

Lessons 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

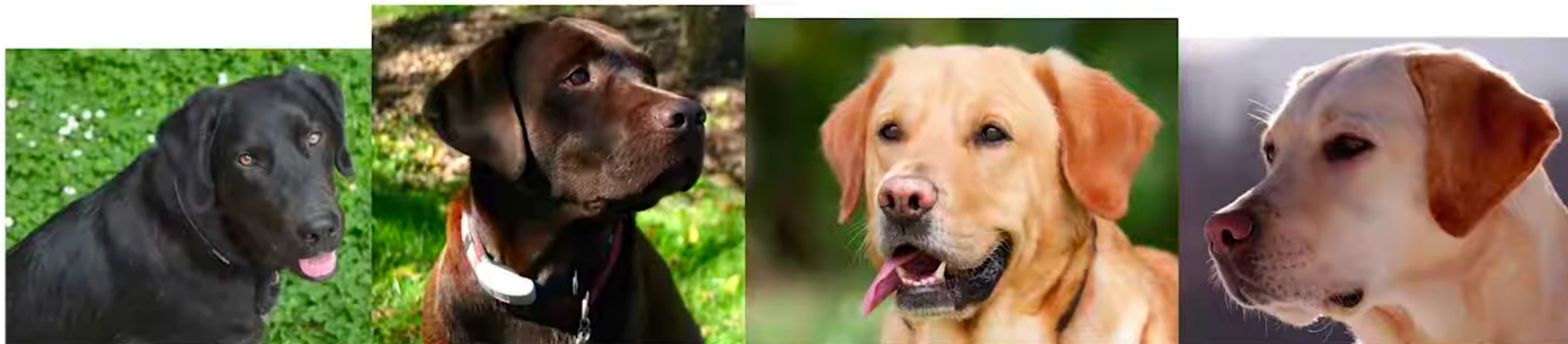
Alleles



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

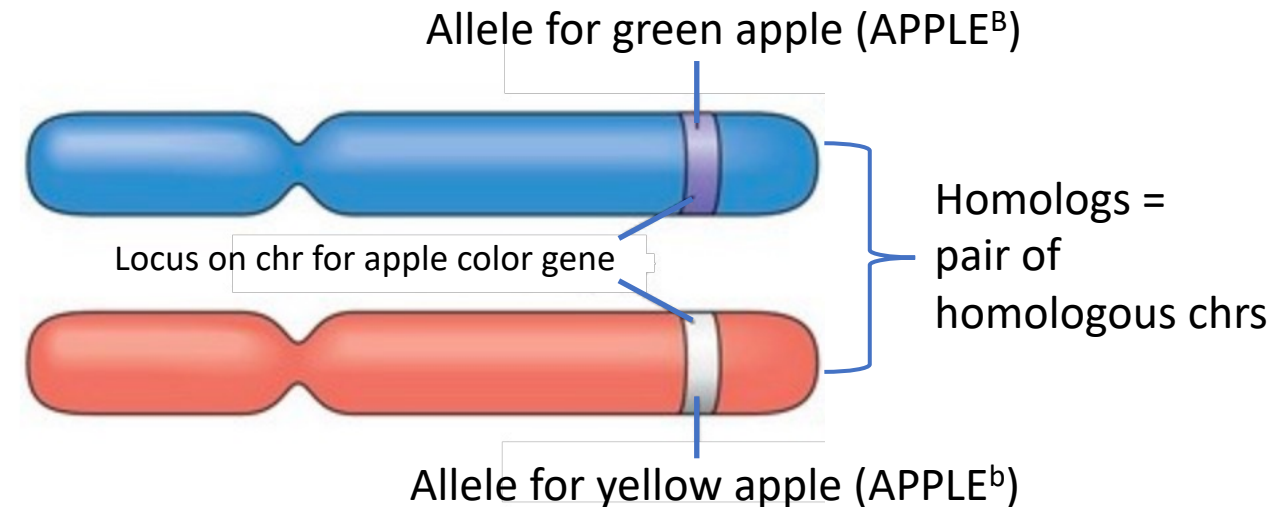
- $APPLE^B \rightarrow$ gives you a green apple
- $APPLE^b \rightarrow$ gives you a yellow apple



- 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 ^B
mRNA →	5' AUGUGGCUCCUGGAUUAA 3'	
protein →	N-Met-Trp-Leu-Leu-Asp-C (stop)	
Template strand →	5' ATGTGGCTCCTGGTTTAA 3' 3' TACACCGAGGACCAATT 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!

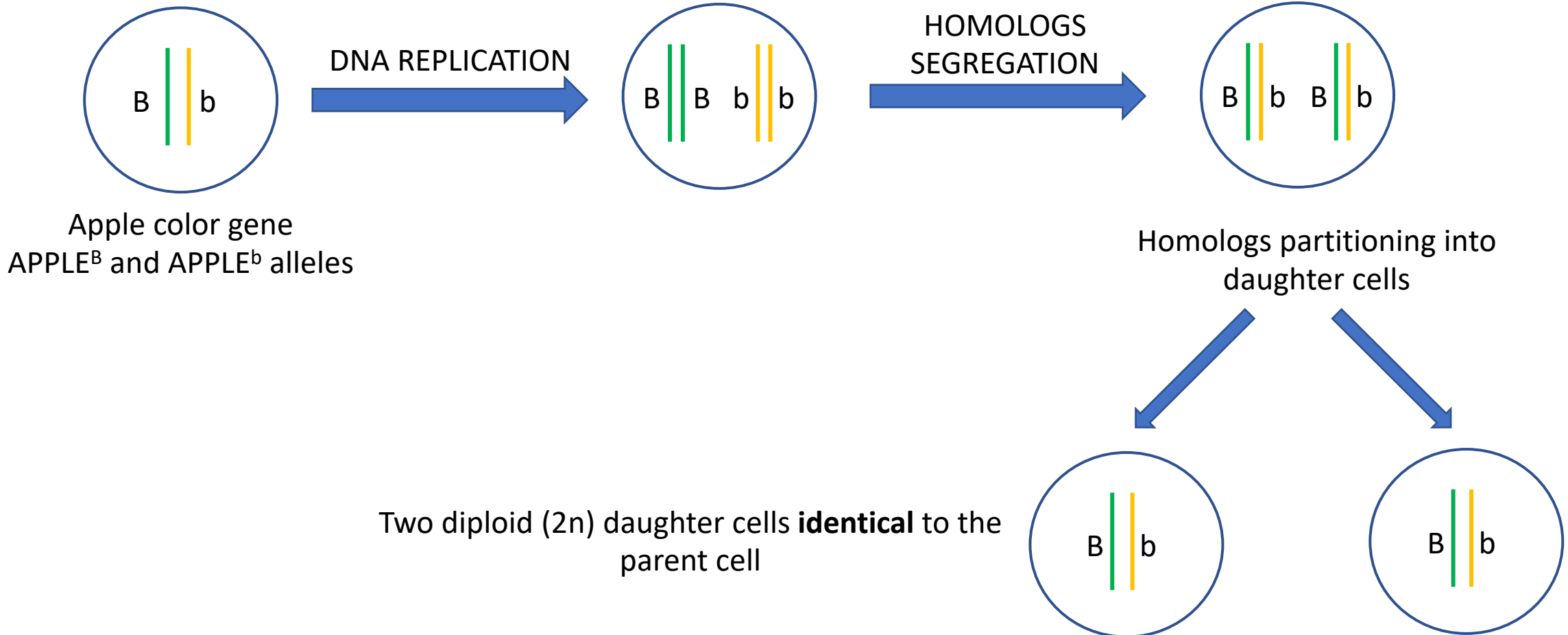
Cell division and alleles

- 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

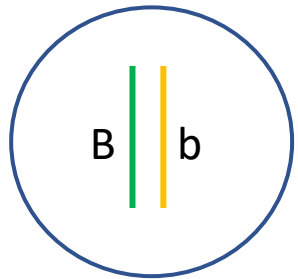
Diploid ($2n$) cell with 1 homolog

Sister chromatids



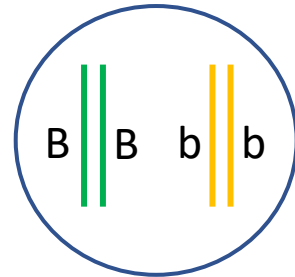
Meiosis (germline inheritance) – revisited & simplified

Diploid (2n) cell with 1 homolog

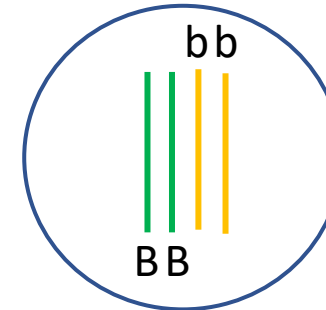


DNA REPLICATION

Sister chromatid



Sister chromatid aggregation*



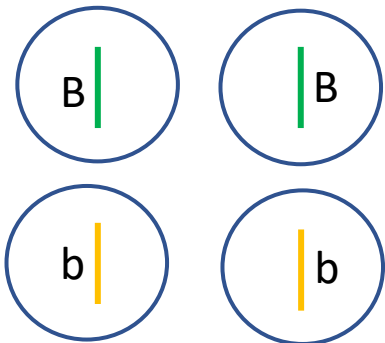
*Chiasma formation and possible crossing-over

Apple color gene
APPLE^B and APPLE^b alleles

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

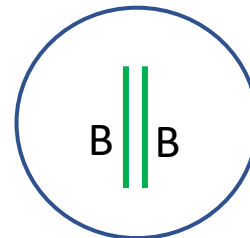
SISTER CHROMATID
SEGREGATION

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



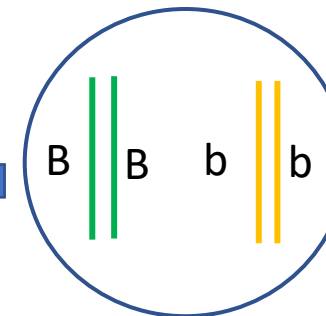
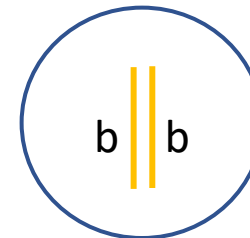
SISTER CHROMATID
PARTITIONING
INTO DAUGHTER CELLS

MEIOSIS 2

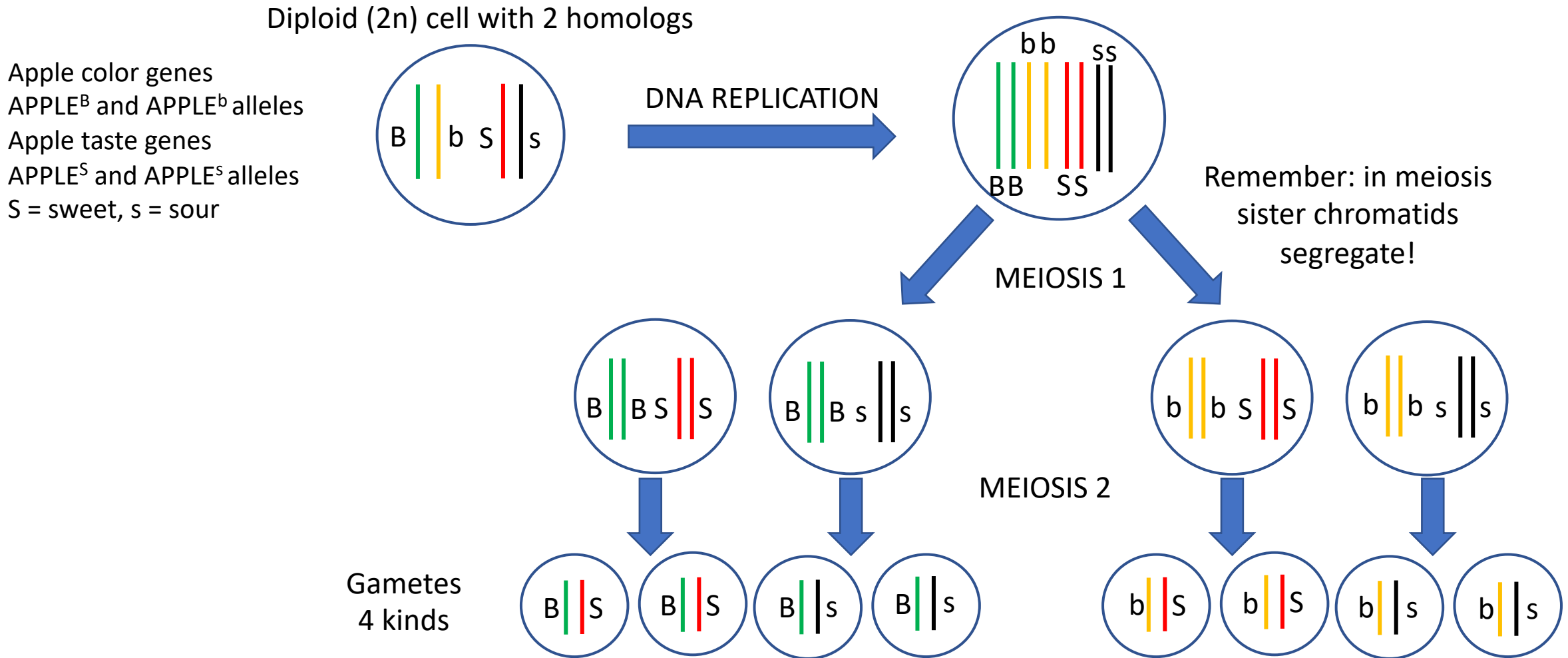


SISTER CHROMATID
PARTITIONING
INTO DAUGHTER CELLS

MEIOSIS 1



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



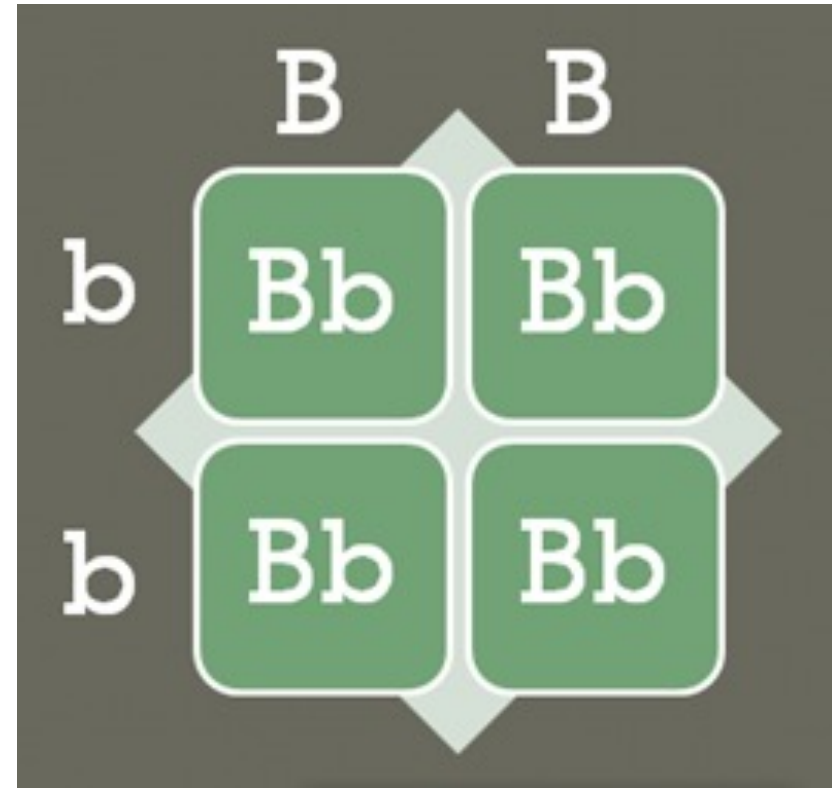
Independent allele segregation

- 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

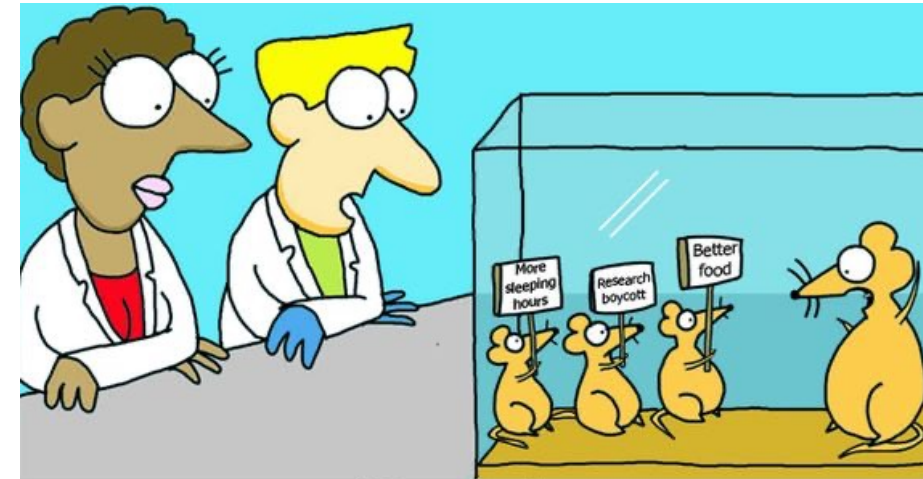


Genetics & crosses

- 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
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- **Gene** = DNA sequence required to make the final product (usually a protein)
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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



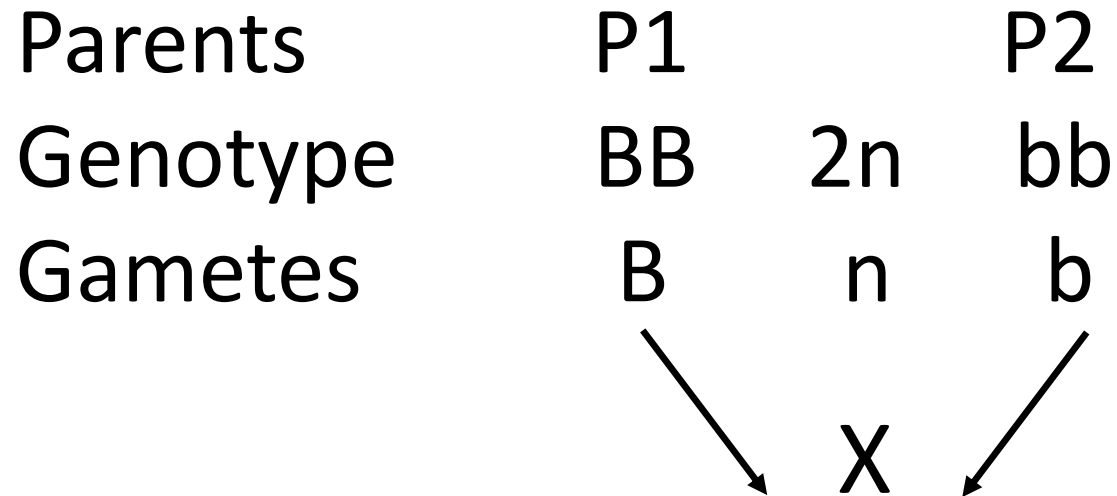
Theoretical breeding
experiment

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 (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

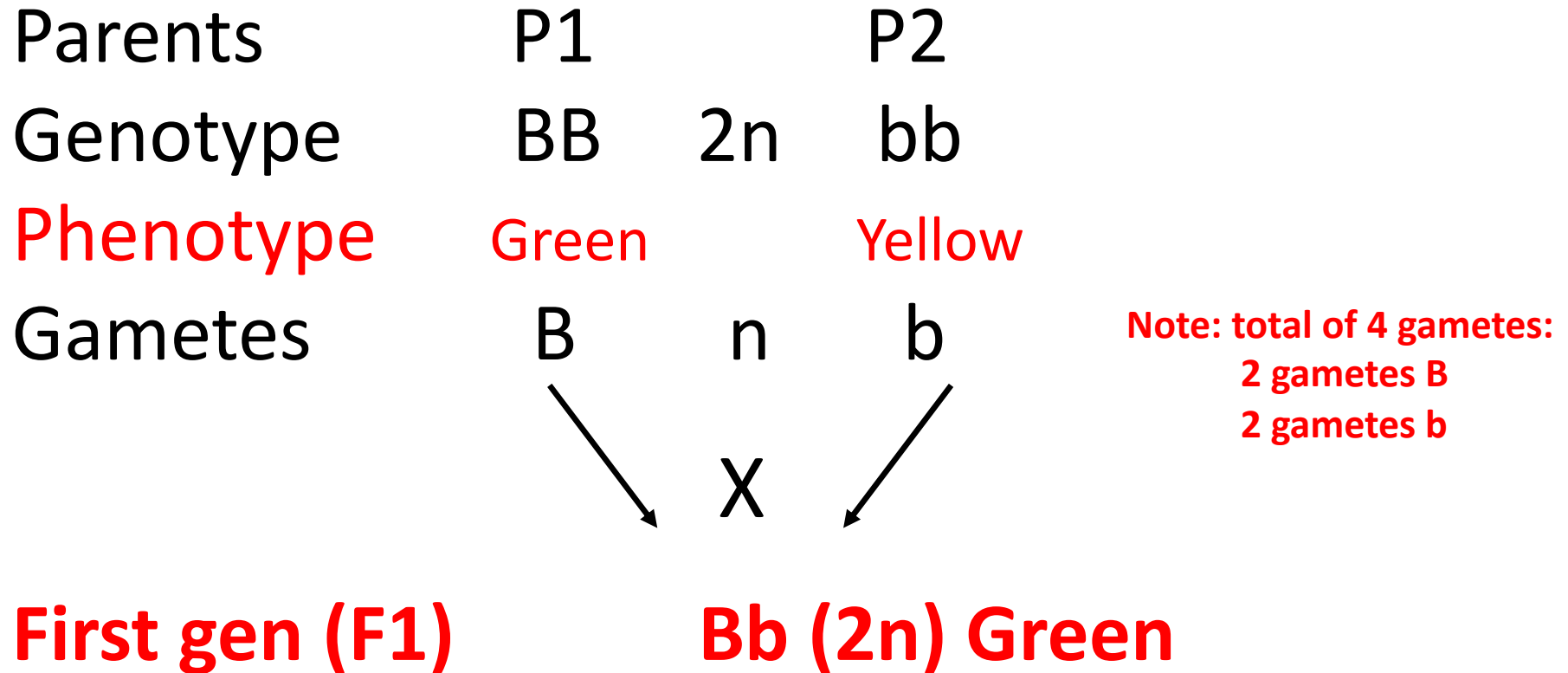
F1 offspring

Shows F1 genotype and proportions

100% Bb

BB, bb = homozygote Bb = heterozygote

Genetic crosses follow phenotypes (traits)



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

Punnett squares

Parents
Genotype P1 x P2
BB bb
Phenotype Green Yellow

Punnett square
representation
Genotype +
phenotype

		Gametes	
		B	b
Gametes	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 F1 = P1 x F1 = P2
 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)

		Gametes	
		B	b
Gametes	P1 P2	B	b
	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
		F1 offspring	
		Bb	Bb

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

		P1 gametes	
		B	b
P2			
	gametes	b	b
		Bb	bb
		F1 offspring	
		Bb	bb

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