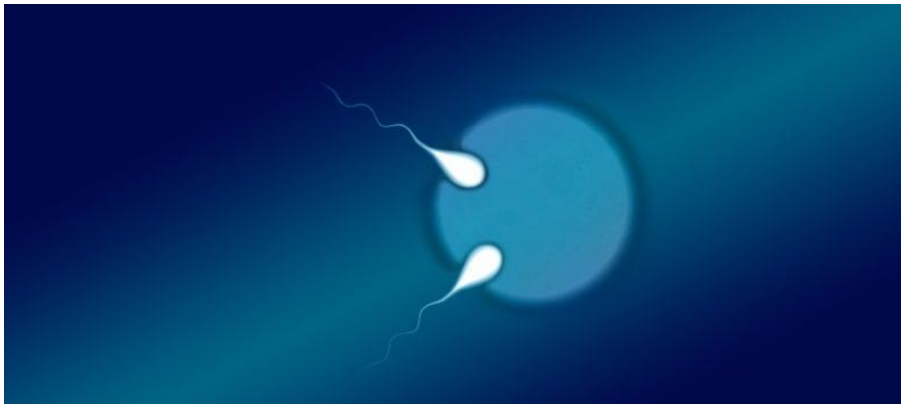
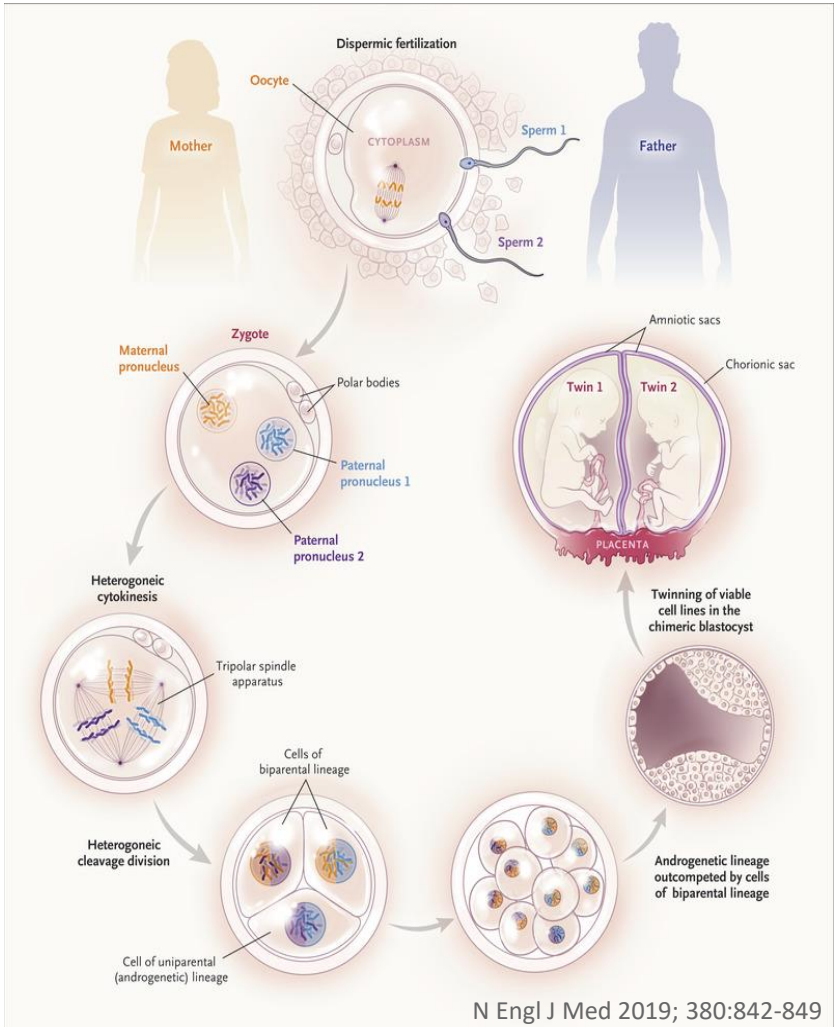
Genetics in the news



"The semi-identical twin boy and girl born in Australia were found to have 100 per cent of their mother's DNA in common, but were only 78 per cent identical in the paternal DNA they carry." cbc.ca 28 Feb 2019

[Doctors Report World's Second Case of Semi-Identical Twins | The Scientist Magazine® (the-scientist.com)]

[Molecular Support for Heterogonesis Resulting in Sesquizygotic Twinning | NEJM] (includes a short video explanation)



N Engl J Med 2019; 380:842-849
DOI: 10.1056/NEJMoa1701313

The Molecular Basis of Inheritance

From Chromosomes to Genomes

BIOL 202

Prof. Laura Nilson, Dept of Biology

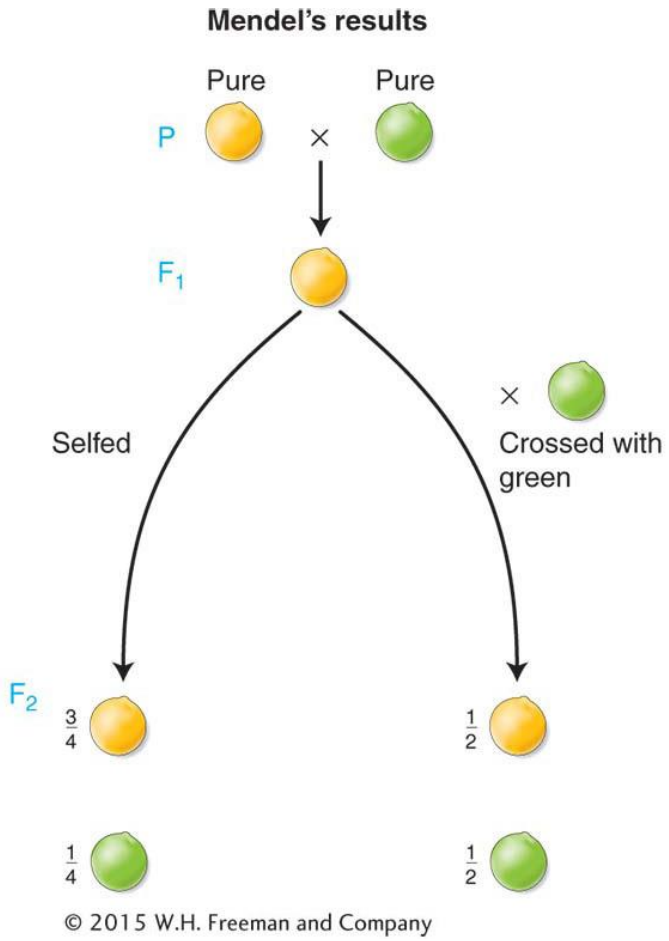
Office hours

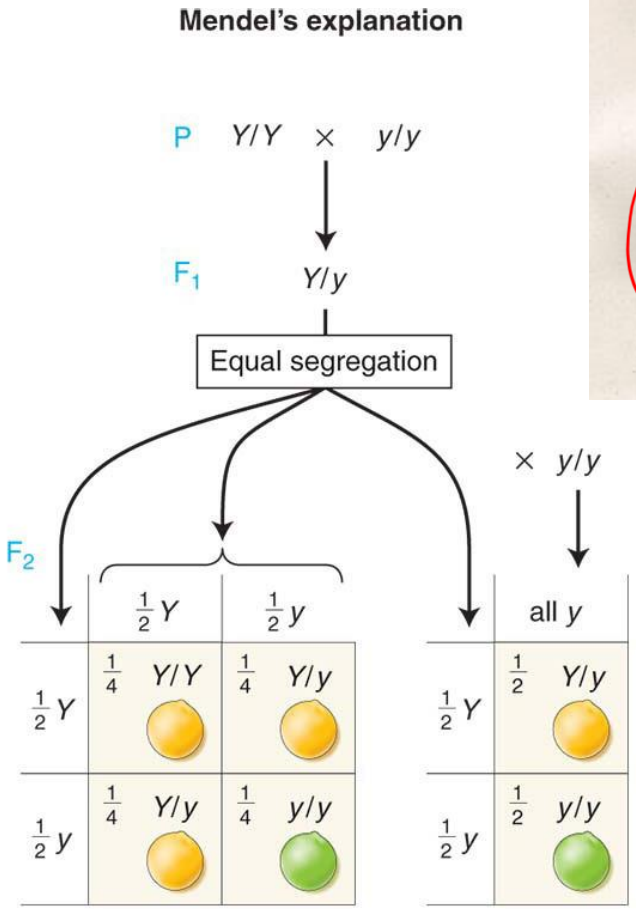
- Fridays 1-2pm (zoom or Stewart Biology, N5/8)
- By appointment
- Before or after class, right here in Leacock 132

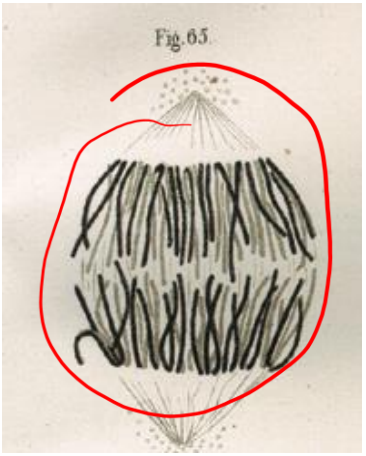
The Molecular Basis of Inheritance

What is the physical basis of the Mendelian law of heredity?

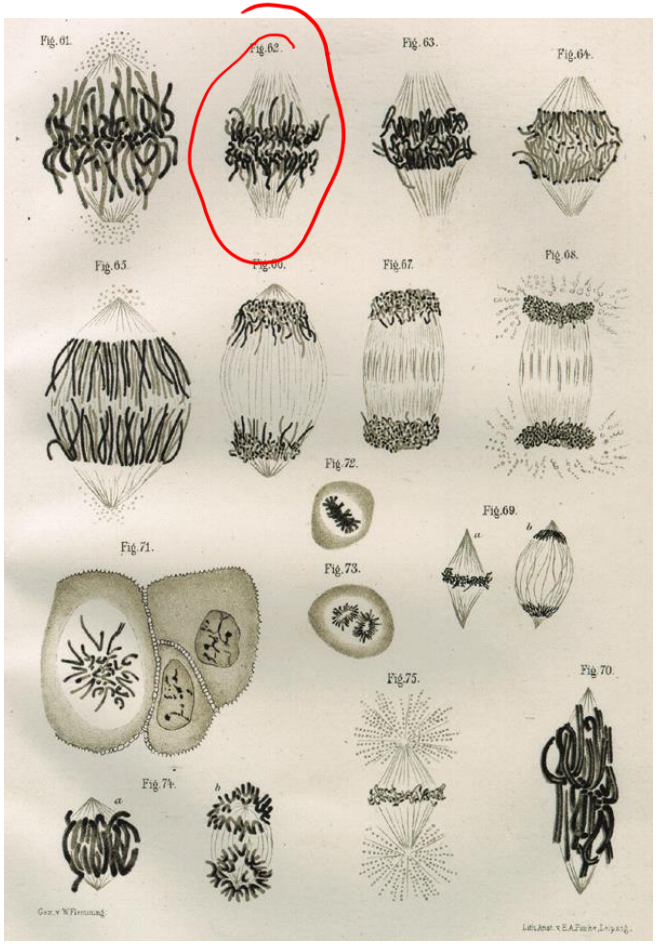
What is the physical basis of the inheritance of traits?







In the late 1800s, many microscopists observed chromosomes. In 1880, W. Roux proposed that they were the physical substrate for inheritance of traits.

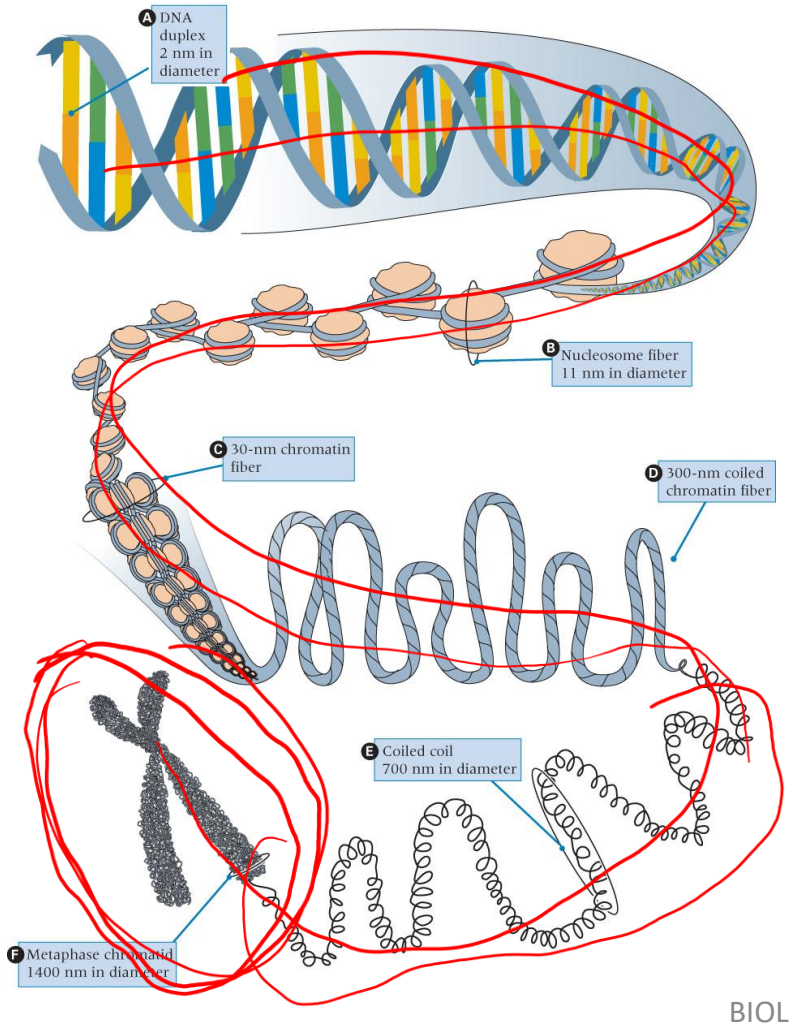


- Chromosomes occur in matched pairs.
- Chromosome pairs segregate 1:1 in meiosis.
- Chromosome pairs segregate independently.

- Segregation patterns correlate with inheritance of traits.

- A correct number of chromosomes is required for development.

Here's an overview of this section



- Large scale changes in chromosome number or structure
- Transposable elements and genome surveillance
- DNA mutations: causes and consequences
- Understanding the genetic code: genetics, bioinformatics, and functional genomics

- Your input: Other topics of interest???

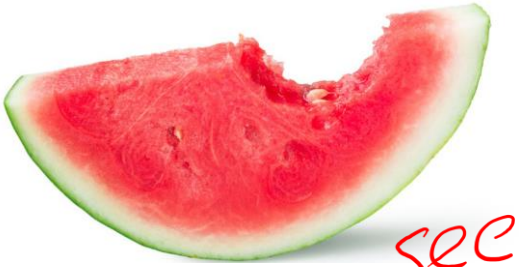
Part 1: Large Scale Chromosomal Changes

Genetics is all around us

What is different between these strawberries?

8n

What do this banana and this watermelon have in common?



seedless
→ sterile

The number of "sets of chromosomes" in an organism is referred to as "ploidy"

TABLE 17-1 Chromosome Constitutions in a Normally Diploid Organism with Three Chromosomes (Identified as A, B, and C) in the Basic Set*

Name	Designation	Constitution	Number of chromosomes
Normal Euploid			
Diploid	*2n*	AA BB CC	6
Aberrant Euploids			
Monoploid	*n*	A B C	3
Triploid	*3n*	AAA BBB CCC	9
Tetraploid	*4n*	AAAA BBBB CCCC	12
Aneuploids			
Monosomic	*2n* – 1	A BB CC	5
		AA B CC	5
		AA BB C	5
Trisomic	*2n* + 1	AAA BB CC	7
		AA BBB CC	7
		AA BB CCC	7

*In the case shown, the number of chromosomes in the basic set (the haploid chromosome number) is three.

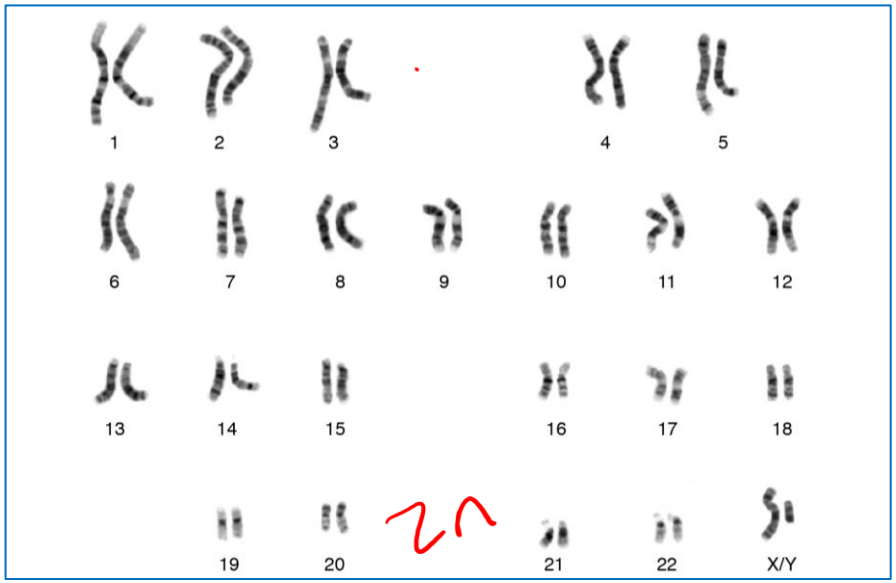
Polyploidy:

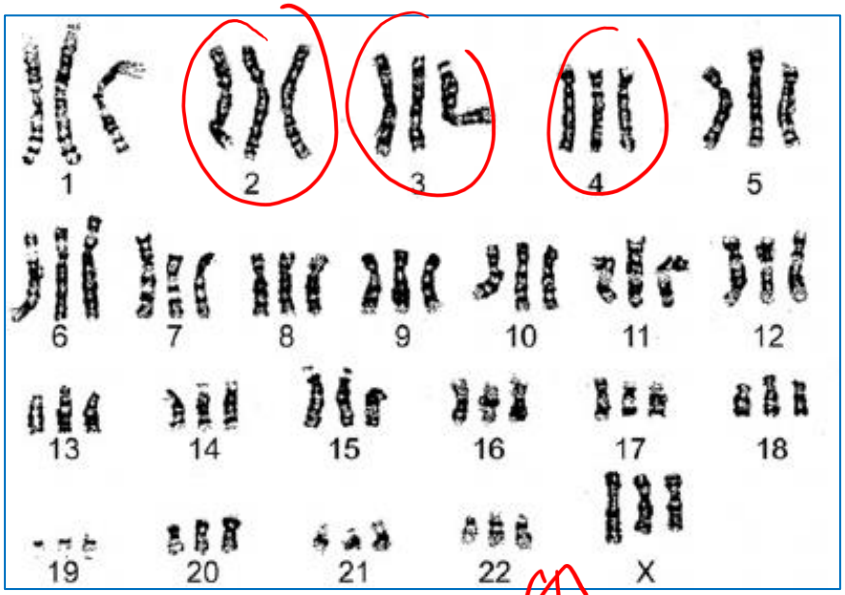
Autopolyploidy

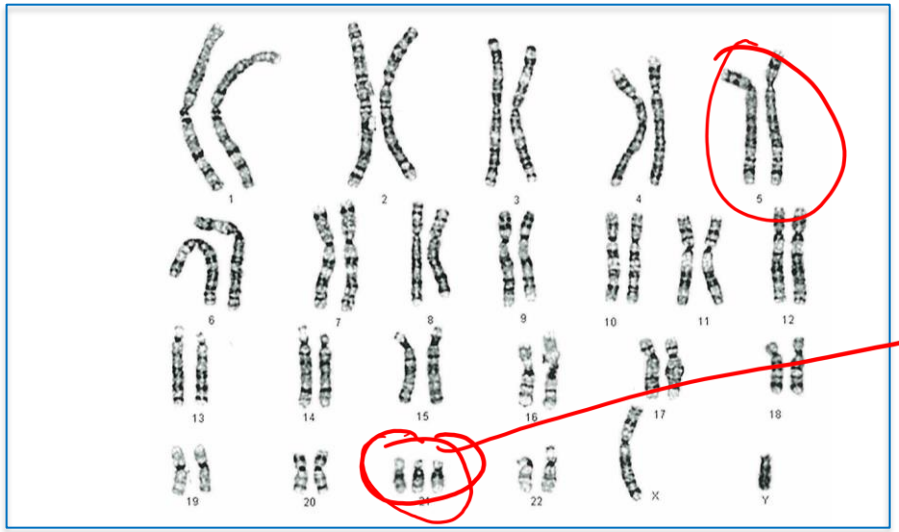
Allopolyploidy

Table 17-1
Introduction to Genetic Analysis, Twelfth Edition

What is the ploidy of these human karyotypes?





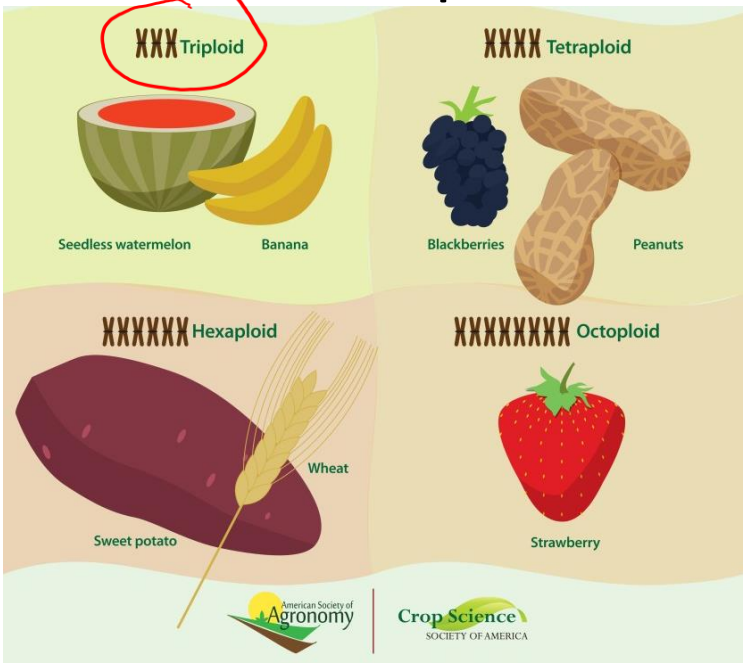


Polyploidy

uncommon in animals

8n

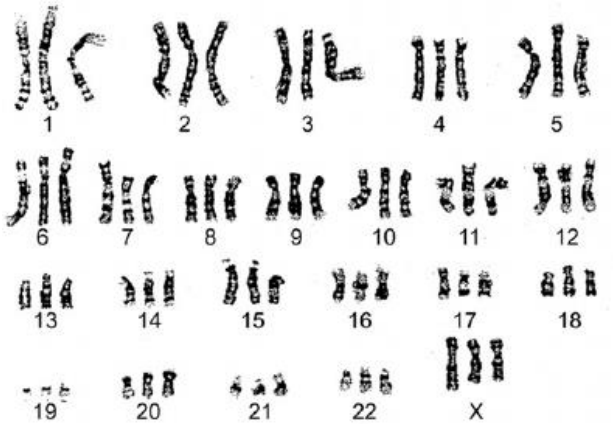
common in plants



What are the consequences of polyploidy?

How does polyploidy arise?

Consequences of autopolyploidy



Triploidy occurs in 2 to 3% of human conceptuses and accounts for approximately 20% of chromosomally abnormal first-trimester miscarriages. **Triploidy** is estimated to occur in 1 of 3,500 pregnancies at 12 weeks', 1 in 30,000 at 16 weeks', and 1 in 250,000 at 20 weeks' gestation.
www.ncbi.nlm.nih.gov › pmc › articles › PMC5511524

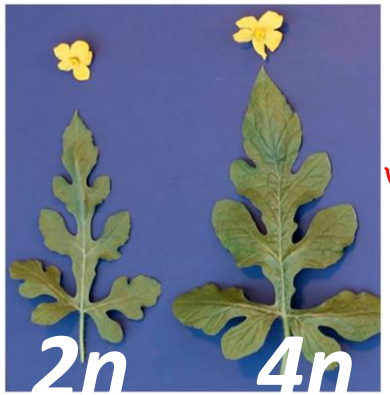
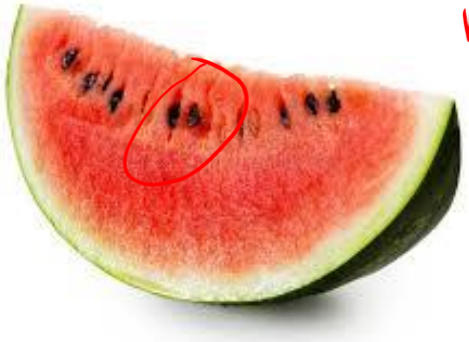


Figure 17-11
Introduction to Genetic Analysis, Eleventh Edition
Michael E. Compton, University of Wisconsin—Platteville

These are "somatic" consequences of polyploidy

Consequences of autopolyploidy



sterility — no viable germ cells

3n

2n

"Germline consequences" of polyploidy:

- gametes are not viable
- the individual is sterile

Why are triploids sterile?

Chromosome segregation during meiosis in diploids vs. triploids

Think Break

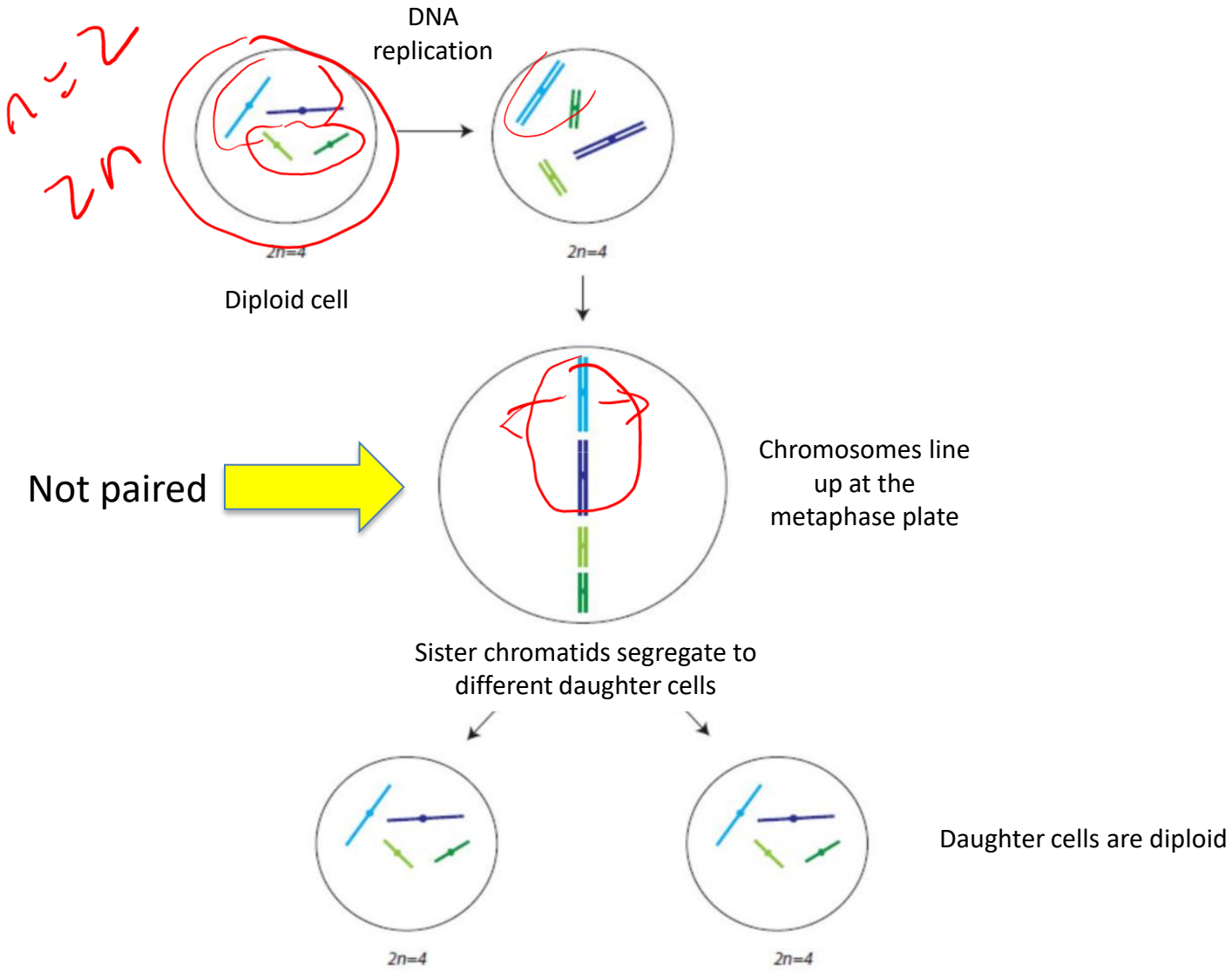
Mini-break from lecture, to:

- Highlight a tricky concept, or
- Help illustrate my reasoning, or
- Assess how well you are following along
- Break up the flow of the lecture
- Pause and refocus

Think Break

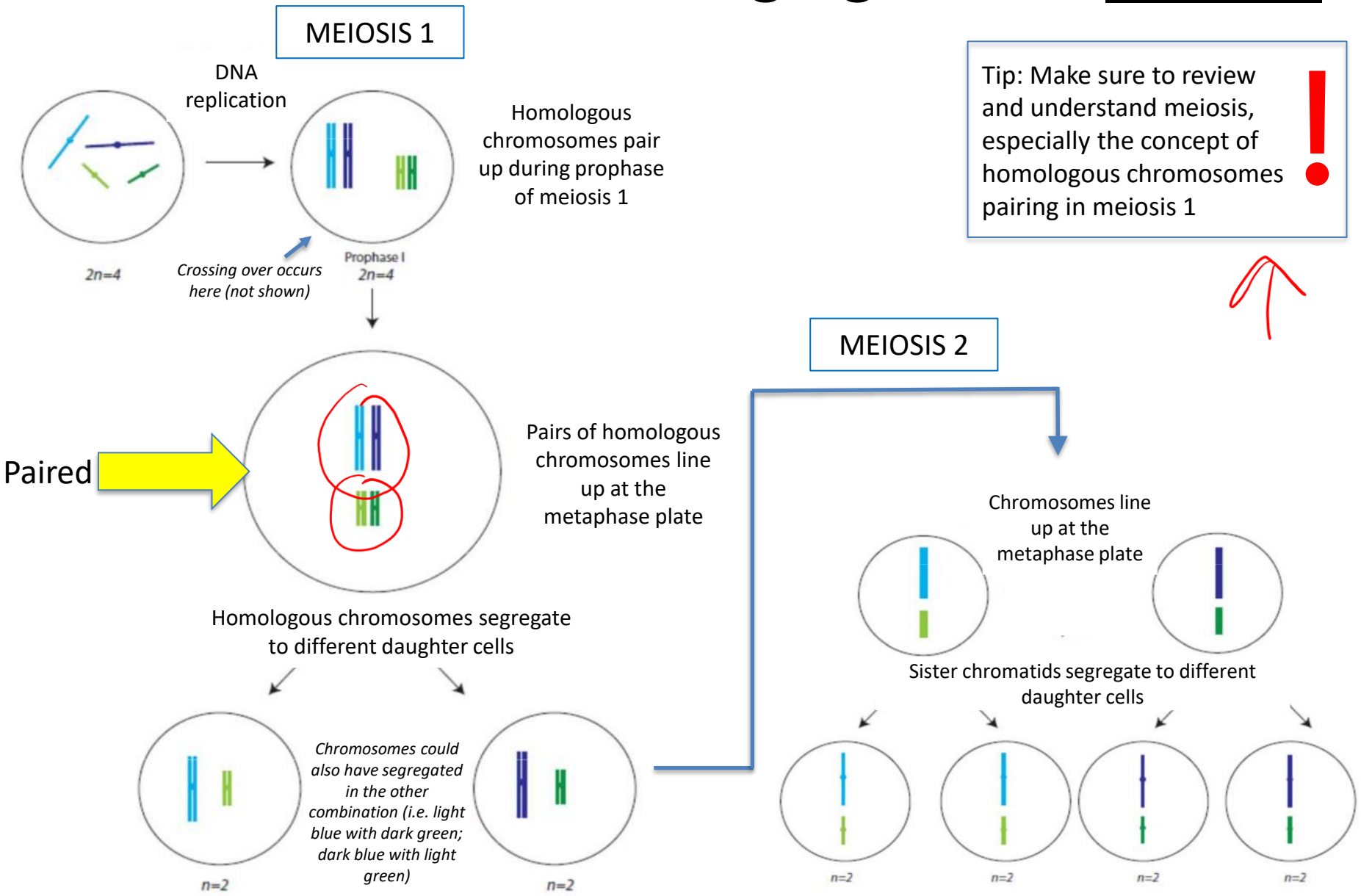
Why are we looking at meiosis?

Review: chromosome segregation in mitosis

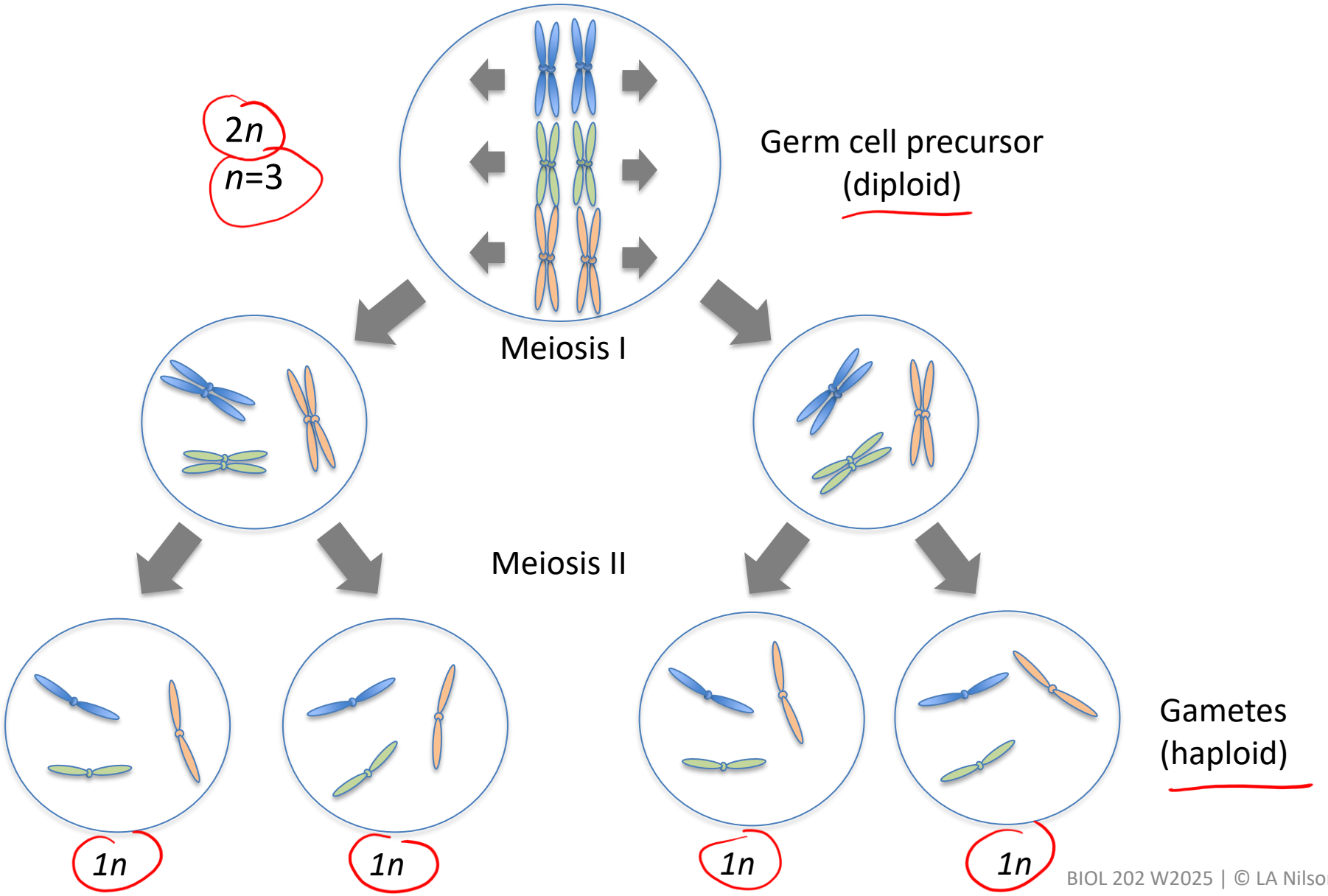


from BIOL 202 primer, available on myCourses

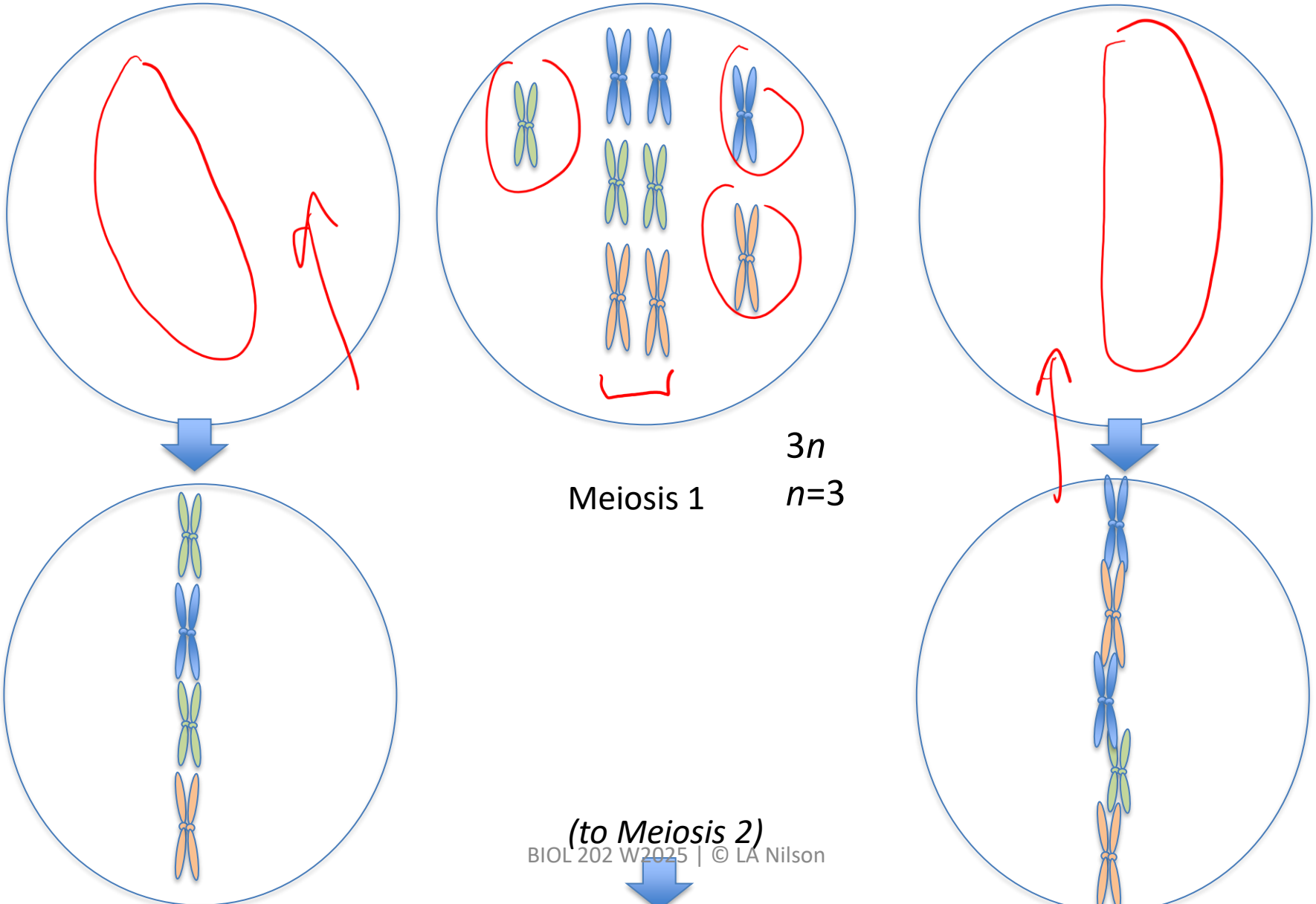
Review: chromosome segregation in meiosis



Meiosis in diploids



Meiosis 1 in triploids



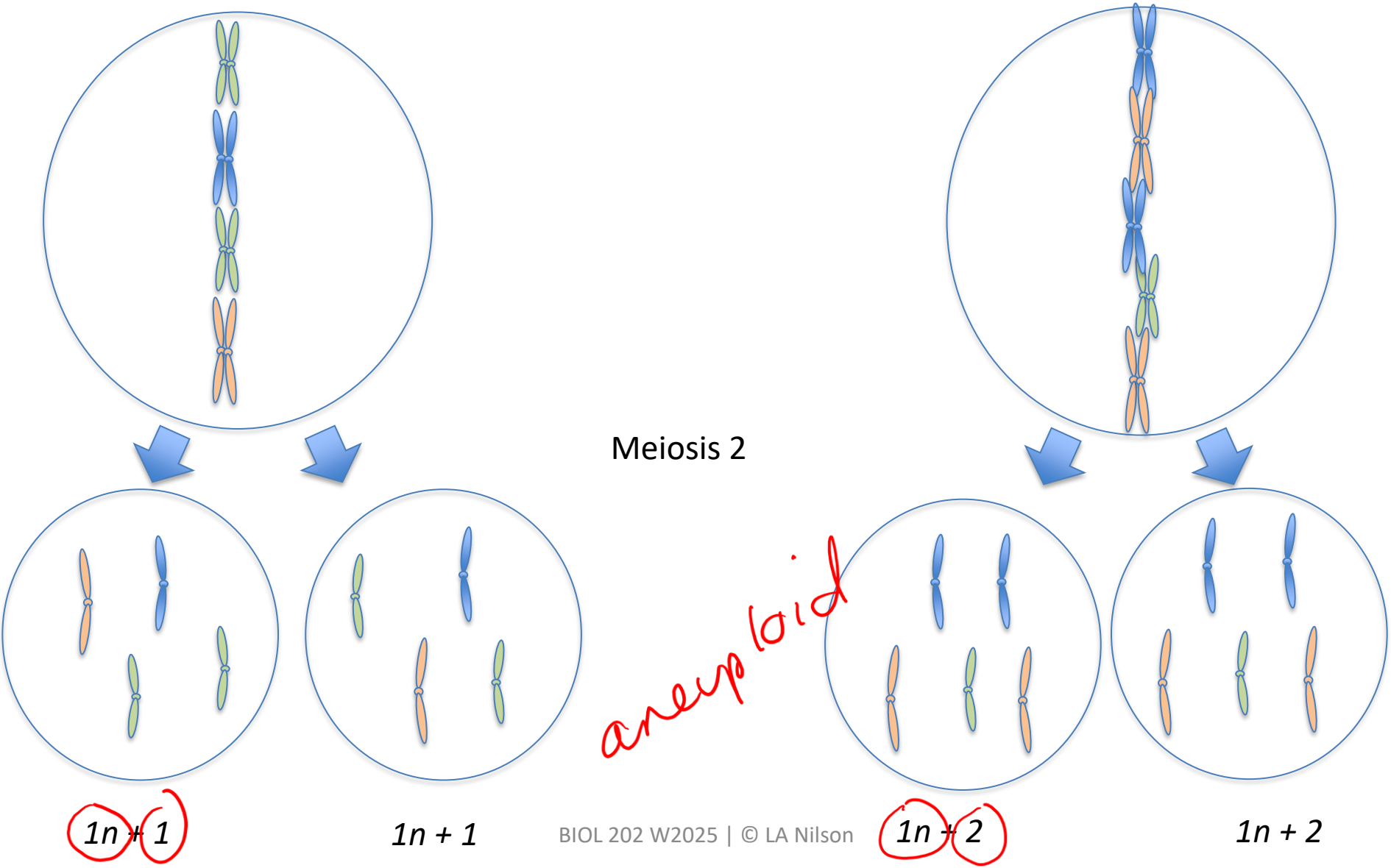
Meiosis 1

$3n$

$n=3$

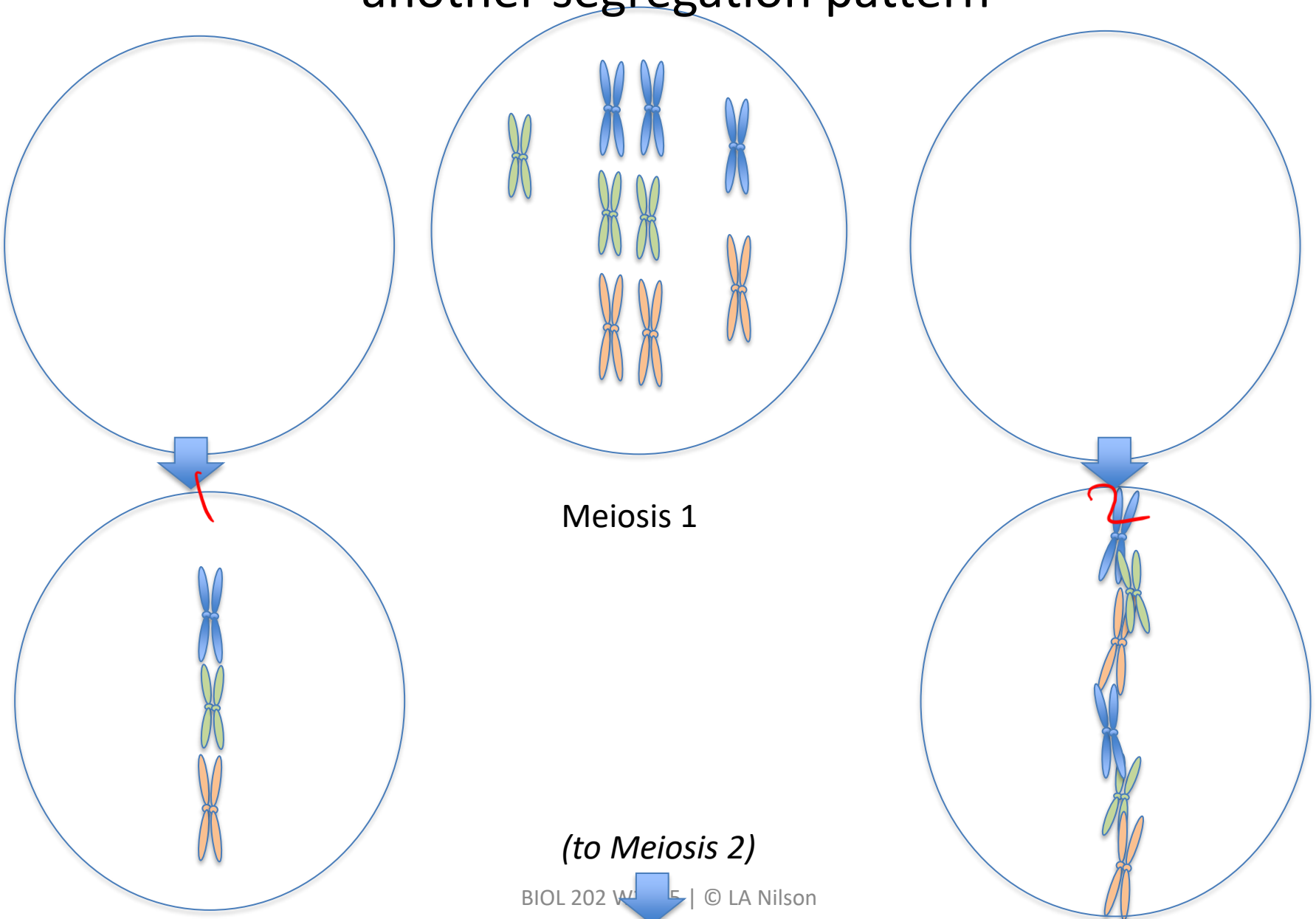
(to Meiosis 2)

Meiosis 2 in triploids



Meiosis in triploids:

another segregation pattern

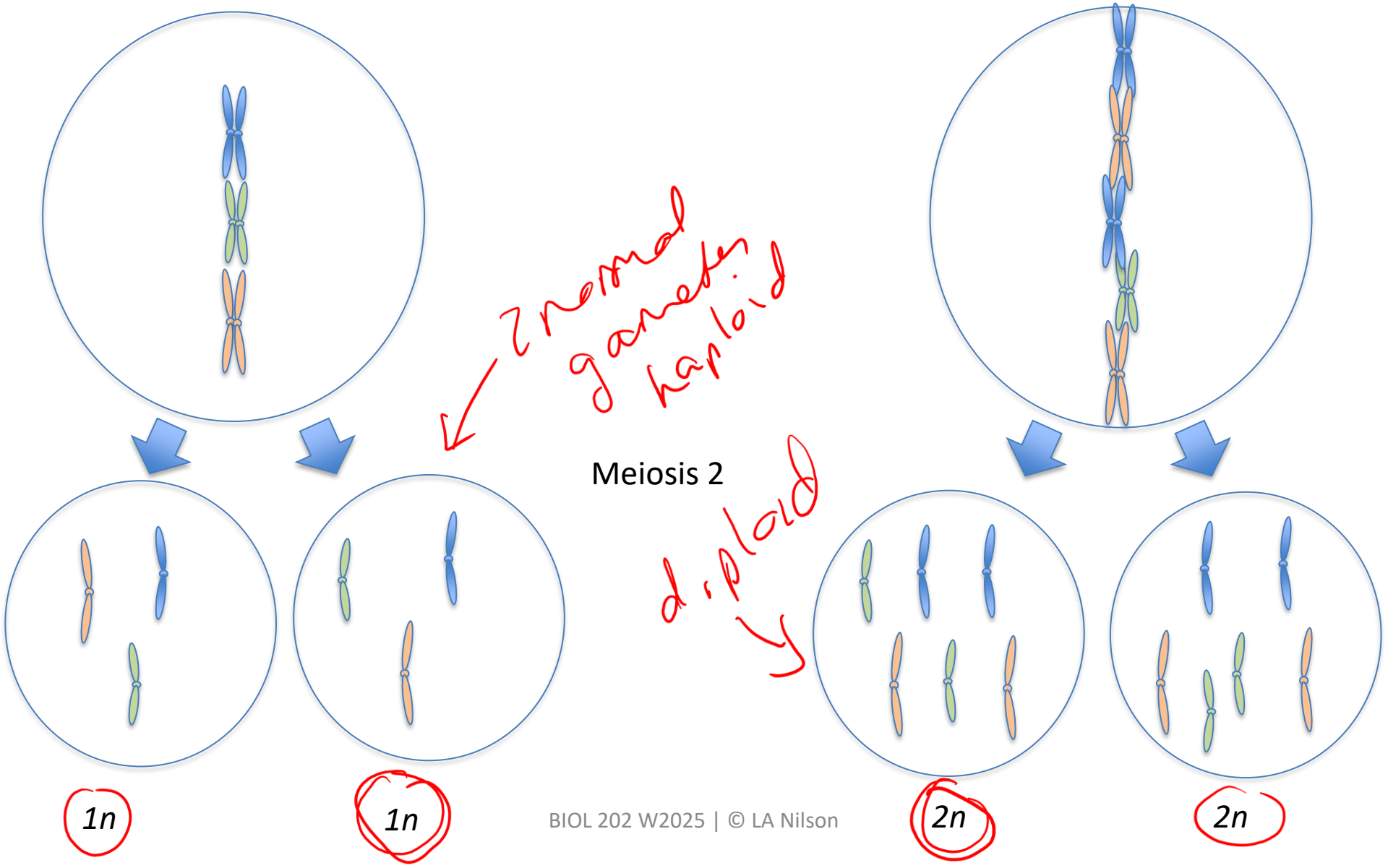


Meiosis 1

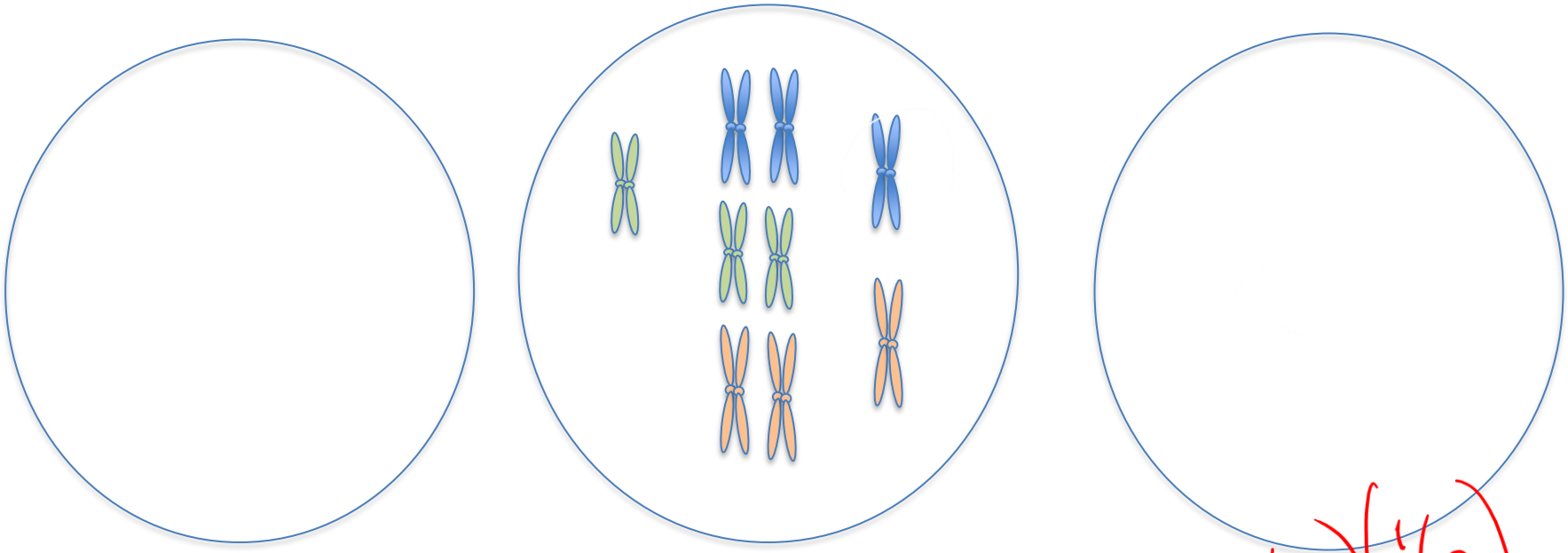
(to Meiosis 2)

Meiosis in triploids:

another segregation pattern



Normal euploid gametes can be generated in a triploid if all of the "extra" chromosomes segregate, **by chance**, to the same daughter cell.



What is the probability of a euploid gamete?

$p(\text{euploid}) = (1)(1/2)(1/2)$

$p = 1/2[n-1]$

$1/4$

Note added after class:

$p = \frac{1}{2}^{(n-1)}$

Triploids rarely produce euploid gametes

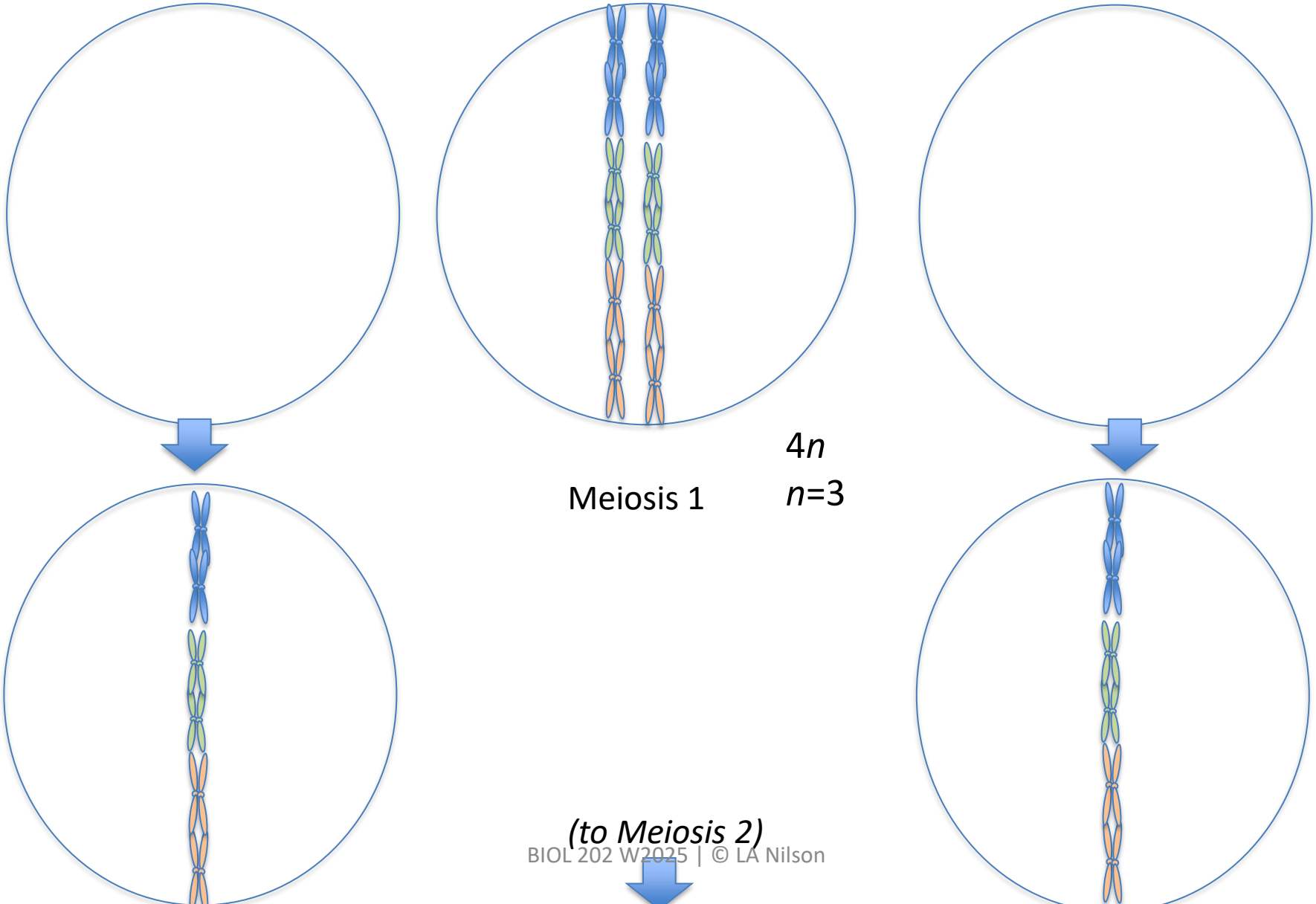
Watermelon n=11

1/1024

What about meiosis in tetraploids?

What do you think will happen?

Meiosis 1 in tetraploids



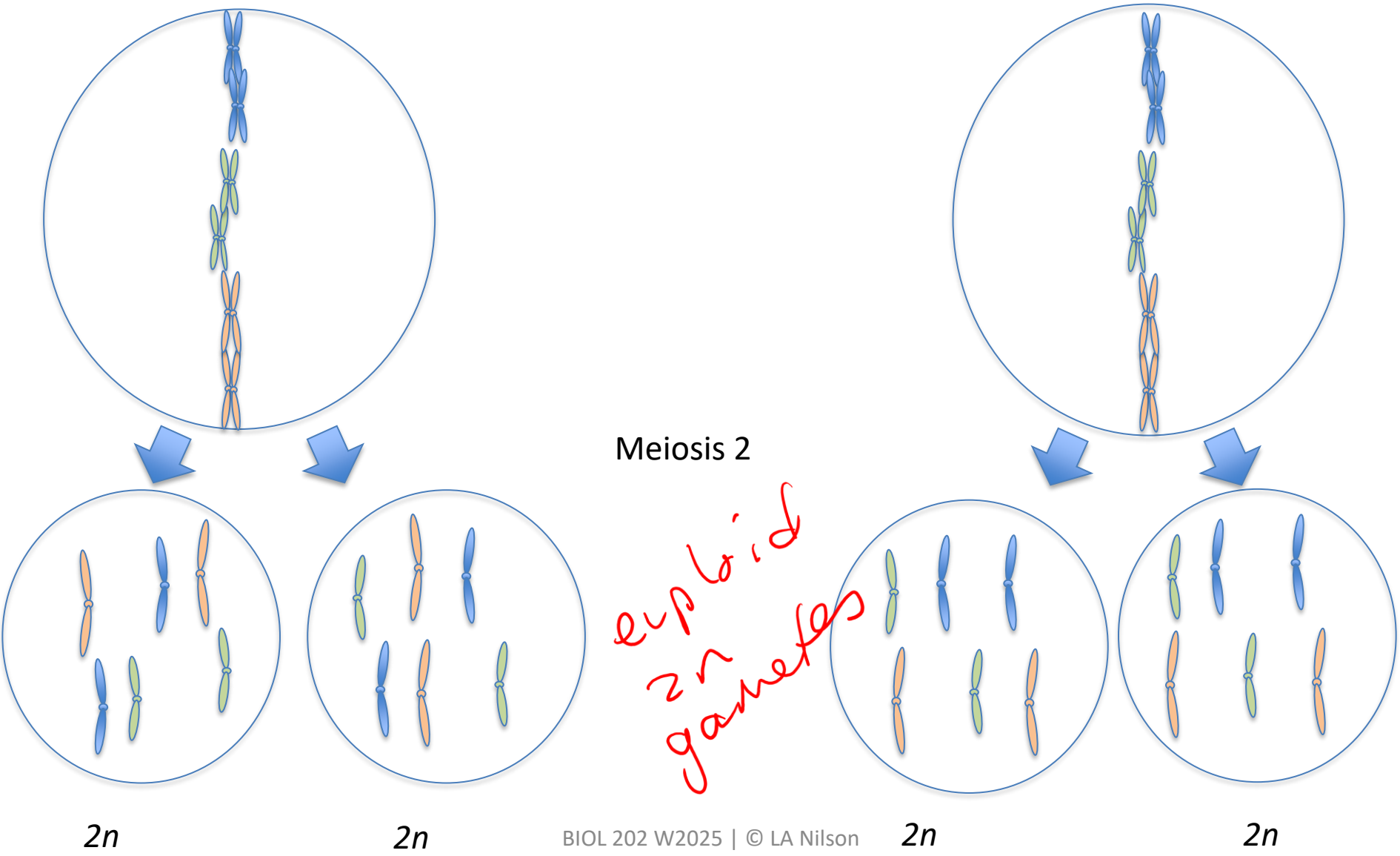
Meiosis 1

4n

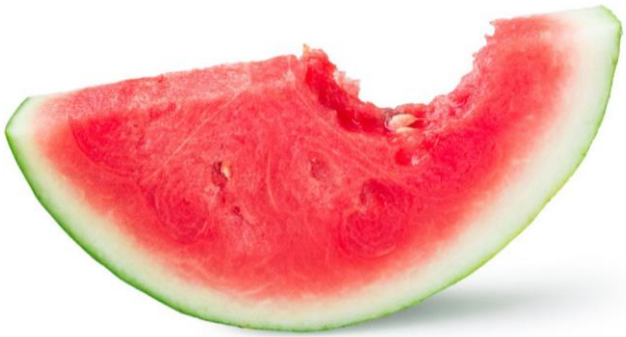
n=3

(to Meiosis 2)

Meiosis 2 in tetraploids

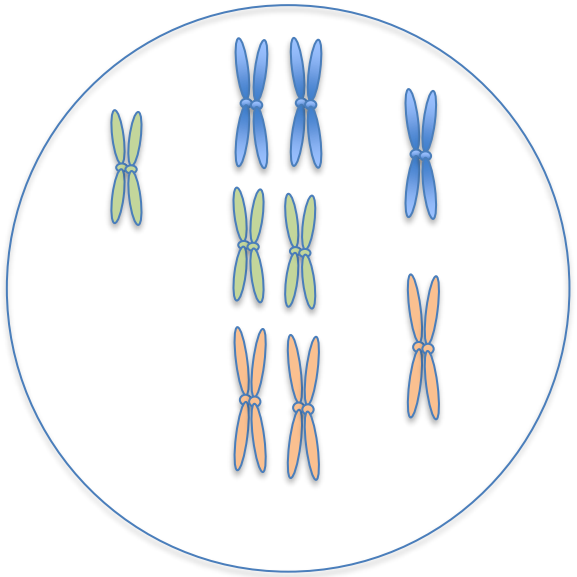


Think Break



Which of these is a tetraploid watermelon? How do you know?

What causes autopolyploidy?



What causes autopolyploidy?

- Induced/intentional
  - chemical disruption of chromosome segregation
  - fertilization of a diploid germ cell with a polyploid germ cell

- Spontaneous
  - errors in meiosis leading to diploid germ cells
  - fertilization by multiple sperm (e.g. dispermy)

Polyploidy can be spontaneous or induced

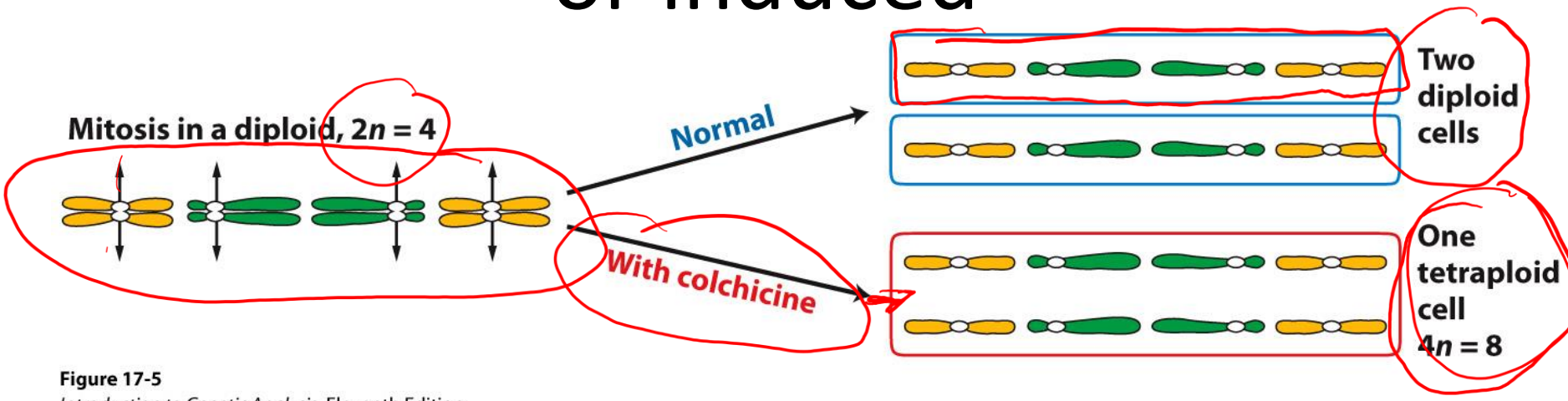
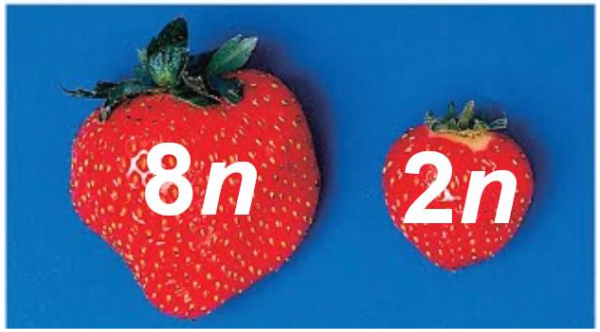
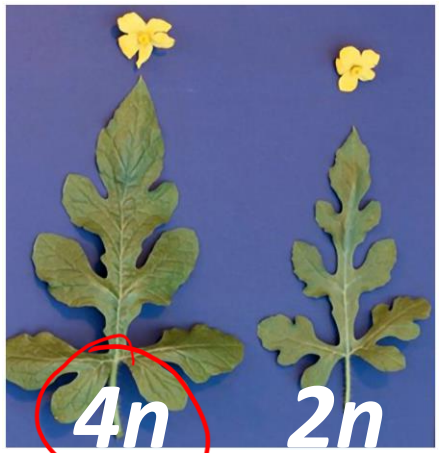


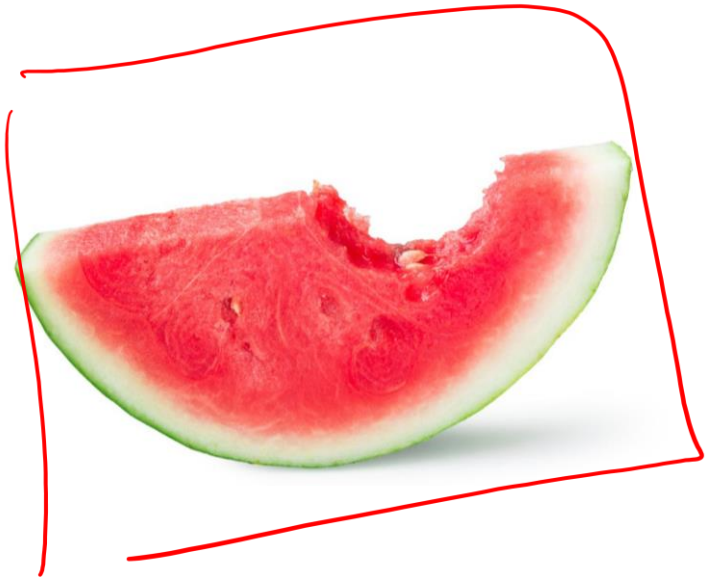
Figure 17-5
Introduction to Genetic Analysis, Eleventh Edition

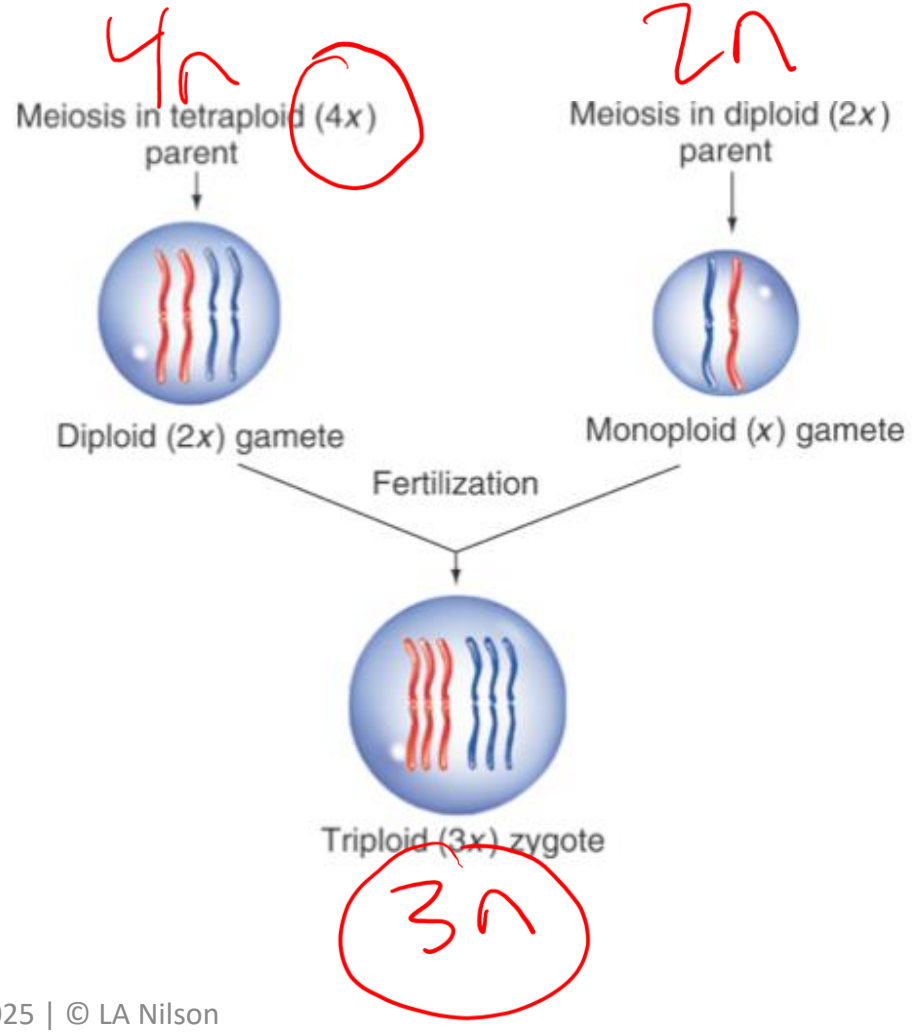




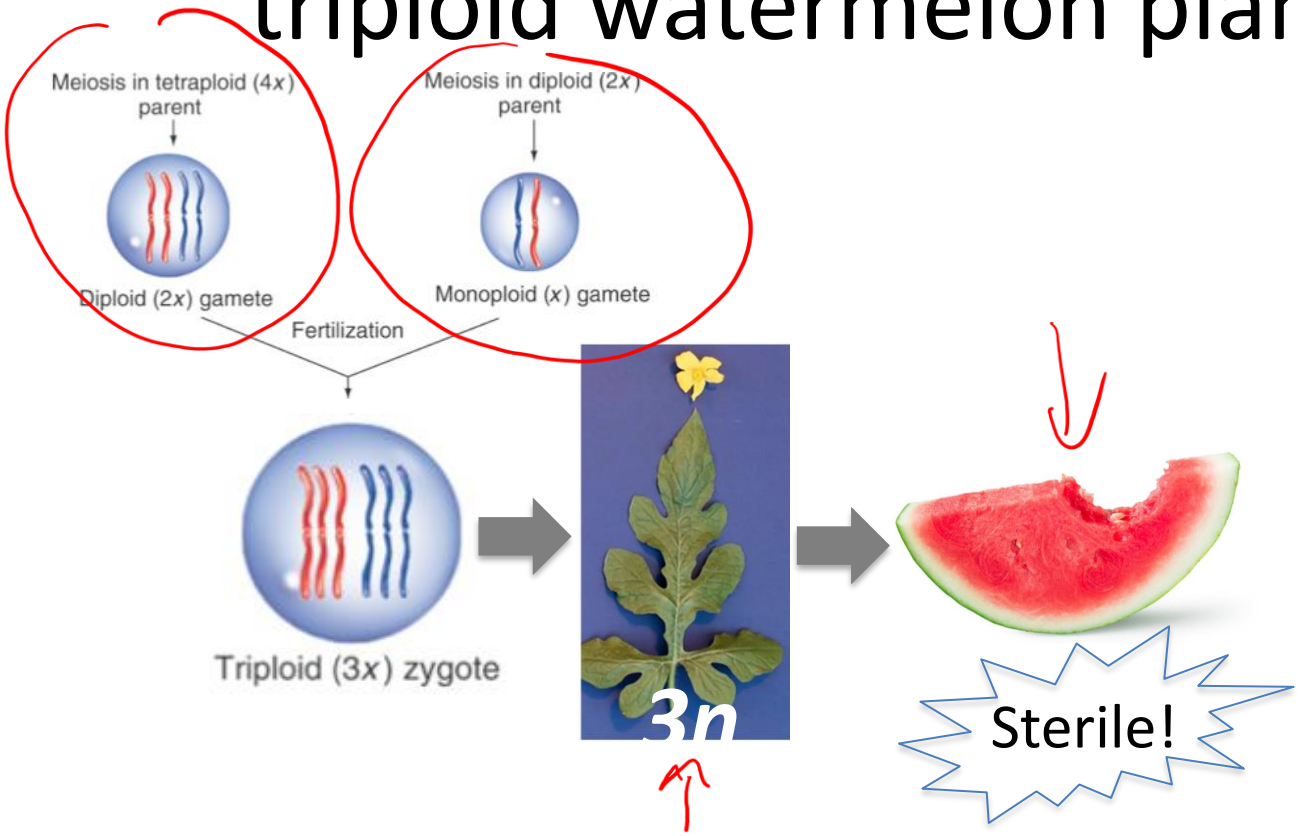
Cell grown to generate a mature plant

Polyploidy can be generated through selective breeding





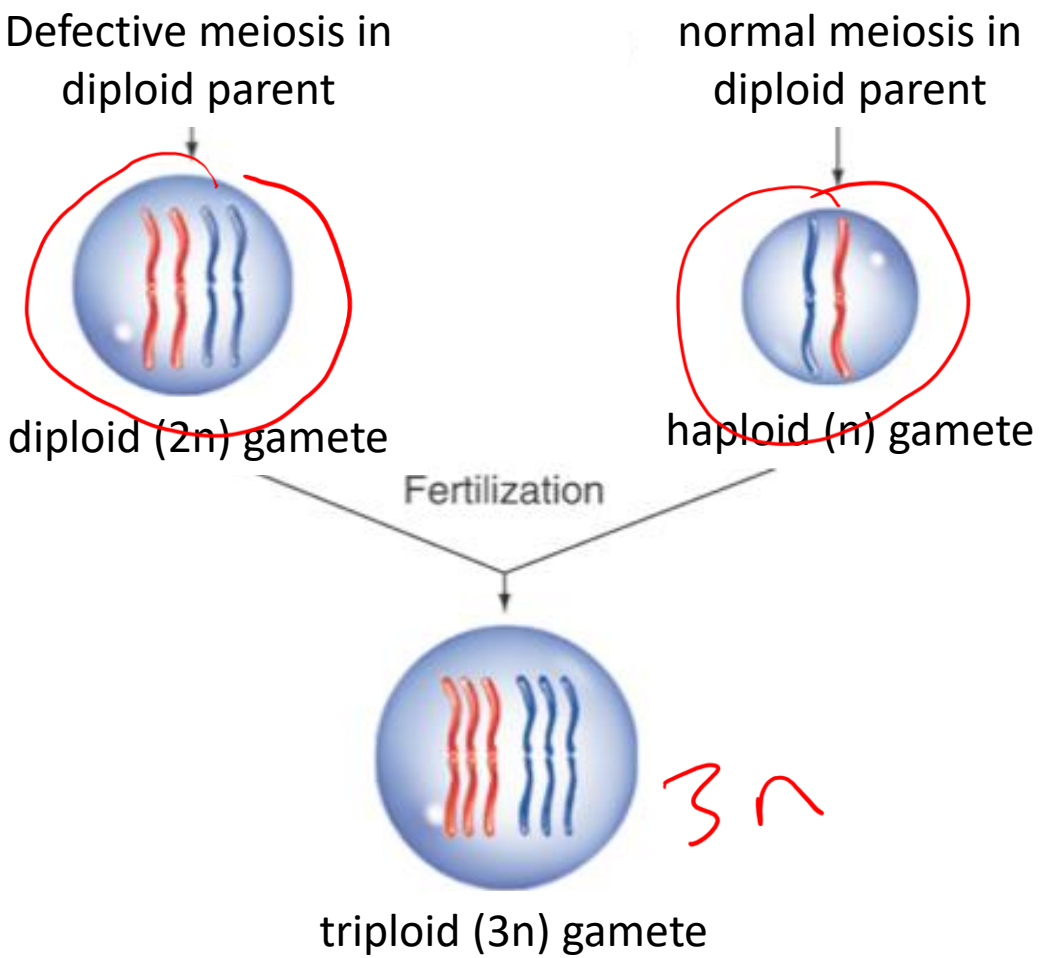
What happens once we have this triploid watermelon plant?

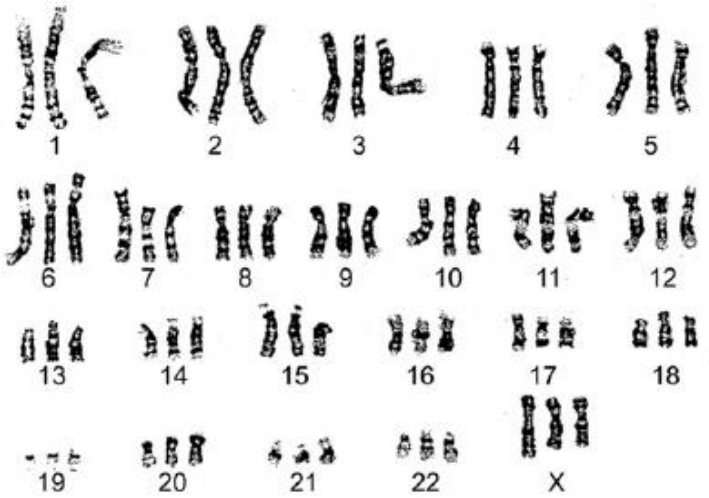


How to produce more seedless watermelons?

Where did the tetraploid plant come from?

Triploidy can be caused by errors in chromosome segregation during meiosis

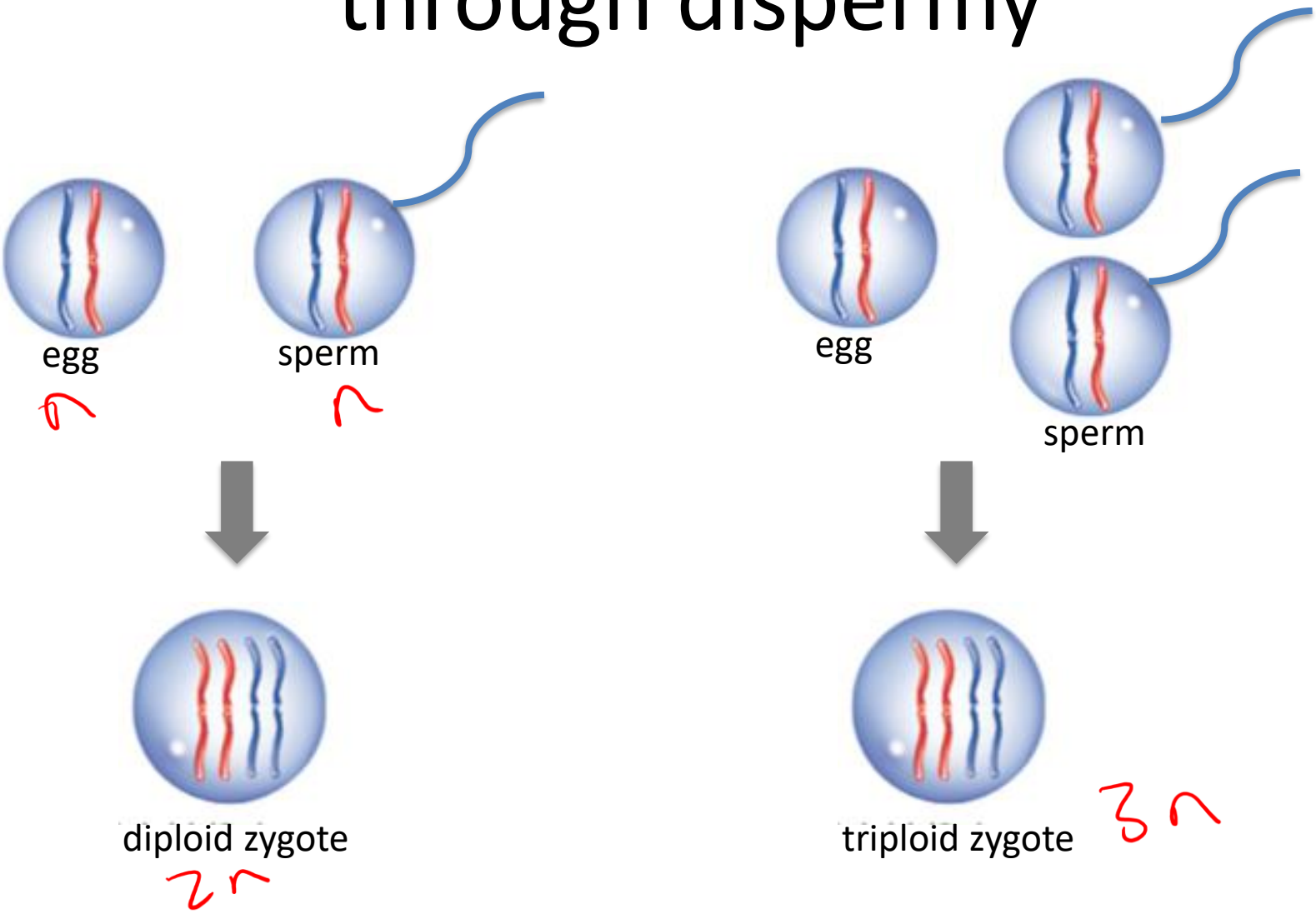




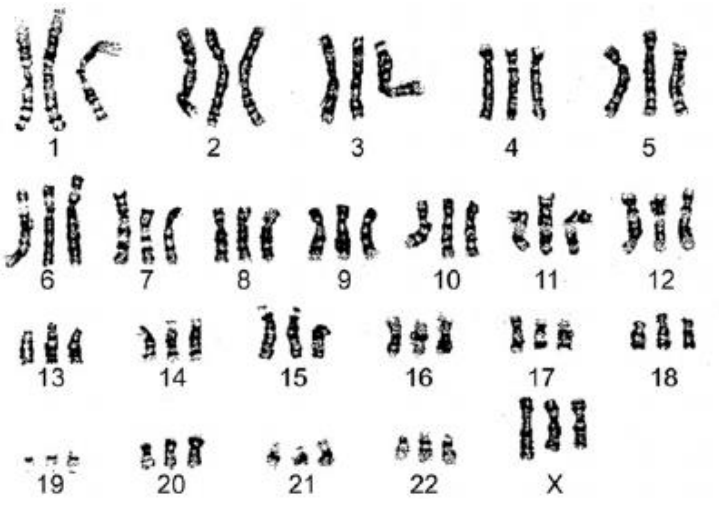
Example: triploidy in humans

Triploidy occurs in 2 to 3% of human conceptuses and accounts for approximately 20% of chromosomally abnormal first-trimester miscarriages. **Triploidy** is estimated to occur in 1 of 3,500 pregnancies at 12 weeks', 1 in 30,000 at 16 weeks', and 1 in 250,000 at 20 weeks' gestation.
www.ncbi.nlm.nih.gov › pmc › articles › PMC5511524

Triploidy can be generated through dispermy



Triploidy can be generated through dispermy



Example: triploidy in humans

Dispermy is the most common cause of human triploidy

Example: [new giant triploid invasive crayfish]

Hypothesis: crowded “petshop” conditions led to dispermy