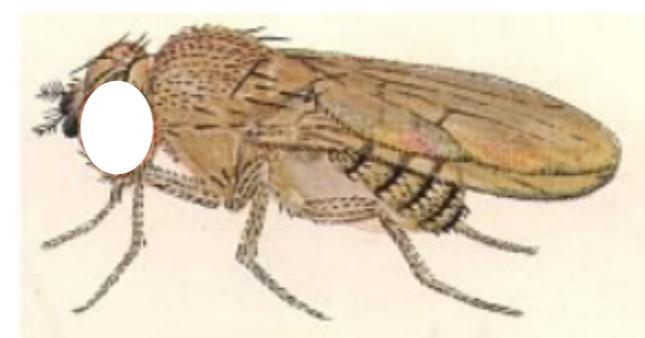


X^wX^{w+} ♀

1/2000
offspring

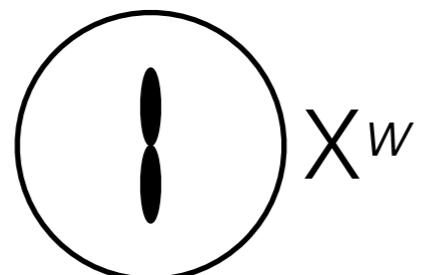
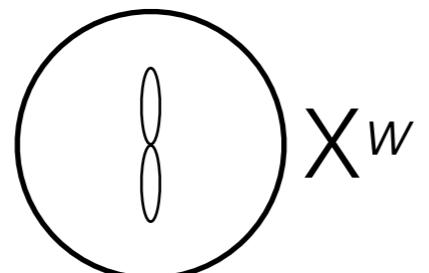
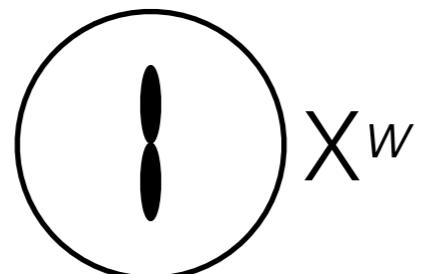
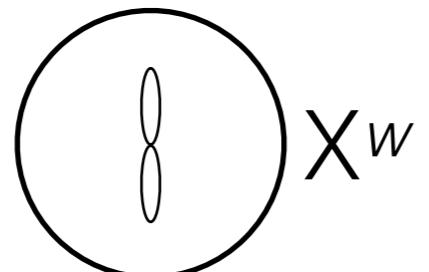


♂

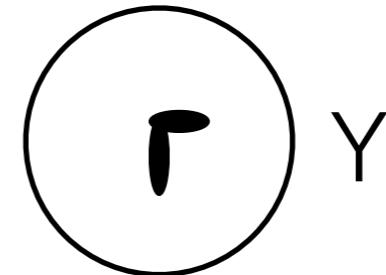
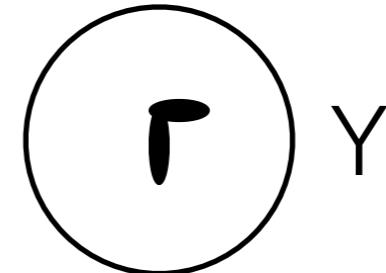
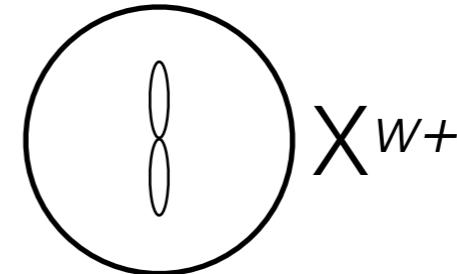
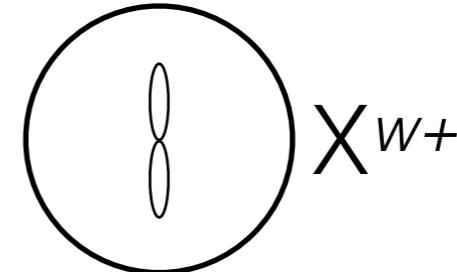


♀

Female
gametes

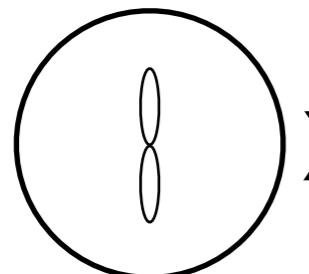
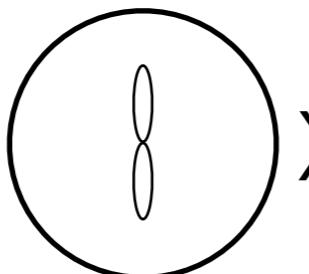
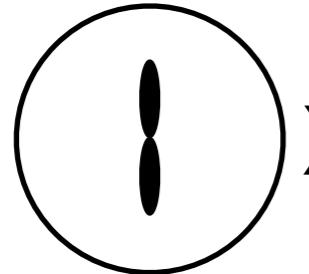
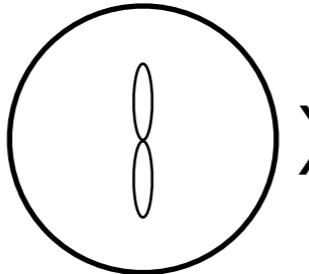
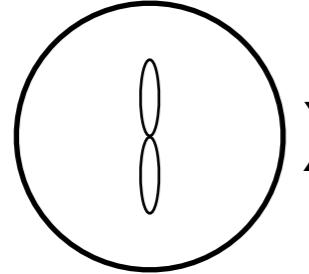
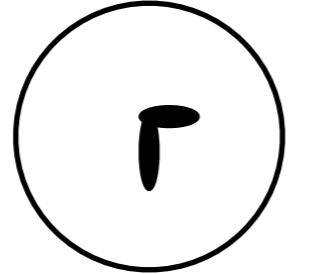
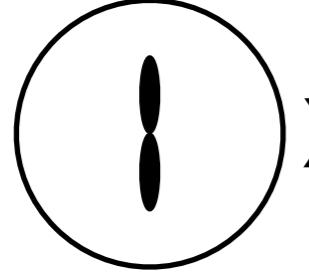
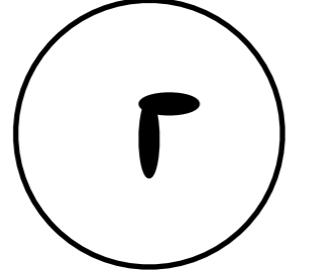
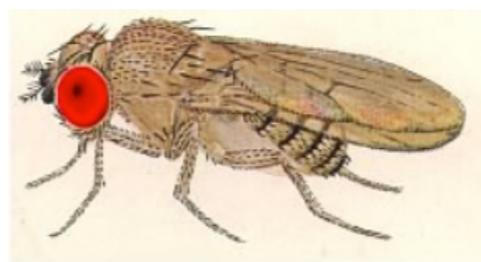


Male
gametes

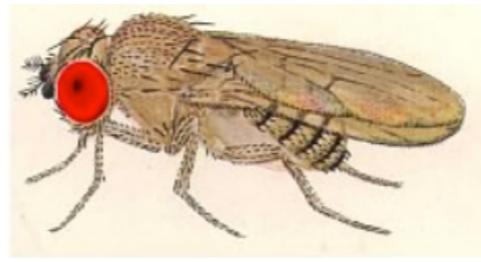
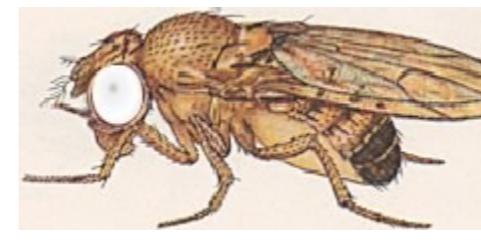
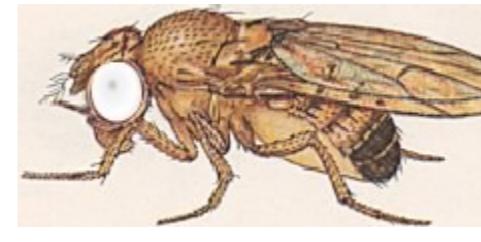


1999/2000
offspring

Female
gametes Male
gametes

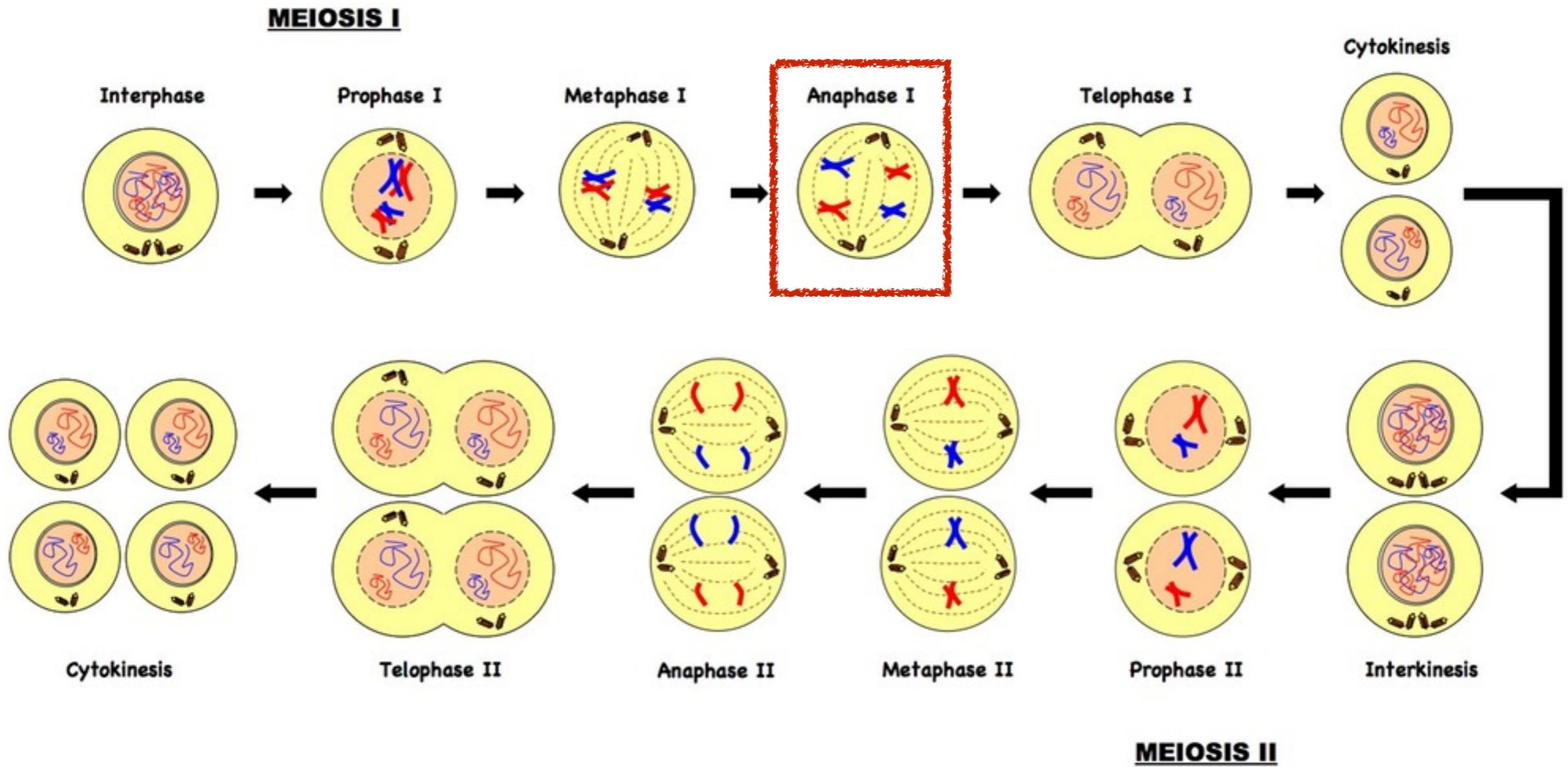
 X^w  X^{w+}  X^w  X^{w+}  X^w  Y  X^w  Y 

Offspring

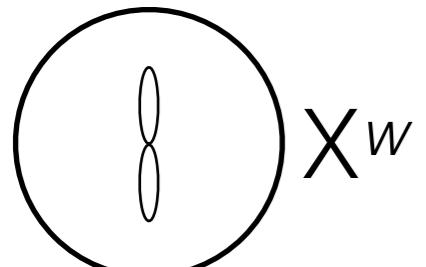
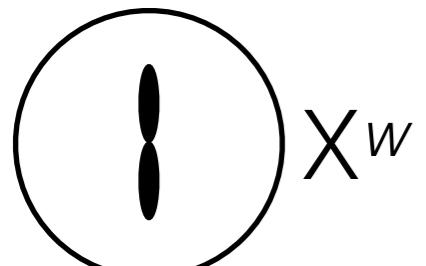
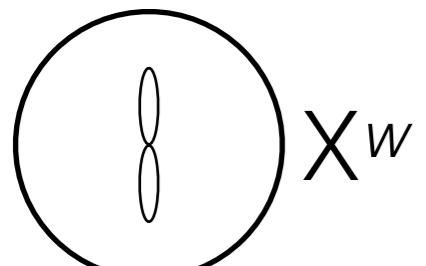
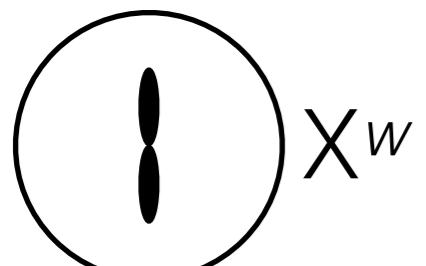
 X^wX^{w+}  X^wX^{w+}  X^wY  X^wY

What is going on with the rare (1/2000) class?

Meiotic non-disjunction I



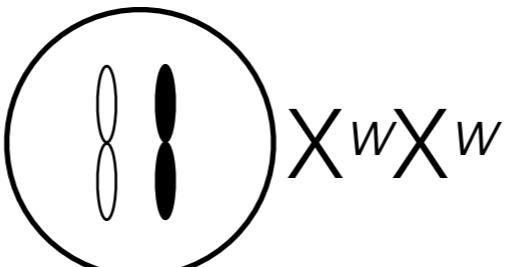
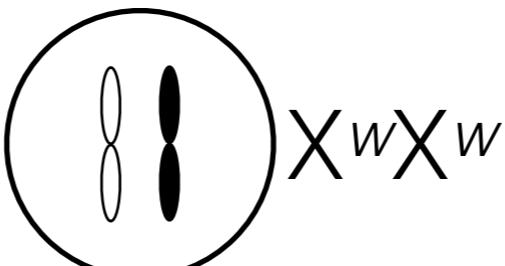
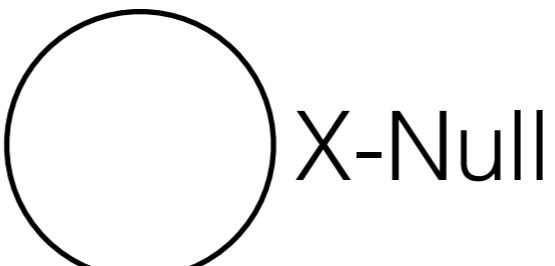
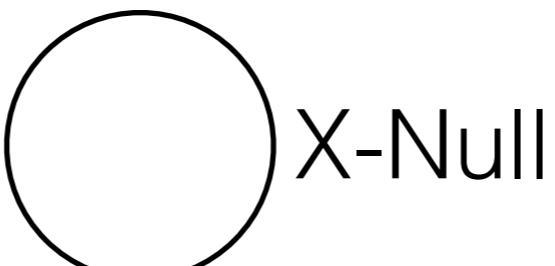
Female
gametes

 X^w  X^w  X^w  X^w

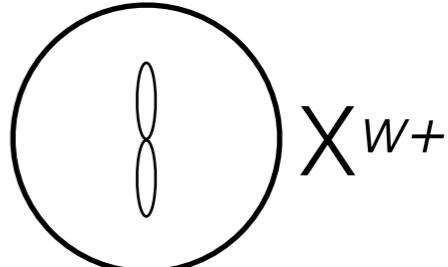
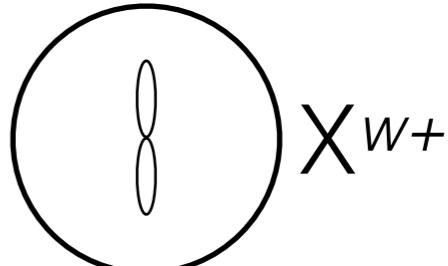
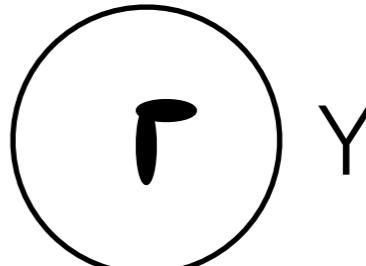
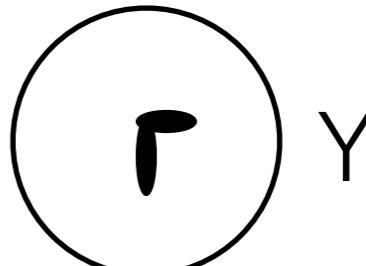
1999/2000
offspring

 $X^{w+}X^w$ X^wY

Meiosis I NDJ
Female gametes

 X^wX^w  X^wX^w  $X\text{-Null}$  $X\text{-Null}$

Male
gametes

 X^{w+}  X^{w+}  Y  Y

1/2000
offspring

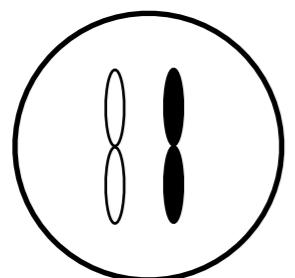
 $X^{w+}0$ X^wX^wY

red male

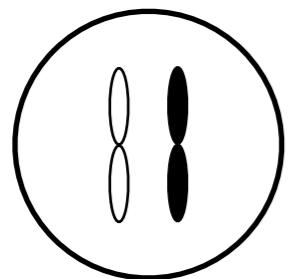
white female

Two different types of female gametes

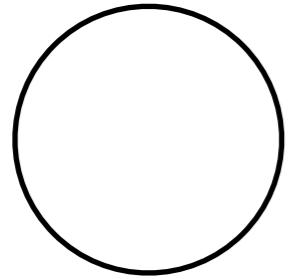
Meiosis I NDJ
Female gametes



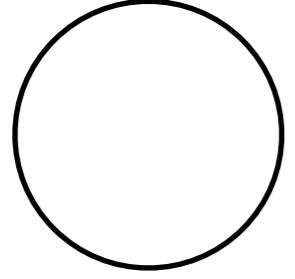
X^wX^w



X^wX^{w+}

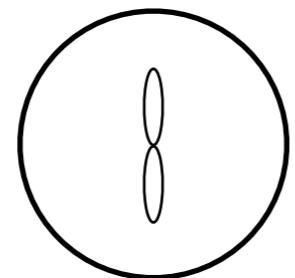


X-Null

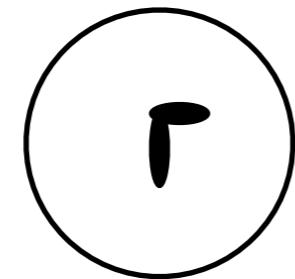


X-Null

Male
gametes



X^{w+}



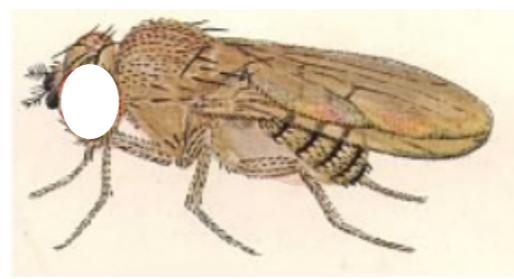
Y



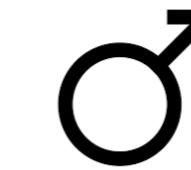
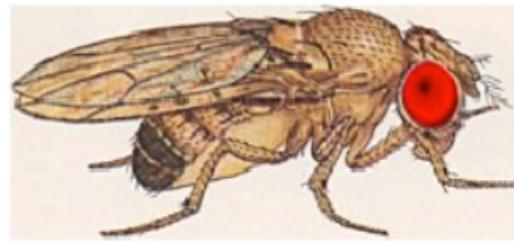
Offspring

Dead

$X^wX^wX^{w+}$



X^wX^wY



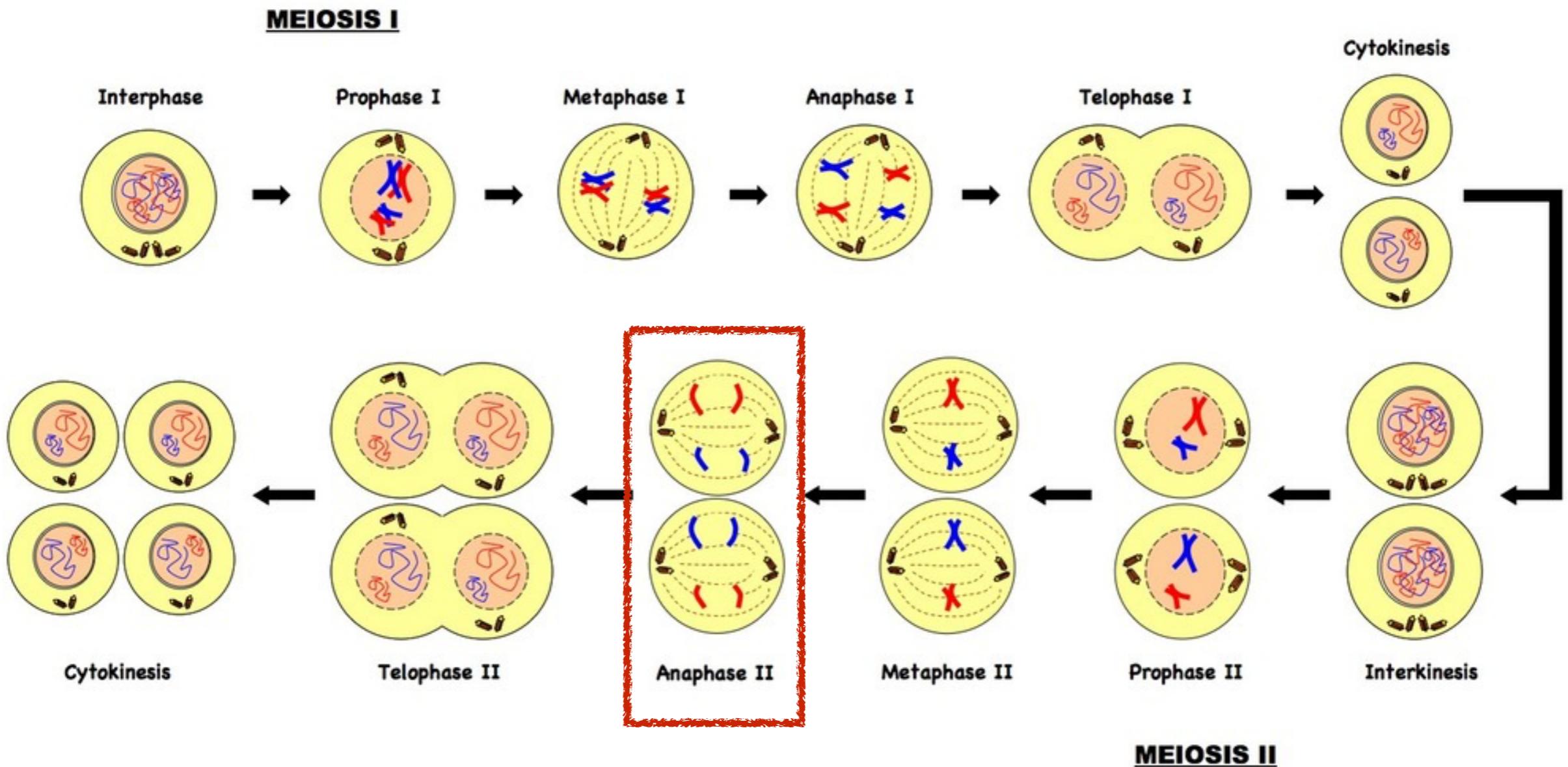
$X^{w+}0$



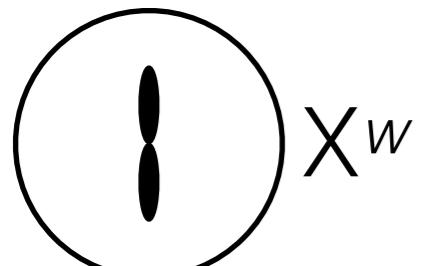
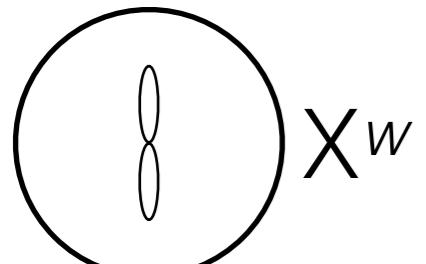
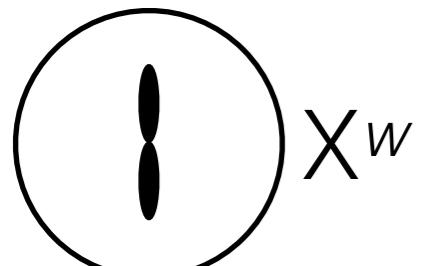
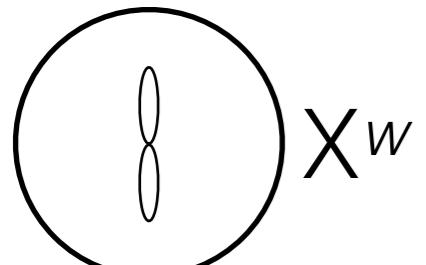
Dead

0Y

Meiotic non-disjunction II



Female
gametes

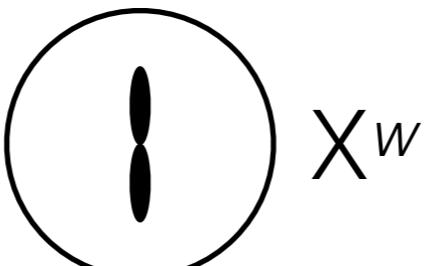
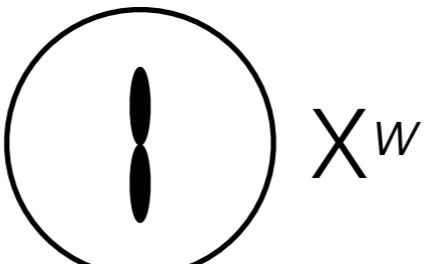
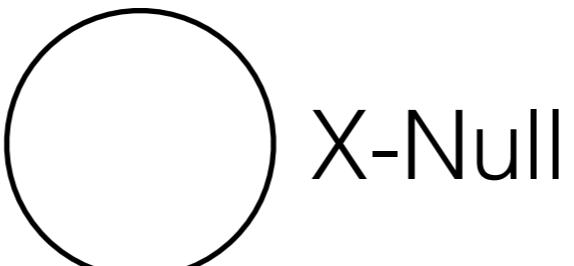


1999/2000
offspring

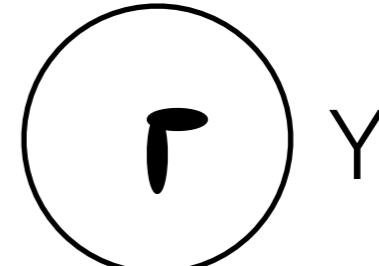
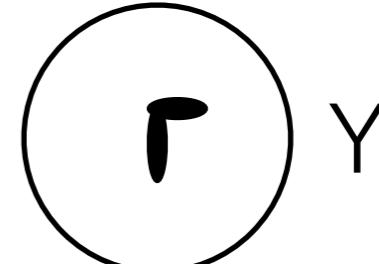
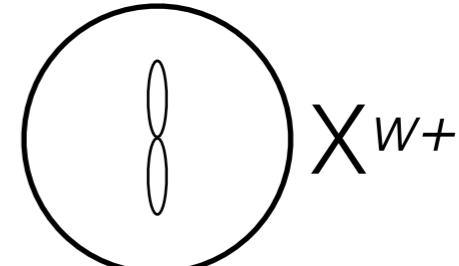
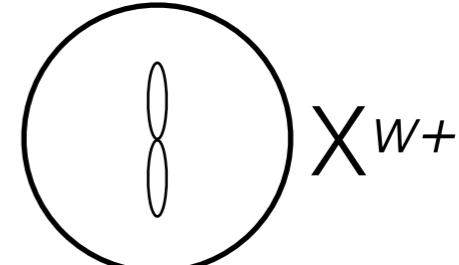
$X^{w+}X^w$

X^wY

Meiosis II NDJ
Female gametes



Male
gametes



1/2000
offspring

$X^{w+}0$

X^wX^wY

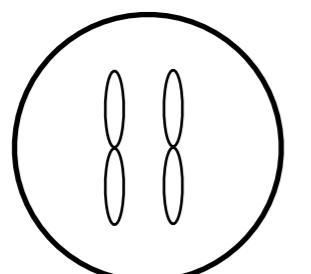
red male

white female

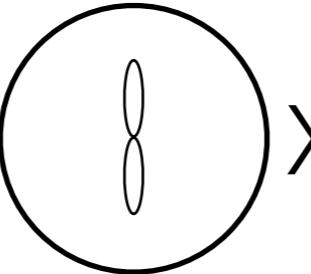
Three different types of female gametes

Meiosis II NDJ
Female gametes

Male
gametes



X^wX^w



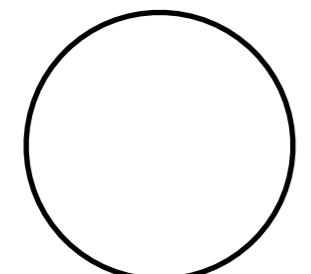
X^{w+}



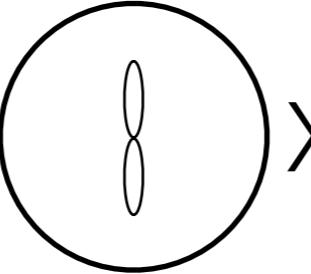
Offspring

Dead

$X^wX^wX^{w+}$



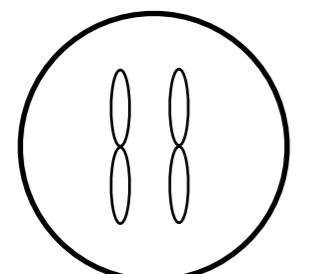
X-Null



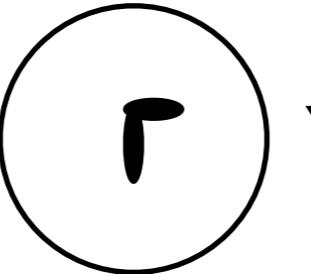
X^{w+}



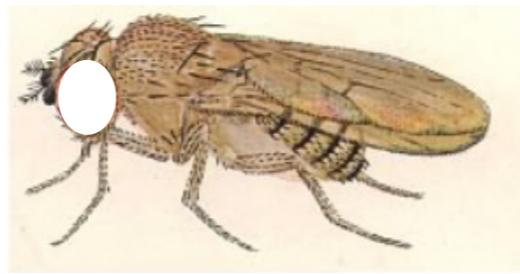
$X^{w+}0$



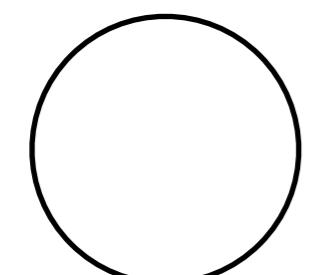
X^wX^w



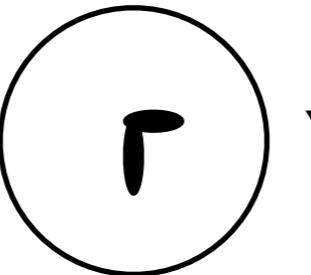
Y



X^wX^wY



X-Null

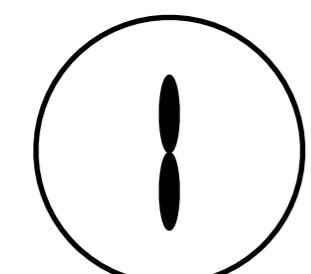


Y

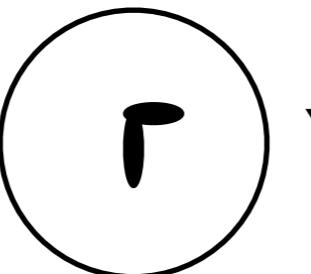


Dead

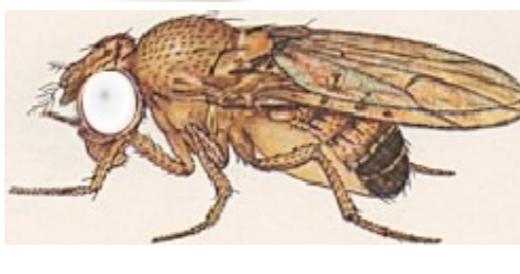
0Y



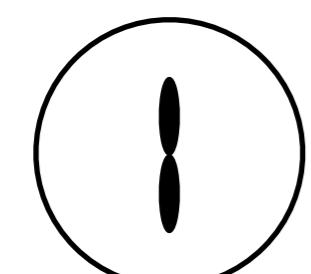
X^w



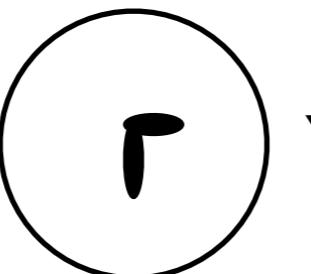
Y



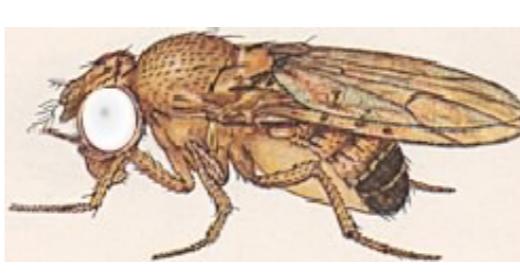
X^wY



X^w



Y



X^wY

The connections between chromosome NDJ and a trait was made by Stevens and Bridges



Courtesy of the Marine Biological Laboratory.
Noncommercial, educational use only.

Nettie Stevens



Calvin Blackman Bridges, 1927.
Photo courtesy of Cold Spring Harbor
Laboratory Archives.

Calvin Bridges



**Polytene
chromosomes**

Why did the first cross not indicate to them that something weird was going on?

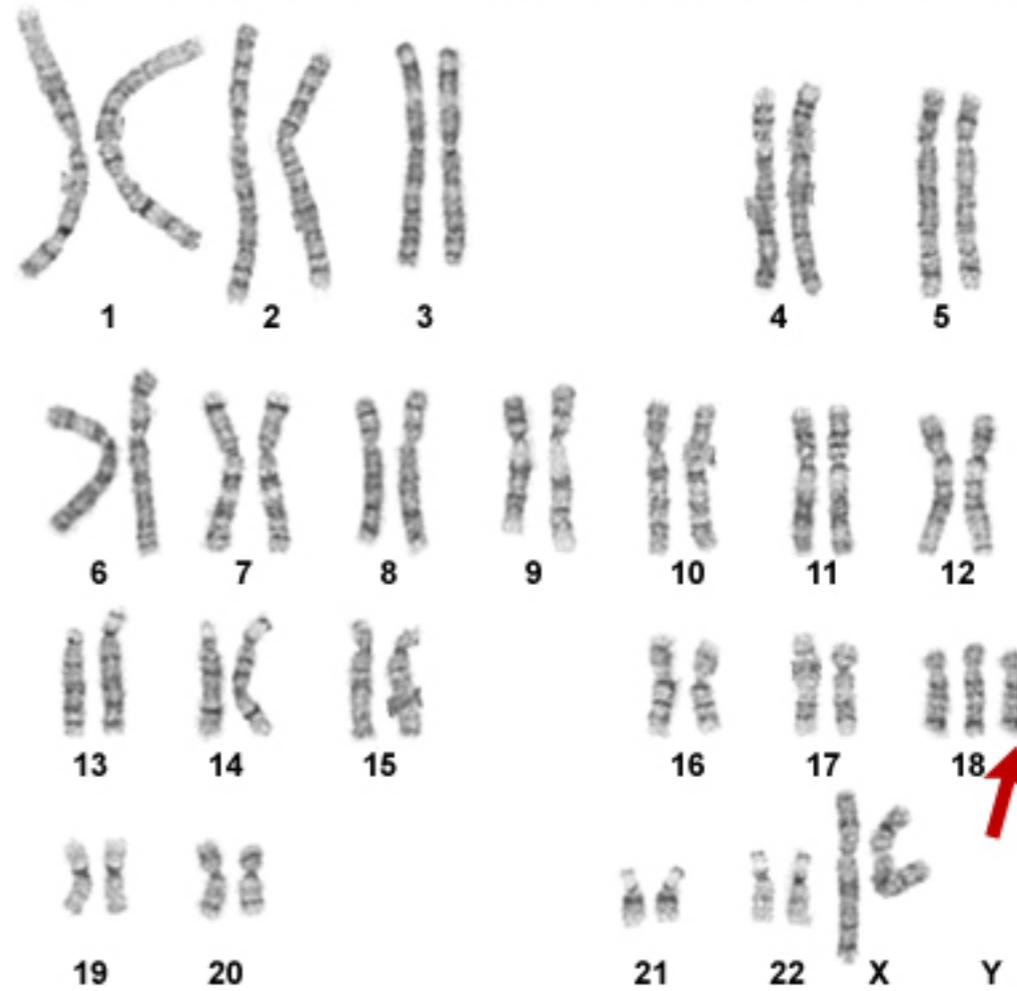
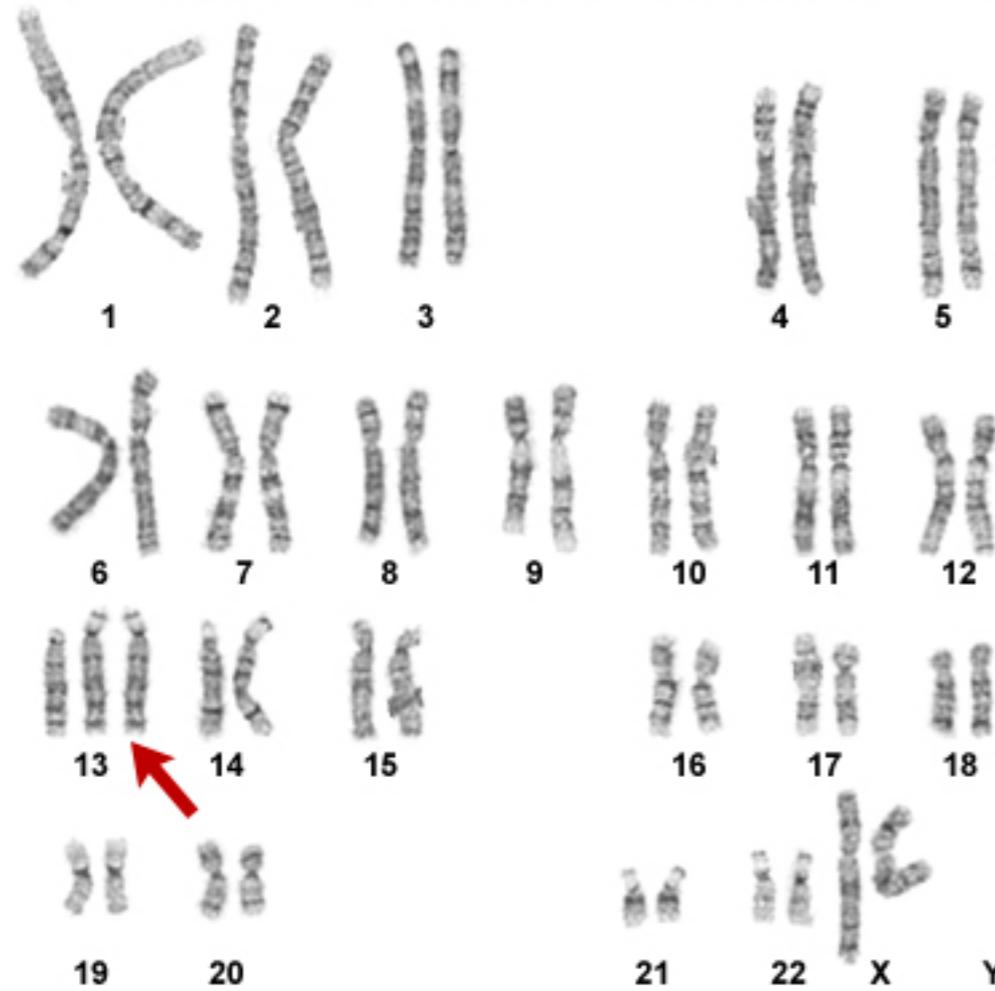
X^wY

X

$X^{w+}X^{w+}$

How can you tell the difference
between Meiosis I NDJ and Meiosis II NDJ?

Non-disjunction is a relatively common error - not just the X chromosome aneuploidy



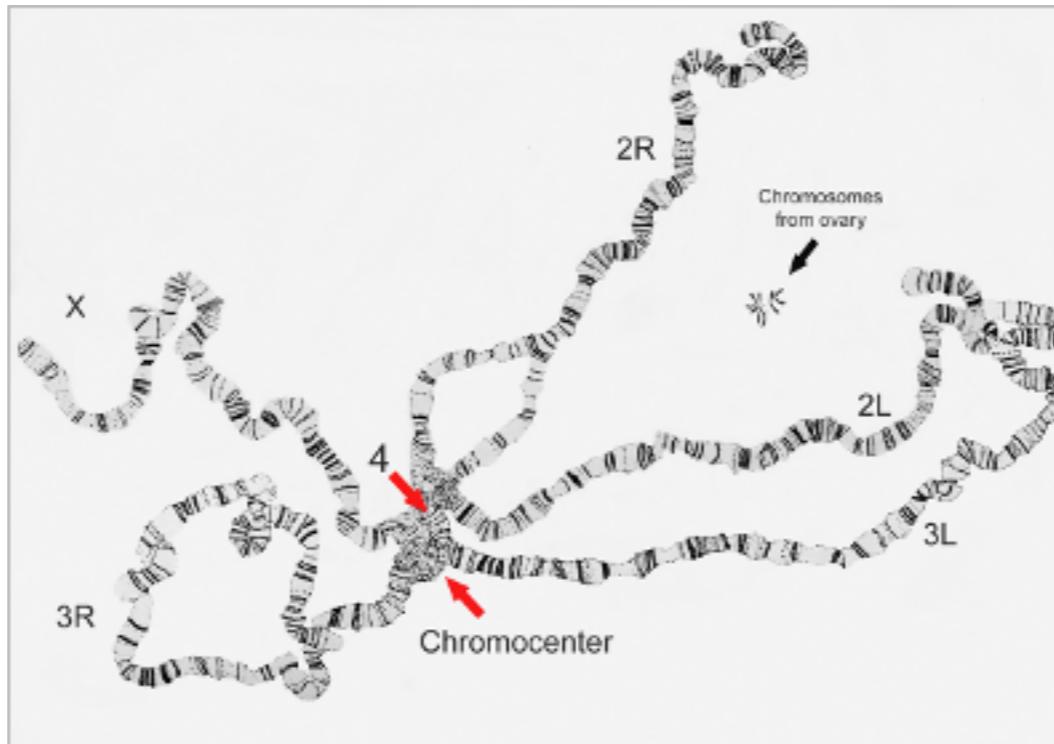
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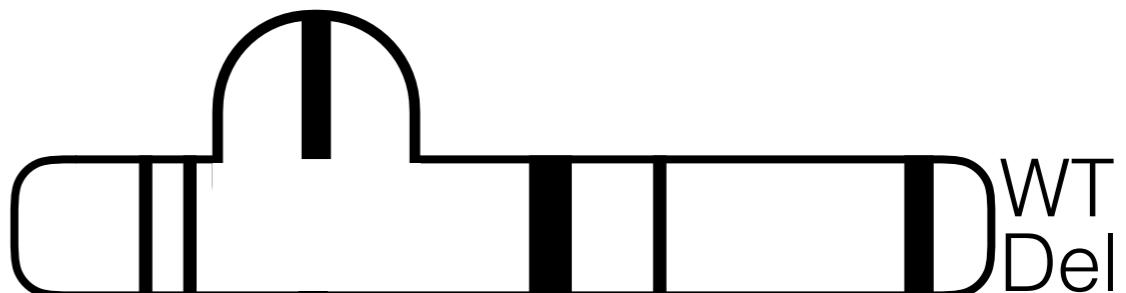
Non-disjunction is a relatively common error - not just the X chromosome aneuploidy



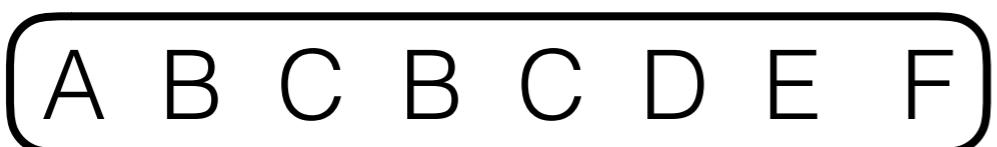
Chromosomal abnormalities



WT



Deletion BC



Duplication BC

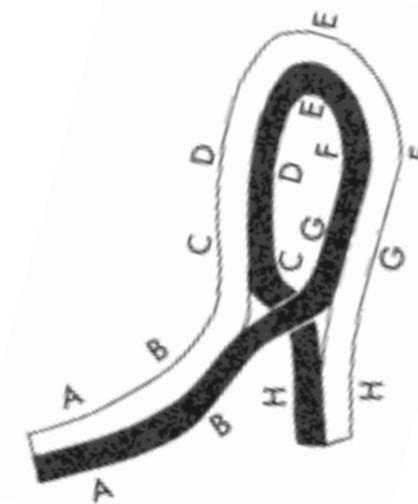
Chromosomal abnormalities



WT



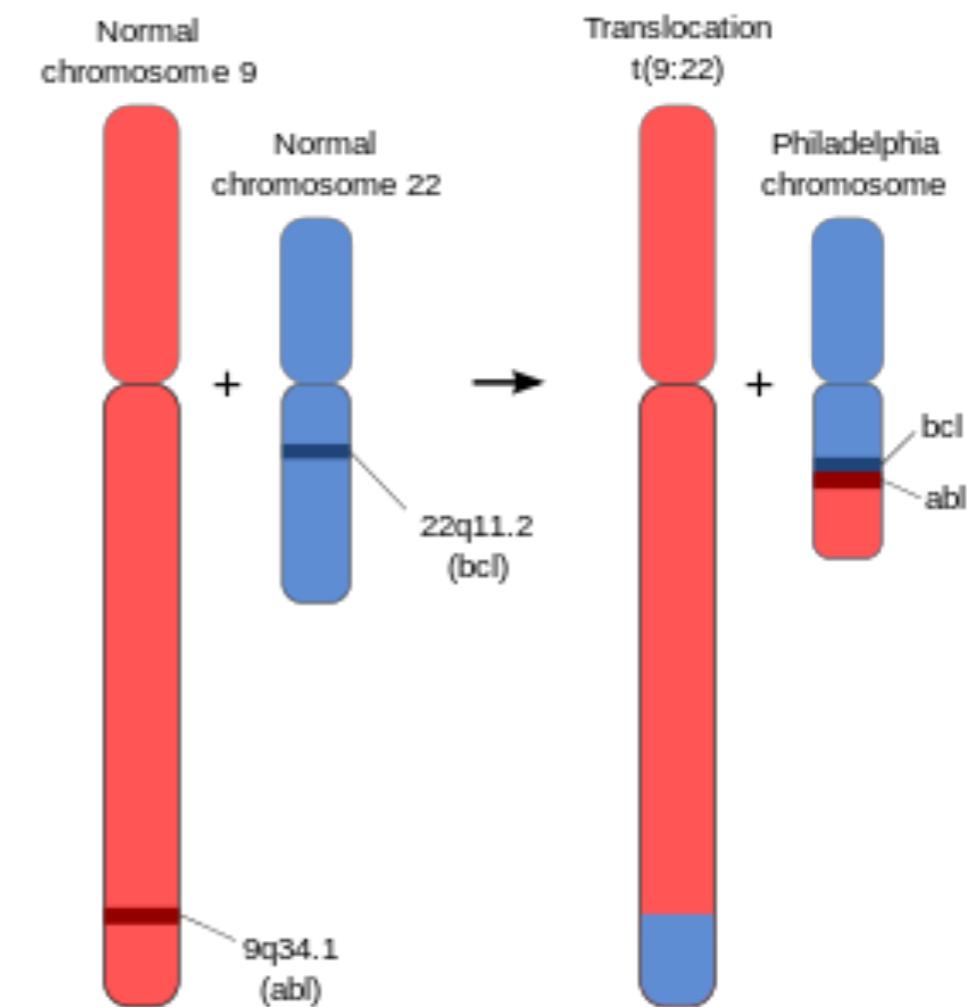
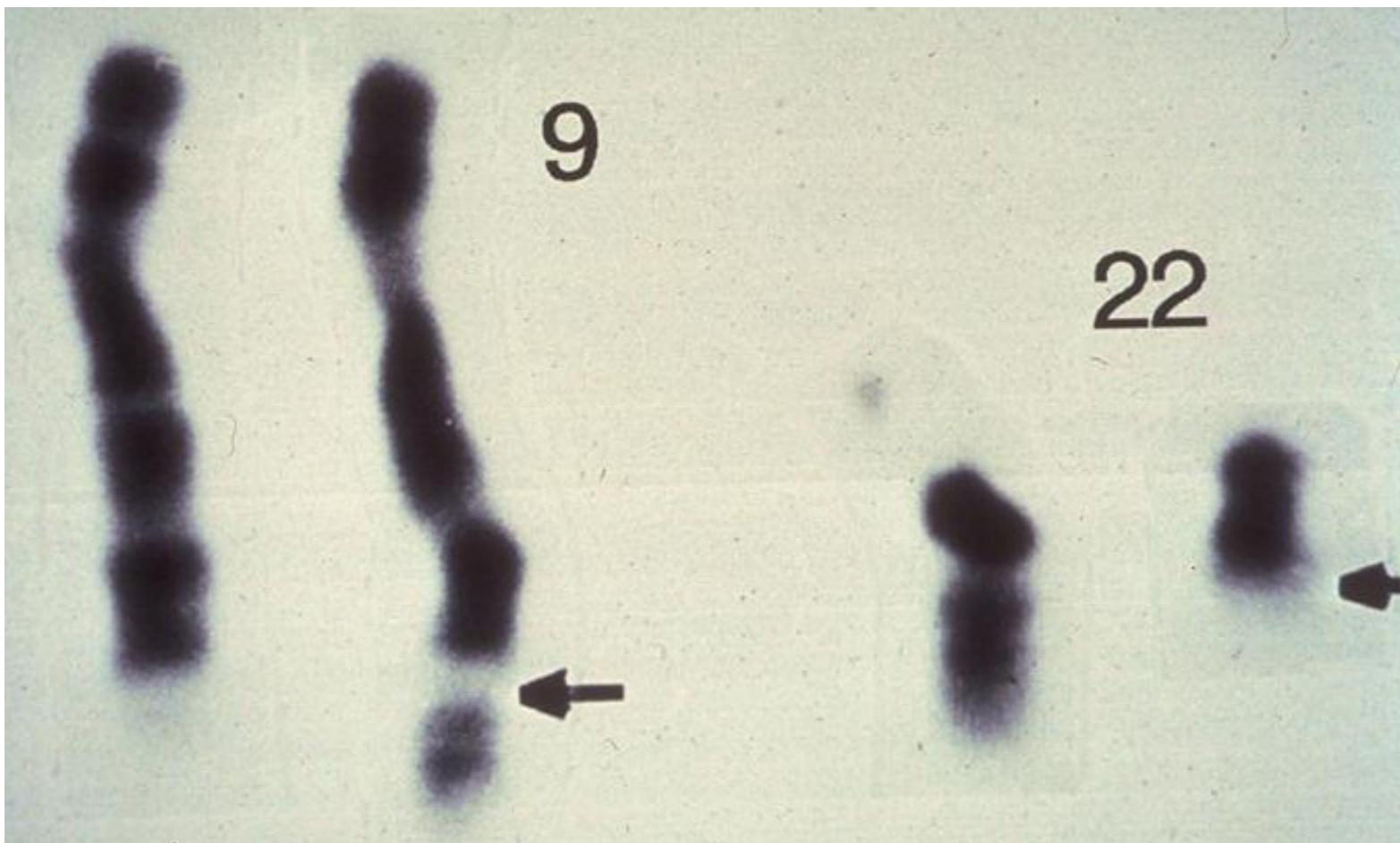
Inversion BCD



Translocation ABC-XYZ

Fusion of two chromosomes

The Philadelphia chromosome: translocation



Janet Rowley