Lecture – 3

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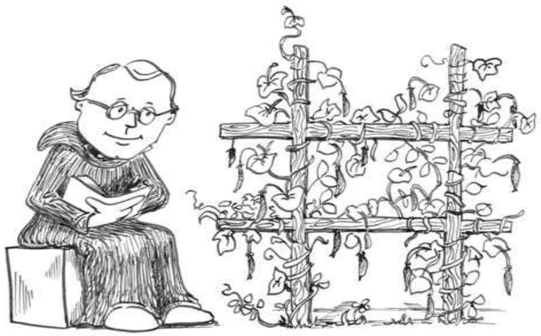
The instruction manual of life

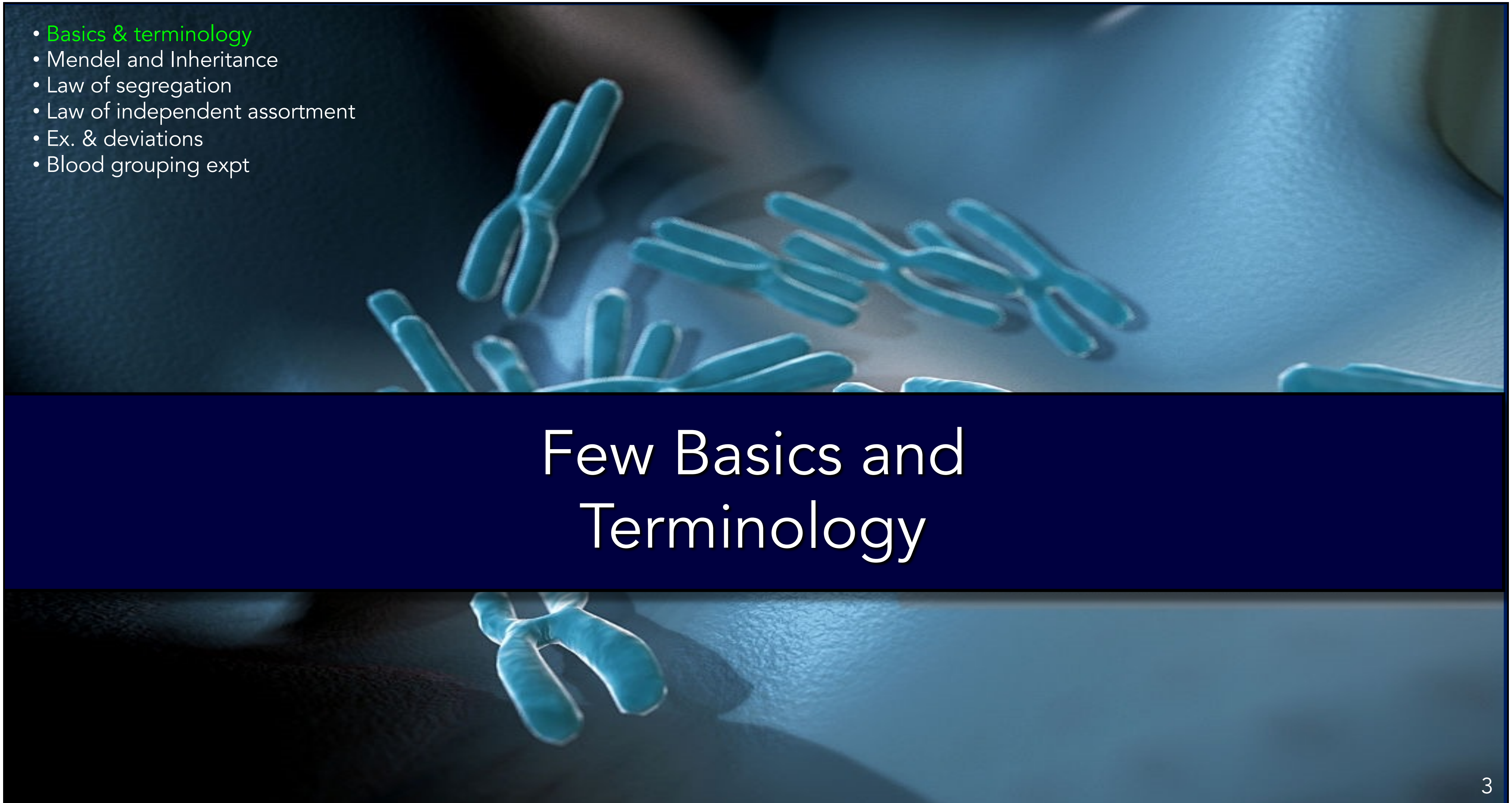


How was it discovered?

Mendel and heredity

Outline

- Few basics & terminologies
- Mendel and Concept of Inheritance
- Law of Segregation
- Law of Independent Assortment
- Examples (& Deviation) of Mendelian Genetics
- Blood grouping experiment



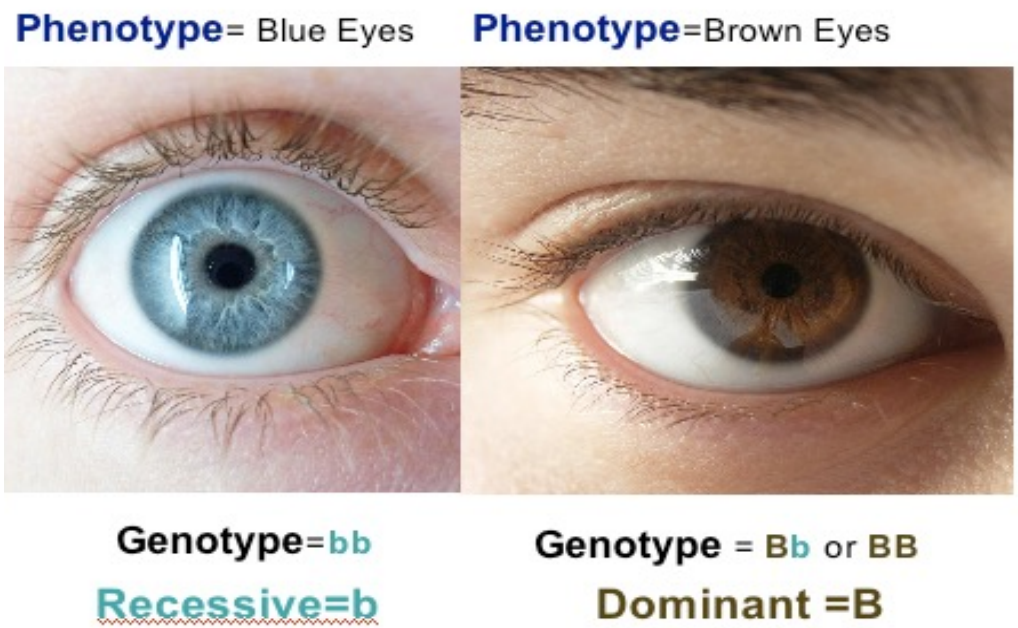
- Basics & terminology
- Mendel and Inheritance
- Law of segregation
- Law of independent assortment
- Ex. & deviations
- Blood grouping expt

Few Basics and Terminology

Genotype & Phenotype

Genotype: An organism's full hereditary information (today, we know this to be the genome, consisting of DNA)

Phenotype: Actual observed properties



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

Natural Variations

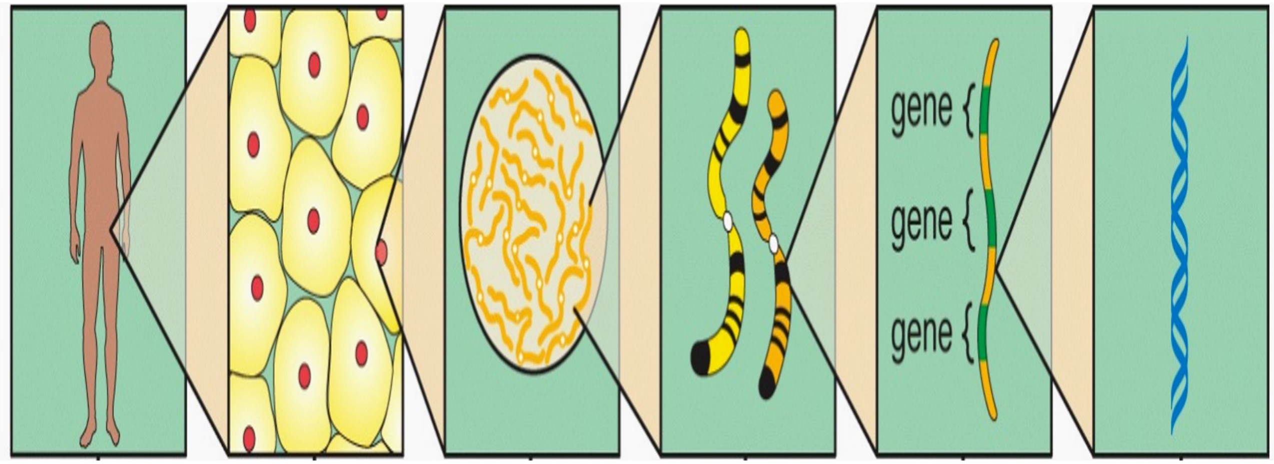
Eyes - Brown, Blue, Green or Gray

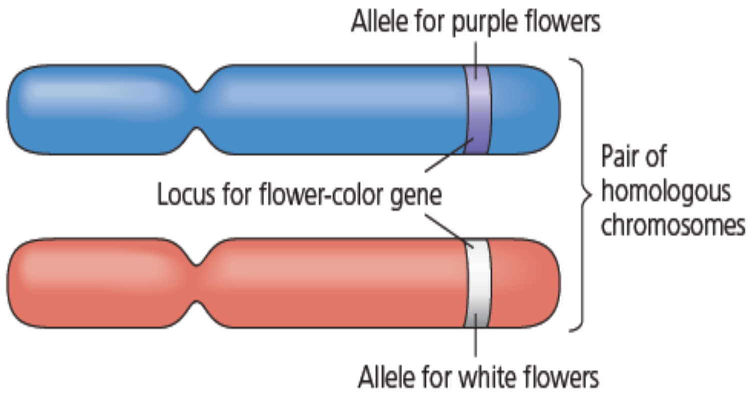
Hair - Black, Brown, Blond or Red

Image - Wikipedia

An Overview of Gene & Alleles

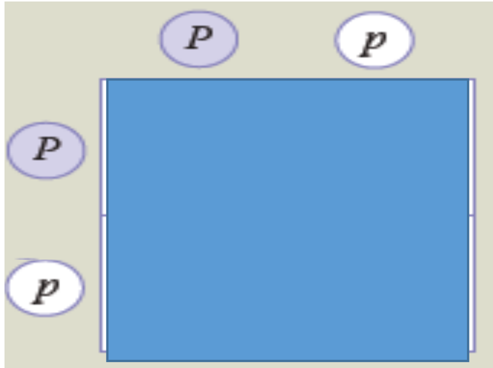
DNA double helix

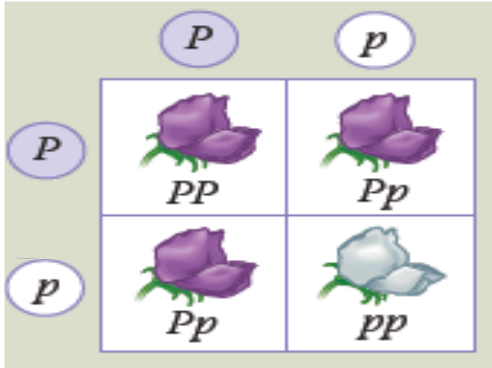




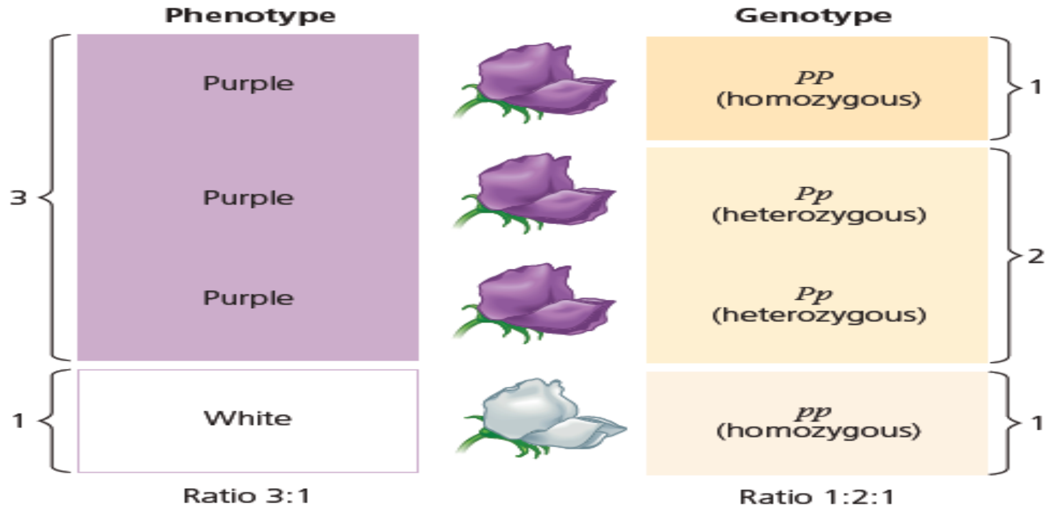
- Gene - sequence of nucleotides at a specific place, or locus, along a particular chromosome = Mendel's "heritable factor"
- Alleles are different forms of a gene inherited from both the parents
  - Each organism harbors two copies of a gene
  - Dominant allele has the effect but recessive has not

Genotype vs. Phenotype





Phenotype ratio? Genotype ratio?



Different alleles: Heterozygous; Identical alleles: Homozygous

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

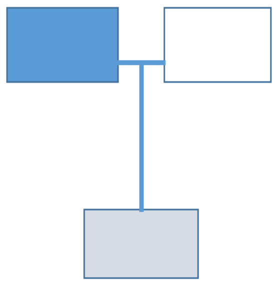
Blending hypothesis - similar to the color palette

- Collection of genes is like deck of cards

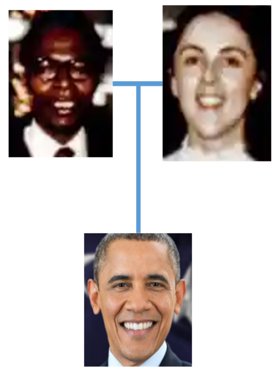
Player 1
↓
Player 2
↓
Player 3

Parent
↓
Son/daughter
↓
Grand son/daughter

Color mixing



Parents
↓
Son/daughter



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

First insights: Mendel and the idea of inheritance

- 1857, Mendel began breeding garden peas
- 1860, Gregor Mendel provided laws of inheritance to define genetic principles

1822-1884

Drawing from the Deck of Genes

Flow of heritable traits

- Before we knew about DNA, the most obvious visible manifestation was the presence of heritable traits
- Phenotypic characters that are passed on from parent to offspring e.g. eye and hair color
- Some phenotypes: probability of getting breast cancer, Alzheimer's disease, etc.
- Today, Uzbekistan is using genetic testing to find future Olympians (phenotypes are based on the sport), Estonia and many countries for marriage councelling

- Basics & terminology ✓
- Mendel and Inheritance
- Law of segregation
- Law of independent assortment
- Genetics & Rules of Probability
- Ex. & deviations
- Blood grouping expt

Mendel and Concept of Inheritance

Mendel discovered basic principles of heredity by breeding garden peas

Gregor Mendel: Elucidation of the principle of heredity

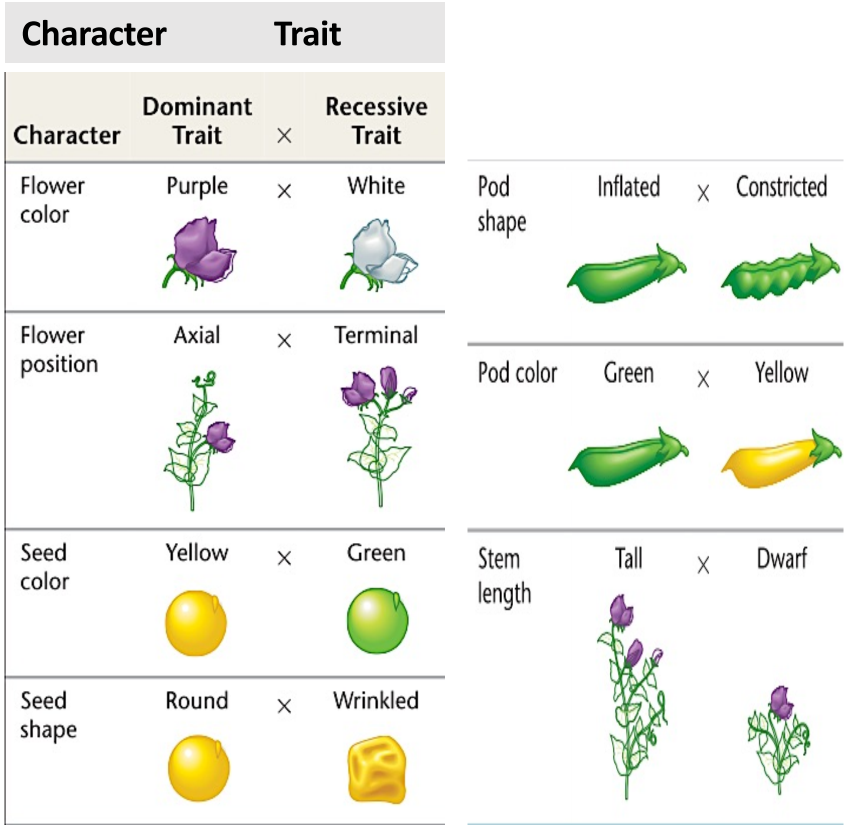
- Mendel’s choice of experimental system -- pea plants

Distinct heritable variation: characters

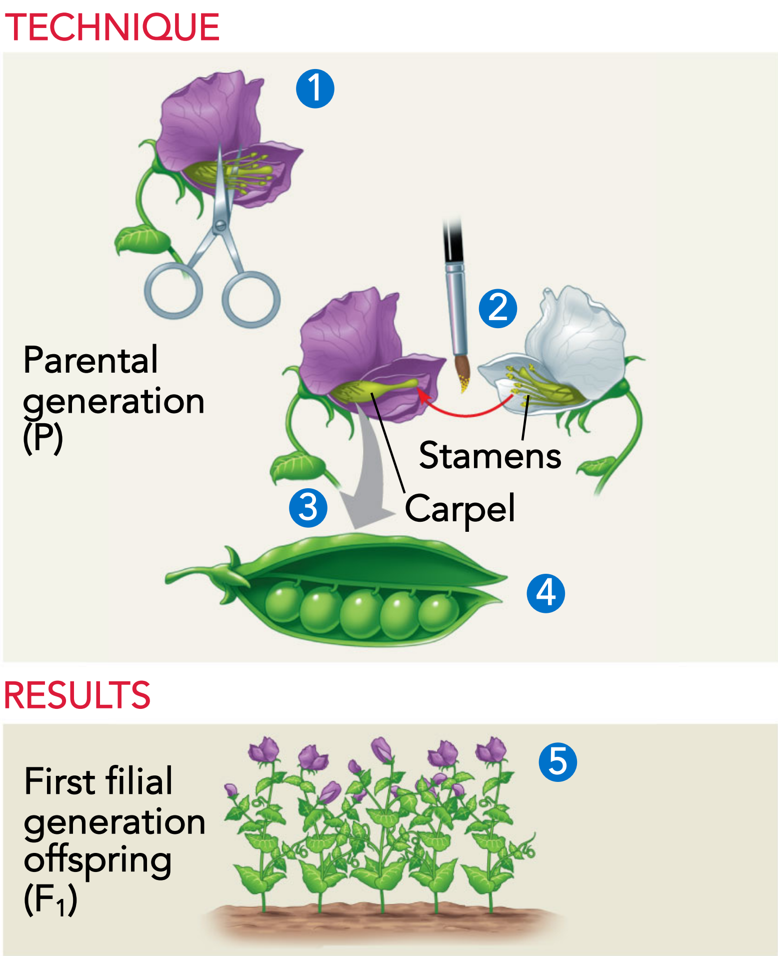
Why pea plants?

He was an avid gardener (& mathematician)

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



What is cross-pollination?



A **carpel** is the ovule and seed producing reproductive organ in flowering plants (like female).
A **stamen** is the pollen-producing reproductive organ of a flower (like male).

Pea flowers have both!

1: Remove the stamens of purple flowers so they cannot pollinate

2 & 3: Pollinate a purple flower carpel with the stamens of a white flower

4: The fertilized purple flower will give rise to seeds which can be planted

5: Observe the offspring for the trait

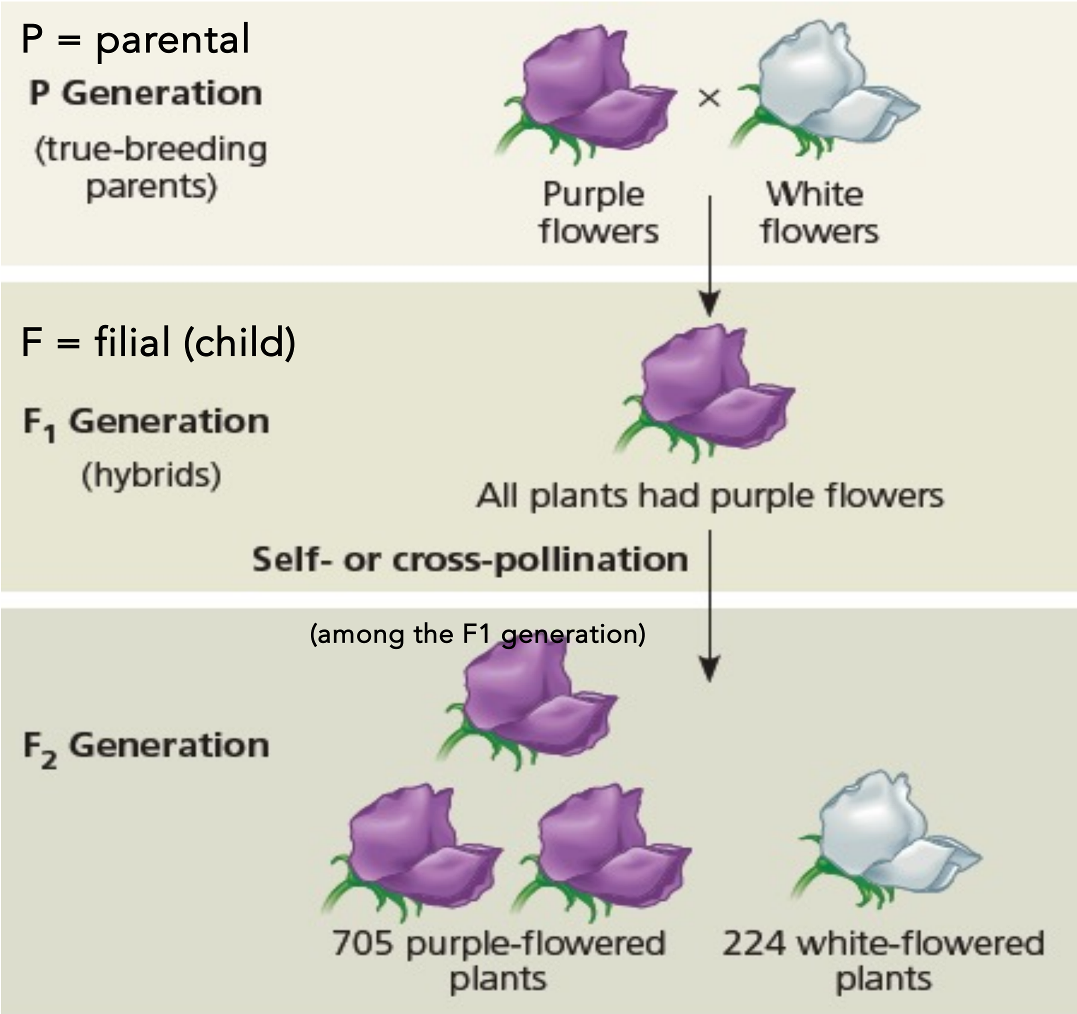
Before starting the real experiment…

Mendel made sure to choose only **true breeding** varieties

Purple-flowered plant X Purple-flowered plant

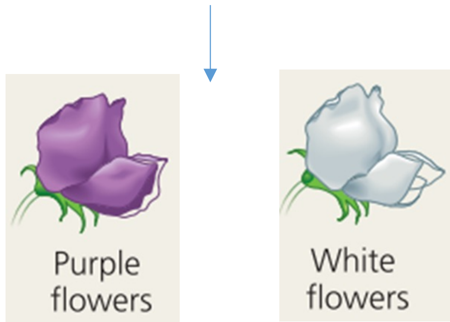
Always purple-flowered plant

Results of mating (crossing) of two contrasting traits



<u>Surprises!</u>

1) <u>Most importantly, no blending!</u>
2) F1 were all purple; where did white go?
3) F2 showed the presence of missing white
4) 3:1 ratio of purple to white in the F2 generation; why this ratio and what does it mean?



Dominant trait Recessive trait

- Basics & terminology ✓
- Mendel and Inheritance ✓
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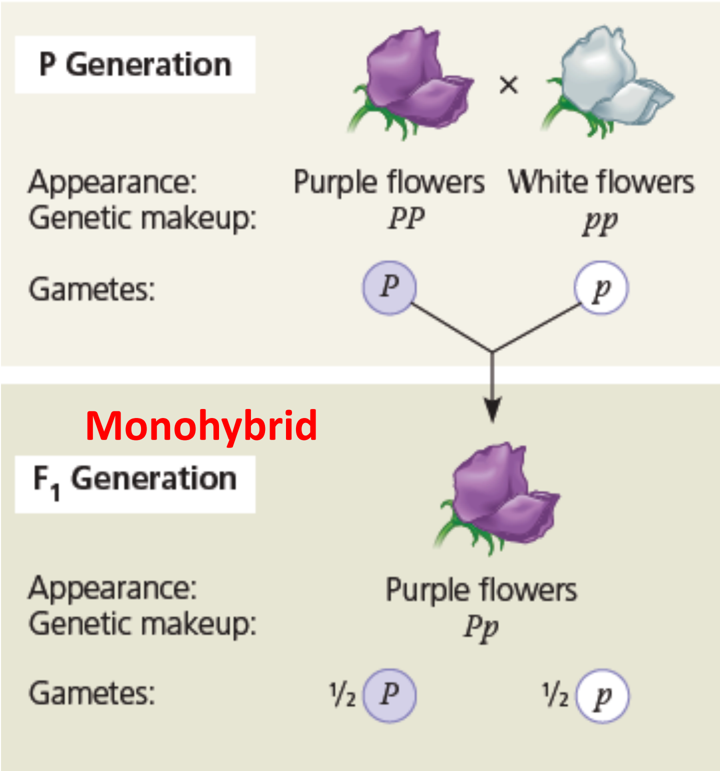
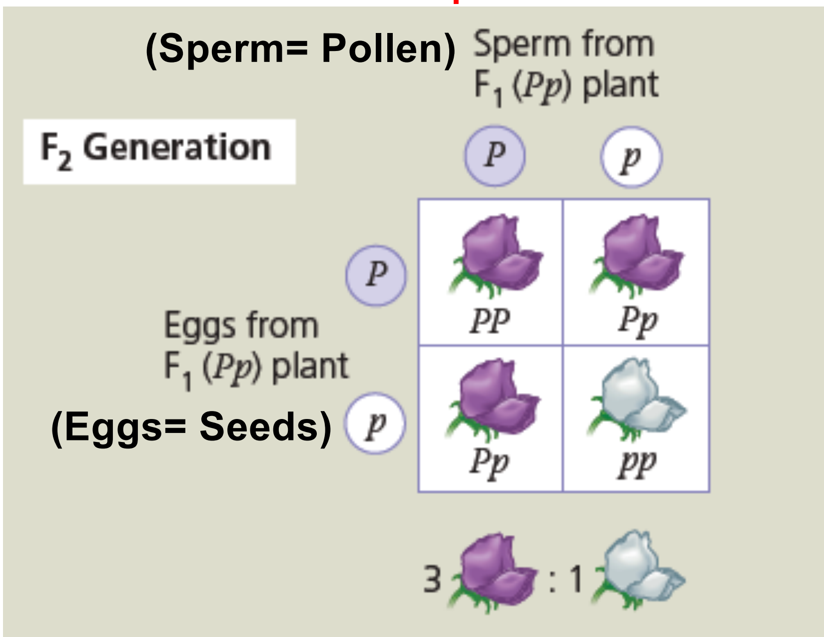
The 3:1 ratio can be explained by the concept of alleles

- Law of segregation -- Two units (called alleles today) for a heritable traits separate from each other during gamete formation and end up in different gametes

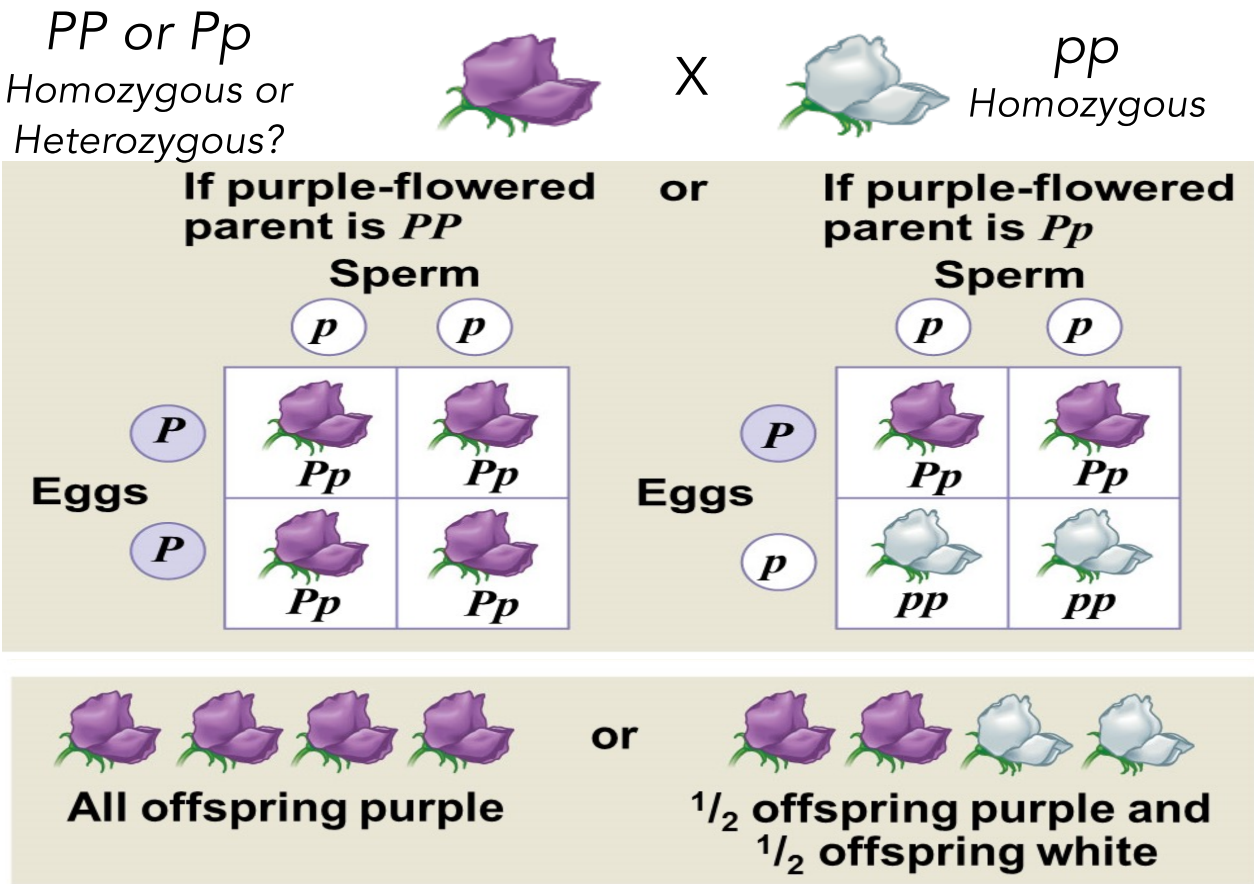


Punnett square



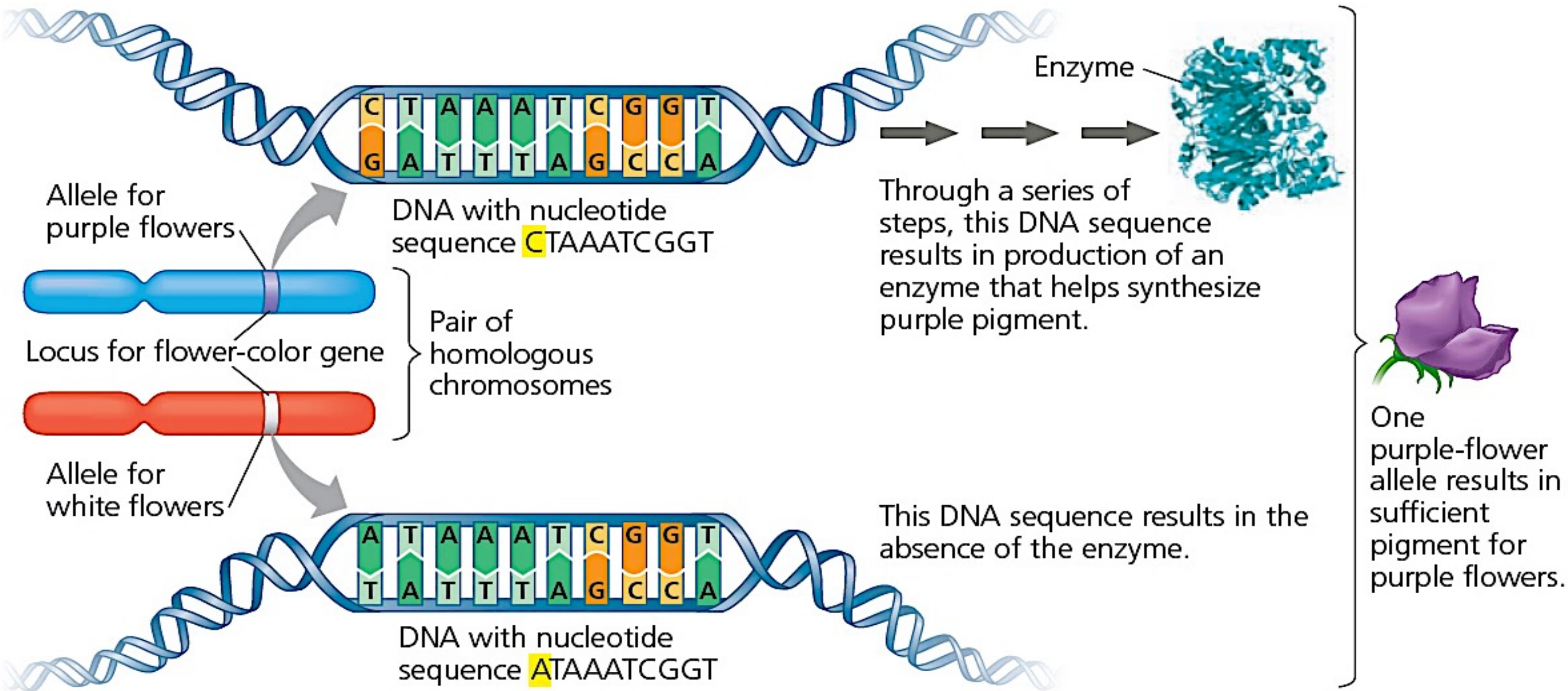
F_2 plants – a cross between 2 F_1 plants

TEST CROSS: Determine Organism's Genotype



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

An Overview of Gene & Alleles



Inferences made by Mendel (1865)

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

These were not consistent with the 'blending' hypothesis of heredity and led to a new 'particulate' hypothesis: genetic material must be in the form of discrete units

- Basics & terminology ✓
- Mendel and Inheritance ✓
- Law of segregation ✓
- Law of independent assortment
- Genetics & Rules of Probability
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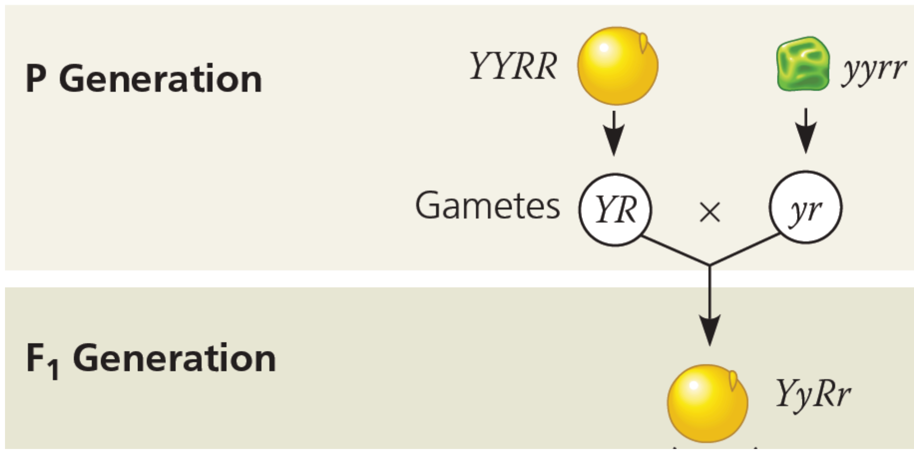
Law of Independent Assortment

Law of Independent Assortment

- 2 traits were simultaneously studied e.g. dihybrid cross

Dominant (*YYRR*) Recessive (*yyrr*)

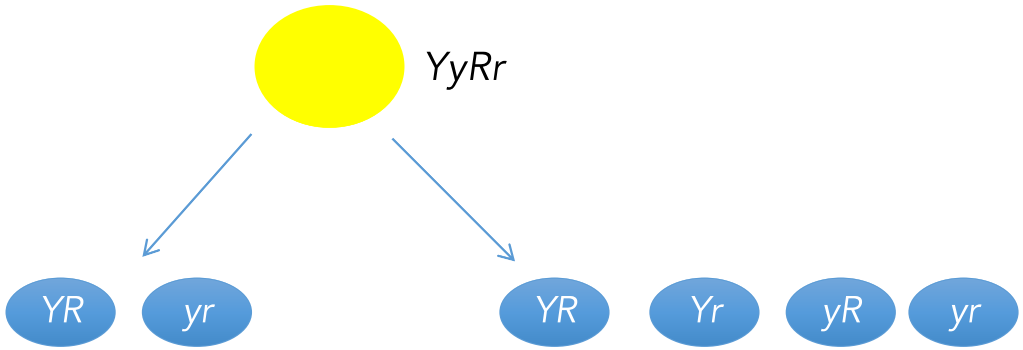
Dihybrid



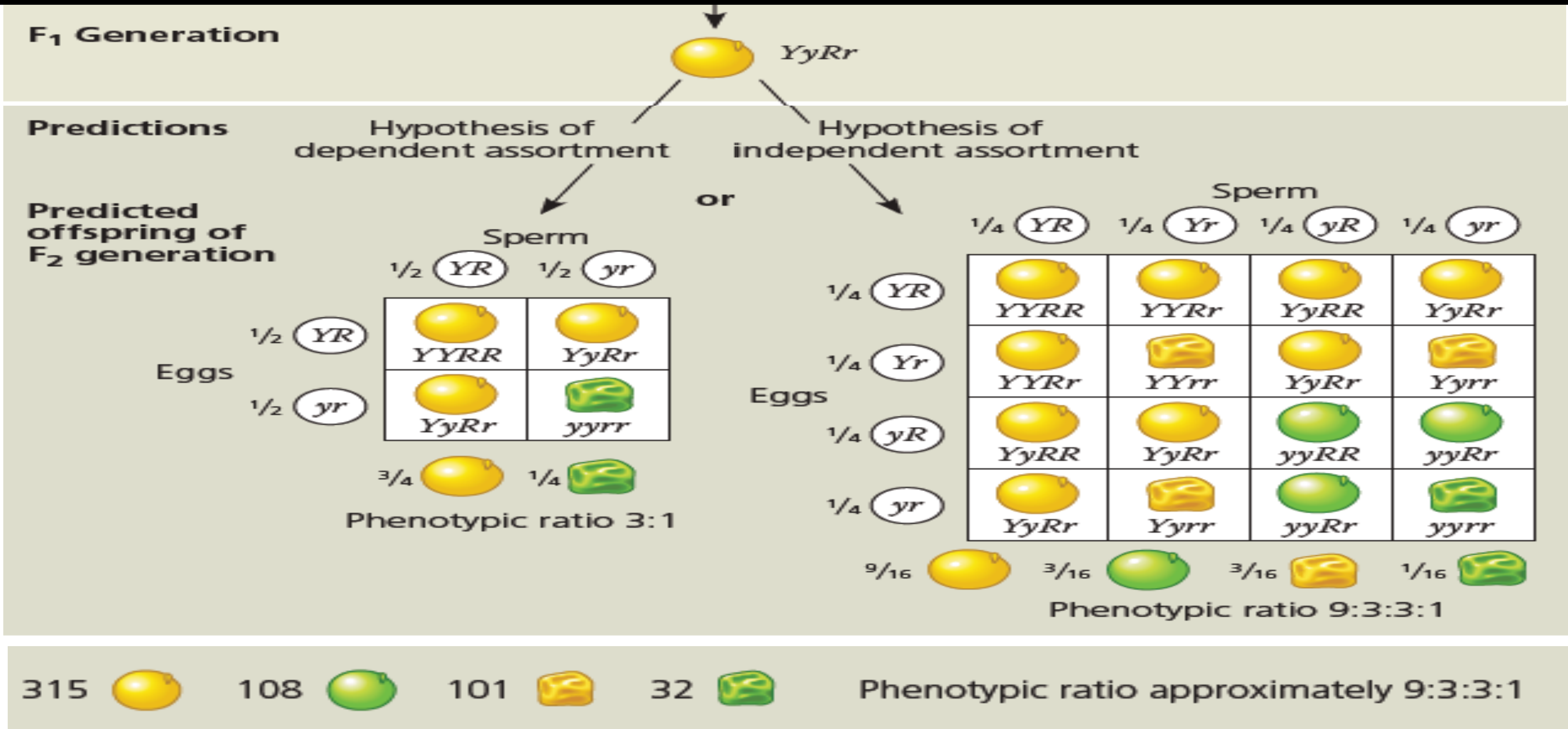
Law of Independent Assortment

Dihybrid Cross: Dependent or Independent Assortment?

F1 Generation



Results of Dihybrid Cross: Law of Independent Assortment



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

- Basics & terminology ✓
- Mendel and Inheritance ✓
- Law of segregation ✓
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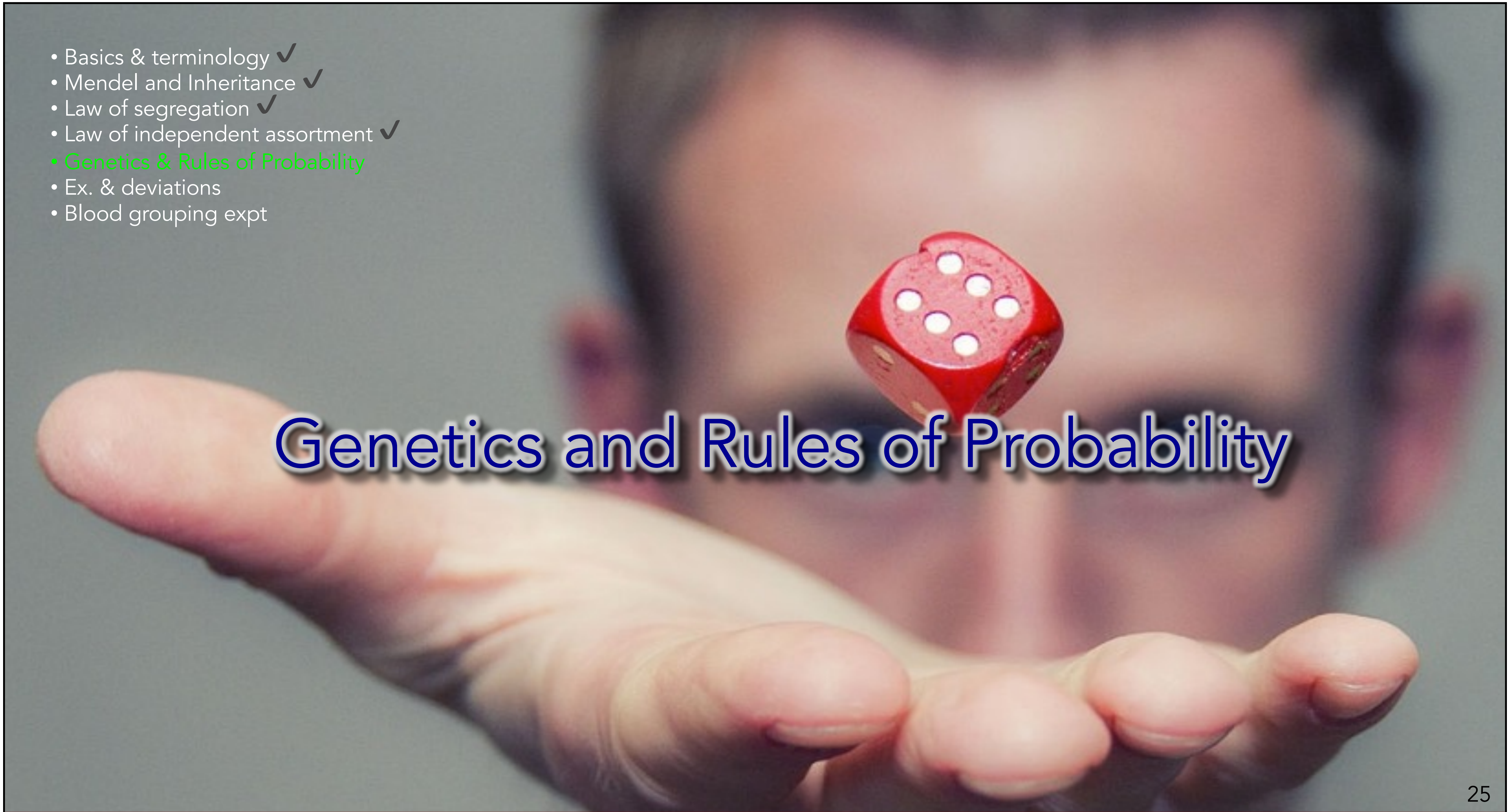


Genetics and Rules of Probability

Basics: Probability

- Probability is a measure of the expectation that an event will occur (probability is always between 0 – 1)

- To determine the probability of any genotype in offspring of two heterozygote – *multiply the individual probability of a specific allele from egg & sperm*

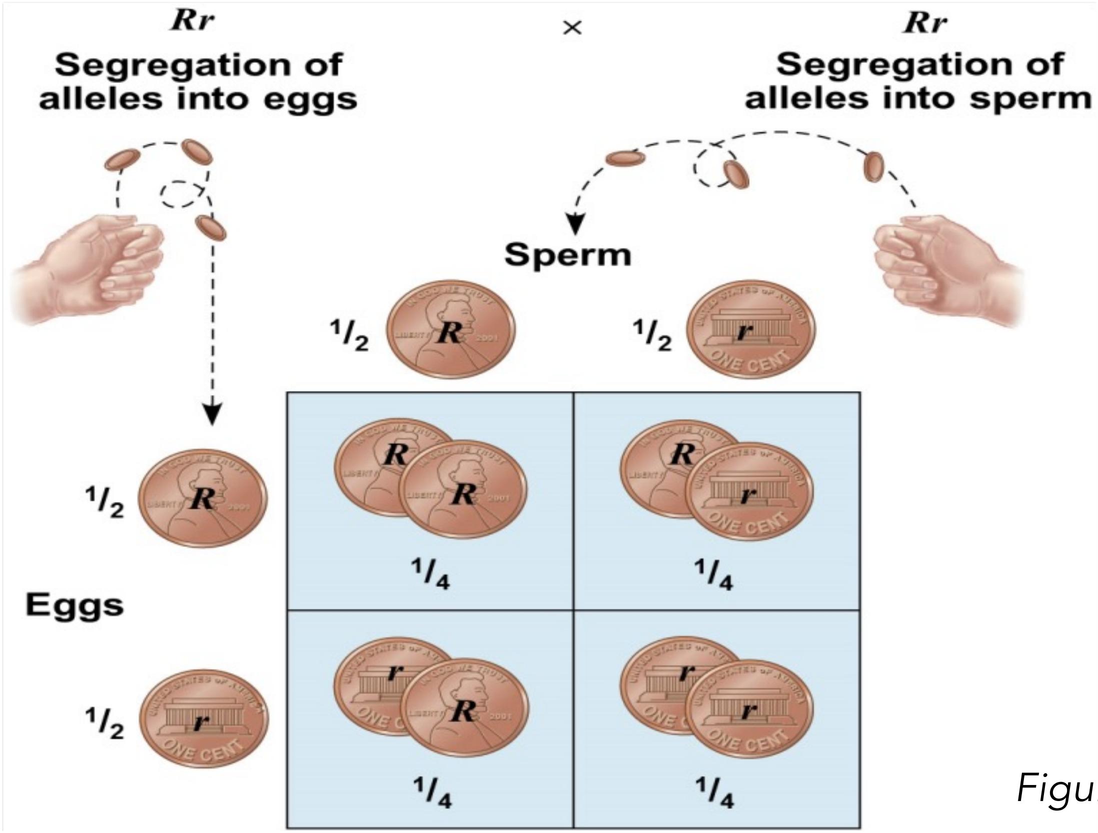
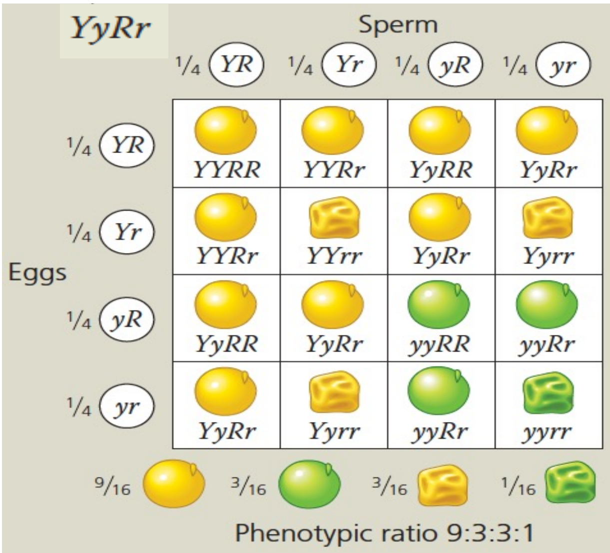


Figure 14.9

Genetics and Rules of Probability

- Remember, Law of Independent Assortment - each allelic pair segregates independently during gamete formation.
- Therefore, a dihybrid or multicharacter cross is equivalent to two or more independent monohybrid crosses happening simultaneously.
- Consider dihybrid cross between YyRr heterozygotes or monohybrid cross of Yy plants (seed color) or Rr plants (seed shape).



Consider each gene separately

Genetics and Rules of Probability

Seed color

	Y	y
Y	*YY*	*Yy*
y	*Yy*	*yy*

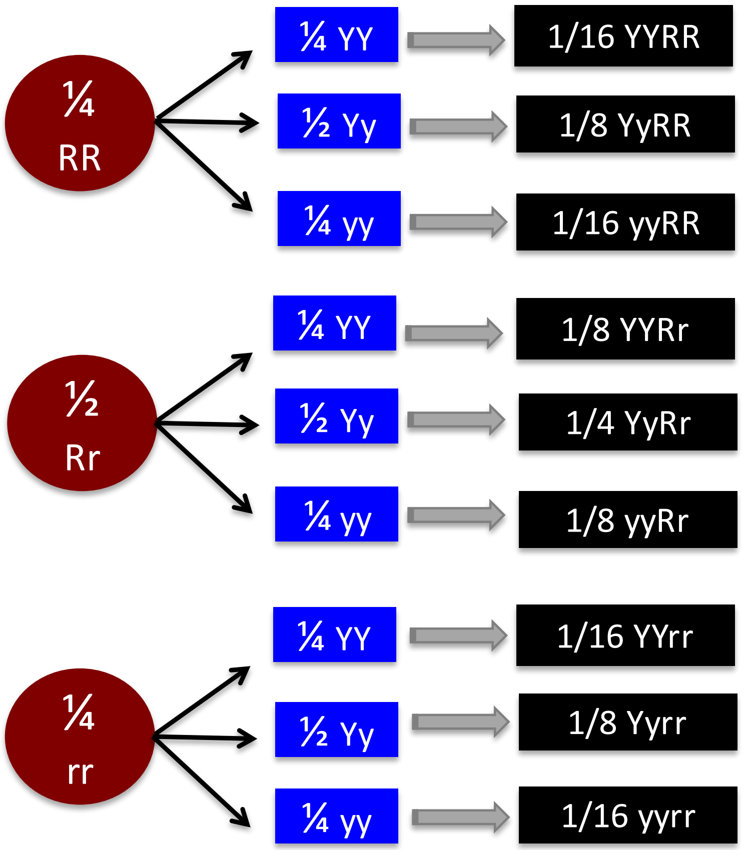
¼ YY, ½ Yy, ¼ yy

Seed shape

	R	r
R	*RR*	*Rr*
r	*Rr*	*rr*

¼ RR, ½ Rr, ¼ rr

- Probability of F2 genotype YYRR ? = ¼ (YY) X ¼ (RR) = 1/16
- Probability of F2 genotype YyRR ? = ½ (Yy) X ¼ (RR) = 1/8



Genetics Examples

- What is the probability of having an Albino child, if both the parents are heterozygous for the albinism?

	A	*a*
A	*AA*	*Aa*
a	*Aa*	*aa*

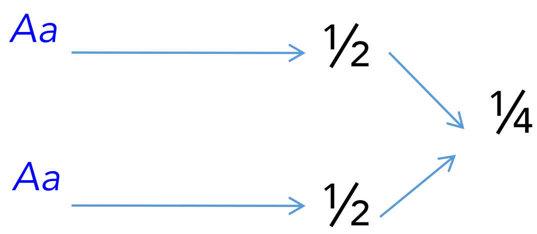


Figure 14.18

Genetics Examples

- Achondroplasia (dwarfism) is due to a dominant allele. What is the probability of having a child with disease if one parent is homozygous recessive and the other is heterozygous?

	D	d
d	Dd Dwarf	dd Normal
d	Dd Dwarf	dd Normal

½

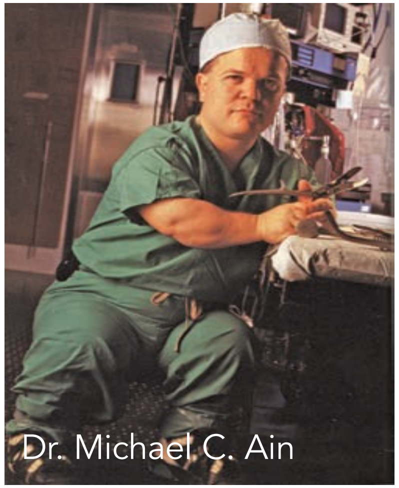


Figure 14.18

Summary: Law of Independent Assortment

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.

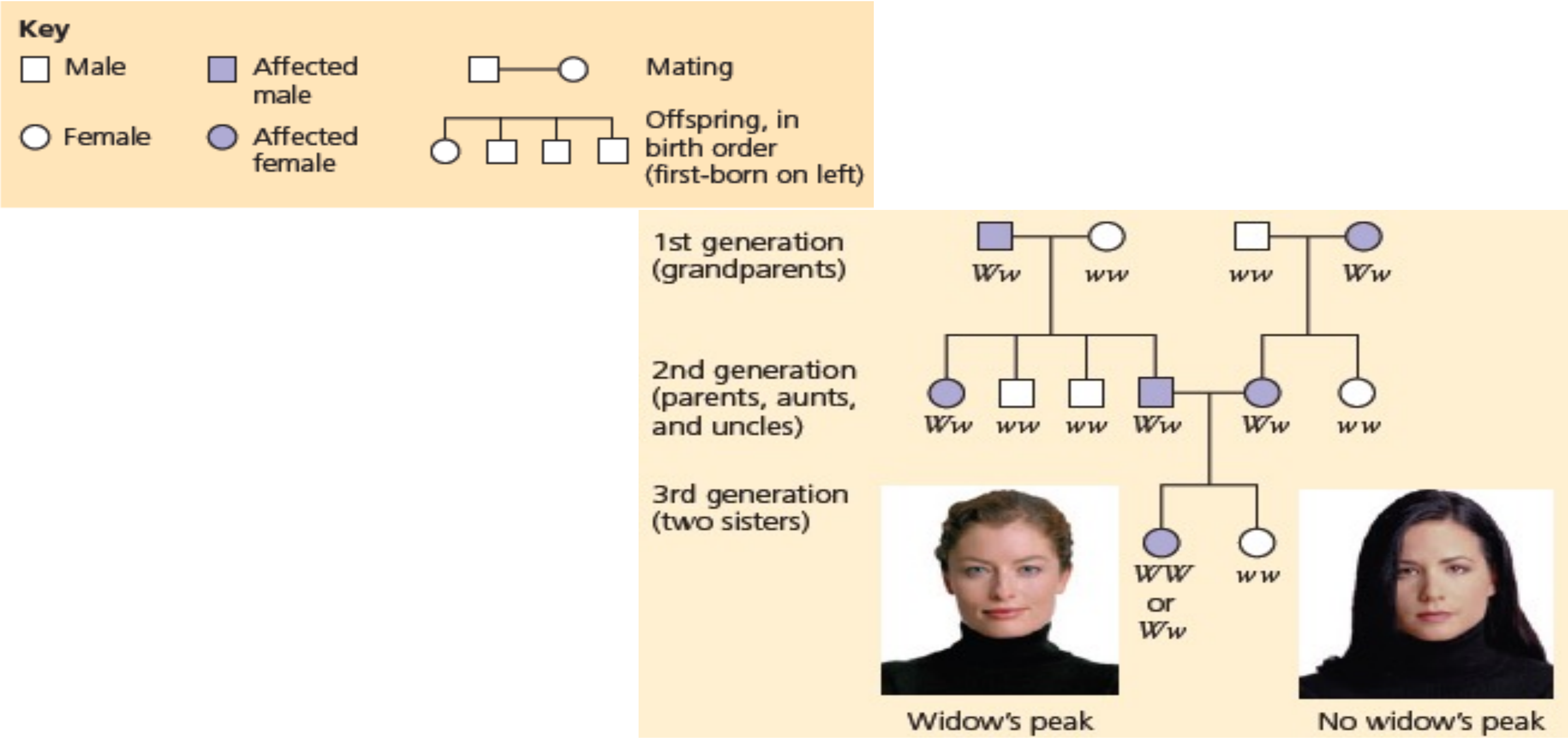

• Basics & terminology ✓
• Mendel and Inheritance ✓
• Law of segregation ✓
• Law of independent assortment ✓
• Genetics & Rules of Probability
• Ex. & deviations
• Blood grouping expt
Examples of Mendelian Genetics

Many Human Traits follow Mendelian Patterns of Inheritance

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

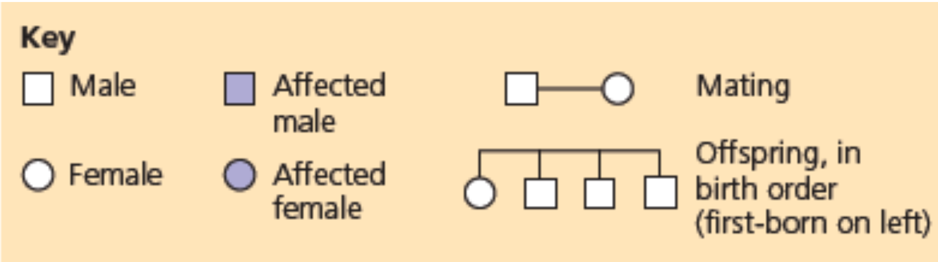
Human traits to follow Mendelian genetics

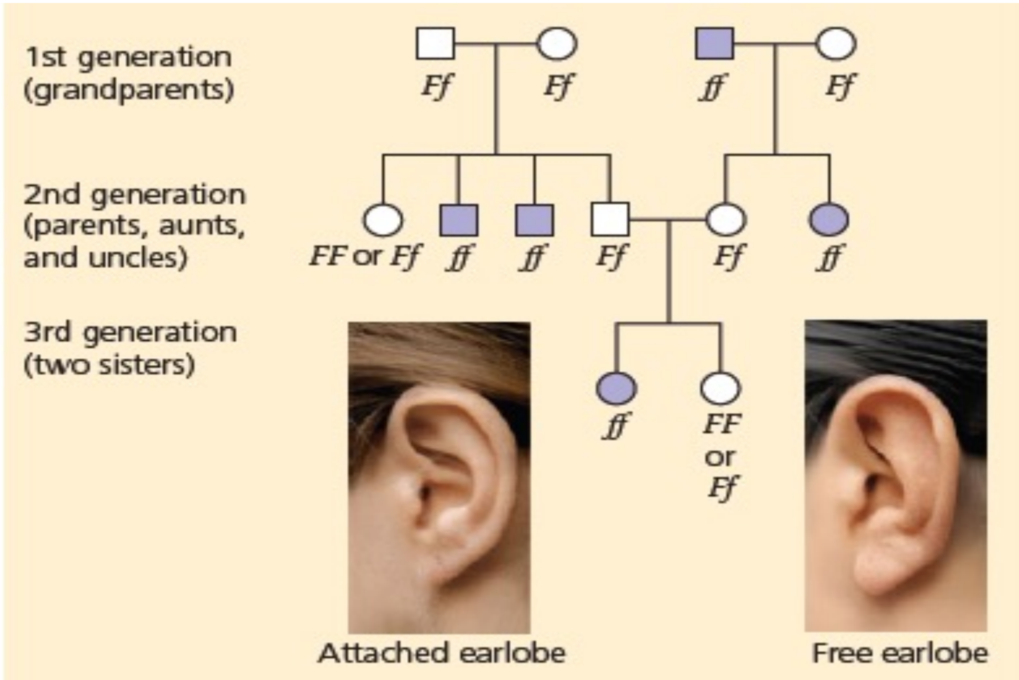
- Widow's peak (dominant trait)



Human traits to follow Mendelian genetics

- Attached earlobe (recessive trait)





Human traits that follow Mendelian genetics

Single gene human diseases:

- Phenylketonuria
- Hemophilia
- Sickle cell disease
- Beta-thalassemia

Online *Mendelian Inheritance in Man (OMIM)* database identifies some 4000 Mendelian disorders.

Other traits:

- Resistance to HIV
- Ability to taste phenylthiocarbamide

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

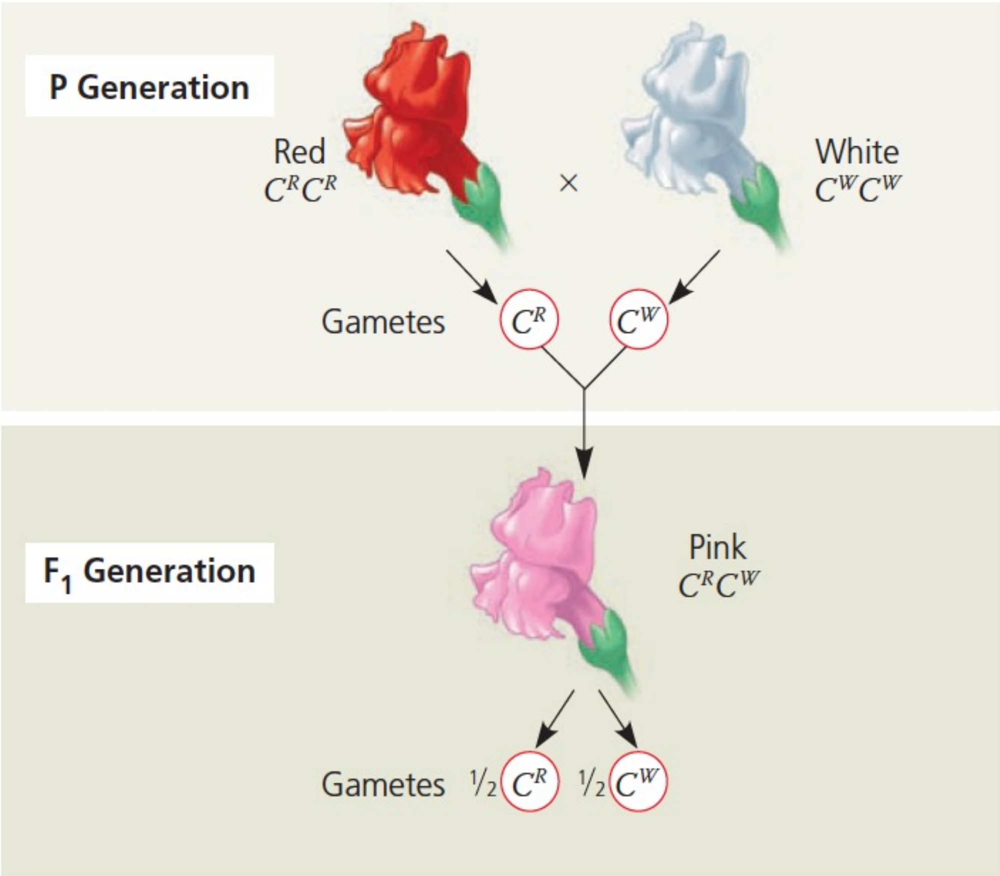
Degrees of Dominance

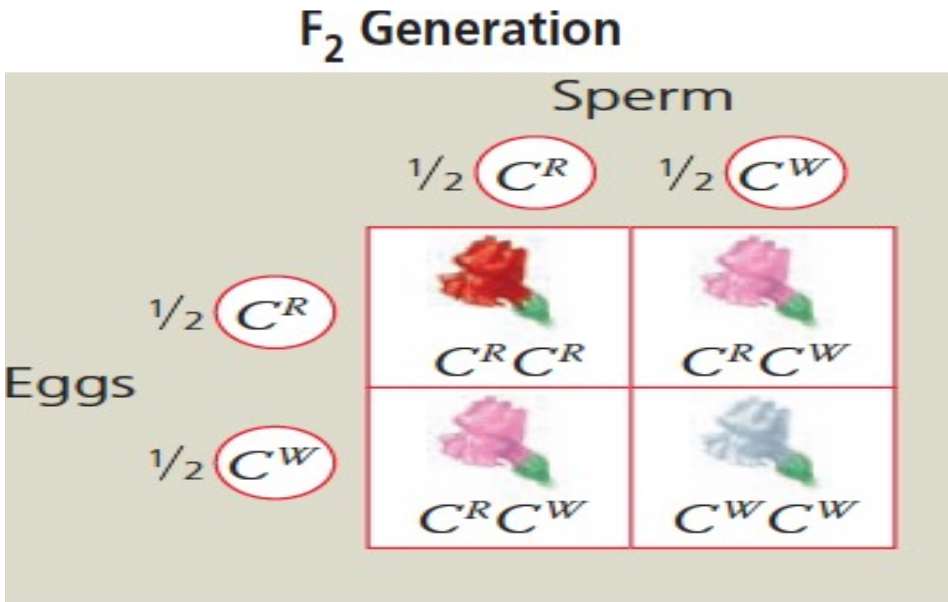
- In Mendelian genetics each character was determined by one gene, for which there are only two alleles (completely dominant or completely recessive)
- Relationship between genotype and phenotype are not always as simple!

- 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

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

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

Parents

Normal *Aa* X Normal *Aa*

Sperm: *A*, *a*

Eggs: *A*, *a*

Eggs \ Sperm	*A*	*a*
A	*AA* Normal	*Aa* Normal (carrier)
a	*Aa* Normal (carrier)	*aa* Albino

Lack of pigmentation

- Basics & terminology ✓
- Mendel and Inheritance ✓
- Law of segregation ✓
- Law of independent assortment ✓
- Genetics & Rules of Probability ✓
- Ex. & deviations ✓
- Blood grouping expt

Multiple Alleles

ABO Blood Grouping

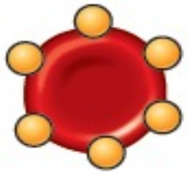
- A person's blood group may be one of four types: A, B, AB or O.
- These letters refer to two carbohydrates—A and B, which are found on RBC surface.
- Blood cells may have carbohydrate A (type A blood), carbohydrate B (type B), both (type AB), or neither (type O).

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^AI^A or I^Ai	I^BI^B or I^Bi	I^AI^B	ii
Red blood cell appearance				
Phenotype (blood group)	A	B	AB	O

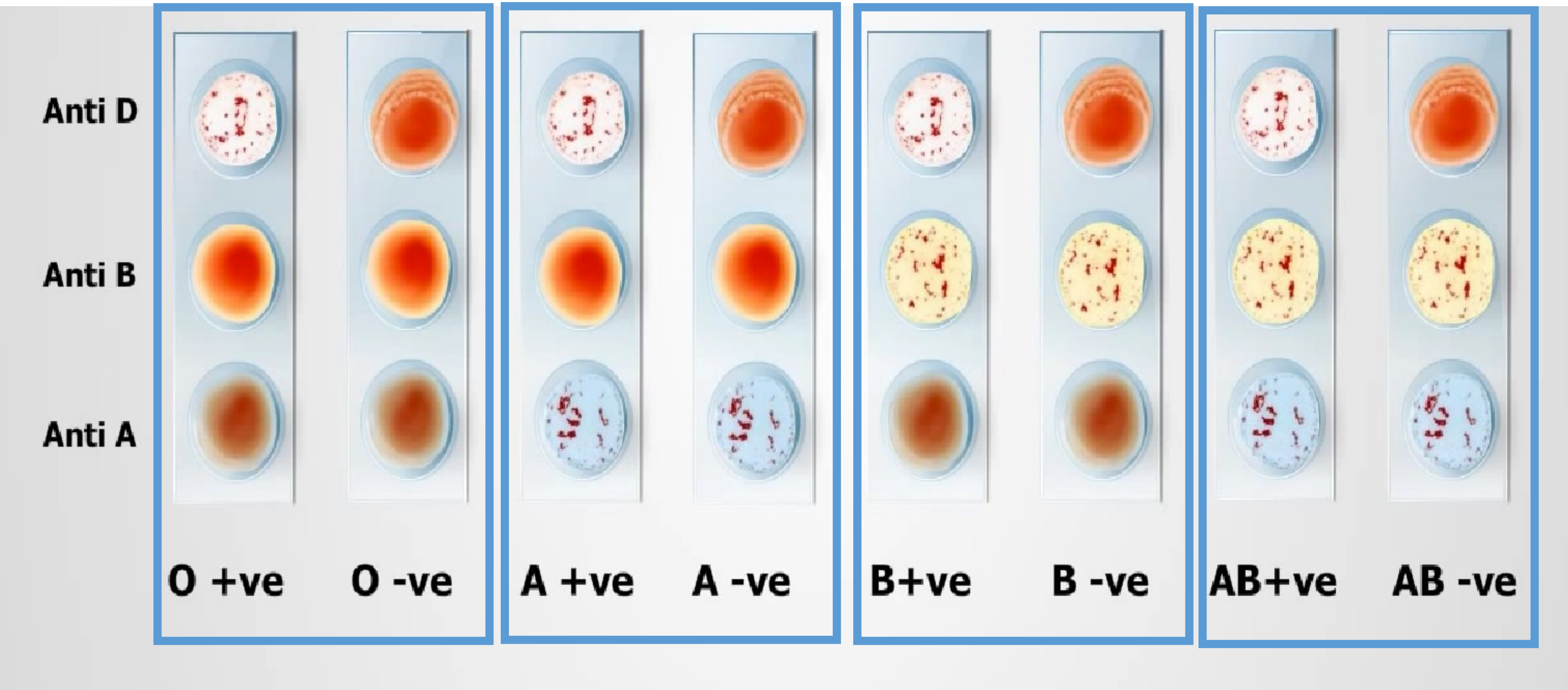
Blood group genotypes and phenotypes

Blood Group & Co-dominance

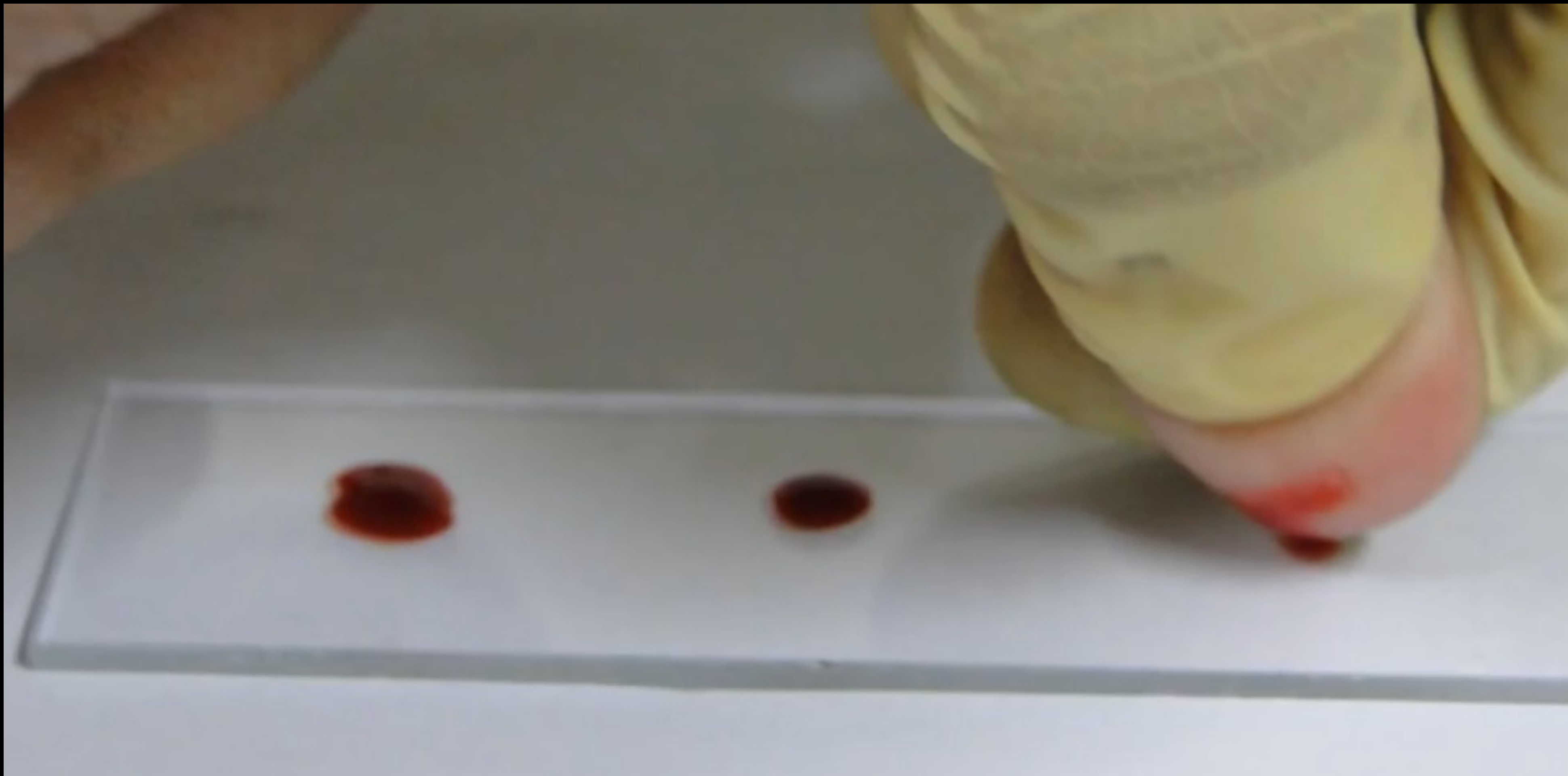
- **A** and **B** blood groups are **dominant** over **O** blood group
- A and B group genes are co-dominant

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

ABO Blood Grouping



Lab Experiment

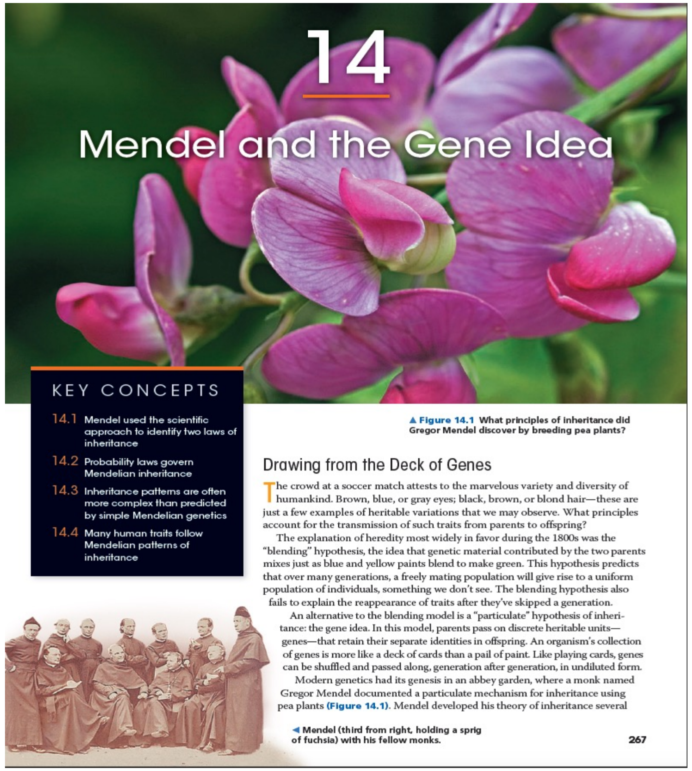


Summary

- **Basics & Terminology:**
  - Genetics studies heredity and traits through genes and alleles, determining genotype and observable phenotype.
- **Mendel and Inheritance:**
  - Gregor Mendel's pea plant experiments unveiled true-breeding traits and laid the foundation for modern genetics.
- **Laws of Inheritance:**
  - Mendel's Law of Segregation explains allele separation, while the Law of Independent Assortment describes gene independence during inheritance.
- **Genetics & Probability:**
  - Probability rules guide predictions of genotypes and phenotypes using tools like Punnett Squares.
- **Examples & Deviations:**
  - Incomplete Dominance, Codominance, and Multiple Alleles showcase variations in genetic expression.
- **Blood Grouping Experiment:**
  - ABO Blood Groups, exhibiting codominance, and the Rh Factor demonstrate practical applications of Mendelian principles in determining blood types.

References

- Campbell Biology - Reece, Urry, Cain, Wasserman, Minorsky, Jackson 10th Edition, Pearson
- *Acknowledgment*
  - Cover images – getty images



14

Mendel and the Gene Idea

KEY CONCEPTS

14.1 Mendel used the scientific approach to identify two laws of inheritance

14.2 Probability laws govern Mendelian inheritance

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

14.4 Many human traits follow Mendelian patterns of inheritance

▲ Figure 14.1 What principles of inheritance did Gregor Mendel discover by breeding pea plants?

Drawing from the Deck of Genes

The crowd at a soccer match attests to the marvelous variety and diversity of humankind. Brown, blue, or gray eyes; black, brown, or blond hair—these are just a few examples of heritable variations that we may observe. What principles account for the transmission of such traits from parents to offspring?

The explanation of heredity most widely in favor during the 1800s was the "blending" hypothesis, the idea that genetic material contributed by the two parents mixes just as blue and yellow paints blend to make green. This hypothesis predicts that over many generations, a freely mating population will give rise to a uniform population of individuals, something we don't see. The blending hypothesis also fails to explain the reappearance of traits after they've skipped a generation.

An alternative to the blending model is a "particulate" hypothesis of inheritance: the gene idea. In this model, parents pass on discrete heritable units—genes—that retain their separate identities in offspring. An organism's collection of genes is more like a deck of cards than a pail of paint. Like playing cards, genes can be shuffled and passed along, generation after generation, in undiluted form.

Modern genetics had its genesis in an abbey garden, where a monk named Gregor Mendel documented a particulate mechanism for inheritance using pea plants (Figure 14.1). Mendel developed his theory of inheritance several

◀ Mendel (third from right, holding a sprig of fuchsia) with his fellow monks.

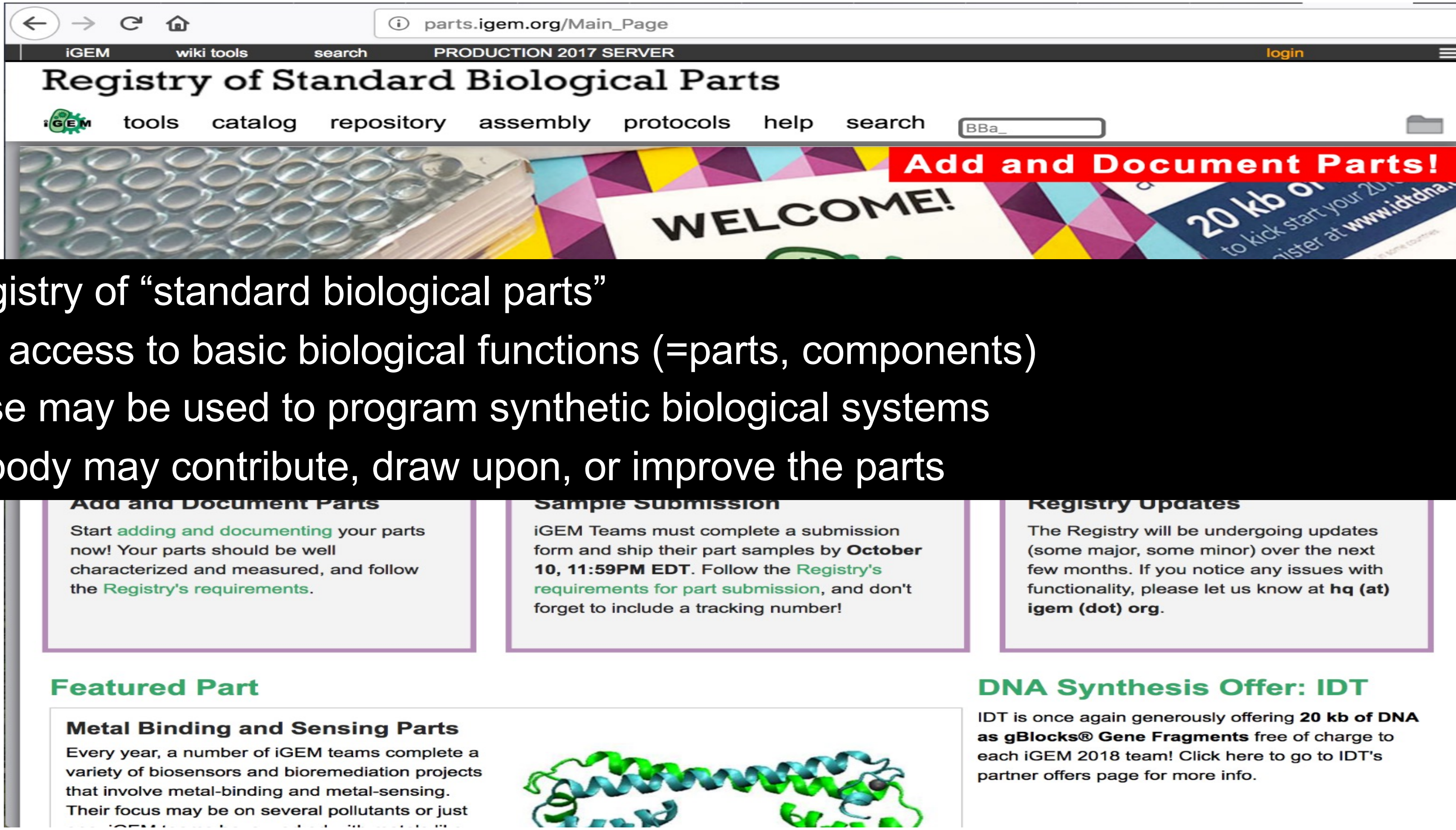
267

Next Lecture…

DNA as a genetic material

Information only… iGEM

International Genetically Engineered Machines (iGEM)



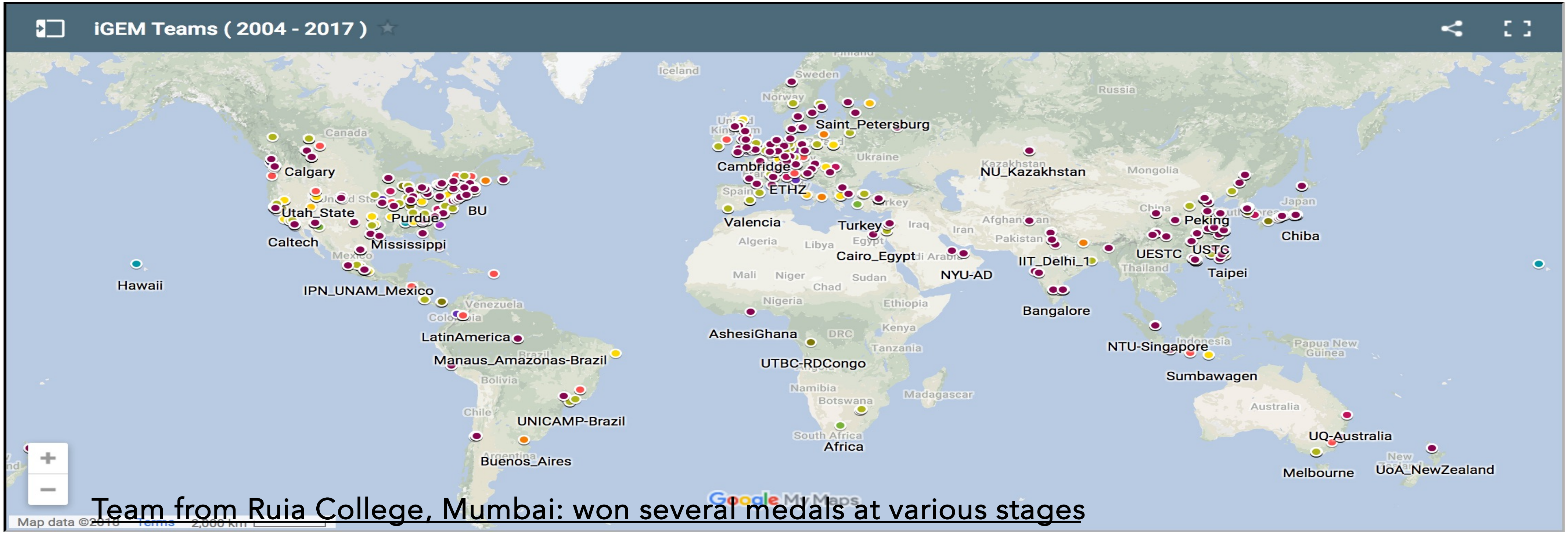
A registry of "standard biological parts"

Free access to basic biological functions (=parts, components)

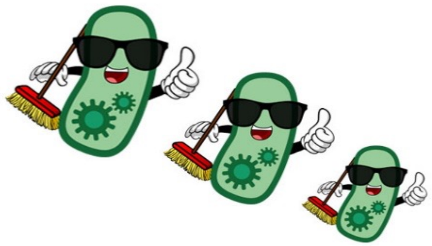
These may be used to program synthetic biological systems

Anybody may contribute, draw upon, or improve the parts

iGEM teams



Team from Ruia College, Mumbai: won several medals at various stages



Bacteria that remove “paan” stains