Chapter 5 Topics

1. Animal Life Cycles
2. Structure of DNA in Eukaryotes and Prokaryotes!
3. Replication of DNA (copying of DNA)
4. Making proteins!
    - Transcription-going from DNA to mRNA
    - Translation-going from mRNA to protein

5. Gene regulation!
6. Sizing up the Genome
7. Mutations
8. Mitosis vs Meiosis (sexual reproduction)
9. **Mendel and Punnett Squares**
10. **Getting more real….**

Chapter 5 Final Topic

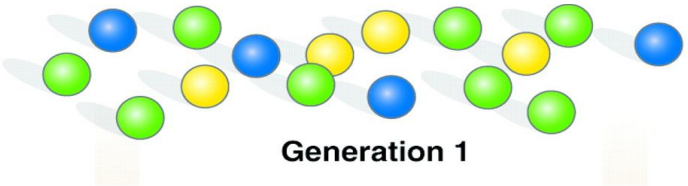
Mendel and Punnet Squares

Everyone was interested in inheritance during 2nd half of 1800s

Mostly people believed in blending inheritance.

What is blending inheritance??

Imagine balls are individuals filled with paint and you mimic mating by randomly pulling them out of a bucket two by two….



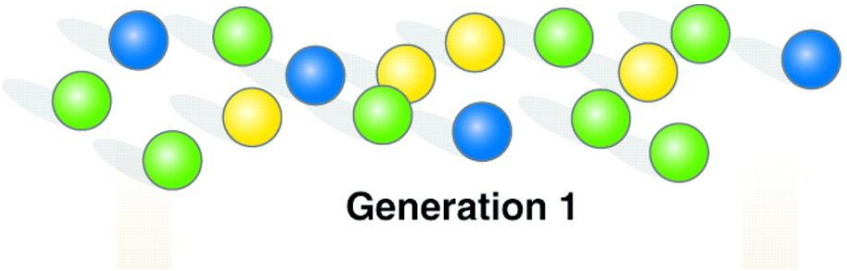
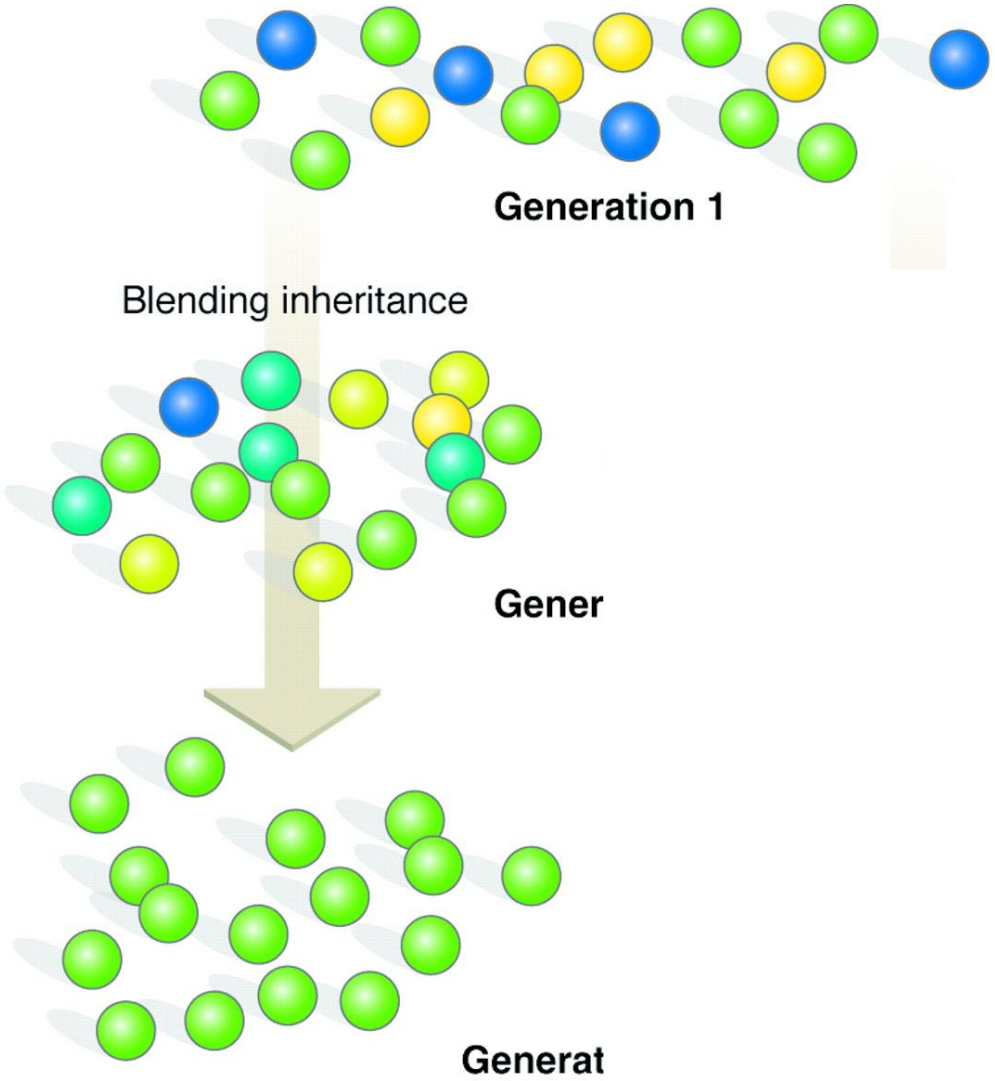


Fig. 1. Difference between the outcomes from blending and from particulate inheritance. In post-Mendelian terms, we assume a single diallelic locus, and hence three diploid genotypes (AA, blue; Aa, green; aa, yellow). Under particulate inheritance, the population's variability is preserved from generation to generation. In contrast, the conventional wisdom of Darwin's day saw offspring inherit a blend of parents' characteristics, here represented as the average of the two parental shadings. The result is that the variability diminishes in successive generations (the variance is halved each generation if mating is at random) SCIENCE MAGAZINE B. MAY



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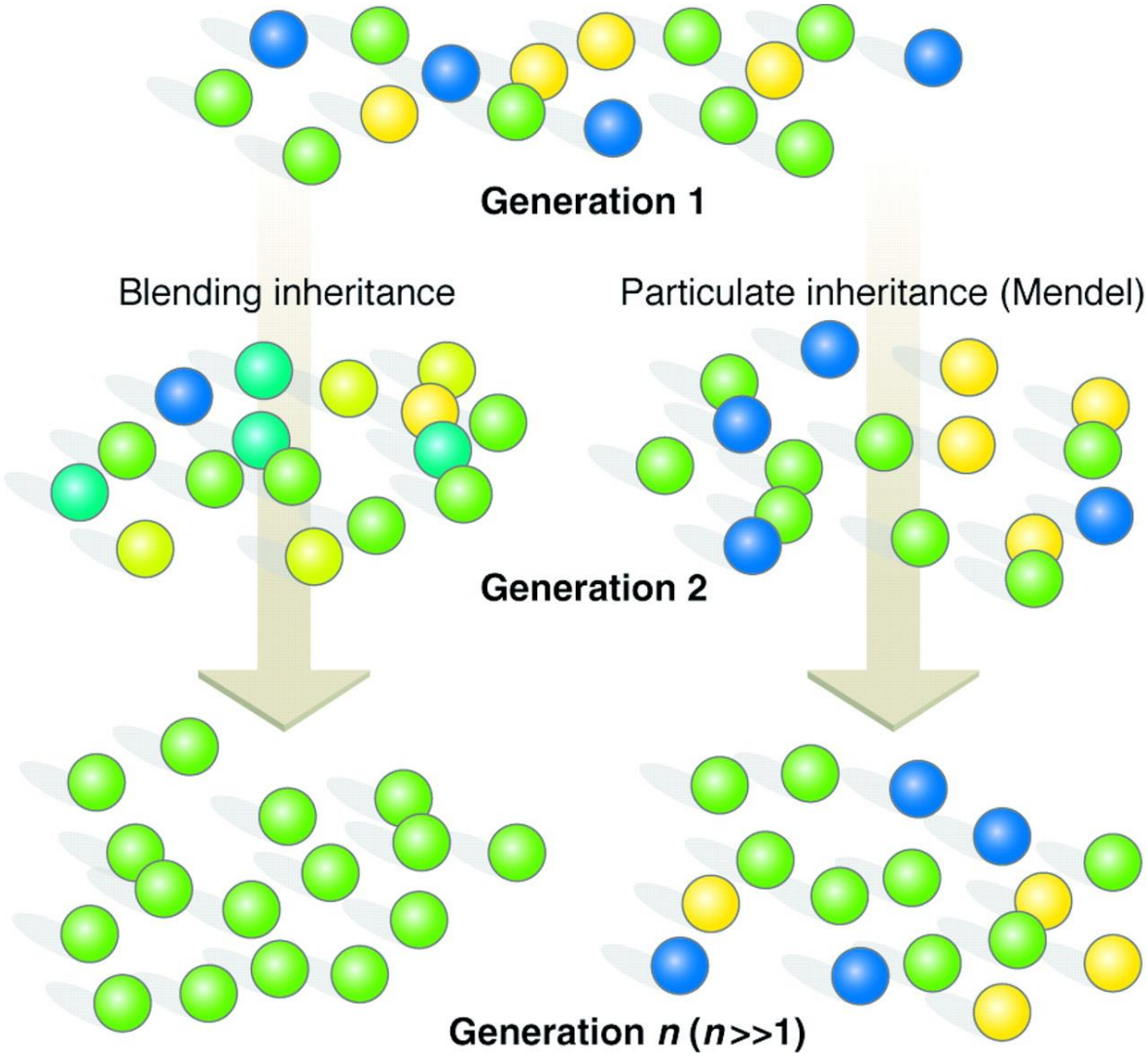

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Why was Mendel able to deduce the “particulate pattern” while others were struggling?

Imagine you are interested in inheritance…….

What measurements could you make on the people in this class?

Pictorial Press Ltd / Alamy Stock Photo

Box Figure 5.2.1 Gregor Mendel (1822–84) first recognized that inherited traits were made possible by factors (now called genes) passed down from parents to offspring.

Why was Mendel successful?

He was successful because he chose ….

- flower color is purple or white
- flower position is axil or terminal
- stem length is long or short
- seed shape is round or wrinkled
- seed color is yellow or green
- pod color is yellow or green

He did not choose traits like

- number of seeds per pod
- pod length
- pod width
- seeds per plant
- seed weight
- plant height

To review…

Mendel chose to track only characters that varied in an "either-or" rather than a more or less manner. His plants had either purple or white flowers, there was nothing intermediate between these two varieties.

Had Mendel focused instead on characters that vary in a continuum among individuals-seed weight for example-he would not have discovered the particulate nature of inheritance!

Considering all that can occur in the pathway from genotype to phenotype it is amazing that Mendel could simplify the complexities to reveal the fundamental principles governing transmission of individual genes from parents to offspring!

1865 Mendel publishes his PARTICULATE VIEW of Inheritance …

Almost completely ignored even though it would have made natural selection work to drive evolution!

Was not until the Modern Synthesis that Mendel's ideas came together with mathematical models of natural selection.

Review of terms…

What is a locus?

What are alleles?

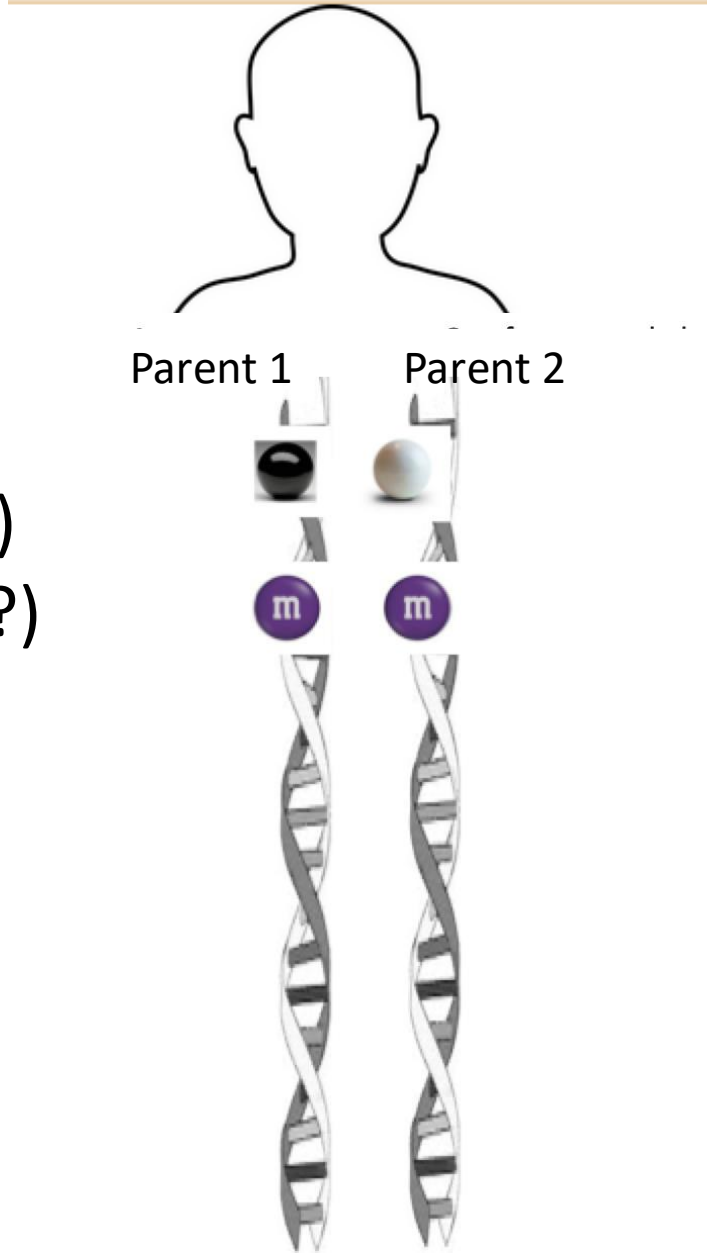
For a diploid organism, how many alleles will there be at each locus?

Homozygous? (Gumball or M+M?)

Heterozygous? (Gumball or M+M?)

Dominant?

Recessive?



This shows two homologous chromosomes in one individual.

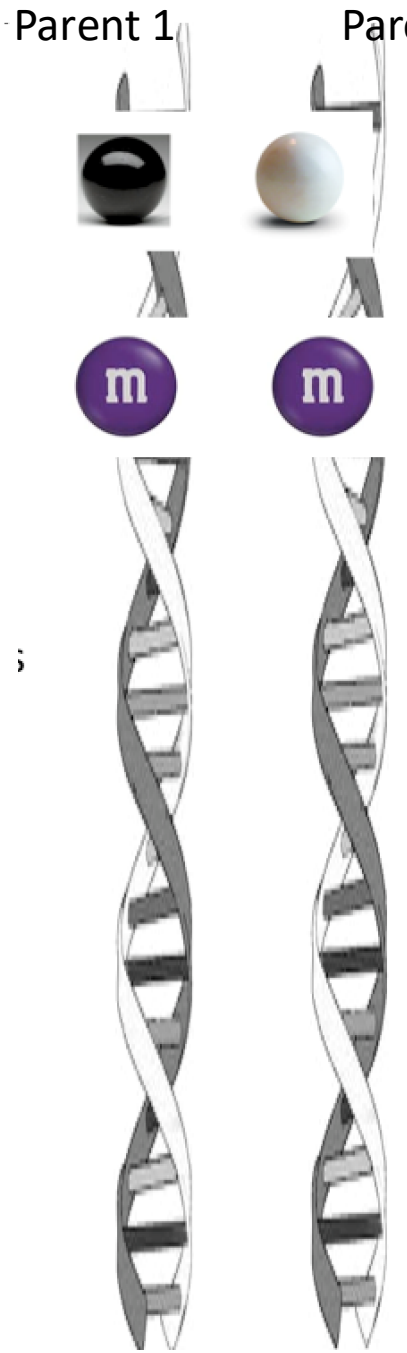
The chromosomes are labeled according to which parent they were passed on from.

We have two copies of each chromosome…

The black allele at the gumball locus

The white allele at the gumball locus

Shown here are two homologous chromosomes of the 23 pairs of chromosomes you have.



At the gumball locus you are heterozygous… Bb (?)

At the m+m locus you are homozygous… PP (?)

IF the black gumball is DOMINANT to the white gumball, this individual's phenotype would be (what color would the be if the gumball locus affected fur color)…..?

We have two copies of each chromosome…

The black allele at the gumball locus
The white allele at the gumball locus

Parent 1 Parent 2

At the gumball locus you are heterozygous…

At the m+m locus you are homozygous…

Shown here are two homologous chromosomes of the 23 pairs of chromosomes you have.

A. For a diploid organism, how many alleles will there be at each locus?

Image below shows 5 pairs of homologous chromosomes in 5 **different individuals**.

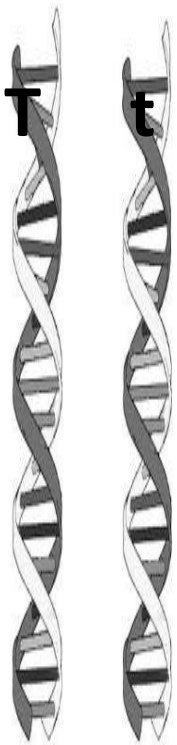
B. Which individuals are heterozygous in this **population** at the gumball locus?

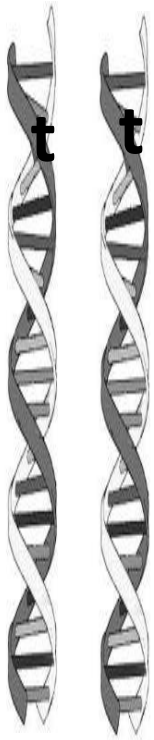
How many alleles are there in this **population** of 5 individuals at the gumball locus?

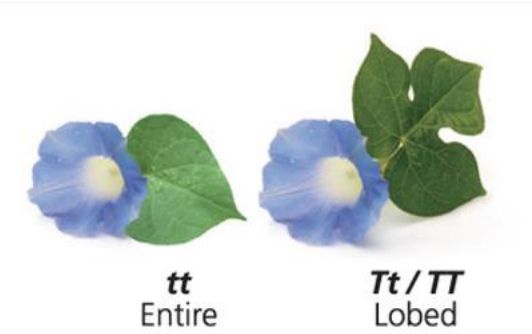
Lets focus on a **single locus ("gene")** on a **single chromosome** (imagine chromosome 1) for plants!

In plants we use upper and lower case letters (usually).

Here are three plants with different genotypes at the "lobed" locus.







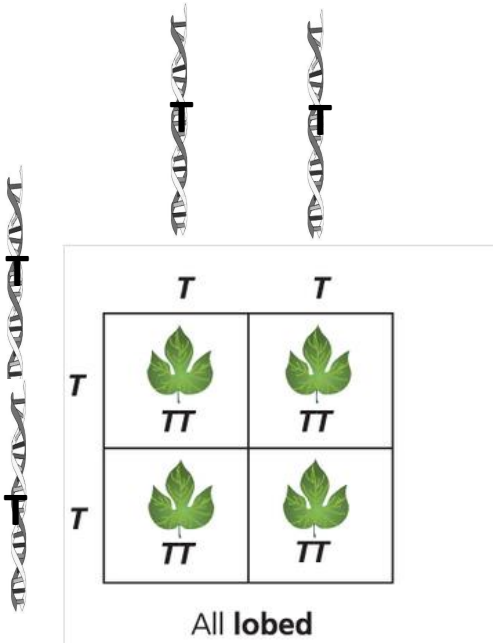
If each of these individuals were making gametes…..

Lets get organized and use a Punnett Square!

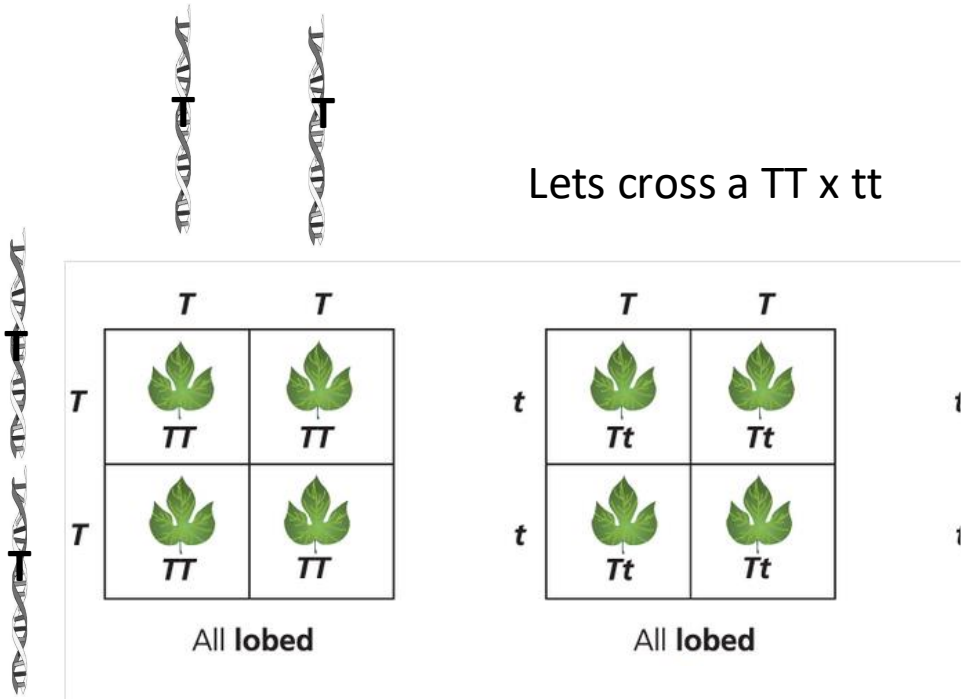
Example #1

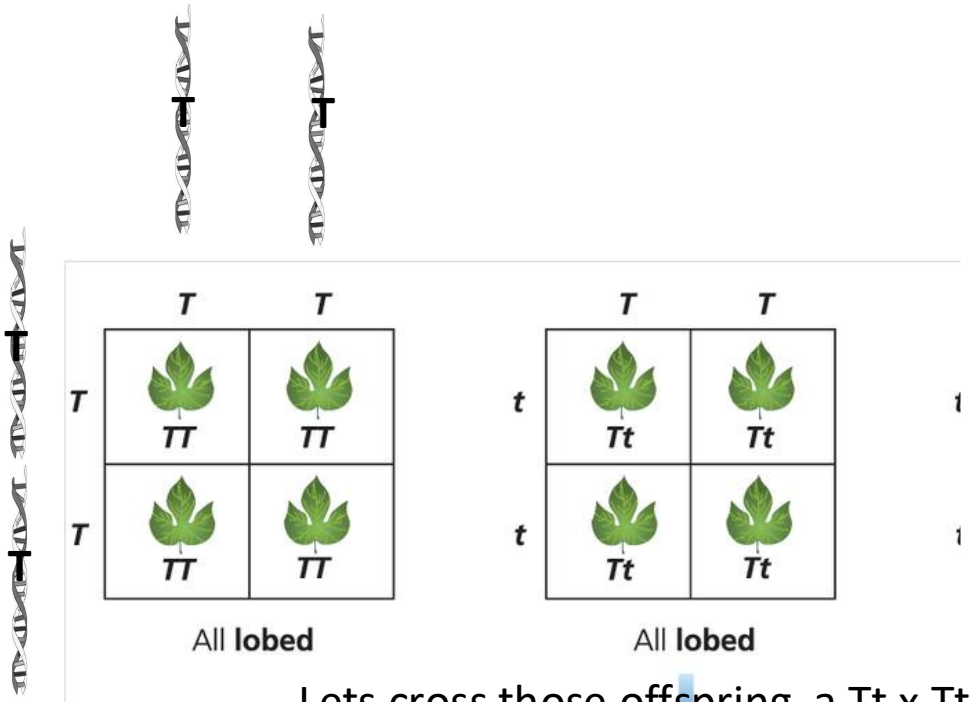
From your text

Lets cross a TT x TT

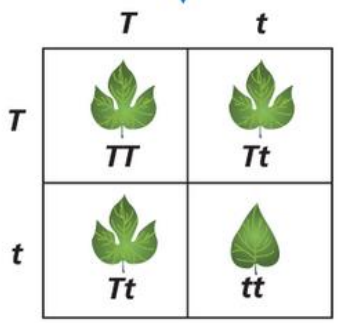


Which is dominant? T or t?





Lets cross those offspring a Tt x Tt

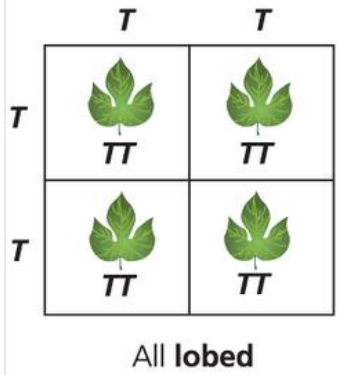


T T

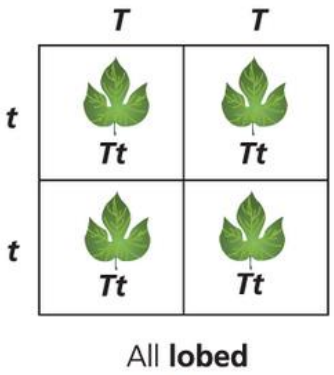
T

T

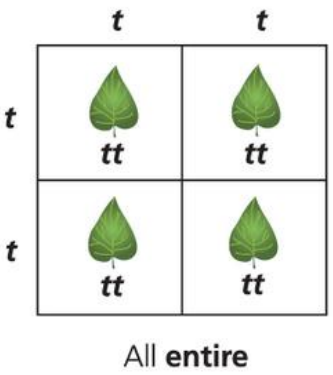
Lets cross a tt x tt



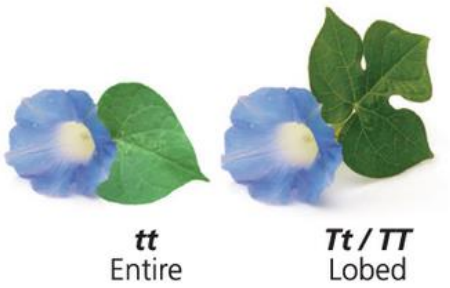
All **lobed**

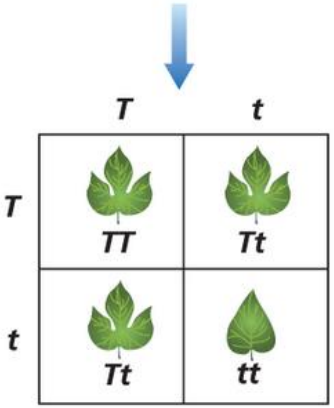


All **lobed**

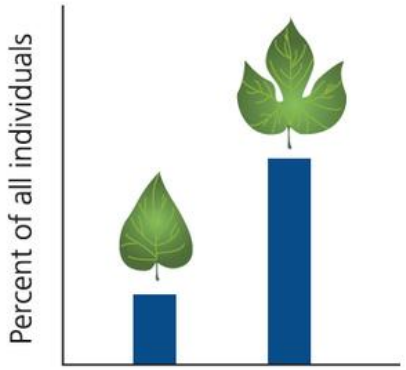


All **entire**





3:1 lobed/entire

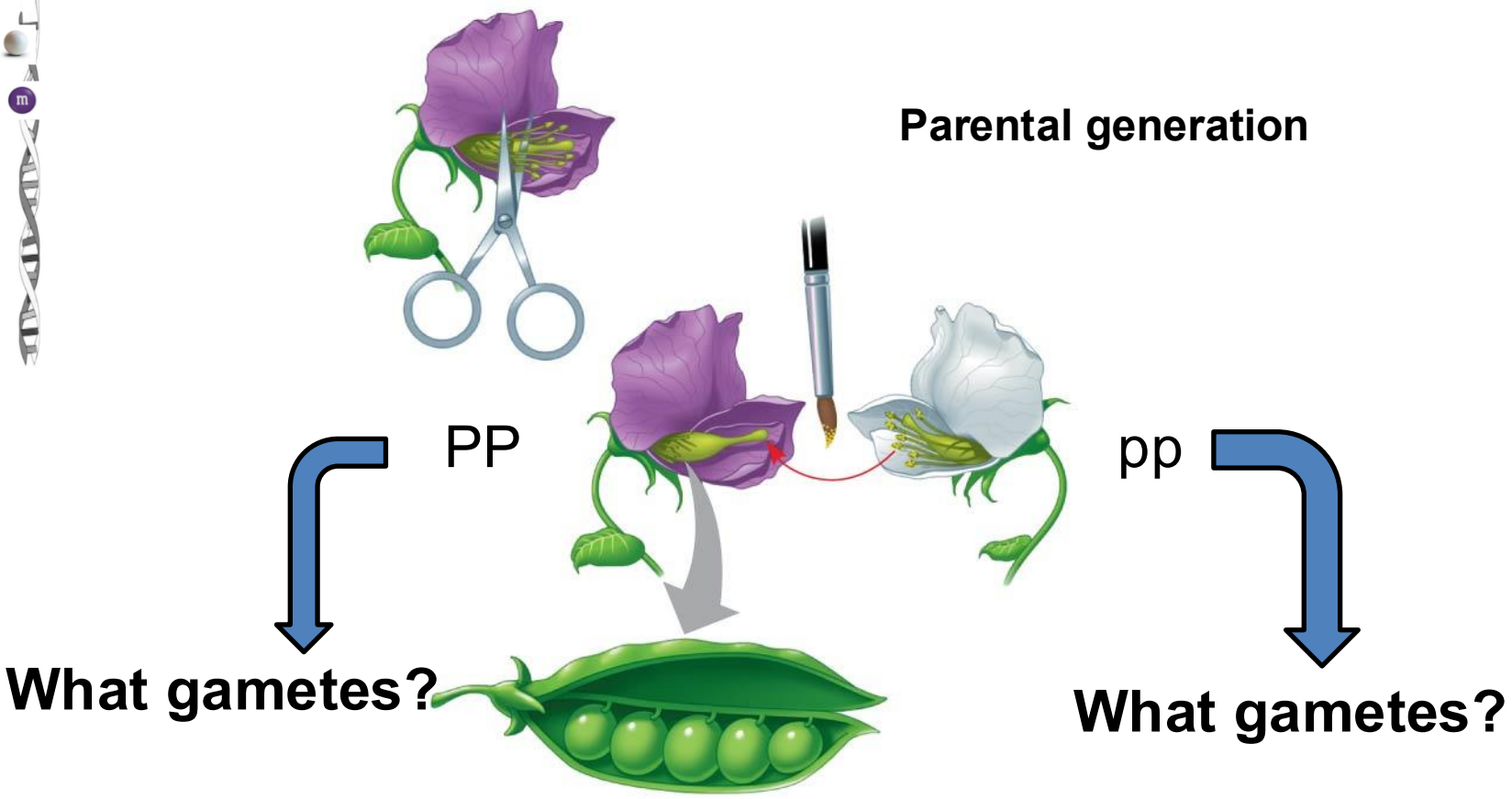


Punnett Square Example #2

I'll add another example-
Flower color in Mendel's peas

In pea plants P (purple flowers) is **dominant** to (p) white flowers…..

So if a plant's genotype at this locus is PP it will be..what color?
If it is Pp it will be...
If it is pp it will be…



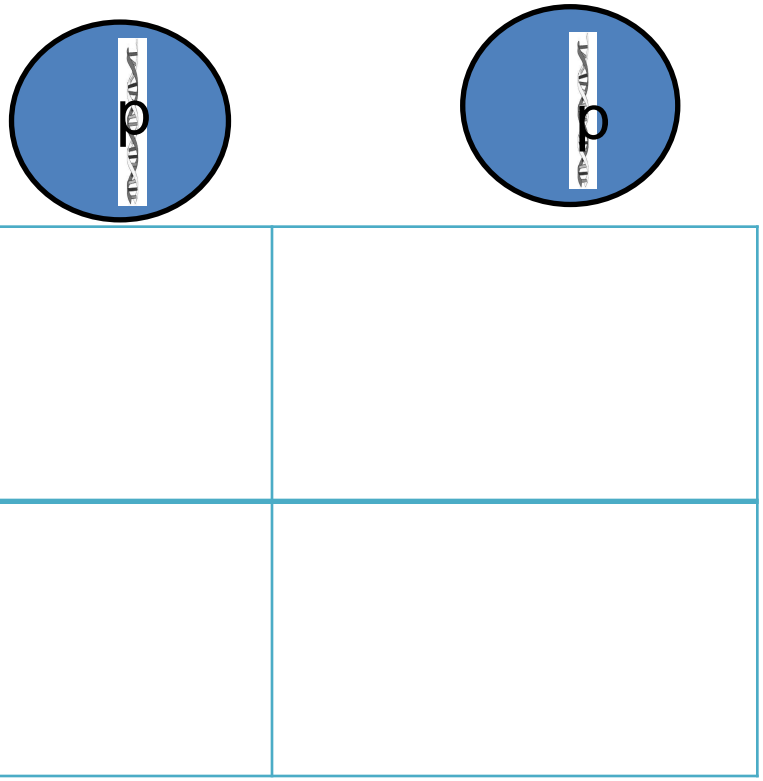
Genotypes of all offspring (F1)?

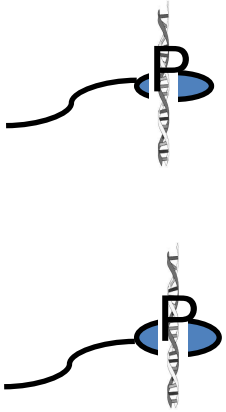
Phenotypes of all offspring?

An organized way to respond to this question of what are the genotypes going to be in the next generation is to use a **Punnett Square**..

PP x **pp** cross…

Practice picturing these as gametes!

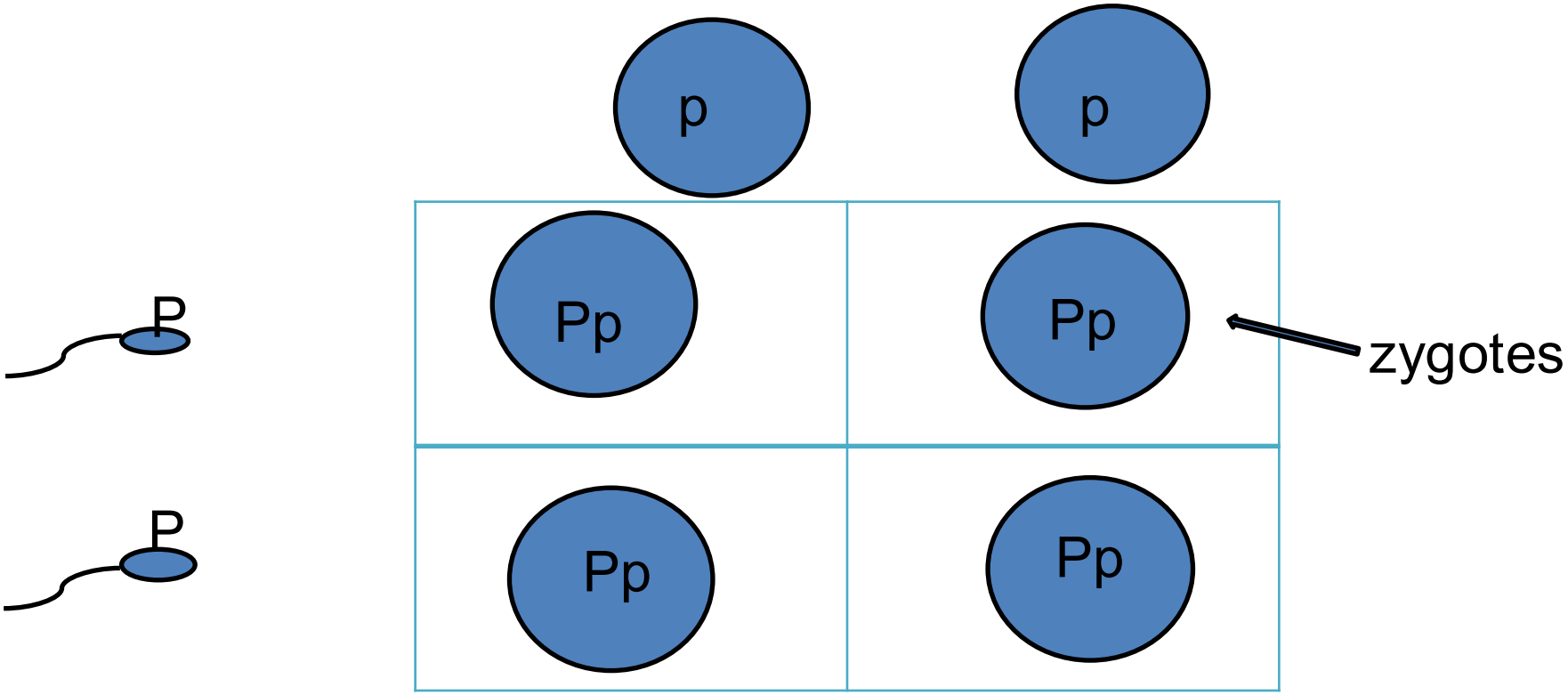




What are genotypes and phenotypes of offspring?

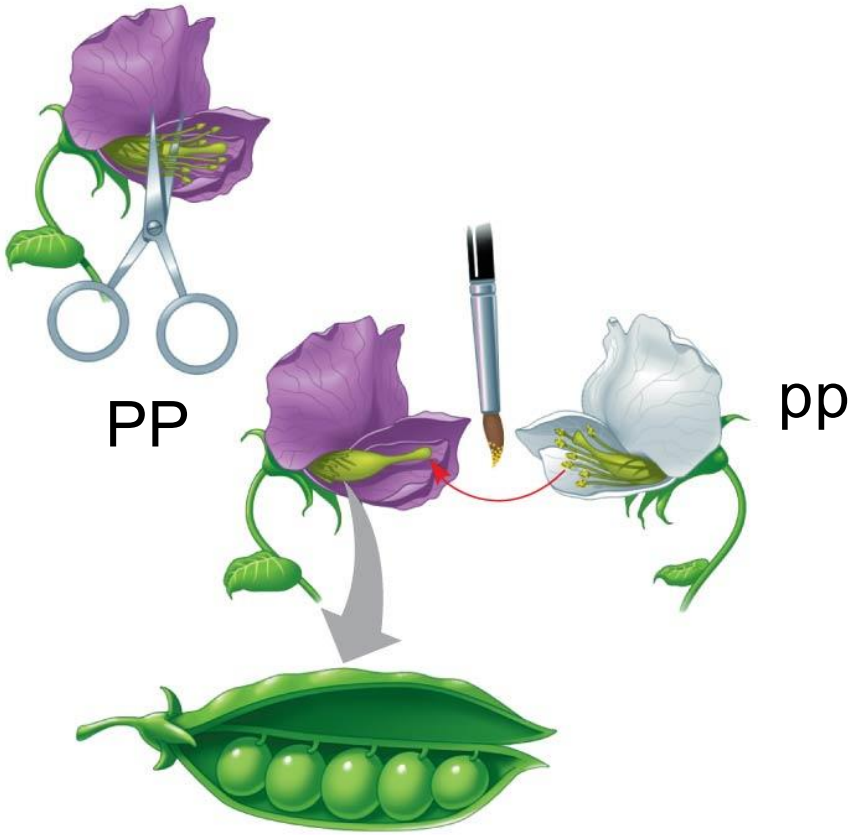
An organized way to respond to this question of what are the genotypes going to be in the next generation is to use a Punnett Square..

PP x **pp** cross…



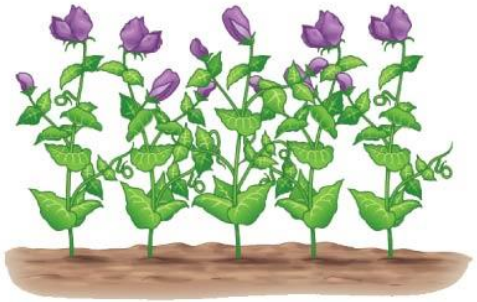
What are genotypes and phenotypes of offspring?

Did I even need a 2 x 2 table??



So these are all **Pp**…right?

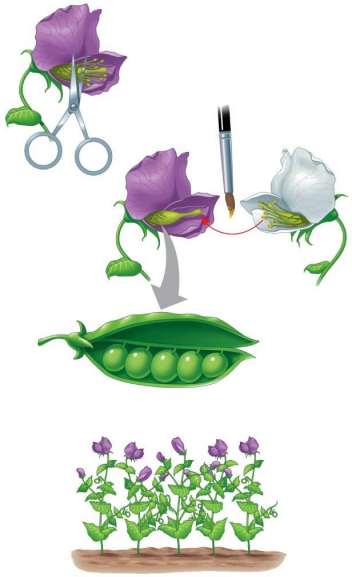
Now lets cross two of these!



F1

What if you interbred the Pp offspring to get a second generation?

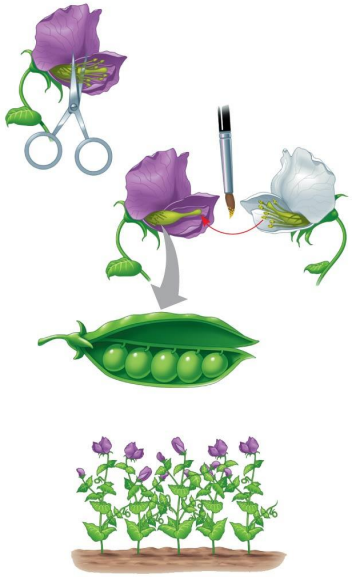
Pp x **Pp** cross…



	P	p
P		
p		

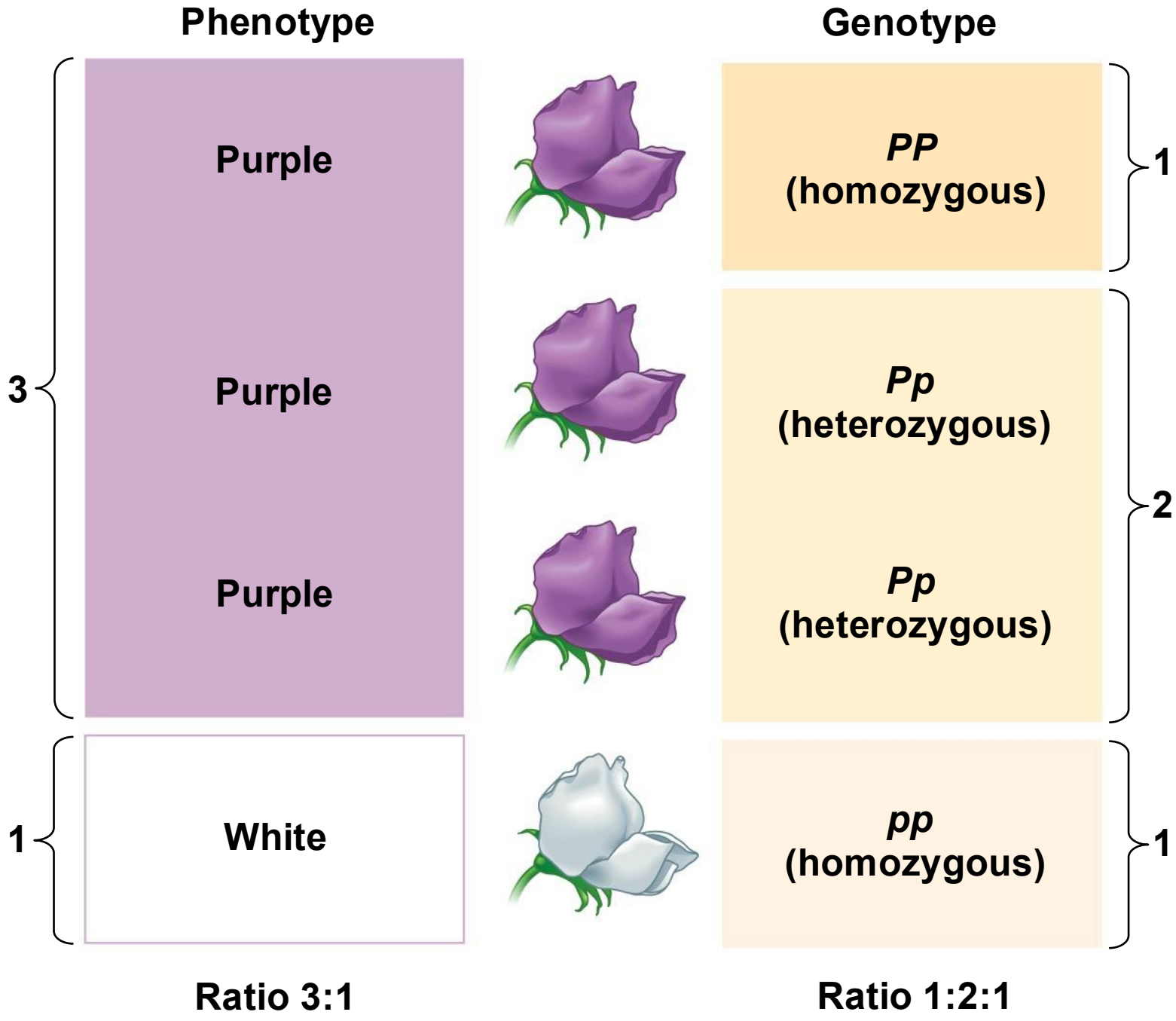
What are genotypes and phenotypes of these F2 offspring?

Pp x Pp cross…
What colors are the next generation‘s flowers if you grew them up?

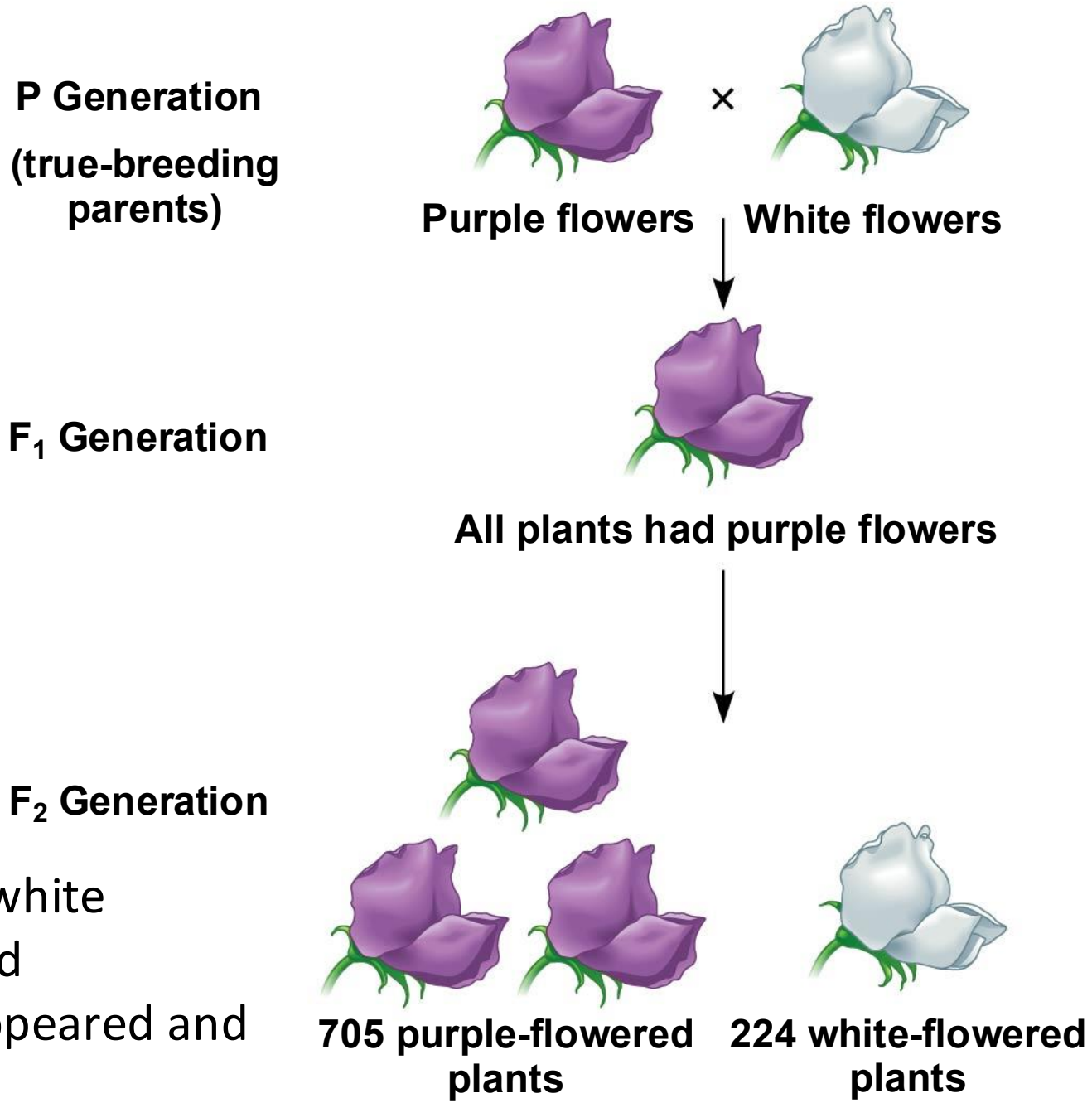


	P	p
P	PP	Pp
p	Pp	pp

What are genotypes and phenotypes of these F2 offspring?


Phenotype
Genotype
Purple
Purple
Purple
White
PP
(homozygous)
Pp
(heterozygous)
Pp
(heterozygous)
pp
(homozygous)
3
1
1
2
1
Ratio 3:1
Ratio 1:2:1

Is this observation compatible with **blending inheritance**????



This reappearance of white flowers was considered amazing!! White disappeared and then reappeared!

Was pretty impressive that through these results Mendel was able to infer what was going on!

He figured out that there were true “particles” that retained their identity.

Was not like paint blending!

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3. Replication of DNA (copying of DNA)
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    - Transcription-going from DNA to mRNA
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5. Gene regulation!
6. Sizing up the Genome
7. Mutations
8. Mitosis vs Meiosis (sexual reproduction)
9. Mendel and Punnett Squares
10. **Getting more real….**

10. Getting more real…..

A. Most real loci have many alleles when you look at a population!

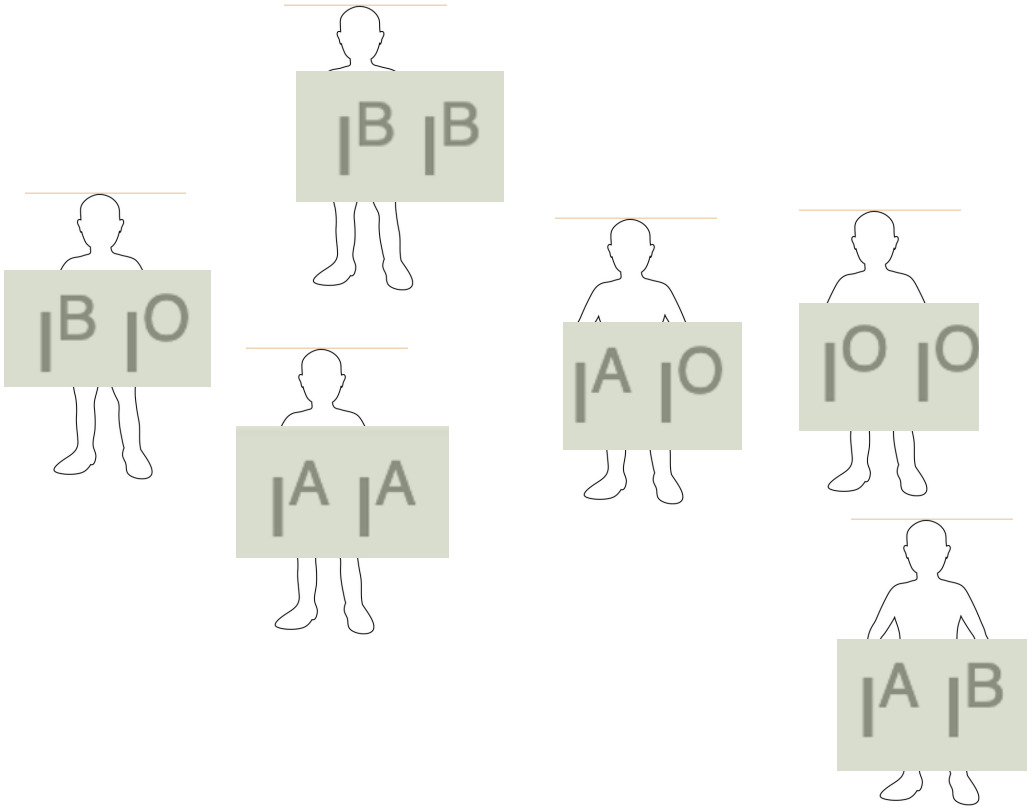
- Blood group alleles… I^A, I^B, and I^O
- So what genotypes would be possible in this population?

Remember each individual only has “2 slots to fill”!

10. Getting more real…..

A. Most real loci have many alleles when you look at a population!

- Blood group alleles… I^A, I^B, and I^O
- So what genotypes would be possible in this population



10. Getting more real…..

A. Most real loci have many alleles when you look at a population!

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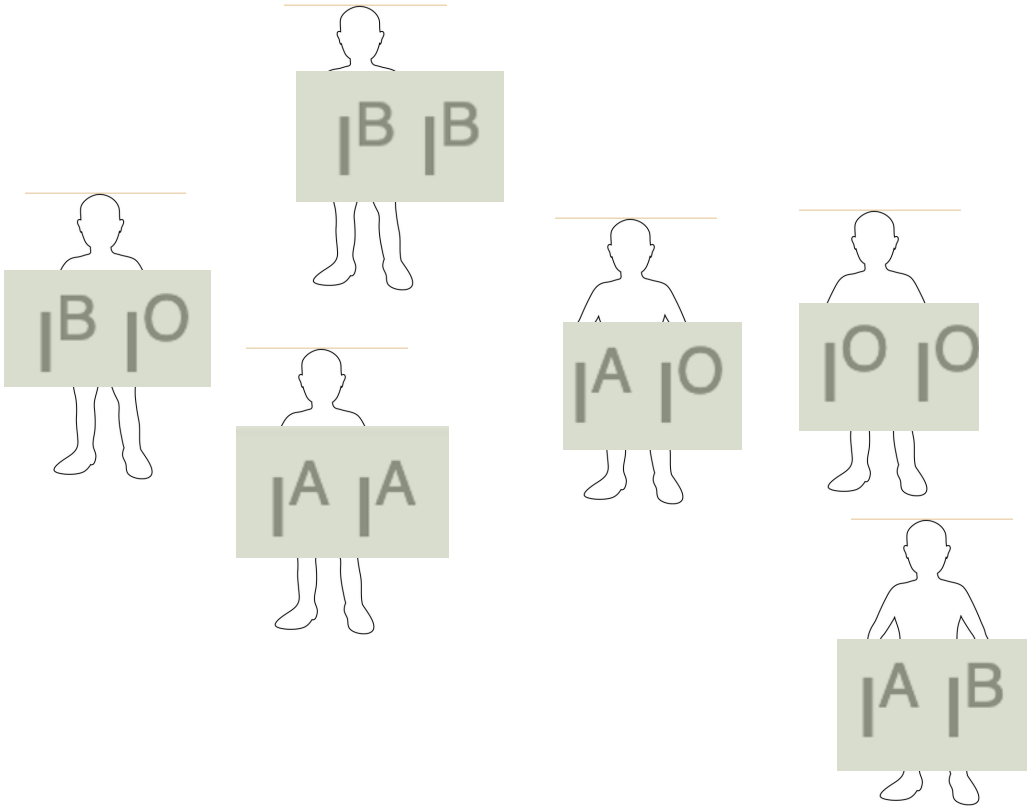


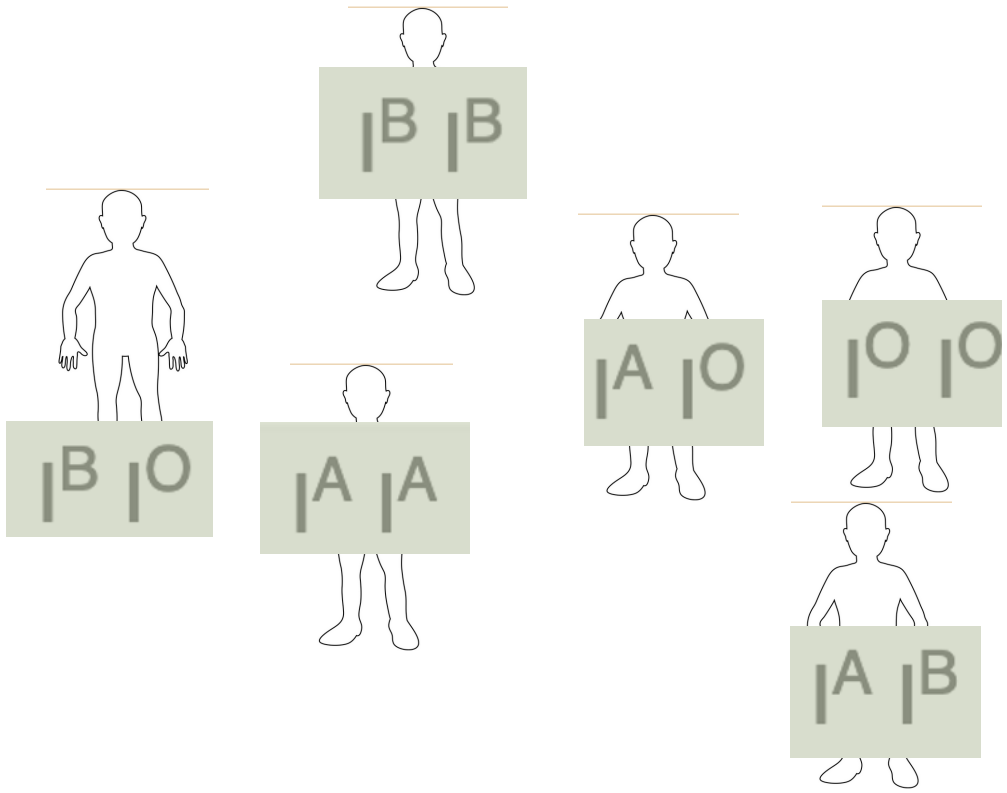
Genotypes in the *population*	Phenotype in the *population*
$I^A I^A$	
$I^A I^O$	
$I^B I^B$	
$I^B I^O$	
$I^A I^B$	
$I^O I^O$	

Now I will tell you what the phenotypes will be!

10. Getting more real…..

A. Most real loci have many alleles when you look at a population!

- Blood group alleles… I^A, I^B, and I^O
- So what genotypes would be possible in this population

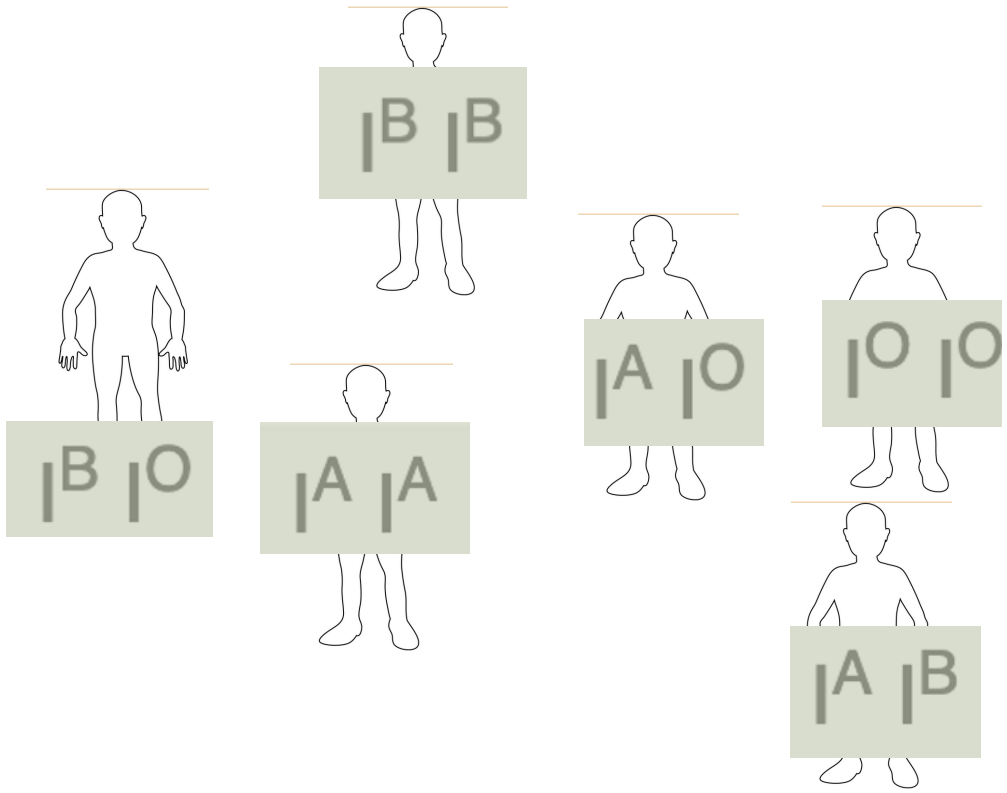


Genotypes in the *population*	Phenotype in the *population*
$I^A I^A$	A
$I^A I^O$	A
$I^B I^B$	B
$I^B I^O$	
$I^A I^B$	
$I^O I^O$	

10. Getting more real…..

A. Most real loci have many alleles when you look at a population!

Genotype	Blood Group
$I^A I^A$	A
$I^A I^O$	A
$I^B I^B$	B
$I^B I^O$	B
$I^A I^B$	AB
$I^O I^O$	O
Genotypes in the *population*	Phenotype in the *population*



10. Getting more real…..

A. Most real loci have many alleles when you look at a population!

- When you have three alleles how many different **genotypes** can you have?
- Major histocompatibility complex (MHC) genes also known as the human leukocyte **antigen** (HLA) may have 100 different alleles in a population at a single locus.

Genotype	Blood Group
$I^A I^A$	A
$I^A I^O$	A
$I^B I^B$	B
$I^B I^O$	B
$I^A I^B$	AB
$I^O I^O$	O

Genotypes in the *population*	Phenotype in the *population*

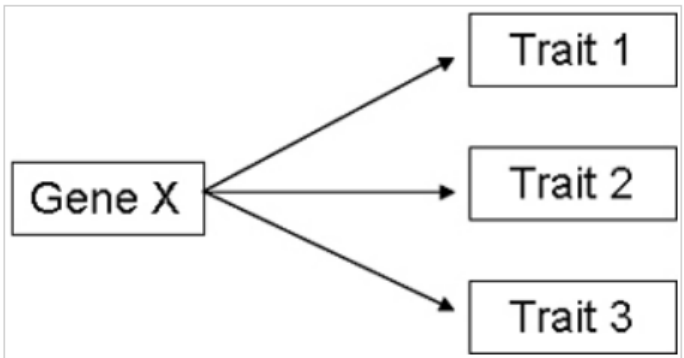
B. Most genes affect multiple traits - Pleiotropy!

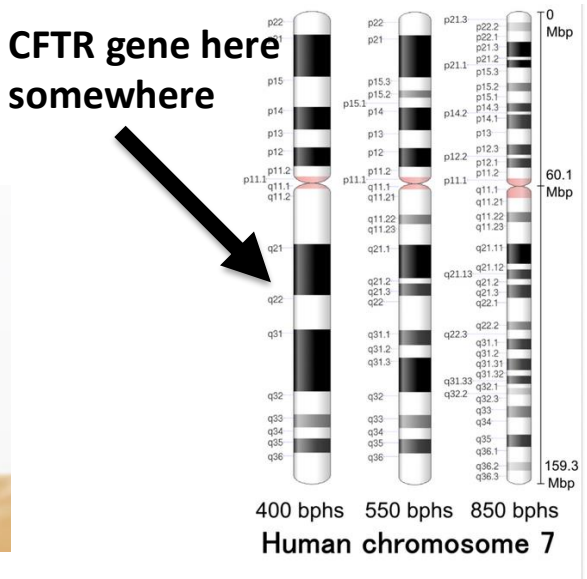
Ex. **Cystic fibrosis** caused by mutations in **CFTR gene** (Which may have over 1900 alleles or different mutations!) affects lungs (get infections) but also affects pancreas and digestive system.

(Cool Fact: This mutation may have been selected for in past because may provide resistance to typhoid!)

https://embryo.asu.edu/pages/cystic-fibrosis-transmembrane-conductance-regulator-cftr-gene

Ex. Fruit flies ***vg* gene** affects wings and the position of bristles on one part of a fly, and decreases the length of a fly's life!





Wikipedia

B. Most genes affect multiple traits - Pleiotropy!

Mutations you looked at this week!

Dmpy
"Mutations in the Caenorhabditis elegans *dpy-13* (dumpy) gene result in a short, chunky body shape."

"dpy-13(e184), carries a small deletion near the middle of this gene. The DNA sequence reveals that ***dpy-13* is a member of the collagen multi-gene family**, and it could encode a polypeptide of 302 amino acids."

In addition to affecting their size and width this mutation also affects their movement, reproduction and metabolism!

Volume 55, Issue 4, 18 November 1988, Pages 567-576

Article

dpy-13: A nematode collagen gene that affects body shape

Nicola von Mende, David Mck. Bird *, Patrice S. Albert, Donald L. Riddle

FYI: Lets look at some of the members of that same collagen gene family in humans! One of those collagen genes (*COL2A1)* is very **pleiotropic**-why?

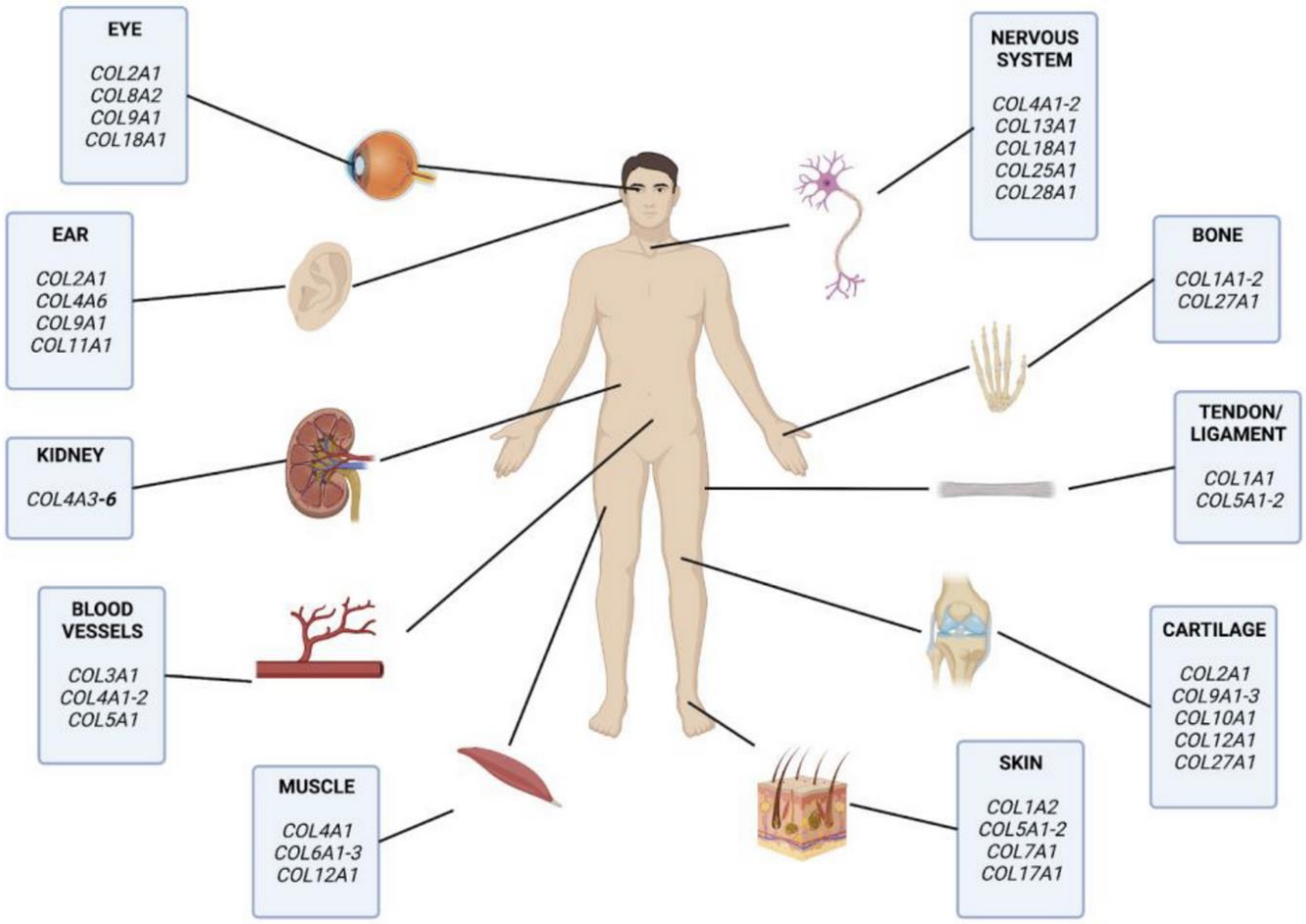


Figure 1. Collagens in Human Tissues. Different types of collagen proteins experience localized expression in various tissue types. For example, *COL2A1* is expressed in tissues with substantial components of cartilaginous fibers.


Article 18 April 2023

The Human Extracellular Matrix Diseasome Reveals Genotype–Phenotype Associations with Clinical Implications for Age-Related Diseases

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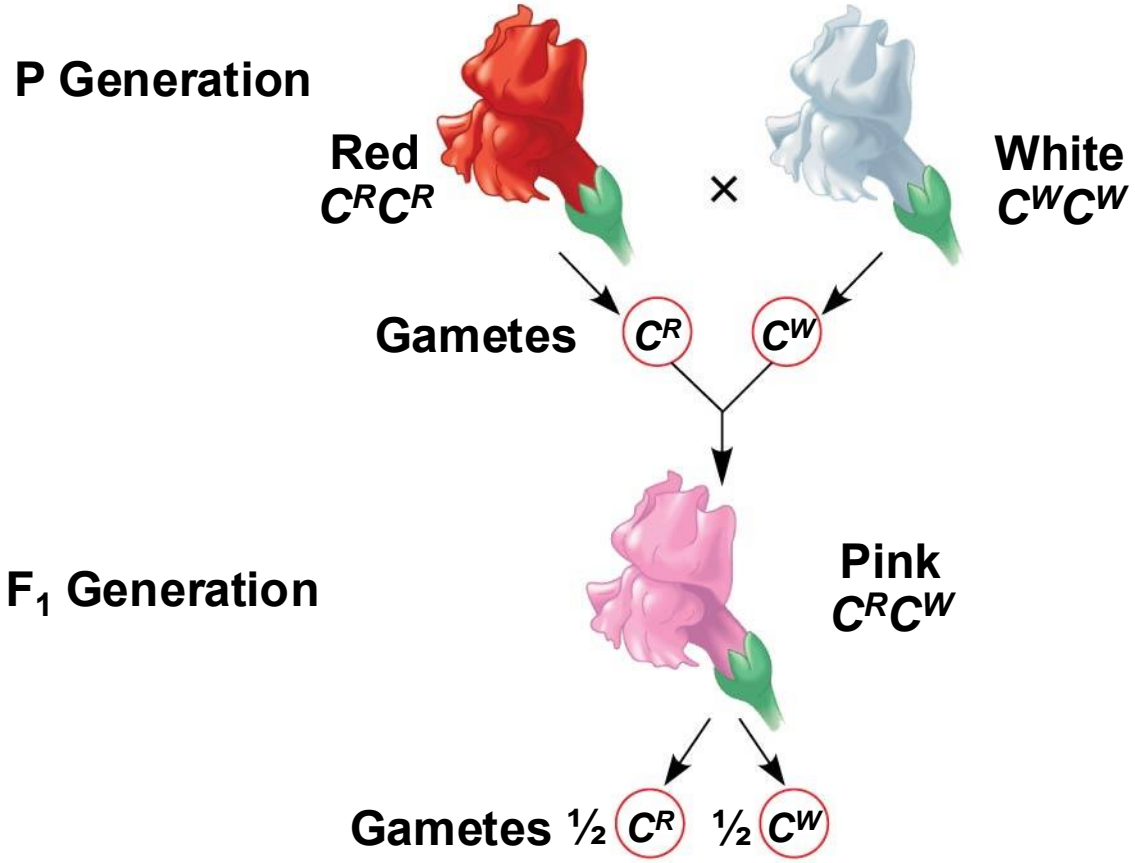
In some cases…two alleles at a particular locus have unusual relationships!

It may **not** be a simple dominant-recessive relationship.



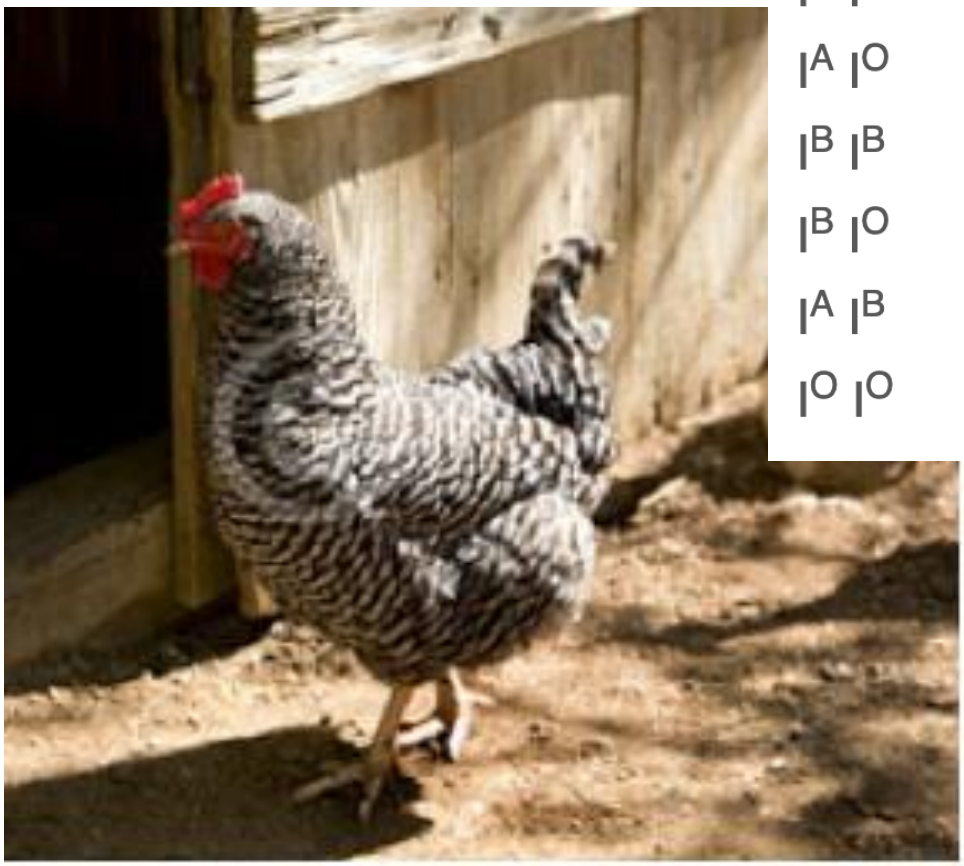
C. Incomplete dominance!

Is this blending inheritance???

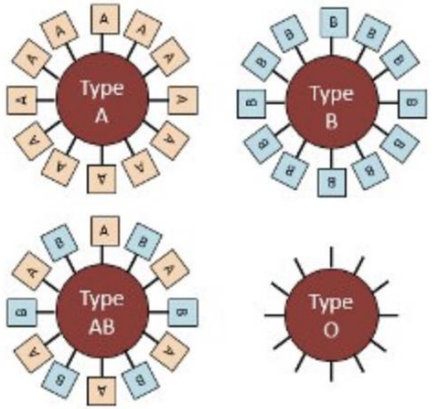
This chicken is heterozygous at a single locus (BW), erminette.

What is going on here? Both alleles are expressed and both traits are “visible”!

D. Codominance



Genotype	Blood Group
$I^A I^A$	A
$I^A I^O$	A
$I^B I^B$	B
$I^B I^O$	B
$I^A I^B$	AB
$I^O I^O$	O

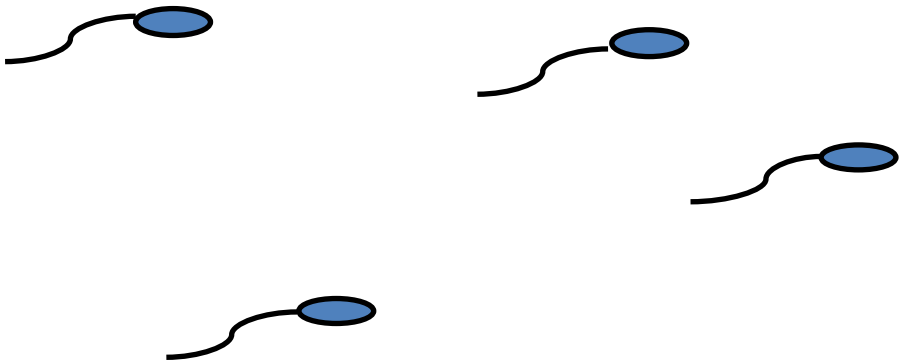


AB blood type is codominant because the red blood cells have the products of both the A and the B alleles of the ABO gene.

https://genetics.thetech.org/ask-a-geneticist/codominant-traits-people

Plus many genes interact with one another….

Plus many genes interact with one another….



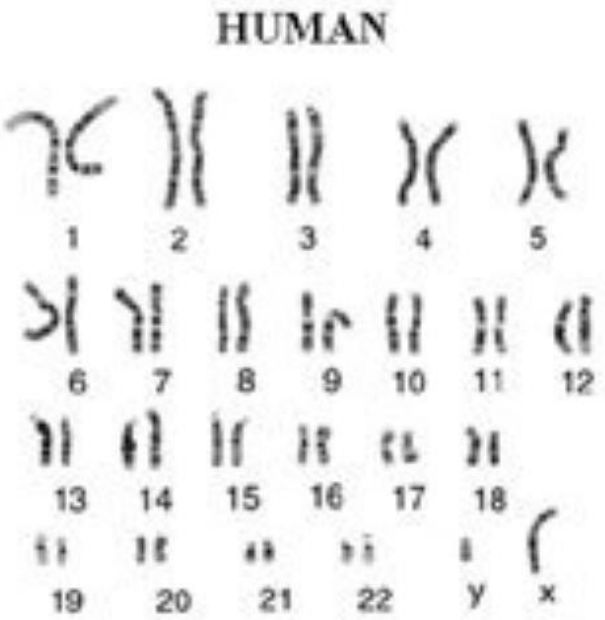
Here are two of the 39 pairs of chromosomes in dogs (#1, #17).

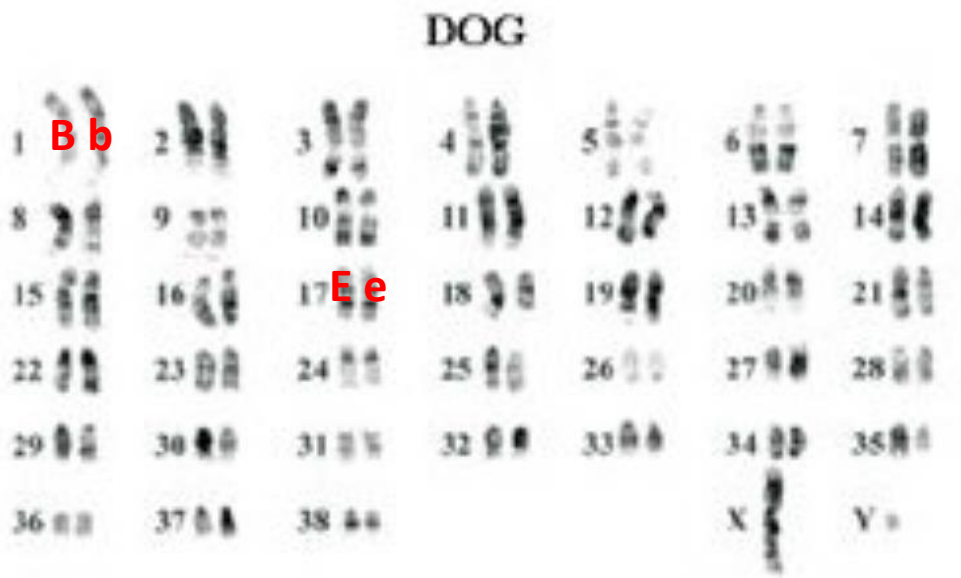
The two genes (the b and e genes) represented are on different chromosomes.

What are the possible genotypes of the gametes (sperm) this dog could produce?

Haploid= N =23 pairs
Diploid= 2N =46

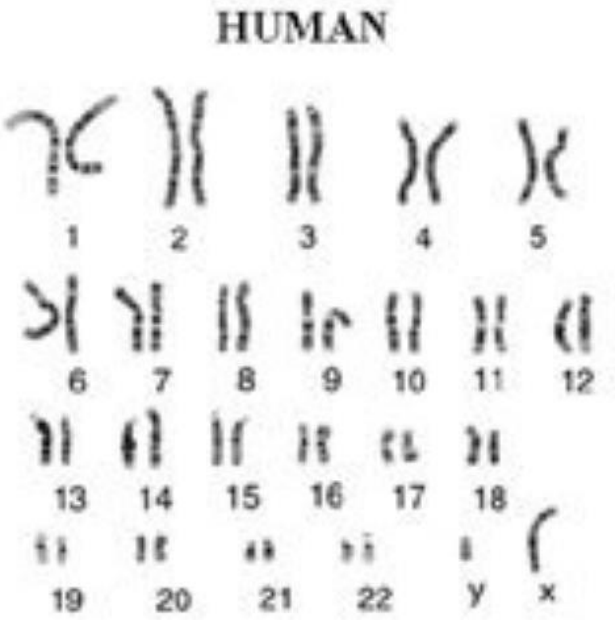
Haploid= N =39 pairs
Diploid= 2N =78

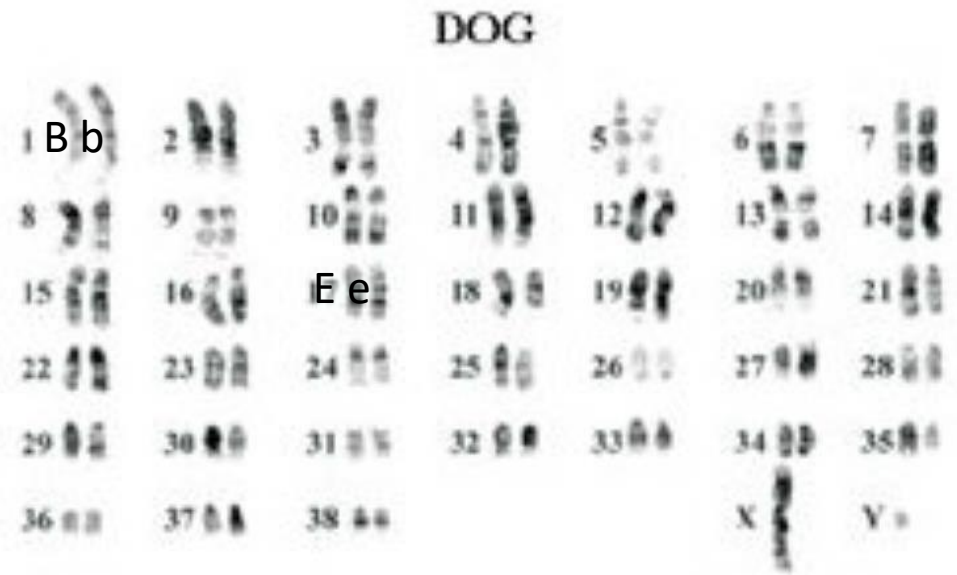




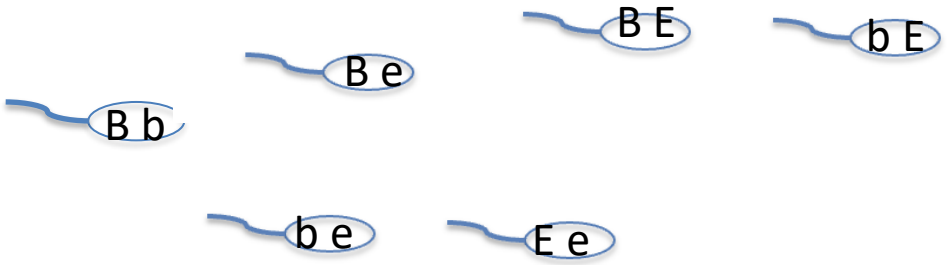
Note these two genes are on two different chromosomes.
What are the possible genotypes of the gametes this dog would produce?

Which of the gametes are not possible???

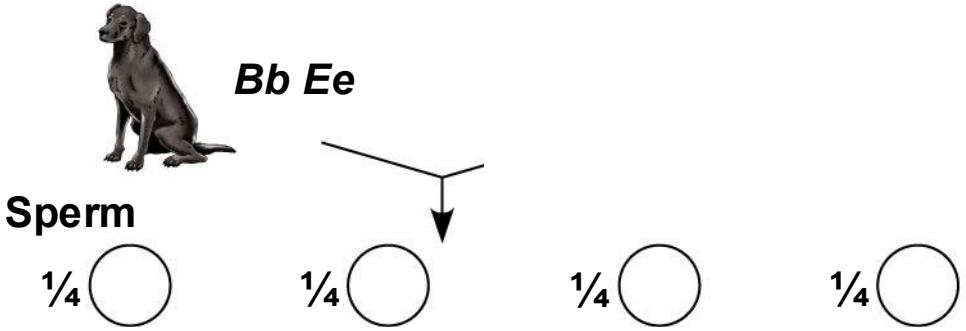




Which of these are NOT possible?!

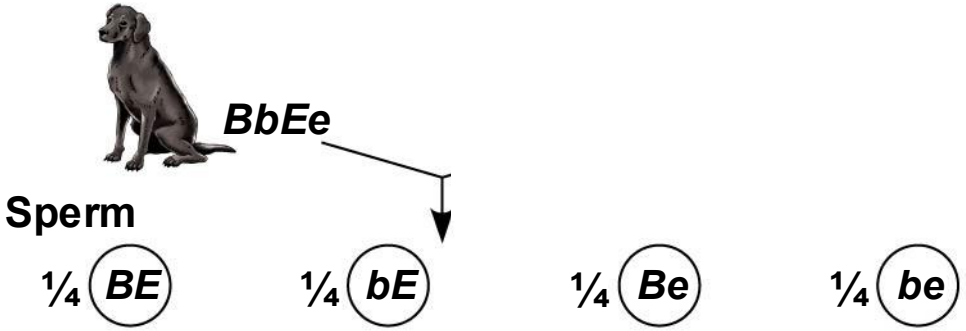


FYI..2 loci on 2 different chromosomes



What would be the possible genotypes of the sperm produced by this male lab?

FYI..2 loci on 2 different chromosomes



BbEe

Sperm

¼ ***BE*** ¼ ***bE*** ¼ ***Be*** ¼ ***be***

Eggs

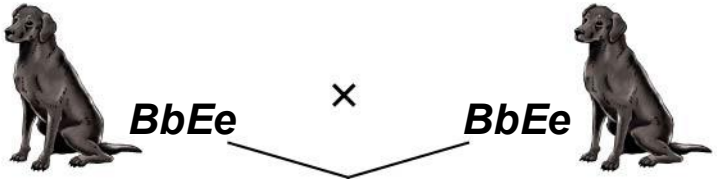
¼ ***BE***

BbEe

¼ ***bE***

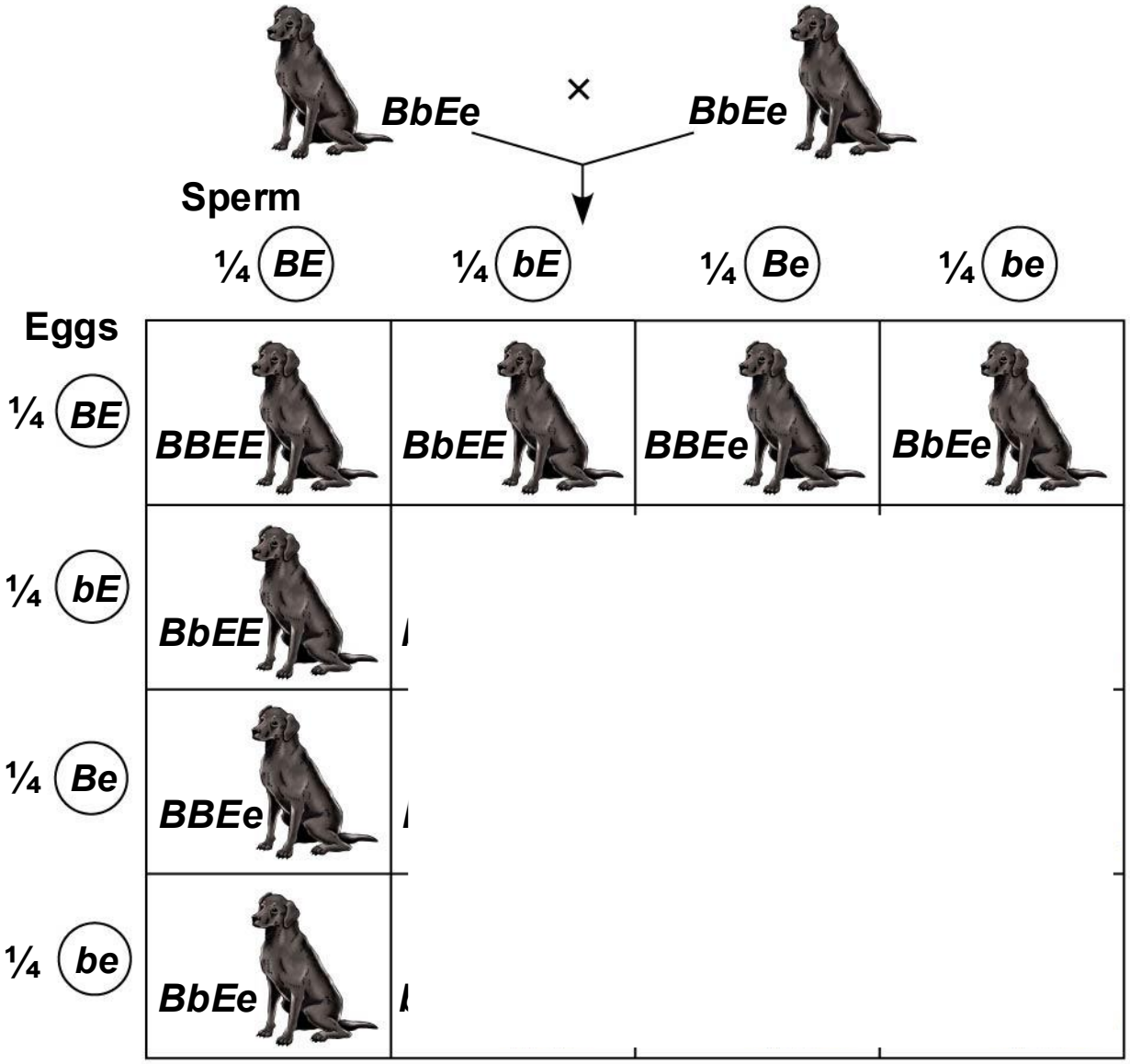
¼ ***Be***

¼ ***be***



Sperm

Eggs	¼ BE	¼ bE	¼ Be	¼ be
¼ BE	BBEE	BbEE	BBEe	BbEe
¼ bE				
¼ Be				
¼ be				



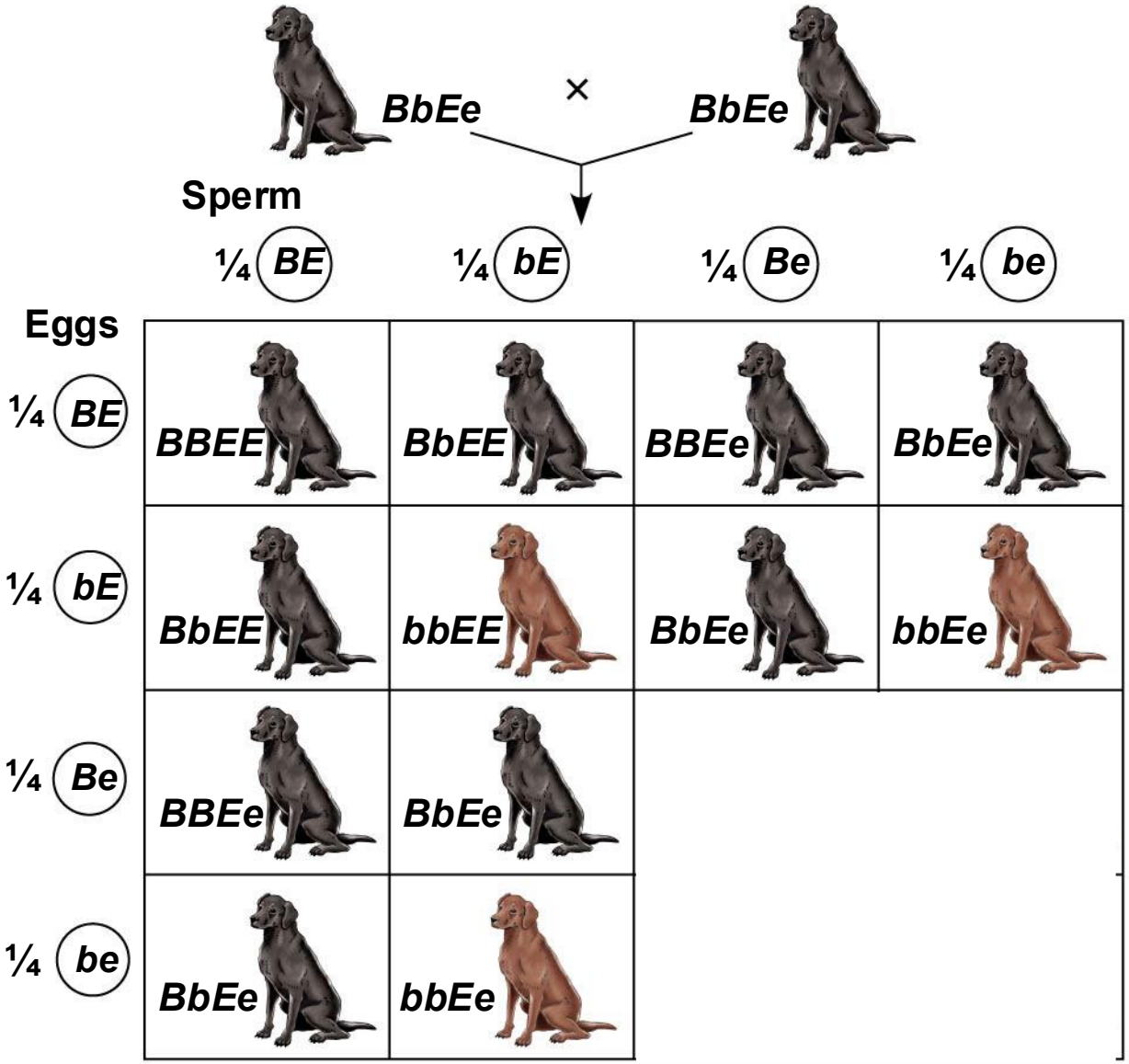
What is the nature of the relationship between B and b?????

What is the nature of the relationship between E and e????

What is going on????

What is the relationship between the two different genes or loci?

Brown lab when homozygous recessive (bb) and either (EE) or (Ee).

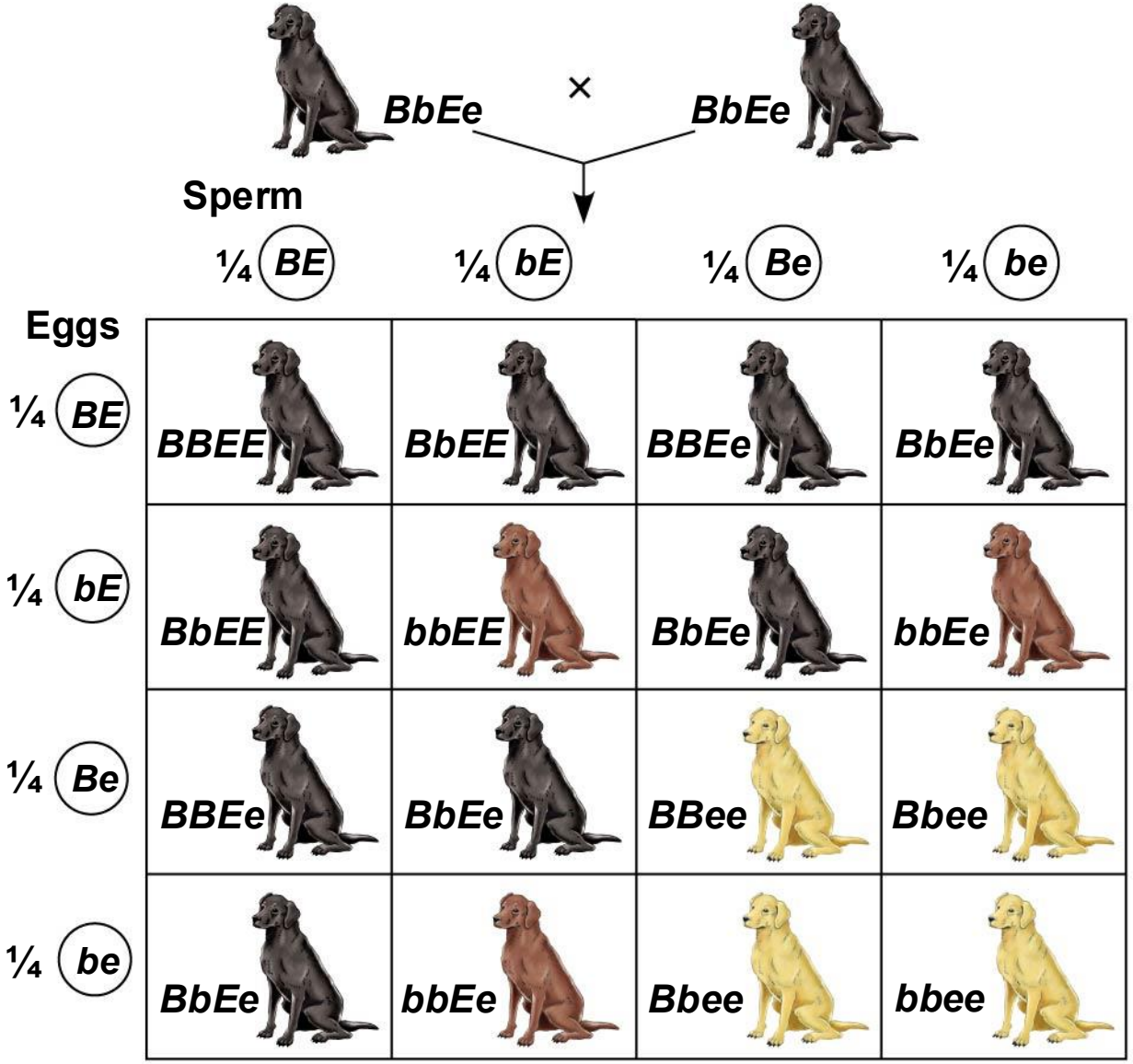


Get yellow lab whenever ee!

(ee shuts down color producing gene so doesn't matter whether it would have been black of brown)

What is this called?

E. Epistasis



https://healthtopics.vetmed.ucdavis.edu/health-topics/canine/inheritance-coat-color-labrador-retriever

F. Traits are usually determined by many more than one or even two loci!

(This is why everyone had trouble studying inheritance in the late 1800's)

With 2 loci (like in the dogs ***BbEe***) what is maximum number of genetically different gametes you could have?

4 different gametes….(BE, Be, bE, be)

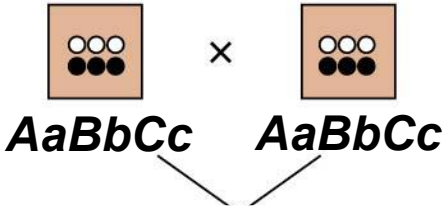
Imagine 3 loci or more!

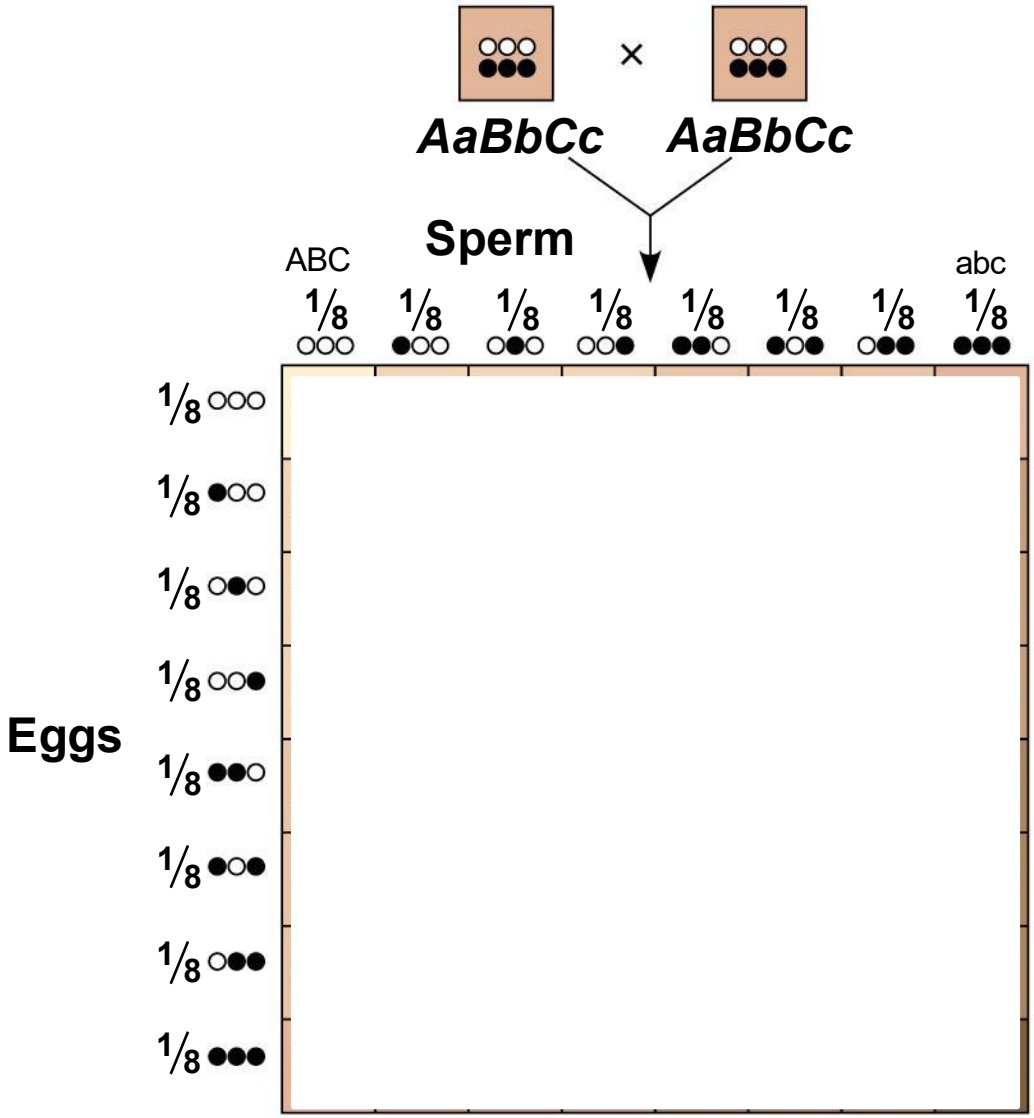
How many genetically different gametes are possible with 3 loci all on separate chromosomes? AaBbCc

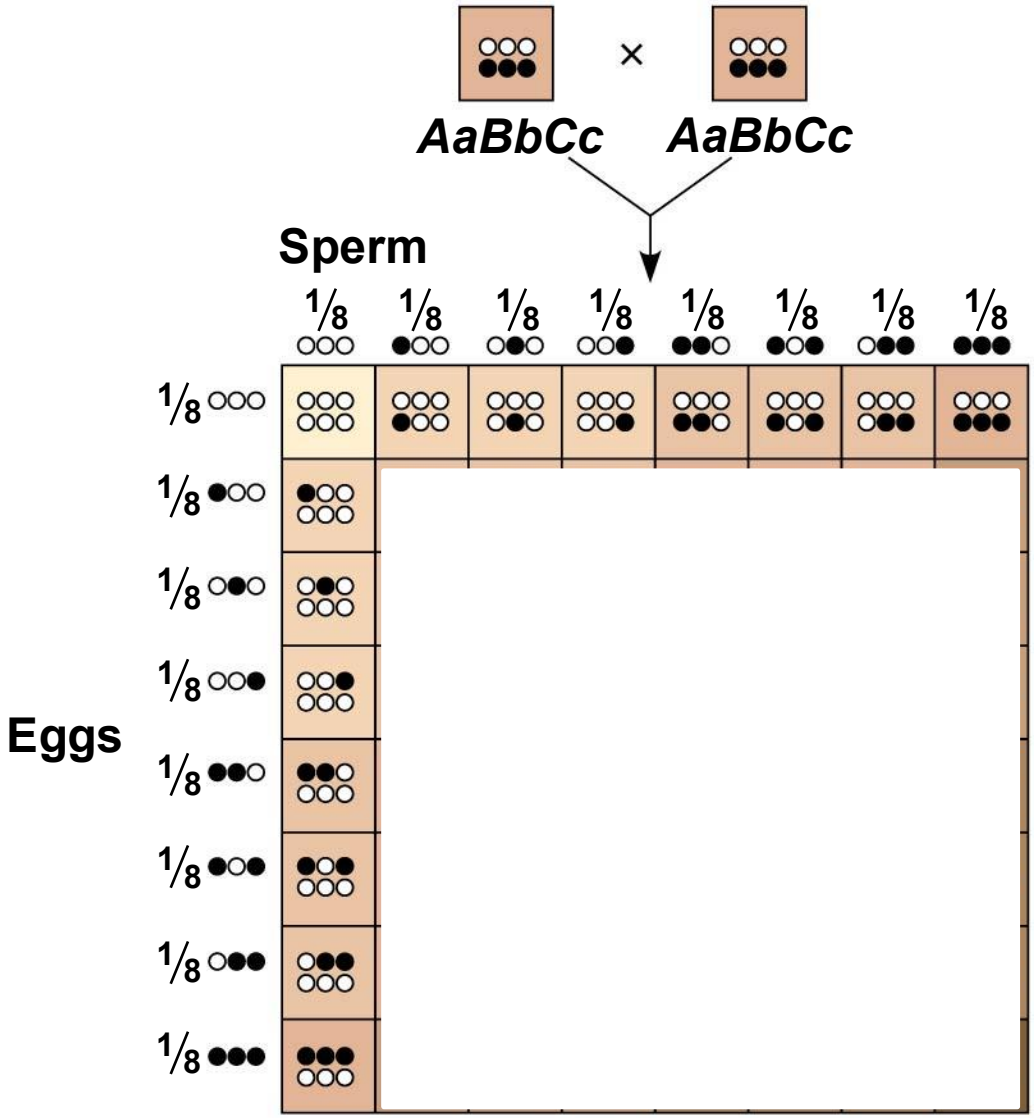
ABC Abc ABc aBC abC AbC aBc abc

What will your Punnett square look like!!

Ex. Skin color is **Polygenic** (another term for quantitative, multilocus, continuous traits)




AaBbCc
×
AaBbCc
Sperm
ABC
abc
1/8
Eggs


AaBbCc
×
AaBbCc
Sperm
Eggs
1/8
1/8
1/8
1/8
1/8
1/8
1/8
1/8
1/8
1/8
1/8
1/8
1/8
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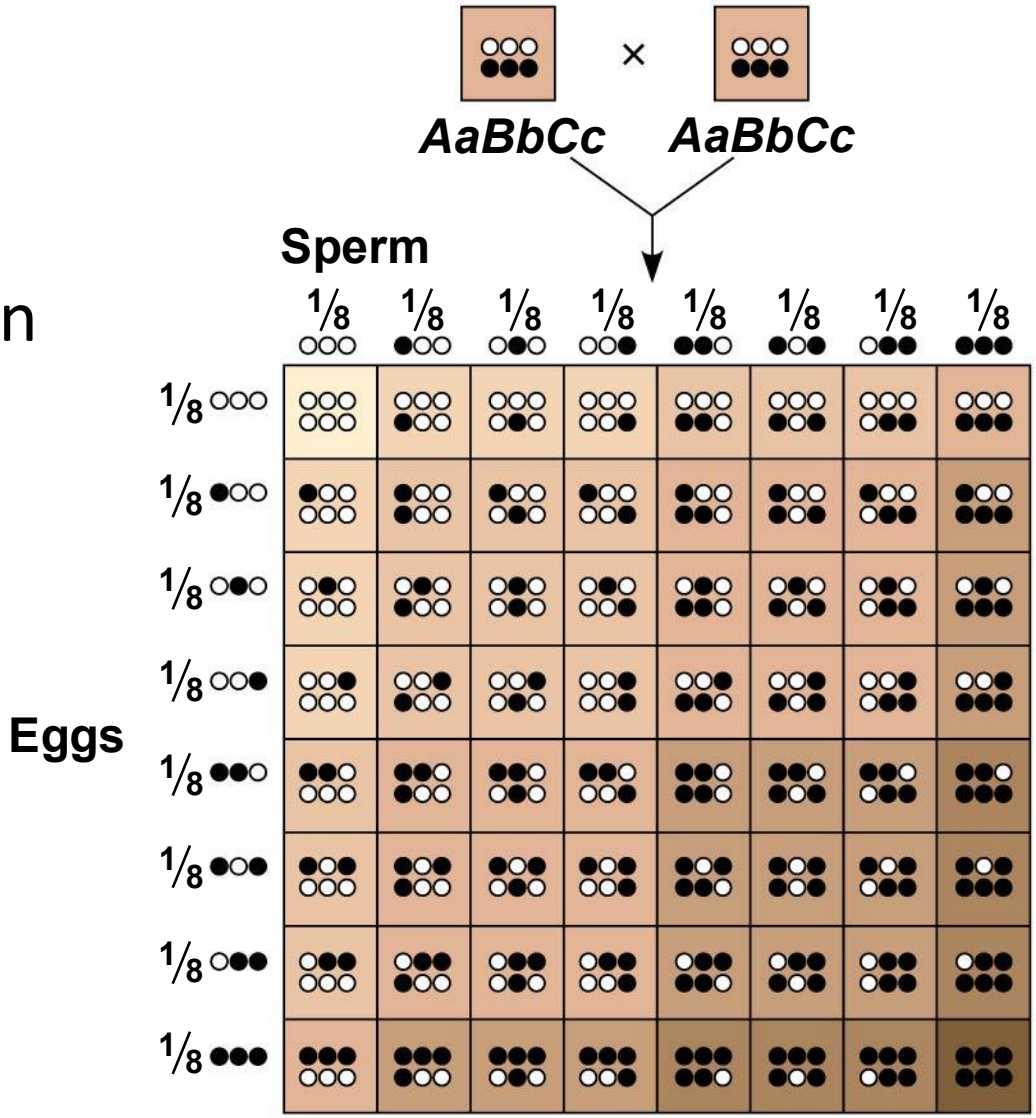
1/8
1/8

Do you think having 3 loci is realistic for skin color?



*"Human skin color is a polygenic trait, meaning multiple gene loci are involved in its expression. At last count, the International Federation of Pigment Cell Societies…has determined that there are a total of **378 genetic loci** involved in determining skin color in humans and mice."*

http://www.gbhealthwatch.com/Trait-Skin-Color.php

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The next (26th) International Pigment Cell Conference (IPCC2026) will be organized by the ASPCR and will be held in New Delhi (India) on **April 30 to May 3, 2026**, at the Hotel The Ashok & Samrat Complex.

Remember how we looked at the heritability of height?

*"The researchers found **12,111 genetic** variants (or regions of the genome) associated with human height, which clustered into small genomic segments that cover 21% of the genome. This confirms earlier predictions[9] that a large proportion of the human genome is involved in shaping height."*

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NEWS AND VIEWS | 12 October 2022

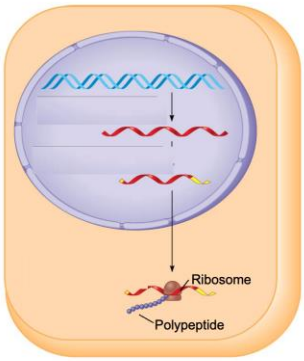
Missing heritability found for height

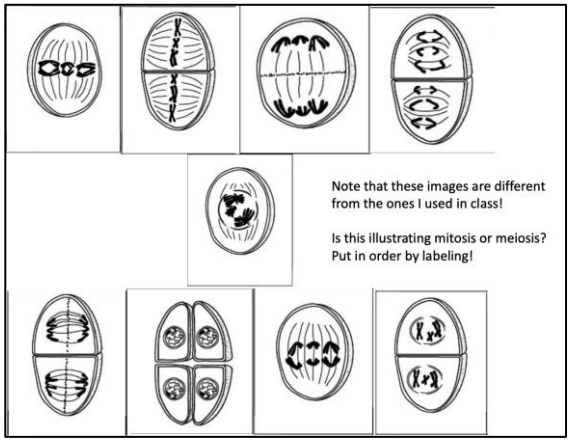
A combined analysis of 281 genome-wide association studies finds 12,111 common DNA variants associated with a person's height – and shows that larger studies will not yield more variants in populations of European ancestry.

By Karoline Kuchenbaecker

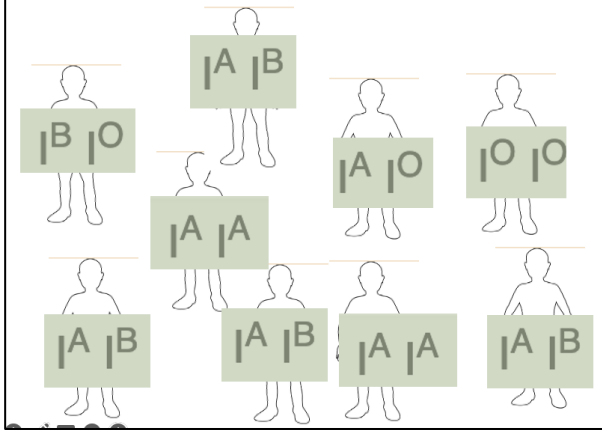
Map the following processes onto the image. Where and when does each take place and what does each do?

- epigenetic modifications
- transcription factors
- post transcriptional modification (RNA processing, RNA splicing)
- microRNA
- post translational modification

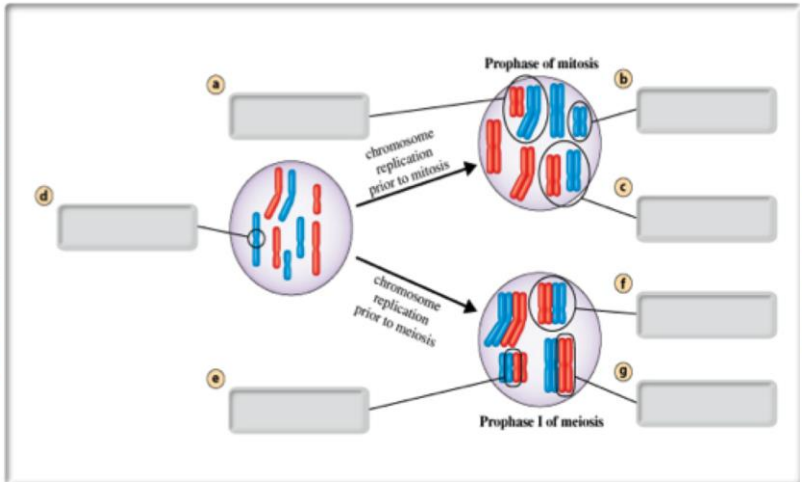




- How many alleles are there in this population at this blood group locus?
- How many possible genotypes are there given the number of alleles in the population?
- Describe the relationship between the A and the O allele when an individual's genotype is AO?
- Describe the relationship between the A and the B allele when an individual's genotype is AB?
- Cross an I^AI^O individual with a I^BI^O individual using a Punnett square and show your work. What would the genotypes and phenotypes be in the next generation?



centromere
homologous chromosomes
non-homologous chromosomes
sister chromatids
nonsister chromatids



genes
locus
traits
genome
gametes
chromosomes
chromatin