Lesson 14
Allele segregation

Alleles

- **Allele** = alternate form of a gene
  - Variation of a gene relative to some reference gene
- Reference gene = wild-type gene
  - Alleles = variations relative to the wild-type gene
- Alleles are due to DNA sequence variations (differences)
  - Alleles are gene variants that govern related traits
  - Alleles ensures traits variations in all species

Alleles

Alleles are gene variants that govern related traits

Alleles and chromosomes

- For example: let's be APPLE the gene that encodes an apple color
  - $APPLE^{B}$ → gives you a green apple
  - $APPLE^{b}$ → gives you a yellow apple

- How do alleles relate to chromosomes (chrs)?

Alleles and chromosomes

- For example: let's be APPLE the gene that encodes an apple color
  - $APPLE^B$ → gives you a green apple
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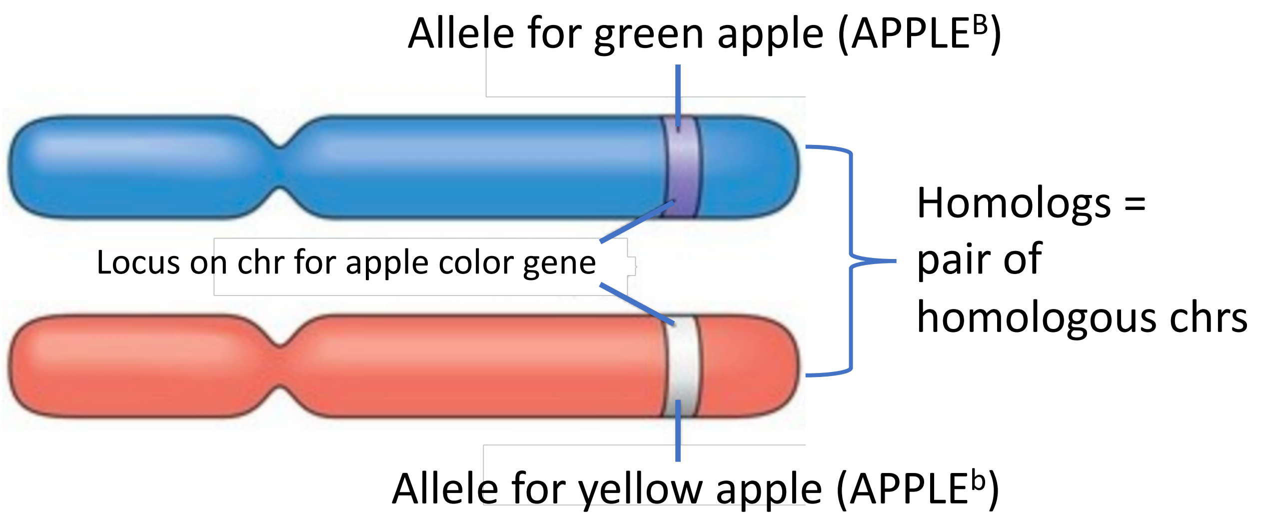
- How do alleles relate to chromosomes (chrs)
- In a diploid cell:
  - each of these alleles would be on one of the matching chrs
  - In diploid cells (2n) each chr pair has <u>**the same or different alleles of particular genes**</u>



Alleles, chromosomes and proteins

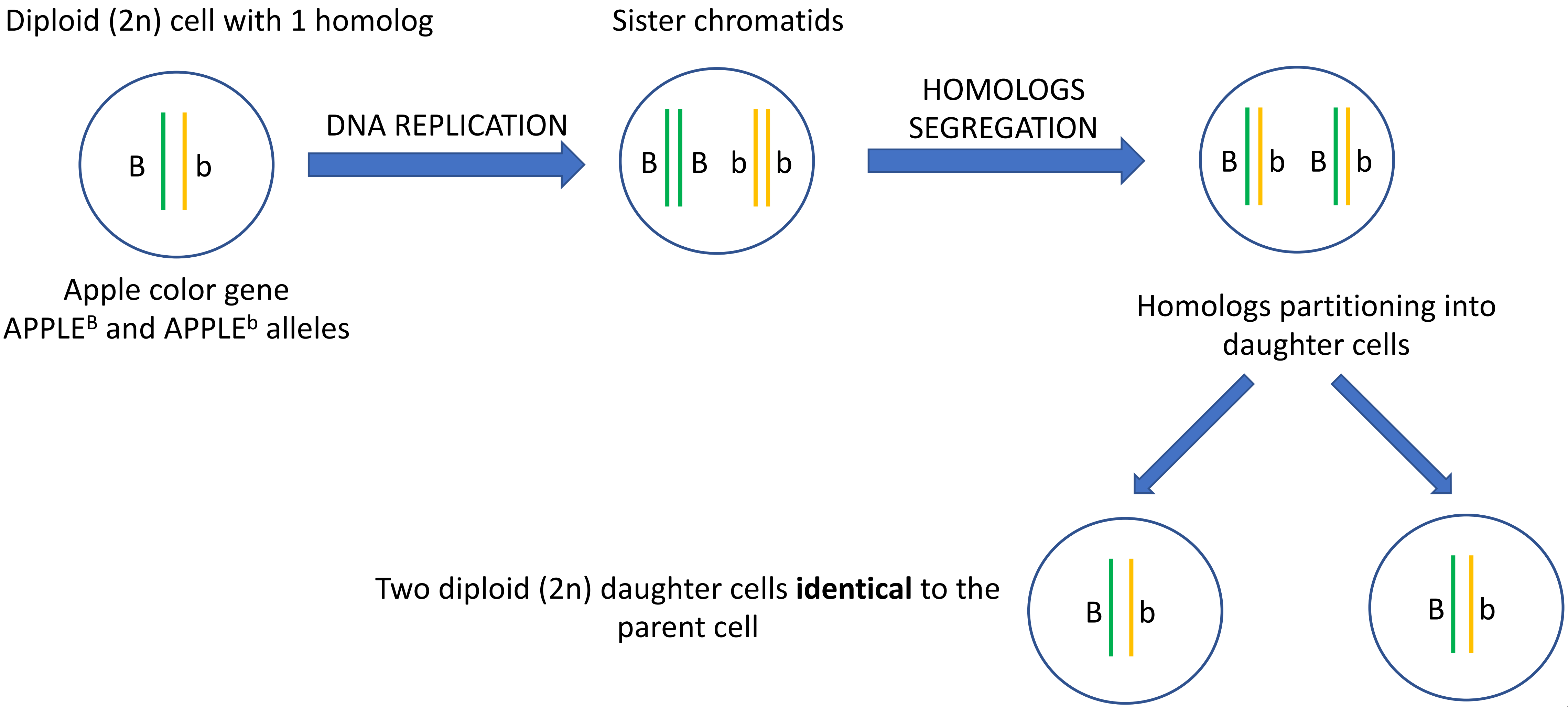
	5’ ATGTGGCTCCTGGATTAA 3’	Gene $APPLE^B$
Template strand →	3’ TACACCGAGGACCTAATT5’	
mRNA →	5’ AUGUGGCUCCUGGAUUAA 3’	
protein →	N-Met-Trp-Leu-Leu-Asp-C (stop)	
	5’ ATGTGGCTCCTGGTTTAA 3’	
Template strand →	3’ TACACCGAGGACCAAATT 5’	Gene $APPLE^b$
mRNA →	5’ AUGUGGCUCCUGGUUUAA 3’	
protein →	N-Met-Trp-Leu-Leu-**Val-C** (stop)	

Alleles $APPLE^B$ and $APPLE^b$ are both apple color genes but encodes for slightly different proteins!

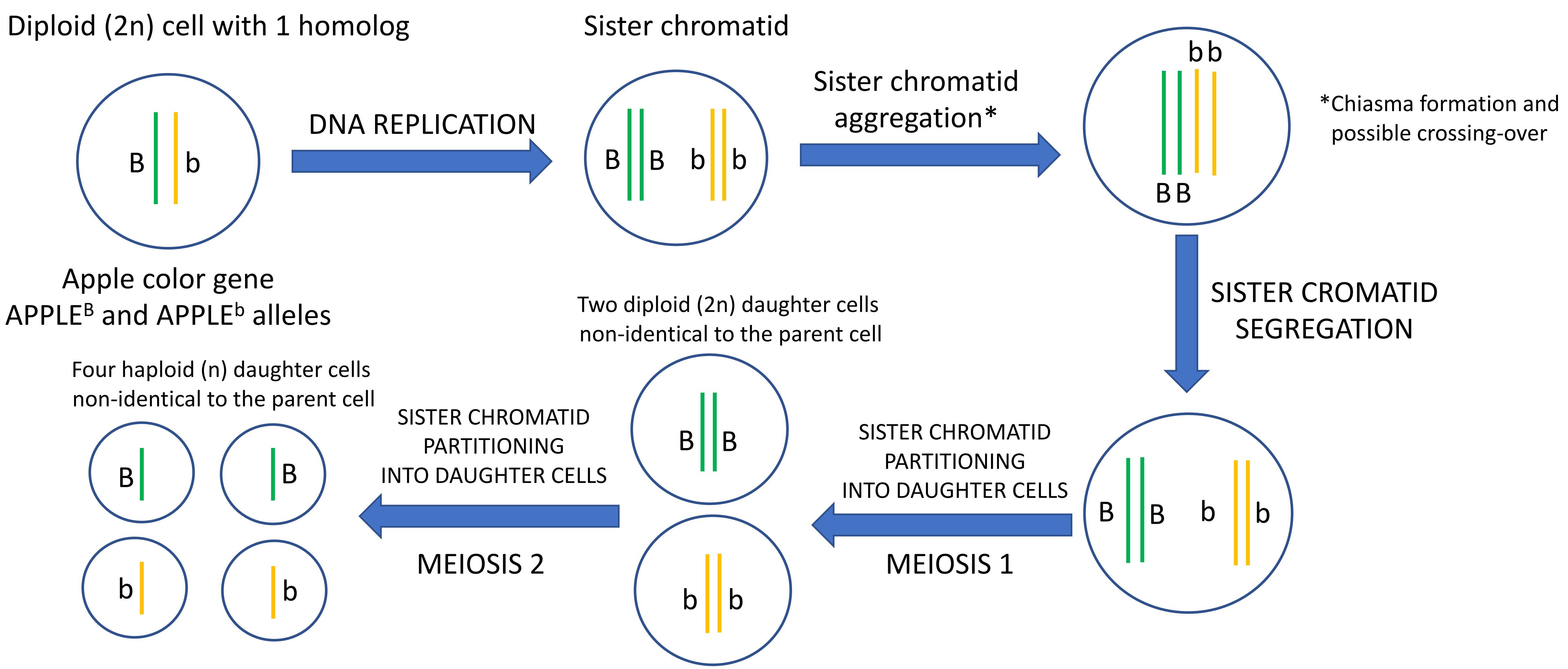
Cell division and alleles

- Somatic (body) cells (diploid) undergo replication via mitosis
- **Mitosis outcome** → daughter cells (diploid) that **have alleles identical to those of the parent cell**
- Germs (sex) cells (diploid) undergo replication via meiosis
- **Meiosis outcome** → daughter cells (gametes = eggs and sperms, haploid) that **DO NOT have alleles identical to the parent cell**

Mitosis (somatic inheritance) - revisited

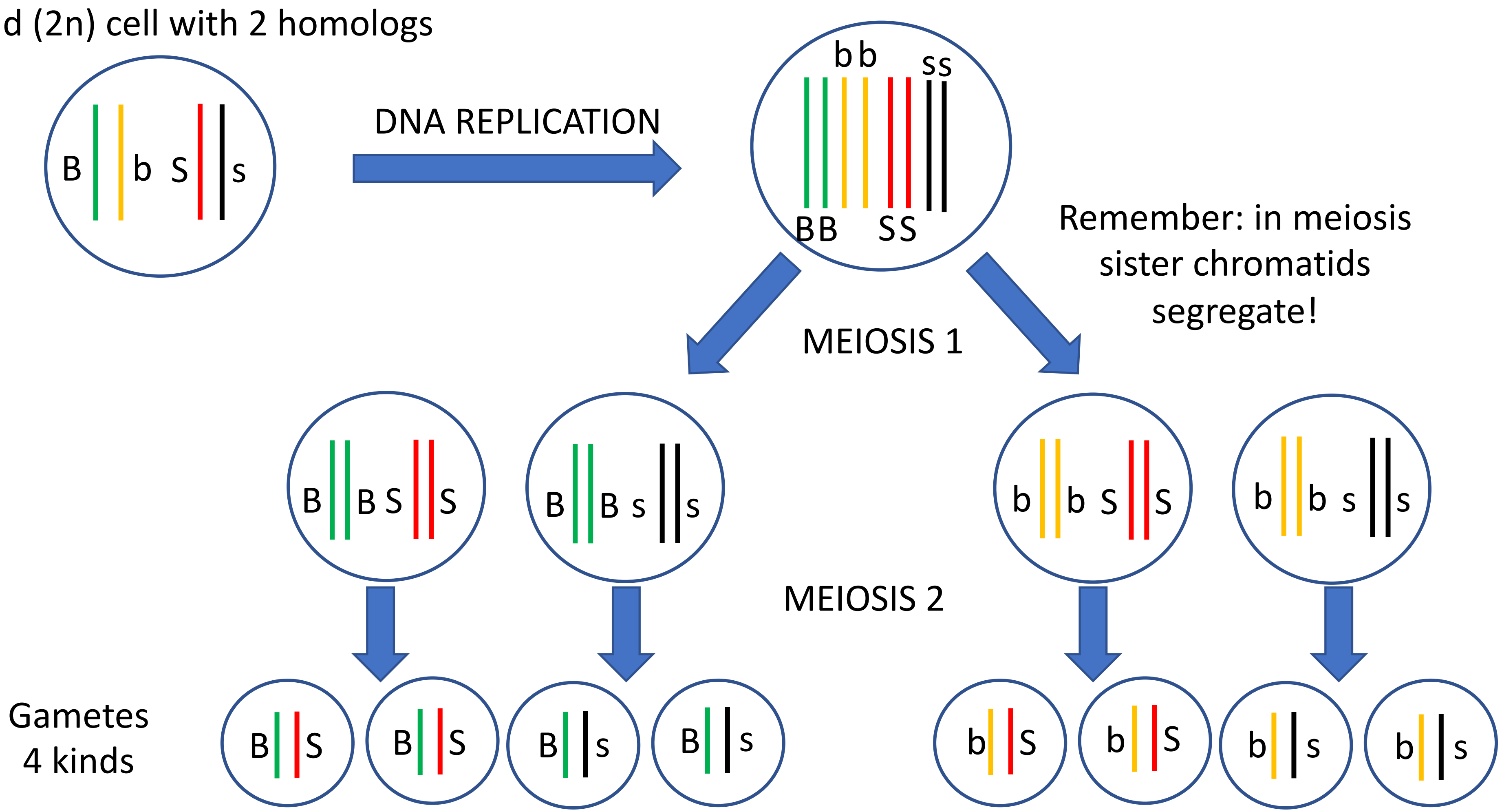


Meiosis (germline inheritance) – revisited & simplified



Meiosis (germline inheritance) – independent allele segregation (simplified)

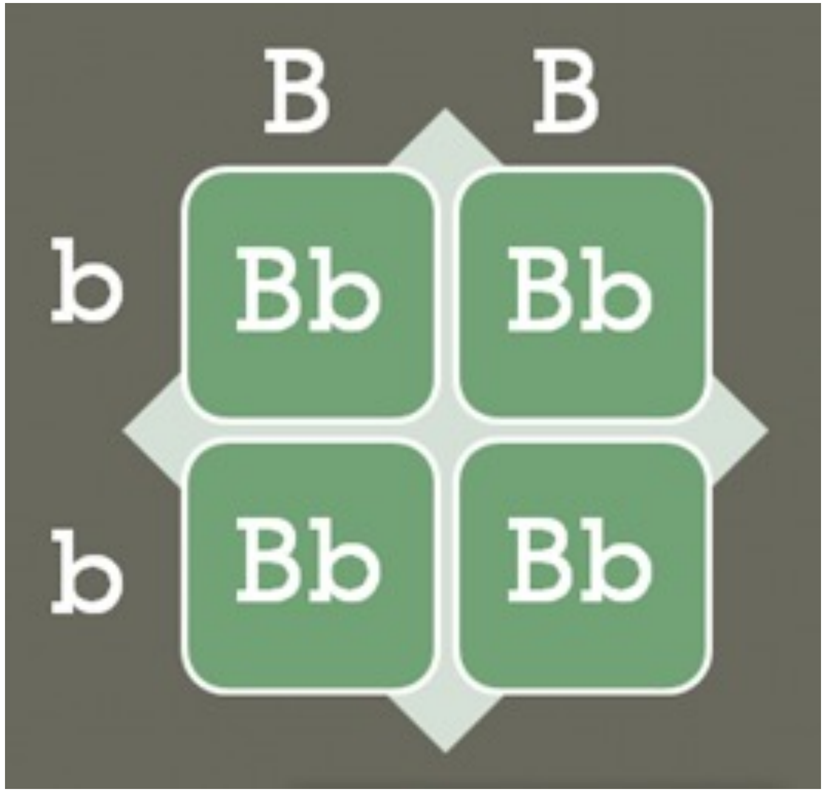
Apple color genes
APPLE^{B} and APPLE^{b} alleles
Apple taste genes
APPLE^{S} and APPLE^{s} alleles
S = sweet, s = sour



Independent allele segregation

- Independent allele segregation dictates what the next generation of the organism is going to look like
  - For the given example, we can have:
    - Green apples with sweet taste ($APPLE^B$,$APPLE^S$)
    - Green apples with sour taste ($APPLE^B$,$APPLE^s$)
    - Yellow apples with sweet taste ($APPLE^b$, $APPLE^S$)
    - Yellow apples with sour taste ($APPLE^b$, $APPLE^s$)

Lesson 15
Punnett squares



Genetics & crosses

- We need to know:
  - What are the DNA sequences that confer a particular trait
  - How traits are inherited
  - How a particular gene – and hence trait – might be regulated
    - Mostly important in diseases
- **Genetics** = set of tools to understand gene functions and inheritance

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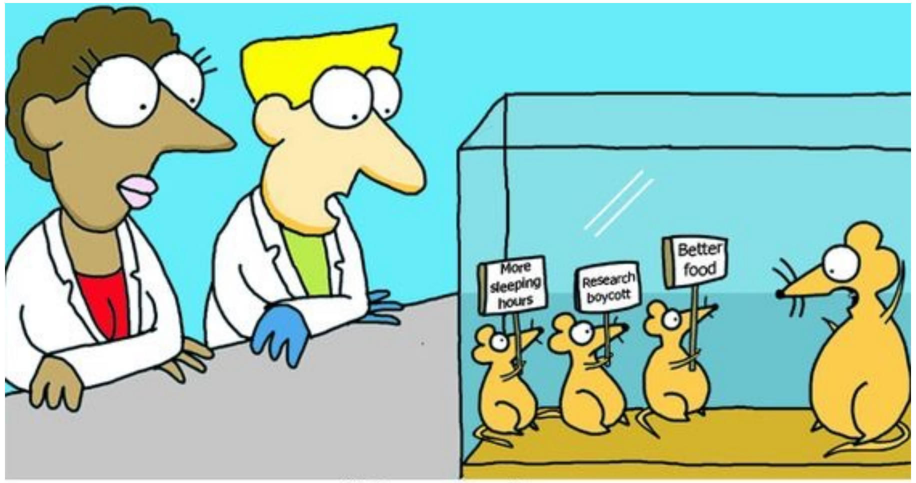

"Our mistake.
We introduced a politician's genes in that one!"

- **Genotype** = full set of genes of an individual
  - In a more narrow sense, the term can be used to refer to the alleles that are carried by an organism (*e.g.*, $APPLE^B$,$APPLE^B$ can also be also called a genotype)
- **Phenotype** = observable characteristics = traits
  - *e.g.*, the color green is associated with the $APPLE^B$,$APPLE^B$ genotype
  - Note that phenotypes are equally, or even sometimes more greatly influenced by environmental effects than genetic effects

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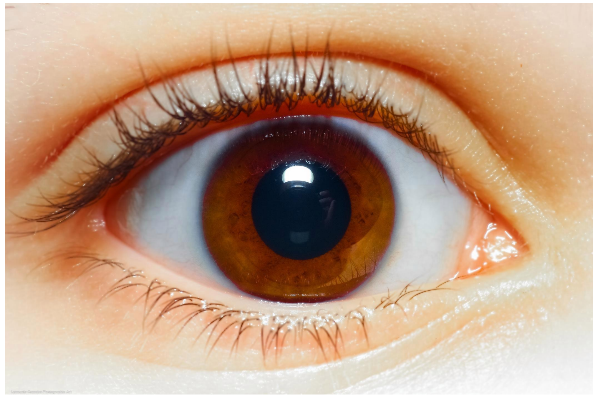
- **Alleles** = alternate gene forms
- **Generation (gen)** = individuals born at the same time from the same parents
  - P gen = parents
  - F1 gen = first generation offspring
  - F2 gen = second generation offspring…..

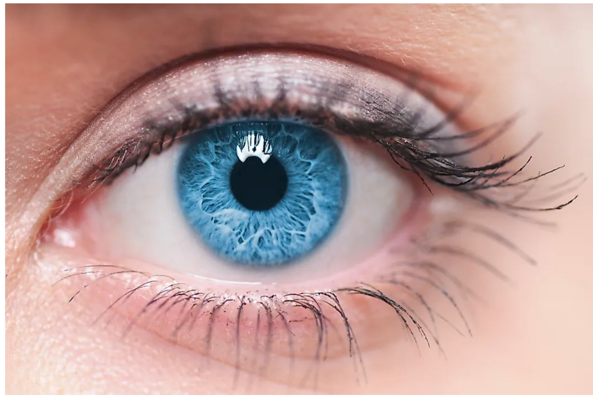
Genetic trait dominance

Traits (observable characteristics) can be:



dominant

co-dominant



recessive

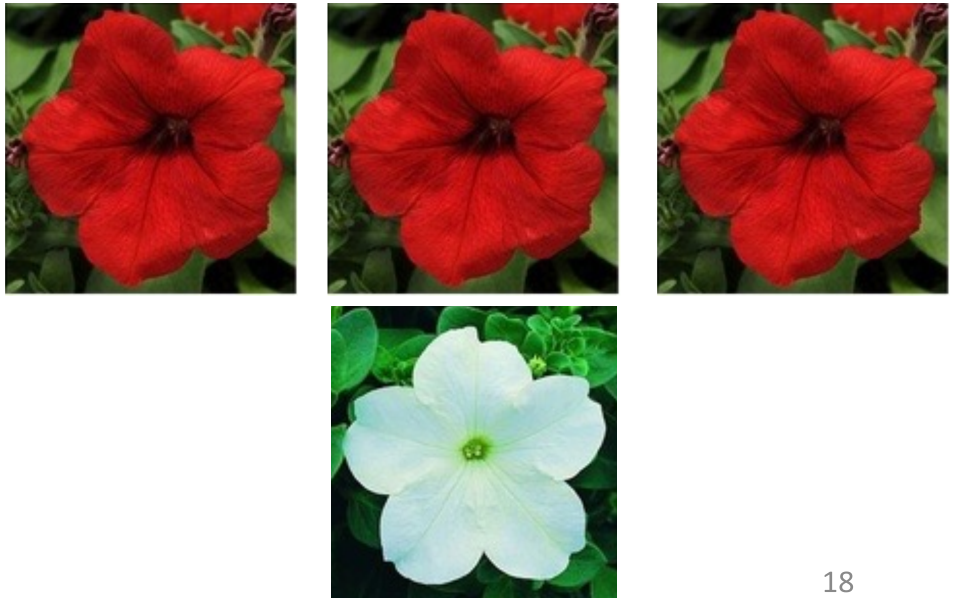
incompletely dominant

Dominant and recessive traits

- In combination traits can be dominant or recessive
- AA = red

- aa = white

- A is **dominant** to a (a is **recessive** to A)
  - AA and Aa will always give you a red flower
  - Only aa will give you a white flower



Codominance and incomplete dominance

- Aa is **codominant**

both alleles are fully expressed
(in different parts of the organism)

- Aa is **incompletely dominant**

the two alleles mix together to create
an entirely different phenotype

Gene crosses – monohybrid crosses

- Genetic crosses that follow a genotype
- **Monohybrid crosses** → (likely) 1 trait and 1 gene involved

  - Parents
  - Genotype
  - Gametes (egg/sperm)
  - First gen offspring

} Theoretical breeding experiment

Gene crosses – monohybrid crosses

- Let us reconsider our apple example
- Let us abbreviate the apple color gene alleles $APPLE^B$ with B (green) and $APPLE^b$ with b (yellow)
  - In genetics:
    - Dominant allele = capital letter (B)
    - Recessive allele = small letter (b)
- Parent apples: P1 and P2
- Genotype of P1: BB (diploid) Genotype of P2: bb (diploid)
- Gametes of P1: B (aploid, 2x) Gametes of P2: b (aploid, 2x)

Gene crosses – monohybrid crosses

Parents	P1		P2
Genotype	BB	2n	bb
Gametes	B	n	b

↘ X ↙

Note: total of 4 gametes:
2 gametes B
2 gametes b

First gen (F1) **Bb (2n) (100%)**

A few more terms

- If your gene has two of the same alleles (*e.g.*, BB or bb)
  - **Homozygous**
- If your gene has two different alleles (*e.g.*, Bb)
  - **Heterozygous**

Parents	P1		P2	
Genotype	BB	2n	bb	**Homozygous**
Gametes	B	n	b	
First gen (F1)		**Bb (2n) (100%)**		**Heterozygous**

Punnett squares

Punnet square representation: genotype

Parents	P1 X	P2
Genotype	BB	bb

P1 / P2	gametes B	gametes B
gametes b	Bb	Bb
gametes b	Bb	Bb

F1 offspring

Shows F1 genotype and proportions
100% Bb
BB, bb = homozygote   Bb = heterozygote

Genetic crosses follow phenotypes (traits)

Parents	P1		P2
Genotype	BB	2n	bb
Phenotype	Green		Yellow
Gametes	B	n	b

X

Note: total of 4 gametes:
2 gametes B
2 gametes b

First gen (F1) **Bb (2n) Green**

In this example green = dominant = B allele and yellow = recessive = b allele
The first gen will all be green apples (Bb)

Punnett squares

	P1 x P2
Parents	P1 x P2
Genotype	BB bb
Phenotype	Green Yellow

Punnett square representation Genotype + phenotype

P1 / P2	Gametes: B	Gametes: B
Gametes: b	Bb green	Bb green
Gametes: b	Bb green	Bb green

F1 offspring

F1 100% heterozygous Bb
F1 100% green

F1 cross

Parents	F1	X	F1
Genotype	Bb		Bb
Phenotype	Green		Green
Gametes	B and b		B and b

Note: total of 4 gametes:
2 gametes B and b
2 gametes B and b

2nd gen (F2) ?

Remember: in this example green = dominant = B allele and yellow = recessive = b allele

Punnett squares

	F1 =P1	x	F1 =P2
Parents Genotype	Bb		Bb
Phenotype	Green		Green

Punnett square representation Genotype + phenotype

3 genotypes: BB, bb, Bb
F2 will be 50% homozygous
25% BB
25% bb
and
50 % heterozygous (Bb)

2 phenotypes: green and yellow
F2 will be 75% green + 25% yellow as B is dominant
(3:1)

P1 / P2	Gametes: B	Gametes: b
Gametes: B	BB green	Bb green
Gametes: b	Bb green	bb yellow

F2 offspring

B is dominant
b is recessive

Test cross

- The parent genotype might be unknown
  - You want to figure this out
- For the apple example:
  - Is a parent genotype BB, Bb or bb?
- **Test cross**: you cross the parent with a true breeding strain (TBS)
- **True breeding strain = individual with two alleles of a recessive gene**
  - Gene b (yellow apple) is recessive, your TBS will be a yellow apple with genotype bb
  - Crossing two TBS both with genotype bb will always and only give you apples with genotype bb and phenotype yellow

Test cross

Parents	P1	X	P2
Genotype	BB? or Bb?		bb (true-breeding)

Test cross: for parental genotype

Obviously, if P1 has a (bb) genotype you will get an all bb and all yellow Punnett square

F1 offspring

P2 gametes \ P1 gametes	B	B
b	Bb	Bb
b	Bb	Bb

If P1 is BB
100% offspring are green (Bb)

F1 offspring

P2 gametes \ P1 gametes	B	b
b	Bb	bb
b	Bb	bb

If P1 is Bb
50% offspring are green (Bb)
50% are yellow (bb)