

# 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

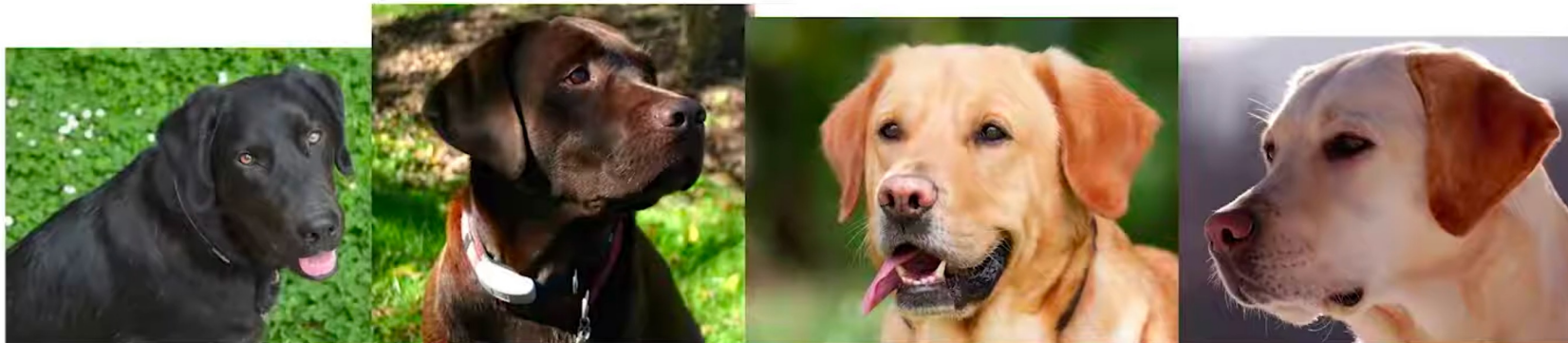
# Alleles



Eye Color Chart



Alleles are gene variants that govern related traits



# Alleles and chromosomes

- For example: let's be **APPLE** the gene that encodes an apple color

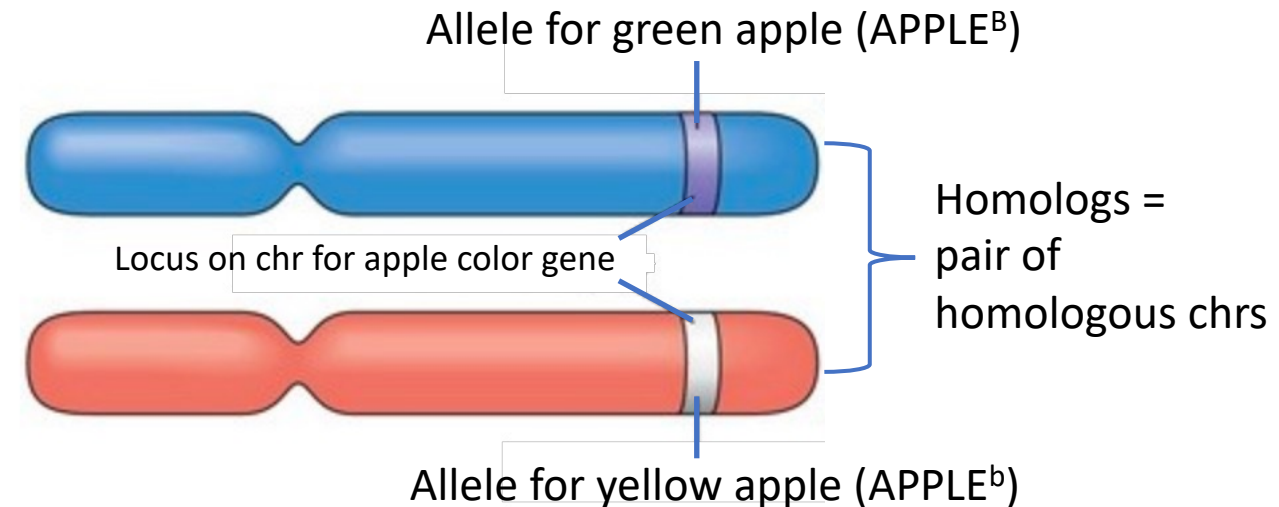
- $APPLE^B \rightarrow$  gives you a green apple
- $APPLE^b \rightarrow$  gives you a yellow apple



- How do alleles relate to chromosomes (chrs)?

# Alleles and chromosomes

- For example: let's be APPLE the gene that encodes an apple color
  - $APPLE^B \rightarrow$  gives you a green apple
  - $APPLE^b \rightarrow$  give you a yellow apple
- 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 **the same or different alleles of particular genes**



# Alleles, chromosomes and proteins

Template strand →	5' ATGTGGCTCCTGGATTAA 3' 3' TACACCGAGGACCTAATT5'	Gene APPLE <sup>B</sup>
mRNA →	5' AUGUGGCUCCUGGAUUAA 3'	
protein →	N-Met-Trp-Leu-Leu-Asp-C (stop)	
Template strand →	5' ATGTGGCTCCTGGTTTAA 3' 3' TACACCGAGGACCAATT 5'	Gene APPLE <sup>b</sup>
mRNA →	5' AUGUGGCUCCUGGUUUAA 3'	
protein →	N-Met-Trp-Leu-Leu-Val-C (stop)	

Alleles APPLE<sup>B</sup> and APPLE<sup>b</sup> are both apple color genes but encodes for slightly different proteins!

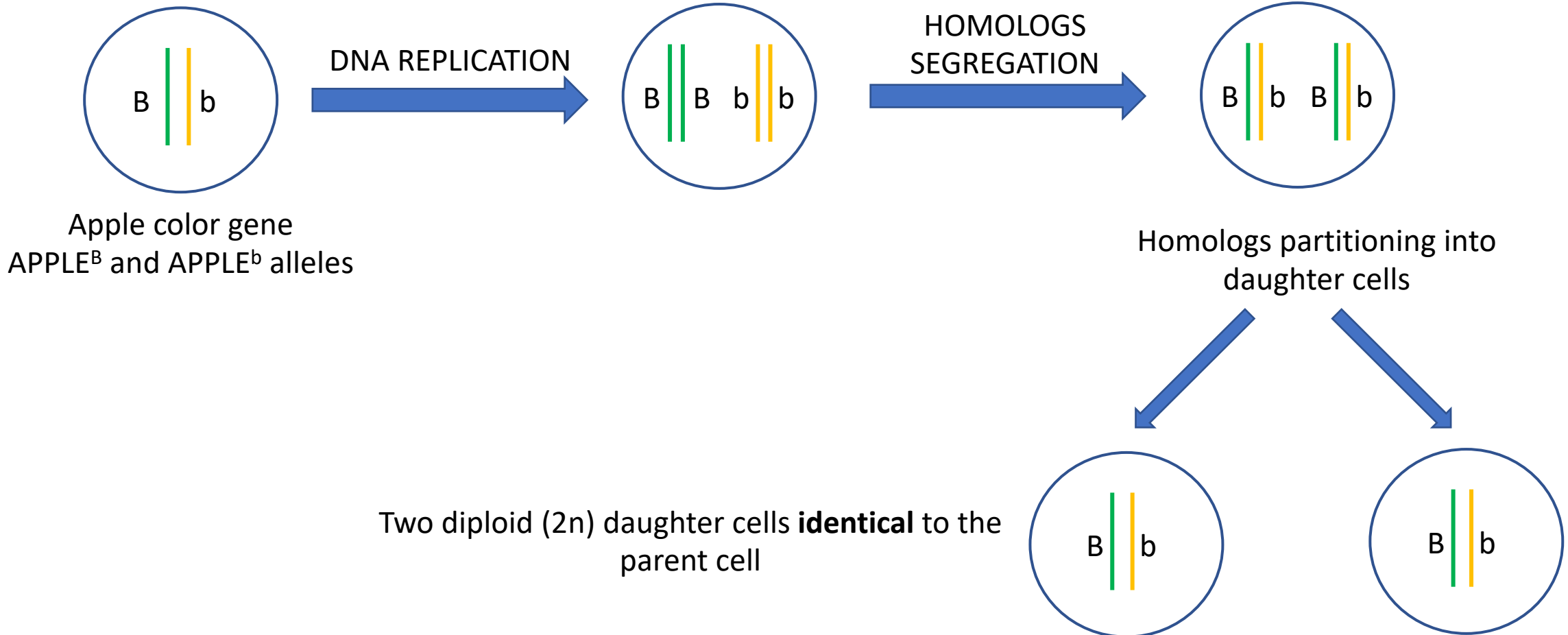
# Cell division and alleles

- 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

Diploid (2n) cell with 1 homolog

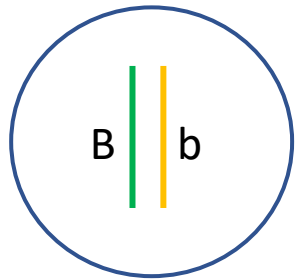
Sister chromatids





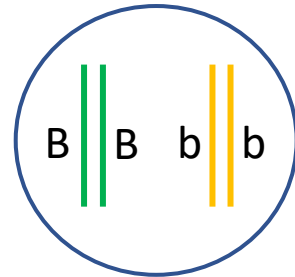
# Meiosis (germline inheritance) – revisited & simplified

Diploid (2n) cell with 1 homolog

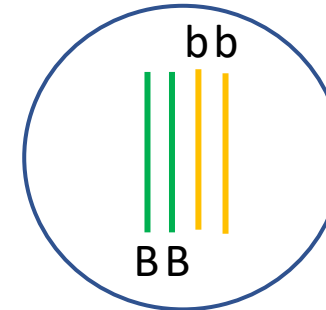


DNA REPLICATION

Sister chromatid



Sister chromatid aggregation\*



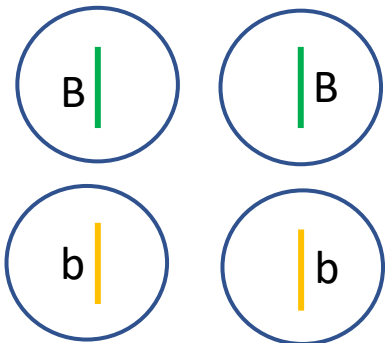
\*Chiasma formation and possible crossing-over

Apple color gene  
APPLE<sup>B</sup> and APPLE<sup>b</sup> alleles

Two diploid (2n) daughter cells  
non-identical to the parent cell

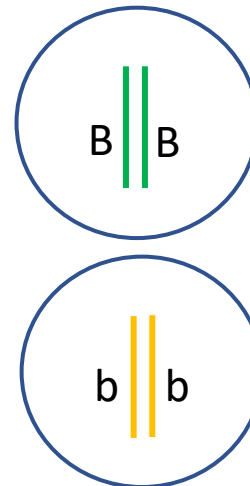
SISTER CHROMATID  
SEGREGATION

Four haploid (n) daughter cells  
non-identical to the parent cell



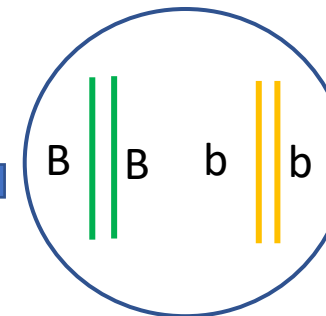
SISTER CHROMATID  
PARTITIONING  
INTO DAUGHTER CELLS

MEIOSIS 2

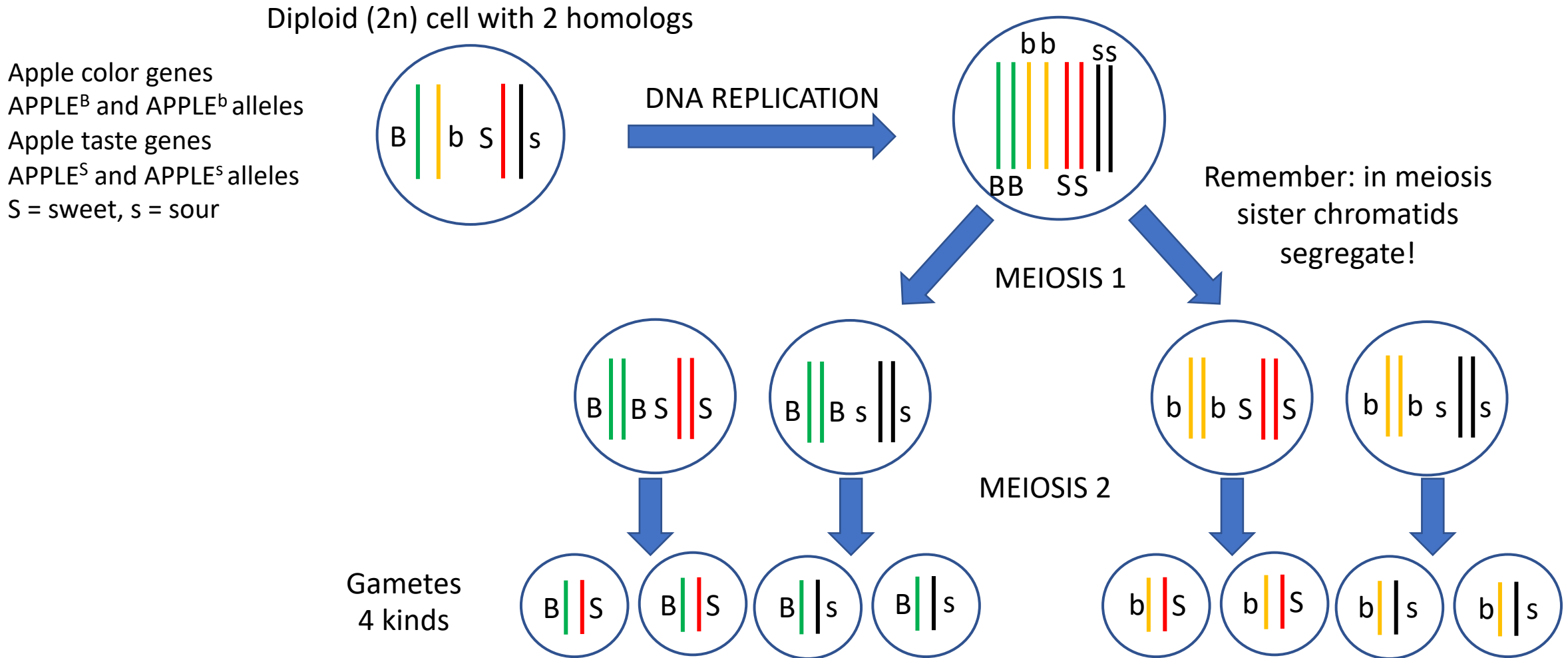


SISTER CHROMATID  
PARTITIONING  
INTO DAUGHTER CELLS

MEIOSIS 1



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



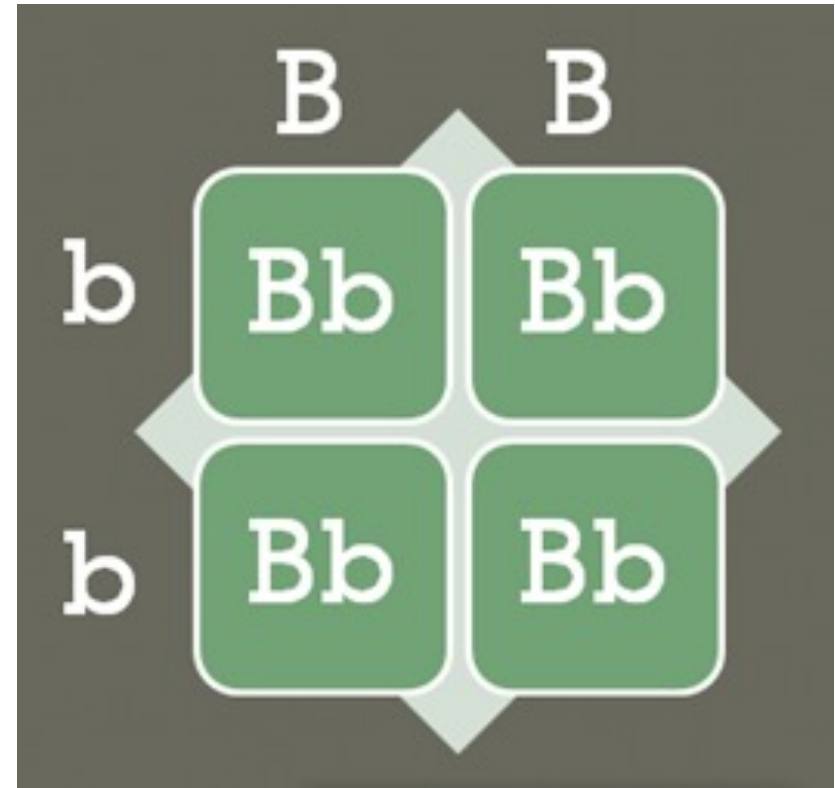
# Independent allele segregation

- 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

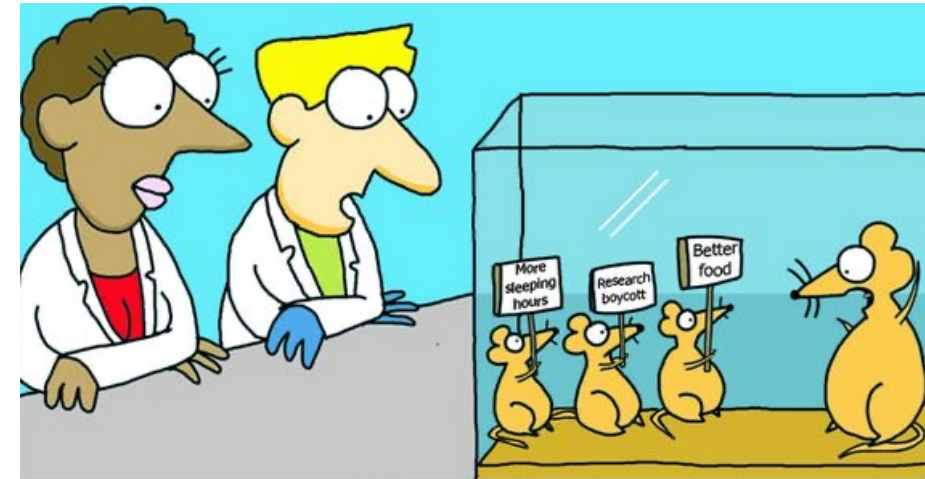


# Genetics & crosses

- 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

# Genetics & crosses

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- **Genetics** = set of tools to understand gene functions and inheritance
- **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



“Our mistake.  
We introduced a politician’s genes in that one!”

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- **Gene** = DNA sequence required to make the final product (usually a protein)
  - In genetics DNA = hereditary unit
- **Alleles** = alternate gene forms

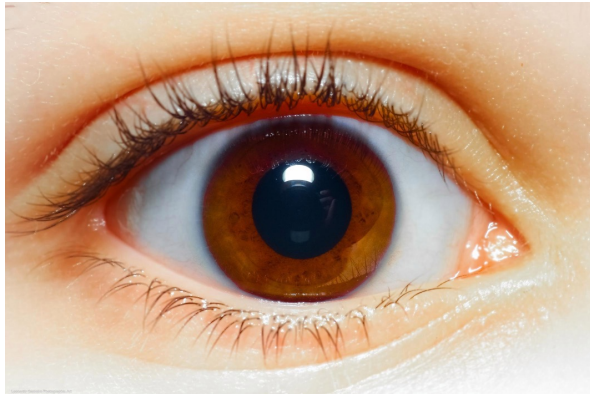
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- **Generation (gen)** = individuals born at the same time from the same parents
  - P gen = parents
  - F1 gen = first generation offspring
  - F2 gen = second generation offspring.....



# 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



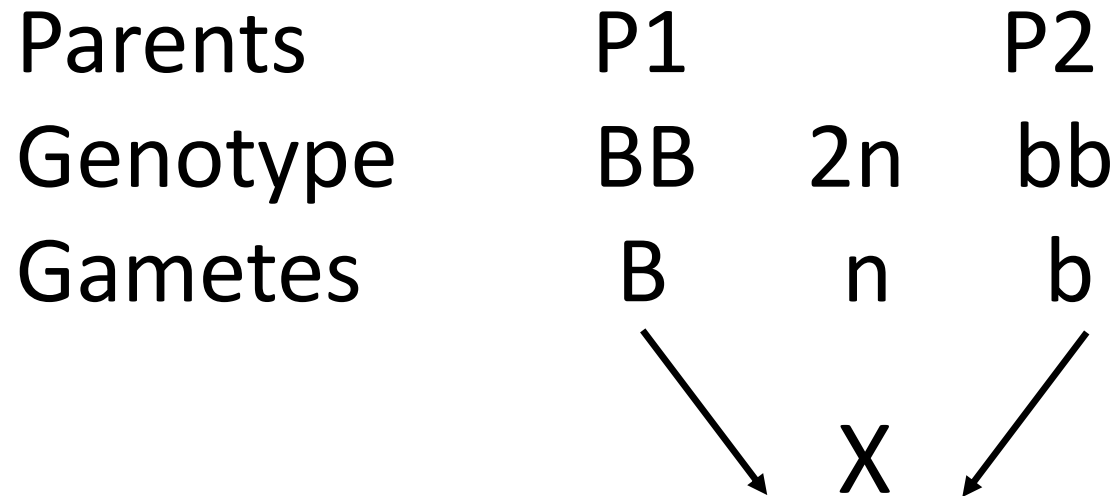
Theoretical breeding  
experiment

# 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 (haploid, 2x) Gametes of P2: b (haploid, 2x)

# Gene crosses – monohybrid crosses



**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

Parents	P1	X	P2
Genotype	BB		bb

Punnett square representation: genotype

		gametes	
		B	B
gametes	b	Bb	Bb
	b	Bb	Bb

F1 offspring

Shows F1 genotype and proportions

100% Bb

BB, bb = homozygote    Bb = heterozygote





# Punnett squares

Parents  
 Genotype P1 x P2  
 BB bb  
 Phenotype Green Yellow

Punnett square representation  
 Genotype + phenotype

		Gametes	
		B	B
Gametes	P1 P2 b	Bb green	Bb green
	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

Parents  
Genotype  
Phenotype

F1 = P1  
Bb  
Green

x

F1 = P2  
Bb  
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)

		Gametes	
		B	b
Gametes	P1 P2 B	BB green	Bb green
	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

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

If P1 is BB

100% offspring are green (Bb)

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

If P1 is Bb

50% offspring are green (Bb)

50% are yellow (bb)