Molecular and Cellular Biology (MCB)

BB101

LECTURE-4

16/ 1/ 2018

Genetics

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Outline

1. Mendel and Concept of Inheritance
2. Law of Segregation
3. Law of Independent Assortment
4. Mendelian Genetics Examples

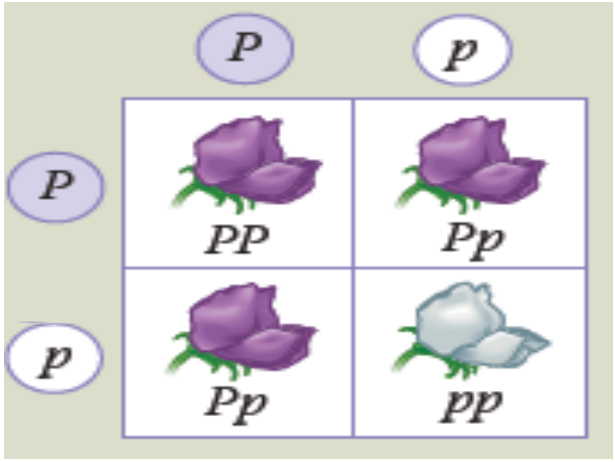
Few Basics and Terminology

Genotype & Phenotype

Genotype: An organisms full hereditary information

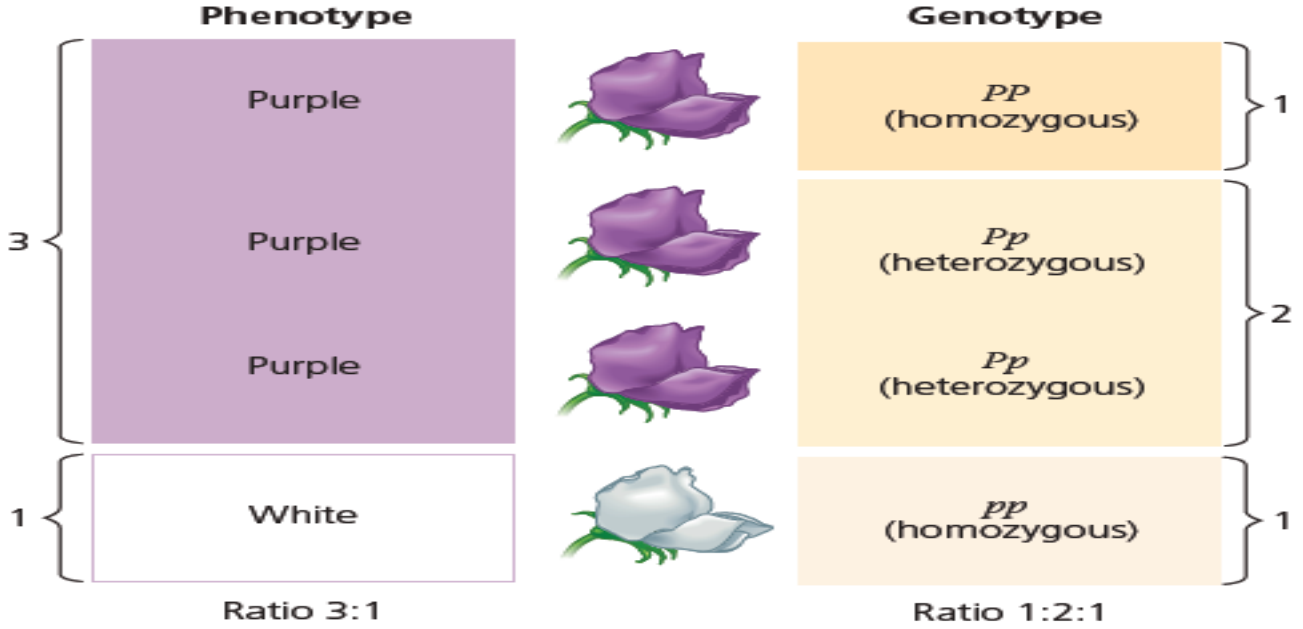
Phenotype: Actual observed properties

Genotype vs. Phenotype



Phenotype ratio?

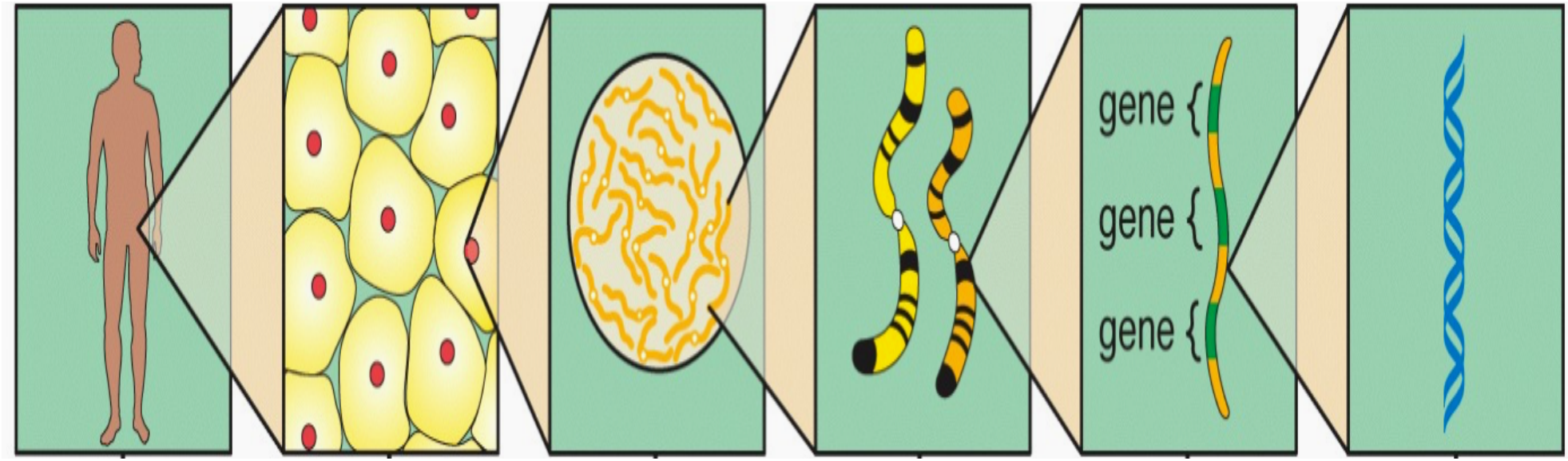
Genotype ratio?



Different alleles: Heterozygous; Identical alleles: Homozygous

An Overview of Gene & Alleles

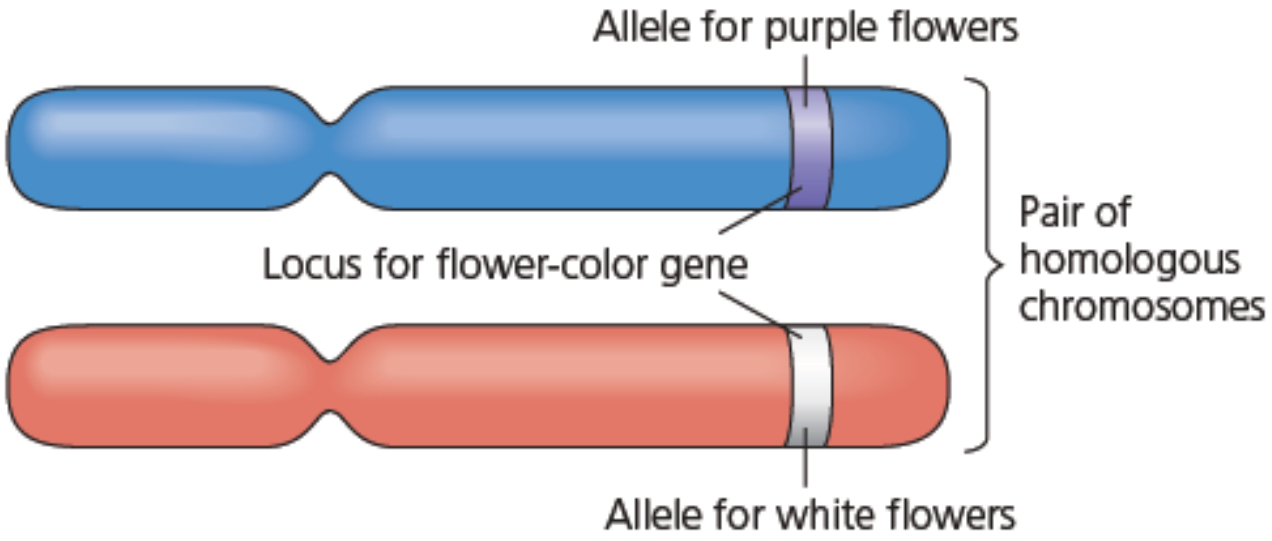
DNA double helix



- Gene - sequence of nucleotides at a specific place, or locus, along a particular chromosome = Mendel's "heritable factor"
- Alleles are different forms of a gene inherited from both the parents
  - Each organism harbors two copies of a gene

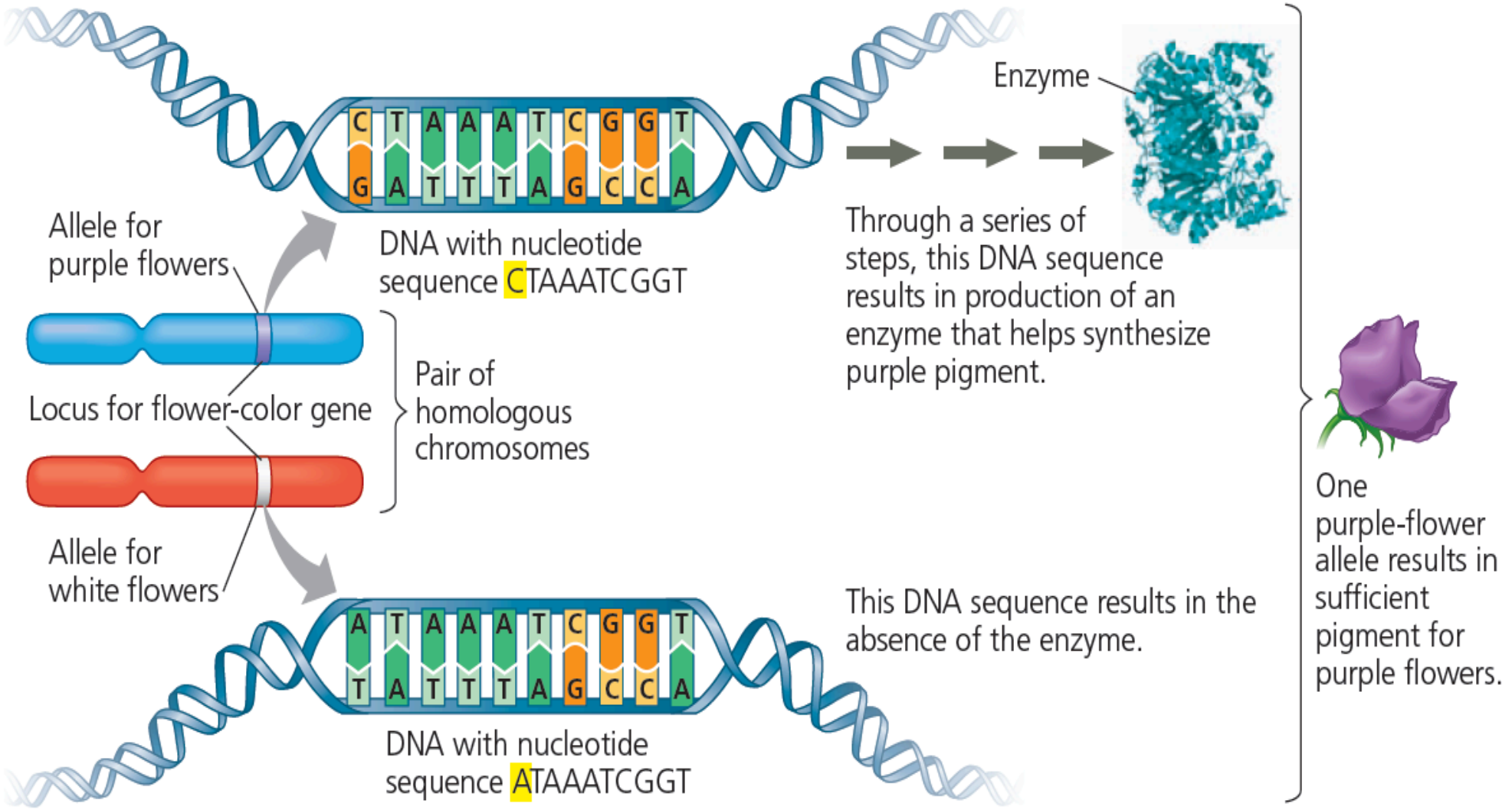
An Overview of Gene & Alleles

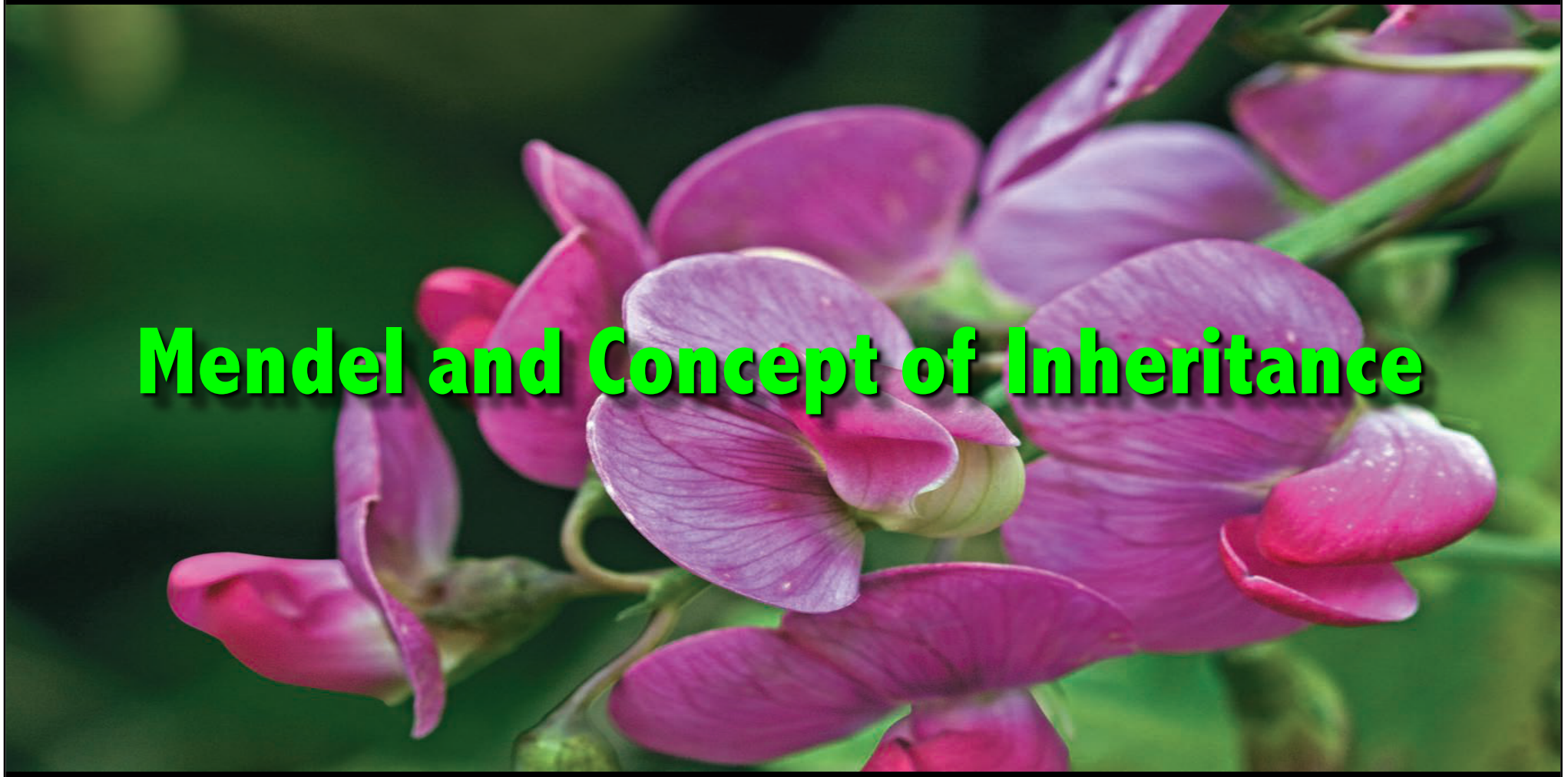
- Alternative variations of genes account for variation ---- called alleles.



- Each organism harbors 2 copies of a gene
- Dominant allele has the effect but recessive has not

An Overview of Gene & Alleles





Mendel and Concept of Inheritance

Mendel discovered basic principles of heredity by breeding garden peas

About Mendel

- A monk named Gregor Mendel documented a particulate mechanism for inheritance using pea plants
- In 1843, at the age of 21, Mendel entered an Augustinian Monastery
- In 1851, he left the monastery to pursue two years of study in physics and chemistry at the University of Vienna
- After attending the university, Mendel returned to monastery and started teaching at a local school

About Mendel & his Experiments

1857, Mendel began breeding garden peas

- Chose only true breeding varieties

Mendel & his Experiments

1857, Mendel began breeding garden peas

• Around 1860, Gregor Mendel provided the laws of inheritance to define the genetic principles

Flow of Heritable Traits

- *What are the Heritable traits?*
- Phenotypic characters that passes from parent to off spring e.g. eye and hair color
- *What are the genetic principles that account for the transmission of such traits?*
- Hereditary rules deciphered by Mendel

Heritable Variations

Natural Variations

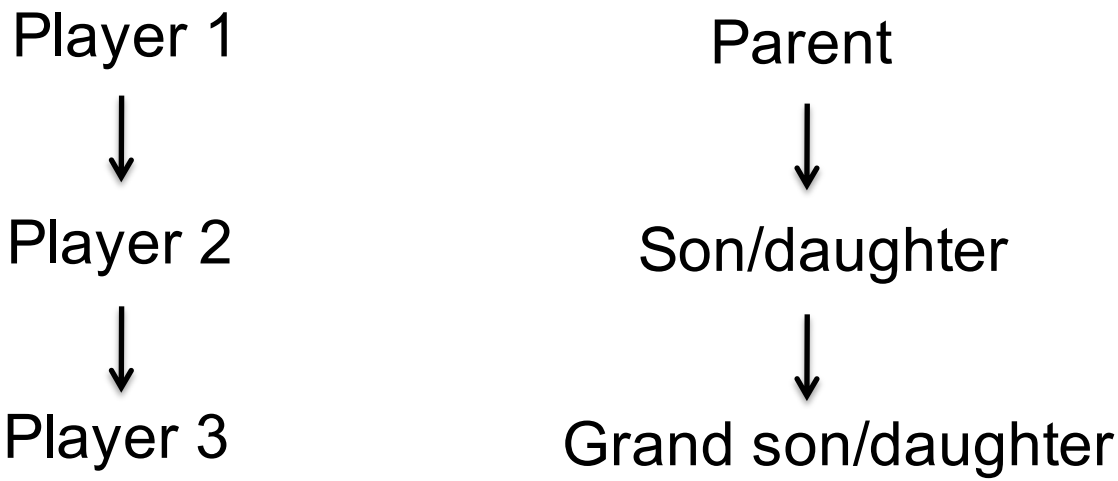
Eyes - Brown, Blue, Green or Gray

Hair - Black, Brown, Blond or Red

Image - Wikipedia

How to Define Heredity?

- Blending hypothesis - genetic material contributed by the two parents mixes just as blue and yellow paints blend to make green.
- Particulate hypothesis ---- leads to idea of gene
- Collection of genes is like deck of cards



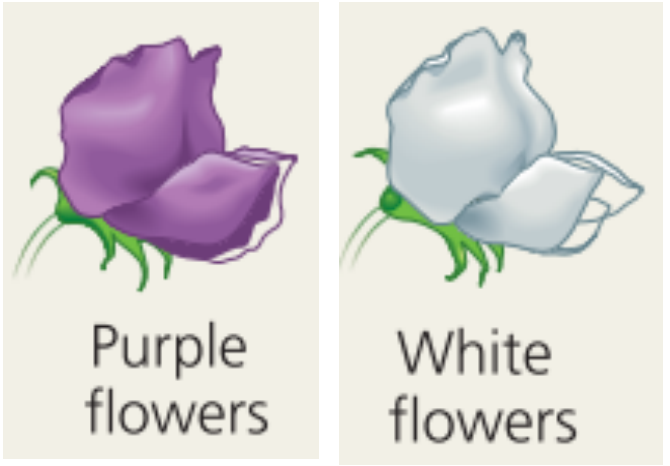
- Genes can be shuffled & passed along, generation after generation

Mendel: Elucidation of Principle of Heredity

- Mendel's choice of experimental system -- pea plants

Why pea plants?

1. Availability in many variations
2. Short generation time
3. Large number of offspring from each mating
4. Easier crossing due to well separated pollen producing and egg bearing organs



Parameters that Mendel made sure

- Chose only true breeding varieties

Purple-flowered plant X Purple-flowered plant

Always purple-flowered plant

Parameters that Mendel Studied

- Chose only those characters showing distinct alternative forms

Character	Dominant Trait	×	Recessive Trait
Flower color	Purple 	×	White 
Flower position	Axial 	×	Terminal 
Seed color	Yellow 	×	Green 
Seed shape	Round 	×	Wrinkled 

Pod shape: Inflated  × Constricted 

Pod color: Green  × Yellow 

Stem length: Tall  × Dwarf 

Character variants are called traits

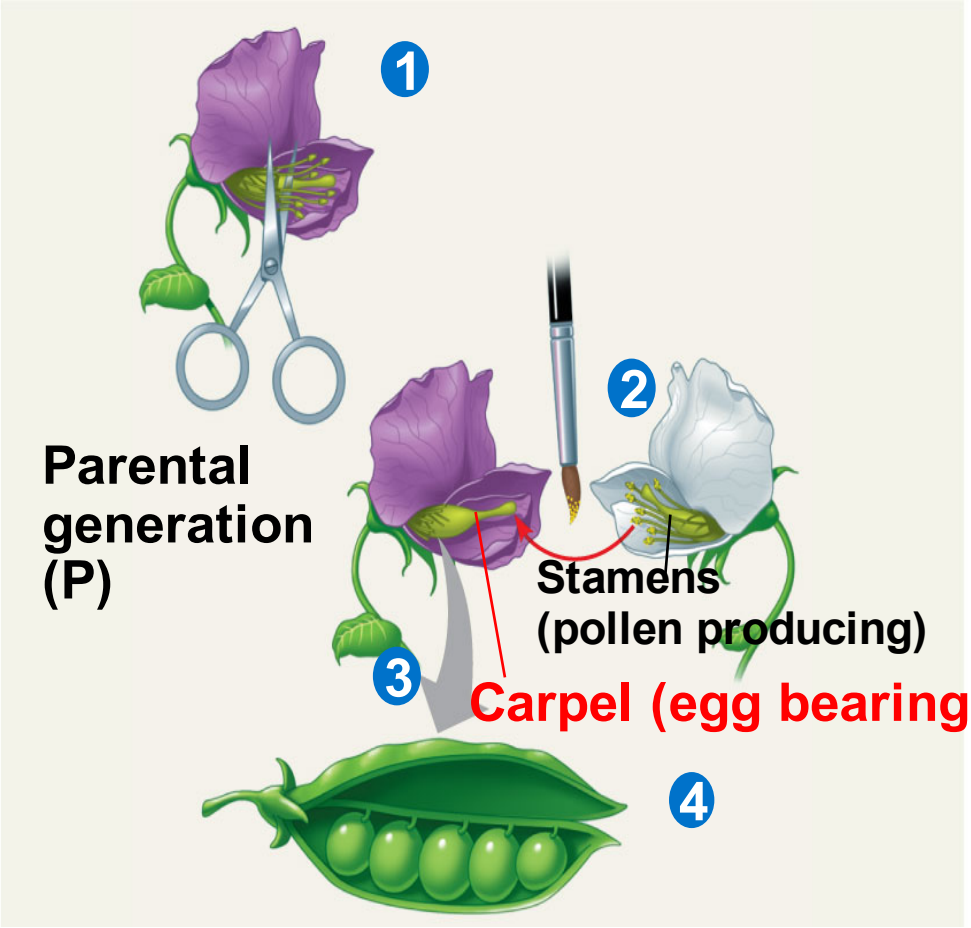
Mendel: Elucidation of Principle of Heredity

1. Inheritance of each trait is determined by "units" or "factors" that are passed on to descendants unchanged

2. Individual inherits one such unit from each parent for each trait
3. A trait may not show up in an individual but can still be passed on to the next generation


Law of Segregation

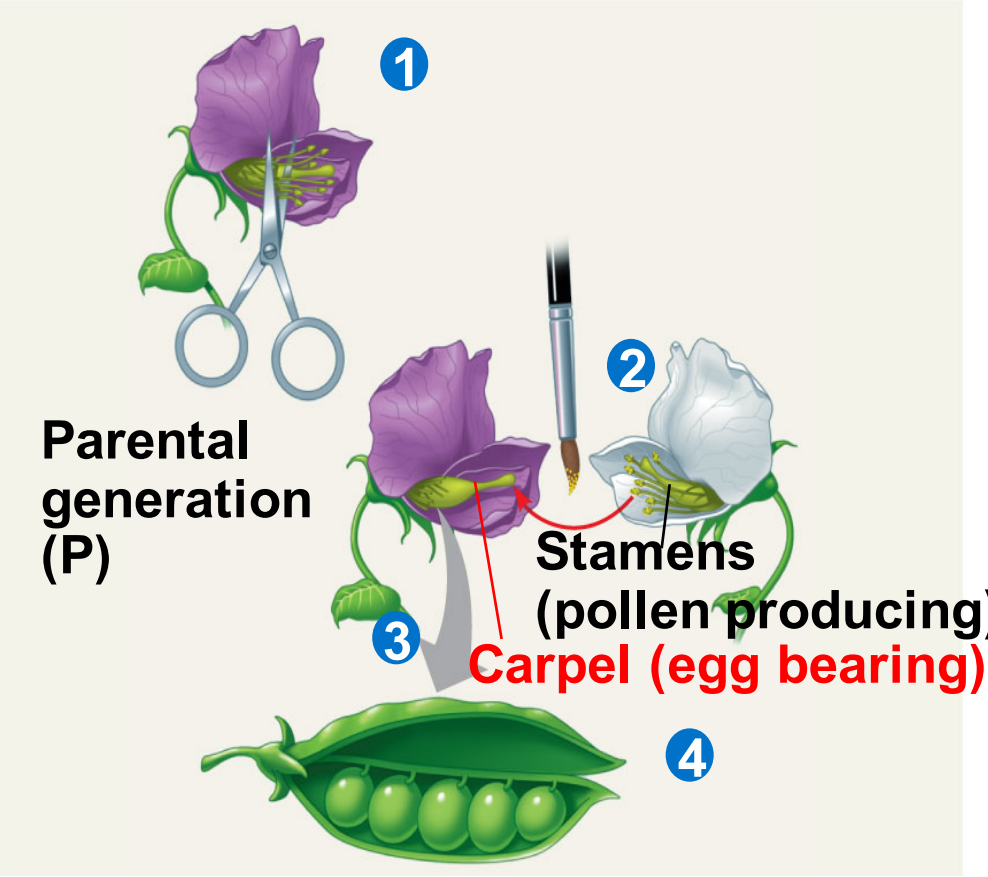
Mendel's Experiments: Crossing (Mating)



A **carpel** is the ovule and seed producing reproductive organ in flowering plants

A **stamen** is the pollen-producing reproductive organ of a flower.

Mendel's Experiments: Crossing (Mating)



- Mendel cross-pollinated two contrasting, true-breeding pea varieties

  e.g. purple-flowered plants and white-flowered plants

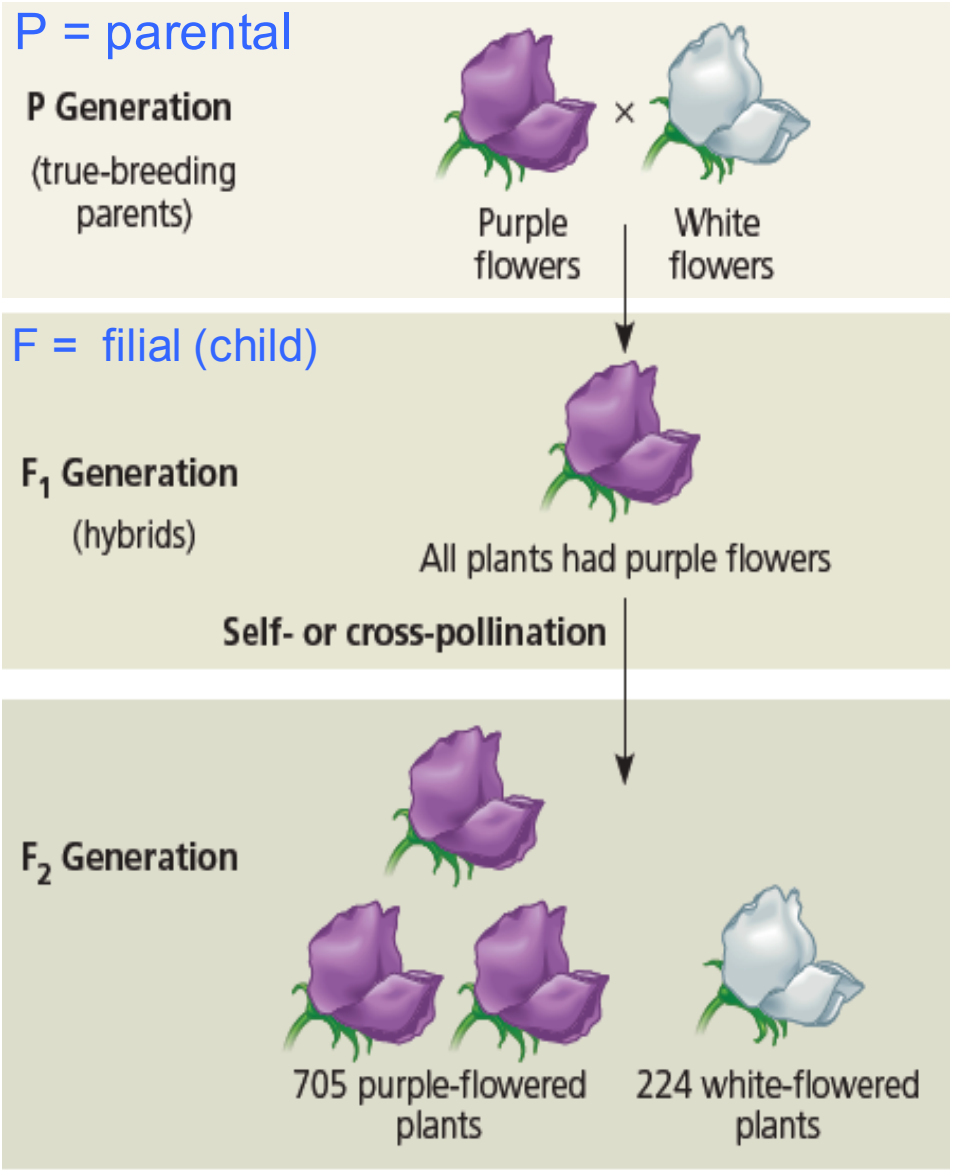
- Hybridization - mating or crossing of two true-breeding varieties

*hybrid offspring are **F1 generation** (first filial generation)*

Results of Crossing Two Contrasting Traits

Dominant trait

Recessive trait



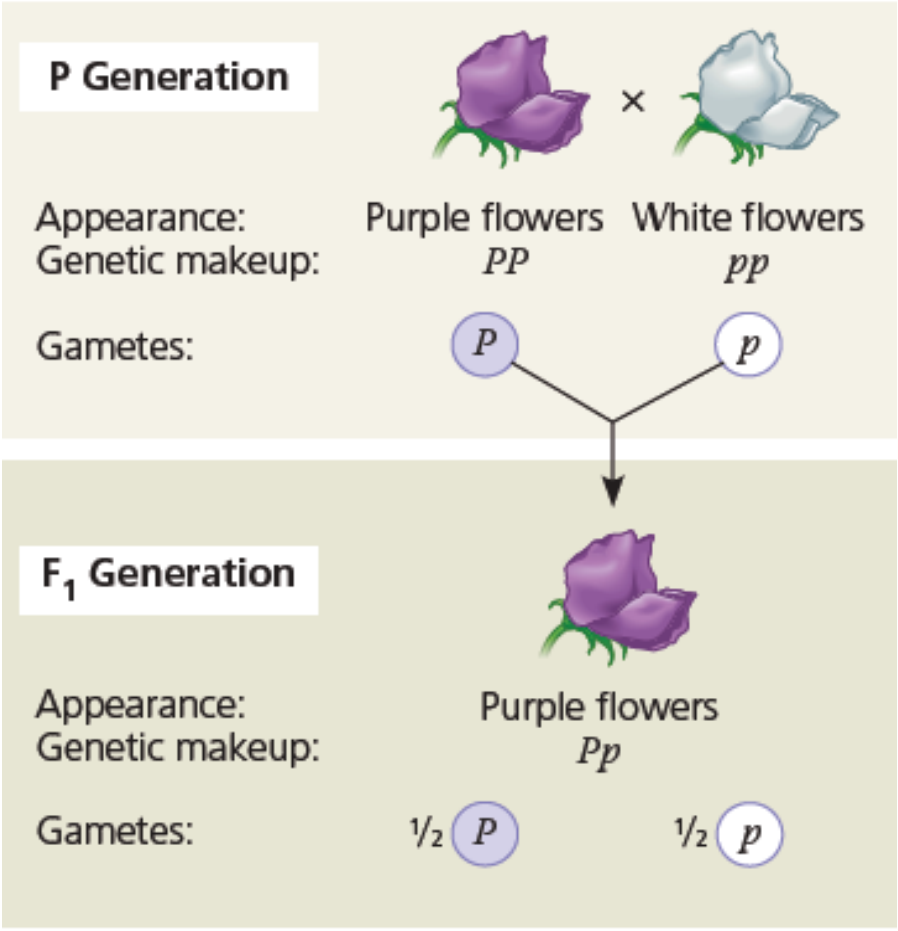
F2 generation

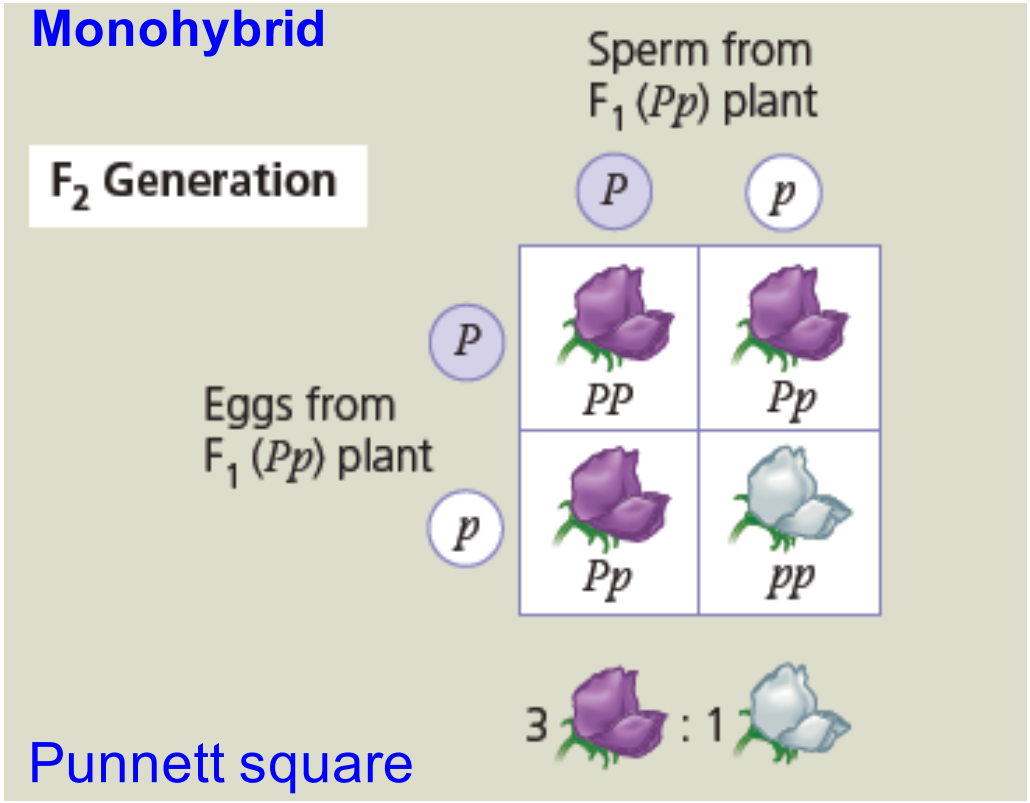
- F1 hybrids to self-pollinate or cross-pollinate with other F1 hybrids

Dominant vs. Recessive trait 3:1

Law of Segregation

- Two alleles for a heritable traits separate from each other during gamete formation and form different gametes
  - *Alternative versions of a gene are called "allele"*





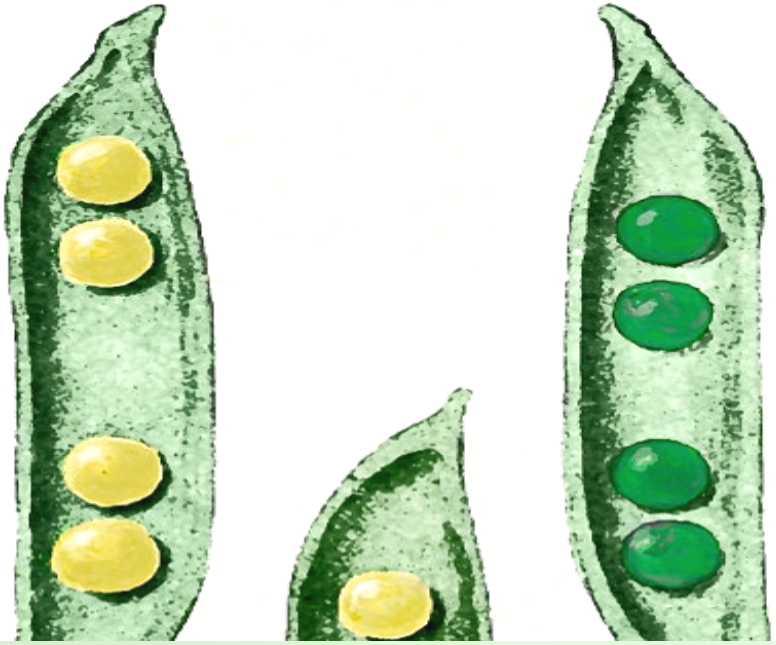
F_2 plants – cross between 2 monohybrids

Law of Segregation

Character	Dominant Trait	×	Recessive Trait	F_2 Generation Dominant: Recessive	Ratio
Flower color	Purple 	×	White 	705:224	3.15:1
Flower position	Axial 	×	Terminal 	651:207	3.14:1

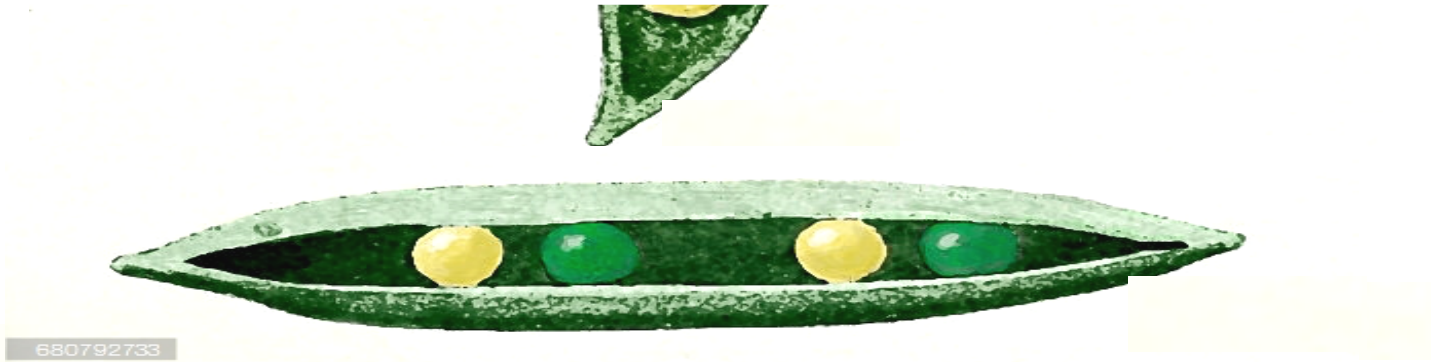
Observation of F_2 plants

The ratio of dominant vs recessive trait is 3:1



How to determine an organism's genotype?

An organism that exhibits a dominant trait, can be either homozygous for the dominant allele or heterozygous

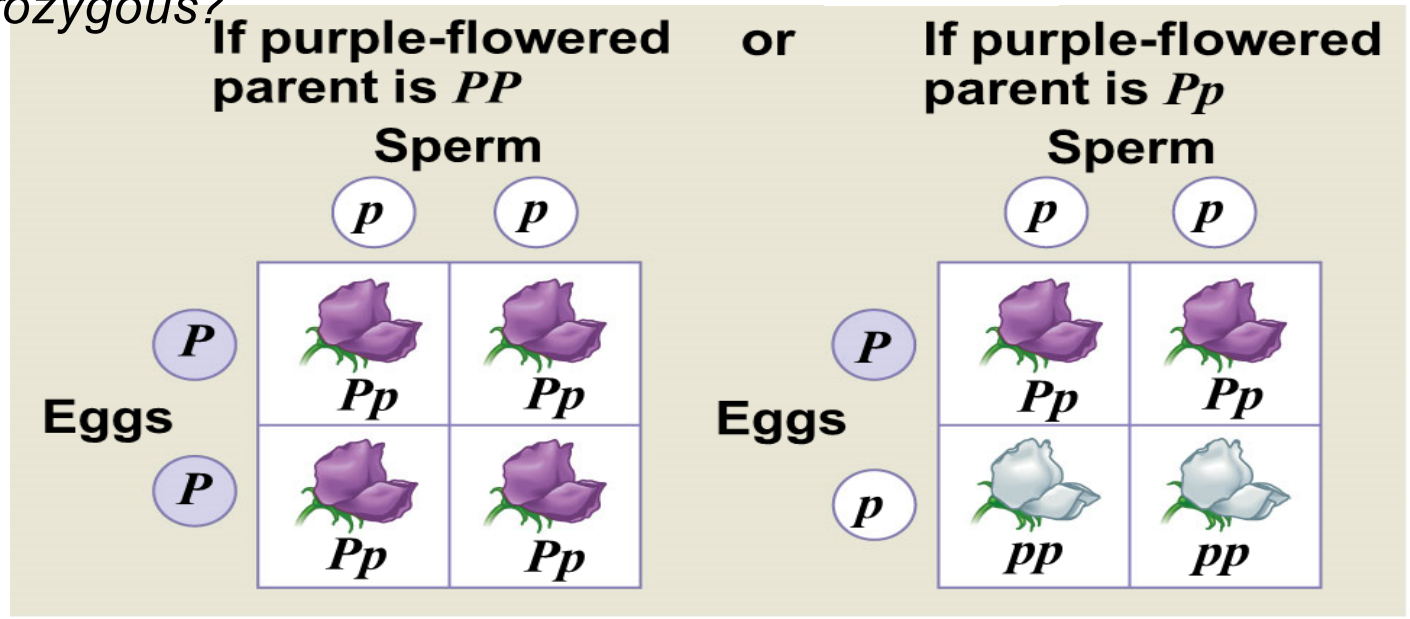


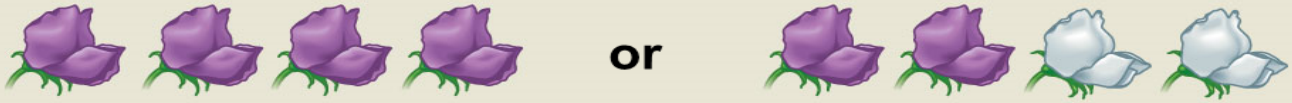
TEST CROSS: Determine Organism's Genotype

PP or Pp
Homozygous or Heterozygous?

X

pp
Homozygous





All offspring purple

or

$^1/_2$ offspring purple and $^1/_2$ offspring white

Breeding an organism of unknown genotype with a recessive homozygote "test cross" because it can reveal genotype of that organism

Conclusions: Section-I

1. The Law of Segregation suggests that genes have alternative forms or alleles.

2. Two alleles for a heritable traits separate from each other during gamete formation.

3. This law explains that 3:1 ratio of F2 phenotypes is found when monohybrids self-pollinate.

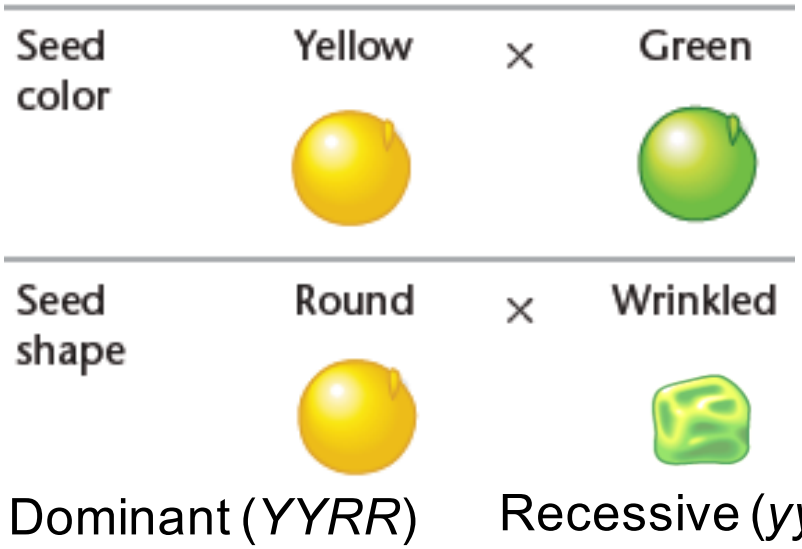
4. A trait may not show up in an individual but can still be passed on to the next generation – TEST CROSS can be used to decipher the genotype.

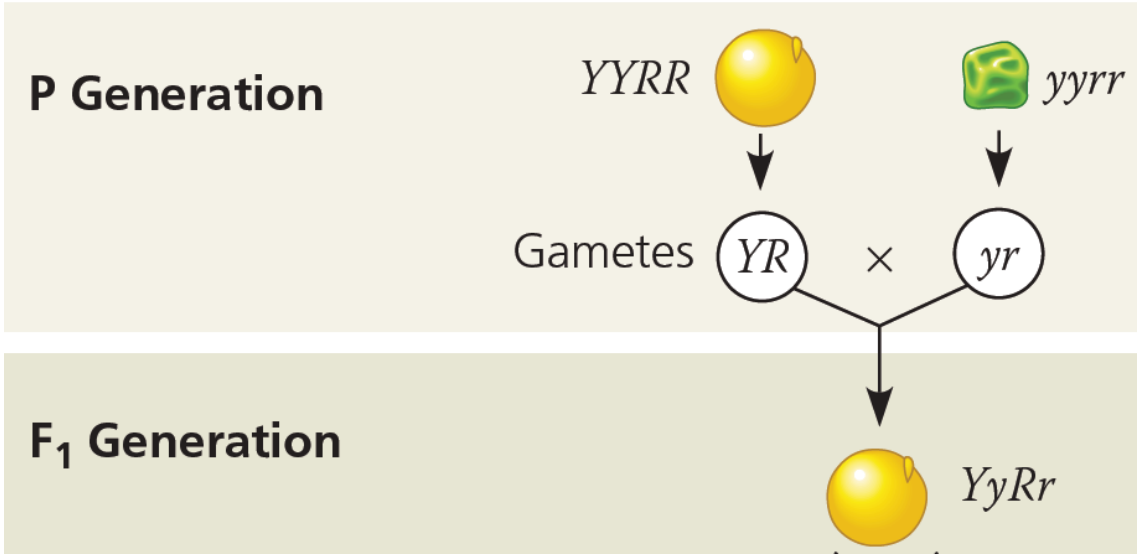
Law of Independent Assortment

Dependent or Independent?

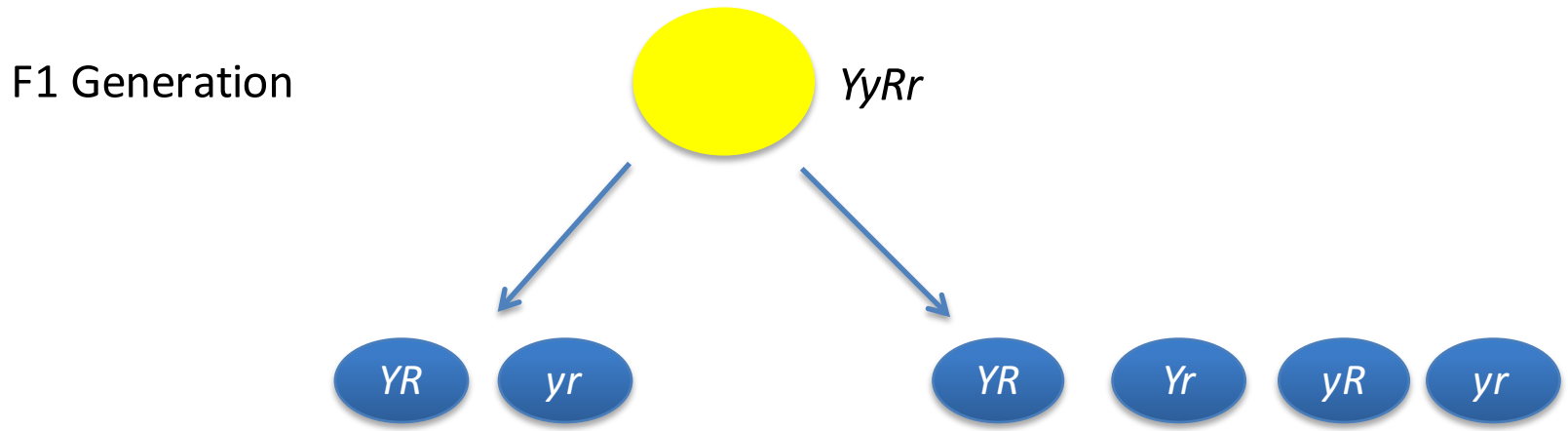
Law of Independent Assortment

- Study of two traits simultaneously – dihybrid cross

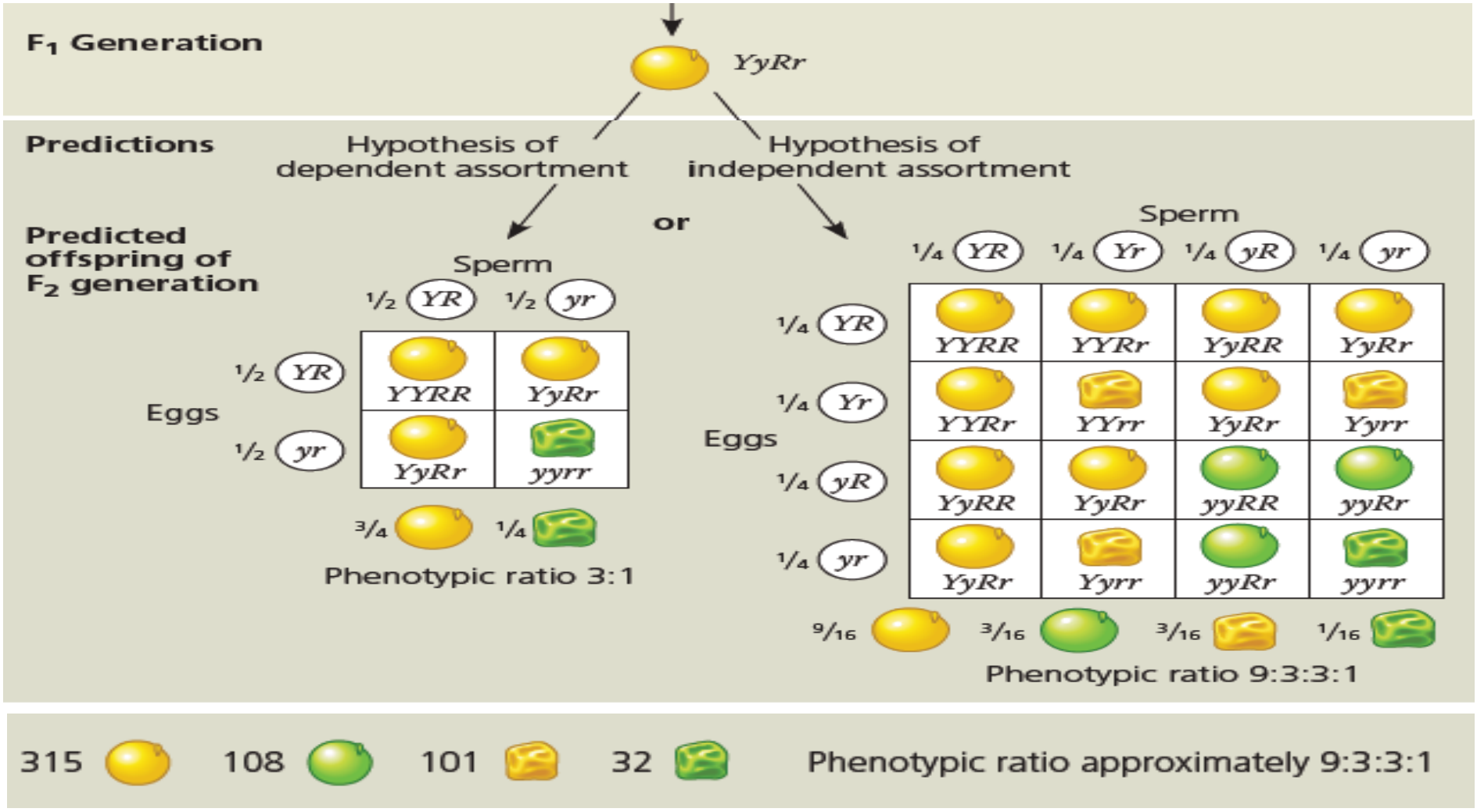




Dihybrid Cross: Dependent or Independent Assortment?



Results of Dihybrid Cross: Law of Independent Assortment



Each pair of alleles segregate independent of other pair of alleles during gamete formation

Conclusions: Section-II

1. Mendel formulated his second law of inheritance by studying two characters at the same time.

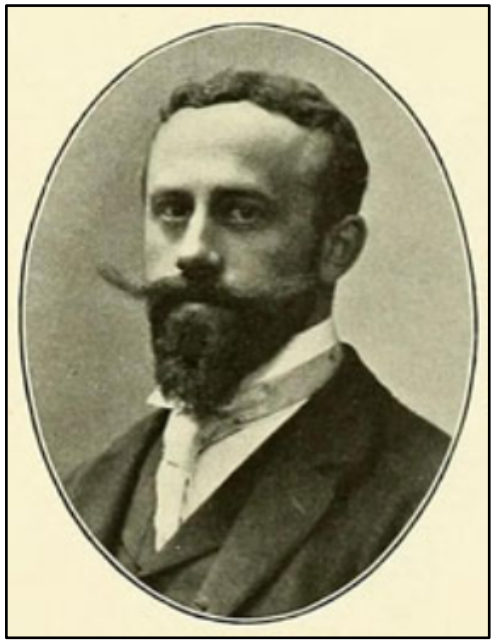
2. He stated that two or more genes assorted independently; each pair of alleles segregate independently during gamete formation.

3. Crossing experiment between dihybrids resulted into offspring having four phenotypes in a 9:3:3:1 ratio.

Mendelism Reconfirmed

- Mendel had originally published his results in 1866; however, his results were unnoticed.
- In 1900 three scientists independently rediscovered Mendel's laws, which was unnoticed by the scientific community.

Hugo de Vries



Erich Tschermak

Carl Correns


Examples of Mendelian Genetics

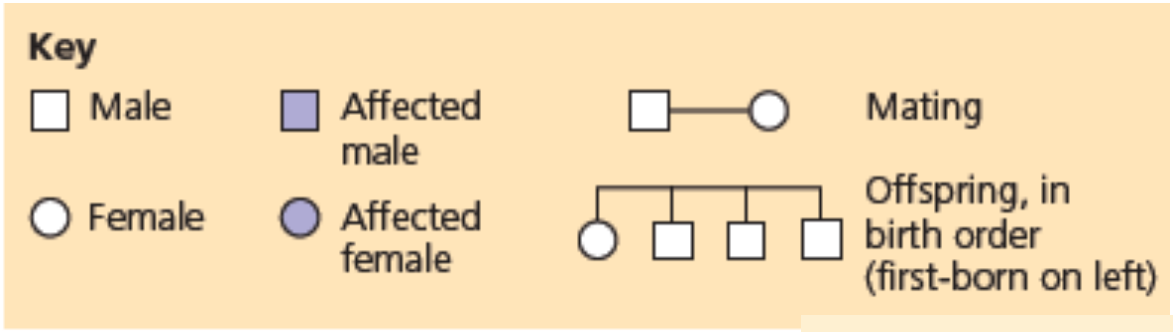
Many Human Traits follow Mendelian Patterns of Inheritance

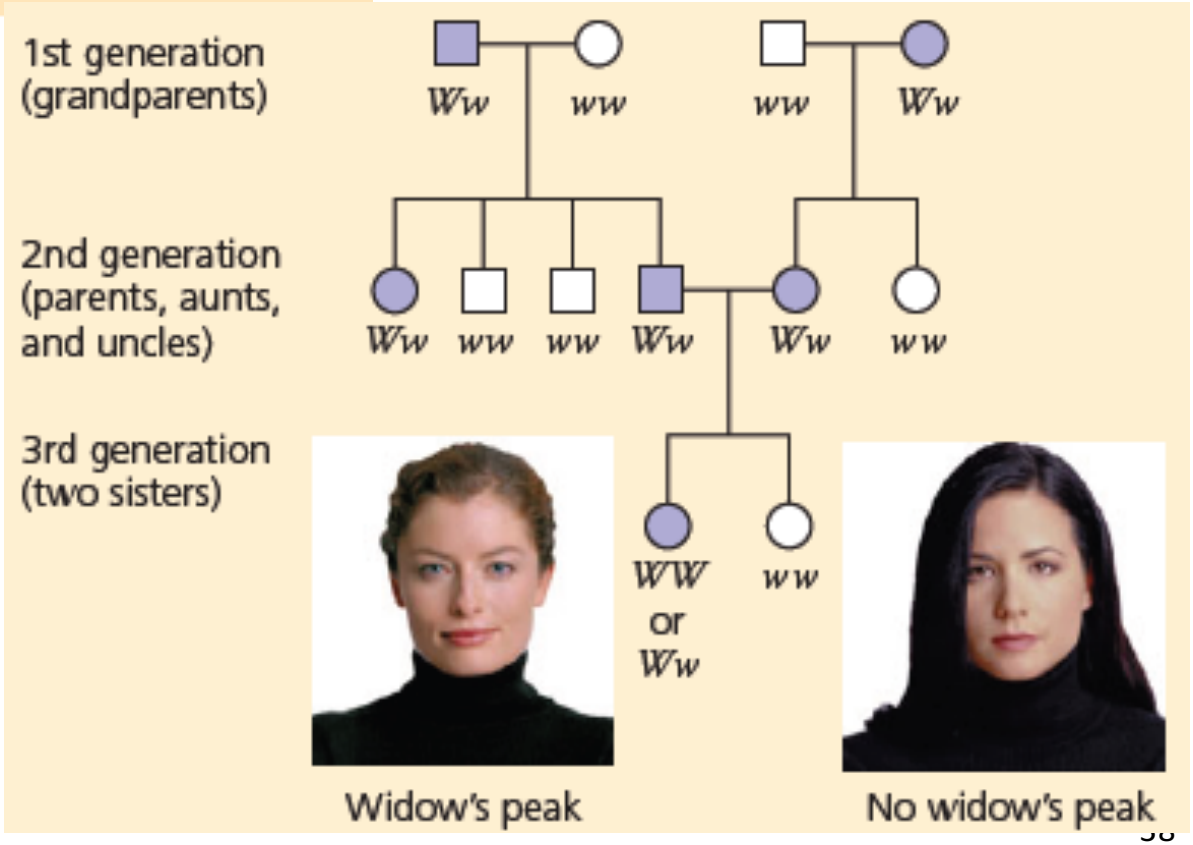
Pedigree analysis

Family history analysis for a particular trait and assembling information in family tree describing traits of parents and children across the generations - the family pedigree

Human traits to follow Mendelian genetics

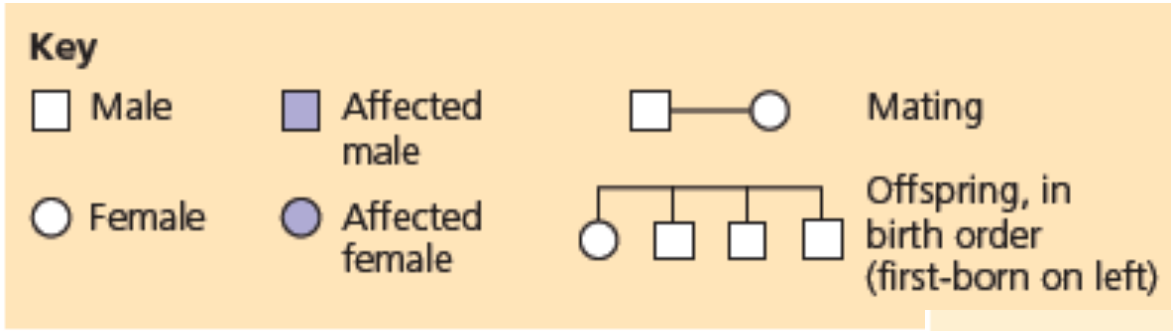
- Widow's peak (dominant trait)

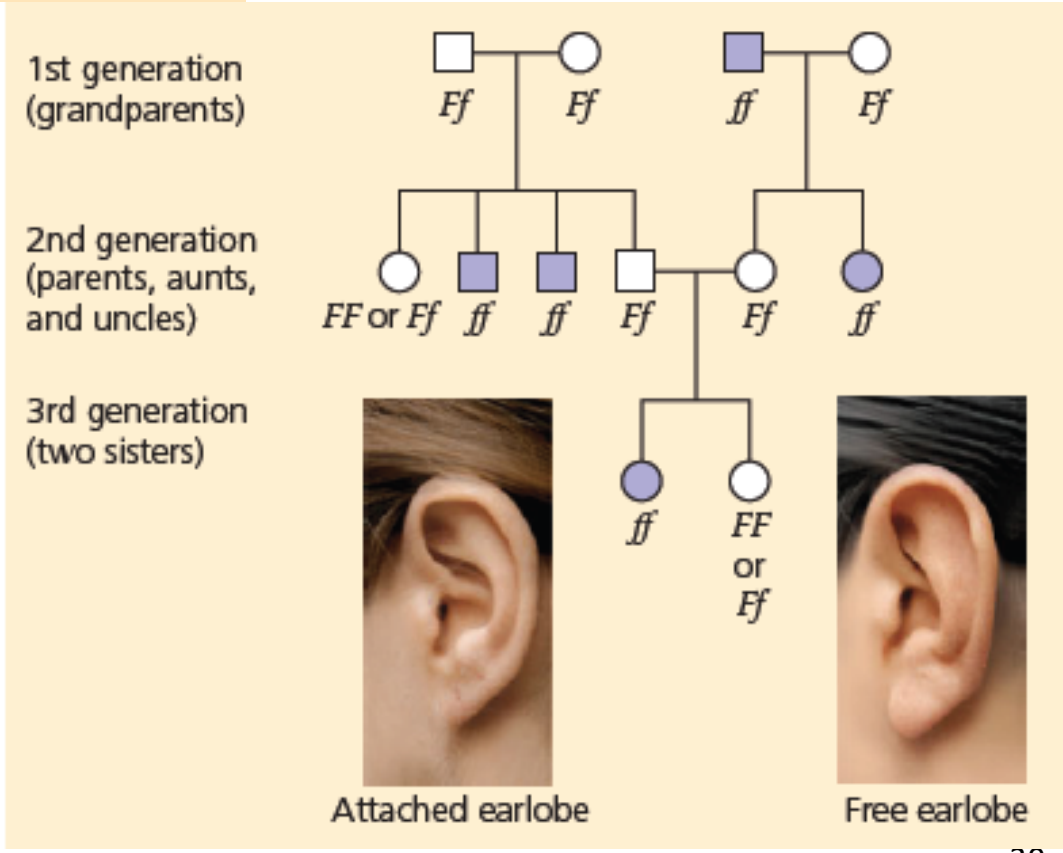




Human traits to follow Mendelian genetics

- Attached earlobe (recessive trait)



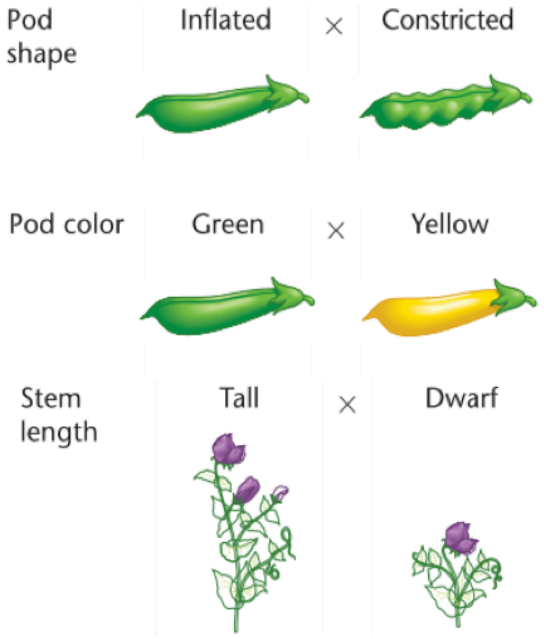


Inheritance patterns are often more complex than predicted by simple Mendelian genetics

Complex Inheritance Patterns

- In Mendelian genetics each character was determined by one gene, for which there are only two alleles (completely dominant or completely recessive)
- Relationship between genotype and phenotype are not always as simple!

Character	Dominant Trait	×	Recessive Trait
Flower color	Purple	×	White
Flower position	Axial	×	Terminal
Seed color	Yellow	×	Green
Seed shape	Round	×	Wrinkled

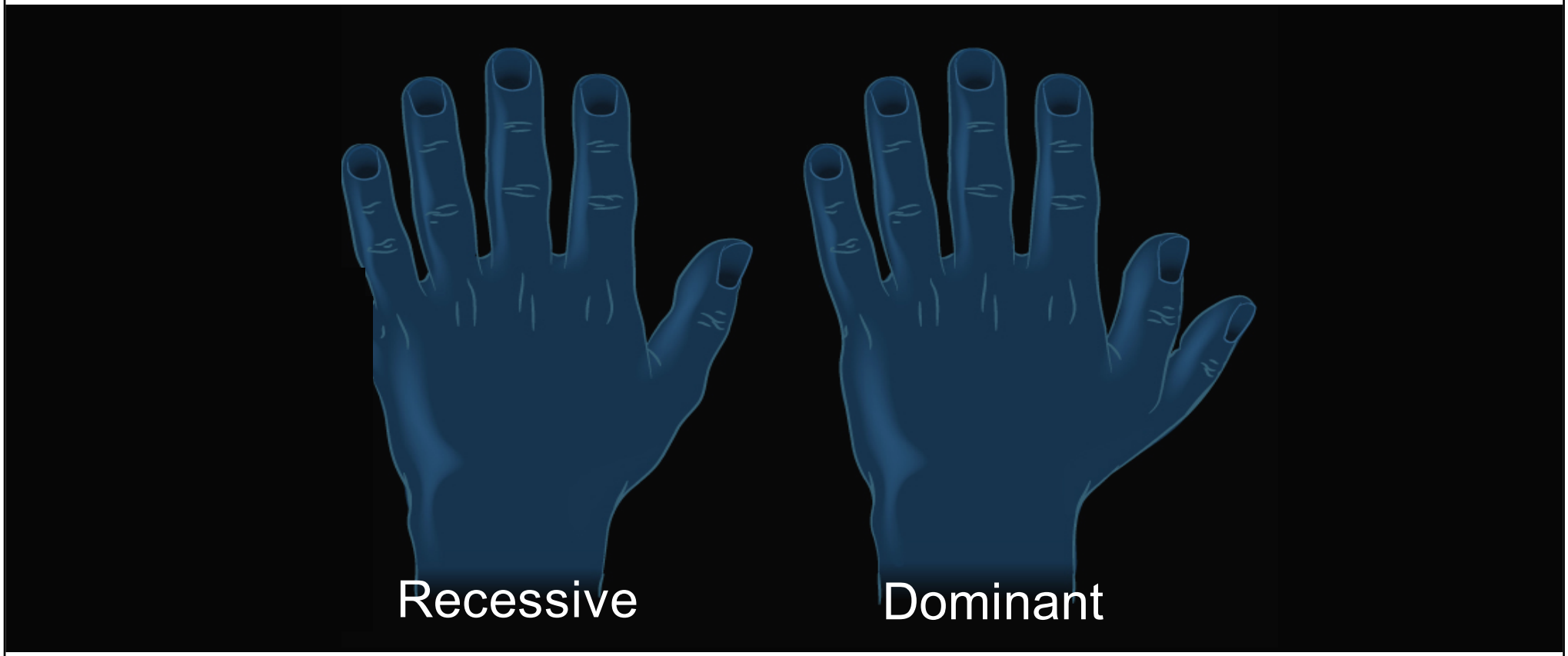


Degrees of Dominance

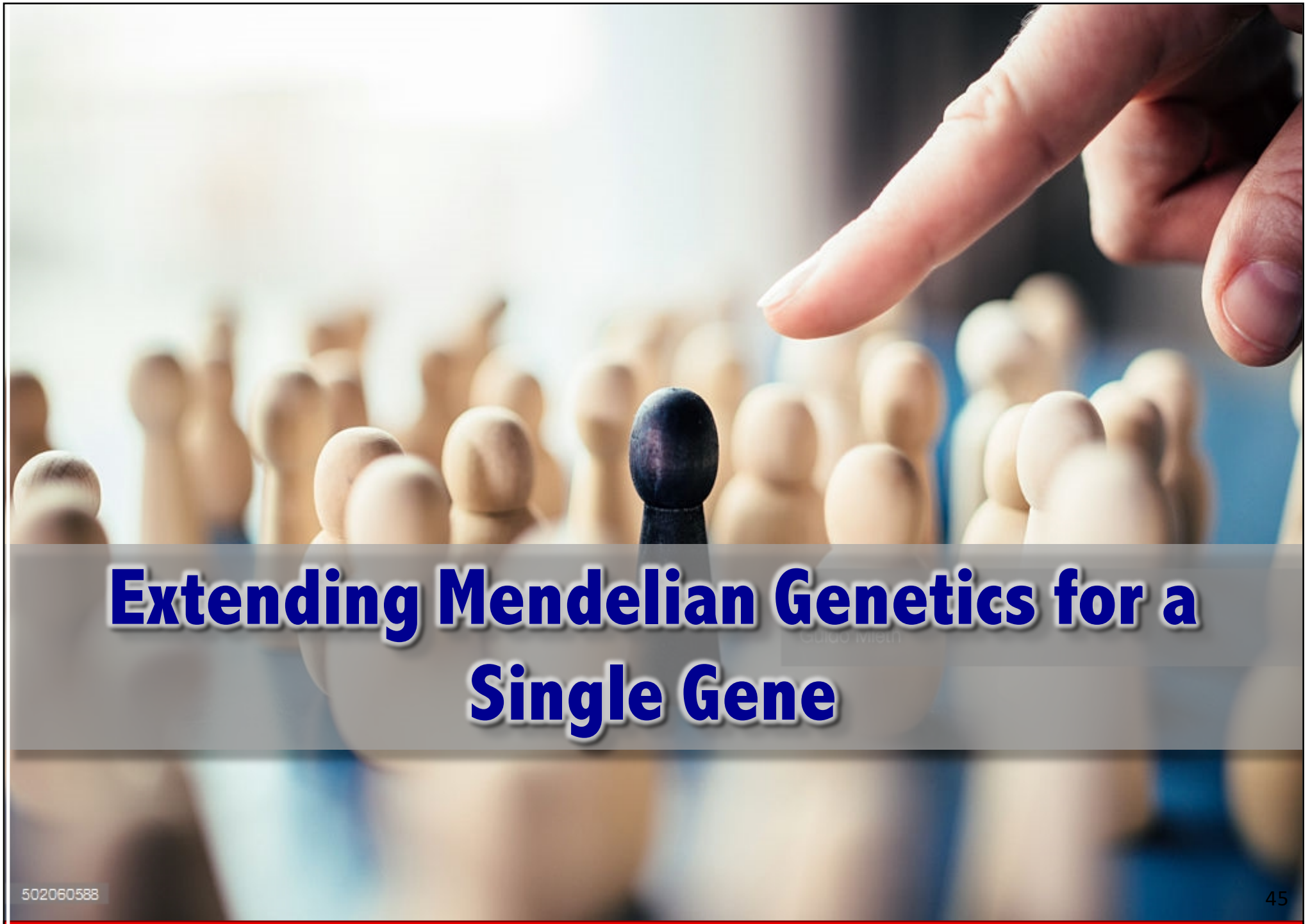
- Complete dominance occurs when phenotypes of the heterozygote and dominant homozygote are identical
- Incomplete dominance the phenotype of F_1 hybrids is somewhere between the phenotypes of the two parental varieties
- Co-dominance two dominant alleles affect the phenotype separately, distinguishable ways

Are dominant allele for a particular character always more common than the recessive allele?

Frequency of Dominant Alleles: Example Polydactyly



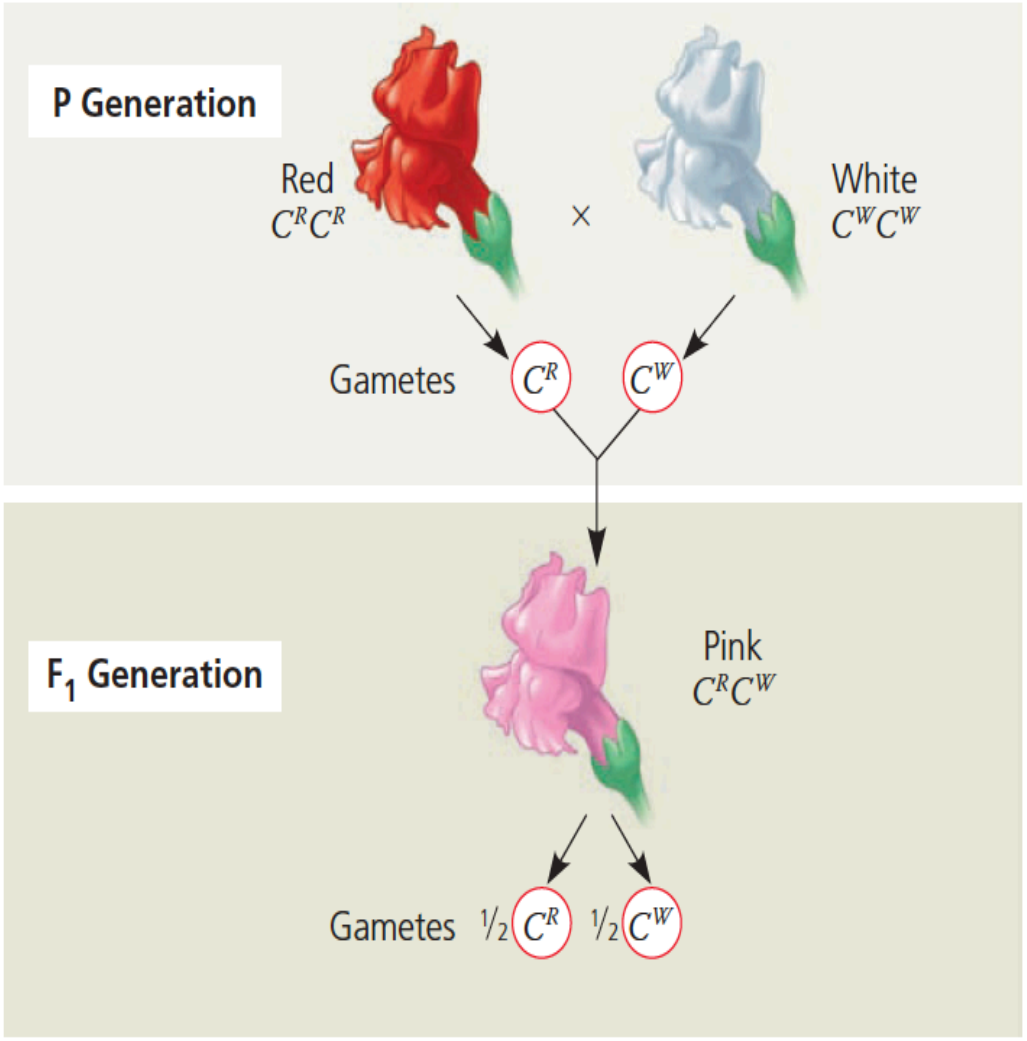
- In US ~ 0.4% children are born with polydactyly (an extra finger or toes)
- Recessive alleles (normal appearance with five digits) is far more prevalent than the dominant allele – Natural selection?

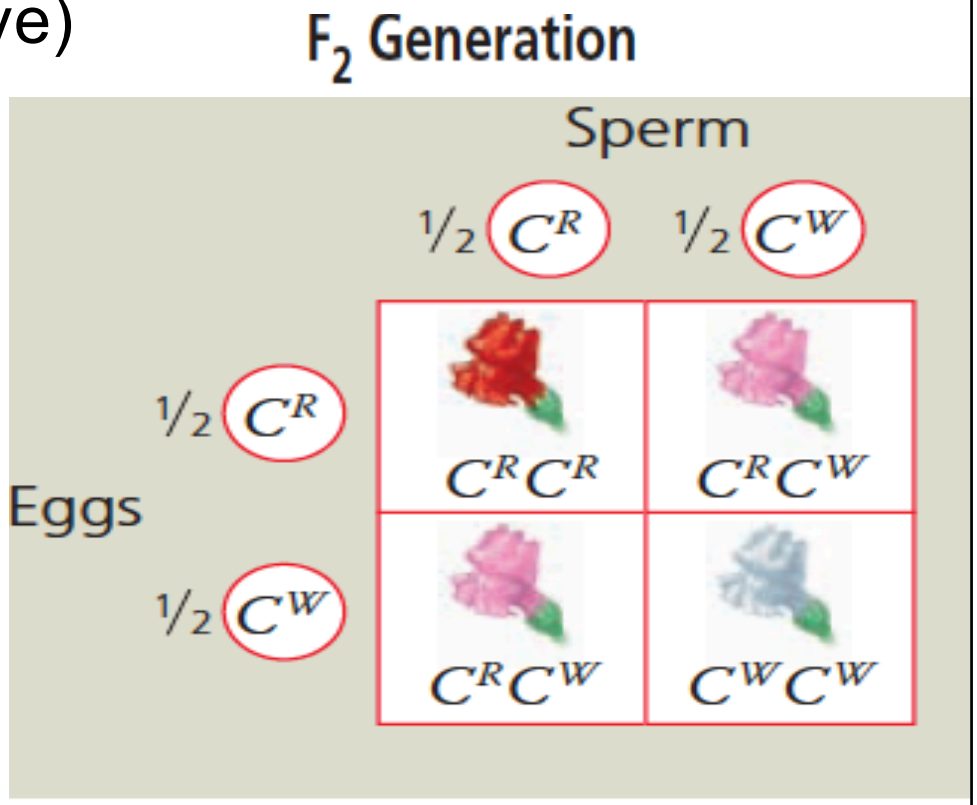


Extending Mendelian Genetics for a Single Gene

Examples of Deviation from Mendelian Genetics

- Incomplete dominance (Red Snapdragons; alleles are not completely dominant or recessive)





F2 generation with a 1:2:1 ratio

Figure 14.10

Albinism - A recessively inherited disorder shows up only in the homozygous (aa) individuals

If parents are carriers of the disorder but themselves have a normal phenotype. What would be the probability of carrier children when there are 4 siblings?

Albinism: A Recessively Inherited Disorder

Parents

Normal *Aa* X Normal *Aa*

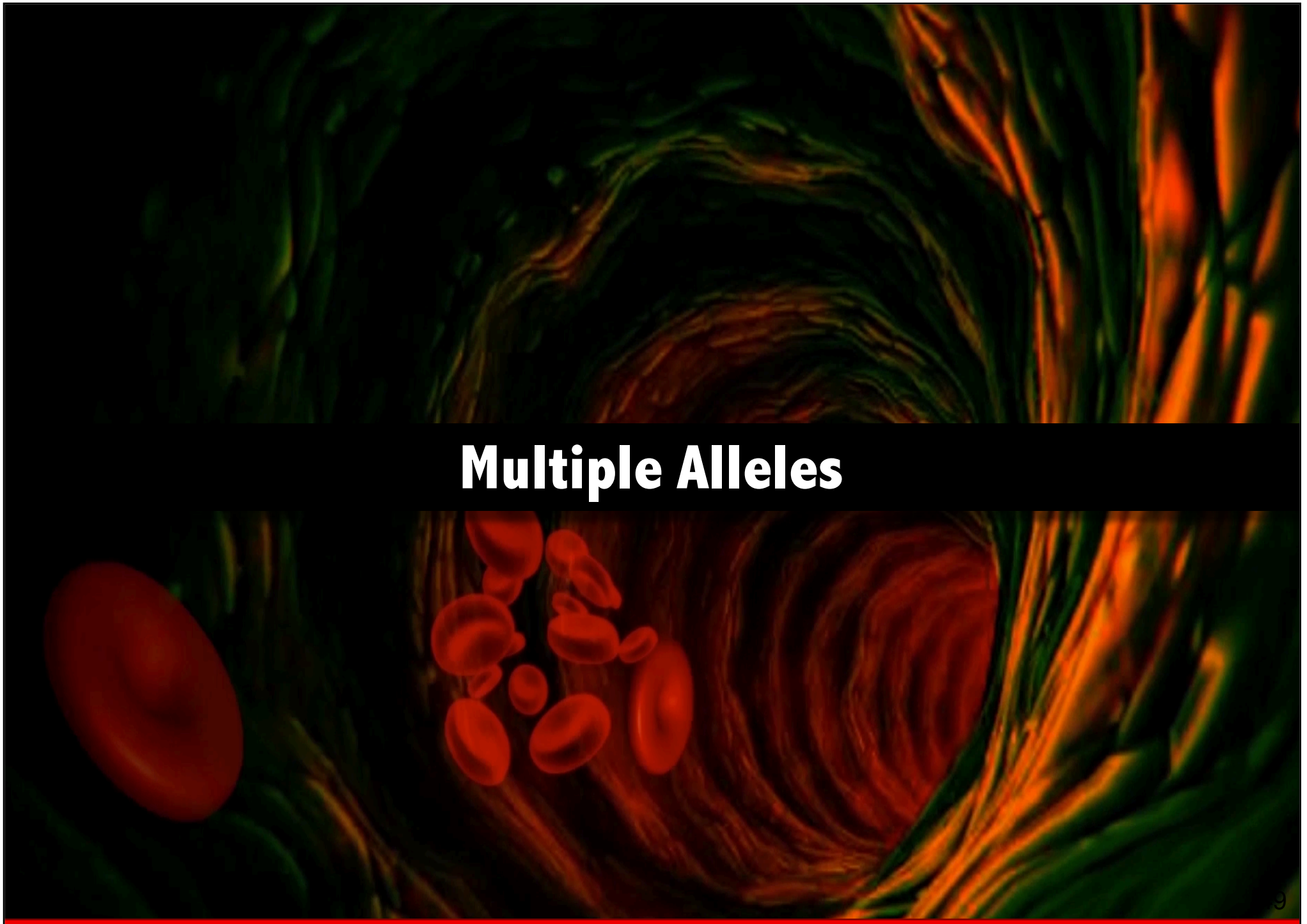
Sperm

Eggs

	A	*a*
A	*AA* Normal	*Aa* Normal (carrier)
a	*Aa* Normal (carrier)	*aa* Albino

Lack of pigmentation

Figure 14.16



Multiple Alleles

ABO Blood Grouping

- A person's blood group may be one of four types: A, B, AB, or O.
- These letters refer to two carbohydrates—A and B, which are found on RBC surface.
- Blood cells may have carbohydrate A (type A blood), carbohydrate B (type B), both (type AB), or neither (type O)

Multiple Alleles for ABO Blood Group

- Multiple alleles (when genes exist in more than 2 allelic forms)

Allele	I^A	I^B	i
Carbohydrate	A 	B 	none

The three alleles for the ABO blood groups and their carbohydrates

Genotype	$I^A I^A$ or $I^A i$	$I^B I^B$ or $I^B i$	$I^A I^B$	ii
Red blood cell appearance				
Phenotype (blood group)	A	B	AB	O

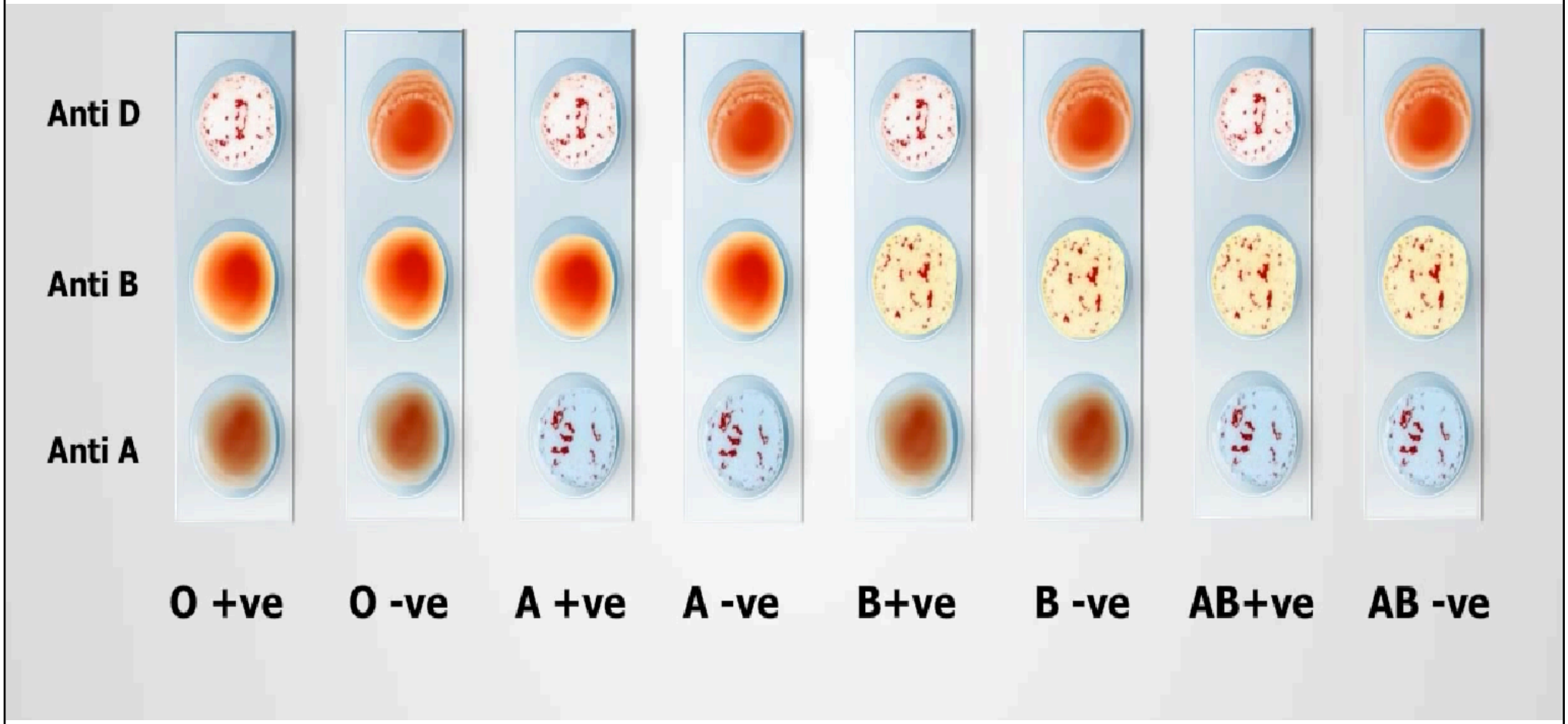
Blood group genotypes and phenotypes

Blood Group & Co-dominance

- **A** and **B** blood groups are **dominant** over **O** blood group
- A and B group genes are co-dominant

Parent Allele	A	B	O
A	AA	AB	AO
B	AB	BB	BO
O	AO	BO	OO

ABO Blood Grouping



Summary

- Gregor Mendel formulated “theory of inheritance” based on experiments with garden peas
- Discrete factors “genes” transmit characteristics from one generation to other
- A diploid individual must contain two copies of genes
  - *each parent transmits one copy to the next generation*
- A trait may not show up in an individual but can still be passed on to the next generation

References

- Campbell Biology - Reece, Urry, Cain, Wasserman, Minorsky, Jackson 10th Edition, Pearson
- Wilmut, I., et al., Nature, 385: 264-267 (1997)

- *Video contents*
  - https://www.youtube.com/watch?v=Mehz7tCxjSE
  - https://www.youtube.com/watch?v=-jKzLLHjRfs
- *Acknowledgment*
  - Cover images – getty images

Next Lecture…

Mendelian Genetics Examples
Chromosomal and Molecular basis of inheritance